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TEXTS AND READINGS IN MATHEMATICS 40

A Course in Applied Stochastic Processes

A. Goswami
B.V. Rao

HINDUSTAN BOOK AGENCY

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Applied Stochastic Processes

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PREFACE: WHAT IS ALL THIS ABOUT?

This book grew out of lectures given by the authors — over various semesters spanning the last decade — at the Indian Statistical Institute, in a course with the same name for students at Masters' level in Statistics. The main aim of the book is to illustrate some applications of elementary theory of Stochastic Processes in various applied fields. We emphasize that this book is only an introduction to applications in certain fields and by no means an extensive account of all possible applications. There are many areas where theory of Stochastic Processes finds important applications that have been left out of the purview of this book. It is only hoped that this book will be able to create the right initiation so that an interested reader may move on to learning applications in his/her field of interest.

This book is intended for both undergraduate as well as graduate students in Statistics. This may be particularly useful for those who are interested in pursuing further studies in applications of Probability Theory. As a further justification, if one is needed, we would like to add the following. Several interesting and deep applications of elementary theory of Stochastic Processes in many different fields have been known for a long time. In fact, it is such applications that have driven much of the research of many eminent probabilists like W. Feller, A. N. Kolmogorov and B. V. Gnedenko, and more recently, S. Karlin, U. Grenander, P. Diaconis and M. Talagrand. Demands of such applications, in turn, have enriched the theory of Stochastic Processes. One case in point is the beautiful application of the theory of Finite Markov Chains in Genetics.

Unfortunately, this applied aspect of Probability Theory, in general, and Theory of Stochastic Processes, in particular, seem to have been largely ignored in the Statistics curriculum of Indian universities. This is evidenced by the lack of any specific course — at either the undergraduate or the masters' level — meant exclusively for such applications. This is despite the fact that the necessary theory of Stochastic Processes is covered in any standard masters' course in Statistics. Part of the reason could be the lack of easily available text books primarily devoted to such applications. We hope that the present book will help remove that void and provide an impetus for people to seriously think of having a course based on such beautiful applications. Even otherwise, this can be used as a supplementary book in any regular course on Stochastic Processes. Here is another way in which we believe the present book may be useful. Imagine a fresh Ph.D. student who wants to carry out research in Applied Stochastic Processes. Where does she start from? The current literature is filled with diverse applications at equally diverse levels — Finite Markov Chains in Learning Models to Diffusion Processes in Population Genetics to Infinite-Dimensional Stochastic Calculus in Mathematical Finance to Large Deviation Methods in Spin Glass Theory — making it difficult to choose a starting point. Hopefully the present book will provide one.

In terms of pre-requisites, the book does not demand much. Although an exposure to elementary Probability Theory would help, it is by no means essential. Chapter 0 is meant to supply the necessary background in Probability. The only real pre-requisite is an exposure to undergraduate Linear Algebra and Calculus, and of course, the necessary motivation.

We would like to highlight one interesting feature of the present book. A substantial amount of basic Martingale Theory and Theory of Diffusion Processes have been presented in this book and we have been able to do this without resorting to any measure- theoretic framework. We have not just 'conveyed the idea without rigour' — in most cases, we have given completely rigorous proofs.

Here is a brief summary of what the reader is in for. Chapter 0 gives a brief introduction to the necessary background in Probability. It almost starts from scratch and takes the reader through to Martingale Theory, Markov Chains, and a little of Diffusion Processes. Chapter 1 discusses the elementary theory of Discrete Time one-dimensional Branching Processes à la Galton-Watson. Much of the material covered here is available in the books of Harris, Feller and Karlin, as referred to at the end of the chapter. Chapter 2 is preparatory to Chapter 3. It contains the necessary introduction to Mathematical Genetics and the relevent Probability Models. An important topic here is the Hardy-Wienberg Laws. Some of the basic concepts in Population Genetics like Selfing, Sibmating, Gene Identity are elaborately discussed in this chapter and some related mathematical analyses are presented. Chapter 3 contains one of the most important and interesting topics of this book. We mainly discuss various Markov Chain models in Population Genetics. Of course the classical Wright-Fisher Model is the starting point. But many other models, not easily available in standard texts, are discussed at length. Towards the end, some nice Diffusion approximations to these Markov Chains are also discussed. Chapter 4 discusses Stochastic models in the spread of Epidemics. Some non-Stochastic models are discussed first to create motivation for their Stochastic counterparts. It is only in this chapter that we use some Continuous Time Markov Chain models. The most important topic in this chapter — at least in our opinion — consists of the Threshold Theorems. These theorems are believed to depict, in a nut shell, the temporal spread of Epidemics. At the end of each chapter, we have given a list of references as suggested supplementary readings. This is primarily aimed at readers who might take an active interest in pursuing further studies in these areas. Each of the chapters contain a fairly large number of exercises and except in Chapter 0, these exercises are given at the end of the chapters. In Chapter 0, the exercises are spread out over various sections. Sometimes the exercises are accompanied by enough hints, whenever deemed necessary. It is advisable for a serious reader to attempt the exercises as far as possible. Many of the exercises are taken from the various sources referred to throughout the book. For the sake of brevity we refrain from specific acknowledgements. The index at the end should be of help for a quick reference to important concepts and definitions. With a few exceptions, items are listed only according to their first appearance.

We usually cover chapters 1 through 4, in a one-semester M.Stat. course in our Institute. Most of Chapter 0 is not necessary in that course, because the students come with a fairly extensive exposure to basic Probability Theory and the theory of Markov Chains. It may be a little too tight to cover all this material in one semester, if one also has to do a substantial part from Chapter 0. In such cases, one of the following two options may be tried: Chapter 0 plus chapters 2 through 4 *or* Chapter 0 plus chapters 1 through 3. There should be other meaningful ways of making a coherent one-semester course out of selected material from this book.

We would like to acknowledge support from our various colleagues at the Institute. Special thanks are due to Professor T. Krishnan for giving the first impetus to undertake the work, supporting it throughout and also for his innumerable queries 'Finished?', usually with a meaningful smile towards the later stages, as he felt that the end of the project was nowhere in sight. This book, perhaps, would not have taken shape without him. We thank Professor K. K. Roy for using a preliminary draft of the book in his course. Professor Arup Bose patiently went through the first draft of the book and pointed out a number of typos and mistakes. We thank him for that. We got many valuable suggestions and pointers to typos from the anonymous referees. We have tried to incorporate many of the suggestions. We are indebted to the referees. Finally we thank Professors R. L. Karandikar and R. Bhatia for making it possible for the book to be published in the TRIM series.

A. Goswami B. V. Rao

Chapter 0

PROBABILITY TOOLS AND TECHNIQUES

0.1 Probabilities and Conditional Probabilities

The theory of probability is a mathematical theory to analyze experiments with multiple outcomes where one does not know a priori which outcome will actually occur. Such experiments are usually called *random experiments.* A natural and accepted way to model such phenomena is to associate a number called *probability* to each possible outcome. These numbers are supposed to reflect the chances of occurrence of the different outcomes. How these numbers are arrived at (more specifically, the numerical value of these probabilities) is not the major concern in developing a mathematical model. It must however be noted that in practical applications of probability models, these numerical values would matter in determining how close the model is to reality. Before we go to the axiomatic definition of probability, here are a few simple and familiar examples.

Example 1: The simplest example of a random experiment is tossing a coin. Here there are two possible outcomes: either the coin lands Head up *or* Tail up. The two possibilities can conveniently be denoted by H and T respectively. A mathematical model would then associate two numbers p and q which will denote the probabilities of H and T respectively. At this point let us agree on the following convention. First, we want the chances to be non-negative numbers and second, we want the chances of all possible outcomes to add up to one. Instead of trying to justify this, let us note that this is consistent with one's intuition of 'chances'. In the absence of a priori knowledge, one is inclined to believe that p and q should be equal, which according to the above convention forces $p = q = \frac{1}{2}$.
The above model can be thought of as an abstraction of any dichotomous experiment, that is, an experiment with two possible outcomes. For example, consider a machine manufacturing bolts where each bolt produced by the

machine has a chance of being defective. Here again we have two outcomes: defective and non-defective. We can still label them as H and T. Of course, in this case $p = q = \frac{1}{2}$ does not appear realistic because any reasonable machine is expected to produce a much larger proportion of non-defective items than defective items.

Example 2: Consider a usual six-faced die with faces numbered 1 through 6. If it is rolled once, any one of the six faces may show up. So there are six outcomes which could be denoted by the numbers 1 through 6. If nothing else is known, it seems intuitively clear that each of these outcomes should have probability 1/6.

Example 3: Pick up a name at random from the telephone directory and consider the first letter. It can be any one of the 26 letters of the alphabet. At the same time, not all the letters are equally likely to appear. For example, one certainly does not expect the letter X to occur as frequently as B. Thus it would not be reasonable to attribute equal probabilities to all the outcomes.

All the above examples show that a random experiment consists of two ingredients: first, the set of possible outcomes, to be called the *sample space* — denoted by Ω, and second, an assignment of probabilities to the various outcomes. Of course, in all the above examples, the set Ω is only a finite set, that is, $\Omega = \{\omega_1, \ldots, \omega_n\}$. In this case probability assignment means assigning non-negative numbers $p_1, \ldots, p_n$ adding up to unity, where the number p_i denotes the probability of the outcome ω_i. We write $P(\{\omega_i\}) = p_i$. Often we will be interested not in individual outcomes but with a certain collection of outcomes. For example, in rolling of a die we may ask: what is the probability that an even-numbered face shows up? In the context of a name being selected from the telephone directory we may ask: what are the chances that the letter is a vowel? These are called *events*. In general an event is any subset of the sample space. The probability of an event A is defined by

$$P(A) = \sum_{\omega \in A} P(\{\omega\})$$

where $P(\{\omega\})$ denotes the probability of the outcome ω.

Example 4: Suppose we roll a die twice. The sample space is

$$\Omega \quad = \quad \{(i,j); 1 \leq i \leq 6; 1 \leq j \leq 6\}$$

We assign equal probabilities to all the 36 outcomes, that is, for any $\omega \in \Omega$, $P(\{\omega\}) = 1/36$. If A is the event described by "first face is even", then A consists of $\{(i,j) : i = 2,4,6; 1 \leq j \leq 6\}$ and $P(A) = 1/2$. If A is described by "sum of the two faces is 5" then A consists of $\{(1,4),(2,3),(3,2),(4,1)\}$ and $P(A) = 1/9$.

As the above example shows, if, in general, we have a finite sample space with all outcomes equally likely, then for any event A, $P(A) = |A|/|\Omega|$ where, for any set B, $|B|$ denotes the number of elements of the set B. In these situations, probability computations become a combinatorial exercise.

In any case, equally likely or not, one can easily verify that probabilities of events satisfy the following properties:

1. $0 \leq P(A) \leq 1$, $P(\Omega) = 1$.
2. $P(A \cup B) = P(A) + P(B)$ whenever $A \cap B = \emptyset$.
   In particular, $P(A^c) = 1 - P(A)$.

So far we have restricted ourselves only to finite sample spaces but the same idea as described in the paragraph following Example 3 applies also to situations where Ω is countably infinite. With $\Omega = \{\omega_1, \omega_2, \ldots\}$ and non-negative numbers $p_1, p_2, \ldots$, adding to unity, one can define $P(A) = \sum_{\omega_i \in A} p_i$ for $A \subset \Omega$, as probability of the event A. One needs only to notice that the sum appearing in the definition of $P(A)$ may now be an infinite series. But with usual caution as necessary while dealing with infinite sums, one can show that the above properties hold and one has moreover,

3. $P(A_1 \cup A_2 \cup \cdots) = P(A_1) + P(A_2) + \cdots$ if $A_i \cap A_j = \emptyset$ for $i \neq j$.

We now give a formal definition of probability.

Definition: Let Ω be a countable set. A *probability* on Ω is a function P defined on all subsets of Ω satisfying the following conditions.
(0) $P(\emptyset) = 0$ and $P(\Omega) = 1$
(1) $P(\cup_i A_i) = \sum_i P(A_i)$ if $A_i \cap A_j = \emptyset$ for $i \neq j$.

The next few exercises list some standard properties that are easy consequences of the definition.

Exercise 1: Let P be a probability on Ω. Then
(a) $0 \leq P(A) \leq 1$; $P(A^c) = 1 - P(A)$; if $A \subset B$ then $P(A) \leq P(B)$.
(b) $P(A \cup B) = P(A) + P(B) - P(A \cap B)$. More generally,

$$P(\bigcup_1^n A_i) = S_1 - S_2 + S_3 - \cdots \tag{1}$$

where S_i denotes the sum of probabilities of i-fold intersections.
(c) If $A_n \uparrow A$ then $P(A_n) \uparrow P(A)$. If $A_n \downarrow A$ then $P(A_n) \downarrow P(A)$.

Exercise 2: For a sequence (B_n) of events, one defines

$$\limsup_n B_n = \bigcap_n \bigcup_{k \geq n} B_k.$$

Show that $\limsup_n B_n$ is the event that B_n occurs for infinitely many n (sometimes described as the events B_n occurring *infinitely often*). Show that if $\sum P(B_n) < \infty$, then $P(\limsup_n B_n) = 0$. This is called *(the first) Borel-Cantelli Lemma.*

Exercise 3: Suppose that p is a non-negative function on Ω such that $\sum_\omega p(\omega) = 1$. Then $P(A) = \sum_{\omega \in A} p(\omega)$ defines a probability on Ω.

From now on, by a random experiment, we mean a pair (Ω, P) where Ω is a non-empty countable set and P is a probability on Ω. The number $P(A)$ represents the probability that the event A will occur when the random experiment is performed. Of course, if the experiment is really performed and we know the exact outcome, there is no need for probabilities. Probability of an event is really an assessment of the chance of occurrence of the event irrespective of whether the experiment is actually conducted and we know the outcome or not. However, sometimes we may have a situation where a random experiment is performed and some partial information is available to us about the outcome and we are to assess the chances of an event taking this additional information into account. It is intuitively clear that we should modify probability assignments of events in the presence of this additional information.

Consider the example of rolling a die twice with all outcomes being equally likely. The probability that the first face is 3 is already known to be $1/6$. But suppose now we have the additional information that the sum of the two faces is 5. This information already tells us that the outcome must be among $(1,4)$, $(2,3)$, $(3,2)$ and $(4,1)$, so that the chance of first face being 3 is now $1/4$. Such probabilities are called *conditional probabilities*. More precisely, if A is the event that the first face is 3 and B is the event that the sum of the two faces is 5, then the unconditional probability of A is $1/6$ whereas the conditional probability of A given that B has occured is $1/4$. This later probability is denoted $P(A \mid B)$. Here is the general definition.

Definition: Let (Ω, P) be a random experiment and let $B \subset \Omega$ be an event with $P(B) > 0$. Then for any event A, the *conditional probability* of A given the event B is defined by

$$P(A \mid B) = \frac{P(A \cap B)}{P(B)}. \tag{2}$$

In the equally likely case (as in the earlier example) this reduces to

$$P(A \mid B) = \frac{|A \cap B|}{|B|}.$$

The following can be easily verified:

Theorem 0.1:

1. *Fix B and let $P_B(A) = P(A \mid B)$, then P_B is a probability on Ω.*
2. *$P(A \cap B \mid C) = P(A \mid B \cap C)\, P(B \mid C)$. More generally,*

$$P(A_1 \cap \cdots \cap A_n \mid A_{n+1}) = \prod_{j=1}^{n} P(A_j \mid A_{j+1} \cap \cdots \cap A_{n+1}).$$

3. If $B_1, \ldots, B_n$ is a partition of Ω then for any event A

$$P(A) = \sum P(A \mid B_i)\, P(B_i).$$

More generally,

$$P(A \mid C) = \sum P(A \mid B_i \cap C)\, P(B_i \mid C).$$

4. If $B_1, \ldots, B_n$ is a partition of Ω then for any event A

$$P(B_i \mid A) = \frac{P(A \mid B_i)\, P(B_i)}{\sum_j P(A \mid B_j)\, P(B_j)}.$$

Exercise 4: $P(A \mid B) \leq P(A)$ if and only if $P(B \mid A) \leq P(B)$. In particular, $P(A \mid B) = P(A)$ if an only if $P(B \mid A) = P(B)$.

Let us return to the example of rolling a die twice. Let, as earlier, A be the event that the first face is 3 and B be the event that the sum of the two faces is 5. Then $P(A \mid B) = 1/4 > 1/6 = P(A)$. So here the additional information has the effect of increasing the chances of A. On the other hand if we consider the event C that the sum is 11, then clearly $P(A \mid C) = 0$, that is, the additional information reduces the chances of A (to indeed zero!). Does it always happen this way? That is, will additional information always change the chances one way or other? The answer is NO. For example if D is the event that the sum is 7, then $P(A \mid D) = 1/6 = P(A)$. That is, the probability of A remains unchanged even if we are told that D has occured. This situation is described by saying that A is *independent* of D. Here is the precise definition.

Definition: Two events A and B are said to be *independent* if $P(A \cap B) = P(A)P(B)$.

Of course when one of the two events, say, B has positive probability then A and B are independent is the same as saying $P(A \mid B) = P(A)$.

Exercise 5: If A, B are independent, then A^c, B are independent ; A, B^c are independent ; A^c, B^c are independent.

Definition: Events A_1, A_2, ..., A_n are said to be *independent* if for any $1 \leq i_1 < i_2 < \cdots < i_k \leq n$

$$P(A_{i_1} \cap A_{i_2} \cap \cdots \cap A_{i_k}) = P(A_{i_1})P(A_{i_2}) \cdots P(A_{i_k}). \tag{3}$$

Exercise 6: Let A_1, A_2, ..., A_n be independent.
(i) If for each i, B_i denotes one of the events A_i or A_i^c then B_1, B_2, ..., B_n are independent.
(ii) If $1 \leq j < n$, $\cap_{1 \leq j} B_i$ is independent of $\cap_{i>j} B_i$. $\cup_{1 \leq j} B_i$ is independent of $\cap_{i>j} B_i$. $\cup_{1 \leq j} B_i$ is independent of $\cup_{i>j} B_i$. Here B_i are as in (i).

The assertions in (ii) above are merely special cases of a more general phenomenon: if $1 \leq j < n$ and C is an event "constructed" out of $A_1, \ldots, A_j$ and

D is an event constructed out of $A_{j+1}, \ldots, A_n$, then C and D are independent events. This is intuitively clear, but a formal proof requires more machinery than what is available at this level.

Often random experiments can be thought of as composed of simpler random experiments in the sense explained below. If you toss a coin twice you can describe the outcomes of the experiment by $\Omega = \{HH, HT, TH, TT\}$. Notice that $\Omega = \{H, T\} \times \{H, T\}$, that is, Ω is the two-fold product of a single toss experiment. More generally, the sample space for 10 tosses of a coin (or a toss of 10 coins) can be thought of as the ten-fold product of $\{H, T\}$. But what is important is that not only the sample space can be thought of as a product, but the probabilities of the outcomes can also be thought of as products. Here is the general method.

Let (Ω_i, P_i), for $1 \leq i \leq n$, be random experiments. Put

$$\Omega = \Omega_1 \times \Omega_2 \times \cdots \times \Omega_n = \{(\omega_1, \ldots, \omega_n) : \forall i, \quad \omega_i \in \Omega_i\}.$$

For $\omega = (\omega_1, \ldots, \omega_n) \in \Omega$, put $P(\{\omega\}) = P_1(\{\omega_1\}) \times \cdots \times P_n(\{\omega_n\})$. One can now define $P(A)$ for any $A \subset \Omega$, thus getting a probability P on Ω.

Exercise 7: If $A = A_1 \times A_2 \times \cdots \times A_n$ then $P(A) = \prod_{i=1}^{n} P_i(A_i)$. Conclude that if $\widetilde{A_i} \subset \Omega$ is the set of all points in Ω whose i-th coordinate is in A_i then $\widetilde{A_1}, \ldots, \widetilde{A_n}$ are independent.

The exercise above really means that events that depend on different coordinates are independent. This, of course, is a consequence of the way the probability P has been defined on Ω. It is clearly possible to construct other probabilities P on Ω, such that $P(\widetilde{A_i}) = P_i(A_i)$ for all i, but independence fails. One can easily see that 10 tosses of a coin with all outcomes equally likely is the same as the ten-fold product of single toss of coin with $P(H) = P(T) = 1/2$.

If $\Omega_1 = \Omega_2 = \cdots = \Omega_n$, then we write $\Omega = \Omega_1^n$. If further $P_1 = P_2 = \cdots = P_n$, then we write $P = P_1^n$. (Ω_1^n, P_1^n) represents n independent repetitions of the experiment (Ω_1, P_1).

0.2 Random Variables and Distributions

In the context of random experiments, the actual outcomes may often be quite abstract. For example, if you toss a coin 10 times, outcomes will be 10-tuples of H's and T's. Often one is interested not in the exact outcome per se but some numerical value associated with each outcome. For example, in case of 10 tosses of a coin, one may be interested in the number of times heads showed up *or* in the number of times a tail was immediately followed by a head. Such numerical values associated with outcomes are what are called *random variables.* This section is devoted to a study of random variables and their distributions.

0.2.1 Distribution of a Random Variable

Definition: A *random variable* is a real-valued function defined on the sample space Ω.

It is customary to denote random variables by X, Y, Z etc. For example, in 10 tosses of a coin, let X denote the total number of heads and Y denote the number of times a tail is immediately followed by a head. Then for the outcome $\omega = HTTHTTTHHH$, $X(\omega) = 5$ and $Y(\omega) = 2$, while for another outcome $\omega' = THHTHTHHTH$, $X(\omega') = 6$ and $Y(\omega') = 4$.

Given a random variable, we can ask what the possible values of the random variable are and the chances (probabilities) of it taking each of those values. This is what is called the *distribution* of the random variable. Since our sample space is countable, any random variable can only take countably many values.

Definition: Let X be a random variable on (Ω, P). Then by the *distribution* of X is meant the set of possible values $D = \{x_1, x_2, \ldots\}$ of the random variable X and the probabilities $\{p(x_1), p(x_2), \ldots\}$ where $p(x_i) = P(\omega : X(\omega) = x_i)$. The right side is often abbreviated as $P(X = x_i)$.

Of course, p can be extended to a function on R by setting $p(x) = P(X = x)$. However, for any $x \notin D$ we have $p(x) = 0$. This p is called the *probability mass function* (p.m.f.) of the random variable X.

Once we know the probability mass function of a random variable X, we can compute for any $A \subset R$, the probability $P(X \in A)$ by the formula

$$P(X \in A) = \sum_{x \in A} p(x).$$

Example 1: Consider n independent tosses of a coin. Assume that in each toss the probability of heads is p. Define X to be the total number of heads obtained. Clearly X is a random variable which can take any integer value from 0 to n. One might wonder: how do we get a random variable even before describing the sample space. We concede that we were jumping steps. So here is our sample space: $(\Omega, P) = (\Omega_1^n, P_1^n)$ where $\Omega_1 = \{H, T\}$; $P_1(H) = p$ and $P_1(T) = 1 - p$. The definition of the random variable X as a real-valued function on Ω should now be clear. It is also easy to verify that the probability mass function of X is given by

$$\begin{aligned} p(x) &= \binom{n}{x} p^x (1-p)^{n-x} && \text{for } x \in \{0, 1, \ldots, n\} \\ p(x) &= 0 && \text{for } x \notin \{0, 1, 2, \ldots, n\} \end{aligned}$$

This random variable is called the *Binomial* random variable with parameters n and p, in short, a $B(n, p)$ random variable. We write $X \sim B(n, p)$ for this. The distribution is called the *Binomial distribution.*

Almost all the information about the random variable X is contained in its distribution (or its p.m.f.) — the underlying sample space or the precise definition of X as a function on the sample space is of no additional importance.

Therefore it is often customary to describe random variables simply by their distributions without any reference to any underlying sample space.

Example 2: Fix a number p with $0 < p < 1$. A random variable X is said to have $G(p)$ distribution — *geometric distribution with parameter* p — if X takes value x with probability $p(1-p)^x$ for $x \in \{0, 1, \ldots\}$. In other words, X has p.m.f.

$$p(x) = p(1-p)^x \quad \text{for} \quad x \in \{0, 1, \ldots\}$$

It is to be understood here and elsewhere that $p(x) = 0$ for all other x. Suppose you have a coin with chance of heads p. If the coin is tossed repeatedly until a head shows up, then the number of tails preceeding the head has this geometric distribution.

Example 3: Here is a generalization of the above example. Again we have a coin for which the chance of a head in each toss is p. Fix an integer $m \geq 1$. Toss the coin until a total of m heads show up. (What is the sample space?) The random variable X is the total number of tails obtained. Clearly X takes values $x = 0, 1, 2, \ldots$ as earlier. A simple combinatorial argument shows that $P(X = x) = \binom{x+m-1}{x-1}(1-p)^x p^m$. This random variable is called a *negative binomial* random variable with parameters (m, p) — in short, $NB(m, p)$ random variable — and the distribution is called the negative binomial distribution (why?). Clearly when $m = 1$, we get the geometric random variable of Example 2.

Example 4: Fix integers N, $n < N$ and $N_1 < N$. A random variable X is said to be $Hyp(N, N_1; n)$ — *hypergeometric with parameters* N, N_1 *and* n — if it takes value x with probability

$$p(x) = \binom{N_1}{x}\binom{N - N_1}{n - x} \Big/ \binom{N}{n}.$$

Of course you have to interpret $\binom{a}{b} = 0$ unless b is an integer with $0 \leq b \leq a$. This arises if you have a bunch of N items of which N_1 are good, the remaining are defective and you select a random sample of size n without replacement. The random variable in question is the number of good items in the sample.

Example 5: Fix a number $\lambda > 0$. A random variable X is said to be $P(\lambda)$, written $X \sim P(\lambda)$ — *Poisson with parameter* λ — if it takes value x with probability $e^{-\lambda}\lambda^x/x!$ for $x = 0, 1, 2, \ldots$. This random variable arises as a limiting case of the number of heads when you toss a coin a large number of times and the chance of heads in each toss is very small. For details see Section 0.3.

Example 6: Roll a fair die twice and let X be the sum of the two numbers obtained. Then X takes values

$$2, \quad 3, \ldots, \quad 7, \quad 8, \ldots, \quad 12$$

with probabilities given respectively by

$$1/36, 2/36, \ldots, 6/36, 5/36, \ldots, 1/36.$$

Suppose that a fair coin is tossed ten times and X is the number of heads. Clearly X can take any one of the values $0, 1, 2, \ldots, 10$ with different probabilities, the actual value depending on the outcome of the ten tosses. But if we were to choose one "representative value" of X without knowing the actual outcome, what would be a good candidate? One possibility is to consider the most probable value, which in this case is 5. However a commonly used and mathematically more tractable quantity is what is known as the *expected value*. As the next definition shows, this is weighted average of the possible values.

Definition: Let X be a random variable with set of values D and p.m.f. $p(x)$ for $x \in D$. If $\sum_{x \in D} |x|\, p(x) < \infty$ (automatically true if D is finite), then X is said to have a finite expectation and the *expected value* of X is defined to be

$$E(X) = \sum_{x \in D} x\, p(x)\,. \tag{4}$$

Thus, expected value of a random variable, when it exists, is the weighted average of its values, weighted by their probabilities. Expected value or *expectation* is also called the *mean value* or the *mean*.

If X is a random variable and $g : R \longrightarrow R$ is a function then clearly $g(X)$ is again a random variable. It is not difficult to check that $g(X)$ has finite expectation iff $\sum_{x \in D} |g(x)|\, p(x) < \infty$ and in that case $E(g(X)) = \sum_{x \in D} g(x)\, p(x)$. This is a very useful formula because we can compute $E(g(X))$ straight from the p.m.f. of X rather than having to go to the p.m.f. of the random variable $g(X)$.

Definition: A random variable X with $E(X^m)$ finite is said to have a *finite m-th moment*, given by $E(X^m)$. For X with a finite second moment, the *variance* of X, denoted $V(X)$, is defined by

$$V(X) = E[(X - EX)^2]\,. \tag{5}$$

The quantity $V(X)$ measures the spread of the distribution of X. For example, $V(X) = 0$ iff the distribution of X is concentrated at one point (that is, X is a constant random variable).

Indicator random variables as defined below form a very simple, yet useful, class of random variables.

Definition: For any event A the *Indicator random variable* of the event is defined as

$$I_A(\omega) = \begin{cases} 1 & \text{if} \quad x \in A \\ 0 & \text{if} \quad x \notin A \end{cases}\,.$$

Clearly the expectation of I_A is $P(A)$.

Exercise 1: If the m-th moment is finite, then so is the n-th, for any $n < m$.

Exercise 2. For a random variable X on a probability space (Ω, P), $E(X)$ exists iff $\sum_{\omega \in \Omega} |X(\omega)|\, P(\{\omega\}) < \infty$ and in that case $E(X) = \sum_{\omega \in \Omega} X(\omega)\, P(\{\omega\})$.

Exercise 3. If $P(X = c) = 1$ for some real number c, then $E(X) = c$.

Exercise 4. If X is a random variable with finite expectation, then $|E(X)| \leq E(|X|)$.

Exercise 5. If X and Y are two random variables defined on the same space and having finite expectations, then

(a) $X \leq Y$ implies $E(X) \leq E(Y)$.

(b) $E(aX + bY) = a\,E(X) + b\,E(Y)$ for any two real numbers a and b. In particular, $E(aX + b) = a\,E(X) + b$.

Exercise 6. If $P(X \geq 0) = 1$ and $E(X) = 0$, then $P(X = 0) = 1$. More generally, if $P(X \geq Y) = 1$ and $E(X) = E(Y)$, then $P(X = Y) = 1$.

Exercise 7. If X_n and X are non-negative random variables defined on the same space and $X_n \uparrow X$, then $E(X_n) \uparrow E(X)$. In case X has infinite expectation, this should be read as $E(X_n) \uparrow \infty$. This is known as Lebesgue's *Monotone Convergence Theorem.*

Exercise 8. Supose that X_n and X are random variables defined on the same space such that $X_n \to X$. Suppose also that there is a random variable Y with finite expectation such that $|X_n| \leq Y$ for all n, that is, all the random variables X_n are dominated in modulus by the random variable Y. Then $E|X_n - X| \longrightarrow 0$. In particular $E(X_n) \to E(X)$. This is called Lebesgue's *Dominated Convergence Theorem.*

Exercise 9. If X and Y are two random variables on the same space such that $E(X \cdot I_A) \geq E(Y \cdot I_A)$ for every event A then $P(X \geq Y) = 1$. In particular, $E(XI_A) = E(YI_A)$ for every A if and only if $P(X = Y) = 1$.

Exercise 10. $V(X) = E(X^2) - (EX)^2$.

Exercise 11. $V(X) = 0$ iff $P(X = c) = 1$ for some constant c.

Exercise 12. $V(aX + b) = a^2V(X)$.

Exercise 13. $V(I_A) = P(A)[1 - P(A)]$.

Exercise 14. If X has finite variance and $E(X) = \mu$, then $E(X - a)^2 \geq V(X)$ for every real a. Thus $E(X - a)^2$ is minimized when $a = \mu$.

Exercise 15. For each of the random variables in Examples 1 through 6, find its expected value and variance.

0.2.2 Joint Distributions

Suppose that X and Y are two random variables defined on the same space. As mentioned earlier, probabilities of events concerning the random variable X (respectively, Y) can be computed from the distribution of X (respectively, of Y). However we may often be interested in probabilities of events that concern both X and Y. For example, 'what is $P(X = Y)$?' or 'what is $P(X + Y = 7)$?' etc. For such probabilities individual distributions of X and Y alone would not suffice. We need to know what is called the *joint distribution* of X and Y.

Definition: Let X and Y be two random variables defined on the same space.

Let D_X and D_Y denote the set of possible values of the random variables X and Y respectively. The set of possible values of the pair (X, Y) are clearly contained in $D_X \times D_Y$. The *joint distribution* of (X, Y) is given by the *joint probability mass function* defined as $p(x, y) = P(X = x, Y = y)$ for $(x, y) \in D_X \times D_Y$.

Consider, for example, tossing a coin 15 times, with the chance of a head in each toss being p. Let X be the number of heads in the first ten tosses and Y be the number of heads in the last ten tosses. Clearly both X and Y are $B(10, p)$ random variables. Here $D_X = D_Y = \{0, 1, \ldots, 10\}$. The joint distribution of (X, Y) would be given by the mass function p on $D_X \times D_Y$. For example, $p(10, 10) = p^{15}$. In general,

$$p(m, n) = \sum_{k=0}^{5} \binom{5}{m-k}\binom{5}{k}\binom{5}{n-k} p^{m+n-k}(1-p)^{15+k-m-n}$$

with the usual convention that $\binom{a}{b} = 0$ unless b is integer with $0 \leq b \leq a$.

From the joint p.m.f. of (X, Y), the individual (marginal) p.m.f. of X and Y can be obtained as follows:

$$p_1(x) = P(X = x) = \sum_{y \in D_Y} p(x, y) \quad \text{for} \quad x \in D_X$$

$$p_2(y) = P(Y = y) = \sum_{x \in D_X} p(x, y) \quad \text{for} \quad y \in D_Y$$

In an analogous way the joint distribution of n random variables (defined on the same space) is given by their joint p.m.f.

$$p(x_1, x_2, \ldots, x_n) = P(X_1 = x_1, X_2 = x_2, \ldots, X_n = x_n)$$

Example 7: Consider an n faced die with p_1, p_2, ..., p_n denoting the probabilities of different faces in a single throw. Roll the die r times and let X_i be the number of times face i shows up. The joint p.m.f. of $(X_1, X_2, \ldots, X_n)$ is given by

$$p(x_1, x_2, \ldots, x_n) = \frac{r!}{x_1! x_2! \cdots x_n!} p_1^{x_1} p_2^{x_2} \cdots p_n^{x_n}$$

for $x_1, x_2, \ldots, x_n$ non-negative integers adding to r. This distribution is called the *multinomial distribution with parameters* $(r; p_1, p_2, \ldots, p_n)$.

Definition: For a pair of random variables X and Y defined on the same space, the *covariance* between X and Y is defined as

$$Cov(X, Y) = E[(X - EX)(Y - EY)]. \tag{6}$$

Further, $E(X^m Y^n)$, for positive integers m and n, are called the various *cross-product moments* of the pair (X, Y).

Exercise 16. $Cov(X, Y) = E(XY) - E(X)E(Y)$.

Exercise 17. $Cov(\sum_i a_i X_i, \sum_j b_j Y_j) = \sum_i \sum_j a_i b_j Cov(X_i, Y_j)$.

Exercise 18. $Cov(X, X) = V(X)$.

Exercise 19. $Cov(X, a) = 0$ for any constant random variable a.

Exercise 20. $Cov(X, Y) \leq \sqrt{V(X)}\sqrt{V(Y)}$.

Exercise 21. $V(\sum_i X_i) = \sum_i V(X_i) + 2\sum_{i<j} Cov(X_i, X_j)$. In particular, if $Cov(X_i, X_j) = 0$ for all $i \neq j$, then $V(\sum_i X_i) = \sum_i V(X_i)$.

0.2.3 Conditional Distributions and Conditional Expectations

Let X be a random variable. For any event A with $P(A) > 0$, the conditional distribution of X given A simply means the conditional probabilities for X taking various values given the event A. Thus the conditional distribution is given by the (conditional) p.m.f. $p(x \mid A) = P(X = x \mid A)$. It is, of course, immediate that this is indeed a probability mass function. The conditional expectation and conditional variance of X given A are just the expectation and variance of this conditional distribution. Clearly all the properties listed in Exercises 4,5,7,8 and 10 through 14 can be formulated and shown to hold with conditional expectation and conditional variance.

Next, let X and Y be two random variables defined on the same space. For y with $P(Y = y) > 0$, we can talk about the *conditional distribution* of X given $Y = y$. This is given by the conditional mass function

$$p(x \mid y) = P(X = x \mid Y = y) = \frac{p(x, y)}{p_2(y)} \,. \tag{7}$$

Here p is the joint p.m.f. of (X, Y) and p_2 is the (marginal) p.m.f. of Y. It is clear that for each y with $p_2(y) > 0$, the function $p(\cdot \mid y)$ is a probability mass function — called the *conditional p.m.f.* of X given $Y = y$.

If X has finite expectation, then the *conditional expectation* of X given $Y = y$ is defined to be

$$E(X \mid Y = y) = \sum x\, p(x \mid y) \,. \tag{8}$$

The assumption of finite expectation ensures the convergence of the right hand side of Equation (8). Thus, the conditional expectation of X given $Y = y$ is just the expectation of X under the conditional distribution given $Y = y$. Clearly $E(X \mid Y = y)$ is a function of y, say $\phi(y)$. The random variable $\phi(Y)$ is denoted by $E(X \mid Y)$. We do this because, in many contexts it is convenient to think of the conditional expectation itself as a random variable. One can similarly define the conditional distribution of Y given $X = x$ and also $E(Y \mid X)$.

It may be noted that if Y is a constant random variable, say, $Y \equiv c$, then the conditional distribution as well as the conditional expectation of X given $Y = c$ reduce to the unconditional distribution and unconditional expectation

of X. The following facts on conditional expectation are easy to verify, and left as exercises.

Exercise 22. $E(E(X \mid Y)) = E(X)$.

Exercise 23. If X has finite expectation and if g is a function such that $Xg(Y)$ also has finite expectation, then show that $E(X\, g(Y) \mid Y) = E(X \mid Y)g(Y)$.

Exercise 24. $E(X - g(Y))^2 \geq E(X - E(X \mid Y))^2$ for any X and g such that X^2 and $(g(Y))^2$ have finite expectations. (Exercise 14 in 0.2.1 is easily seen to be a special case of the above.)

Exercise 25. For any function g such that $g(X)$ has finite expectation, $E(g(X) \mid Y = y) = \sum g(x)\, p(x \mid y)$.

Exercise 26. $|E(X \mid Y)| \leq E(|X| \mid Y)$.

The above notions of conditional distribution and conditional expectation naturally extend to the case of more then two random variables. To be precise, if $X_1, X_2, \ldots, X_n$ are random variables on the same space, one can, in a natural way, talk about the conditional joint distribution of k of these random variables given the others. For instance, the conditional joint distribution of $(X_1, \ldots, X_k)$, given $X_{k+1} = x_{k+1}, \ldots, X_n = x_n$ is defined by

$$p(x_1, \ldots, x_k \mid x_{k+1}, \ldots, x_n) = \frac{P(X_1 = x_1, \ldots, X_n = x_n)}{P(X_{k+1} = x_{k+1}, \ldots, X_n = x_n)}\,.$$

provided, of course, $P(X_{k+1} = x_{k+1}, \ldots, X_n = x_n) > 0$, and for each such $(x_{k+1}, \ldots, x_n)$, the function $p(\cdot \mid x_{k+1}, \ldots, x_n)$ is a p.m.f. — called the *conditional joint p.m.f.* of $(X_1, \cdots, X_k)$, given $X_{k+1} = x_{k+1}, \ldots, X_n = x_n$.

If g is a k-variable function such that $Y = g(X_1, \ldots, X_k)$ has finite expectation, then the conditional expectation of Y given $X_{k+1} = x_{k+1}, \ldots, X_n = x_n$, has a natural definition, namely

$$\begin{aligned} &E(Y \mid X_{k+1} = x_{k+1}, \ldots, X_n = x_n) \\ &\qquad = \sum_{x_1, \ldots, x_k} g(x_1, \ldots, x_k)\, p(x_1, \ldots, x_k \mid x_{k+1}, \ldots, x_n)\,. \end{aligned}$$

In particular, one can talk about the conditional expectation of X_1 given $X_2, \ldots, X_n$ or conditional expectation of $X_1^2 + X_2^2$ given X_3, X_5, and so on.

Exercise 27. $E(E(X \mid Y, Z) \mid Y) = E(X \mid Y)$. More generally

$$E(E(X \mid X_1, \ldots, X_n) \mid X_1, \ldots, X_{n-1}) = E(X \mid X_1, \ldots, X_{n-1})\,.$$

Here $E(Y \mid X_{k+1}, \ldots, X_n)$ denotes the random variable $\phi(X_{k+1}, \ldots, X_n)$ where ϕ is the $(n-k)$-variable function defined by

$$\phi(x_{k+1}, \ldots, x_n) = E(Y \mid X_{k+1} = x_{k+1}, \ldots, X_n = x_n)\,.$$

If these things look a little abstract there is no cause for alarm. Simply try to understand the meaning of the conditional expectation of X_1 given

$X_2, \ldots, X_n$ or the conditional expectation of $X_1^2 + X_2^2$ given X_3 and X_4. Here is a useful exercise left to be proved by the reader. This is often referred to as the *smoothing property* of conditional expectation.

$$E(E(g(X,Y)\,|\,Z,W)\,|\,Z) = E(g(X,Y)\,|\,Z)\,.$$

Or more generally, if $U = g(X_1, \ldots, X_m)$ then

$$E(E(U\,|\,Y_1, \ldots, Y_n)\,|\,Y_1, \ldots, Y_{n-1}) = E(U\,|\,Y_1, \ldots, Y_{n-1})\,. \tag{9}$$

Indeed, one may think of (9) as equivalent to Exercise 27 above. What this says is the following. In order to get the conditional expectation of a random variable given $Y_1, Y_2, \ldots, Y_{n-1}$, one may first calculate its conditional expectation given $Y_1, Y_2, \ldots, Y_n$ and then take the conditional expectation of this random variable given $Y_1, Y_2, \ldots, Y_{n-1}$. Here is an application.

Example 8: Toss a fair coin a Poisson number of times. Find the conditional expectation of the time of occurrence of the first Head, given the total number of Heads. More precisely, let N be a random variable having the Poisson distribution with parameter λ. Suppose that a fair coin is tossed N times. Let X be the number of Heads obtained and T be the time of occurrence of the first Head. In case there are no Heads, T is defined to be one plus the number of tosses, that is to say, $T = 1 + N$ in case $X = 0$. Of course, if $N = 0$, then $X = 0$ automatically so that $T = 1$. We want $E(T\,|\,X = x)$ for each $x \geq 0$.

The plan is the following. We first compute $E(T\,|\,X, N)$ and then compute its conditional expectation given X. By the smoothing propertry this will be the same as $E(T\,|\,X)$.

For integers $0 \leq x \leq n$, let $f(n,x) = E(T\,|\,N = n, X = x)$. In case $x = 0$, by our convention made above, $f(n,0) = 1 + n$ clearly. For $1 \leq x \leq n$, $f(n,x)$ is simply the expected waiting time till the first head, given that n tosses of a fair coin has resulted in a total of x heads. For the sake of completeness, we set $f(n,x) = 0$ (or any other value, for that matter) for $x > n \geq 0$. We now proceed to obtain a recurrence relation among the $f(n,x)$. For $1 \leq x \leq n$, we obtain, by conditioning on the outcome of the first toss,

$$f(n,x) = \alpha + \beta\,,$$

where

$$\alpha = E(T\,|\,x \text{ heads in } n \text{ tosses, first is heads}) \cdot P(\text{first heads}\,|\,N = n, X = x)\,,$$

$$\beta = E(T\,|\,x \text{ heads in } n \text{ tosses, first is tails}) \cdot P(\text{first tails}\,|\,N = n, X = x)\,.$$

A routine calulation now shows that

$$\alpha = \frac{\binom{n-1}{x-1}}{\binom{n}{x}} = \frac{x}{n} \quad \text{and}$$

$$\beta = [1 + f(n-1,x)]\,\frac{\binom{n-1}{x}}{\binom{n}{x}} = \frac{n-x}{n}\,[1 + f(n-1,x)]\,,$$

giving us the recurrence relation

$$f(n,x) = 1 + \frac{n-x}{n} f(n-1,x)\,.$$

Since $f(x,x) = 1$, we get by induction on n, that for $n \geq x$,

$$f(n,x) = \frac{n+1}{x+1}\,.$$

(Try to directly compute the conditional expectation $E(T \,|\, N = n, X = x)$.) Thus $E(T|X,N) = (N+1)/(X+1)$. To calculate the conditional expectation of this given $X = x$ we calculate the conditional distribution of N given $X = x$. Clearly, $P(N < x \,|\, X = x) = 0$ and for $n \geq x$,

$$P(N = n \,|\, X = x) = e^{-\lambda/2} (\lambda/2)^{n-x} \frac{1}{(n-x)!}$$

As a consequence for $x \geq 1$,

$$E(T \,|\, X = x) = E[\,(N+1)/(X+1) \;|\; X = x] = \frac{x + \frac{\lambda}{2} + 1}{x+1} = 1 + \frac{\lambda}{2(x+1)}$$

Even though given $X = 0$, T equals $1 + N$ and $E(N) = \lambda$, it does not mean that $E(T \,|\, X = 0) = 1 + \lambda$; indeed $E(T \,|\, X = 0) = 1 + \frac{\lambda}{2}$ (why?).

0.2.4 Independence

Definition: Random variables $X_1, X_2, \ldots, X_n$ are said to be *independent* if for any $x_1, x_2, \ldots, x_n$,

$$P(X_1 = x_1, \ldots, X_n = x_n) = P(X_1 = x_1) \cdots P(X_n = x_n)\,. \tag{10}$$

Thus independence requires that the joint p.m.f. is just the product of the marginal probability mass functions. Moreover (10) is clearly equivalent to saying that for sets $B_1, B_2, \ldots, B_n$, the events $\{(X_i \in B_i), 1 \leq i \leq n\}$ are independent. Also, independence of $X_1, X_2, \ldots, X_n$ clearly implies independence of $X_{j_1}, X_{j_2}, \ldots, X_{j_m}$ for any $1 \leq j_1 < j_2 < \cdots < j_m \leq n$. With some work, one can also show the following. Let $1 \leq i_1 < i_2 < \cdots < i_{k-1} \leq n$ and consider the random variables $Y_1, Y_2, \cdots, Y_k$ defined as $Y_1 = g_1(X_1, X_2, \ldots, X_{i_1})\,, Y_2 = g_2(X_{i_1+1}, \ldots, X_{i_2})\,, \ldots,\ Y_k = g_k(X_{i_{k-1}+1}, \ldots, X_n)$, for functions $g_1, g_2, \ldots, g_k$. Then independence of $X_1, X_2, \ldots, X_n$ implies that of $Y_1, Y_2, \ldots, Y_k$. Here are some more consequences of the definition of independence that the reader should work out.

Exercise 28. If $X_1, X_2, \ldots, X_n$ are independent, then the conditional joint distribution of any subset of them, given the others, is the same as the unconditional joint distribution.

Exercise 29. A constant random variable is independent of any random variable. Moreover, a random variable is independent of itself if and only if it is a

constant random variable.

Exercise 30. If $X_1, X_2, \ldots, X_n$ are independent random variables with finite expectations, then the product $\Pi_{i=1}^n X_i$ also has finite expectation and $E(\Pi_{i=1}^n X_i) = \prod_{i=1}^n E(X_i)$.

Exercise 31. If X and Y are independent with finite expectations, then $Cov(X,Y) = 0$. In particular, if X and Y have finite variances, then $V(X+Y) = V(X) + V(Y)$.

Exercise 32. Give an example of random variables X and Y such that $Cov(X,Y) = 0$, but X and Y are not independent.

Exercise 33. Suppose that X and Y are independent random variables and suppose that g is a function such that $Z = g(X,Y)$ has finite expectation, then $E(Z \,|\, Y = y) = E(g(X,y))$. More generally, if $X_1, X_2, \ldots, X_n$ are independent random variables and g is a function such that $Z = g(X_1, X_2, \ldots, X_n)$ has finite expectation, then

$$E(Z \,|\, X_{k+1} = x_{k+1}, \ldots, X_n = x_n) = E(g(X_1, \cdots, X_k, x_{k+1}, \ldots, x_n)) .$$

Exercise 34. In fifteen tosses of a fair coin, let X_1 be the number of heads in the first three tosses, X_2 be the number of tails in the next six tosses, and X_3 be the number of heads minus the number of tails in the last six tosses. Show that X_1, X_2 , X_3 are independent. Find $E(X_1 X_2 X_3)$.

0.3 Generating Functions

Let $(a_k)_{k\geq 0}$ be a sequence of numbers with $0 \leq a_k \leq 1$ for all k. Then clearly for any $t \in (-1,1)$ the series $\sum_{k=0}^{\infty} a_k t^k$ converges absolutely. The function $A(t) = \sum_{k=0}^{\infty} a_k t^k$ defined for $t \in (-1,1)$ is called the *generating function* of the sequence $(a_k)_{k\geq 0}$. By the uniqueness of the Taylor expansion, the function $A(t)$ determines the sequence (a_k) completely. Indeed, the function $A(t)$ is infinitely differentiable on $(-1,1)$ and $a_k = A^{(k)}(0)/k!$ where $A^{(k)}(0)$ is the k-th derivative of the function $A(t)$ at $t = 0$. Moreover as $t \uparrow 1$, $A(t)$ also increases and the limit $\lim_{t\uparrow 1} A(t)$ is finite iff $\sum a_k$ converges. In fact $\lim_{t\uparrow 1} A(t) = \sum a_k$. We denote this limit by $A(1)$. It should however be noted that in case $\sum a_k$ does not converge, then $A(t)$ increases to ∞. In this latter case also, we say that the limit $A(1) = \lim_{t\uparrow 1} A(t)$ exists and equals infinity. It is known from calculus that the derivative of the function $A(t)$ also has a power series expansion in the interval $(-1,1)$ given by $A'(t) = \sum_{k=1}^{\infty} k a_k t^{k-1}$. In fact, one can similarly get power series expansions for higher order derivatives. Once again as $t \uparrow 1$, $A'(t)$ has a finite limit iff $\sum ka_k$ converges and $\lim_{t\uparrow 1} A'(t) = \sum_{k\geq 1} ka_k$. This equality remains valid even if the right-hand side does not converge. We denote this limit by $A'(1)$. In general we will always use the notation $A^{(k)}(1)$ for the limit $\lim_{t\uparrow 1} A^{(k)}(t)$, finite or not.

By the *convolution* of two sequences $(a_k)_{k\geq 0}$ and $(b_k)_{k\geq 0}$ is meant the new sequence $c_k = (a * b)_k$ defined by $c_k = \sum_{l=0}^{k} a_l b_{k-l}$. It is easy to see that the generating function of the convolution of two sequences equals the product of the corresponding generating functions. That is, $C(t) = A(t)B(t)$.

A particularly interesting case arises when the sequence $(a_k)_{k\geq 0}$ is the probability mass function of a non-negative integer-valued random variable X. In that case, $A(t)$ is denoted by $\varphi_X(t)$ and is called the *probability generating function (p.g.f.)* or *generating function (g.f.)* of X. From our earlier discussion, it follows that the distribution of a non-negative integer valued random variable is completely determined by its p.g.f. Indeed, for such a random variable X, $P(X = k) = \varphi_X^{(k)}(0)/k!$. Clearly $\varphi_X(1) = 1$. Also

$$\varphi'_X(1) = \lim_{t\uparrow 1} \varphi'_X(t) = E(X)\,, \tag{11}$$

whether this expectation is finite or not. It is left as an exercise to show that, in case X has finite variance,

$$V(X) = \varphi''_X(1) + \varphi'_X(1) - [\varphi'_X(1)]^2 \tag{12}$$

Exercise 1. (i) If $X \sim B(n,p)$, then show that its p.g.f is $\varphi_X(t) = (1-p+pt)^n$. (ii) If $X \sim P(\lambda)$, then show that $\varphi_X(t) = e^{-\lambda(1-t)}$. (iii) If $X \sim NB(k,p)$, then show that $\varphi_X(t) = p^k(1-qs)^{-k}$.

Exercise 2. If X and Y are independent non-negative integer valued random variables, then show that $\varphi_{X+Y}(t) = \varphi_X(t)\varphi_Y(t)$.

Exercise 3. Show that (i) the sum of independent $B(n,p)$ and $B(m,p)$ random variables is $B(n+m,p)$; (ii) the sum of two independent Poisson random variables is again Poisson; (iii) the sum of independent $NB(k,p)$ and $NB(l,p)$ random variables is again NB.

Exercise 4. Let $X_1, X_2, \ldots$ and N be non-negative integer-valued random variables. Suppose that, for every $k \geq 1$, the $(k+1)$ random variables $X_1, X_2, \ldots, X_k$ and N are independent. Suppose further that the X_i have a common distribution with p.g.f. $\psi(t)$. Define $Z = \sum_{i\leq N} X_i$, with the convention that if $N = 0$, then this sum is zero. Show that the p.g.f. of Z is $\varphi_N(\psi(t))$, where φ_N is the p.g.f. of N. In particular, show that if each $X_i \sim B(1,p)$ and $N \sim P(\lambda)$, then $Z \sim P(\lambda p)$.

Exercise 5. Let $\varphi(s)$ be the p.g.f. of a random variable X. Let $q_k = P(X > k)$ for $k \geq 0$. Then the function $Q(s) = \dfrac{1-P(s)}{1-s}$ is the generating function of the sequence $(q_k)_{k\geq 0}$.

Suppose that $X_n \sim B(n,p_n)$, and denote np_n by λ_n. Then the p.g.f. of X_n is

$$\varphi_n(t) = (1-p_n+p_nt)^n = (1+p_n(t-1))^n = \left[1+\frac{\lambda_n}{n}(t-1)\right]^n .$$

If we assume that $\lambda_n \to \lambda$ then clearly

$$\varphi_n(t) \to \varphi(t) = e^{\lambda(t-1)}$$

which is the p.g.f. of $P(\lambda)$ random variable. From this it looks plausible that the distribution of X_n converges to a $P(\lambda)$ distribution. That is, for $k \geq 0$,

$$P(X_n = k) \to e^{-\lambda}\lambda^k/k!$$

This is indeed true and is actually a consequence of the next theorem.

Theorem: *For each $n \geq 1$, let φ_n be the generating function of a sequence of numbers $(a_{n,k})_{k\geq 0}$. In order that $\lim_{n\to\infty} a_{n,k} = a_k$ exists for each k, it is necessary and sufficient that $\lim_{n\to\infty} \varphi_n(s) = \varphi(s)$ exists for each $s \in (0,1)$. In that case, φ is actually the generating function of the sequence $(a_k)_{k\geq 0}$.*

Remark: It may be noted that even when the φ_n are p.g.f. of a sequence of random variables, the limit function φ need not be a p.g.f. — that is, even if $\sum_k a_{n,k} = 1$ for each n, the sequence a_k may not be a probability distribution (consider $\varphi_n(s) = s^n$). Of course $\sum a_k \leq 1$ will always hold.

Proof of Theorem: Let $\varphi_n(s) = \sum_k a_{n,k}s^k$. First assume that for each k, $a_{n,k} \to a_k$ as $n \to \infty$. Clearly $0 \leq a_k \leq 1$ for each k. Let $\varphi(s)$ be the generating function of the sequence (a_k). Fix $s \in (0,1)$ and $\epsilon > 0$ be given. Choose k_0 large enough so that $s^{k_0} < \frac{1}{4}\epsilon(1-s)$. Since $\lim_n a_{n,k} = a_k$ for each k, we choose n_0 so that for $n \geq n_0$, $|a_{n,k} - a_k| \leq \frac{\epsilon}{2k_0}$. Then

$$|\varphi_n(s) - \varphi(s)| \leq \sum_{k=1}^{k_0-1} |a_{n,k} - a_k|s^k + \sum_{k\geq k_0} |a_{n,k} - a_k|s^k \,.$$

By choice of k_0, the second term is smaller than $\epsilon/2$ and, for all $n \geq n_0$, the first term is smaller than $\epsilon/2$. Thus $|\varphi_n(s) - \varphi(s)| \leq \epsilon$ for all $n \geq n_0$, showing that $\varphi_n(s) \to \varphi(s)$ for each $s \in (0,1)$.

Conversely, suppose that $\varphi_n(s) \to \varphi(s)$ for each s with $0 < s < 1$. Clearly $0 \leq \varphi(s) \leq 1$ and $\varphi(s)$ is non-decreasing in s. In particular $\lim_{s\downarrow 0} \varphi(s) = a_0$ (say) exists. Further,

$$|a_{n,0} - a_0| \leq |a_{n,0} - \varphi_n(s)| + |\varphi_n(s) - \varphi(s)| + |\varphi(s) - a_0| \,, \tag{13}$$

and

$$|a_{n,0} - \varphi_n(s)| = \sum_1^\infty a_{n,k}s^k \leq \frac{s}{1-s} \,.$$

Therefore, given $\epsilon > 0$, we can choose s close enough to zero so that the first and third terms of the right side of (13) are each less than $\epsilon/3$. Now choose n large enough so that the second term is smaller than $\epsilon/3$. Thus we conclude that $a_{n,0} \to a_0$. Now note that

$$\frac{\varphi_n(s) - a_{n,0}}{s} \to \frac{\varphi(s) - a_0}{s} \quad \text{for } 0 < s < 1 \,.$$

It is easy to see that $g_n(s) = \dfrac{\varphi_n(s) - a_{n,0}}{s}$ is the generating function of the sequence $(a_{n,k+1})_{k\geq 0}$ so that by the same argument as above we can conclude that

$$\lim_{s\downarrow 0} \frac{\varphi(s) - a_0}{s} = a_1, \quad \text{say}$$

exists and moreover $\lim_n a_{n,1}$ exists and equals a_1. One can use induction to show that for each k, $\lim_{n\to\infty} a_{n,k} = a_k$ (say) exists.

Referring now to the *only if* part of the theorem we conclude that $\varphi_n(s)$ must converge, for each $s \in (0,1)$, to the generating function of the sequence (a_k), which therefore has to be the function $\varphi(s)$. This completes the proof of the theorem. ∎

The concept of a generating function as discussed above extends naturally to higher dimensions. We will briefly outline the definition and basic facts. Also for the sake of simplicity we confine ourselves to the case of multivariate probability generating functions.

Let $X_1, X_2, \ldots, X_d$ be random variables, defined on the same space, each taking non-negative integer values. Let their joint probability mass function be $p(k_1, k_2, \ldots, k_d)$. The *joint probability generating function (joint p.g.f.)* of $(X_1, X_2, \ldots, X_d)$ is the function φ defined on $[-1,1]^d$ defined by

$$\varphi(t_1, \ldots, t_d) = E(t_1^{X_1} \cdots t_d^{X_d}) = \sum_{k_1,\ldots,k_d} p(k_1, \ldots, k_d) t_1^{k_1} \cdots t_d^{k_d}. \tag{14}$$

It is not difficult to see that the series above converges absolutely. The function φ can also be shown to have partial derivatives of all orders and

$$p(k_1, \ldots, k_d) = \frac{1}{k_1! \cdots k_d!} \varphi^{(k_1,\ldots,k_d)}(0, \ldots, 0), \tag{15}$$

where $\varphi^{(k_1,\ldots,k_d)}$ denotes $D_1^{k_1} \cdots D_d^{k_d} \varphi$ with the usual notation that for $i = 1, \ldots, d$ and $k \geq 0$, D_i^k is the k-th order partial derivative with respect to the i-th variable. Thus for example, with $d = 3$,

$$\varphi^{(1,2,1)}(0,0,0) = \frac{\partial}{\partial t_1} \frac{\partial^2}{\partial t_2^2} \frac{\partial}{\partial t_3} \varphi(t_1, t_2, t_3)|_{(t_1=0, t_2=0, t_3=0)}.$$

Equation (15) shows that, as in the case of one dimension, the joint distribution of $(X_1, \ldots, X_d)$ is completely determined by the joint p.g.f. φ. Note that $\varphi(1, \ldots, 1) = 1$ by definition. One can also find all the moments, including cross-product moments of $(X_1, X_2, \ldots, X_d)$ from φ. For example,

$$E(X_1^2 X_2) = \varphi^{(2,1,0,\ldots,0)}(1, \ldots, 1) + \varphi^{(1,1,0,\ldots,0)}(1, \ldots, 1).$$

Also for any i, $1 \leq i \leq d$, $\varphi(t_1, \ldots, t_{i-1}, 1, t_{i+1}, \ldots, t_d)$ is precisely the joint p.g.f. of the random variables $(X_1, \ldots, X_{i-1}, X_{i+1}, \ldots, X_d)$.

In case $X_1, X_2, \ldots, X_d$ are independent with p.g.f.s $\varphi_1, \varphi_2, \ldots, \varphi_d$ respectively, then the joint p.g.f. of $(X_1, \ldots, X_d)$ is easily seen to be

$$\varphi(t_1, t_2, \ldots, t_d) = \varphi_1(t_1)\varphi_2(t_2)\cdots\varphi_d(t_d)\,. \tag{16}$$

In fact the condition (16) is also sufficient for independence. More generally, one can factor φ, the joint p.g.f of $(X_1, \ldots, X_d)$, as

$$\varphi(t_1, \ldots, t_d) = \widetilde{\varphi}(t_1, \ldots, t_i)\,\overline{\varphi}(t_{i+1}, \ldots, t_d)$$

if and only if $(X_1, \ldots, X_i)$ is independent of $(X_{i+1}, \ldots, X_d)$. Moreover, the functions $\widetilde{\varphi}$ and $\overline{\varphi}$ in the above factorization are the joint p.g.f.s of $(X_1, \ldots, X_i)$ and $(X_{i+1}, \ldots, X_d)$ respectively except possibly for some multiplicative constants. For example, the functions $3\widetilde{\varphi}$ and $\overline{\varphi}/3$ would also give a factorization.

The continuity theorem proved for one dimension has the following multivariate analogue.

Theorem: *For each $n \geq 1$, let φ_n be the joint p.g.f. of $(X_1^n, \ldots, X_d^n)$. In order that $\lim_{n\to\infty} P(X_1^n = k_1, \ldots, X_d^n = k_d)$ exists for all d-tuples $(k_1, \ldots, k_d)$ it is necesary and sufficient that for all $(t_1, \ldots, t_d) \in (0,1)^d$, the limit*

$$\lim_{n\to\infty} \varphi_n(t_1, \ldots, t_d) = \varphi(t_1, \ldots, t_d), \quad \textit{(say)}$$

exists. In this case, φ is actually the function

$$\varphi(t_1, \ldots, t_d) = \sum_{k_1,\ldots,k_d} a(k_1, \ldots, k_d) t_1^{k_1} \cdots t_d^{k_d},$$

where

$$a(k_1, \ldots, k_d) = \lim_{n\to\infty} P(X_1^n = k_1, \ldots, X_d^n = k_d)\,.$$

Barring complications arising out of d-dimensional variables, the idea of proof is no different from the one dimensional case. We omit the proof. In general the limit function $\varphi = \lim \varphi_n$ need not be a joint p.g.f.

Exercise 6. Show that the p.g.f. of the d-dimensional multinomial distribution with parameters $n, p_1, p_2, \ldots, p_d$ is $(p_1 t_1 + \cdots + p_d t_d)^n$.

Exercise 7. If for each $n \geq 1$, $(X_{n,1}, \ldots, X_{n,d})$ is multinomial with parameters $(n, p_{n1}, \ldots, p_{nd})$ and if $np_{ni} \to \lambda_i$ for $1 \leq i \leq d-1$, then show that $(X_{n,1}, \ldots, X_{n,d-1})$ has a limiting distribution as $n \to \infty$ and find the limiting distribution.

0.4 Continuous Random Variables

So far we have considered random variables with values in a finite or a countably infinite set. But in many applications it is necessary to go beyond that. For

example, consider picking a point at random from the interval $(0, 1]$. Here by picking a point at random we mean that any point "is as likely" to be picked as any other. The selected point X would then represent a random variable whose possible value can be any real number in $(0, 1]$. How do we describe the distribution of such a random variable? First of all, since any point is as likely to be picked as any other point, $P(X = x)$ should be the same for *all* x. Noting that there are infinitely many points x, one can easily argue that $P(X = x) = 0$ for all $x \in [0, 1]$. Thus, if we wanted to define the probability mass function p of the random variable X, the only candidate would be $p(x) = 0$ for all x. Certainly the distribution of the random variable X cannot be captured by such a function.

So, instead of prescribing probabilities of events through probabilities of individual outcomes that constitute an event, one may hope to prescribe probabilities of all events at one go. In other words, one may think of directly specifying $P(X \in A)$ for various subsets $A \subset [0, 1]$. But clearly, that is a tall task! However, there is a general theory — known as *measure theory* — which says that it is sufficient to specify $P(X \in A)$ only for intervals $A \subset [0, 1]$ which, in turn, uniquely determine $P(X \in A)$ for a large class of sets A, known as *measurable sets.* One may still wonder what if we want $P(X \in A)$ for a non-measurable set A. However, there is no real need to worry! The class of measurable sets is really huge — almost any set A one is likely to come across for the purpose of computing $P(X \in A)$ is going to be a measurable set. Having said all these let us add that mere recognition and acceptance of this fact will do for the rest of this book. We do not make any explicit use of measure theory.

Continuing with our example and again noting that the point is selected at random, one can easily deduce for any $0 \leq a < b \leq 1$, we must have $P(a < X < b) = b - a$. In fact, the above is just a consequence of the fact that $P(X \in I)$ equals $P(X \in J)$ whenever I and J are intervals of same length.

Of course, for any random variable X, prescribing the probabilities $P(X \in A)$ for intervals A and hence prescribing the distribution of X could also be done by simply specifying the function

$$F(x) = P(X \leq x) \tag{17}$$

for all $x \in R$. This function F is called the *probability distribution function* of the random variable X and has the following properties:

(i) $0 \leq F(x) \leq 1$ for all x and $F(x) \leq F(y)$ whenever $x \leq y$,
(ii) $\lim_{x \to -\infty} F(x) = 0$, $\lim_{x \to +\infty} F(x) = 1$, and
(iii) F is right-continuous, that is, $\lim_{y \downarrow x} F(y) = F(x)$.

It may be noted that $\lim_{y \uparrow x} F(y) = P(X < x)$, so that $P(X = x) = F(x) - \lim_{y \uparrow x} F(y)$. From all these it should be clear that $F(x)$ determines $P(X \in A)$ for every interval A (and hence for all measurable sets A).

In the example of picking a point at random, the corresponding distribution function is

$$F(x) = \begin{cases} 0 & \text{if } x \leq 0 \\ x & \text{if } 0 \leq x \leq 1 \\ 1 & \text{if } x \geq 1 \end{cases} .$$

A *continuous random variable* is one whose distribution function is continuous. From the properties of F listed above, it follows that a random variable X is continuous if and only if $P(X = x) = 0$ for all $x \in R$. It is in this sense that continuous random variables are diametrically opposite to discrete random variables.

0.4.1 Probability Density Function

One special class of continuous random variables are those for which the distribution function is given by

$$F(x) = \int_{-\infty}^{x} f(y)\, dy \tag{18}$$

where f is a non-negative funcion with $\int_{-\infty}^{\infty} f(y)\, dy = 1$. Such a function f is called a *probability density function* (p.d.f., in short). Probabilities involving X can be calculated from its density function by the formula $P(X \in A) = \int_A f(y)\, dy$. Such probability distributions are called *absolutely continuous* distributions and the corresponding random variable is also called absolutely continuous. It may be noted that probability density function of a distribution (or, of a random variable) is not unique. (Changing the value of f at a finite number of points would not change the integrals appearing in (18) and therefore, would give the same F!)

Unlike probability mass function, the probability density function does not represent any probability. However, it has the approximate interpretation

$$P(X \in (x, x + \delta x)) \sim f(x)\delta x\,.$$

This should explain why f is called the density function as opposed to mass function of the discrete case. For a random variable X with density function f the expected value is defined by the formula

$$E(X) = \int_{-\infty}^{\infty} x\, f(x)\, dx\,, \tag{19}$$

provided the integral exists. We allow the integral to equal $+\infty$ or $-\infty$. But there is a caveat! Two infinities cannot be added unless they have the same sign. We define, more generally,

$$E(g(X)) = \int_{-\infty}^{\infty} g(x)\, f(x)\, dx\,, \tag{20}$$

provided the integral exists. The expected value so defined can be shown to satisfy all the properties that were proved to be true for discrete random variables. As in the discrete case, the m-th moment of X is defined to be $E(X^m)$ and the variance is defined as $V(X) = E(X^2) - (EX)^2$.

Exercise 1. Fix numbers $a < b$. Let f be the function which is $1/(b-a)$ for points in the interval (a, b) and zero for points outside the interval. Show that this is a probability density function. Calculate the corresponding distribution function. This is called the *Uniform distribution* on (a, b), denoted $\mathcal{U}(a, b)$ and a random variable with this distribution is called a $\mathcal{U}(a, b)$ random variable. Find the expected value and variance of such a random variable.

Exercise 2. Fix any number $\lambda > 0$. Consider the function f which is zero for negative numbers and is $\lambda \exp(-\lambda x)$ for non-negative numbers x. Show that this is a probability density function. Calculate the corresponding distribution function. This is called the *Exponential distribution with parameter* λ, written $\mathcal{E}xp(\lambda)$. For a $\mathcal{E}xp(\lambda)$ random variable X, find (i) $P(X > 10.25)$, (ii) $P((X - 3)^2 > 1)$. Also find $E(X)$, $V(X)$ and $E(e^{tX})$ for $t \in R$.

Exercise 3. Fix any real number μ and any strictly positive number σ. Let

$$f(x) = \frac{1}{\sigma\sqrt{2\pi}} e^{-\frac{1}{2\sigma^2}(x-\mu)^2}$$

for $-\infty < x < +\infty$. This is a probability density function (not easy to show this fact). Corresponding distribution is called the *Normal distribution* with parameters μ and σ^2, written $\mathcal{N}(\mu, \sigma^2)$. The distribution function cannot be calculated explicitly. Show that a $\mathcal{N}(\mu, \sigma^2)$ random variable X has mean μ and variance σ^2. Also show that $E(e^{tX}) = \exp[\mu t + \frac{1}{2}\sigma^2 t^2]$ for $t \in R$.

In case $\mu = 0$, $\sigma = 1$ in Exercise 3 above, the distribution is called *Standard Normal Distribution.* In this case, the distribution function is usually denoted by $\Phi(x)$ and the density function is denoted by $\phi(x)$.

Exercise 4. Repeat Exercises 4–14 of Section 0.2.1, assuming that all the random variables are absolutely continuous.

0.4.2 Joint Density Function

For two continuous random variables X and Y defined on the same space, we may be interested in probabilities of events that concern both X and Y. For computing such probabilities, knowing the individual density functions of X and Y alone would not suffice. We need to know what is called the *joint density function* of X and Y.

Definition: Let X and Y be two random variables defined on the same space. The pair (X, Y) is said to have a *joint density function* $f(x, y)$ if f is a non-negative function such that for any $x, y \in R$,

$$P(X \leq x, Y \leq y) = \int_{-\infty}^{y} \int_{-\infty}^{x} f(u, v)\, du dv.$$

Clearly such an f satisfies $\int_{-\infty}^{\infty}\int_{-\infty}^{\infty} f(u,v)\,du\,dv = 1$. Probabilities involving the pair (X,Y) can be computed from the formula $P((X,Y)\in A) = \int\int_A f(x,y)dx\,dy$.

From the joint density of (X,Y) the individual (marginal) densities of X and Y can be recovered as follows:

$$f_1(x) = \int f(x,y)dy, \quad f_2(y) = \int f(x,y)dx.$$

In an analogous way the joint density of n random variables (defined on the same space) is defined to be a non-negative function f of n variables such that

$$P(X_1 \le x_1, \ldots, X_n \le x_n) = \int_{-\infty}^{x_n} \cdots \int_{-\infty}^{x_1} f(u_1, \ldots, u_n)\,du_1 \cdots du_n.$$

Here also the individual density of each X_i can be obtained from the joint density f by a formula analogous to the bivariate case. An important point to note in this connection is that the existence of a joint density for $(X_1, \ldots, X_n)$ implies that each X_i has a density; however the converse is not true. For example, if X has $\mathcal{U}(0,1)$ distribution and $Y = X$, then both X and Y have densities, but the pair (X,Y) does not have a joint density (why?).

For $(X_1, \ldots, X_n)$ with joint density f, the expected value of any function of $(X_1, \ldots, X_n)$ can be computed by the formula,

$$E(g(X_1, \ldots, X_n)) = \int \cdots \int g(x_1, \ldots, x_n) f(x_1, \ldots, x_n) dx_1 \cdots dx_n,$$

provided, of course, that the integral exists.

For a pair (X,Y) with joint density f, the covariance between X and Y is defined by the same formula (21) of Section 0.2.2 and all the properties listed in Exercises 16–21 there remain valid.

0.4.3 Conditional Density

For a pair (X,Y) with joint density f the *conditional density* of X given $Y = y$ is defined to be

$$f_1(x \,|\, y) = \frac{f(x,y)}{f_2(y)} \quad \text{if} \quad f_2(y) > 0.$$

For y with $f_2(y) = 0$, one may define $f_1(x \,|\, y)$ to equal any density function, for example, one may put $f_1(x \,|\, y) = f_1(x)$. Here f_1 and f_2 are the marginal densities as defined in the previous section. One can easily check that $f_1(x \,|\, y)$ is a density (in x) for every y. The distribution given by this density is called the *conditional distribution* of X given $Y = y$. One can similarly define the conditional distribution of Y given $X = x$. It is also not difficult to extend this concept to the case of n random variables with a joint density.

For a random variable X with density f and for any event A with $P(A) > 0$, one can define the conditional density of X given A. However, there is no explicit formula for this density in general. One is only guaranteed of the existence of this density by a result known as *Radon-Nikodym Theorem* which is beyond the scope of this book. However, in the special case when $A = \{X \in B\}$ the conditional density of X given A is given by $f(x|A) = f(x)/P(A)$ if $x \in B$ and equals zero otherwise.

As in Section 0.2.3, expectation and variance of the conditional distribution are known as *conditional expectation* and *conditional variance* respectively. As before, it is sometimes convenient to think of conditional expectation itself as a random variable, denoted by $E(X|Y)$, which has the same interpretation as in Section 0.2.3. Also, all the properties in Exercises 4,5,7,8, 10–14 and 22–26 go through.

0.4.4 Independence

Definition: Random variables $(X_1, \ldots, X_n)$ with a joint density f are said to be *independent* if for any $x_1, x_2, \ldots, x_n$,

$$f(x_1, \ldots, x_n) = f_1(x_1) \cdots f_n(x_n) .$$

Thus the random variables are independent if the joint density factors into product of the marginal probability densities $f_1, \ldots, f_n$. This can be shown to be equivalent to the condition that for sets $B_1, B_2, \ldots, B_n$, the n events $(X_i \in B_i)$, $1 \leq i \leq n$ are independent. Also independence of $X_1, X_2, \ldots, X_n$ clearly implies independence of $X_{j_1}, X_{j_2}, \ldots, X_{j_m}$ for $1 \leq j_1 < j_2 < \cdots < j_m \leq n$. With some work one can also show the following. Consider k indices $1 \leq i_1 < i_2 < \cdots < i_{k-1} \leq n$ and let $Y_1, Y_2, \ldots, Y_k$ be random variables defined as follows: $Y_1 = g_1(X_1, \ldots, X_{i_1}), Y_2 = g_2(X_{i_1+1}, \ldots, X_{i_2}), \cdots$, $Y_k = g_k(X_{i_{k-1}+1}, \ldots, X_n)$, for functions $g_1, g_2, \ldots, g_k$. Then independence of $X_1, X_2, \ldots, X_n$ implies that of $Y_1, Y_2, \ldots, Y_k$.

It is left as an exercise to verify that properties 28–33 of Section 0.2.4 remain valid here also.

0.5 Sequences of Random Variables

Let Y be a random variable with finite mean μ. Let $Y_1, Y_2, \ldots$ be independent observations on the variable Y, that is, for each n, the random variables $Y_1, \ldots, Y_n$ are independent each having the same distribution as Y. One says that $Y_1, Y_2, \ldots$ is a sequence of *independent and identically distributed*, abbreviated as i.i.d., random variables. Let X_n denote the average of the first n observations, that is, $X_n = (Y_1 + \cdots + Y_n)/n$. This X_n is also called the observed mean or the sample mean, based on n observations. An important question is : what happens to these observed means as n, the sample size, becomes large? A classical result in probability (known as the *law of large numbers*) is that

the observed means converge to the common population mean μ. It should be noted that the observed means X_n are random variables. Thus, one has to know what is meant by the convergence of a sequence of random variables. In this section, we discuss some of the various concepts of convergence that are used in probability.

In what follows, $(X_n)_{n\geq 1}$ will stand for a sequence of random variables defined on the same space.

Definition: We say that X_n *converges in probability* to a random variable X, and write $X_n \xrightarrow{P} X$, if for each $\epsilon > 0$, $P(|X_n - X| > \epsilon) \to 0$ as $n \to \infty$.

That is, given any $\epsilon > 0$ however small, the chances that X_n deviates from X by more than ϵ become smaller and smaller as n gets larger and larger. However this should not be misinterpreted as X_n remaining close to X eventually for almost all sample points. One can construct an example where $X_n \xrightarrow{P} X$ but $X_n(\omega) \not\longrightarrow X(\omega)$ for any sample point ω! [see Exercise 5 below]. This motivates the next definition.

Definition: We say that X_n *converges with probability one* to X, and write $X_n \longrightarrow X$ w.p.1, if for all sample points ω, outside a set of zero probability, $X_n(\omega) \to X(\omega)$ as $n \to \infty$.

Another mode of convergence that will be useful for us, is the following.

Definition: Let $p \geq 1$ be a number. We say that X_n *converges in p-th mean* to X, written $X_n \xrightarrow{L_p} X$, if $E|X_n - X|^p \to 0$ as $n \to \infty$. This mode of convergence is also referred to as convergence in L_p.

We shall now see the relationships between these three modes of convergence. The last two modes of convergence are stronger than the first one. Indeed if $X_n \xrightarrow{p} X$ then for any $\epsilon > 0$,

$$P(|X_n - X| > \epsilon) = P(|X_n - X|^p > \epsilon^p) = E\left(I_{\{|X_n - X|^p > \epsilon^p\}}\right)$$

$$\leq E\left(\frac{|X_n - X|^p}{\epsilon^p} I_{\{|X_n - X|^p > \epsilon^p\}}\right) \leq \frac{E|X_n - X|^p}{\epsilon^p} \longrightarrow 0,$$

by hypothesis. Note that here ϵ is fixed and n becomes large.

Hidden in the above argument is the fact that for any random variable Z and any $\epsilon > 0$

$$P(|Z| \geq \epsilon) \leq \frac{E|Z|}{\epsilon}, \tag{21}$$

a useful inequality, known as *Markov's inequality*. This inequality can be easily proved using Exercise 5(a) of Section 0.2.1.

Next suppose that $X_n \longrightarrow X$ w.p.1; that is, there is a set A of probability zero such that for $\omega \notin A$, $X_n(\omega) \to X(\omega)$. Let $\epsilon > 0$. Then for any n,

$$\{|X_n(\omega) - X(\omega)| > \epsilon\} \quad \subset \quad \bigcup_{k>n} (|X_k(\omega) - X(\omega)| > \epsilon)$$

and the set on the right side decreases as n increases and the limiting set is contained in A (because for any ω in the limiting set $X_n(\omega) \not\longrightarrow X(\omega)$). Since $P(A) = 0$ it follows that

$$\lim_{n\to\infty} P(|X_n(\omega) - X(\omega)| > \epsilon) = 0\,.$$

Convergence with probability 1 and convergence in L_p are, in general, not comparable. However, here is a useful result.

$$\text{If } X_n \longrightarrow X \text{ w.p.1} \quad \text{and} \quad E(\sup_n |X_n|^p) < \infty, \qquad \text{then } X_n \xrightarrow{L_p} X\,.$$

Indeed one can replace $X_n \longrightarrow X$ w.p.1 by the weaker hypothesis $X_n \xrightarrow{P} X$. So we will assume only this. Denote the random variable $\sup_n |X_n|^p$ by Z. It is not difficult to see that $X_n \xrightarrow{P} X$ yields that $P(|X|^p \leq Z) = 1$. [Show that for any $\epsilon > 0$, $P(|X|^p > Z + \epsilon) = 0$]. Thus $|X_n - X|^p \leq 2^p Z$ Note that the hypothesis says that Z has finite expectation. Therefore given $\delta > 0$, we can choose $\lambda > 0$ so that $E(ZI_{(Z>\lambda)}) < 2^{-p}\delta/3$. We can also choose $\epsilon > 0$ so that $\epsilon^p < \delta/3$. Now choose n_0 such that for $n \geq n_0$ we have, $P(|X_n - X| \geq \epsilon) \leq \delta 2^{-p}/3\lambda$. Now for $n \geq n_0$,

$$\begin{aligned} E|X_n - X|^p &\leq E(|X_n - X|^p I_{|X_n - X| \leq \epsilon}) + E(|X_n - X|^p I_{|X_n - X| > \epsilon}) \\ &\leq \epsilon^p + 2^p E(Z I_{|X_n - X| > \epsilon}) \\ &\leq \epsilon^p + 2^p E(Z I_{Z \leq \lambda} I_{|X_n - X| > \epsilon}) + 2^p E(Z I_{Z > \lambda}) \\ &\leq \epsilon^p + 2^p \lambda P(|X_n - X| > \epsilon) + 2^p E(Z I_{Z > \lambda})\,. \end{aligned}$$

Each term on the right side is at most $\delta/3$, completing the proof.

The reader must have already realized that Lebesgue's Dominated Convergence Theorem as given in Exercise 8, Section 0.2.1, is just a special case of the above.

Exercise 1. If $X_n \xrightarrow{P} X$, then show that $X_n^{19} \xrightarrow{P} X^{19}$. More generally, if f is a continuous function, then $f(X_n) \xrightarrow{P} f(X)$. What if, convergence in probability is replaced by convergence in L_p or by convergence with probability one?

Exercise 2. If $X_n \xrightarrow{L_p} X$ then $X_n \xrightarrow{L_r} X$, for $1 \leq r \leq p$.

Exercise 3. If $X_n \xrightarrow{P} X$ and $Y_n \xrightarrow{P} Y$, then show that $X_n + Y_n \xrightarrow{P} X + Y$ and $X_n Y_n \xrightarrow{P} XY$. What if $\xrightarrow{P}$ is replaced by $\xrightarrow{L_p}$?

Exercise 4. Let $X_n \xrightarrow{P} X$. Show that there is a subsequence (n_k) such that $X_{n_k} \longrightarrow X$ w.p.1. (Choose n_k so that $P(|X_{n_k} - X| > 2^{-k}) < 2^{-k}$.)

Exercise 5. Consider a $\mathcal{U}(0,1)$ variable X. Consider the following sequence of random variables: $Z_1 = I_{(X<1/2)}$; $Z_2 = I_{(X>1/2)}$; $Z_3 = I_{(X<1/4)}$; $Z_4 = I_{(1/4<X<1/2)}$; $Z_5 = I_{(1/2<X<3/4)}$; $Z_6 = I_{(3/4<X<1)}$; etc. It should be clear(?)

how the subsequent Z_n are defined. Show that Z_n does not converge with probability one, but converges in probability to zero.

In conclusion, let us go back to the convergence of observed means to the population mean. Classical Laws of Large Numbers say that convergence here takes place with probability one. In other words, if $Y_1, Y_2, \ldots$ are i.i.d with common finite mean μ, then $X_n = (Y_1 + \cdots + Y_n)/n \longrightarrow \mu$ w.p.1. In fact, this result remains valid even without the assumption of finiteness of the mean, as long as the Y_i are non-negative. The proof of this result is quite involved for presenting here. Instead, we show that convergence in probability holds. For this, let us further assume that $V(Y) < \infty$. In this case, by Markov inequality

$$P(|X_n - \mu| > \epsilon) = P(|X_n - \mu|^2 > \epsilon^2) \leq \frac{1}{\epsilon^2} E|X_n - \mu|^2 = \frac{1}{\epsilon^2} \frac{V(Y)}{n} \to 0\,.$$

In the above, we have used the fact that $V(Y_1 + \cdots + Y_n) = nV(Y)$ because of the i.i.d. hypothesis. It is possible to do away with the finite variance assumption, but the argument becomes a little more complicated. As a special case of the above, if X_n is $B(n,p)$ then $X_n/n \xrightarrow{P} p$.

Exercise 6. If $X_n \sim B(n,p)$ show that $\sum E(X_n - np)^4/n^4 < \infty$ and hence conclude, using Borel-Cantelli lemma that $X_n/n \longrightarrow p$ w.p.1.

Another important mode of convergence is *convergence in distribution.* Since we do not need it for our applications, we do not discuss it. However in Section 0.3, we had an illustration of this kind of convergence. To be specific, what was shown there is that if $X_n \sim B(n,p_n)$ where $np_n \to \lambda$ as $n \to \infty$, then X_n converges 'in distribution' to a $P(\lambda)$ random variable. A classical result in probability, involving the notion of convergence in distribution is what is known as *Central Limit Theorem.* Here is what it says. If $Y_1, Y_2, \ldots$ are i.i.d. random variables with mean μ and finite variance σ^2, then $X_n = (Y_1 + Y_2 + \cdots + Y_n)/\sqrt{n}$ converges in distribution to a $\mathcal{N}(\mu, \sigma^2)$ random variable, that is, for any real number a, $P(X_n \leq a) \longrightarrow \int_{-\infty}^{a} f(u)du$, where f is the density function given in Exercise 3 of Section 0.4.1.

0.6 Characterstic Functions

Definition: For any random variable X, the function $\varphi_X(t) = E(e^{itX})$ defined for $-\infty < t < +\infty$ is called the *characterstic function* of X.

To make sense of this definition, one needs to extend the notion of expectation to a complex-valued random variable. If $Z = U + iV$ where U and V are real random variables with finite expectations, one defines $E(Z)$ to be the complex number $E(U) + iE(V)$. With this definition, it is easy to see that the property

$$E(\alpha_1 Z_1 + \alpha_2 Z_2) = \alpha_1 E(Z_1) + \alpha_2 E(Z_2)$$

holds, where α_1, α_2 are complex numbers and Z_1, Z_2 are complex random variables. The property $|E(Z)| \leq E(|Z|)$ also holds where, as usual, for a complex number z, $|z| = \sqrt{(Re\, z)^2 + (Im\, z)^2}$. Here is a quick proof of the above inequality. It is easy to see that $E(Z) = \alpha |E(Z)|$ for some complex number α with $|\alpha| = 1$. Thus

$$|E(Z)| = \overline{\alpha} E(Z) = E(\overline{\alpha} Z) = E(Re\,(\overline{\alpha} Z)) \leq E(|\overline{\alpha} Z|) = E(|Z|)$$

One can use this to show that the Lebesgue's Dominated Convergence Theorem (see Section 0.2) holds for complex random variables as well.

Returning to characterstic functions it may be noted that φ_X is a complex-valued function of a real variable t, given by the formula

$$\varphi_X(t) = E(\cos(tX)) + i\, E(\sin(tX))\,. \tag{22}$$

Clearly, the real random variables $\cos(tX)$ and $\sin(tX)$ are bounded and hence have finite expectations for all t. From (22) it follows that

(1) $\varphi_X(0) = 1$ and $\varphi_{aX+b}(t) = e^{itb} \varphi_X(at)$.

(2) $\varphi_X(-t) = \overline{\varphi_X(t)} = \varphi_{-X}(t)$. In particular, $\varphi_X(t)$ is a real-valued function if X has a symmetric distribution, that is, X and $-X$ have the same distribution.

Using $|E(e^{itX})| \leq E(|e^{itX}|)$ one also gets

(3) $|\varphi_X(t)| \leq 1$ for all t.

For any real t and h,

$$|\varphi_X(t+h) - \varphi_X(t)| \leq E(|e^{itX}(e^{ihX} - 1)|) = E(|e^{ihX} - 1|)$$

and by the Dominated Convergence Theorem the last expression goes to zero as $h \to 0$. Thus we have proved

(4) $\varphi_X(t)$ is a continuous function — in fact, it is uniformly continuous.

One of the important features of the characterstic function of a random variable X is that the distribution of X is completely determined by its characterstic function φ_X. In other words, two random variables with different distributions cannot have the same characterstic function. We give below the formula, known as the *Inversion formula*, that determines the distribution function F of a random variable X from its characterstic function φ_X.

(5) For any two continuity points $a < b$ of F,

$$F(b) - F(a) = \lim_{T \to \infty} \frac{1}{2\pi} \int_{-T}^{T} \frac{e^{-ita} - e^{-itb}}{it} \varphi_X(t)\, dt$$

(6) Moreover, if $\int |\varphi_X(t)|dt < \infty$, then the random variable X has a bounded continuous density function given by

$$f(x) = \frac{1}{2\pi} \int_{-\infty}^{\infty} e^{-itx} \varphi_X(t) dt$$

The proof of (5) is somewhat involved and hence omitted here. Interested reader may consult Chung [2005]. Here is a sketch of a proof of (6). Using an analogue of the Dominated Convergence Theorem valid for general integrals, one can show that if $|\varphi_X|$ has finite integral then the inversion formula can be written as

$$\begin{aligned} F(b) - F(a) &= \frac{1}{2\pi} \int_{-\infty}^{\infty} \frac{e^{-ita} - e^{-itb}}{it} \varphi_X(t) dt \\ &= \frac{1}{2\pi} \int_{-\infty}^{\infty} \left(\int_a^b e^{-itx} dx \right) \varphi_X(t) dt. \end{aligned}$$

Interchanging the order of integration now (which can again be justified in view of $\int |\varphi_X(t)|dt < \infty$), one gets

$$F(b) - F(a) = \int_a^b f(x)dx \quad \text{where} \quad f(x) = \frac{1}{2\pi} \int_{-\infty}^{\infty} e^{-itx} \varphi_X(t) dt.$$

Thus X has density $f(x)$ which is bounded because

$$|f(x)| \leq \frac{1}{2\pi} \int |\varphi_X(t)|dt < \infty.$$

Continuity of f follows from the Dominated Convergence Theorem alluded to above.

One consequence of the one-one correspondence between characterstic functions and distributions is that the converse of (2) holds. In other words

(7) $\varphi_X(t)$ is real-valued function if and only if X has a symmetric distribution, that is, X and $-X$ have the same distribution.

(8) Of course, for independent random variables X and Y, we have $\varphi_{X+Y} = \varphi_X \cdot \varphi_Y$.

It is easy to see that if $X \sim B(n,p)$ then $\varphi_X(t) = (q + pe^{it})^n$. Now if X and Y are independent random variables and $X \sim B(n,p)$ and $Y \sim B(m,p)$ then the characterstic function of $X + Y$ turns out to be

$$\varphi_{X+Y}(t) = \varphi_X(t) \cdot \varphi_Y(t) = (q + pe^{it})^{m+n}$$

from which we can immediately conclude that $X+Y$ must have $B(m+n,p)$ distribution.

Similarly one can show that if $X \sim \mathcal{N}(0,1)$, then $\varphi_X(t) = E(\cos tX) = e^{-t^2/2}$. From this one can deduce that if $X \sim \mathcal{N}(\mu,\sigma^2)$, then $\varphi_X(t) = e^{it\mu - \frac{1}{2}t^2\sigma^2}$. Therefore, if $X \sim \mathcal{N}(\mu;\sigma^2)$ and $Y \sim \mathcal{N}(\nu,\tau^2)$ are independent then by computing φ_{X+Y}, one can conclude that $X+Y$ is $\mathcal{N}(\mu+\nu,\sigma^2+\tau^2)$.

Characterstic functions can also be used to compute moments of the distribution, when they exist. Here is how the method works. Using the power series expansion $e^{itX} = \sum_{n=0}^{\infty} \frac{(itX)^n}{n!}$ and taking expectations, one gets $\varphi_X(t) = E(\sum \frac{(itX)^n}{n!})$. Assume for the time being that the expectation and the infinite sum could be interchanged. That would give

$$\varphi_X(t) = \sum_{n=0}^{\infty} i^n E(X^n) \frac{t^n}{n!},$$

that is, $\varphi_X(t)$ has a power series expansion in t in which, the coefficient of t^n is $\frac{i^n}{n!}E(X^n)$. From the general theory of power series it would follow that $E(X^n) = \varphi_X^{(n)}(0)/i^n$. It is possible to justify the above formal calculations (using simply Dominated Convergence Theorem and appropriate Mean Value Theorem) and here is the precise result.

(9) If X has finite n-th moment then φ_X has derivatives of orders upto and including n everywhere and, for every $k \leq n$, $E(X^k) = \varphi_X^{(k)}(0)/i^k$.

A very important use of characterstic functions consists of proving convergence in distribution. This is achieved through, what is known as, *Lèvy's Continuity Theorem.* The theorem asserts the equivalence of convergence in distribution and pointwise convergence of characterstic functions. Since we have not formally defined the notion of convergence in distribution, we would not go into the details of this result. The interested reader may consult Chung [2005].

Exercise 1. Calculate the characterstic functions of the following random variables: $P(\lambda)$, $\mathcal{U}[0,1]$, $\mathcal{E}xp(\lambda)$ and $\mathcal{N}(\mu,\sigma^2)$.

Exercise 2. If X has the *double exponential* density

$$f(x) = \frac{1}{2}\, e^{-|x|}, \qquad -\infty < x < \infty,$$

find the characterstic function of X. Use this and property (6) to find the characterstic function of *Cauchy* distribution given by the density

$$f(x) = \frac{1}{\pi}\, \frac{1}{1+x^2}, \qquad -\infty < x < \infty.$$

Exercise 3. Show that $\varphi_X(t_0) = 1$ for some $t_0 > 0$ if and only if X is discrete

with values in the set $\{2\pi n/t_0 : n = 0, \pm 1, \pm 2, \ldots\}$. More generally, show that $|\varphi_X(t_0)| = 1$ for some $t_0 > 0$ if and only if X is discrete with values in the set $\{(2\pi n + \theta)/t_0 : n = 0, \pm 1, \pm 2, \ldots\}$ for some real number θ.

Exercise 4. If φ is a characterstic function, show that both $|\varphi|^2$ and $Re\ \varphi$ are characterstic functions.

Exercise 5. Let $(X_n)_{n\geq 1}$ be a sequence of i.i.d. random variables with common characterstic function φ. Let N be a $P(\lambda)$ random variable independent of the sequence (X_n). Find the characterstic function of $Y = X_1 + \cdots + X_N$. If $N = 0$, we define Y to be zero.

0.7 Martingales

In this section we discuss a special class of sequences of random variables known as *martingales*. Martingales constitute a very important and widely useful class of processes. We do not intend to present here an extensive coverage of this topic. Instead we only list a few basic properties of martingales which will be needed for our purposes. A reader interested to learn more can see the book of Leo Breiman.

Consider a sequence of independent tosses of a fair coin. Before each toss you are allowed a bet. If the toss results in heads then you win the amount you wagered; otherwise you loose the same amount. Note that you are allowed to change your wagers at each toss and moreover, your decision is allowed to be based on the outcomes of previous tosses. This can be mathematically formalized by means of a sequence $\epsilon_1, \epsilon_2, \ldots$ of random variables where ϵ_n denotes your wager amount for the n-th toss. If we denote the outcomes of the tosses themselves by a sequence $\eta_1, \eta_2, \ldots$, where each η_i can be $+1$ or -1, then the actual amount you win at the n-th toss is $\eta_n \epsilon_n$. Clearly η_n are i.i.d. random variables. The condition on the ϵ_n is that ϵ_1 is a constant and, for $n \geq 2$, ϵ_n is a random variable that is allowed to depend *only* on $\eta_1, \eta_2, \ldots, \eta_{n-1}$. As an additional technical condition, we shall also assume that each ϵ_n has finite expected value. One of the interesting features of the game is that if the coin is fair, then the game is also fair in the following sense. Denote by X_n, your accumulated fortune upto and including the n-th toss, that is, $X_n = \sum_{i=1}^{n} \epsilon_i \eta_i$. Define $X_0 = 0$. One can easily deduce that, at any stage if you want to find the conditional expectation of your accumulated fortune after the next toss, given all the information upto and including the present time, it equals your present accumulated fortune. That is to say that if you play one more game, it would, on the average, make you neither better off nor worse off. The word 'on an average' is important here, because in the actual play you would really either win or loose. The point is that you cannot be certain of either and the mean change in fortune, based on available information, is zero. This is the mathematical formulation of fairness in the game. This leads to the following formal definition.

All the random variables that we consider below are defined on the same space. Also they are all assumed to be discrete and to have finite expectations. Although the condition that they are discrete is not necessary in general, however it allows us to avoid some technicalities.

Definition: A sequence $(X_n)_{n\geq 0}$ of random variables is said to be a *martingale* if, for every n,

$$E(X_n \mid X_0, X_1, \ldots, X_{n-1}) = X_{n-1}. \tag{23}$$

In particular $E(X_n)$ is same for all n.

We will see plenty of examples of martingales in our applications in the subsequent chapters. However, here are some simple examples.

Example 1: Let $(\eta_i)_{i\geq 1}$ be a sequence of independent random variables with zero means. Set, $X_0 = 0$, and for $n \geq 1$, $X_n = \sum_{i=1}^{n} \eta_i$. Then $(X_n)_{n\geq 0}$ is easily seen to be a martingale. We could easily replace each X_n by $X_n + Z$ where Z is a random variable with finite mean, independent of $(\eta_i)_{i\geq 1}$, and still have a martingale.

Example 2: Let $(\eta_i)_{i\geq 1}$ and Z be as above. Let $(\epsilon_i)_{i\geq 1}$ be a sequence of bounded random variables with ϵ_n depending only on $\{Z, \eta_1, \ldots, \eta_{n-1}\}$ for each n. Set $X_n = Z + \sum_{i=1}^{n} \epsilon_i \eta_i$, $n \geq 0$. Then $(X_n)_{n\geq 0}$ is a martingale. The condition that ϵ_i are bounded is just to ensure that $\epsilon_i \eta_i$ has finite expectation and can be relaxed by the latter. The example given at the beginning of this section with the η_i representing the outcomes of succesive tosses of a coin is just a special case.

Example 3: Let $(\eta_i)_{i\geq 1}$ be as in Example 1, with the additional assumption that $V(\eta_i) = \sigma_i^2 < \infty$. Then $X_0 = 0$ and $X_n = (\sum_1^n \eta_i)^2 - \sum_1^n \sigma_i^2$, $n \geq 1$, defines a martingale. In particular, if each η_i takes the values ± 1 with probability $1/2$ each, then $(\sum_1^n \eta_i)^2 - n$ is a martingale.

Example 4: Here is the famous Polyà Urn Scheme. Start with an urn containing b black balls and r red balls. A ball is drawn at random, its colour noted and then the ball is replaced along with an additional ball of the same colour. This process is repeated. Note that, at each stage the total number of balls in the urn increases by one so that after n turns, the urn will have $b + r + n$ balls. Denoting X_n to be the proportion of red balls in the urn after n turns, with, of course, $X_0 = r/(b + r)$, it is not difficult to check that we get a martingale.

Example 5: Let $(\eta_i)_{i\geq 1}$ be an i.i.d sequence, taking the values ± 1 with probabilities $1/2$ each. Denote $S_n = \sum_{1\leq i\leq n} \eta_i$. Then for any $\theta \in (0, 1)$, the sequence $(X_n)_{n\geq 0}$ defined as $X_0 \equiv 1$ and for $n \geq 1$, $X_n = 2^n \theta^{(n+S_n)/2} (1-\theta)^{(n-S_n)/2}$, defines a martingale. Indeed this is a special case of the next Example.

Example 6: Let $(\eta_i)_{i\geq 1}$ be a sequence of discrete random variables and for each

$n \geq 1$, let $p_n(u_1, \ldots, u_n)$ be the (true) joint p.m.f. of $(\eta_1, \ldots, \eta_n)$. Suppose that $\widetilde{p}_n(u_1, \ldots, u_n)$, $n \geq 1$, be a sequence of joint p.m.f.s satisfying

(i) $\sum_{u_{n+1}} \widetilde{p}_{n+1}(u_1, \ldots, u_n, u_{n+1}) = \widetilde{p}_n(u_1, \ldots, u_n)$ and

(ii) $\widetilde{p}_n(u_1, \ldots, u_n) = 0$ whenever $p_n(u_1, \ldots, u_n) = 0$.

Then $X_0 = 1$ and for $n \geq 1$, $X_n = \widetilde{p}_n(\eta_1, \ldots, \eta_n)/p_n(\eta_1, \ldots, \eta_n)$ can be seen to define a martingale (taking the ratio 0/0 to be 0). Complicated though this example looks, here is the context in which it arises. The $\widetilde{p}_n$ can be thought of as the joint p.m.f.s under some proposed alternative distribution of the sequence $(\eta_i)_{i \geq 1}$. A statistician wants to test the validity of this alternative. Standard tools of statistics often use the X_n (known as *likelihood ratio* in statistical parlance) to test such hypotheses.

Most of the basic theory of martingales is due to J. L. Doob. We proceed to present some of the basic results on martingales, which we need in the sequel. The first result is about convergence with probability one for a martingale. One of the main tools for the proof of this result is an inequality known as *Doob's upcrossing inequality.*

Let $x_0, x_1, \ldots, x_n$ be a finite sequence of real numbers. For $a < b$, let $u_n(a, b)$ denote the number of 'upcrossings' of the interval (a, b) by the sequence. For example, suppose $n = 7$ and $x_0 \leq a, a < x_1 < b, x_2 \geq b, x_3 \geq b, a < x_4 < b, x_5 \leq a, x_6 \leq a$ and $x_7 \geq b$. Then there are exactly two upcrossings. More generally, $u_n(a, b) = k$ if there exist exactly k pairs (and no more) of indices $0 \leq m_1 < n_1 < \cdots < m_k < n_k \leq n$ such that $x_{m_i} \leq a$ and $x_{n_i} \geq b$ for $i = 1, \ldots, k$. Here is a convenient formula for counting the number of upcrossings. Define $v_0 \equiv 1$ and for $0 \leq i \leq n$, $v_{i+1} = 0$ or v_i or 1 according as $x_i \leq a$ or $a < x_i < b$ or $x_i \geq b$. It is then easy to see that $u_n(a, b) = \sum_{i=1}^{n} (v_{i+1} - v_i)^+$. The reader can easily verify the inequality $(b-a)(v_{i+1} - v_i)^+ \leq (x_i - a)(v_{i+1} - v_i)$ for $i = 1, \ldots, n$. This immediately gives $(b-a)u_n(a, b) \leq \sum_{i=1}^{n} (x_i - a)(v_{i+1} - v_i)$. In the above, we have used the notation c^+ to denote $\max\{c, 0\}$ — known as 'the positive part' of a real number c.

Suppose now $X_0, X_1, \ldots, X_n$ are random variables and denote the corresponding number of upcrossings by $U_n(a, b)$, which is also a random variable now. Further the corresponding v_i are now denoted by V_i. For each $i \geq 1$, V_i is also a random variable and depends only on $X_0, \ldots, X_{i-1}$. From the above inequality it follows that, if the X_i have finite means, then $(b-a)E[U_n(a, b)] \leq \sum_{i=1}^{n} E[(X_i - a)(V_{i+1} - V_i)] = \sum_{i=1}^{n} E[(X_i - a)V_{i+1}] - \sum_{i=1}^{n} E[(X_i - a)V_i]$.

Assume now that we have a martingale $(X_i)_{i \geq 0}$ and apply the above to the random variables $X_0, \ldots, X_n$. For each $i = 1, \ldots, n$, we have

$$\begin{aligned} E[(X_i - a)V_i] &= E[E\{(X_i - a)V_i \,|\, X_0, \ldots, X_{i-1}\}] \\ &= E[V_i\, E\{(X_i - a) \,|\, X_0, \ldots, X_{i-1}\}] = E[(X_{i-1} - a)V_i]. \end{aligned}$$

In the above we have used properties of conditional expectations stated in Exercises 22 and 23 of Section 0.2.3 and the martingale property. Using this we get

$$\begin{aligned}(b-a)E[U_n(a,b)] &\leq \sum_{i=1}^{n} E[(X_i - a)V_{i+1}] - \sum_{i=1}^{n} E[(X_{i-1} - a)V_i] \\ &= E[(X_n - a)V_{n+1}] - E[(X_0 - a)V_1].\end{aligned}$$

Since the second term in the final expression is easily seen to be non-negative and the first term is $\leq E(|X_n - a|) \leq E|X_n| + |a|$, we have proved

Theorem (Doob's Upcrossing Inequality): *For any martingale $(X_i)_{i\geq 0}$ and for any $a < b$, $E[U_n(a,b)] \leq (E|X_n| + |a|)/(b-a)$, for all n.*

We are now ready to prove the convergence theorem known as *Doob's Martingale Convergence Theorem.* We need the following simple observation whose proof is left as an exercise. Given any sequence $(x_i)_{i\geq 0}$ of real numbers, the sequence converges if and only if for every pair of rational numbers $a < b$, $u(a,b) \stackrel{\text{def}}{=} \sup_n u_n(a,b) < \infty$. Here by the convergence of a sequence we mean that it either converges to a real number *or* diverges to $+\infty$ *or* diverges to $-\infty$.

Suppose now that $(X_i)_{i\geq 0}$ is a martingale and let $a < b$ be a pair of rational numbers. Consider the sequence of random variables $(U_n(a,b))_{n\geq 1}$ as defined earlier. Clearly this is a non-decreasing sequence of random variables taking non-negative integer values. Thus $U(a,b) = \lim_{n\to\infty} U_n(a,b)$ is well defined (possibly taking the value $+\infty$). Moreover by the Monotone Convergence Theorem, $E[U(a,b)] = \lim_{n\to\infty} E[U_n(a,b)] \leq (\sup_n E|X_n| + |a|)/(b-a)$, where the last inequality uses the upcrossing inequality. Therefore if the martingale $(X_i)_{i\geq 0}$ is assumed to satisfy the condition $\sup_n E|X_n| < \infty$, we will get $E[U(a,b)] < \infty$. This will of course imply $P[U(a,b) = +\infty] = 0$; equivalently, $P[U(a,b) < \infty] = 1$. Since this is true for every pair of rational numbers $a < b$ (and there are only countably many such pairs), we have $P[U(a,b) < \infty$ for every pair of rationals $a < b] = 1$. But this will imply by the earlier observation that $P\{\, X_i \text{ converges } \} = 1$. Further, denoting $Z = \liminf_i |X_i|$, an easy application of the Monotone Convergence Theorem (and the definition of lim inf) gives $E(Z) \leq \liminf_i E|X_i| \leq \sup_n E|X_n| < \infty$. This would imply that if X_i converges to X with probability 1, then X has finite mean. We have thus proved

Theorem (Doob's Martingale Convergence Theorem): *If $(X_n)_{n\geq 0}$ is a martingale with $\sup_n E|X_n| < \infty$, then X_n converges with probability 1 to a random variable X which has finite expectation.*

We are now going to prove that if moreover, $\sup_n E|X_n|^2 < \infty$ then the convergence takes place also in L_2, that is, $E(X^2) < \infty$ and $E(X_n - X)^2 \to 0$

as $n \to \infty$. An immediate consequence of this, which will be used by us, is the following: if $(X_n)_{n\geq 0}$ is a martingale such that $|X_n| \leq c$ for all n, where c is a finite constant, then X_n converges to X with probability 1 as well as in L_2 (hence in L^1) and, in particular, $EX = EX_0$.

For the proof, we first need the following basic result on the expectation of non-negative random variables.

Lemma: *For any non-negative random variable X with finite expectation, one has* $E(X) = \int_0^\infty P(X > \lambda)\, d\lambda$.

Proof: In case X has a density, say $f(x)$, then by an interchange of integrals,

$$\int_0^\infty P(X > \lambda)\, d\lambda = \int_0^\infty \int_\lambda^\infty f(x)dx\, d\lambda = \int_0^\infty f(x) \int_0^x d\lambda\, dx = \int_0^\infty xf(x)dx\,.$$

which equals $E(X)$ as stated. Next let us consider a discrete random variable X taking finitely many values say, $x_1 < \cdots < x_k$ with probabilities $p_1, \ldots, p_k$ respectively. In this case, it is easy to see that

$$\begin{aligned}\int_0^\infty P(X > \lambda)d\lambda = \quad & x_1 + (x_2 - x_1)(1 - p_1) + (x_3 - x_2)(1 - p_1 - p_2) + \\ & \cdots + (x_n - x_{n-1})p_n,\end{aligned}$$

and the right hand side clearly simplifies to $\sum x_i p_i = E(X)$.
If X is a non-negative discrete random variable taking an infinite number of values, say $x_1, x_2, \ldots$, then we can define a sequence (X_n) of non-negative random variables increasing to X with each X_n taking only finitely many values. To be precise, for each n, X_n is defined to be equal to X whenever X takes values from $\{x_1, \ldots, x_n\}$, and is defined to be zero otherwise. An application of the earlier case and Monotone Convergence Theorem completes the proof. ∎

Suppose that $p \geq 1$ and $E(|X|^p) < \infty$. Then by the above Lemma applied to the non-negative random variable $|X|^p$, we get

$$E(|X|^p) = \int_0^\infty P(|X|^p > \lambda)d\lambda = \int_0^\infty P(|X| > \lambda^{1/p})d\lambda.$$

An easy change of variable now leads to

Corollary: For any $p \geq 1$ and any random variable X with $E(|X|^p) < \infty$, $E(|X|^p) = p \int_0^\infty \lambda^{p-1} P(|X| > \lambda)d\lambda$.

Now let $(X_n)_{n\geq 0}$ be a martingale. For each n, let $M_n = \max_{i\leq n} |X_i|$. Fix $\lambda > 0$ and consider the event $A = \{M_n > \lambda\}$. An upper bound for $P(A)$ is

provided by what is known as *Doob's Maximal Inequality* given below.

Lemma (Doob's Maximal Inequality): $P(A) \leq E(|X_n| \cdot I_A)/\lambda$.

Proof: Let $A_0 = \{|X_0| > \lambda\}$ and $A_i = \{|X_0| \leq \lambda, \ldots, |X_{i-1}| \leq \lambda, |X_i| > \lambda\}$, for $1 \leq i \leq n$. Then $A_0, \ldots, A_n$ are disjoint and $A = \bigcup_i A_i$, so that, $P(A) = \sum P(A_i) \leq \frac{1}{\lambda} \sum E(|X_i| I_{A_i})$. An easy consequence of the martingale property of (X_n) is that, for any $i \leq n$, $X_i = E(X_n \,|\, X_0, \ldots, X_i)$. This would therefore give

$$\begin{aligned} P(A) &\leq \frac{1}{\lambda} \sum E(|E(X_n \,|\, X_0, \ldots, X_i)| I_{A_i}) \\ &\leq \frac{1}{\lambda} \sum E(E(|X_n| \,|\, X_0, \ldots, X_i) I_{A_i}) \\ &= \frac{1}{\lambda} \sum E(E(|X_n| I_{A_i} \,|\, X_0, \ldots, X_i)) \\ &= \frac{1}{\lambda} \sum E(|X_n| I_{A_i}) = \frac{1}{\lambda} E(|X_n| I_A). \end{aligned}$$

∎

Applying now the Corollary above with $p = 2$ and $X = M_n$ followed by the Lemma, we get

$$\begin{aligned} E(M_n^2) &\leq 2 \int_0^\infty E(|X_n| I_{\{M_n > \lambda\}})\, d\lambda = 2E\left(|X_n| \int_0^\infty I_{\{M_n > \lambda\}}\, d\lambda\right) \\ &= 2E(|X_n| M_n) \leq 2\sqrt{E(X_n^2)}\sqrt{E(M_n^2)}. \end{aligned}$$

This leads to $E(M_n^2) \leq 4E(X_n^2)$. If now (X_n) is an L_2-bounded martingale, that is, $\sup_n E(X_n^2) < \infty$, then it follows that

$$E\left(\sup_n |X_n|^2\right) \leq 4 \sup_n E(X_n^2) < \infty\,,$$

whence by Dominated Convergence Theorem we get

Theorem: *If $(X_n)_{n \geq 0}$ is an L_2-bounded martingale, then it converges with probability one to a random variable X having finite second moment and moreover, the convergence is also in L_2, that is, $\lim_n E(X_n - X)^2 = 0$.*

0.8 Markov Chains

Consider a system which evolves with time in a random way. For the sake of simplicity, let us consider the set of times to be discrete. Let us also assume that the set S of possible states of the system is countable. S will be called the *state space* of the system and the individual states (i.e. elements of S) will be denoted by i, j, k etc. Let X_n denote the (random) state of the system at time n, $n = 0, 1, 2, \ldots$. We are assuming that each X_n is an S-valued random variable and the entire evolution of the system is described by the sequence $(X_n)_{n \geq 0}$. In particular, X_0 is the *initial state* of the system. Study of such systems in this generality, without any further assumptions, will not lead to any interesting

theory. Usually one imposes additional restrictions on the joint distribution of the sequence to get different kinds of "stochastic processes". One such condition, studied in the last section, is that $E(X_n | X_0, X_1, \ldots, X_{n-1}) = X_{n-1}$ for all n, which gave rise to what are called martingales. In this section, we study one other extremely important and useful condition that leads to a class of processes known as *Markov chains.*

0.8.1 Markov Chains: Transition Probabilities

The property that is imposed can be briefly referred to as 'lack of memory', by which we mean that given the present state of the system the 'future' evolution does not depend on the 'past' history. In mathematical terms, this means that for any two non-negative integers n and m, the conditional distribution of X_{n+m} given $X_0, X_1, \ldots, X_n$ depends only on X_n. It turns out that one needs only to assume this for $m = 1$. Thus we have the following definition.

Definition: A sequence $(X_n)_{n \geq 0}$ of random variables taking values in a countable set S is acIled a *Markov chain* on the state space S if, for any $n \geq 0$ and any $i_0, i_1, \ldots, i_{n-1}, i, j \in S$,

$$P(X_{n+1} = j \,|\, X_0 = i_0, \ldots, X_{n-1} = i_{n-1}, X_n = i) = P(X_{n+1} = j \,|\, X_n = i)\,.$$

Further, the chain is called *time homogeneous*, if the above conditional probabilities do not depend on n and hence are also equal to $P(X_1 = j \,|\, X_0 = i)$.

Markov chains appearing in most applications also happen to be time homogeneous. A rich theory exists for such chains. We shall restrict ourselves to only Markov chains which are time homogeneous. We will denote, for any $i, j \in S$, the probability $P(X_1 = j \,|\, X_0 = i)$ by p_{ij}. Writing the distribution of X_0 as $\{\mu_i, i \in S\}$, it is not difficult to see that all the finite dimensional joint distributions for $(X_n)_{n \geq 0}$ are completely determined by the quantities μ_i, $i \in S$ and p_{ij}, $i, j \in S$. Specifically, for any n and any collection of states $i_0, i_1, \ldots, i_n$,

$$P(X_0 = i_0, X_1 = i_1, \ldots, X_n = i_n) = \mu_{i_0} p_{i_0 i_1} \cdots p_{i_{n-1} i_n}\,.$$

$\{\mu_i, i \in S\}$ is called the *initial distribution* and $\{p_{ij}, i, j \in S\}$ are called the *transition probabilities* or *one-step transition probabilities*, to be exact. From the definition it is clear that,

(1) $\mu_i \geq 0$ for all i and $\sum_i \mu_i = 1$,

(2) $p_{ij} \geq 0$ for all $i, j \in S$ and $\sum_j p_{ij} = 1$ for all $i \in S$.

It is often convenient to represent the initial distribution by a row vector $\mu = (\mu_i; i \in S)$ and the transition probabilities by the *transition matrix* $P = ((p_{ij}))_{i,j \in S}$. The property (2) above simply says that P has non-negative entries with each row sum equal to one. Such matrices are called *stochastic matrices.* Much of the theory of Markov chains rests on an analysis of its transition

matrix P and not so much on μ. The matrix P^n, the n-th power (in the sense of matrix multiplication) of the matrix P, is called the *n-step transition matrix* simply because its (i,j)-th element $p_{ij}^{(n)}$ gives the probability of transition from i to j in n steps, that is,

$$p_{ij}^{(n)} = P(X_{n+m} = j \,|\, X_m = i) = P(X_n = j \,|\, X_0 = i)\,.$$

One can easily verify this for $n = 2$ and then use induction to complete the proof.

One useful consequence, known as the *Chapman-Kolmogorov equations*, is

$$p_{ij}^{(m+n)} = \sum_k p_{ik}^{(m)} p_{kj}^{(n)}\,. \tag{24}$$

This can of course be verified directly from Markov property. For the sake of completeness we need to set $P^{(0)} = I$, the identity matrix which is also consistent with the notion of zero-step transition.

A simple yet useful property of a Markov chain $(X_n)_{n\geq 0}$ is that if f is any real function on the state space S satisfying $\sum_j p_{ij} f(j) = f(i)$ for all i, then the sequence $(f(X_n))_{n\geq 0}$ is a martingale, provided, of course, the sum $\sum f(i)\mu_i$ is convergent. Functions f satisfying $\sum_j p_{ij} f(j) = f(i)$ for all i, are known as *harmonic functions* for the transition matrix P.

Example 1: Let $\xi_1, \xi_2, \ldots$ be i.i.d. integer-valued random variables with common distribution $P(\xi_1 = j) = \alpha_j$. Let X_0 be any integer valued random variable independent of the ξ sequence. For $n \geq 1$, let $X_n = X_0 + \sum_{l=1}^{n} \xi_l$. Then (X_n) is a Markov chain with state space $S =$ the set of integers and transition probabilities $p_{ij} = \alpha_{j-i}$. The n-step transition probabilities are also not difficult to get. An elegant formula for these can be obtained using the following notation. For any two probability distributions $\alpha = (\alpha_j)$ and $\beta = (\beta_j)$ on integers, let $\alpha * \beta$ denote the distribution defined by $(\alpha * \beta)_j = \sum_i \alpha_i \beta_{j-i}$. In particular, α^{*n} is defined recursively by $\alpha^{*n} = \alpha^{*(n-1)} * \alpha$. With this notation, the n-step transition probabilities of the chain (X_n) above are given by $p_{ij}^{(n)} = \alpha_{(j-i)}^{*n}$.

It may be noted that here p_{ij} as well as $p_{ij}^{(n)}$ depend on i and j only through $j - i$. In fact, these are the only Markov chains with this property. In other words, if $(X_n)_{n\geq 0}$ is a Markov chain whose transition probabilities p_{ij} depend only on $i - j$, then the random variables $\xi_n = X_n - X_{n-1}$, $n \geq 1$, are i.i.d. The proof is easy. Such Markov chains are called *random walks*. A special case is when the random variables ξ_j take only two values $+1$ and -1. This gives what is known as *simple random walk*. A further special case when $P(\xi_j = +1) = p(\xi_j = -1) = 1/2$, is called *simple symmetric random walk*. Feller's book (vol.1) gives an extensive and interesting account of such random walks. His analysis is based entirely on what is known as 'path counting' and

is therefore easily accessible. Interested reader should consult this book. We will include parts of this material at the end of this section.

Example 2: Consider an urn with a total of D tokens — some numbered $+1$ and some -1. The composition of the urn changes over time as follows. At each turn, a token is picked at random from the urn and its sign changed and put back in the urn. Denote by X_n the number of $+1$ at time n. It is clear that X_n is a Markov chain with state space $S = \{0, 1, \ldots, D\}$ and transition probabilities

$$p_{i,i+1} = 1 - \frac{i}{D} = 1 - p_{i,i-1} .$$

Thus, from a state i, transitions are possible only to states $i-1$ or $i+1$ in one step. Of course if $i = 0$ (respectively D) then the system moves surely to 1 (respectively $D-1$). It is not difficult to see that the two-step transition probabilities are given by:

$$p^{(2)}_{i,i+2} = (1 - \frac{i}{D})(1 - \frac{i+1}{D}), \qquad p^{(2)}_{i,i-2} = \frac{i}{D}\frac{i-1}{D},$$

$$p^{(2)}_{ii} = 1 - \frac{i(i-1)}{D^2} - \frac{(D-i)(D-i-1)}{D^2} .$$

Exercise 1. Suppose X_0 is uniformly distributed on the state space S in the above example. Calculate the distributions of X_1, X_2 and also the joint distribution of (X_1, X_2). Do the same when $X_0 \sim B(D, 1/2)$.

Example 3 ($0-1$ chain): Consider a machine which can be in two states, 'on' and 'off'. Also if the machine is 'on' today, then the probability is α that it will be 'off' tomorrow. Similarly, β is the probability of transition from 'off' state to 'on' state in one day. Denote the 'on' and 'off' states by 0 and 1 respectively. Denoting by X_n the state of the machine on day n, $(X_n)_{n\geq 0}$ is a Markov chain with state space $S = \{0, 1\}$ and transition matrix

$$P = \begin{pmatrix} p_{00} & p_{01} \\ p_{10} & p_{11} \end{pmatrix} = \begin{pmatrix} 1-\alpha & \alpha \\ \beta & 1-\beta \end{pmatrix} .$$

We assume that $\alpha + \beta > 0$ (what happens if $\alpha + \beta = 0$?). A trite calculation gives

$$p^{(n)}_{01} = p^{(n-1)}_{00}\alpha + p^{(n-1)}_{01}(1-\beta) = \alpha + p^{(n-1)}_{01}(1-\alpha-\beta) ,$$

from which one deduces

$$p^{(n)}_{01} = \frac{\alpha}{\alpha+\beta}[1 - (1-\alpha-\beta)^n] .$$

Similar calculation shows that

$$p^{(n)}_{10} = \frac{\beta}{\alpha+\beta}[1 - (1-\alpha-\beta)^n] .$$

If one further assumes that $\alpha + \beta < 2$ (what happens if $\alpha + \beta = 2$?), so that $|1 - \alpha - \beta| < 1$, then one gets $p_{01}^{(n)} \to \frac{\alpha}{\alpha+\beta}$ and $p_{10}^{(n)} \to \frac{\beta}{\alpha+\beta}$. Since $p_{00}^{(n)} = 1 - p_{01}^{(n)}$ we deduce that $p_{00}^{(n)} \to \frac{\beta}{\alpha+\beta}$. Similarly $p_{11}^{(n)} \to \frac{\alpha}{\alpha+\beta}$.

From all these, one can deduce that, if $0 < \alpha + \beta < 2$, then the Markov chain has the limiting distribution $\pi = \langle \pi_0, \pi_1 \rangle = \langle \frac{\beta}{\alpha+\beta}, \frac{\alpha}{\alpha+\beta} \rangle$, whatever be the initial distribution of the chain. This distribution represents what is called the 'equilibrium' or 'steady-state' distribution of the chain. It is so called because π also happens to be the only distribution on S with the property that if X_0 has distribution π, then for every n, X_n has the same distribution π. This conclusion also follows from the equations describing $p_{ij}^{(n)}$.

This example leads one to the natural question. Is the above kind of phenomenon true for all Markov chains? That is, does every Markov chain have a limiting distribution? Is the limiting distribution unique (i.e. independent of initial conditions)? Are these limiting distributions, if any, also steady-state distributions in the above sense? The cases $\alpha + \beta = 0$ and $\alpha + \beta = 2$ should convince the reader that the answers to the above questions cannot always be in the affirmative.

To better understand the nature of problems and to identify some of the cases where we do have affirmative answer, we first need to discuss 'classification' of the states. This will be done in the next section. But let us now make a little digression to discuss some interesting properties of simple random walk as described in Example 1.

Simple Random Walk: Recall simple random walk as discussed in Example 1. Here is an illustration of the path-counting argument and the final result will be used in Chapter 4.

In the context of random walks, a path from $(0,0)$ to (n,a) is a polygonal line with vertices (i, s_i) for $0 \leq i \leq n$, with $s_0 = 0$, $s_n = a$ and $s_i - s_{i-1} = \pm 1$ for $1 \leq i \leq n$. It is clear that a has to be an integer between $-n$ and $+n$. Similarly one can talk about paths from (m,a) to $(m+n,b)$, for integers m and n with $n > 0$. Such paths are called paths of length n. It is clear that the total number of paths of length n starting from a given point is 2^n. Also the number of paths from $(0,0)$ to (n,a) is $\binom{n}{\frac{n+a}{2}}$.

A fact, often called the *reflection principle* is that, for integers $a, b > 0$, the number of paths from $(0,a)$ to (n,b) that touch or cross the X-axis is the same as the total number of paths from $(0,-a)$ to (n,b) and hence equals $\binom{n}{\frac{n+a+b}{2}}$. This is done by establishing a one-one correspondence between the two sets of paths. From this one can easily deduce that for any integer $a > 0$, the number of paths from $(0,0)$ to (n,a) that do not hit the X-axis equals $\binom{n-1}{\frac{n+a}{2}-1} - \binom{n-1}{\frac{n+a}{2}}$. Incidentally, this formula also gives a solution to what

is known as the *ballot problem.*

We now turn to an important property of simple, but not necessarily symmetric, random walk. In other words, we consider the Markov chain on the set of integers with transition probabilities $p_{i,i+1} = p = 1 - p_{i,i-1}$. For any integer a, let $T_a = \inf\{n \geq 1 : X_n = a\}$, that is, T_a is the hitting time of a. Fix integers $a < i < b$. Let

$$\varphi(i) = P(T_a < T_b \,|\, X_0 = i) \quad \text{for} \quad a < i < b$$

and $\varphi(a) = 1$ and $\varphi(b) = 0$.

Exercise 2.

(i) Show that for $a < i < b$, $\varphi(i) = p\varphi(i+1) + (1-p)\varphi(i-1)$

(ii) Denoting $d_i = \varphi(i) - \varphi(i-1)$, show that for $a + 1 < i \leq b$,

$$pd_i = (1-p)d_{i-1},$$

and hence that

$$d_i = \left(\frac{1-p}{p}\right)^{i-a-1} d_{a+1}\,.$$

(iii) Assume that $p = 1/2$ and show that $\varphi(i) = 1 + (i-a)d_{a+1}$ for $a \leq i \leq b$, and hence deduce that $\varphi(i) = (b-i)/(b-a)$.

(iv) Assume that $p \neq 1/2$ and denote $(1-p)/p$ by α. Show that

$$\varphi(i) = 1 + \frac{p}{1-2p}\, d_{a+1}\, \left[\alpha^{i-a} - 1\right]$$

for $a \leq i \leq b$ and hence deduce that

$$\varphi(i) = \frac{\alpha^b - \alpha^i}{\alpha^b - \alpha^a}\,.$$

(v) If $p > 1/2$ then show that

$$P(T_a < \infty \,|\, X_0 = i) \quad = \quad \alpha^{i-a} < 1\,.$$

(vi) If $p \leq 1/2$ then show that

$$P(T_a < \infty \,|\, X_0 = i) \quad = \quad 1\,.$$

0.8.2 Classification of States: Recurrence and Transience

For any state i, let us define two random variables, possibly taking value $+\infty$, as follows:

$$T_i = \min(n \geq 1 : X_n = i), \qquad N_i = \#(n \geq 1 : X_n = i)\,.$$

In the definition of T_i, if there is no $n \geq 1$ such that $X_n = i$ we take $T_i = +\infty$. If there are infinitely many such n, then of course, $N_i = +\infty$. T_i represents the time of the first visit to i and N_i represents the total number of visits to i. In both of these, the time point 0 is not counted. It is clear that the events $(T_i < \infty)$ and $(N_i \geq 1)$ are same.

For i and j in S, let

$$f_{ij}^{(n)} = P(T_j = n \,|\, X_0 = i) = P(X_n = j; X_l \neq j, 1 \leq l < n \,|\, X_0 = i), \tag{25}$$

$$f_{ij} = \sum_{n=1}^{\infty} f_{ij}^{(n)} = P(T_j < \infty \,|\, X_0 = i)\,. \tag{26}$$

It is clear from the definitions that $p_{ij}^{(n)} \geq f_{ij}^{(n)}$. In fact one has the following identity often known as the *renewal equation* :

$$p_{ij}^{(n)} \quad = \quad \sum_{m=1}^{n} f_{ij}^{(m)} p_{jj}^{(n-m)}\,. \tag{27}$$

To prove the equation, one has to simply write the event $(X_n = j)$ as the disjoint union of events $\bigcup_{m=1}^{n} (T_j = m, X_n = j)$ and calculate the probabilities of these events by applying the Markov property.

All the states of the Markov chain are classified into two kinds as defined below.

Definition: A state i is called *recurrent* if $f_{ii} = 1$ and is called *transient* otherwise. A state i is called *absorbing* if $p_{ii} = 1$.

Thus a state i is recurrent if the chain starting from i is sure to return to i at some future (possibly random) time. Naturally, for a transient state i there is a positive probability of never returning to i. Clearly every absorbing state is recurrent.

Since the two events $(T_i < \infty)$ and $(N_i \geq 1)$ are identical, it follows that a state i is recurrent iff $P(N_i \geq 1 \,|\, X_0 = i) = 1$, that is, the chain starting from the recurrent state i is sure to make at least one visit to i. We will, in fact, show that starting from a recurrent state i, the chain actually makes infinitely many visits to i with probability one. Intuitively, this should be obvious from the Markov property. To do this rigorously and get some other results we need the following identity:

For states i and j and any $m \geq 1$,

$$P(N_j \geq m \,|\, X_0 = i) = f_{ij} \cdot f_{jj}^{m-1}\,. \tag{28}$$

For $m = 1$, this is just the definition of f_{ij}. Let us prove it for $m = 2$. Clearly,

$$P(N_j \geq 2 \,|\, X_0 = i) = \sum_{n=1}^{\infty} \sum_{n'=1}^{\infty} P(A_{n,n'} \,|\, X_0 = i),$$

where $A_{n,n'} = \left\{ \begin{array}{l} X_n = X_{n+n'} = j; \\ X_p \neq j \text{ for } 1 \leq p < n \text{ and for } n+1 \leq p < n+n' \end{array} \right\}$.

By using the properties of conditional probability and the Markov property, each summand reduces to the product

$$P(X_n = j, X_p \neq j, 1 \leq p < n | X_0 = i) \cdot P(X_{n'} = j, X_p \neq j, 1 \leq p < n' | X_0 = j),$$

so that

$$P(N_j \geq 2 | X_0 = i) = \sum_{n=1}^{\infty} \sum_{n'=1}^{\infty} f_{ij}^{(n)} f_{jj}^{(n')} = f_{ij} f_{jj} .$$

The proof for a general m is similar. Do it for $m = 3$ to make sure that you understand.

Notice that the events $(N_j \geq m)$ are decreasing as m increases with limit being the event $(N_j = \infty)$. If j is a transient state then $f_{jj} < 1$, so that for every i, $P(N_j = \infty | X_0 = i) = 0$. That is, a transient state can be visited at most a finite number of times, no matter where the chain starts. On the other hand, if j is recurrent, then $P(N_j = \infty | X_0 = i) = f_{ij}$. In particular $P(N_j = \infty | X_0 = j) = 1$, as stated earlier.

It also follows that if j is a recurrent state, then $E(N_j | X_0 = i) = 0$ or ∞ according as $f_{ij} = 0$ or $f_{ij} > 0$. In particular, $E(N_j | X_0 = j) = \infty$ if j is recurrent. On the other hand, if j is transient then

$$P(N_j = m | X_0 = i) = f_{ij}(1 - f_{jj}) f_{jj}^{m-1} \qquad \text{for } m = 1, 2, \ldots .$$

Since $f_{jj} < 1$, we have $E(N_j | X_0 = i) = f_{ij}/(1 - f_{jj})$. In particular, it follows that $E(N_j | X_0 = i) < \infty$.

The above analysis leads to another characterization of transience and recurrence, namely, *a state j is recurrent if and only if the series $\sum_n p_{jj}^{(n)}$ diverges.* To see this, one defines random variables $(Y_n, n \geq 1)$ as $Y_n = 1$ if $X_n = j$ and $Y_n = 0$ otherwise. Then $N_j = \sum_{n=1}^{\infty} Y_n$, so that, for any i,

$$E(N_j | X_0 = i) = \sum E(Y_n | X_0 = i) = \sum_{n=1}^{\infty} p_{ij}^{(n)} .$$

The above-stated characterization of recurrence follows now by taking $i = j$.

Further, for a transient state j, the series $\sum p_{ij}^{(n)}$ converges to $\dfrac{f_{ij}}{1 - f_{jj}}$ for every state i. In particular, $p_{ij}^{(n)} \to 0$ as $n \to \infty$.

This last observation can be used to deduce that if the state space is finite then there has to be at least one recurrent state. This is clear since $\sum_{j \in S} p_{ij}^{(n)} = 1$ always for every n. Therefore if S is finite, $\lim_{n \to \infty} \sum_{j \in S} p_{ij}^{(n)} = 1$ also. This makes it impossible that $p_{ij}^{(n)} \to 0$ for all j.

Given states i and j we say that i *leads to* j, in symbols $i \hookrightarrow j$, if $f_{ij} > 0$. This can be seen to be equivalent to requiring that $f_{ij}^{(n)} > 0$ for some $n \geq 1$, which in turn is the same as requiring $p_{ij}^{(n)} > 0$ for some $n \geq 1$. It is a simple consequence of the Chapman-Kolmogorov equations that if $i \hookrightarrow j$ and $j \hookrightarrow k$, then $i \hookrightarrow k$.

An important result is that *a recurrent state does not lead to a transient state.* More specifically, if i is recurrent and $i \hookrightarrow j$ then j is recurrent and $f_{ij} = f_{ji} = 1$. We only need to prove the result when $i \neq j$. We first prove that $f_{ji} = 1$. Since $i \hookrightarrow j$, $p_{ij}^{(n)} > 0$ for some $n \geq 1$. Let m be the smallest such n. Then we can get states $i_1, i_2, \ldots, i_{m-1}$, all different from i, such that $p_{ii_1}p_{i_1i_2}\cdots p_{i_{m-1}j} = \alpha > 0$. Suppose, if possible, $f_{ji} < 1$, that is, $P(X_n \neq i \quad \forall n \,|\, X_0 = j) = \beta > 0$. But $P(X_n \neq i \quad \forall n \,|\, X_0 = i)$ is at least as much as

$$P(X_1 = i_1, \ldots, X_{m-1} = i_{m-1}, X_m = j \text{ and } X_{m+n} \neq i \,\forall n \geq 1 \,|\, X_0 = i)\,.$$

By Markov property, it is easy to see that the right hand side equals $\alpha\beta > 0$, contradicting the recurrence of i. Thus we must have $f_{ji} = 1$. In particular, $j \hookrightarrow i$. Now the recurrence of j is derived as follows. Let $m' \geq 1$ be such that $p_{ji}^{(m')} > 0$. Then

$$\sum_{n=1}^{\infty} p_{jj}^{(n)} \quad \geq \quad \sum_{n=1}^{\infty} p_{ji}^{(m')} p_{ii}^{(n)} p_{ij}^{(m)}$$

and the right hand side diverges because both $p_{ji}^{(m')}$ and $p_{ij}^{(m)}$ are strictly positive and i is recurrent. This implies divergence of the left hand side and hence recurrence of j. That $f_{ij} = 1$ is obtained now by reversing the roles of i and j.

Results of Exercise 2 in the previous section really tell us that for a simple random walk with $p > 1/2$, $f_{ij} < 1$ for all $i > j$. Using the fact that $i \hookrightarrow j$ for any two states i and j, we deduce that all states are transient in case $p > 1/2$. One can similarly show that the same is true if $p < 1/2$. It would be an interesting exercise to go back to the formula for d_i and examine what happens in case $p = 1/2$. The reader should be able to show that now $f_{ij} = 1$ for all $i \neq j$ and deduce from this that all states are recurrent.

0.8.3 Decomposition of the State Space: Irreducible Closed Classes

The limiting and steady state behaviour of a Markov chain is intimately connected with a partition of the state space. One way to get the decomposition is to define an equivalence relation between states. Given states i and j, we will say that they are *communicating* if either $(i = j)$ or $(i \hookrightarrow j$ and $j \hookrightarrow i)$. It is easy to see that this is an equivalence relation so that the whole state space S is partitioned as a disjoint union of equivalence classes. These equivalence classes are called *communicating classes.*

From the earlier result it is clear that in a communicating class either all states are recurrent or all states are transient. A communicating class is called *recurrent* (respectively, *transient*) if all states in the class are recurrent (respectively, transient). It is natural to ask how to interpret these equivalence classes in terms of the behaviour of the chain. Let us make a definition first.

Definition: A set $C \subset S$ is said to be *closed* or *stochastically closed* if $p_{ij} = 0$ for $i \in C$ and $j \notin C$ or equivalently, for $i \in C$, $\sum_{j \in C} p_{ij} = 1$.

The condition in the definition above can easily be seen to be equivalent to $\sum_{j \in C} p_{ij}^{(n)} = 1$ for all $i \in C$ and for all $n \geq 1$. This really means that if the chain starts from $i \in C$, then with probability one it remains in C for ever. More precisely,

$$P(X_n \in C \quad \forall n \geq 1 \,|\, X_0 \in C) = 1\,.$$

The state space S is trivially a closed set. A singleton set $C = \{i\}$ is closed iff i is an absorbing state. It is also easy to see that any recurrent communicating class is closed and moreover it does not have a proper subset which is closed. It is therefore natural to ask "what are the minimal closed subsets of S?"

A closed set C is called *irreducible* if $i \hookrightarrow j$ for every i and j in C. It is easy to see that a closed irreducible set C is minimal in the sense that no proper subset of C is closed. A closed communicating class is irreducible. In particular, any recurrent communicating class is closed and hence irreducible. One can not say the same thing about transient classes simply because they may not be closed. In fact, as already remarked, a finite transient class can never be closed. An infinite transient class may or may not be closed.

Exercise 3. Let the state space S be the set of all integers. The transition matrix is given by $P_{i,i+1} = 3/4$ and $p_{i,i-1} = 1/4$. (This is just the simple random walk with $p = 3/4$.) Show that the chain is transient, S is a transient class and S is closed.

Exercise 4. Let the state space S be the set of all non-negative integers. The transition matrix is given by: $p_{00} = 1$ and, for $i \geq 1$, $p_{i,i+1} = 3/4$, $p_{i,i-1} = 1/4$. Then the set of strictly positive integers is a transient class, but not closed.

In passing let us also note that there may not be any closed irreducible set. For example, let $S = \{0, 1, \ldots\}$ and $p_{i,i+1} = 1$. One can easily see that sets of the form $\{k, k+1, \ldots\} \subset S$ are all closed and these are the only closed sets. Thus no closed set is irreducible. Of course, such a behaviour is impossible for a finite state space Markov chain. In fact, for a finite state space Markov chain, the structure is fairly simple. Since finite state space chains are all that we will be needing for our applications, from now on

let us specialize to the case where the state space is finite.

In this case, we have a unique decomposition of the state space S as follows. $S = S_R \cup S_T$, where S_R is the set of recurrent states (necessarily non-empty)

and S_T those of transient states (possibly empty). Further $S_R = \bigcup_{l=1}^{k} C_l$ where each C_l is an irreducible closed set. If the chain starts in C_l it remains there forever visiting each state an infinite number of times with probability one. Thus if $S_T = \emptyset$, we may, depending on the initial state of the chain, study the chain only on a reduced state space, namely, one of the C_l.

In fact, even if $S_T \neq \emptyset$, a chain starting in S_T will, after a finite (random) number of steps, has to enter one of the C_l — and, of course, will remain there from then on. The long-term behaviour of the chain will therefore be still determined by the analysis of the chain on the restricted state space C_l. The only other things that are pertinent in this case are: How long does it take to enter one of the classes C_l and what are the probabilites of entering the different classes? We address these questions first.

Let $i \in S_T$ and $1 \leq l \leq k$. Let

$$\begin{aligned} \alpha_{il} &= P(X_n \in C_l \text{ for some } n \,|\, X_0 = i) \\ &= P(X_n \in C_l \text{ for all large } n \,|\, X_0 = i)\,. \end{aligned}$$

From what has been said above $\sum_{l=1}^{k} \alpha_{il} = 1$. Let us also denote for $i \in S_T$ and for $1 \leq l \leq k$,

$$\beta_{il} = \sum_{j \in C_l} p_{ij} = P(X_1 \in C_l \,|\, X_0 = i)\,.$$

It is now easy to see from the Markov property that

$$\alpha_{il} = \beta_{il} + \sum_{j \in S_T} p_{ij}\alpha_{jl}\,. \tag{29}$$

In other words, for each l, the numbers $(\alpha_{il})_{i \in S_T}$ satisfy the system of linear equations given by (29). In fact, one can show that it is the unique solution. It is convenient to write this system of equations in matrix form. Let Q denote the submatrix of P of order $S_T \times S_T$ defined as $Q = (p_{ij})_{i,j \in S_T}$. It is convenient to think of the rows and columns of Q indexed by states in S_T. For fixed l, $1 \leq l \leq k$, let $\widetilde{\alpha}_l$ and $\widetilde{\beta}_l$ be the column vectors of size S_T with entries α_{il} and β_{il} respectively for $i \in S_T$. Then $\widetilde{\alpha}_l$ is the unique solution of the equation

$$\widetilde{\alpha}_l = \widetilde{\beta}_l + Q\widetilde{\alpha}_l\,.$$

The uniqueness is a consequence of invertibility of the matrix $(I - Q)$, which in turn follows from the fact that the series $I + Q + Q^2 + \cdots$ is convergent and is indeed the inverse of $I - Q$ (finiteness of S_T plays a role here). Here I is the identity matrix. As a consequence

$$\widetilde{\alpha}_l = (I - Q)^{-1}\widetilde{\beta}_l\,.$$

The duration of time that a chain takes, before it enters S_R, is

$$\tau = \min\{n \geq 1 : X_n \in S_R\}.$$

We want to get a formula for the expected value of τ starting from different transient states. For $i \in S_T$, let

$$m_i = E(\tau \,|\, X_0 = i),$$

and let $\widetilde{m}$ be the column vector of size S_T with entries m_i. Denoting $\widetilde{e}$ to be the column vector of size S_T with all entries 1, it is easy to see that $\widetilde{m}$ satisfies the equation

$$\widetilde{m} \quad = \quad \widetilde{e} + Q\widetilde{m},$$

from which the unique solution for $\widetilde{m}$ emerges as

$$\widetilde{m} = (I - Q)^{-1}\widetilde{e}.$$

From the above analysis, it is clear that the matrix $(I - Q)^{-1}$ plays a fundamental role and is appropriately called the *fundamental matrix* of the chain, denoted by N. The above method is often referred to as the *fundamental matrix method.* A particularly useful special case — which would also be singularly relevant for applications in Markov models in genetics — is what are known as *absorbing chains.*

Definition: A Markov chain on a finite state space, for which all the recurrent states are absorbing, is called an *absorbing chain.*

For an absorbing chain, each C_l obtained in the decomposition of S_R, consists of a single absorbing state. Entering the different C_l really means getting absorbed in one of the absorbing states. For each $i \in S_T$, the numbers α_{il}, for $1 \leq l \leq k$, are called the *absorption probabilities* starting from i.

As seen earlier, we can only solve for α_{il} simultaneously for all $i \in S_T$ with l held fixed. In other words each vector $\widetilde{\alpha}_l$ is supposed to be solved separately for each l. However, in case there are only two absorbing states — that is $k = 2$ — then solving for one l is enough (why?).

In case of absorbing chains, it is notationally convenient to list the states so that the absorbing states come before the transient states. To avoid triviality, we assume that there is at least one transient state. With this ordering of states, the transition matrix P takes the form

$$P \quad = \quad \begin{pmatrix} I & O \\ R & Q \end{pmatrix},$$

where Q is as before. The entries of NR are precisely the absorption probabilities α_{il} for $i \in S_T$ and $1 \leq l \leq k$. Entries of $N\widetilde{e}$ are precisely the mean times till absorption.

Exercise 5. Consider a chain with four states and transition matrix

$$P = \begin{pmatrix} 1 & 0 & 0 & 0 \\ 0 & 1 & 0 & 0 \\ 1/4 & 0 & 1/2 & 1/4 \\ 1/4 & 1/4 & 1/4 & 1/4 \end{pmatrix}$$

Calculate the fundamental matrix, the absorption probabilities and mean times till absorption.

Exercise 6. For an absorbing chain, show that the vector of variances of the time till absorption starting from different transient states is given by $\widetilde{V} = (2N - I)\widetilde{m} - \widetilde{m}^2$ where $\widetilde{m}^2$ denotes the vector whose entries are the squares of the entries of $\widetilde{m}$.

0.8.4 Ergodic Chains: Limiting Behaviour and Stationary Distributions

We go back to a general finite state space Markov chain and recall the decomposition of the state space

$$\begin{aligned} S &= S_R \cup S_T \\ &= \bigcup_{l=1}^{k} C_l \cup S_T \end{aligned}$$

where S_T and S_R are the sets of transient and recurrent states respectively and C_l, $1 \leq l \leq k$, are the closed irreducible subsets of S_R.

As noted already, the long-term behaviour of the chain is determined by the analysis of the chain on the restricted state spaces C_l. This is what we want to pursue now. Accordingly, let us assume that the whole state space S of the chain is one single closed irreducible class of recurrent states. Such a chain is called an *irreducible recurrent chain*, also sometimes referred to as an *ergodic chain*. For such a chain, we are going to show that there exists a unique $\pi = \{\pi_i, i \in S\}$ with $\pi_i > 0$ and $\sum \pi_i = 1$ such that $\pi P = \pi$ or equivalently, there is a unique probability π on S such that if $X_0 \sim \pi$ then $X_1 \sim \pi$ (in fact, $X_n \sim \pi$ for all n). It is this property that is described by saying that π is an *invariant distribution* for the chain — also called a *steady-state* or an *equilibrium* or a *stationary* distribution. Formally,

Definition: By an *invariant distribution* of a Markov chain is meant a probability distribution π on the state space S for which the equality $\pi P = \pi$ holds.

Exercise 7. If $X_0 \sim \pi$ where π is an invariant distribution, show that the joint distribution of (X_n, X_{n+1}) is same for all n. Generalize from pairs to triplets etc.

A Markov chain may not have any invariant distribution. In case it has, it may have more than one invariant distributions. It is not difficult to show that symmetric simple random walk has no invariant distribution. On the

other extreme, the Markov chain on $S = \{0, 1\}$, with $p_{00} = p_{11} = 1$, is an easy example of a chain with plenty of invariant distributions! Our following analysis will show, among other things, that a finite state space Markov chain will always have at least one invariant distribution.

Indeed, we will show that for an ergodic chain with finitely many states there is one and only one invariant distribution. This invariant distribution π will also turn out to be the limiting distribution of the chain in the following sense:

$$\text{for all} \quad i, j \in S, \quad \lim_n \frac{1}{n} \sum_{l=1}^{n} p_{ij}^{(l)} \quad = \quad \pi_j \,. \tag{30}$$

It may be noted that the above limit does not depend on i. In other words, the effect of the initial distribution wears off in the long run.

We first prove that the limits in the left-hand side of (30) exist and are free of i. We start with some notations. For any state i, $P_i(A)$ will denote the conditional probability of the event A, given $X_0 = i$. Thus $P_i(A)$ can be thought of as the probability of the event A when the initial distribution is concentrated at i. Expectation wih respect to this probability will be denoted by E_i. Let us, from now on, fix a state j. Let $T_1, T_2, \ldots$ be the times of successive visits to the state j. Only times greater than or equal to one are considered for these visits. That is,

$$T_1 \quad = \quad \min\{n \geq 1 : X_n = j\},$$

and for $r \geq 2$,

$$T_r \quad = \quad \min\{n > T_{r-1} : X_n = j\} \,.$$

Since S is a closed irreducible recurrent class, $P_i(T_r < \infty \text{ for all } r) = 1$. Set $Z_0 = T_1$ and for $r \geq 1$, $Z_r = T_{r+1} - T_r$. We claim that the Markov property of (X_n) implies that for any i, the random variables $Z_1, Z_2, \ldots$ are i.i.d. under P_i. Moreover, the common distribution is that of T_1 under P_j (and hence does not depend on the initial state i). To see this, note first that $P_i(Z_1 = l_1 \,|\, Z_0 = l_0)$ equals the conditional probability of $(X_{l_0+m} \neq j, 0 < m < l_1; X_{l_0+l_1} = j)$, given $(X_0 = i; X_m \neq j, 0 < m < l_0; X_{l_0} = j)$ which by Markov property at time l_0 equals

$$P(X_m \neq j, 0 < m < l_1; X_{l_1} = j \,|\, X_0 = j) = P_j(T_1 = l_1) \,.$$

Next, $P_i(Z_2 = l_2 \,|\, Z_0 = l_0, Z_1 = l_1)$ equals the conditional probability of the event A given the event B, where

$$A = \{X_{l_0+l_1+m} \neq j, 0 < m < l_2, X_{l_0+l_1+l_2} = j\},$$

and

$$B = \left\{ \begin{array}{l} X_0 = i, X_{l_0} = X_{l_0+l_1} = j, X_m \neq j, 0 < m < l_0, \\ X_{l_0+m} \neq j, 0 < m < l_1 \end{array} \right\} .$$

Once again, by the Markov property at time $l_0 + l_1$, the above conditional probability equals

$$P(X_m \neq j, 0 < m < l_2; X_{l_2} = j \,|\, X_0 = j) = P_j(T_1 = l_2)\,.$$

One can use similar technique to show that for any $r \geq 1$,

$$P_i(Z_{r+1} = l_{r+1} \,|\, Z_0 = l_0, \ldots, Z_r = l_r) = P_j(T_1 = l_{r+1})\,.$$

From this, our claim about the P_i distribution of the sequence $(Z_r)_{r\geq 1}$ can easily be proved.

Now an application of the strong law of large numbers yields

$$\frac{Z_1 + \cdots + Z_r}{r} \longrightarrow E_j(T_1) \quad \text{as } r \to \infty$$

with P_i-probability one, for any i. It should be remarked that the strong law of large numbers used here does not need any apriori assumption on finiteness of the expectation $E_j(T_1)$. This is because the random variables Z_i are all non-negative. (See the paragraph following Exercise 5 in Section 0.5.) By the definition of the sequence $Z_1, Z_2, \ldots$, we have

$$Z_1 + \cdots + Z_r = T_{r+1} - T_1\,.$$

Thus

$$\frac{T_{r+1} - T_1}{r} \longrightarrow E_j(T_1) \quad \text{as } r \to \infty,$$

with P_i-probability one. But then

$$\frac{T_r - T_1}{r} = \frac{T_r - T_1}{r-1}\,\frac{r-1}{r} \longrightarrow E_j(T_1) \quad \text{as } r \to \infty,$$

with P_i-probability one. Since $P_i(T_1 < \infty) = 1$ and hence $\frac{T_1}{r} \longrightarrow 0$ as $r \to \infty$, we have with P_i-probability one

$$\frac{T_{r+1}}{r} \longrightarrow E_j(T_1) \quad \text{as well as} \quad \frac{T_r}{r} \longrightarrow E_j(T_1) \quad \text{as } r \to \infty\,.$$

For each $n \geq 1$, let us consider the random variable

$$N_n \quad = \quad \#\{1 \leq l \leq n : X_l = j\}\,.$$

Since we have an ergodic chain, $N_n \to \infty$ as $n \to \infty$ with P_i-probability one, for any i, so that

$$\frac{T_{N_n+1}}{N_n} \longrightarrow E_j(T_1) \quad \text{and} \quad \frac{T_{N_n}}{N_n} \longrightarrow E_j(T_1) \quad \text{as} \quad n \to \infty\,.$$

But by definition of N_n, one clearly has $T_{N_n} \leq n < T_{N_n+1}$ so that

$$\frac{T_{N_n}}{N_n} \quad \leq \quad \frac{n}{N_n} \quad \leq \quad \frac{T_{N_n+1}}{N_n}\,.$$

Thus we have, for every i, $\dfrac{n}{N_n} \to E_j(T_1)$ as $n \to \infty$ or, equivalently,

$$\frac{N_n}{n} \quad \longrightarrow \quad \frac{1}{E_j(T_1)} \quad \text{as} \quad n \to \infty,$$

with P_i-probability one. Since $0 \leq \frac{N_n}{n} \leq 1$ for all n, we have by the dominated convergence theorem that

$$\frac{1}{n} E_i(N_n) \quad \longrightarrow \quad \frac{1}{E_j(T_1)} \quad \text{as} \quad n \to \infty\,.$$

It is easy to identify $E_i(N_n)$ as $\sum_{l=1}^{n} p_{ij}^{(l)}$. This proves that $\lim_{n\to\infty} \frac{1}{n} \sum_{l=1}^{n} p_{ij}^{(l)}$ exists for all i and j, and equals $1/E_j(T_1)$. Denote this quantity by π_j. It is clear that $\pi_j \geq 0$ for each j. Of course, $\pi_j = 0$ is not yet ruled out but will be ruled out soon. Also, since each P^n is a stochastic matrix of finite order one gets $\sum \pi_j = 1$. Thus $\pi = (\pi_j : j \in S)$ is a probability on the state space. We next show that $\pi P = \pi$, or in other words, $(\pi P)_j = \pi_j$ for each j. Fix any arbitrary state k.

$$\begin{aligned}(\pi P)_j &= \sum_i \pi_i p_{ij} = \sum_i \lim_n \frac{1}{n} \sum_{l=1}^{n} p_{ki}^{(l)}\, p_{ij} = \lim_n \frac{1}{n} \sum_{l=1}^{n} \sum_i p_{ki}^{(l)} p_{ij} \\ &= \lim_n \frac{1}{n} \sum_{l=1}^{n} p_{kj}^{(l+1)} = \lim_n \frac{1}{n} \left[\sum_{l=1}^{n} p_{kj}^{(l)} - p_{kj}^{(1)} + p_{kj}^{(n+1)} \right] = \pi_j\,.\end{aligned}$$

This shows that our π is indeed an invariant distribution. To show uniqueness, let $\widetilde{\pi} = (\widetilde{\pi}_j)$ be any invariant distribution. From $\widetilde{\pi} P^n = \widetilde{\pi}$, one easily gets that for each j,

$$\frac{1}{n} \sum_{l=1}^{n} \sum_i \widetilde{\pi}_i p_{ij}^{(l)} \quad = \quad \widetilde{\pi}_j\,.$$

Letting $n \to \infty$, the left-hand side equals $\sum_i \widetilde{\pi}_i \pi_j = \pi_j$ showing that $\widetilde{\pi} = \pi$. It is only appropriate to draw the attention of the reader to an important fact lest it be overlooked. In our analysis above, we have repeatedly taken the liberty of interchanging sum and limits at our will. This was sponsored by the assumption of finiteness of the state space. The case of infinite state space could be a very different ball game.

Thus we have proved that for an ergodic finite state Markov chain, there is a unique invariant distribution π which is also the limiting distribution in the sense of (30). Indeed our proof also shows that for each j, $1/\pi_j$ is nothing but the expected time to return to j, given that the chain started at j. As noted already, this expected value could potentially be infinite for some j, leading to $\pi_j = 0$.

We now go a step further and show that for ergodic finite state Markov chains, $\pi_j > 0$ for all j which, in turn, would also imply that, starting from any

state j, the expected time to return is finite. Indeed, suppose that for some j, $\pi_j = 0$. Fix any $i \neq j$ and an $l \geq 1$ such that $p_{ij}^{(l)} > 0$. Since $\pi P^l = \pi$, we have $\pi_j = \sum_k \pi_k p_{kj}^{(l)} \geq \pi_i p_{ij}^{(l)}$, so that $\pi_i = 0$. Thus $\pi_j = 0$ for some j implies that $\pi_i = 0$ for all i which contradicts $\sum \pi_i = 1$.

For an irreducible recurrent chain we already knew that starting from a state j, we are sure to return to j sometime or the other. What we have just shown is that if the state space is moreover finite then the expected time to return is also finite. This property is often referred to in the literature as *positive recurrence.* This is not true in general, that is, a recurrent state may fail to be positive recurrent, if the state space is infinite. Such recurrent states are called *null recurrent.*

Another natural question that arises out of (30) is : why do we not consider simply the $\lim_n p_{ij}^{(n)}$ instead of the averages $\frac{1}{n}\sum p_{ij}^{(l)}$ as was done above? It is not difficult to see that $\lim_n p_{ij}^{(n)}$ may fail to exist, in general. In fact, a two state chain with transition matrix

$$P = \begin{pmatrix} 0 & 1 \\ 1 & 0 \end{pmatrix}$$

will illustrate this. What is happening in this example is that, for any i and j, exactly one of $p_{ij}^{(n)}$ and $p_{ij}^{(n+1)}$ is positive for each n. In fact, for $i = j$, $p_{ij}^{(n)}$ is positive (indeed, equals 1) if and only if n is even, while for $i \neq j$, this happens if and only if n is odd.

Usually, it is only such periodic behaviour of the chain, as illustrated in the example above, that prevents the existence of $\lim_n p_{ij}^{(n)}$. We are not going to pursue the periodicity properties and their consequences here. However, for subsequent applications, we are going to describe now (without proofs) what happens if such periodic behaviour is ruled out.

For a Markov chain, a state j is said to be *aperiodic* if $\{n \geq 1 : p_{jj}^{(n)} > 0\}$ has greatest common divisor 1. It is immediate that none of the two states in the above example are aperiodic — the g.c.d is 2 for both. It can be shown that in an irreducible Markov chain, either all states are aperiodic or none are and, in the first case, the chain is said to be *aperiodic.* Now we can state the main result without proof.

Theorem: *If an ergodic finite state chain is aperiodic, then for all states i and j, the limit $\lim_{n\to\infty} p_{ij}^{(n)}$ exists and equals π_j where $\pi = (\pi_j, j \in S)$ is the unique invariant distribution.*

In effect what it says is that for an aperiodic ergodic chain, π is the limiting distribution of the chain, irrespective of how it starts.

Exercise 8. Consider a Markov chain with r states. Suppose that the transition matrix has the property that each column sum is one (remember that for a transition matrix each row sum is one). If the chain is irreducible then

show that the uniform distribution on the state space is the unique stationary distribution. What do you infer about the expected times to return in this case? What if it is not irreducible?

Exercise 9. Let a be a probability vector with strictly positive entries and length 10. Consider a Markov chain with 10 states. Let the transition matrix have identical rows, each row being a. What is the stationary distribution? What chain are we talking about? What if the vector is not strictly positive?

Exercise 10. Consider a chain with 4 states and the following transition matrix.

$$\begin{pmatrix} 1/2 & 1/2 & 0 & 0 \\ 1/3 & 1/3 & 1/3 & 0 \\ 1/4 & 1/4 & 1/4 & 1/4 \\ 0 & 0 & 1/2 & 1/2 \end{pmatrix}$$

Denoting by τ the time (≥ 1) of the first visit to the state 2, calculate $E_i(\tau)$ for each state i. Suppose that μ is an initial distribution on the state space. Calculate $E_\mu(\tau)$.

0.8.5 Absorbing Chains: Limiting Behaviour, Rate of Convergence

Recall that an absorbing chain is a finite state Markov chain consisting only of absorbing and transient states. Since the state space is finite, there is at least one absorbing state. To avoid trivialities, we assume that there is at least one transient state also. Specifically, let us assume that there are m states, $\{1, 2, \ldots, m\}$, of which the first k are absorbing and the remaining transient. As already seen, the transition matrix has the structure

$$P = \begin{pmatrix} I & O \\ R & Q \end{pmatrix},$$

where I is the identity matrix of order k and R and Q are of orders $(m-k) \times k$ and $(m-k) \times (m-k)$ respectively. As already observed, the fundamental matrix $N = (I - Q)^{-1}$ plays an important role. For example, the matrix NR equals $((\alpha_{ij}))$ where α_{ij} for $k+1 \leq i \leq m$ and $1 \leq j \leq k$ are the absorption probabilities. For an absorbing state j, it follows from the continuity property of probability that

$$\alpha_{ij} = \lim_{n \to \infty} P_i(X_n = j) = \lim_n p_{ij}^{(n)}. \tag{31}$$

Recall that, for $k+1 \leq j \leq m$, $\lim_n p_{ij}^{(n)} = 0$ for all i. All of these can be stated in matrix form as

$$P^n \longrightarrow \begin{pmatrix} I & O \\ A & O \end{pmatrix}$$

where $A = NR = ((\alpha_{ij}))$.

We now want to show that the convergence in (31) happens geometrically fast and also calculate the exact rate of convergence. Interest in the rate of convergence lies in the fact that just like the expected time till absorption, this rate also gives another indication as to how fast the chain gets trapped in one of the absorbing states.

To begin with, let us recall that a number λ (possibly complex) is called an *eigenvalue* of P if there is a non-null vector $\widetilde{u}$ (with possibly complex entries) such that $\widetilde{u}P = \lambda\widetilde{u}$. Such a non-null vector $\widetilde{u}$ is called a *left eigenvector* corresponding to the eigenvalue λ. Recall also that the set of all vectors $\widetilde{u}$ with $\widetilde{u}P = \lambda\widetilde{u}$ is a vector space, called the *left eigenspace* associated to λ.

Let us now observe that a transition matrix P cannot have an eigenvalue λ with $|\lambda| > 1$. If possible, suppose $|\lambda| > 1$ and $\widetilde{u} = (u_1, u_2, \ldots, u_m)$ is a non-null vector with $\widetilde{u}P = \lambda\widetilde{u}$. Then clearly, $\widetilde{u}P^n = \lambda^n\widetilde{u}$ for all $n \geq 1$. Let j be such that $u_j \neq 0$. We then get a contradiction from the fact that

$$\sum_i u_i p_{ij}^{(n)} = \lambda^n u_j \,,$$

where the left-hand-side remains bounded by $\sum |u_i|$ for all n, while the right-hand-side is unbounded.

We next show that if P is the transition matrix of an absorbing chain with k absorbing states, then the dimension of the eigenspace associated to $\lambda = 1$ is exactly k. Indeed, let $\widetilde{u}$ be any vector with $\widetilde{u}P = \widetilde{u}$ which, of course, implies $\widetilde{u}P^n = \widetilde{u}$ for all n. Then, for any $j \geq k+1$, $u_j = \sum u_i p_{ij}^{(n)} \to 0$ as $n \to \infty$ showing that $u_{k+1} = \cdots = u_m = 0$. Thus, the dimension of the eigenspace is at most k. On the other hand, for each i, $1 \leq i \leq k$, the vector $\widetilde{u}^i$ with i-th coordinate equal to 1 and other coordinates 0, can easily seen to be a left eigenvector corresponding to $\lambda = 1$. So there are exactly k linearly independent left eigenvectors for $\lambda = 1$.

Finally, for P as above, we show that $\lambda = -1$ cannot be an eigenvalue. Suppose that $\widetilde{u}$ satisfies $\widetilde{u}P = -\widetilde{u}$ and hence $\widetilde{u}P^n = (-1)^n\widetilde{u}$ for all n. For $j \geq k+1$, $(-1)^n u_j = \sum u_i p_{ij}^{(n)}$ again yields that $u_j = 0$. For $j \leq k$,

$$-u_j = \sum_{i=1}^{m} u_i p_{ij} = \sum_{i=1}^{k} u_i p_{ij} = u_j$$

implying again that $u_j = 0$. Thus any $\widetilde{u}$ satisfying $\widetilde{u}P = -\widetilde{u}$ must be null.

To continue with our discussion of the rate of convergence, let us assume that there are m real eigenvalues $\lambda_1, \ldots, \lambda_m$ (not necessarily distinct) of P, with associated left eigenvectors $\widetilde{u}^1, \ldots, \widetilde{u}^m$, which are linearly independent. As shown earlier, we can and do take $\lambda_1 = \lambda_2 = \cdots = \lambda_k = 1$ and $\widetilde{u}^i$, $1 \leq i \leq k$, as defined above. If the remaining eigenvalues are listed in decreasing order of magnitude, we will clearly get

$$1 = \lambda_1 = \cdots = \lambda_k > |\lambda_{k+1}| \geq |\lambda_{k+2}| \geq \cdots \geq |\lambda_m| \,.$$

Denote by L the $m \times m$ matrix whose i-th row is $\widetilde{u}^i$. The above equations can be reformulated as

$$LP = DL$$

where $D = \text{Diag}(\lambda_1, \ldots, \lambda_m)$. Since the vectors $\widetilde{u}^i$ are linearly independent, the matrix L is invertible and therefore

$$P = L^{-1} D L. \tag{32}$$

Readers initiated to linear algebra would quickly recognize the above as the *spectral representation* of P. Our assumption therefore really amounts to P having a spectral representation. It follows from (32) that

$$P^n = L^{-1} D^n L,$$

where clearly $D^n = \text{Diag}(\lambda_1^n, \ldots, \lambda_m^n)$. In particular, for any $1 \leq i, j \leq m$,

$$p_{ij}^{(n)} = \sum_{r=1}^{m} \lambda_r^n (L^{-1})_{ir} L_{rj}.$$

Since $\lambda_1 = \cdots = \lambda_k = 1$, we have

$$\begin{aligned} |p_{ij}^{(n)} - \sum_{r=1}^{k} (L^{-1})_{ir} L_{rj}| &\leq \sum_{r=k+1}^{m} |\lambda_r|^n |(L^{-1})_{ir} L_{rj}| \\ &\leq |\lambda_{k+1}|^n \sum_{r=k+1}^{m} |(L^{-1})_{ir} L_{rj}|. \end{aligned}$$

Of course, $|\lambda_{k+1}| < 1$ implies that the left side goes to zero as $n \to \infty$ and the convergence is geometrically fast with rate not larger than $|\lambda_{k+1}|$. Incidentally this argument also shows that

$$\sum_{r=1}^{k} (L^{-1})_{ir} L_{rj} = \lim_{n \to \infty} p_{ij}^{(n)}$$

for all i and j. Of course, if i is a transient state (that is, $i \geq k+1$) and j is an absorbing state (that is, $j \leq k$), then this quantity is precisely α_{ij}, the probability of absorption in j starting from i. We leave it as an exercise to verify that (i) if i is absorbing, then $\sum_{r=1}^{k} (L^{-1})_{ir} L_{rj}$ equals δ_{ij} and (ii) if j is transient then this is zero.

Exercise 11. In the above discussion of convergence rate for absorbing chain, we assumed spectral representation for the transition matrix. However, a transition matrix need not always admit a spectral representation (32). Show that the following transition matrices do not admit spectral representation. For chains with these transition matrices, find the rates of convergence.

$$P = \begin{pmatrix} 1 & 0 & 0 & 0 \\ 2/3 & 0 & 1/3 & 0 \\ 2/3 & 0 & 0 & 1/3 \\ 2/3 & 1/3 & 0 & 0 \end{pmatrix}, \qquad P = \begin{pmatrix} 1 & 0 & 0 & 0 \\ 1/2 & 0 & 1/2 & 0 \\ 1/2 & 0 & 0 & 1/2 \\ 2/3 & 1/3 & 0 & 0 \end{pmatrix}.$$

0.9 Continuous Time Processes

Discrete time stochastic processes are used to describe evolution of systems that change only at discrete instants of time. The relevant time set is the set of time points at which changes may take place, and, is usually taken as $\{0, 1, 2, \ldots\}$. In the earlier two sections, we discussed some special types of such processes, namely, martingales and Markov chains.

In this section, we discuss stochastic processes evolving over a continuum of time or in other words, *continuous time stochastic processes.* Even though the process evolves over a continuous time, distinction would be made as to the nature of the evolution. Let us consider two simple examples to make the distinction clear.

Imagine a telephone exchange through which calls pass at random instants of time. If we consider the number of calls passing through upto time t, then we have a stochastic process (X_t) over time set $t \in [0, \infty)$. However, the state space of the process is $\{0, 1, 2, \ldots\}$, which is a discrete set and the process evolves only through jumps.

A different example would be the kinetic movement of a gas molecule where the position of the particle changes continuously with time and not through jumps. In other words, here the state space is also a continuum.

In the first subsection, we will discuss a special class of processes of the first type, namely *Markov chains in continuous time.* Such processes will be used in connection with temporal spread of epidemics in Chapter 4.

The second subsection would be devoted to a special class of processes of the second type — known as *diffusion processes.* It is worth noting here that a diffusion process may sometimes serve also as an approximation to a discrete time Markov chain and often allows us to get good approximations to quantities of interest, related to the original discrete chain. Indeed, it is mainly this application of diffusion processes which will be used in connection with Markov models in genetics in Chapter 3.

The interval $[0, \infty)$ is usually taken as the time set for a continuous time process. Thus, we consider a family of random variables X_t, indexed by all real numbers $t \geq 0$, each taking values in a set S. This constitutes a continuous time process and, in analogy with discrete time processes, is denoted $(X_t)_{t \geq 0}$. The set S is called the *state space* of the process. The notion of Markov property for stochastic processes has been already encountered in the discrete set-up. It simply means that at any point of time, given the present state of the process, the future evolution is (conditionally) independent of the history of the past. A simple formulation of this idea in the continuous time case is as follows.

Definition: A process $(X_t)_{t \geq 0}$ with state space S is said to be a *Markov process* if, for any choice $0 \leq s_1 < s_2 < \cdots < s_n < s < t + s$ of time points, the conditional distribution of the random variable X_{t+s}, given $(X_{s_1}, \ldots, X_{s_n}, X_s)$,

depends only on X_s, that is, for $B \subset S$,

$$P(X_{t+s} \in B \,|\, X_{s_1} = x_1, \ldots, X_{s_n} = x_n, X_s = x) = P(X_{t+s} \in B \,|\, X_s = x)\,.$$

If, furthermore, these conditional distributions are the same for all s, that is, the right side of the above equation depends only on t and not on s, then the process $(X_t)_{t\geq 0}$ is said to be *time-homogeneous*.

In the above definition, the set B can be any subset of S in case S is a countable set. However, as discussed in Section 0.4, one has to be more selective, in case S is not countable. Of course, for almost any conceivable B, the above property has to hold.

We will consider here only time-homogeneous Markov processes. Thus, for all $t > 0$ and $s \geq 0$, we have

$$P(X_{t+s} \in B \,|\, X_s = x) = P(X_t \in B \,|\, X_0 = x)\,.$$

Let us denote this by $P_t(x, B)$. Clearly, for each $t \geq 0$ and each point x in the state space S, $P_t(x, \cdot)$ is a probability distribution on S. This family of distributions, as t and x vary — called the family of *transition probabilities* — play the same role as that of the transition matrix and its powers for a Markov chain in capturing the evolutionary mechanism of the whole process $(X_t)_{t\geq 0}$.

Just like in the case of discrete Markov chains, the family of transition probabilities $P_t(x, \cdot)$ here also satisfy the Chapman-Kolmogorov equations, which now reads as

$$P_{t+s}(x, B) = \int P_s(y, B) P_t(x, dy) \quad \text{for all } t \geq 0, s \geq 0\,.$$

The interpretation of the integral is not difficult. It is simply a notation for $E(P_s(X_t, B) \,|\, X_0 = x)$. We will return to this in the next two subsections and see that the equations take on simpler forms under special assumptions.

0.9.1 Continuous Time Markov Chains

In this section, we assume that the state space is countable, that is, each X_t is a discrete random variable taking values in a countable set S. The time-homogeneous Markov property reduces to

$$P(X_{t+s} = j \,|\, X_{s_1} = i_1, \ldots, X_{s_n} = i_n, X_s = i) = P_{ij}(t) = P(X_t = j \,|\, X_0 = i)$$

for all $0 \leq s_1 < s_2 < \cdots < s_n < s < t + s$ and $i, j \in S$. It can be shown that the above equation actually implies that for any $s > 0$, the conditional distribution of $(X_{t+s})_{t\geq 0}$, given $(X_u)_{u<s}$ and $X_s = i$, is the same as that of $(X_t)_{t\geq 0}$, given $X_0 = i$. In particular,

$$P(X_{t+s} = j \,|\, X_u, u < s; X_s = i) = P_{ij}(t)\,.$$

If $P(t)$ denotes the $S \times S$ matrix whose (i, j)-th entry is $P_{ij}(t)$, then each $P(t)$ is clearly a stochastic matrix. Thus we have a family $\{P(t), t \geq 0\}$ of stochastic

matrices. Here $P(0)$ is the identity matrix of size S. The Chapman-Kolmogorov equations are easily seen to correspond to the *semigroup* property

$$P(t+s) = P(t) \cdot P(s) .$$

As mentioned earlier, the family $(P(t))_{t\geq 0}$ plays the same role as the sequence $(P^n)_{n\geq 0}$ of the n-step transition matrices in case of discrete Markov chains. The notable difference is that while the P^n are all determined by the one-step transition matrix P, it is not clear how to get one single matrix that will determine all the $P(t)$ for $t \geq 0$. We are now going to show how to do this.

One may recall that if $P : [0,\infty) \longrightarrow R$ is a continuous function with $P(t+s) = P(t) \cdot P(s)$ and $P(0) = 1$, then $P'(0)$ exists and determines $P(t)$ for all values of t. Indeed if $P'(0) = Q$, then $P(t) = e^{Qt}$ for all t. In particular $P'(t) = P(t) \cdot Q = Q \cdot P(t)$ for all t. Indeed this differential equation along with the initial condition $P(0) = 1$ also characterizes the function $P(t) = e^{Qt}$.

Our present situation is quite similar to this except that, instead of real-valued functions, we are dealing with an $S \times S$ matrix-valued function $P(t)$. We want to show that under certain conditions, $P(t)$ also satisfies the matrix differential equations

$$P'(t) = Q \cdot P(t) = P(t) \cdot Q$$

for some matrix $Q = (q_{ij})$. In other words, we have the following two systems of equations

$$P'_{ij}(t) = \sum_k q_{ik} P_{kj}(t) \qquad i,j \in S \tag{33}$$

$$P'_{ij}(t) = \sum_k P_{ik}(t) q_{kj} \qquad i,j \in S \tag{34}$$

Unlike in the real-valued case, the two systems are not identical. System (33) is always true and is known as Kolmogorov's *Backward Equations*. System (34) which is true under some additional regularity conditions, is known as Kolmogorov's *Forward Equations*, also known as *Fokker-Planck* equations. We proceed to give a derivation of the above equations, assuming that the state space is finite. Indeed, it is only in the proof of the forward equations that the finiteness of the state space will be used. Our derivation will also identify the matrix Q, frequently known as the *Q-matrix* of the chain.

We first prove two basic lemmas which will give us a description of how the chain evolves with time. Let

$$T = \inf\{t > 0 : X_t \neq X_0\} .$$

In other words, T is the first time the system leaves the initial state.

Lemma 1: *For any* $s, t \geq 0$,

$$P(T > t+s \,|\, X_0 = i) = P(T > t \,|\, X_0 = i) P(T > s \,|\, X_0 = i) .$$

Proof: First assume that $s > 0$ and $t > 0$.

$$\begin{aligned} P(T > t+s \,|\, X_0 = i) &= P(T > t+s, T > s \,|\, X_0 = i) \\ &= P(T > t+s, X_s = i, T > s \,|\, X_0 = i) \\ &= P(T > s, X_s = i \,|\, X_0 = i) \\ &\quad \times P(T > t+s \,|\, X_0 = i, T > s, X_s = i) \\ &= P(T > s \,|\, X_0 = i) \cdot P(T > t \,|\, X_0 = i)\,, \end{aligned}$$

where the equality $P(T > t+s \,|\, X_0 = i, T > s, X_s = i) = P(T > t \,|\, X_0 = i)$ follows from the assumed Markov property. The case $s = 0$ and/or $t = 0$ follows by taking limits. ∎

A consequence of the above is that, for any $i \in S$, there is a $\lambda_i \in [0, \infty]$ such that, $P(T > t \,|\, X_0 = i) = e^{-\lambda_i t}$ for all $t \geq 0$. In particular, $P(T > 0 \,|\, X_0 = i)$ is either one or zero (according as λ_i is finite or not). Also, the case $\lambda_i = 0$ corresponds to $P(T = \infty \,|\, X_0 = i) = 1$. Clearly, $0 < \lambda_i < \infty$ refers to an exponential distribution as encountered in Section 0.4. However, we agree here to use the term exponential distribution in a broad sense even when λ_i equals 0 or ∞.

Lemma 2: *For any i, j with $i \neq j$ and any $s > 0$,*

$$P(T > s, X_T = j \,|\, X_0 = i) = P(T > s \,|\, X_0 = i) P(X_T = j \,|\, X_0 = i)\,.$$

Proof: This is clearly true if $P(T > 0 \,|\, X_0 = i) = 0$. We assume therefore that $P(T > 0 \,|\, X_0 = i) = 1$.

$$\begin{aligned} P(T > s, X_T = j \,|\, X_0 = i) &= P(T > s, X_T = j, X_s = i \,|\, X_0 = i) \\ &= P(X_s = i, T > s \,|\, X_0 = i) \times P(X_T = j \,|\, X_0 = i, X_s = i, T > s) \\ &= P(T > s \,|\, X_0 = i) P(X_T = j \,|\, X_0 = i, T > 0) \\ &= P(T > s \,|\, X_0 = i) P(X_T = j \,|\, X_0 = i)\,. \end{aligned}$$

∎

The content of the two lemmas is the following. For every state i, there is a number $\lambda_i \in [0, \infty]$ and transition probabilities p_{ij} for $j \neq i$. If the chain starts in the state i, it remains there for an exponentially distributed random time T_1 with mean $1/\lambda_i$ and then moves to state j with probability p_{ij}, independently of T_1. Subsequently, the chain behaves as if started from state j. It may be pointed out that $\lambda_i = \infty$ corresponds to $P(T_1 = 0 \,|\, X_0 = i) = 1$, meaning that the chain instantaneously jumps from the state i. Such states are called *instantaneous states*. It can be shown that this contingency is not possible in a finite state chain. In general, we assume that there are no such states. It may also be pointed out that $\lambda_i = 0$ corresponds to $P(T_1 = \infty \,|\, X_0 = i) = 1$, meaning that the chain starting at i remains there forever. That is, i is *absorbing*. Thus only $\lambda_i > 0$ corresponds to the case when the waiting time in state i is a proper exponential random variable. In any case, from the above description it is clear that the evolution of the chain is completely captured

by the parameters $(\lambda_i, i \in S)$ and the stochastic matrix $((p_{ij}))_{i,j\in S}$ with zero diagonal entries.

From the above description, it should also be clear that if $T_1, T_2, \ldots$ represent the successive (random) times of jumps of the chain then the sequence of random variables defined as

$$Y_0 = X_0, \quad Y_n = X_{T_n} \quad \text{for } n \geq 1$$

would form a discrete time Markov chain with state space S. The one-step transition probabilities of the Markov chain are given by p_{ij}, if $\lambda_i > 0$. For i such that $\lambda_i = 0$, we have $p_{ii} = 1$.

The chain $(Y_n)_{n\geq 0}$ is usually called the *embedded chain.* For many of the properties of the continuous time chain, like classification of states, asymptotic behaviour and existence of invariant distributions, it suffices to examine only the embedded chain. Of course, some important features that explicitly make use of the waiting times at various states would not be captured by the embedded chain. For more on embedded chains, the reader may look at the book of Bhattacharya and Waymire.

We now proceed towards proving Kolmogorov's backward equations (33). Let $i \in S$ be such that $\lambda_i > 0$. Then for any $j \in S$ and any $t > 0$, we have by conditioning on the time of the first jump from i,

$$\begin{aligned} P_{ij}(t) &= \sum_{k\neq i} \int_0^t \lambda_i e^{-\lambda_i s} p_{ik} P_{kj}(t-s)ds + e^{-\lambda_i t}\delta_{ij} \\ &= \sum_{k\neq i} e^{-\lambda_i t} p_{ik} \int_0^t \lambda_i e^{\lambda_i u} P_{kj}(u)du + e^{-\lambda_i t}\delta_{ij}\,. \end{aligned}$$

Note that, in case $j = i$, the process starting from i may be in state $j\,(= i)$ at time t by simply waiting at the initial state at least till time t. The term $e^{-\lambda_i t}\delta_{ij}$ occurs to take care of this contingency. Of course, for $j \neq i$, this contingecy does not arise and therefore the term has no contribution. Here δ_{ij} is the usual Kronecker delta, that is, δ_{ij} equals 1 or 0 according as $i = j$ or $i \neq j$. The above equation shows that $P_{ij}(t)$ is continuous in t. In case the state space is finite, this is immediate because each summand is continuous in t. In general, one needs to use the Dominated Convergence Theorem. The continuity of $P_{ij}(t)$, in turn, gives differentiability also and indeed, the sum on the right side can be differentiated term by term. This requires the fundamental theorem of calculus as well as the Dominated Convergence Theorem. The upshot is

$$\begin{aligned} P'_{ij}(t) &= -\lambda_i \left(\sum_{k\neq i} e^{-\lambda_i t} p_{ik} \int_0^t \lambda_i e^{\lambda_i u} P_{kj}(u)du + e^{-\lambda_i t}\delta_{ij} \right) \\ &\qquad + \sum_{k\neq i} e^{-\lambda_i t} p_{ik} \lambda_i e^{\lambda_i t} P_{kj}(t) \\ &= -\lambda_i P_{ij}(t) + \sum_{k\neq i} \lambda_i p_{ik} P_{kj}(t)\,. \end{aligned}$$

In other words, denoting

$$q_{ik} = \lambda_i p_{ik} \quad \text{for} \quad k \neq i; \quad \text{and} \quad q_{ii} = -\lambda_i, \tag{35}$$

we get

$$P'_{ij}(t) = \sum_k q_{ik} P_{kj}(t)$$

Clearly the same equation also holds in case $\lambda_i = 0$ because in that case $P_{ij}(t) = \delta_{ij}$ so that $P'_{ij}(t) = 0$. Thus we have proved the backward equations (33) with q_{ij} for $i, j \in S$ defined by (35).

We now proceed to derive the forward equations (34). Let us first observe that a consequence of the Equations (33) is that

$$P'_{ij}(0) = q_{ij} \quad \text{for all } i, j \in S\,.$$

Of course the derivative at zero is only the derivative from the right, that is,

$$\lim_{h \downarrow 0} \frac{P_{ij}(h) - \delta_{ij}}{h} \quad = \quad q_{ij}\,. \tag{36}$$

By the Chapman-Kolmogorov equations $P_{ij}(t+h) = \sum_k P_{ik}(t) P_{kj}(h)$ so that,

$$\frac{P_{ij}(t+h) - P_{ij}(t)}{h} = \sum_{k \neq j} P_{ik}(t) \frac{P_{kj}(h)}{h} + P_{ij}(t) \frac{P_{jj}(h) - 1}{h}$$

Now letting $h \downarrow 0$ and using (36), one obtains the forward equations. It is in the last step — interchanging the limit and sum — that finiteness of the state space is used. It should be noted that because of the differentiability of $P_{ij}(t)$, the limit $\lim_{h \downarrow 0} \frac{P_{ij}(t+h) - P_{ij}(t)}{h}$ equals $P'_{ij}(t)$ for all $t > 0$.

The matrix $Q = ((q_{ij}))$ is often called the *infinitesimal matrix* or *rate matrix* or *Q-matrix* of the chain. This Q-matrix has the property that all the off-diagonal entries are non-negative and each row sum equals zero. Accordingly the diagonal entries must be non-positive and are determined by the off-diagonal entries. The equation (35) shows that the Q-matrix is determined by the parameters $(\lambda_i, i \in S)$ and $(p_{ij}, i, j \in S, i \neq j)$. What is more important is that the Q-matrix, in turn, determines these parameters. Indeed $\lambda_i = -q_{ii} = \sum_{j \neq i} q_{ij}$, and, for any i, j with $j \neq i$, $p_{ij} = -q_{ij}/q_{ii}$. Of course, if $q_{ii} = 0$, then clearly for each j, q_{ij} is also zero and the above ratio should be interpreted as zero. In a nutshell, the Q-matrix of a chain completely captures the evolution of the chain. The elements of the Q-matrix are often called the *transition rates*, not to be confused with transition probabilities.

A simple but important class of continuous time Markov chains are what are known as *Birth and Death* chains. The state space is $\{0, 1, 2, \ldots\}$. The transition rates are given as follows:

$$q_{i,j} = 0 \quad \text{for all } i, j \text{ with } |i - j| > 1;$$

$$q_{0,1} = b_0; \qquad q_{i,i+1} = b_i, \quad q_{i,i-1} = d_i \quad \text{for } i \geq 1 .$$

It is clear that $\lambda_i = b_i + d_i$, so that the chain starting at i, waits there for an exponential time with mean $1/(b_i + d_i)$, at the end of which it jumps to $i-1$ or $i+1$ with probabilities $d_i/(b_i + d_i)$ and $b_i/(b_i + d_i)$ respectively. If we think of i as population size, then a jump to $(i-1)$ can be treated as death whereas a jump to $(i+1)$ can be regarded as birth. So the population evolves only through a death or a birth. Obviously from size 0, there can only be a birth. The parameters b_i (respectively, d_i) are called the *birth rates* (respectively, *death rates*). The Kolmogorov equations take on a simple form and are often not too difficult to solve. For example, the forward equations will now read

$$P'_{ij}(t) = b_{j-1}P_{i,j-1}(t) + d_{j+1}P_{i,j+1}(t) - (b_j + d_j)P_{ij}(t) .$$

If furthermore $b_i = 0$ for all i, that is, there are no births, the underlying chain is called a *pure death chain.* Clearly, 0 would always be an absorbing state for such a chain. For some special forms of the birth and death rates, the reader may consult the book of Karlin.

0.9.2 Diffusion Processes

To simplify matters, we will assume that the state space of the process is a bounded interval I and, more importantly, that for each t and x, the distribution $P_t(x, \cdot)$ is absolutely continuous with density $p(t, x, \cdot)$. The probability density functions $p(t, x, \cdot)$ — known as the *transition densities* —are then easily seen to satisfy an equation similar to (24) of Section 0.8, namely, that for all $t, s > 0$,

$$p(t+s, x, y) \quad = \quad \int_I p(t, x, z)p(s, z, y)dz . \tag{37}$$

These are the *Chapman-Kolmogorov equations* for transition densities in the continuous time case.

Suppose now that we have a process $(X_t)_{t\geq 0}$ that satisfies, in addition to the above, the following properties:

$$E(X_{t+h} - X_t \,|\, X_t = x) \quad = \quad a(x)h + o(h), \tag{38}$$

$$E(|X_{t+h} - X_t|^2 \,|\, X_t = x) \quad = \quad b(x)h + o(h), \tag{39}$$

$$E(|X_{t+h} - X_t|^k \,|\, X_t = x) \quad = \quad o(h), \qquad \text{for } k \geq 3 . \tag{40}$$

Recall that a function $g(h)$ is said to be of *smaller order* than h, written $o(h)$, if $g(h)/h \to 0$ as $h \to 0$. For subsequent use, let us also recall that $g(h)$ is said to be *at most of the order of* h, written $O(h)$, if $g(h)/h$ remains bounded as $h \to 0$. In both places we are only considering the behaviour near zero.

That the left sides of the equations (38) through (40) are independent of t is, of course, a consequence of the time-homogeneity property. Here $a(\cdot)$ and $b(\cdot)$ are two functions on the state space I and are known as the *drift coefficient*

and *diffusion coefficient* respectively. The equations (38)–(40) can equivalently be expressed in terms of the transition densities as:

$$\int (y-x)p(h,x,y)dy = a(x)h + o(h), \tag{41}$$

$$\int |y-x|^2 p(h,x,y)dy = b(x)h + o(h), \tag{42}$$

$$\int |y-x|^k p(h,x,y)dy = o(h), \quad \text{for } k \geq 3\,. \tag{43}$$

Definition: By a *diffusion process*, we will simply mean a time homogeneous Markov process $(X_t)_{t\geq 0}$ with transtion density $p(t,x,y)$ that satisfies the properties (37) and (41)–(43).

A substantial and mathematically deep theory of diffusion processes exists. See, for example, the book of Bhattacharya and Waymire. One of the major concerns of the theory is to show that, under suitable conditions on the functions $a(\cdot)$ and $b(\cdot)$, a unique diffusion process with required properties exists which, moreover, has nice additional features like, for example, having 'continuous paths'. Further, by imposing suitable conditions on $a(\cdot)$ and $b(\cdot)$, one can also ensure that the transition density of the resulting diffusion is sufficiently smooth in the state variables x and y. However, the mathematical depth of formal diffusion theory is inappropriate at this level, and also, high technical rigour is somewhat unnecessary for our present purposes. Accordingly, we choose not to get into the theory here. We will assume much of what we need and, instead, try to focus on how to apply it. In particular, we assume without question that a unique diffusion process with given drift and diffusion coefficients does exist and that its transtion density $p(t,x,y)$ is twice continuously differentiable in both the state variables x and y.

Before proceeding any further, let us also assume that the state space I of the diffusion process is the unit interval $[0,1]$. Now let g be any twice continuously differentiable function on $[0,1]$ with $g(0) = g(1) = g'(0) = g'(1) = 0$. Using (37) we have

$$\int g(z)p(t+h,x,z)\,dz = \int\int g(z)p(t,x,y)p(h,y,z)\,dy\,dz\,. \tag{44}$$

Using the Taylor expansion of g around y, namely,

$$g(z) = g(y) + (z-y)g'(y) + \frac{1}{2}(z-y)^2 g''(y) + O(|z-y|^3)$$

on the right side of (44), we get

$$\begin{aligned} \int g(y)p(t,x,y)\,dy &+ \int g'(y)[\int (z-y)p(h,y,z)\,dz]\,dy \\ &+ \tfrac{1}{2}\int g''(y)p(t,x,y)[\int (z-y)^2 p(h,y,z)\,dz]\,dy \\ &+ \int p(t,x,y)[\int O(|z-y|^3)p(h,y,z)\,dz]\,dy\,. \end{aligned}$$

Making use of (41)–(43), equation (44) can now be rewritten as

$$\int g(y)[p(t+h,x,y)-p(t,x,y)]\,dy$$
$$=\left[\int g'(y)p(t,x,y)a(y)\,dy+\frac{1}{2}\int g''(y)p(t,x,y)b(y)\,dy\right]h+o(h)\,.$$

Dividing both sides by h and taking limits as $h\downarrow 0$, we obtain

$$\int g(y)\frac{\partial}{\partial t}[p(t,x,y)]\,dy$$
$$=\int g'(y)a(y)p(t,x,y)\,dy+\frac{1}{2}\int g''(y)b(y)p(t,x,y)\,dy\,.$$

Applying integration by parts once on the first term of the right side and twice on the second term, and, using the assumed boundary conditions satisfied by g, we get

$$\int g(y)\frac{\partial}{\partial t}p(t,x,y)\,dy$$
$$=\int g(y)\{-\frac{\partial}{\partial y}(a(y)p(t,x,y))+\frac{1}{2}\frac{\partial^2}{\partial y^2}(b(y)p(t,x,y))\}\,dy\,.$$

Since this equation is valid for all functions g satisfying the assumed conditions, we must have

$$\frac{\partial}{\partial t}p(t,x,y)=-\frac{\partial}{\partial y}(a(y)p(t,x,y))+\frac{1}{2}\frac{\partial^2}{\partial y^2}(b(y)p(t,x,y))\,. \tag{45}$$

This partial differential equation (45) for the transition density function is known as the Kolmogorov's *Forward Equation* or the *Fokker-Planck Equation* and is of fundamental importance in diffusion theory and its applications. A similar equation, called the Kolmogorov's *Backward Equation* for the transition density, can be derived much more easily as follows.

From (37), we have

$$p(t+h,x,y)=\int p(h,x,z)p(t,z,y)\,dy \tag{46}$$

Using the Taylor expansion of $p(t,z,y)$ as a function of z around the point $z=x$, that is, the expansion

$$p(t,z,y)=p(t,x,y)+(z-x)\frac{\partial p(t,x,y)}{\partial x}+\frac{1}{2}(z-x)^2\frac{\partial^2 p(t,x,y)}{\partial x^2}+O(|z-x|^3)$$

on the right side of (46), we get

$$p(t+h,x,y)=p(t,x,y)+\frac{\partial p(t,x,y)}{\partial x}\int(z-x)p(h,x,z)\,dz$$
$$+\frac{1}{2}\frac{\partial^2 p(t,x,y)}{\partial x^2}\int(z-x)^2p(h,x,z)\,dz+\int O(|z-x|^3)p(h,x,z)\,dz\,.$$

Using properties (41)–(43) again, we obtain

$$p(t+h,x,y) - p(t,x,y) = \left\{ a(x)\frac{\partial p(t,x,y)}{\partial x} + \frac{1}{2}\frac{\partial^2 p(t,x,y)}{\partial x^2} \right\} h + o(h)\,.$$

Dividing both sides by h and taking limits as $h \downarrow 0$ leads finally to

$$\frac{\partial p(t,x,y)}{\partial t} = a(x)\frac{\partial p(t,x,y)}{\partial x} + \frac{1}{2}b(x)\frac{\partial^2 p(t,x,y)}{\partial x^2} \tag{47}$$

which is the so called backward equation and will be more useful in the sequel.

We now proceed to show some examples as to how the equation (47) can be used to evaluate certain quantities of interest related to the underlying diffusion. Let us consider, for example, the function

$$F(t,x,y) = \int_0^y p(t,x,z)\,dz, \qquad 0 < x < 1\,.$$

Clearly $F(t,x,y) = P(X_t \leq y \,|\, X_0 = x)$. It follows easily from (47) that the function F satisfies the differential equation

$$\frac{\partial F(t,x,y)}{\partial t} = a(x)\frac{\partial F(t,x,y)}{\partial x} + \frac{1}{2}b(x)\frac{\partial^2 F(t,x,y)}{\partial x^2}\,. \tag{48}$$

This, of course, involves several interchanges of differentiation and integration. But, as mentioned earlier, we will not worry about such technical issues. We will simply put it on record that they can all be justified with some work.

Suppose now that for the diffusion process under study, both the states 0 and 1 are absorbing states. For $i = 0, 1$, let $A_i(t,x)$ denote the probability that the diffusion process starting at state x gets absorbed in state i at or before time t. It is clear then that

$$A_0(t,x) = \lim_{y \downarrow 0} F(t,x,y) \quad \text{and} \quad A_1(t,x) = 1 - \lim_{y \uparrow 1} F(t,x,y)\,.$$

By passing to the limits in (65) as $y \downarrow 0$ or as $y \uparrow 1$ we obtain,

$$\frac{\partial A_i(t,x)}{\partial t} = a(x)\frac{\partial A_i(t,x)}{\partial x} + \frac{1}{2}b(x)\frac{\partial^2 A_i(t,x)}{\partial x^2}\,. \tag{49}$$

It should be noted that though both $A_0(t,x)$ and $A_1(t,x)$ satisfy the same partial differential equation, the solutions would be different (as they should be) because they satisfy different boundary conditions, namely, $A_0(t,0) = 1$ and $A_0(t,1) = 0$ whereas $A_1(t,0) = 0$ and $A_1(t,1) = 1$.

Let us denote by $A_i(x)$, for $i = 0, 1$, the probability that the process starting at the state x ever gets absorbed in the state i. Clearly

$$A_i(x) \quad = \quad \lim_{t \uparrow \infty} A_i(t,x)\,.$$

By a standard result of calculus, since $\lim_{t\uparrow\infty} A_i(t,x)$ exists, $\dfrac{\partial A_i(t,x)}{\partial t} \to 0$ as $t \to \infty$. It thus follows, by letting $t \to \infty$ in (49), that $A_i(x)$ satisfies the differential equation

$$a(x)\frac{dA_i(x)}{dx} + \frac{1}{2}b(x)\frac{d^2A_i(x)}{dx^2} = 0\,. \tag{50}$$

It should again be noted that, although $A_0(x)$ and $A_1(x)$ satisfy the same differential equation, the boundary conditions are different for the two. For $A_0(x)$, for example, the boundary conditions are $A_0(0) = 1$ and $A_0(1) = 0$. Using these, one can easily solve (50) explicitly to get

$$A_0(x) = \frac{\int_x^1 \psi(y)\,dy}{\int_0^1 \psi(y)\,dy}\,, \tag{51}$$

where

$$\psi(y) = \exp\left\{-2\int_0^y \frac{a(z)}{b(z)}\,dz\right\}. \tag{52}$$

Similarly, for $A_1(x)$, using the boundary conditions $A_1(0) = 0$ and $A_1(1) = 1$, one gets

$$A_1(x) = \frac{\int_0^x \psi(y)\,dy}{\int_0^1 \psi(y)\,dy}\,. \tag{53}$$

Of course, $A_1(x) = 1 - A_0(x)$, as it should be.

Having thus obtained simple formulae for the absorption probabilities, let us next turn to the time until absorption. Let τ denote the random variable representing the time until absorption. Let us write

$$A(t,x) = A_0(t,x) + A_1(t,x)$$

where $A_i(t,x)$ are as defined earlier. Then $A(t,x)$ also satisfies the same partial differential equation (49). Notice, however, that $A(t,x)$ is just the probability that $\tau \le t$ given $X_0 = x$; in other words, $A(t,x)$ is the probability distribution function (in t) of τ, conditional on the initial state being x. Suppose now that for each $x \in (0,1)$, this conditional distribution is absolutely continuous with density function $\varphi(t,x)$, $t \ge 0$. Since $A(t,x)$ satisfies the equation (49) we will then have

$$\varphi(t,x) = \frac{\partial A(t,x)}{\partial t} = a(x)\frac{\partial A(t,x)}{\partial x} + \frac{1}{2}b(x)\frac{\partial^2 A(t,x)}{\partial x^2},$$

so that

$$\varphi(t,x) = a(x)\frac{\partial}{\partial x}\left\{\int_0^t \varphi(s,x)\,ds\right\} + \frac{1}{2}b(x)\frac{\partial^2}{\partial x^2}\left\{\int_0^t \varphi(s,x)\,ds\right\}.$$

On differentiating with respect to t (and, of course, assuming again that integration with respect to s and differentiation with respect to x in the above equation can be interchanged) one obtains that

$$\frac{\partial\varphi(t,x)}{\partial t} = a(x)\frac{\partial\varphi(t,x)}{\partial x} + \frac{1}{2}b(x)\frac{\partial^2\varphi(t,x)}{\partial x^2}. \tag{54}$$

Suppose now that we are interested in the mean time till absorption, that is, in

$$T(x) = E(\tau \mid X_0 = x) = \int_0^\infty t\varphi(t,x)\,dt. \tag{55}$$

Let us assume that $t\varphi(t,x) \to 0$ as $t \to \infty$. One then has

$$1 = \int_0^\infty \varphi(t,x)\,dt = [t\varphi(t,x)]\Big|_{t=0}^{t=\infty} - \int_0^\infty t\frac{\partial\varphi(t,x)}{\partial t}\,dt = -\int_0^\infty t\frac{\partial\varphi(t,x)}{\partial t}\,dt.$$

Now using (54) we have

$$1 = -\int_0^\infty t\left\{a(x)\frac{\partial\varphi(t,x)}{\partial x} + \frac{1}{2}b(x)\frac{\partial^2\varphi(t,x)}{\partial x^2}\right\}dt.$$

Assuming once again that the t-integration and x-differentiation can be interchanged, one obtains $T(x)$ to satisfy the ordinary differential equation

$$a(x)\frac{dT(x)}{dx} + \frac{1}{2}b(x)\frac{d^2T(x)}{dx^2} = -1.$$

The obvious boundary conditions now are $T(0) = T(1) = 0$. Using the standard method of integrating factors, one obtains the solution to be

$$T(x) = -2\int_0^x \psi(z)\left(\int_0^z \frac{1}{b(y)\psi(y)}\,dy\right)dz + 2\frac{\int_0^x \psi(z)\,dz}{\int_0^1 \psi(z)\,dz}\int_0^1 \psi(z)\left(\int_0^z \frac{1}{b(y)\psi(y)}\,dy\right)dz, \tag{56}$$

or equivalently

$$T(x) = -2\int_0^x \frac{1}{b(y)\psi(y)}\left(\int_y^x \psi(z)\,dz\right)dy + 2\frac{\int_0^x \psi(z)\,dz}{\int_0^1 \psi(z)\,dz}\int_0^1 \frac{1}{b(y)\psi(y)}\left(\int_y^1 \psi(z)\,dz\right)dy, \tag{57}$$

where ψ is as defined in (52). After some algebra, this solution can equivalently be expressed in the form

$$T(x) \quad = \quad \int_0^1 t(x,y)\,dy, \tag{58}$$

where

$$t(x,y) \quad = \quad \begin{cases} 2A_0(x) \left[b(y)\psi(y) \int_0^y \psi(z)\,dz \right]^{-1} & \text{if} \quad 0 \le y \le x \\ 2A_1(x) \left[b(y)\psi(y) \int_y^1 \psi(z)\,dz \right]^{-1} & \text{if} \quad x \le y \le 1 \end{cases} \tag{59}$$

where $A_i(x)$ are as defined earlier. The above representation is not fortuitous. It can be shown, although we skip it here, that the function $t(x,y)$ has the following interpretation. For $0 \le y_1 < y_2 \le 1$, the integral $\int_{y_1}^{y_2} t(x,y)\,dy$ is the mean time that the diffusion process starting at x spends in the interval (y_1, y_2). In particular, if g is a well-behaved function on the state space, then

$$E\left(\int_0^\tau g(X_s)\,ds \mid X_0 = x \right) \quad = \quad \int_0^1 g(y)t(x,y)\,dy\,.$$

For each fixed non-absorbing state x, the function $t(x,\cdot)$ is what is called the *sojourn time density* of the diffusion starting at the state x.

We end this section here by simply mentioning that it is possible to derive the higher moments of the absorption time — more generally, of $\int_0^\tau g(X_s)\,ds$ — by proceeding in exactly the same way, except that the formulae become complicated.

0.10 References/Supplementary Readings

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[8] Ross, S. M. [1982]: *Stochastic Processes*, John Wiley & Sons.

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For supplementary reading on the material discussed in this chapter the reader may refer to the above books in the manner listed below.
Sections 0.1 through 0.6: [2], [3], [4], [5], [7], [8], [9].
Section 0.7 : [2], [6].
Section 0.8 : [1], [5], [6], [7], [8].
Section 0.9 : [1].

Chapter 1

BRANCHING PROCESSES

1.1 Introduction: Background and Definitions

Historically, it was in 1874 that Sir Francis Galton and H.W.Watson, while investigating the problem of "the extinction of family names" in England, formulated a simple but elegant mathematical model for the evolution of a family over successive generations. This was the first significant attempt to apply probability theory in order to study the effects of random fluctuations on the development of families or populations. It is this model that later came to be known as the *Galton-Watson Branching Process* and formed the basis of many subsequent extensions and generalizations.

Let us imagine objects that give birth to objects of the same kind; they may be men or bacteria reproducing by the usual biological methods, or neutrons in a chain reaction, and so on. We start with an initial set of objects, to be called the *zeroth generation.* Each member of this generation produces a certain number of offsprings and the aggregate of all these offsprings constitute the *first generation.* Each member of the first generation, in turn, produces offsprings, giving rise to the *second generation.* This is how the process of evolution continues — from one generation to the next. One can visualise this as a "tree" where each generation "branches off" to the next generation.

Formulating a stochastic model for this process of evolution simply means introducing a specific chance mechanism to govern the branchings that take place at different stages. Galton and Watson proposed the following model.

Any member of a generation produces offsprings according to some fixed distribution $\widetilde{p}$. Here $\widetilde{p} = (p_0, p_1, p_2, \ldots)$ is a probability distribution on the set of non-negative integers $\{0, 1, 2, \ldots\}$. This distribution remains the same over generations. Further, different members produce offsprings independently of one another.

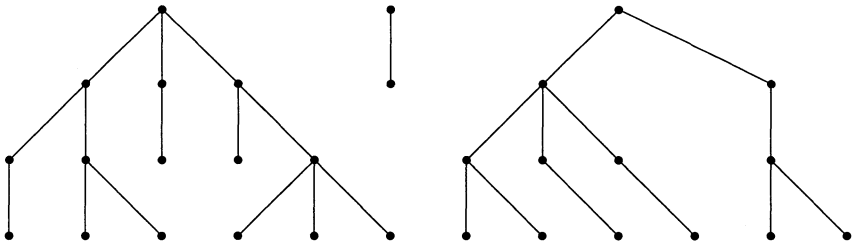

Figure 1.1: Successive Generations

In other words, the number of offsprings produced by different members (belonging to different or the same generation) are independent non-negative integer-valued random variables with a common distribution $\widetilde{p}$.

Denoting, for each $n \geq 0$, the total number of objects in the n-th generation by X_n, we get a sequence $(X_n)_{n\geq 0}$ of non-negative integer valued random variables. This discrete time stochastic process is what is called the *Galton-Watson Branching Process.* One is interested in various probabilistic questions about this process, concerning both finite-time as well as asymptotic behaviour. For example, on one hand, one may be interested in the probability distribution of X_n for fixed n. In particular, one may ask what is $E(X_n)$, the expected size of the n-th generation, or, what is $V(X_n)$, the variance. On the other hand, the singlemost important question about the process (X_n) is: what is the probability that X_n tends to 0 as n tends to infinity. Since the X_n take only non-negative integer values, the above probability is same as the probability that the X_n become all zero after some stage. Thus, it is the probability of *eventual extinction* of the family.

Before we begin a systematic analysis of the above and other questions, let us understand the probabilistic mechanism described above a little more clearly and convince ourselves that it completely determines the stochastic process $(X_n)_{n\geq 0}$ without any ambiguity. First of all, the initial set of objects is assumed to be fixed, so that, X_0 is a degenerate random variable, say, $X_0 = k_0$, a positive integer. (This assumption can be dispensed with and one can take X_0 to be a random variable with some distribution.) Each of these k_0 objects produce offsprings according to the distribution $\widetilde{p}$, independently of one another. Denoting by Y_i^0, the number of offsprings produced by the i-th individual of the initial population, $Y_1^0, \ldots, Y_{k_0}^0$ are i.i.d. random variables with common distribution $\widetilde{p}$. The total number of objects in the first generation is then the random variable $X_1 = Y_1^0 + \cdots + Y_{k_0}^0$. If $X_1 = 0$, which happens if and only if all the Y_i^0 equal zero, evolution stops there and the family becomes extinct at the first generation; all the subsequent X_n are defined to be zero in this case. On the other hand, if X_1 takes a positive value, say, $X_1 = k_1$,

then, conditional on this event the (distribution of the) random variable X_2 equals (that of) the random variable $Y_1^1 + \cdots + Y_{k_1}^1$ where Y_i^1, $1 \leq i \leq k_1$, are i.i.d. random variables with common distribution $\widetilde{p}$ and are independent of the random variables Y_i^0. In other words, given $X_1 = k_1 > 0$, the random variables Y_i^1, $1 \leq i \leq k_1$, represent the number of offsprings produced by the k_1 individuals of the first generation, so that $X_2 = Y_1^1 + \cdots + Y_{k_1}^1$ represents the size of the second generation. Once again, if $X_2 = 0$, all the subsequent X_n are defined to be zero and the family becomes extinct at the second generation. Otherwise, if $X_2 = k_2 > 0$, say, then conditional on the values of X_1 and X_2, the random variable X_3 is distributed simply as $Y_1^2 + \cdots + Y_{k_2}^2$, where Y_i^2, $1 \leq i \leq k_2$ are again i.i.d. random variables with common distribution $\widetilde{p}$ and are also independent of all the Y_i^0 as well as the Y_i^1. In general, for any $n \geq 1$, the distribution of X_{n+1}, conditional on the evolution upto the n-th generation, is as follows. If $X_n = 0$, then X_{n+1} is defined to be equal to zero, while if $X_n = k_n > 0$, then X_{n+1} is distributed as $Y_1^n + \cdots + Y_{k_n}^n$ where Y_i^n, $1 \leq i \leq k_n$ are i.i.d. random variables with common distribution $\widetilde{p}$ and are independent of all the Y_i^j, $0 \leq j \leq n-1$, $1 \leq i \leq k_j$. Thus, for every $n \geq 0$, we do have the conditional distribution of X_{n+1}, given $(X_0, \ldots, X_n)$. Noting that $X_0 \equiv k_0$, this completely determines (at least in principle) the joint distribution of $(X_0, X_1, \ldots, X_n)$, for every n, and hence the probability distribution of the entire proces $(X_n)_{n \geq 0}$. In practice, however, it may be quite complicated to write down these joint distributions. The reader may try her hand with (X_0, X_1, X_2), for example, to get a feel for the computations.

In the above, we described the conditional distributions, at every stage, of the size of the next generation, given the entire history of the evolution upto the present generation. However, one could not have failed to notice that these conditional distributions depend only on the size of the present generation, and that the basic rule remains the same for all the different generations. So, here we are ! Our branching process $(X_n)_{n \geq 0}$ is indeed a time-homogeneous Markov chain as discussed in Section 0.8. A formal proof of the following proposition (if still needed) is left to the reader.

Proposition 1.1: *The branching process $(X_n)_{n \geq 0}$ is a time-homogeneous Markov chain with the set of non-negative integers as state space. The initial state is k_0 and the transition probability matrix $P = ((p_{ij}))$ given by $p_{00} = 1$, and for $i > 0$, $p_{ij} = P(Y_1 + \cdots + Y_i = j)$, where $Y_1, \ldots, Y_i$ are i.i.d. random variables with common distribution $\widetilde{p}$.*

One could have, as well, started by defining the branching process as simply a Markov chain with transition probabilities as in Proposition 1.1. But our way of defining it seems to give a clear picture of the model. Moreover, even though our process turns out to be a Markov chain, the standard Markov chain techniques do not seem to be of much help in the study of this process. This is primarily because those techniques rest heavily on analyzing the structure of the transition matrix, while for the present process, entries of the transition matrix are almost impossible to write down explicitly.

1.2 Method of Probability Generating Functions

An elegant analysis of branching processes is obtained through an ingeneous use of the good old probability generating functions. The definition and an account of the properties of probability generating functions (p.g.f.'s, in short) were given in Section 0.3. From the definition of the process $(X_n)_{n\geq 0}$, as given in Section 1, it is clear that the entire distribution of the process is completely determined by two quantities — the initial size k_0 and the underlying probability distribution $\widetilde{p}$ for producing offsprings. Henceforth, we will often refer to $\widetilde{p}$ as the "*progeny distribution*" or the "*offspring distribution*". Let ϕ denote the p.g.f. of the progeny distribution

$$\phi(s) = \sum_{i\geq 0} p_i s^i, \quad \text{for } 0 \leq s \leq 1 \tag{1}$$

The function ϕ turns out to be of central importance in the analysis of the process (X_n), as we shall see. For this reason, the function ϕ is referred to as the *'progeny generating function'* of the process.

We shall begin with a basic result. For each $n \geq 1$, let the joint p.g.f. of $(X_0, X_1, \ldots, X_n)$ be denoted by $g_n^{(k_0)}$. The dependence on k_0 is made explicit for reasons to be clear. However, we have chosen to suppress the dependence on $\widetilde{p}$ (equivalently, on ϕ) for the simple reason that throughout our entire analysis, the progeny distribution will be assumed to be some fixed $\widetilde{p}$.

Theorem 1.2: *For any $n \geq 1$ and any $(s_0, s_1, \ldots, s_n)$ with $0 \leq s_i \leq 1$,*

$$g_n^{(k_0)}(s_0, s_1, \ldots, s_n) = g_{n-1}^{(k_0)}(s_0, \ldots, s_{n-2}, s_{n-1}\phi(s_n)) \,. \tag{2}$$

Proof: By the definition of $g_n^{(k_0)}$ and the properties of conditional expectation,

$$\begin{aligned} g_n^{(k_0)}(s_0, s_1, \ldots, s_n) &= E(s_0^{X_0} s_1^{X_1} \cdots s_n^{X_n}) \\ &= \sum_{k_1,\ldots,k_{n-1}} s_0^{k_0} s_1^{k_1} \cdots s_{n-1}^{k_{n-1}} E(s_n^{X_n} \,|\, X_0 = k_0, \ldots, X_{n-1} = k_{n-1}) \\ &\qquad \cdot P(X_0 = k_0, \ldots X_{n-1} = k_{n-1}) \,. \end{aligned}$$

Since the conditional distribution of X_n, given $X_0 = k_0, \ldots, X_{n-1} = k_{n-1}$, is the distribution of the sum of k_{n-1} i.i.d. random variables with common distribution $\widetilde{p}$ (this is valid even if $k_{n-1} = 0$), the above expression equals

$$\begin{aligned} &\sum_{k_1,\ldots,k_{n-1}} s_0^{k_0} s_1^{k_1} \cdots s_{n-1}^{k_{n-1}} (\phi(s_n))^{k_{n-1}} P(X_0 = k_0, \ldots, X_{n-1} = k_{n-1}) \\ &\qquad = E\left(s_0^{X_0} \cdots s_{n-2}^{X_{n-2}} (s_{n-1}\phi(s_n))^{X_{n-1}}\right) \\ &\qquad = g_{n-1}^{(k_0)}(s_0, \ldots, s_{n-2}, s_{n-1}\phi(s_n)) \end{aligned}$$

completing the proof of the theorem. ∎

To understand the implications of the above theorem, let us consider $g_2^{(k_0)}$, the joint p.g.f. of (X_0, X_1, X_2). By the above theorem, one has $g_2^{(k_0)}(s_0, s_1, s_2) = g_1^{(k_0)}(s_0, s_1\phi(s_2))$. For $g_3^{(k_0)}$, repeated application of the theorem would give $g_3^{(k_0)}(s_0, s_1, s_2, s_3) = g_2^{(k_0)}(s_0, s_1, s_2\phi(s_3)) = g_1^{(k_0)}(s_0, s_1\phi(s_2\phi(s_3)))$. The next corollary seeks to generalize this.

Corollary 1.3: *For any $n > 1$, and for any $s_0, \ldots, s_n$ with $0 \leq s_i \leq 1$,*

$$g_n^{(k_0)}(s_0, s_1, \ldots, s_n) = g_1^{(k_0)}(s_0, s_1\phi(s_2\phi(\cdots(s_{n-1}\phi(s_n))\cdots)) . \tag{3}$$

[Do not get intimidated by too many brackets and dots; if you have understood it for $n = 2, 3, 4$, you have understood it for general n.]

The corollary above shows that the joint p.g.f. of $(X_0, \ldots, X_n)$ is known for any n, once we have an expression for the joint p.g.f. of (X_0, X_1), which is what comes next.

Theorem 1.4: *For any s_0, s_1 with $0 \leq s_0, s_1 \leq 1$,*

$$g_1^{(k_0)}(s_0, s_1) = (s_0\phi(s_1))^{k_0} . \tag{4}$$

Proof: By definition, $g_1^{(k_0)}(s_0, s_1) = E(s_0^{X_0} s_1^{X_1})$. But $X_0 \equiv k_0$ and X_1 is distributed as the sum of k_0 many i.i.d. random variables with common p.g.f. ϕ. Therefore,

$$E(s_0^{X_0} s_1^{X_1}) = s_0^{k_0} E(s_1^{X_1}) = s_0^{k_0}(\phi(s_1))^{k_0} = (s_0\phi(s_1))^{k_0} .$$

In particular, with $k_0 = 1$, we have $g_1^{(1)}(s_0, s_1) = s_0\phi(s_1)$, so that, for any $k_0 \geq 1$,

$$g_1^{(k_0)}(s_0, s_1) = (g_1^{(1)}(s_0, s_1))^{k_0} = (s_0\phi(s_1))^{k_0} . \tag{5}$$

∎

Now appealing to Corollary 1.3 and Equation (5), it follows that, for every $n \geq 1$,

$$g_n^{(k_0)}(s_0, s_1, \ldots, s_n) = [g_n^{(1)}(s_0, s_1, \ldots, s_n)]^{k_0} . \tag{6}$$

What (6) means is that if we consider k_0 independent branching processes $(X_n^1)_{n\geq 0}, \ldots, (X_n^{k_0})_{n\geq 0}$, each with initial size 1 and common progeny distribution $\widetilde{p}$, then the process $(\widetilde{X}_n)_{n\geq 0}$ defined by $\widetilde{X}_n = \sum_{i=1}^{k_0} X_n^i$, $n \geq 0$ is, in distribution, the same as the branching process $(X_n)_{n\geq 0}$ with initial size k_0 and progeny distribution $\widetilde{p}$. In other words, a branching process with initial population size k_0 and progeny distribution $\widetilde{p}$ is just the "sum" of k_0 independent branching processes, each with initial population size 1 and progeny distribution $\widetilde{p}$. This should be intuitively clear. After all, if we fix attention on any one of the k_0 individuals of the 0-th generation and consider the successive generations originating from the offsprings of that individual only, we will clearly get a branching process with initial size 1 and progeny distribution $\widetilde{p}$. So the overall scenario would be that of k_0 such branching processes evolving

simultaneously. Moreover, since the k_0 members of the 0-th generation act independently, the corresponding branching processes would be independent. And, of course, the n-th generation of the whole family would simply be the aggregate of the n-th generation of the k_0 subfamilies. It all sounds very simple and logical, so much so as to make the dry mathematics done above appear uncalled for. Well, if you had known and believed it all along, you may still find the mathematical argument useful (at least in drawing a non-believer into your camp!).

Anyway, the point we sought to make is that, in the study of branching processes, it is alright to limit ourselves to the case $k_0 = 1$. This is what we shall do from now on. Thus in what follows,

(X_n) denotes a branching process with $X_0 \equiv 1$ and progeny distribution $\widetilde{p}$.

Having decided this, there is no need to hang on to the superscripts in the g_n defined earlier. We will simply write g_n (instead of $g_n^{(1)}$) for the joint p.g.f. of $(X_0, X_1, \ldots, X_n)$. Our earlier results, when specialized to this case, can be summarized as

Theorem 1.5: *For every $n \geq 1$,*

$$\begin{aligned} g_n(s_0, s_1, \ldots, s_{n-1}, s_n) &= g_{n-1}(s_0, s_1, \ldots, s_{n-2}, s_{n-1}\phi(s_n)) \\ &= \cdots\cdots\cdots \\ &= g_1(s_0, s_1\phi(s_2\phi(\cdots s_{n-1}\phi(s_n)\cdots))) \\ &= s_0\phi(s_1\phi(s_2\cdots s_{n-1}\phi(s_n)\cdots)) \end{aligned} \tag{7}$$

and consequently,

$$g_n(s_0, s_1, \ldots, s_n) = s_0\phi(g_{n-1}(s_1, \ldots, s_n))\,. \tag{8}$$

The recurrence relations (7) and (8) between the successive g_n capture the essence of the evolution mechanism of a branching process. Let us denote, for every $n \geq 0$, the p.g.f. of X_n by ϕ_n. Since $X_0 \equiv 1$, one of course has $\phi_0(s) = s$. For any $n \geq 1$, clearly $\phi_n(s) = g_n(1, 1, \ldots, 1, s)$. Relations (7) and (8) now yield

Theorem 1.6: *For every $n \geq 1$,*

$$\phi_n(s) = \phi_{n-1}(\phi(s)) = \phi(\phi_{n-1}(s))\,. \tag{9}$$

Thus the ϕ_n, $n \geq 0$ are simply the successive iterates of the function ϕ, that is, $\phi_0(s) = s$, $\phi_1(s) = \phi(s)$, $\phi_2(s) = \phi(\phi(s))$, and so on. It is, however not always possible to get simple explicit expressions for the successive iterates of a given p.g.f. ϕ. Examples of a situation where it is possible are given in Exercise 9. Nevertheless, the relations (9) can be used very fruitfully to answer a number of important theoretical questions about the process, as we will presently see.

1.3 Moments of the Successive Generation Sizes

To start with, let us ask how the expected population size changes from generation to generation. We will assume, henceforth, that the underlying progeny distribution $\widetilde{p}$ has a finite mean m, that is, $\sum_{k\geq 0} kp_k = m < \infty$. This, of course, implies that $\phi'(1) = m$.

Since $X_0 \equiv 1$, we have $E(X_0) = 1$. Also, X_1 is clearly a random variable with distribution $\widetilde{p}$, so $E(X_1) = m$. Thus, the first generation will have expected size m. Again, each of these m individuals produce offsprings according to the distribution $\widetilde{p}$, so each is expected to produce m offsprings. The expected size of the second generation should, therefore, be m^2. Proceeding in this way, in general, the expected size of the n-th generation should be m^n. That this is indeed the case will now be proved using (9). Since ϕ is assumed to have a finite derivative at 1, it is easy to show, by induction and by using (9), that ϕ_n, for each n, has a finite derivative at 1. Indeed, simple chain rule applied to (9) gives $\phi_n'(1) = \phi_{n-1}'(\phi(1)).\phi'(1) = \phi_{n-1}'(1).\phi'(1)$, since $\phi(1) = 1$. This means that each X_n has finite expectation, and, that for each n, $E(X_n) = mE(X_{n-1})$. Since $E(X_1) = m$, it follows that for each n,

$$E(X_n) = m^n\,. \tag{10}$$

We will now employ similar methods to get a formula for the variance of X_n, namely $V(X_n)$. For this, we assume that the distribution $\widetilde{p}$ has a finite second moment and denote its variance by σ^2. This of course means that ϕ has a second derivative which at the point 1 is given by $\phi''(1) = \sigma^2 + m^2 - m$. Using (9) again, it follows that each ϕ_n has a finite second derivative and one has

$$\phi_n''(s) = \phi_{n-1}''(\phi(s))[\phi'(s)]^2 + \phi_{n-1}'(\phi(s))\phi''(s)\,.$$

In particular, at $s = 1$, using $\phi(1) = 1$, $\phi'(1) = m$, $\phi_{n-1}'(1) = m^{n-1}$ and $\phi''(1) = \sigma^2 + m^2 - m$, one obtains

$$\begin{aligned}
\phi_n''(1) &= \phi_{n-1}''(1)m^2 + m^{n-1}(\sigma^2 + m^2 - m) \\
&= [\phi_{n-2}''(1)m^2 + m^{n-2}(\sigma^2 + m^2 - m)]m^2 + m^{n-1}(\sigma^2 + m^2 - m) \\
&= \phi_{n-2}''(1)m^4 + (\sigma^2 + m^2 - m)(m^n + m^{n-1}) \\
&= \cdots\cdots\cdots \\
&= \phi_1''(1)m^{2n-2} + (\sigma^2 + m^2 - m)(m^{2n-3} + \cdots + m^n + m^{n-1}) \\
&= (\sigma^2 + m^2 - m)(m^{2n-2} + \cdots + m^{n-1})\,.
\end{aligned}$$

Thus

$$\begin{aligned}
V(X_n) &= \phi_n''(1) + \phi_n'(1) - (\phi_n'(1))^2 \\
&= (\sigma^2 + m^2 - m)(m^{2n-2} + \cdots + m^{n-1}) + m^n - m^{2n} \\
&= \sigma^2 m^{n-1}(m^{n-1} + \cdots + m + 1) + m^n(m-1)(m^{n-1} + \cdots + m + 1) \\
&\quad + m^n(1 - m^n) \\
&= \sigma^2 m^{n-1}(m^{n-1} + \cdots + m + 1),
\end{aligned}$$

giving us

$$V(X_n) = \begin{cases} n\sigma^2 & \text{if} \quad m = 1 \\ \sigma^2 m^{n-1} \frac{m^n - 1}{m-1} & \text{if} \quad m \neq 1 \end{cases} . \tag{11}$$

We have thus proved

Theorem 1.7: *If the progeny distribution $\widetilde{p}$ has a finite mean m, then, for all $n \geq 1$, $E(X_n) = m^n$. Moreover if $\widetilde{p}$ has a finite second moment also, then $V(X_n) = n\sigma^2$ or $\sigma^2 m^{n-1}(m^n - 1)(m-1)^{-1}$ according as $m = 1$ or $m \neq 1$, where σ^2 is the variance of $\widetilde{p}$.*

Higher order moments can also be obtained in a similar fashion, provided we assume the existence of such moments for $\widetilde{p}$.

1.4 Extinction Probability

The singlemost important question, at least historically, about the branching process (X_n) is what was posed originally by F.Galton: to find the probability of extinction of the family. *Extinction* here means the event that at some generation no further offsprings are produced, so that none of the future generations ever come to exist. Analytically, it is the event

$$A = \bigcup_{n \geq 1} \{X_n = X_{n+1} = \cdots = 0\} .$$

Since the X_n take only integer values, A also equals the event $\{\lim_{n \to \infty} X_n = 0\}$. We shall denote the probability of this event by q. This is what is called the *extinction probability*. Our question is: how to find q for a branching process with a given progeny distribution $\widetilde{p}$ (equivalently, for a given p.g.f. ϕ)?

Observe that, by the definition of the process (X_n), the event $\{X_n = 0\}$ is the same as the event $\{X_n = X_{n+1} = \cdots = 0\}$, so that q is simply the probability of the event that $X_n = 0$ for some n. Moreover, the events $\{X_n = 0\}$ are monotone increasing with n, so that $P(X_n = 0)$ also increases with n. An alternative analytical way to argue this is as follows. Denote $q_n = P(X_n = 0)$. Clearly, $q_n = \phi_n(0)$ for each n. Now, $X_0 \equiv 1$, so that $q_0 = 0$. Next, $q_1 = P(X_1 = 0) = p_0 \geq 0 = q_0$. Again, $q_2 = \phi_2(0) = \phi(\phi(0)) = \phi(p_0) \geq \phi(0) = q_1$, using the fact that $\phi(s) = \sum_{k \geq 0} p_k s^k$ is a non-decreasing function of s on [0,1]. In general, having proved that $q_n \geq q_{n-1}$, one obtains similarly that $q_{n+1} = \phi_{n+1}(0) = \phi(\phi_n(0)) = \phi(q_n) \geq \phi(q_{n-1}) = \phi(\phi_{n-1}(0)) = \phi_n(0) = q_n$. Induction completes the proof.

It is now clear from above that $q = \lim_{n \to \infty} P(X_n = 0) = \lim_{n \to \infty} q_n$. In fact $q_n \uparrow q$ as $n \uparrow \infty$. Moreover, for each n, $q_n = \phi(q_{n-1})$. Letting $n \to \infty$ on both sides and using continuity of ϕ, it follows that q must satisfy $q = \phi(q)$. Since $0 \leq q \leq 1$, in order to find q, one must solve the equation

$$\phi(s) = s \qquad \text{for } 0 \leq s \leq 1. \tag{12}$$

Since $\phi(1) = 1$, $s = 1$ is always a solution of (12). Galton observed this and unfortunately, made the mistake of concluding that q is always equal to 1. The point he missed is that the Equation (12) may have other solutions in the interval [0,1], and, all that one can say at this stage is that one of these solutions is q — but which one among these solutions is q, remains open.

As a step towards further identification of q, we now show that q is the *smallest* solution of (12) in the interval [0,1]. In other words, if π is any solution of (12) in [0,1], then $q \leq \pi$. To prove this, it sufficies to show that $q_n \leq \pi$ for each n (since $q = \lim_{n\to\infty} q_n$). We use induction and the fact that ϕ is non-decreasing on [0,1]. First of all, $q_0 = 0 \leq \pi$, and, $q_1 = \phi(0) \leq \phi(\pi) = \pi$. To complete induction, assume that $q_n \leq \pi$ and we have $q_{n+1} = \phi(q_n) \leq \phi(\pi) = \pi$. Thus we have proved

Theorem 1.8: *For a branching process with ϕ as the p.g.f. of the progeny distribution, the extinction probability q is the smallest solution of the equation $\phi(s) = s$ in $[0, 1]$.*

Before proceeding any further, let us see some examples of simple applications of the above. Consider a society that follows the policy of "two children per family". We want to examine the chances of survival of a family name in such a society. To bring a branching process into the scene, we make the following assumptions: (1) Family names are carried only by the male offsprings. (We do not mean to preach patriarchy — this is, after all, a reality in most societies.) (2) Each couple gives birth to exactly two children. This may perhaps seem unrealistic. Our rationale is that we basically want our family names to survive and, therefore, given the stipulation of the society, each couple will try to produce two offsprings. Fertility and related issues are ignored. (3) The probabilities of an offspring being a male or a female are equal. (4) Sexes of different offsprings are determined independently. The assumptions (3) and (4) are the standard rules of genetics, as we shall see in the next chapter.

If we start with one male member of a family and concentrate on his male offsprings, then their male offsprings and so on, we get a branching process with its progeny distribution given by $p_0 = 1/4$, $p_1 = 1/2$, $p_2 = 1/4$ and $p_3 = p_4 = \cdots = 0$. This has p.g.f. $\phi(s) = \frac{1}{4} + \frac{1}{2}s + \frac{1}{4}s^2$. It is easy to see that $s = 1$ is the only solution of (12) in [0,1], and, therefore $q = 1$ in this case. Thus, among the descendents of any one particular male member of a family, it is with probability one that sooner or later there would not be anyone left to carry his family name. Since the same fate awaits the family name of every member of the society, each family name is sure to be wiped out eventually. (Have we proved that our family planning programme, if strictly followed, spells doom for our society?)

If instead, the society followed a policy of "three children per family", then we would have $\phi(s) = \frac{1}{8} + \frac{3}{8}s + \frac{3}{8}s^2 + \frac{1}{8}s^3$. One can check that now (12) has two roots in [0,1], namely, $s = 1$ and $s = \sqrt{5} - 2$. By Theorem 1.8, the

extinction probability equals $\sqrt{5} - 2$ (~ 0.234, approximately). Thus, there is a positive probability for a family name to survive. For example, if there are k male members in a family now, the reader should have no difficulty in deducing from (12) and the remark thereafter, that the probability of survival of that family name is approximately $1 - (0.234)^k$.

It is clear from the above examples that in order to get q by appealing to Theorem 1.8, we have to somehow know what are all the solutions of equation (12) in [0,1]. However, except in some very simple cases as above, it is almost always impossible to explicitly solve equation (12). This is a serious limitation of Theorem 1.8, from the point of view of applicability. It would be very nice if we could come up with a simpler criterion which would enable us at least to tell whether q equals 0 or 1 or whether q lies strictly in between (even though the exact value may not be known). This is what we take up next.

We begin by eliminating a trivial (and, hence uninteresting) case, namely, when $p_0 = 0$. In this case, any individual in any generation produces at least one offspring, so that the extinction is completely ruled out. Analytically speaking, $p_0 = 0$ implies that $P(X_1 \geq 1) = 1$, and, also $P(X_{n+1} \geq X_n) = 1$, for all n. Therefore $P(X_n = 0) = 0$ for each n, and, hence $q = 0$. In fact, since q satisfies (12) so that $q = 0$ if and only if $\phi(0) = 0$, that is, $p_0 = 0$. In what follows, we assume, therefore that $p_0 > 0$.

Theorem 1.9: *Assume $p_0 > 0$. Then $s = 0$ is never a solution of* (12) *and $s = 1$ is always a solution. If $m \leq 1$, then* (12) *has no solution in the open interval (0,1), while if $m > 1$, then there is one and exactly one such solution. Consequently, if $m \leq 1$, then the extinction probability equals 1, while if $m > 1$, the extinction probability is strictly between 0 and 1.*

Proof: That $s = 0$ is not a solution of (12) if $p_0 > 0$ and that $s = 1$ is always a solution has been already seen. To prove the other assertions, we consider two cases separately.

Case 1: $p_0 + p_1 = 1$.

In this case, $\phi(s) = p_0 + p_1 s$, so that its graph is a straight line L_1, say, as shown. Since $\phi(0) = p_0 > 0$ and $\phi(1) = 1$, the line L_1 meets the line $L : \psi(s) = s$ at exactly one point, namely, when $s = 1$. Thus, the only solution of the equation (12) in the interval [0,1] is $s = 1$. Note that, in this case, $m = p_1 < 1$, and, the extinction probability is 1 as asserted.

Case 2: $p_0 + p_1 < 1$.

We first show that, whatever be m, the equation (12) has at most one solution in the open interval (0,1). Note that ϕ has derivatives of all orders in the open interval (0,1) and, in particular, $\phi''(s) = \sum_{k \geq 2} k(k-1) p_k s^{k-2}$, for $s \in (0,1)$. Since this power series in s has all coefficients non-negative and at least one coefficient strictly positive, it follows that $\phi''(s) > 0$ for all $s \in (0,1)$. Therefore, $\phi'(s)$ is strictly increasing on the open interval (0,1). Now, suppose, if possible, equation (12) has two distinct roots, say, α and β, with $\alpha < \beta$, in the open interval (0,1). Then α, β and 1 are three distinct zeros of the function $\phi(s) - s$ and $\alpha < \beta < 1$. By the mean value theorem, there must exist points γ and

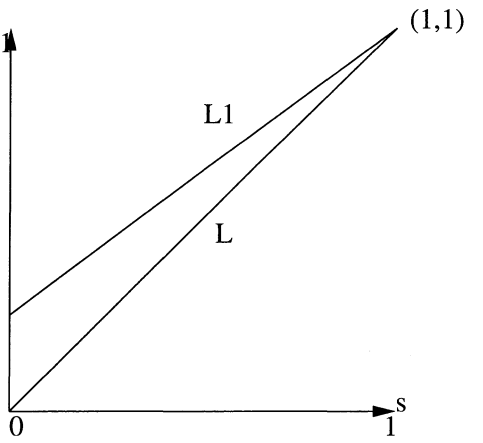

Figure 1.2: $L1 : \varphi(s) = p_0 + p_1 s; \quad L : \psi(s) = s$

δ with $\alpha < \gamma < \beta$ and $\beta < \delta < 1$, such that $\phi'(\gamma) = \phi'(\delta) = 1$. But this is impossible since $\phi'(s)$ is strictly increasing on the interval (0,1).
We now complete the proof of the theorem by showing that if the function $\phi(s) - s$ has no zeros in the open interval (0,1), then m must be ≤ 1, whereas, if $\phi(s) - s$ has a zero in (0,1), then we must have $m > 1$.

So, suppose first that the function $\phi(s) - s$ has no zeros in the open interval (0,1). It then follows that $\phi(s) - s > 0$ for all $s \in (0,1)$. This is because $\phi(0) - 0 = p_0 > 0$. So, if for some $s_0 \in (0,1)$, $\phi(s_0) - s_0 \leq 0$, then the intermediate value theorem applied to the continuous function $\phi(s) - s$ would assert that $\phi(s) - s$ must have a zero inside the interval $(0, s_0]$, thereby contradicting our hypothesis. Now, since $\phi(s) > s$ for all $s \in (0,1)$, we have $(1-\phi(s)) \,/\, (1-s) < 1$ for all $s \in (0,1)$. Taking limit as $s \uparrow 1$, we obtain $\phi'(1) \leq 1$, that is $m \leq 1$.
If, on the other hand, $\phi(s) - s$ has a zero, say at s_0, in the open interval (0,1), then, in view of the fact that $s = 1$ is always a zero of the function $\phi(s) - s$, mean value theorem will assert the existence of a point s_1 in the open interval $(s_0, 1)$, such that $\phi'(s_1) = 1$. Since $\phi'(s)$ is strictly increasing on (0,1), we immediately have $\phi'(1) = \lim_{s \uparrow 1} \phi'(s) > \phi'(s_1) = 1$, that is $m > 1$. ∎

Remarks:

1. Note that in the examples discussed above m equals 1 in the first case and equals $\frac{3}{2}$ (>1) in the second case.

2. We know that $q = \lim_{n \to \infty} P(X_n = 0) = \lim_{n \to \infty} \phi_n(0)$, by definition. But, as a matter of fact, if $p_0 > 0$, then for every $s \in [0,1)$, $\phi_n(s) \to q$ as $n \to \infty$. This can be seen as follows. First of all, since Equation (12) does not have a solution in $[0, q)$ and since $\phi(0) > 0$, it follows from the intermediate value theorem and the monotonicity of the function ϕ, that, for every $s \in [0, q]$, one has $s \leq \phi(s) \leq \phi(q) = q$. Using induction, one gets the string of inequalities

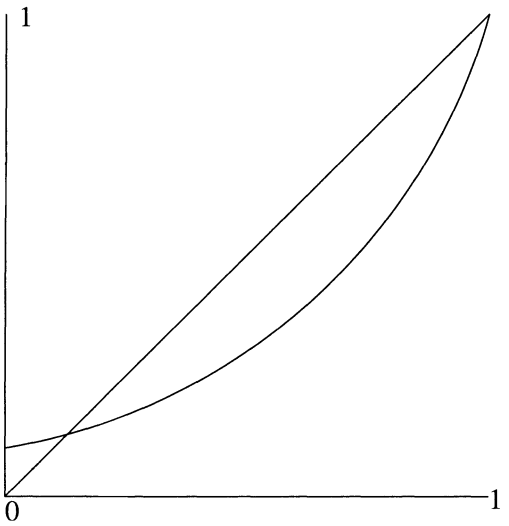

Figure 1.3: $m > 1$

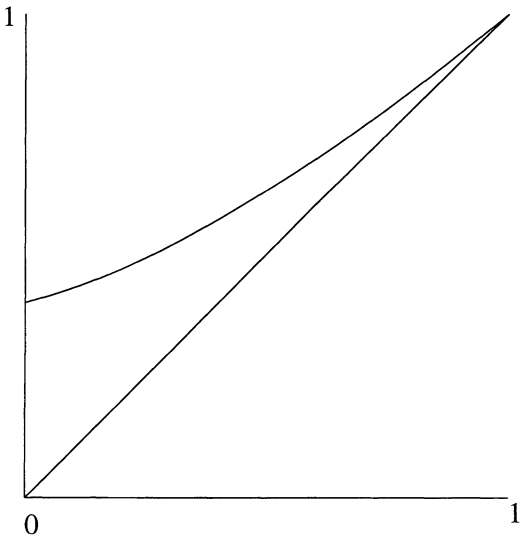

Figure 1.4: $m < 1$

$s \leq \phi(s) \leq \phi_2(s) \leq \phi_3(s) \leq \cdots \leq \phi(q) = q$ for every $s \in [0,q]$. But, at the same time $\phi_n(0) \to q$ as $n \to \infty$, and $\phi_n(s) \geq \phi_n(0)$ for all $s \geq 0$. All these would imply that, for each $s \in [0,q]$, $\phi_n(s) \uparrow q$ as $n \uparrow \infty$. In particular, when $q = 1$ (which happens if and only if $m \leq 1$), we have $\phi_n(s) \uparrow q = 1$ as $n \uparrow \infty$, for every $s \in [0,1]$. When $q < 1$ (equivalently $m > 1$), the convexity of the function ϕ implies that, for every $s \in (q,1)$, $1 > s \geq \phi(s) \geq \phi(q) = q$, and by induction, $1 > s \geq \phi(s) \geq \phi_2(s) \geq \phi_3(s) \geq \cdots \phi(q) = q$. It follows that for every $s \in (q,1)$, $\lim_{n\to\infty} \phi_n(s)$ exists. Clearly the limit satisfies Equation (12). However, it is strictly less than one and hence must equal q. Thus $\phi_n(s) \downarrow q$ as $n \uparrow \infty$, for each $s \in (q,1)$ also.

So far we have focussed our attention entirely on the methods of finding the probability of extinction. Its complementary event is the event of "survival". By definition, this is simply the event that all the X_n are different from zero. It turns out, however, that in case this event occurs, the X_n actually have to grow indefinitely large. One has to, of course, set aside one trivial exceptional case, namely, when $p_1 = 1$, in which case $P(X_n = 1 \text{ for all } n) = 1$. We will now show that, if $p_1 < 1$, then, with probability one, the sequence (X_n) either goes to zero or diverges to infinity as $n \to \infty$; it is not possible for the successive generation sizes to remain positive and at the same time bounded.

Theorem 1.10: *Assume that* $p_1 < 1$. *Then for every* $k \geq 1$, $P(X_n = k) \to 0$ *as* $n \to \infty$. *Moreover,* $P(X_n \to \infty) = 1 - q = 1 - P(X_n \to 0)$.

Proof: We are going to use standard Markov chain terminology and notations. We show that for the chain (X_n), each of the states $\{1,2,\ldots\}$ is a transient state. For this, we simply have to show that $f_{kk} < 1$ for each $k \in \{1,2,\ldots\}$, where, following the Markov chain theory of Section 0.8, f_{kk} denotes the conditional probability $P(X_{n+j} = k \text{ for some } j \geq 1 \,|\, X_n = k)$. This probability, of course, does not depend on n because of time homogeneity. If $p_0 = 0$, then noting that $p_{ik} = 0$ for any $i > k$, one gets $f_{kk} = P(X_{n+1} = k \,|\, X_n = k) = p_1^k < 1$. On the other hand, if $p_0 > 0$, that is, $1 - p_0^k < 1$, we get $f_{kk} \leq 1 - P(X_{n+1} = 0 \,|\, X_n = k) < 1$. Noting that $X_0 \equiv 1$, it now follows from the general theory that, for $k \in \{1,2,\ldots\}$, $\lim_{n\to\infty} P(X_n = k) = \lim_{n\to\infty} P(X_n = k \,|\, X_0 = 1) = 0$, and also that, $P(X_n = k \text{ for infinitely many } n) = P(X_n = k \text{ for infinitely many } n \mid X_0 = 1) = 0$. From this last assertion, one easily deduces that, for any positive integer L, however large, $P(X_n \in \{1,2,\ldots L\} \text{ for infinitely many } n) = 0$. This, of course, means that, with probability one, either $X_n \to 0$ or $X_n \to \infty$ as $n \to \infty$. The proof is complete. ∎

1.5 Asymptotic Behaviour

In the last section, we saw that, when $m > 1$, a family starting with one ancestor has a positive probability of surviving (Theorem 1.9), and moreover, if it does survive, then the generation sizes grow indefinitely large (Theorem

1.10). The so called *Malthusian Law* then expects the sizes to grow eventually at a geometric rate. We now show mathematically that this is indeed the case. One actually has $X_n \sim Wm^n$ for large n, where W is a random variable which is zero only when $X_n \longrightarrow 0$.

Theorem 1.11: *Assume that* $m \neq 0$, *and, let* $W_n = \dfrac{X_n}{m^n}$, *for* $n \geq 0$.

(a) The sequence (W_n) *of random variables converges to a random variable* W *with probability one. If moreover,* $m > 1$ *and* $E(X_1^2) < \infty$, *then the convergence takes place in* L_2 *also, and* $E(W) = 1$ *and* $V(W) = V(X_1)(m^2 - m)^{-1}$.

(b) If X_1 *is non-degenerate with* $E(X_1^2) < \infty$, *then the random variable* W *of (a) takes the value zero only when* (X_n) *converges to zero.*

Proof: From definition, the conditional distribution of X_{n+1}, given $X_0 = x_0$, $X_1 = x_1$, ..., $X_n = x_n$, is just that of the sum of x_n-many i.i.d. random variables with common mean m. It follows that

$$E(X_{n+1} \,|\, X_0 = x_0, X_1 = x_1, \ldots, X_n = x_n) = m \cdot x_n \,,$$

that is,

$$E\left(\frac{X_{n+1}}{m^{n+1}} \,\middle|\, X_0 = x_0, X_1 = x_1, \ldots, X_n = x_n\right) = \frac{x_n}{m^n} \,.$$

Since the events $(X_0 = x_0, X_1 = x_1, \ldots, X_n = x_n)$ and $(W_0 = x_0, W_1 = x_1/m, \ldots, W_n = x_n/m^n)$ are identical, the above equality is same as

$$E\left(W_{n+1} \,|\, W_0 = x_0, W_1 = \frac{x_1}{m}, \ldots, W_n = \frac{x_n}{m^n}\right) = \frac{x_n}{m^n} \,.$$

In the notation of Section 0.2.3, this amounts to

$$E(W_{n+1} \,|\, W_0, W_1, \ldots, W_n) = W_n \,.$$

Thus $(W_n)_{n \geq 0}$ is a non-negative martingale. By Doob's martingale convergence theorem of Section 0.7, the sequence (W_n) converges, with probability one, to a random variable W with $E(W) < \infty$. In case $m > 1$ and $E(X_1^2) < \infty$, we have from (11), that

$$E(W_n^2) = 1 + V\left(\frac{X_n}{m^n}\right) = 1 + V(X_1).(1 - m^{-n})(m^2 - m)^{-1},$$

so that

$$\sup_n E(W_n^2) = 1 + V(X_1)(m^2 - m)^{-1} < \infty \,.$$

This implies that (W_n) converges to W in L_2 also. Moreover, in this case, $E(W) = \lim_{n \to \infty} E(W_n) = 1$ (since $E(W_n) = 1$ for all n) and $V(W) = \lim_{n \to \infty} V(W_n)$ $= \lim_{n \to \infty} V(X_1)(1 - m^{-n})(m^2 - m)^{-1} = V(X_1)(m^2 - m)^{-1}$.

For part (b), observe first that $X_n \to 0$ (equivalently, $X_n = 0$ for all large n) implies that $W = 0$. Thus, in case $m \leq 1$, one has $P(W = 0) = P(X_n \to 0) = 1$.

We now show that in the case $m > 1$ also, $P(W = 0)$ equals the extinction probability q. First of all, it is a consequence of (8) that

$$E(s_1^{X_1} s_2^{X_2} \cdots s_n^{X_n}) = \phi(s_1 E(s_2^{X_1} \cdots s_n^{X_{n-1}})) .$$

Observe that by the smoothing property of conditional expectation, as in Section 0.2.3, the left-hand side of the above equation equals

$$\sum_{k \geq 0} s_1^k E(s_2^{X_2} \cdots s_n^{X_n} \mid X_1 = k) p_k ,$$

while, by the definition of the function ϕ, the right-hand side equals

$$\sum\nolimits_{k \geq 0} [s_1 E(s_2^{X_1} \cdots s_n^{X_{n-1}})]^k p_k .$$

Thus, we have

$$\sum_{k \geq 0} s_1^k E(s_2^{X_2} \cdots s_n^{X_n} \mid X_1 = k) p_k = \sum_{k \geq 0} [s_1 E(s_2^{X_1} \cdots s_n^{X_{n-1}})]^k p_k .$$

Therefore, for each $k \geq 0$, $E(s_2^{X_2} \cdots s_n^{X_n} \mid X_1 = k) = (E(s_2^{X_1} \cdots s_n^{X_{n-1}}))^k$, that is, the joint p.g.f. of the conditional distribution of $(X_2, \ldots, X_n)$ given $X_1 = k$, equals the k-th power of the joint p.g.f. of the unconditional joint distribution of $(X_1, \ldots, X_{n-1})$. This means that, given $X_1 = k$, the random vector $(X_2, \ldots, X_n)$ is conditionally distributed as the sum of k independent random vectors, each distributed like the vector $(X_1, \ldots, X_{n-1})$. One concludes that $P(W_n \to 0 \mid X_1 = k) = [P(W_n \to 0)]^k$. Since

$$P(W_n \to 0) = \sum_k P(W_n \to 0 \mid X_1 = k) \cdot p_k ,$$

it follows that the probability $q^* = P(W_n \to 0)$ is a solution of the equation (12). But, since under the hypothesis, $V(W) = V(X_1)(m^2 - m)^{-1} > 0$, q^* can not be equal to 1. This forces q^* to be equal to q. ∎

Thus, we see that $\dfrac{X_n}{m^n}$ converges with probability one to a random variable W. Also, if the progeny distribution $\widetilde{p}$ has a strictly positive finite variance, then $W = 0$ only when $X_n \to 0$. Thus, in this last case, we indeed have $X_n \sim W \cdot m^n$ asymptotically as asserted earlier. In case $m \leq 1$ of course, the statement $X_n \sim W \cdot m^n$ holds irrespective of whether $\widetilde{p}$ has finite variance or not. Thus the condition that EX_1^2 is finite, becomes important really in the case $m > 1$ only. Levinson (see Harris for details) has given an example with $EX_1^2 = \infty$, where $W \equiv 0$ even though $m > 1$ (and, hence $P(X_n \to 0) < 1$). Under the condition $0 < V(X_1) < \infty$, we know that if $m > 1$, then on the set where $X_n \not\to 0$ (which has positive probability), the random variable W takes strictly positive values. Our next result shows that W is actually non-degenerate on this set.

Theorem 1.12: *Under the condition $m > 1$ and $0 < V(X_1) < \infty$, the conditional variance of W, given $W > 0$, is strictly positive.*

Proof: We want to show that $E(W^2 \mid W > 0) - (E(W \mid W > 0))^2 > 0$. Observe first that the function $\psi(s) = (1-q)^{-1}\{\phi(s(1-q)+q) - q\}$ is a p.g.f. with $\psi(0) = 0$ and $\psi^{(r)}(0) = (1-q)^{r-1}\phi^{(r)}(q)$ for $r \geq 1$. It is also easy to verify that $\psi''(1) < \infty$ and that the variance of the underlying distribution, namely, $\psi''(1) + \psi'(1) - (\psi'(1))^2$ is strictly positive, or equivalently, that $(1-q)\phi''(1) - m^2 + m > 0$. Since $P(W > 0) = 1 - q$, the assertion is proved.

∎

In view of Theorem 1.11(a), the distribution of the random variable W would be of interest in order to study the distribution of X_n for large n. The next result gives one preliminary fact about the distribution of W.

Theorem 1.13: *Under the conditions of Theorem 1.11, the characteristic function of W, namely, the function $f(t) = E(e^{itW})$, $-\infty < t < \infty$, satisfies the relation*

$$f(mt) = \phi(f(t)). \tag{13}$$

Moreover, if $m > 1$, then this is the only characteristic function satisfying (13) *corresponding to a distribution with mean 1.*

Perhaps, we have got a little explaining to do regarding the right side of (13). Usually when we talk of the p.g.f. of a distribution, we think of it as a function defined on the interval [0,1]. But it should be clear that $\phi(z)$ makes perfect sense also for complex numbers z with $|z| \leq 1$. The recurrence relation (9) remains intact even if we allow such complex variables for the function ϕ.

Proof: In view of the remark made in the above paragraph, the relations (9) yield

$$E(e^{itX_n}) = \phi_n(e^{it}) = \phi(\phi_{n-1}(e^{it})) = \phi(E(e^{itX_{n-1}})),$$

for all $n \geq 1$ and all $t \in (-\infty, \infty)$. Denoting the characteristic function of W_n by f_n, it follows that

$$f_n(mt) = E\left[e^{(itX_n/m^{n-1})}\right] = \phi\left(E\left[e^{(itX_{n-1}/m^{n-1})}\right]\right) = \phi(f_{n-1}(t)).$$

But W_n converges to W with probability one, and hence, by the dominated convergence theorem, $f_n(t)$ converges to $f(t)$ for each t. Since ϕ is continuous, the relation (13) follows.
To prove the other part, let f and g be two characteristic functions satisfying $f'(0) = g'(0) = i$, and,

$$f(mt) = \phi(f(t)) \quad \text{and } g(mt) = \phi(g(t)), \quad \text{for all } t \in (-\infty, \infty).$$

From the first condition, one clearly has $f(t) - g(t) = th(t)$ where h is a continuous function with $h(0) = 0$. Now,

$$|mt\,h(mt)| = |f(mt) - g(mt)| = |\phi(f(t)) - \phi(g(t))| \leq m|f(t) - g(t)| = m|th(t)|,$$

the inequality following from the fact that $|\phi'(z)| \leq m$ for all $|z| \leq 1$. Thus we have $|h(mt)| \leq |h(t)|$, or, by induction, for any fixed t, $|h(t)| \leq |h(t/m^n)|$ for all $n \geq 1$. By continuity of h, we get, for all t, $|h(t)| \leq |h(0)| = 0$. Thus, $f(t) = g(t)$ for all t. ∎

Using Theorem 1.13, it is possible to derive a lot of information about the distribution of W. Among many other things one can show, for example, that the distribution of W is absolutely continuous except for a point mass at zero. For this and other things the book of Harris is the best source.

1.6 The Total Progeny of a Branching Process

The material in this section is also available in Feller, who attributes it to a paper of I. J. Good and gives the exact reference. Here the objects of interest are the random variables Z_n, defined for $n \geq 1$, as

$$Z_n = 1 + X_1 + X_2 + \cdots + X_n, \tag{14}$$

where (X_n) is a branching process as defined in the previous sections with $X_0 \equiv 1$. Z_n denotes the total number of descendants upto and including the n-th generation. Note that the original ancestor (forming the zeroth generation) is also included in Z_n. Clearly, the Z_n are non-decreasing with n. Letting $n \to \infty$, we get the *size of the total progeny*, $Z = 1 + X_1 + X_2 + \cdots$, which may or may not be finite. Let ν_n denote the p.g.f. of Z_n. Since $Z_1 = 1 + X_1$ and X_1 has p.g.f. ϕ, we have $\nu_1(s) = s\phi(s)$. In general, by conditioning on X_1 and observing that, given $X_1 = k$, the random variable $X_1 + X_2 + \cdots + X_n$ is (conditionally) distributed as the sum of k many i.i.d. copies of the random variable Z_{n-1}, one obtains the recurrence relation

$$\nu_n(s) = s\phi(\nu_{n-1}(s)) \,. \tag{15}$$

From (15), it is easy to see by induction that, for any s with $0 < s < 1$, the sequence $\{\nu_n(s)\}$ is monotone non-increasing and therefore, $\nu(s) = \lim_{n\to\infty} \nu_n(s)$ exists. By the Theorem discussed in Section 0.3, we know that $\nu(s)$ is the generating function of a sequence $\{\rho_k, k \geq 1\}$ of non-negative numbers with $\sum \rho_k \leq 1$. Indeed, $\rho_k = \lim_{n\to\infty} P(Z_n = k) = P(Z = k)$ for $k = 1, 2, 3, \ldots$. It also follows from (15) that $\nu(s)$ satisfies

$$\nu(s) = s\phi(\nu(s)) \quad \text{for} \quad 0 < s < 1, \tag{16}$$

so that, for fixed $s \in (0, 1)$, the value of $\nu(s)$ is a root of the equation (in t)

$$t = s\phi(t) \,. \tag{17}$$

We now show that (17) has a unique root for every fixed s in (0,1) and, moreover, that this root $\nu(s)$ is such that $\lim_{s\uparrow 1} \nu(s) = q$, where q is the extinction probability of the branching process (X_n).

Let us now dispose of the two extreme cases, namely, $p_0 = 0$ and $p_0 = 1$. When $p_0 = 0$, we know that $\phi(t) \leq t$ for all $t \in [0,1]$, so that $s\phi(t) \leq st < t$ for $0 < t \leq 1$. Therefore, $t = 0$ is the only solution of (17). Thus $\nu(s) = 0$ for all $s \in (0,1)$ and $\nu(1) = \lim_{s\uparrow 1} \nu(s) = 0$. Of course, we know that if $p_0 = 0$, then $q = 0$. When $p_0 = 1$, one has $\phi(t) \equiv 1$, so that $t = s$ is the only solution of the equation (17). Thus $\nu(s) = s$ for every $s \in (0,1)$ and therefore, $\lim_{s\uparrow 1} \nu(s) = 1$ which is, indeed, the extinction probability q in this case.

Now, let us consider the case $0 < p_0 < 1$. In this case, the function $g(t) = s\phi(t)$ is strictly increasing and convex (not necessarily strictly convex) with $g(0) = sp_0 > 0$ and $g(1) = s < 1$. Thus the function $f(t) = g(t) - t$ is a continuous function on $[0,1]$ with $f(0) > 0$ and $f(1) < 0$, so that there is at least one zero of $f(t)$ in $(0,1)$. Suppose, if possible, $0 < t_1 < t_2 < 1$ are two zeros of f. Then for some $t_0 \in (t_1, t_2)$, we have $f'(t_0) = 0$, that is, $g'(t_0) = 1$, which, by convexity, implies that $g'(t) \geq 1$ for $t \geq t_0$. By the Mean Value Theorem, $g(1) - g(t_2) = (1 - t_2)g'(\xi)$ for some $\xi \in (t_2, 1)$. But this, along with $g'(\xi) \geq 1$, will imply $g(1) - 1 \geq g(t_2) - t_2 = 0$, a contradiction.

Thus (17) has a unique solution in $(0,1)$ for each $s \in (0,1)$. Since $f(q) = s\phi(q) - q = (s-1)q < 0$, it also follows that the solution above lies in $(0,q)$. In view of (16), this unique solution is $\nu(s)$. In particular, for each $s \in (0,1)$, we have $0 < \nu(s) < q$, so that $\nu(1) = \lim_{s\uparrow 1} \nu(s) \leq q$. But, on the other hand, by letting $s \uparrow 1$ in equation (16), we observe that $\nu(1) = \lim_{s\uparrow 1} \nu(s)$ satisfies the relation $\nu(1) = \phi(\nu(1))$. By Theorem 1.8, it follows that $\nu(1) \geq q$. Thus, we have $\nu(1) = q$.

In particular, $\nu(s)$ is a probability generating function if and only if the extinction probability equals 1. Our findings can thus be summarized as

Theorem 1.14: *Let ρ_k be the probability that the total progeny consists of k elements, that is, $\rho_k = P(Z = k)$, for $k = 1, 2, \ldots$. Then*
(a) $\sum_{k\geq 1} \rho_k$ equals the extinction probability q and $1 - q$ is the probability of an infinite progeny.
(b) The generating function $\nu(s) = \sum_{k\geq 1} \rho_k s^k$, $0 < s < 1$, is given by the unique positive root of (17) *and $\nu(s) \leq q$.*

We next turn towards the expected total progeny. Of course, since the total progeny is given by $Z = 1 + X_1 + X_2 + \cdots$ and $E(X_n) = m^n$, one could argue that $E(Z) = \sum_{n\geq 0} m^n$ which is finite if and only if $m < 1$ and, in that case, it equals $(1-m)^{-1}$. This, however, requires the use of $E(\sum_{n\geq 0} X_n) = \sum_{n\geq 0} E(X_n)$ which needs monotone convergence theorem of Section 0.2.1. It is possible to bypass it using the generating function $\nu(s)$ as follows. First of all, if $m > 1$, we know that $q < 1$, so that there is positive probability for the total progeny to be infinite and therefore, it can not have finite expectation. In case $m \leq 1$, the special case $p_1 = 1$ is again trivial, because in that case $X_n \equiv 1$ for all $n \geq 1$, so that the total progeny is infinite with probability one and hence can not have finite expectation. So, we finally consider the case when $m \leq 1$ and $p_1 < 1$.

Since $q = 1$ in this case, the total progeny is finite with probability one and $\nu(s)$ is indeed its p.g.f. Now from (16), one gets $\nu'(s) = \phi(\nu(s))[1 - s\phi'(\nu(s))]^{-1}$ for $0 < s < 1$. Letting $s \uparrow 1$, one obtains that $\nu'(1) = \lim_{s\uparrow 1} \nu'(s)$ is finite and equals $(1-m)^{-1}$ if and only if $m = \phi'(1) = \lim_{s\uparrow 1} \phi'(s)$ is strictly less than 1. Thus we have proved

Theorem 1.15: *The total progeny of a branching process has finite expectation if and only if the mean m of the progeny distribution is strictly smaller than 1 and, in that case, the expected total progeny equals $(1-m)^{-1}$.*

1.7 Some Examples and Diverse Applications

In this section, we give some illustrations of how the theoretical model of branching process as described in the previous sections finds applications in a number of widely diverse situations.

(a) Survival of Family Names: Special cases of this application have already been discussed in detail following Theorem 1.8. As argued there, for this application, only male descendants count; they play the role of the objects and p_k is the probability for a newborn boy to become the progenitor of exactly k boys. Our scheme introduces two artificial simplifications. Fertility may not remain constant over generations and so the progeny distribution $\widetilde{p}$ changes, in reality, from generation to generation (see Section 1.8 (a)). Secondly, common inheritance and common environment are bound to produce similarities among brothers which is contrary to our assumption of stochastic independence. Our model can be refined to take care of these limitations, but the essential features remain unaffected. Our theory allows us to derive the probability of finding k carriers of the family name in the n-th generation, and, in particular, the probability of extinction of the family line. As noted before, survival of family names seems to have been the first chain reaction studied by probabilistic methods.

(b) Nuclear Chain Reactions: This application came into being in connection with the atomic bomb. The following description, as given in Feller, is supposed to be due to E. Schroedinger (1945). The objects here are neutrons, which are subject to chance hits by other particles. Once hit, a neutron creates k new neutrons. Denoting by α, the probability that a particle scores a hit sooner or later, we have a branching process with progeny distribution $\widetilde{p}$ given by $p_0 = 1 - \alpha$, $p_k = \alpha$ and $p_j = 0$ for all $j \neq 0, k$. At worst, the first particle remains inactive and the process never takes off. At best, there will be k particles of the first generation, k^2 particles of the second generation, and so on. If α is near one, the number of particles is likely to increase very rapidly. Of course, this model is simplistic. From the point of view of physical reality, for a very large number of particles, the probability of fission can not remain constant, and also, stochastic independence is impossible.

(c) Electron Multipliers: This is a variant of the above example and the

early treatments were given by Shockley and Pierce (1938) and Woodward (1948). For detailed reference, see the book of Harris. An electron multiplier is a device for amplifying a weak current of electrons. Each electron, as it strikes the first of a series of plates, gives rise to a random number of electrons, which strike the next plate and produce more electrons, and, so on. The number of electrons produced at successive plates have been treated as a Galton-Watson branching process. The complications due to random noise in the instrument can also incorporated in the model.

(d) Genes and Mutations: This application can perhaps be better understood after the reader has gone through Chapter 2 on general genetics. Every gene of a given organism has a chance to reappear in $1, 2, 3 \ldots$ direct descendants and our scheme describes the process, neglecting, of course, variations within the population and with time. Following R. A. Fisher, consider a corn plant which is father to some 100 seeds and mother to an equal number. Suppose that a spontaneous mutation in the plant had produced a single gene of a new kind, which plays the role of a 0-th generation object. We want to study the chances of survival and spread of this mutant gene. Now, if the population size remains constant, an average of 2 among the 200 seeds will develop to a plant, that is, each seed has a chance of $1/100$ to develop to a plant. Also, each seed has probability $1/2$ of receiving a particular gene. Thus the probability of the mutant gene being represented in exactly k new plants is the same as that of k successes in 200 independent Bernoulli trials with success probability $1/200$. it appears reasonable to assume the progeny distribution $\widetilde{p}$ is approximately a Poisson distribution with parameter 1.

(e) Waiting Lines: Interesting applications of branching proceses occur in queuing theory. The following is motivated by D. G. Kendall (see Feller for exact reference). Imagine a counter where customers arrive to get some kind of service. A customer arriving when the server is free, is attended to immediately; otherwise, he joins the queue (waiting line). The server continues service without interruption as long as there are customers in the queue requiring service. Let us assume for simplicity that customers can arrive only at integer time points and only one at a time. Suppose that the arrivals are regulated by Bernoulli trials, so that at any time point n, the probability that a customer arrives is p, while $1 - p$ is the probability that no arrival takes place. On the other hand, let us assume that the successive service times are independent integer-valued random variables with common distribution $\{\beta_k\}$ and p.g.f. $b(s) = \sum_{k \geq 1} \beta_k s^k$.

Suppose that a customer arrives at time 0 and finds the server free. His service time starts immediately. If it has duration k, the counter becomes free at time k, provided that no new customers have arrived at times $1, 2, \ldots, k$. Otherwise, the service continues without interruption. By "busy period" is meant the duration of uninterrupted service commencing at time 0. We show how the theory of branching processes may be used to analyze the duration of the busy period.

The customer arriving at time 0 initiates the busy period and will be called

the ancestor. The first generation consists of the customers arriving prior to or at the time of termination of the ancestor's service time. If there are no such direct descendants, then the process stops. Otherwise, the direct descendants are served successively, and during their service times, their direct descendants, if any, join the queue. We thus have a branching process such that the probability q of extinction equals the probability of termination of the busy period and the total progeny consists of all the customers (including the ancestor) served during the busy period. Needless to say, only queues with $q = 1$ are desirable in practice.

(f) Duration of the Busy Period: The preceeding example treats the number of customers during a busy period, but the actual duration of the busy period is of greater practical interest. This can be obtained by the elegant device, due to I. J. Good (see Feller for exact reference), of considering time units as elements of a branching process. We say that the time point n has no descendants if no new service starts at time n, where as, if a new service starts at time n and if this service lasts r time units, then the time points $n+1, n+2, \ldots, n+r$ are counted as direct descendants of time point n. Suppose that at time 0, the server is free. A little reflection shows that the branching process originated by the time point 0 (the ancestor) either does not come off at all or else lasts exactly for the duration of the uninterrupted service time initiated by a new customer; thus, the total progeny equals 1 with probability $1 - p$, while with probability p, it equals the duration of the busy period commencing at time 0.

1.8 Possible Generalizations and Extensions

(a) Variable Generating Functions: In our branching process model, it was assumed that all the objects always produce offsprings according to the same distribution. In particular, the offspring distribution remained the same over generations. To bring our model closer to reality, in some situations, we may instead allow generational variations and suppose that an object in the n-th generation produces offsprings according to the progeny generating function $\phi^{(n)}(s)$. In this case, the p.g.f. of X_n would be

$$\phi_n(s) = \phi^{(0)}(\phi^{(1)}(\cdots \phi^{(n-2)}(\phi^{(n-1)}(s)) \cdots)) . \tag{18}$$

The formulae (7) and (8) can be generalized in a similar way. The details can be found in the book of Harris.

(b) Family Trees: In our branching process as described in the previous sections, the sole consideration was the total number of objects in the successive generations. It does not distinguish between the different family trees, as long as the numbers in the various generations are the same. Consider, for example, the three families shown in the figure.
In all the three cases, $X_1 = 3$ and $X_2 = 2$. However, if we stipulate, regarding two dots linked to the same vertex, that the higher one corresponds to the older child, then (i), (ii) and (iii) represent three different family structures. It

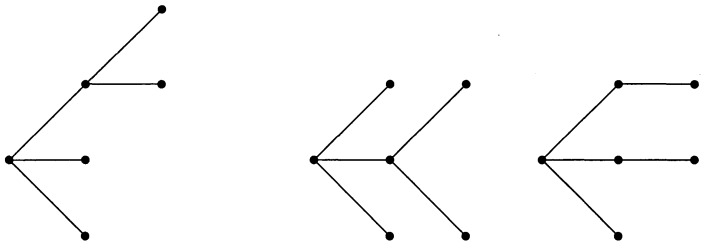

Figure 1.5: (i) (ii) (iii)

is possible to modify the definition of the branching process so as to keep track of the family trees. The book of Harris (pages 122-125) contains a detailed discussion.

(c) Multi-type Galton-Watson Process: Certain applications — for example, in cosmic ray cascade experiments, in reproduction of certain bacteria, etc. — require consideration of processes involving several type of objects. A Galton-Watson process with k types of objects, where k is a fixed positive integer, models a process of evolution where in each generation, objects of k different types may be present. Each object produces a certain number of offsprings of each type, thus giving rise to a k-dimensional random vector. Offspring distributions are therefore given by the joint p.g.f. of these random vectors. Denoting by $\phi_i(s_1, s_2, \ldots, s_k)$, $i = 1, 2, \ldots, k$ the p.g.f. of the offspring distribution of an object of type i, one can now define a Galton-Watson process $(X_n)_{n\geq 0}$, where, for each $n \geq 0$, X_n is the k-dimensional random vector representing the total number of objects of the k different types in the n-th generation. As before, it is again easy to see that $(X_n)_{n\geq 0}$ is a time-homogeneous Markov chain whose transition probabilities are completely determined by the functions ϕ_i. Once the initial confusion, if any, arising out of the simultaneous evolution of multiple types of objects disappears, the analysis can easily be seen to proceed in a manner analogous to the previous sections, and, one obtains generalizations of all the earlier results. Just to mention one, note that here, instead of a single mean m of the offspring distribution, we have a mean matrix $M = ((m_{ij}))$ of order $k \times k$, where m_{ij} denotes the expected number of offspring of type j produced by an object of type i. From the usual theory of matrices, one knows that if M is *positively regular* — that is, if for some integer N, M^N has all its entries strictly positive — then it has a strictly positive eigenvalue ρ, which is simple and greater in absolute value than any other eigenvalue. This eigenvalue ρ plays a role similar to that of m of previous sections, in determining extinction probabilities. The book of Harris (pages 34-49) contains a good account of such multi-type branching processes. A deeper analysis can be found in the book of C. J. Mode.

(d) Continuous Time, Age-dependent Branching Process: There are various possible formulations of branching processes in continuous time, some of which require rather involved analysis. We mention here the simplest one, namely, the *age-dependent branching process.* An object born at time 0 has a random life length L which has a distribution G. At the end of its life, it is replaced by a random number of similar objects of age 0, the probability being p_k that the number of new objects is k. The probabilities $p_k, k \geq 0$ are assumed not to depend either on the age of the object when it is replaced or on the number and ages of the other objects present. The process continues as long as a non-zero number of objects are present. Let X_t denote the number of objects present at time t. This will give us a continuous time process $(X_t)_{t\geq 0}$. In general, the process (X_t) is not markovian. However if the lifetime distribution G is exponential, then it does become a continuous time Markov process. It turns out that the p.g.f. ϕ of the distribution $\{p_k\}$ again plays a crucial role in the analysis of this process. Denoting the p.g.f. of X_t by ϕ_t, one can, for instance deduce the following analogue of (9)

$$\phi_t(s) = s(1 - G(t)) + \int_{(0,t]} \phi(\phi_{t-u}(s))dG(u)\,. \tag{19}$$

For this and a detailed analysis of the process $(X_t)_{t\geq 0}$, we refer the reader to the book of Harris.

1.9 Exercises

1. Let $(X_n)_{n\geq 0}$ be a branching process with $X_0 = 1$. For an arbitrary but fixed positive integer k, define the sequence $Y_r = X_{rk}, r = 0, 1, 2, \ldots$. Show that Y_r is a branching process. If ϕ is the p.g.f. of X_1, then show that ϕ_k, the k-th iterate of ϕ, is the p.g.f. of Y_1.

2. Let $f(s) = 1 - p(1-s)^\beta$ where p, β are constants and $0 < p,\ \beta < 1$. Show that f is a p.g.f. and its iterates are given by

$$f_n(s) = 1 - p^{1+\beta+\beta^2+\cdots+\beta^{n-1}}(1-s)^{\beta^n}.$$

for $n = 1, 2, 3, \ldots$.

3. Suppose that f is a p.g.f. Suppose that h is a function such that $g(s) = h^{-1}(f(h(s)))$ is well defined and is a p.g.f. Show that the n-th iterate of g is given by $g_n(s) = h^{-1}(f_n(h(s)))$. Verify that you can take $f(s) = \dfrac{s}{m-(m-1)s}$ and $h(s) = s^k$. Here k is a positive integer and $m > 1$. Show that

$$g_n(s) = \frac{s}{[m^n - (m^n - 1)s^k]^{1/k}}\,.$$

4. Recall that Z is the size of the total progeny (including the ancestor) in a branching process. Assuming only that $EZ < \infty$, show that $E(\sum_1^\infty X_i) = EX_1.EZ$. Conclude that $EX_1 < 1$ and $EZ = (1 - EX_1)^{-1}$.

5. In a branching process assume that the progeny distribution satisfies $p_i = 0$ for $i > 2$. Assume also that $p_2 > 0$. Show that the probability of extinction equals $\min(p_0/p_2, 1)$.

6. At time 0, a blood culture starts with one red cell. At the end of one minute the red cell dies and is replaced by 2 red cells *or* 1 red and 1 white cell *or* 2 white cells with probabilities 1/4, 2/3 and 1/12 respectively. Each red cell lives for one minute and gives birth to offspring in the same way while each white cell lives for one minute and dies without reproducing. Assume that individual cells behave independently. What is the expected number of white cells that have appeared (and died) by the end of 10 minutes. Show that the probability that the entire culture dies eventually is 1/3.

7. (a) A mature individual produces offspring according to the p.g.f. $\phi(s)$. Suppose that we have a population of k immature individuals each of which grows to maturity with probability p and then reproduces independently of other individuals. Show that the p.g.f. of the total number (immature) of individuals at the beginning of next generation is $(1 - p + p\phi(s))^k$.

   (b) Given that there are k mature individuals in the parent generation show that the p.g.f. of the mature individuals in the next generation is given by $[\phi(1 - p + ps)]^k$.

   (c) Show that the two p.g.f. in (a) and (b) have the same mean but not necessarily the same variance. Can you explain the discrepancy in the variance?

8. Consider a branching process with initial size N and p.g.f. $\phi(s) = q + ps$, where $0 < p < 1$, $q = 1 - p$. If T is the first time when the population becomes extinct then show that

$$P(T = n) = (1 - p^n)^N - (1 - p^{n-1})^N .$$

9. Fix $b > 0$, $c > 0$, $b + c < 1$. Consider the branching process with $X_0 = 1$ and progeny distribution given by $p_i = bc^{i-1}$ for $i = 1, 2, 3, \ldots$, $p_0 = 1 - \sum_{i=1}^{\infty} p_i$.

   (a) Show that $p_0 = \dfrac{1 - b - c}{1 - c}$.

   (b) Show that the p.g.f. is $\phi(s) = \dfrac{1 - (b + c)}{1 - c} + \dfrac{bs}{1 - cs}$.

   (c) Show that $m = \dfrac{b}{(1 - c)^2}$.

   (d) Show that if $s_0 = \dfrac{1 - b - c}{c(1 - c)}$, then $\phi(s_0) = s_0$.

From now on assume that $m \neq 1$.

(e) Show that $\dfrac{\phi(s) - s_0}{\phi(s) - 1} = \dfrac{s - s_0}{m(s-1)}$.

(f) Show that

$$\phi_n(s) = \frac{s_0 - \dfrac{s - s_0}{m^n(s-1)}}{1 - \dfrac{s - s_0}{m^n(s-1)}} = 1 - m^n \frac{1 - s_0}{m^n - s_0} + \frac{m^n \left(\dfrac{1 - s_0}{m^n - s_0}\right)^2 s}{1 - \left(\dfrac{m^n - 1}{m^n - s_0}\right) s}.$$

(g) Show that $P(X_n = 0) = 1 - m^n \dfrac{1 - s_0}{m^n - s_0}$.

(h) If T is the time of extinction, then show that

$$P(T = n) = m^{n-1} s_0 \frac{(m-1)(1-s_0)}{(m^n - s_0)(m^{n-1} - s_0)} \text{ for } n = 1, 2, \ldots .$$

From now on assume that $m = 1$.

(i) Show that $\phi_n(s) = \dfrac{nc - [(n+1)c - 1]s}{1 + (n-1)c - ncs}$.

(j) Show that $P(X_n = 0) = \dfrac{nc}{1 + (n-1)c}$.

(k) Show that $P(T = n) = \dfrac{c(1-c)}{[1 + (n-1)c][1 + (n-2)c]}$.

10. Suppose that a society adopts the following policy. Allow each couple to have two children. If both children are girls, then allow them to have a third child. Find the probability of survival of a given family name under the usual assumptions.

11. A man founds a society for the upholding of moral standards. Each year with probability p, he admits one person of sufficient high moral standard as a member. The probability is $1 - p$, that no new members are admitted in a year. At the same time, any existing member who is guilty of moral lapse must resign. The probability of this happening to a member in a year is λ. The founder himself is not considered a member of the society. Let ϕ_n denote the p.g.f. of the (random) number of members of the society at the end of n-th year. Show that

$$\phi_{n+1}(s) = (ps + 1 - p)\phi_n((1 - \lambda)s + \lambda).$$

Calculate $\phi_1(0)$ and $\phi_2(0)$.

12. Consider a queueing system as discussed in Example (e) of Section 7 and the branching process defined there. Find the progeny generating function $\phi(s)$ for this process. Deduce that $m = p\mu$ where μ is the expected duration of service for a customer. Find conditions for (a) the busy period to terminate with probability one and (b) the expected total number of customers during a busy period to be finite.

13. Consider the same set-up as in the above exercise, but now consider the branching process as discussed in Example (f) of Section 7. Find the progeny generating function. Hence find the generating function of the duration of the busy period.

1.10 References/Supplementary Readings

[1] Cox, D. R and Miller, H. D [1965] : *The Theory of Stochastic Processes*, Methuen Co, London.

[2] Feller, W. [1968] : *An Introduction to Probability Theory and its Applications*, vol. I, Third edition, John Wiley & Sons.

[3] Harris, T. E [1963] : *The Theory of Branching Processes*, Springer-Verlag.

[4] Karlin, S. [1966] : *First Course in Stochastic Processes*, Academic Press.

[5] Karlin, S. and Taylor, H. M. [1975] : *First Course in Stochastic Processes*, Second edition, Academic Press.

[6] Mode, C. J [1971] : *Multitype Branching Processes : Theory and Applications*, American Elsevier, New York.

All the material discussed in this chapter can be found in [2] and [3]. Interested reader can consult the other references for additional material as well as applications.

Chapter 2

BASIC MATHEMATICAL GENETICS

In this chapter, we start with a brief review of a few relevant facts from genetics, which is done in Section 1. We shall only recall just enough genetics that is needed in order to form an idea of the phenomena to be modelled in the subsequent sections. In Section 2, we proceed to the mathematical analysis of the variations in gene frequencies in a population. This includes, in particular, the *Hardy-Wienberg Laws.* Section 3 is devoted to a discussion of the phenomenon of *inbreeding* and the concept of *gene identity.* Malecot's models on *random mating* are taken up in the last section.

2.1 Basic Genetics

Interestingly, the first studies as to how traits or characterstics are passed on from parents to offsprings, were carried out by a saint in a monastery in Austria around the time 1860-1870; his name was Gregor Mendel. While experimenting with pea plants, he was intrigued by the following questions. How does a plant know whether it should be tall or dwarf, whether it should produce green seeds or yellow seeds, whether its seed coat should be grey or white? After a series of experiments, he arrived at the following conclusions. For each trait, there are what are called "determiners" — chemicals which make the plant exhibit that trait — and these determiners occur in pairs. During the formation of the reproductive cells, these pairs segregate or seperate out (*Law of segregation*), while during the fertilization process, one determiner from each parent join together, and it is this newly formed pair that is passed on to the offspring. This joining together does not take place according to any fixed preassigned plan; instead, one determiner from the father and one determiner from the mother join at random (*Law of Random Assortment*). Further the determiners for different characterstics are passed on independently (*Law of Independent Assortment*). This last speculation is, however, not entirely true.

Before going into the pertinent features of the offspring formation that are relevant for the mathematical analysis, we take a glimpse into cell biology and cell division. Human cell can be thought of as a miniature factory, where the raw material (food supply) is in the *cytoplasm* and the executives (that give instructions) are in the "air-conditioned" room called the *nucleus.* The nucleus is separated from the cytoplasm by a nuclear membrane. The nucleus contains, among other things, certain thread-like substances called *Chromosomes.* The chromosomes consist of what is known as *Chromatin* ("chroma" means colour; when a particular dye is applied, these threads pickup red colour and become visible in a microscope). In normal human cell, there are 23 pairs of such chromosomes in each cell nucleus, that is, 46 chromosomes arranged in pairs. Each pair is called a *homologous* pair. The chromosomes in each pair have the same physical appearance but not necessarily the same chemical composition. During the reproductive stage, a cell division called *Meiosis* takes place ("meioo" means reduction and "osis" means execessive growth, so that meiosis literally means reduction growth). This happens as follows. Firstly, the chromatin content doubles. This is called *interphase I.* Thus; 46 sister pairs of chromosomes appear. Then, in what is called *interphase II,* some fibres also start appearing. Then two sister pairs get attached to each fibre; this is called *metaphase I.* Then in *anaphase I,* the fibres break, leaving one sister pair attached to one part of the fibre. This is followed by *telephase I,* when constrictions appear in the cell and the cell slowly breaks into two cells. Now each of the cells has 23 sister pairs. Indeed, each has 23 of the original 46 chromosomes, but each in double dose. Then again *metaphase II* occurs, when each sister pair gets attached to a fibre. *Anaphase II* comes next, when each fibre breaks, leaving one of the chromosomes of the sister pair attached to one part of the fibre. Finally, in *telephase II,* each cell breaks into two, each having 23 chromosomes. Thus, starting from one original cell having 23 pairs of chromosomes, we finally have four cells, each having just 23 chromosomes. These are called *Gametes.*

Figure 1 illustrates the process of formation of gametes, starting from one original cell containing two pairs of chromosomes, namely the $(1a, 1b)$ pair and the $(2a, 2b)$ pair. It should be pointed out that the assortment taking place during anaphase I and telephase I is really random. Thus, starting with the same cell, as in Figure 2.1, the two cells obtained after telephase I could have been different as shown in Figure 2.2.

Though there are some structural differences between the male cell division and the female cell division at the reproductive stage, these differences do not concern our analysis. When a male gamete joins a female gamete, we get what is called a *Zygote.* Thus a zygote has 23 pairs of chromosomes. This zygote now divides and multiplies by the usual cell division process, called *Mitosis,* to form the offspring.

After this brief digression into cell biology and cell division, we now go on to discuss the salient features of basic genetics that are going to be relevant for us.

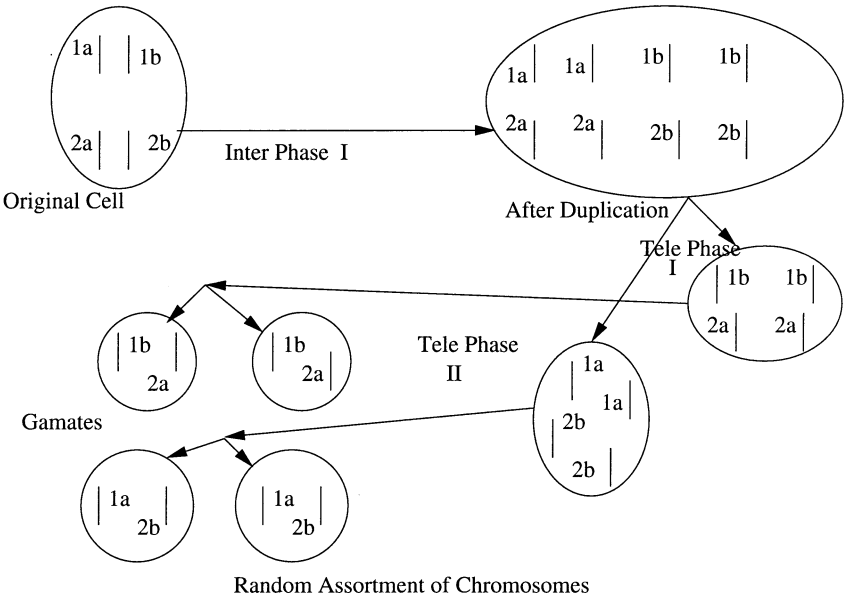

Figure 2.1: Cell Division

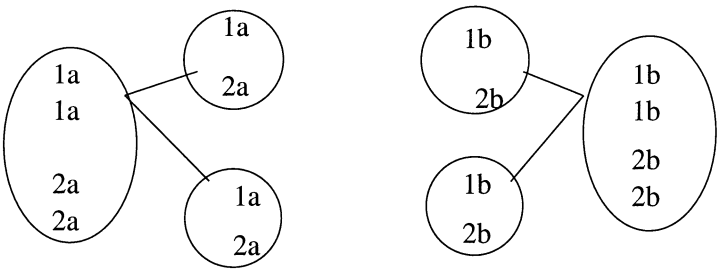

Figure 2.2: Chromosome Assortment

(1) The nucleus of a living cell contains a certain number of chromosomes. The number is the same for all individuals in a given species. In some species, they occur in pairs. Such species are called *Diploid Species.* But there are also species which are *Haploids* (chromosomes occuring singly), or *Triploids* (chromosomes occuring in triplets) and, more generally, *Polyploids.* We mostly consider diploid species. Among the different diploid species, the number of chromosome pairs vary widely. For example, human cells have 23 pairs, dogs have 39 pairs, fruitfly (Drasophila Melanogaster) has 4 pairs, pea plants have 7 pairs and so on.

(2) It is the chromosomes that govern the hereditary characterstics of the species. The quantity that governs a particular characterstic is called a *'Gene'* and the position on a chromososme where the gene is located is called the *Locus* of the gene. For example, in Drasophila, which has 4 pairs of chromosomes, a part of the third chromosome is the locus for 'hairy body' gene. This means that if a particular chemical is present at this locus, the fly will have hairy body. Loci appear in pairs on *homologous chromosomes* (an exception will appear later). In Drasophila, for example, consider the third homologous pair. The same position on each of the chromosomes of this pair is the locus for the 'hairy body' gene. The natural question that arises is that, if one chemical in one locus says 'yes hairy body' and the other one says 'no hairy body', then what happens? It depends on which is *dominant.* This is clarified next.

(3) The various forms in which a gene can occur are called *Alleles.* The different combinations of alleles that can occur for a particular gene on the homologous pair of chromosomes are known as the different *Genotypes.* The different ways in which the genotypes manifest themselves physically are called the different *Phenotypes.* Thus genotypes refer to the actual combinations of alleles of a gene, while phenotypes reflect their outward expressions. Let us look at some examples.

Example 1: For pea plants, consider the characterstic of height. The gene that determines this particular trait has two alleles. We denote them by T and t. The allele T dictates the plant to be 'tall', while the command of t is 'dwarf'. Now a plant can have any one of the three combinations — TT or Tt or tt — on its homologous pair. We are not distinguishing Tt from tT here. These are the three genotypes. Physically, however, it is found that both the TT and Tt combinations result in tall plants, whereas only the tt combination gives dwarfs. Thus there are only two phenotypes, namely, 'tall' and 'dwarf'. It is as if, the command of the allele T dominates over that of the allele t. Quite naturally therefore, in this case, the allele T is said to be *dominant* and the allele t is called *recessive.*

Example 2: The gene that determines the colour in snapdragon flowers has two alleles R (for 'red') and r (for 'no red'). As in Example 1, there are three genotypes, namely RR, Rr and rr. Plants which have RR produce red flowers, whereas plants that have rr produce white flowers. Plants of the genotype

Rr are found to produce pink flowers, intermediate to red and white. Thus, the three genotypes give rise to three different phenotypes also. So, here is a situation where out of the two alleles, no one seems to dominate over the other. In this case, we say that the alleles R and r are *codominant.*

Example 3: As a final example, we consider the Landsteiner blood group system. The gene that determines the blood group has three alleles, called O, A and B. So we have six different genotypes, namely, OO, OA, OB, AA, AB, BB. There are four phenotypes, namely, the L^O group (genotype OO), the L^A group (genotypes OA and AA), the L^B group (genotypes OB and BB) and the L^{AB} group (genotype AB). Here the alleles A and B are codominant whereas the allele O is recessive with respect to both A and B. Thus, genotypes OA and OB manifest in the same phenotypes as AA and BB respectively (namely, L^A and L^B groups respectively), but the genotype AB manifests in a different (intermediate?) phenotype, namely, the L^{AB} group. Of course, the phenotypic classification here is not as transparent as in the earlier examples. For example, pink is evidently intermediate to white and red, as in Example 2, but here what is L^{AB} group? Here is a brief description of the basis of this classification. In immunology one encounters two terms, *antigens* and *antibodies.* Antibodies are protiens in blood plasma manufactured to fight antigens, while antigens are those chemicals whose presence forces the body to manufacture antibodies to fight against these antigens (is it circular?). There are two different kinds of antigens called A and B that may or may not be present in blood. The basic rule is that, you will not manufacture antibodies to fight your own antigens. These four types of phenotypes described above correspond to blood having no antigens at all or having only antigen A or having only antigen B or having both A and B.

(4) A normal human cell contains 23 pairs of chromosomes. The chromosome pairs 1,2,...,22 are called *autosomes* (the chromosome pairs can be arranged and numbered). The 23rd pair is called the *sex-chromosome* pair. This pair consists of either two long ones or one long and one short. In particular, the two chromosomes in this pair may not even physically look alike. The long chromosome is called X and the short one is called Y. This pair determines the sex of a person in the following way: XX persons are females and XY persons are males. Thus, during reproduction, a mother always passes on an X chromosome to the offspring whereas the father may pass on either an X or a Y chromosome, thus determining the sex of the offspring. A gene that has its locus on one of the autosomes is called an *autosomal gene.* On the other hand, a gene that has its locus on the sex chromosome is called a *sex-linked gene.* More precisely, if a gene has its locus on the X chromosome, then it is called an *X-linked gene,* whereas genes having locus on the Y chromosome are called *Y-linked genes.* For example, the gene that determines the trait of colour blindness is an X-linked gene. This gene has two alleles, namely, C (normal vision) and c (colourblind). Thus males have two possible genotypes, namely, CY and cY, while females have three genotypes, namely, CC, Cc and cc. Between the two alleles C is found to be dominant and c recessive. Thus

CY, CC and Cc are the combinations that correspond to normal vision, while persons with genotypes cY and cc are colourblind. The gene that is responsible for what is known as *Haemophilia* (failure of blood-clotting mechanism), is also X-linked with two alleles, namely, H (normal) and h (haemophilic), of which H is dominant. On the other hand, traits like hairy ears, bald head, etc., are believed to be caused by Y-linked genes.

(5) We must immediately point out that every rule has exceptions. For example, sometimes a man may have what is called '*Trisomy 21*' or '*Down's syndrome*'; such a person has three chromosomes numbered 21, thus having a total of 47 chromosomes in all. Sometimes a female may have '*superfemale syndrome*'; having three X chromosomes. Similarly a man may have '*Klinefelter's syndrome*', having XXY. Also, it is not always true that the minimum number of chromosomes is 46. For example, a female may have '*monosomy X*' or '*Turner's syndrome*'; this means having only one X chromosome, thus having 45 chromosomes in all. Sometimes, all the 46 chromosomes may be there, but some of them may be partly missing. Sometimes, for example, the long arm of the 22-nd pair may be missing; this situation leads to a kind of cancerous Leukemia. Of course, all these are some sort of aberrations and will be excluded from consideration in the sequel.

(6) If parents pass on exact replica of what they have to the offspring, as enunciated earlier, then we will have an immediate contradiction to the evolutionary theory of Charles Darwin. In reality, there are sometimes misprints in passing on the message coded in a gene to an offspring. This phenomenon is called *Mutation*. For example, consider an autosomal gene with two alleles A and a. Suppose the father is (of genotype) AA and mother is (of genotype) aa. Then, if we strictly adhere to the theory laid down earlier, every offspring must receive an allele A from the father, and an allele a from the mother and consequently will be (of the genotype) Aa always. In practice, however, it is quite likely that during the gamete or zygote formation stage, the allele A may be 'converted' (with a very small but non-zero chance) to a. Such a conversion will result in the offspring being aa.

To understand the phenomenon of mutation a little better, let us consider the gene that controls the manufacture of haemoglobin. First of all, the chromosome has, besides sugar, nitrogen, etc., a sequence of chemicals acting as symbols for a code of instructions. The chemicals are Adenine symbolized as A, Guanine as G, Uracil or Thiamine as U and Cytosine as C. There are twenty basic amino acids used in the manufacture of proteins and other compounds. It will be helpful to think of a chromosome as a piece of paper with instructions written on it. In reality, these instructions are in the form of a sequence of symbols. Each triplet of these symbols A, G, U, C codes an amino acid (or does not code anything at all). Also, more than one triplet may code the same amino acid; this is called the *degeneracy of the genetic code*. For example, CAA and CAG both code the amino acid *Glutamine*, while GUC, GUA and GUG all code the amino acid *Valine*. When it is time to manufacture haemoglobin, the

relevant gene sends a message (actually, replica of itself) into the *protoplasm*, where the message is read tripletwise and the corresponding amino acids are brought and arranged. The gene that controls the manufacture of haemoglobin has many alleles, of which S (normal) and s (sickle) are two. The only difference between them is that, at a particular location in the string of triplets, the allele S has the code for glutamine, whereas s has the code for valine. Thus, a change of just one triplet would result in a change of an allele S into s (or of s into S). All this discussion is just to impress upon the reader that a mutation of one allele into another may often be accomplished by just a change of one single triplet in a string of thousands. Such a misprint is, of course, not hard to conceive. For example, it is found that radiation, an overdose of harmful chemicals, etc., are capable of causing this. When we talk of radiation, we do not necessarily mean being directly exposed to it. There are indirect ways of being affected by radiation. This could, for instance, be caused by drinking the milk of a cow which ate grass that somehow got contaminated by radiation fallout.

Thus, an allele A can mutate to an allele a and an allele a can mutate to A. In fact, mutation is the main source for genetic diversity. Sometimes, an allele mutates to something new giving rise to a slight variation. This leads to interesting theoretical possibilities as discussed in the next paragraph. If this new allele is 'good', the offspring and their progeny propogate this new allele in the population. If the new allele is deleterious, then it will disappear sooner or later from the population. Sometimes, this new allele may not only be good, but also advantageous for survival, in which case this new variation takes over. This is what is really behind the phenomenon of "survival of the fittest" in the context of evolutionary theory.

(7) As mentioned in the last paragraph, it is quite possible for an allele of a gene to mutate into a form that is not existing at present in the population. This means that the possible number of alleles for a gene may potentially be infinite (very very large), but only finitely many (in fact, only a few) show up (because we have a finite population). We shall not discuss this possibility of infinitely many alleles, though it is an exciting idea. We refer the interested reader to the book of Kingman.

This is all the basic genetics that will be needed to get the motivation and understand the mathematics that follows. If one is interested to know more on genetics, there are a number of good books, some of which are listed in the references at the end of this chapter.

We conclude this section with a simple illustration of how the above ideas work. Suppose that a woman has normal vision, but we are told that her father was colourblind. From this, can we say what the genotype of the woman is? First of all, since she has normal vision, her genotype must be either CC or Cc. But the fact that her father was colourblind, tells us that his genotype must have been cY. He must have passed on X (and not Y) to the daughter

and so must have passed on c. So the woman must have genotype Cc.

2.2 Random Mating: Hardy-Weinberg Laws

We shall now proceed towards some mathematical analysis. They concern the variations in gene frequencies from generation to generation, and are of theoretical interest.

2.2.1 Autosomal Genes

Let us consider a bisexual diploid population (like humans) and an autosomal gene with two alleles A and a. Suppose that, initially in both males as well as females in the populaion, the relative frequencies of the various genotypes are $AA : Aa : aa = u : 2v : w$ with $u + 2v + w = 1$. Such a population structure is sometimes denoted as $u\,AA + 2v\,Aa + w\,aa$. The question that we ask is: what will be the relative frequencies of various genotypes (*genotypic frequencies*, in short) in the next generation? Obviously, the genotypes of the offsprings of a mating depend on the specific genotypes of the parents. Thus, the relative frequencies of various mating types (*mating frequencies*, in short) are necessary. Let us assume that the population is paired into couples to act as parents of the next generation. The mechanism of the pairing is called a *mating system.* Thus an important constituent of any mathematical model would be a stipulation of the proportions of different mating couples. One such simple stipulation is that, for every choice of genotypes α and β, the proportion of mating couples with α type males and β type females — to be symbolically denoted as $\alpha M \times \beta F$ ($\alpha \times \beta$, in short) — equals the proportion of α type males in the population multiplied by the proportion of β type females in the population. Such a mating system is called *random mating.* The idea is that, if one male and one female are selected at random from the population for mating, then the chance of getting $\alpha \times \beta$ mating is given precisely by the product above. Since the two parents play symmetric role (autosomal gene!), there is no need to distinguish between the mating types $\alpha M \times \beta F$ and $\beta M \times \alpha F$. The two together will be denoted by $\alpha \times \beta$. Table 2.1 below lists the various mating types with their relative frequencies and the offspring genotype frequencies for each mating type.

Table 2.1

mating type	mating frequency	offspring genotypic frequency
$AA \times AA$	u^2	$1AA$
$aa \times aa$	w^2	$1aa$
$AA \times Aa$	$4uv$	$\frac{1}{2}AA + \frac{1}{2}Aa$
$aa \times Aa$	$4vw$	$\frac{1}{2}Aa + \frac{1}{2}aa$
$AA \times aa$	$2uw$	$1Aa$
$Aa \times Aa$	$4v^2$	$\frac{1}{4}AA + \frac{1}{2}Aa + \frac{1}{4}aa$

By the rule of total probability, the genotypic frequencies in the offspring population are $(u+v)^2AA + 2(u+v)(v+w)Aa + (v+w)^2aa$. Denoting $(u+v)$ by p and $(v+w)$ by q, these genotypic frequencies may be written as $p^2AA + 2pq\,Aa + q^2aa$. Now, if the same process is repeated to this population (to be called the first generation), then the second generation will consist of $\widetilde{p}^2AA + 2\widetilde{p}\widetilde{q}Aa + \widetilde{q}^2aa$ where $\widetilde{p} = p^2 + pq$ and $\widetilde{q} = pq + q^2$. But, of course, since $p+q=1$, we have $\widetilde{p} = p$ and $\widetilde{q} = q$. Thus, from the first generation onwards, all the subsequent generations will have composition $p^2AA + 2pq\,Aa + q^2aa$. In other words, the genotypic frequencies achieve stability right from the first generation, irrespective of what the composition of the initial population was.

Of course, in what we did above, random mating was the basic assumption. But, there were some other implicit assumptions too, which we now describe.

Firstly, the possibility of mutation was ruled out. For a given parental type, we simply followed the Mendelian rule to derive the offspring genotypic frequencies. If mutation is to be allowed, this has to be appropriately modified, resulting in a change in the third column of Table 2.1. An illustration of how to handle mutations in this set-up will be given later.

Secondly, *Natural Selection* was also ruled out. This means that we implicitly assumed all the genotypes to be equally fit to survive and reproduce. This, however, may not be true in reality. Consider, for example, a situation where the genotypes aa are sterile. Then, obviously, the mating types $Aa \times aa$, $AA \times aa$ and $aa \times aa$ do not produce any offsprings. Therefore, as far as next generation is concerned, we may as well pretend as if the aa genotype is dead from the parental population. So it will be just a matter of restricting oneself to a parental population with only two genotypes AA and Aa (with their relative frequencies appropriately normalized), and proceeding exactly as above. But this is only an extreme illustration. In general, we may only know that the aa genotypes are less fit than the AA. The main problem then is how to quantify the notion of "less fit". One way to do this is to say that only a proportion of the aa genotypes survive to maturity and reproduce. Naturally therefore, before discussing the mating types, we must first re-evaluate the relative frequencies of the different genotypes in the matured population. This is, for example, the case for sickle cell haemoglobin discussed in the earlier section. It is found that the fitness of the ss genotypes is much less compared to that of the SS genotypes. This is due to the fact that if the haemoglobin changes shape, then it cannot carry enough oxygen and, as a result, makes the person weak and intrinsically tired. Certain tribes in Africa are found to contain a considerable proportion of ss genotypes. In this context, we should warn the reader that reality is often much more complex than simple mathematical models. For instance, with sickle cell, the genotype SS is also "less fit" in the sense that it is more susceptible to certain kinds of malarial parasites.

Thirdly, we have implicitly assumed the population to be *closed*, that is, there is neither migration from the population nor any immigration from outside into the population. But, for our purposes, migration of a part of the

population can be thought of as death and brought under fitness assumptions. Similarly, immigration can also be handled via fitness.

Finally, we assumed that the different generations are *non-overlapping.* To be precise, we are trying to evaluate the frequencies of the various genotypes in the different generations. But, what is a generation? To start with, we assumed that there is a generation G_0, who produce the next generation G_1. Then matings from only G_1 produce the next generation G_2, and so on. In reality, however, this may not be the case. In certain societies, a man getting married to his sister's daughter is quite customary. Such systems do not fit into our model.

To make sense of random mating we must really assume that the male-female ratio in the population is 1:1. Also, the population must be potentially infinite. To see what happens otherwise, let us think of an extreme case. Suppose a population has 3 males and 3 females, each consisting of $1AA + 1Aa + 1aa$. Clearly all the six possible mating types listed in our Table 2.1, do not arise in reality. Our model of random mating precludes such possibilities. We do not need to make any additional explicit assumptions to this effect, because the notion of random mating itself takes care of it. Whatever be the underlying mechanism involved, the assumption of random mating describes to us the relative frequencies of various mating types, and, that is all that we need for our calculations. Thus, the notion of random mating is not really as simple as it appears to be. Later, we will see that, if the population is finite, then this kind of stationarity of genotypic frequencies cannot arise.

The assumption made about both sexes having the same genotypic frequencies to start with, is not essential. One can do away with it and proceed in exactly the same way as above to arrive at the following general result.

Theorem 2.1 (Hardy-Weinberg Law for Autosomal Genes): *Consider a diploid bisexual closed population and an autosomal gene with two alleles A and a. Assume that there is no mutation and that all genotypes are equally fit. Suppose that initially we have the genotypic frequencies $uAA + 2vAa + waa$ in males and $\overline{u}AA + 2\overline{v}Aa + \overline{w}aa$ in females. Then under random mating, the first generation consists of $p\overline{p}\,AA + (p\overline{q} + \overline{p}q)Aa + q\overline{q}\,aa$ in both males and females, where $p = u + v$, $\overline{p} = \overline{u} + \overline{v}$, $q = 1 - p$ and $\overline{q} = 1 - \overline{p}$. From the second generation onwards, the population consists of $p_0^2 AA + 2p_0q_0Aa + q_0^2 aa$ in both males and females, where $p_0 = \frac{1}{2}(p + \overline{p})$ and $q_0 = \frac{1}{2}(q + \overline{q})$. Thus, the genotypic frequencies remain stationary from the second generation onwards.*

To prove the above theorem, we may proceed as earlier, preparing a table giving offspring types. This method is called the 'random mating method' in the literature. There is another method of calculations, known as *random union of gametes*, which leads to the same final result. We shall illustrate this method now. Males consist of $uAA + 2vAa + waa$. Each AA male produces two A genes; each Aa male produces one A gene and one a gene; and each aa male produces two a genes. Thus, if you think of a male gene pool, then it consists of A genes and a genes in the proportion $pA + qa$, where $p = u + v$ and

$q = 1 - p$. Similarly, the female gene pool consists of $\overline{p}A + \overline{q}a$, where $\overline{p} = \overline{u} + \overline{v}$ and $\overline{q} = \overline{v} + \overline{w} = 1 - \overline{p}$. The principle of random union of gametes says that, the offspring genotype is obtained by selecting at random one gene from the male gene pool and one from the female gene pool. If we do this, then clearly the offspring population will consist of $p\overline{p}AA + (p\overline{q} + \overline{p}q)Aa + q\overline{q}aa$ for both sexes. Of course, it is legitimate to ask whether the model of random mating is equivalent to the principle of random union of gametes. This is, indeed, the case. The equivalence is discussed in detail in the book of Edwards.

2.2.2 X-linked Genes

Let us now consider an X-linked gene with two alleles A and a. As discussed earlier, the females, in this case, have three possible genotypes as usual, whereas the males have only two genotypes AY and aY (A and a, for short). Suppose that, initially females have the genotypic composition $uAA + 2vAa + waa$ and males have $p_0A + q_0a$. Assuming random mating, calculations similar to Table 2.1 show that, the first generation will have the genotypic compositions $p_0p_1AA + (p_0q_1 + q_0p_1)Aa + q_0q_1aa$ in females and $p_1A + q_1a$ in males, where $p_1 = u + v$ and $q_1 = 1 - p_1$. One can deduce, in general, that the nth generation will consist of $p_nA + q_na$ among males and $p_np_{n-1}AA + (p_nq_{n-1} + q_np_{n-1})Aa + q_nq_{n-1}aa$ among females. This leads to the recurrence relation

$$p_{n+1} = p_np_{n-1} + \frac{1}{2}(p_nq_{n-1} + q_np_{n-1}) = \frac{1}{2}(p_n + p_{n-1})\,.$$

Thus, to see what happens eventually, we should solve the difference equation above and find the limit as $n \to \infty$. Denoting $\alpha_n = p_{n+1} - p_n$, the above equation reduces to $\alpha_n = -\frac{1}{2}\alpha_{n-1}$. It follows that $\alpha_n = (-\frac{1}{2})^n\alpha_0$, for all $n \geq 1$, yielding $p_{n+1} = p_0 + \alpha_0 \sum_{k=0}^{n} (-\frac{1}{2})^k$. Taking limit as $n \to \infty$, we get

$$\lim_{n\to\infty} p_n = p_0 + \alpha_0 \sum_{k=0}^{\infty} (-2)^{-k} = p_0 + \frac{2}{3}\alpha_0 = \frac{2}{3}p_1 + \frac{1}{3}p_0\,.$$

We have thus proved

Theorem 2.2 (Hardy-Weinberg Law for X-linked Genes): *Consider a diploid bisexual closed population and an X-linked gene with two alleles A and a. Assume that there is no mutation and that all genotypes are equally fit. Suppose that the population initially consists of $p_0A + q_0a$ males and $u\,AA + 2v\,Aa + w\,aa$ females. Then, under random mating, the population would eventually consist of $\alpha\,A + (1-\alpha)\,a$ males and $\alpha^2 AA + 2\alpha(1-\alpha)Aa + (1-\alpha)^2 aa$ females, where $\alpha = \frac{2}{3}(u + v) + \frac{1}{3}p_0$.*

To see a concrete application of this theorem, suppose, for instance, that A is for normal vision and a for colourblindness. In the limiting population, if the proportion of colourblind males is β, then only a proportion β^2 of females are expected to be colourblind.

2.3 Random Mating with Mutations

Let us now turn to situations involving mutations. To make calculations simpler, we will work with the gamete pools and use the method of random union of gametes. Suppose that we have an initial population of gametes with the composition p_0A+q_0a and that, in every generation, a proportion μ $(0 < \mu < 1)$ of a genes mutate to A. If the nth generation consists of $p_nA + q_na$, then by assumption, $q_{n+1} = (1-\mu)q_n$. Thus, $q_n = (1-\mu)^n q_0 \to 0$ as $n \to \infty$, so that $p_n \to 1$. Thus, as expected, the population eventually consists entirely of A genes alone.

Let us now consider two-way mutations. Assume that, in each generation, a proportion μ $(0 < \mu < 1)$ of a genes mutate to A and a proportion γ $(0 < \gamma < 1)$ of A genes mutate to a. Of course, this raises the question as to whether it is possible for an a gene to mutate to A which again mutates immediately to a. Since the chances of mutation are usually very small, we assume that this does not happen. The second question that arises is about the order in which the two kinds of mutations take place. We assume both types of mutations to take place simultaneously, that is, a proportion of a genes mutate to A, and at the same time, a proportion of A genes mutate to a. As before, we start initially with a gene pool consisting of $p_0A + q_0a$. Now, if the nth generation consists of $p_nA + q_na$, then clearly, $p_{n+1} = (1-\gamma)p_n + \mu q_n = (1-\mu-\gamma)p_n + \mu$, or equivalently, $p_{n+1} - \frac{\mu}{\mu+\gamma} = (1-\mu-\gamma)(p_n - \frac{\mu}{\mu+\gamma})$. This gives

$$p_n = \frac{\mu}{\mu+\gamma} + (1-\mu-\gamma)^n \left(p_0 - \frac{\mu}{\mu+\gamma}\right).$$

Since $0 < \mu, \gamma < 1$, so that $|1-\mu-\gamma| < 1$, we have $p_n \to \frac{\mu}{\mu+\gamma}$ as $n \to \infty$. Thus, the population will eventually stabilize at $\frac{\mu}{\mu+\gamma}A + \frac{\gamma}{\mu+\gamma}a$, a configuration that does not depend on the initial structure. As a theoretical curiosity the reader may ask what happens if $\mu = \gamma = 1$?

2.3.1 Fitness Coefficient

We conclude this section with an illustration of how to take fitness constraints into account. Let us consider a diploid bisexual population and an autosomal gene with two alleles A and a. As usual, there are three genotypes in both males and females. Individuals with genotype AA or aa are referred in the literature as *Homozygotes*, while individuals with genotype Aa are referred as *Heterozygotes*. To start with, suppose that both males and females have genotypic frequencies $p_0^2AA + 2p_0q_0Aa + q_0^2aa$.

Let us now bring in a fitness assumption. We assume that the heterozygotes are more fit to survive than the homozygotes. More specifically, suppose that only a proportion σ of the AA genotypes and a proportion η of aa genotypes survive upto maturity. There is no restriction on the Aa genotypes, that is, they all survive. In the literature, this situation is referred to as *heterozygotic advantage*. Roughly speaking, the explanation is that if you have two

different alleles present, then you are fit to survive in any environment where any one of the alleles can survive; so you are better off than a homozygote. Thus, we assume that the "*fitness coefficients*" of the zygotes AA, Aa and aa are σ, 1 and η respectively with, of course, $0 < \sigma < 1$ and $0 < \eta < 1$. Unlike mutation, where the total population remains the same, fitness constraints bring in deaths, so that the population has to be renormalized. Thus, the matured initial population consists of $\frac{\sigma p_0^2}{\omega_0} AA + \frac{2p_0q_0}{\omega_0} Aa + \frac{\eta q_0^2}{\omega_0} aa$, where $\omega_0 = \sigma p_0^2 + 2p_0q_0 + \eta q_0^2$. Thus the gene pool in the matured initial population consists of $\frac{\sigma p_0^2 + p_0q_0}{\omega_0} A + \frac{p_0q_0 + \eta q_0^2}{\omega_0} a = p_1 A + q_1 a$, say. Then under random mating (equivalently, random union of gametes) and the usual conditions, the first generation would consist of $p_1^2 AA + 2p_1q_1 Aa + q_1^2 aa$, and the process repeats. Thus, if the nth generation initially consists of $p_n^2 AA + 2p_nq_n Aa + q_n^2 aa$, then the genotypic frequency of the $(n+1)$th generation, before maturity, would be $p_{n+1}^2 AA + 2p_{n+1}q_{n+1} Aa + q_{n+1}^2 aa$, where

$$p_{n+1} = \frac{\sigma p_n^2 + p_nq_n}{\sigma p_n^2 + 2p_nq_n + \eta q_n^2} \quad \text{and} \quad q_{n+1} = 1 - p_{n+1}\,. \tag{1}$$

To understand what happens eventually to the population, we have to know the dynamics of the map

$$f(x) = \frac{\sigma x^2 + x(1-x)}{\sigma x^2 + 2x(1-x) + \eta(1-x)^2}\,, \quad \text{for} \quad 0 \le x \le 1,$$

because Equation (1) simply says that $p_{n+1} = f(p_n)$. Routine algebra shows that

$$f(x) - x = \frac{x(1-x)(2-\sigma-\eta)(x^* - x)}{\sigma x^2 + 2x(1-x) + \eta(1-x)^2}\,, \tag{2}$$

where $x^* = \frac{1-\eta}{2-\sigma-\eta}$, from which it follows that the solutions of the equation $f(x) = x$ are precisely

$$x = 1, \quad x = 0 \quad \text{and} \quad x = x^*\,.$$

Thus, the only equilibrium states of the population are $1AA$, $1aa$ and $x^{*^2} AA + 2x^*(1-x^*)Aa + (1-x^*)^2 aa$. That is, if the population is in any one of these states, then it remains so forever. In other words, if p_0 takes any one of the three values 1 or 0 or x^*, then $p_n = p_0$ for all n. We shall now show that for any value of p_0 with $0 < p_0 < 1$, the population reaches the third equilibrium state eventually, that is, $p_n \longrightarrow x^*$ as $n \to \infty$. Thus the first two equilibrium states, namely, $p_0 = 1$ and $p_0 = 0$, are unstable equilibria, meaning that a slight perturbation from these equilibria causes the population to drift away from them. The third equilibrium state is, of course, a (global) stable equilibrium, meaning that starting from any other non-equilibrium state, the population gets drawn to this state eventually. Note that the third equilibrium state is one in which both genes are present, that is, a *polymorphic* state.

To prove the result claimed above, first note that (2) implies that for all x with $0 < x < x^*$, $f(x) - x > 0$, so that $f(x) > x$. Moreover, since $2 - \sigma - \eta < 2$,

we have $x(1-x)(2-\sigma-\eta) < 2x(1-x)$, whereas, $\sigma x^2 + 2x(1-x) + \eta(1-x)^2 > 2x(1-x)$. Thus, the coefficient of $(x^* - x)$ on the right hand side of (2) is strictly smaller than 1, giving us $f(x) - x < x^* - x$, and hence $f(x) < x^*$. Thus we have, for all x with $0 < x < x^*$, $x < f(x) < x^*$. Now, let $p_0 = x$ for any x with $0 < x < x^*$, and define $p_n = f(p_{n-1})$ for $n \geq 1$. Then $\{p_n\}$ forms an increasing sequence and hence has a limit $\overline{p}$, say. Since $p_n = f(p_{n-1})$ for all n, and f is continuous, this limit $\overline{p}$ must satisfy $\overline{p} = f(\overline{p})$. Of course, $\overline{p} > p_0 > 0$. Also $p_n < x^*$ for all n, so that $\overline{p} \leq x^*$. All these imply that $\overline{p}$ must equal x^*. A similar argument can be used for the case when $x^* < p_0 < 1$. Of course, if $p_0 = x^*$, then $p_n = x^*$ for all n, so that $p_n \to x^*$ trivially. This completes the proof.

It is possible to treat mutation and fitness constraints simultaneously. Most of what has been discussed in this section may also be found in the books of Feller, Karlin, Edwards and Jacquard. Incidentally, Jacquard's book considers other mating systems also.

2.4 Inbreeding – Autozygosity

So far, we have been preoccupied with random mating. It is time that we discussed some other mating systems and try to see what happens with such systems. In particular, we now consider populations that are inbred. We would somehow try to measure the extent of inbreeding. In this context, we now introduce an important concept, namely, that of *genes identical by descent.*

2.4.1 Genes Identical by Descent

If a system is inbred, then the same genes should keep on appearing over generations. After all, offsprings get their genes from their parents, who in turn get theirs from their parents, and so on. Inbreeding roughly means mating between close relatives. Thus, if you take a mating couple, they have some common ancestor, whose gene they are both likely to be carrying and are likely to pass on that common gene to an offspring. In that case, the two genes the offspring receives are just two copies of the same gene of an ancestor in the family. We say that this offspring has genes *identical by descent.* Thus, an individual of genotype $\alpha\beta$ is said to have *genes identical by descent* if, α and β are just replicas of the same single gene passed on by an ancestor. Note that this is much more than saying that $\alpha\beta$ is a homozygote. To clarify ideas, let us look at an example illustrated in Figure 2.3.

Consider a parent F (father) having $\alpha\beta$ as a homologous pair of genes on chromosome 1 and its homologue $\tilde{1}$. Suppose that he has three offsprings named I, II and III; and that they had their genotypes formed as follows. The gene α from F's chromosome 1 is passed on to the chromosome 1 of the offsprings I and III while the offspring II, on its chromosome 1 gets the gene β from F's chromosome $\tilde{1}$. For all three of them, mother's gene gets passed on to

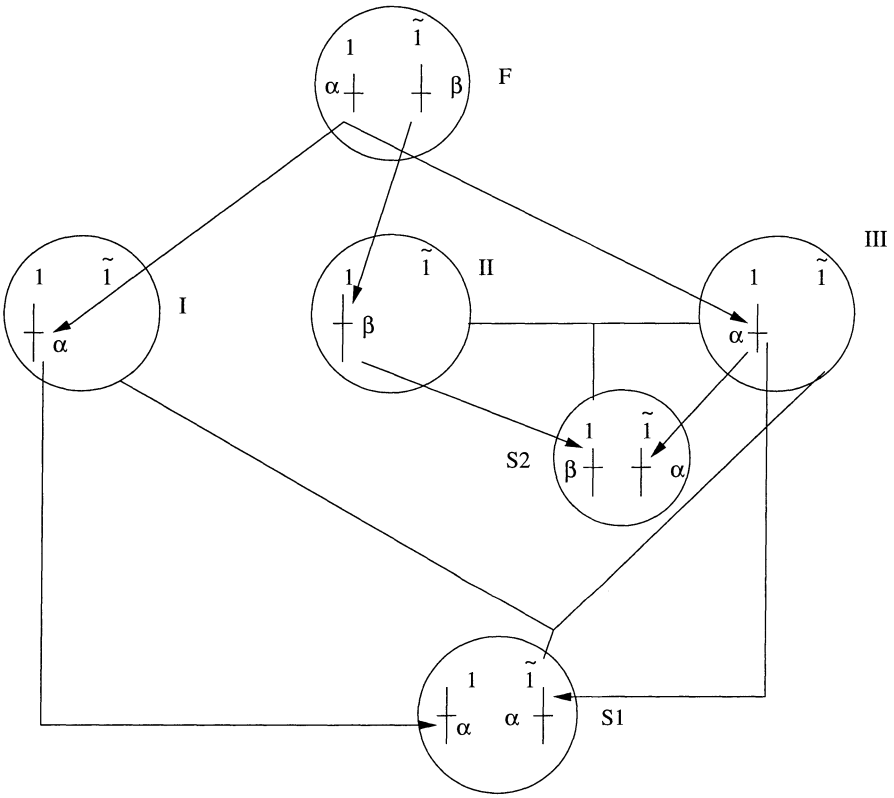

Figure 2.3: Genes Identical by Descent

chromosome $\tilde{1}$. Suppose now, that a mating of I and III produces S_1, and that, to this offspring, both I and III pass on their α gene from chromosome 1. Thus S_1 will have genotype $(F1\alpha, F1\alpha)$. If, on the other hand, II and III mate to produce S_2 and if again, to this offspring, both pass on genes from their chromosome 1, then S_2 has genotype $(F\tilde{1}\beta, F1\alpha)$. By the definition given above, S_1 clearly has genes identical by descent. The ancestor F is a proof of this. On the other hand, S_2 does not have genes identical by descent unless F has so. Of course, S_2 is always a homozygote if F is.

Let us now try to understand why this concept of 'genes identical by descent' is important. So far we have distinguished different alleles of a gene, by using different names, for example, A and a, but have not distinguished one A from another. This may have seemed alright so far. After all, are all the A not the same? Strictly speaking, the answer is NO. To understand this, consider the gene responsible for the manufacture of haemoglobin. The allele A causes manufacture of normal haemoglobin. So A actually consists of a sequence of triplets of A, G, U and C. As we already know, triplets code amino acids, but for many amino acids, there are different codes. Because of this, there can be two entirely different sequences of triplets, which have the same functional property, that is, they decode to the same sequence of amino acids. Thus, when we talk of genes identical by descent, we demand that, in particular, they carry the same sequence of triplets. Another reason for considering this concept is that this is one method of investigating the extent of inbreeding in a population. This point will be explored shortly. You may, of course ask: how does one find out, for a given individual, whether genes are identical by descent? How does one go back in his family line and produce proof for such identicality? Well, we do not. This is not a laboratory concept. We make some assumptions and proceed. The end results turn out to be appealing.

2.4.2 Coefficient of Parentage and Inbreeding Coefficient

Following Malecot and Wright, we now define the concept of *coefficient of parentage* between two individuals. If I and J are two individuals of type $\alpha\beta$ and $\gamma\delta$ respectively, then we define the coefficient of parentage between I and J to be

$$\phi_{IJ} = \frac{1}{4}[P(\alpha \equiv \gamma) + P(\alpha \equiv \delta) + P(\beta \equiv \gamma) + P(\beta \equiv \delta)]\,.$$

Here '$\alpha \equiv \gamma$' means that α and γ are identical by descent. In other words, if you select at random one from the two genes of I and one from the two genes of J, then ϕ_{IJ} is precisely the chance that the selected genes are identical by descent. Note that if the individuals I and J mate to produce an offspring, then the possible genotypes of the offspring are $\alpha\gamma$, $\alpha\delta$, $\beta\gamma$ and $\beta\delta$, each with probability $\frac{1}{4}$. Therefore ϕ_{IJ} equals the probability that this offspring has genes identical by descent. This leads us to make the following definition. If S is an individual with parents I and J, then the *coefficient of inbreeding* of S,

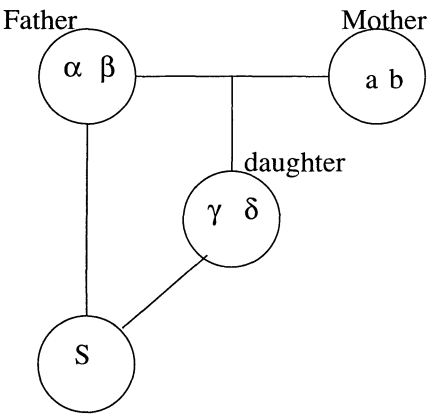

Figure 2.4: φ_{FD}

denoted by F_S, is defined to be the coefficient of parentage between its parents I and J, that is,

$$F_S = \phi_{IJ}\,.$$

Of course, as noted already, F_S gives the chances that S has genes identical by descent. We shall now put these ideas to work through some examples.

Example 1: Consider a plant F of type $\alpha\beta$. Suppose we do *selfing*, that is, mate the plant with itself. This is possible with plants. What is ϕ_{FF}? To apply the definition, it should first be noted that, here γ is same as α and δ is same as β, that is, $\gamma \equiv \alpha$ and $\delta \equiv \beta$. Suppose first that α and β are identical by descent. Then, clearly, $\phi_{FF} = \frac{1}{4}(1+1+1+1) = 1$. If, on the contrary, α and β are known to be not identical by descent, then $\phi_{FF} = \frac{1}{4}(1+0+0+1) = \frac{1}{2}$. On the other hand, if instead of having any definite information about α and β, we only knew that the chances of their being identical by descent is p, then by the rule of total probability, one obtains $\phi_{FF} = 1.p + \frac{1}{2}.(1-p) = \frac{1}{2}(1+p)$.

Example 2: Let us consider a father F and his daughter D as illustrated in Figure 2.4. We wish to calculate ϕ_{FD}. Let the father be of type $\alpha\beta$, the mother of the type ab and the daughter of the type $\gamma\delta$, with γ being obtained from the father and δ from the mother. Let an offspring of F and D be denoted by S. If, to start with, we know that all the four genes α, β, a, and b are identical by descent, then it is clear that $\phi_{FD} = 1$. However, we may not have any definite information to start with. In such situations, the convention will be to assume that the genes are not identical by descent. Thus, we assume here that no two of the genes α, β, a and b are identical by descent. You will, of course, agree that it makes no sense to make the same assumption about, say, the genes α, β, γ and δ. After all, we do know that one of α, β is definitely identical to γ. In view of this last fact, we have $P(\alpha \equiv \gamma) + P(\beta \equiv \gamma) = 1$, and, of course, by our assumption $P(\alpha \equiv \delta) + P(\beta \equiv \delta) = 0$. This gives $\phi_{FD} = \frac{1}{4}$. Note that, in

this calculation, the genotype of the mother turns out to be of no relevance at all.

2.4.3 Selfing

As explained earlier, *Selfing* means mating a plant with itself. You may wonder why we are considering populations other than human, and, even if we do, why we are interested in such mating systems. Well, this is the simplest kind of inbreeding and is of theoretical interest. An understanding of simple phenomenon leads to some grasp and control on the more complicated phenomena.

So, let us start with a plant population. In the absence of any prior information, we assume that F_0, the inbreeding coefficient of any individual in the initial population, is zero; that is, no plant in the initial population has genes identical by descent. Do selfing and get generation 1; do selfing of plants of generation 1 and get generation 2, and so on. Let F_n denote the inbreeding coefficient of generation n. This makes sense, because whatever individual you take in the nth generation, its inbreeding coefficient is the same; it depends only on n and not on the individual. This is so, because we are following a regular pattern. If you do not agree now, just wait and convince yourself that, in our calculation of F_n, the individual is irrelevant. We first claim that $F_1 = \frac{1}{2}$. Indeed, take any individual S of the first generation, whose parent in generation 0 is of type $\alpha\beta$, say. By assumption, α and β are not identical by descent. Therefore, the only way S can have genes identical by descent is by being $\alpha\alpha$ or $\beta\beta$, and the probability of this is $\frac{1}{2}$. In general, by the last remark in Example 1 of the previous section, it follows that

$$F_{n+1} = \frac{1 + F_n}{2}, \quad \text{for } n \geq 0\,.$$

Using $F_0 = 0$, the unique solution of the above equation is given by

$$F_n = \frac{1}{2} + \ldots + \frac{1}{2^n} = 1 - \frac{1}{2^n}, \quad \text{for } n \geq 0\,.$$

Thus, in particular, as $n \to \infty$, $F_n \longrightarrow 1$, at a geometric rate. Often, one talks about what is called the *panmictic index*, denoted by P_n, which is defined as $1 - F_n$. 'Pan' means 'all' and 'mictic' means 'mixing'; thus 'panmictic' means 'all mixing'. This is an index that tells us how much the population is mixing. Clearly, in this case, $P_n \longrightarrow 0$ as $n \to \infty$. Thus, the net result is that eventually all the genes will be identical by descent in each individual; in particular, the population will become homozygous.

2.4.4 Sibmating

SIB is an abbreviation for siblings, brothers and sisters. So the mating system to be considered now is one in which only brothers and sisters mate. Such a mating system is known as *Sibmating*. Start with a population, to be called

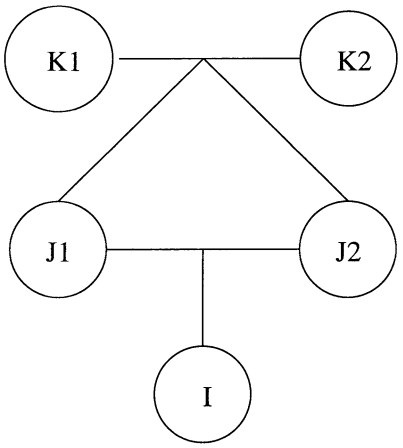

Figure 2.5: Sibmating

generation 0. Assume that, initially no two genes in the *entire population* are identical by descent. In particular, $F_0 = 0$. Here F_n for $n \geq 0$, as in Section 2.4.3, stands for the inbreeding coefficient for the nth generation. Apply sibmating on generation 0 to get generation 1. Of course, you may say that, if we know nothing about generation 0, then how do we know who the sibs are. Well, our answer is that in that case, use any mating system (other than selfing) to get generation 1. Convince yourself that in any case $F_1 = 0$ and hence does not make any difference in the sequel. Now mate siblings in generation 1 to get generation 2, and so on. Thus, for all $n \geq 2$, generation n is obtained by mating of siblings of generation $(n-1)$. We now proceed to obtain a recurrence relation between the successive F_n. We start with a few observations.

Fact 1: For any individual K,

$$\phi_{KK} = \frac{1}{2}(1 + F_K)\,. \tag{3}$$

This is just what was observed in Example 1 of Section 2.4.2; the p there is precisely F_K by definition.

Fact 2: Let $\langle KL \rangle$ denote an offspring of K and L. Then for any individual M,

$$\phi_{\langle KL \rangle M} = \frac{1}{2}(\phi_{K,M} + \phi_{L,M})\,. \tag{4}$$

The proof of this is left as an exercise. One has to just apply the definition and simplify. It should be noted that M in (4) can be any individual, possibly even K or $\langle KL \rangle$.

Let us now go back to our problem. Fix $n \geq 2$, and let I be an individual in generation n with parents J_1 and J_2 in generation $(n-1)$. Note that J_1 and

J_2 are sibs, so that they have common parents, say, K_1 and K_2 in generation $(n-2)$. Then, $F_n = F_I = \phi_{J_1J_2} = \phi_{\langle K_1K_2\rangle J_2} = \frac{1}{2}(\phi_{K_1J_2} + \phi K_2J_2)$ by (4) above. Now write J_2 as $\langle K_1K_2\rangle$ and apply (3) and (4) to get

$$F_n = \frac{1}{4}(\phi_{K_1K_1}+\phi_{K_1K_2}+\phi_{K_2K_1}+\phi_{K_2K_2}) = \frac{1}{4}\left(\frac{1+F_{K_1}}{2} + 2\phi_{K_1K_2} + \frac{1+F_{K_2}}{2}\right).$$

Now, both K_1 and K_2 are from generation $(n-2)$, so that $F_{K_1} = F_{K_2} = F_{n-2}$. Also $\phi_{K_1K_2} = F_{\langle K_1K_2\rangle} = F_{n-1}$. These give us the recurrence relation

$$F_n = \frac{1}{4}(1 + F_{n-2} + 2F_{n-1}),$$

or

$$4F_n - 2F_{n-1} - F_{n-2} = 1\,.$$

This (non-homogeneous) difference equation can be solved as follows. We first make the substitution $F_n = 1 + \alpha_n$ to convert this into the homogeneous equation

$$\alpha_n - \frac{1}{2}\alpha_{n-1} - \frac{1}{4}\alpha_{n-2} = 0\,.$$

The characterstic polynomial of this equation is $x^2 - \frac{1}{2}x - \frac{1}{4}$, whose two roots are given by $\frac{1}{4}(1\pm\sqrt{5})$. So the general solution of the above difference equation is

$$\alpha_n = c_1\left(\frac{1+\sqrt{5}}{4}\right)^n + c_2\left(\frac{1-\sqrt{5}}{4}\right)^n.$$

Now note that the panmictic index $P_n = 1 - F_n = -\alpha_n$. So writing d_1 and d_2 for $-c_1$ and $-c_2$ respectively, we obtain

$$P_n = d_1\left(\frac{1+\sqrt{5}}{4}\right)^n + d_2\left(\frac{1-\sqrt{5}}{4}\right)^n.$$

Now, we apply the initial condtions $F_0 = F_1 = 0$ (equivalently, $P_0 = P_1 = 1$) to get $d_1 + d_2 = 1$ and $(1+\sqrt{5})d_1 + (1-\sqrt{5})d_2 = 4$. These yield

$$d_1 = \frac{\sqrt{5}+3}{2\sqrt{5}} \quad \text{and} \quad d_2 = \frac{\sqrt{5}-3}{2\sqrt{5}}\,.$$

Thus, P_n (and hence, F_n) is completely determined. Note that, just like in the case of selfing, here also $P_n \to 0$ as $n \to \infty$ and the convergence is still geometric, though now at the rate $(\frac{1+\sqrt{5}}{4})^n$, as compared to 2^{-n} in selfing. Thus, eventually each individual in the population will have genes identical by descent. In fact, eventually all the genes (at that locus) in the population will be identical by descent (why?). In particular, the population would become homozygous.

Thus, in both selfing as well as sibmating systems, the panmictic index P_n goes to 0, as n tends to ∞, at a geometrically fast rate, but the rate of

convergence is $\frac{1}{2^n}$ in the case of selfing and $(\frac{1+\sqrt{5}}{4})^n$ in the case of sibmating. Since $\frac{1+\sqrt{5}}{4} > \frac{1}{2}$, we conclude that in the latter case, the convergence of P_n to 0 is slower. This phenomenon has an interesting extension.

Let us call two individuals to be *0th order cousins*, if they have the same parents. Thus sibs are 0th order cousins. In general, *mth order cousins* can be defined as follows. Individuals A and B are mth order cousins, if one parent of A and one parent of B are $(m-1)$th order cousins. One can then show that, in a system where only mth order cousins mate, the panmictic index P_n goes to zero as n goes to infinity and the convergence is geometric; however, the rate becomes slower with increasing m. Thus, for example, in a first order cousin mating system, P_n goes to zero at a rate slower than that in sibmating and, in a 7th order cousin mating system, the rate is much much slower. It is, therefore, no wonder why in some societies, mth order cousins with $m \leq 6$, are forbidden to marry. The idea is to delay the process of homozygosity or equivalently, (as explained earlier) to increase the fitness of the offspring.

2.5 Random Mating in a Finite Population

The above discussions underline the fact that in regular inbreeding systems, the population will eventually consist of *autozygous* individuals, that is, individuals carrying genes identical by descent. In a sense, this feature persists even under random mating as long as we are dealing with finite populations. This striking observation was made by R.A.Fisher, S.Wright and G.Malecot. We shall follow Malecot.

2.5.1 Constant Population Size Model

Imagine a diploid bisexual population, where the number of males and the number of females, M and $\widetilde{M}$ respectively, remain constant over generations. The mating system we consider is as follows. The genotype of an individual in any generation is determined by taking one gene at random from each of a randomly chosen male and a randomly chosen female of the previous generation. $(M + \widetilde{M})$ such independent choices, with replacement, create the M males and the $\widetilde{M}$ females of the next generation. Equivalently, in any generation, consider the $2M$ male genes and the $2\widetilde{M}$ female genes. Independent random selection of one gene from each of the two gene pools determines the genotype of an individual in the next generation. M such independent choices, with replacement, create the M males of the next generation and $\widetilde{M}$ selections create the $\widetilde{M}$ females. We shall refer to this mating system as *random mating for finite populations.*

Denote by F_n, the inbreeding coefficient for generation n. We now proceed to obtain a recurrence relation for the F_n. Let us consider an individual in the nth generation, say, A with parents B and C in the $(n-1)$th generation. We claim that the chances of A getting both his genes from one single person of the

$(n-2)$th generation is $\frac{1}{4M} + \frac{1}{4\widetilde{M}}$. To prove this, we argue as follows. Let us fix an individual, say L, in the $(n-2)$th generation and ask, what the probability is for A to get both his genes from L? If L is a male, then this happens if and only if L is the father of both B and C and both pass on to A, the genes they received from L. Clearly, the chance of this happening is $\frac{1}{M} \times \frac{1}{M} \times \frac{1}{2} \times \frac{1}{2} = \frac{1}{4M^2}$. Similarly, if L is a female, the chance would simply be $\frac{1}{4\widetilde{M}^2}$. Since the $(n-2)$th generation consists of M males and $\widetilde{M}$ females, we conclude that the probability of A getting both his genes from the same individual in the $(n-2)$th generation to be $M \times \frac{1}{4M^2} + \widetilde{M} \times \frac{1}{4\widetilde{M}^2} = \frac{1}{4M} + \frac{1}{4\widetilde{M}}$, as asserted.

Given that A gets his genes from the same individual in the $(n-2)$th generation, the chances of A having genes identical by descent is $(1+F_{n-2})/2$, by Example 1 of Section 2.4.2. In case A got his genes from two different individuals of the $(n-2)$th generation, the chances of A having genes identical by descent would just be equal to the coefficient of parentage of those individuals (in the $(n-2)$th generation), which, in turn, is same as the inbreeding coefficient of their offspring (in the $(n-1)$th generation), namely F_{n-1}. Thus, we obtain

$$F_n = \left(\frac{1}{4M} + \frac{1}{4\widetilde{M}}\right)\left(\frac{1+F_{n-2}}{2}\right) + \left(1 - \frac{1}{4M} - \frac{1}{4\widetilde{M}}\right) F_{n-1}\,.$$

Let us denote $\frac{1}{N} = \frac{1}{4M} + \frac{1}{4\widetilde{M}}$, that is, N is the harmonic mean of $2M$ and $2\widetilde{M}$. N is usually called the *effective sample size* in this context. Thus the above equation can be written as

$$F_n = \frac{1}{2N}(1+F_{n-2}) + \left(1 - \frac{1}{N}\right) F_{n-1}\,.$$

Letting $P_n = 1 - F_n$ as usual, we have

$$P_n = \left(1 - \frac{1}{N}\right) P_{n-1} + \frac{1}{2N} P_{n-2}\,.$$

This homogeneous linear difference equation is solved by using the standard method of characterstic polynomial. The roots of the characterstic polynomial $x^2 - (1 - \frac{1}{N})x - \frac{1}{2N}$ in this case are given by

$$x_1 = \frac{1}{2}\left[1 - \frac{1}{N} + \sqrt{1 + \frac{1}{N^2}}\right] \quad \text{and} \quad x_2 = \frac{1}{2}\left[1 - \frac{1}{N} - \sqrt{1 + \frac{1}{N^2}}\right],$$

and therefore the general solution of the difference equation is given by

$$P_n = \alpha x_1^n + \beta x_2^n\,.$$

The coefficients α and β are determined by the initial conditions. For example, if we assume that, to start with, no two genes in the initial population are identical by descent, that is, $P_0 = P_1 = 1$, then we get $\alpha + \beta = 1$ and $\alpha x_1 + \beta x_2 = 1$, determining α and β.

In any case, since both x_1 and x_2 are (in absolute value) strictly less than one, it is clear that $P_n \to 0$ as $n \to \infty$. Thus, eventually the population consists only of *autozygotes*, that is, individuals carrying genes identical by descent. In fact, something much stronger is true, namely, that eventually only one of the initial genes survives and appears in all individuals. But this requires Markov chain methods, which will be taken up in the next chapter. Needless to say that this is interesting because, at each stage, we are selecting at random from the existing genes. It is the finiteness of the population which somehow forces it to proceed towards autozygosity. Again, from the view point of Markov chains, this is not all that surprising.

2.5.2 Varying Population Size Model

Even though we agree to work with finite populations only, the assumption in Section 2.5.1, that is, the number of males and females remain constant from generation to generation, undoubtedly looks artificial. Let us, therefore, relax this assumption, and, let these numbers vary with generation, with M_n and $\widetilde{M}_n$ denoting the number of males and females respectively for the nth generation. We consider the same mating system as in the previous section and, denote by F_n and P_n, the inbreeding coefficient and the panmictic index respectively, for the nth generation. Then, by the same argument as before, we obtain here

$$P_{n+2} = \left(1 - \frac{1}{N_n}\right) P_{n+1} + \frac{1}{2N_n} P_n, \tag{5}$$

where $N_n = \left(\frac{1}{4M_n} + \frac{1}{4\widetilde{M}_n}\right)^{-1}$ denotes the effective population size of the nth generation.

As usual, we assume that $P_0 = P_1 = 1$. It now follows from (5), by induction, that $P_n > 0$ for all n. Of course, we cannot explicitly hope to solve for P_n any longer. Let us put $P_n = a_0 \cdot a_1 \cdot \cdots \cdot a_n$, that is, we put $a_n = \frac{P_n}{P_{n-1}}$ for $n \geq 2$ and $a_0 = a_1 = 1$. We then claim that, for each n, $\frac{1}{2} \leq a_n \leq 1$. This is, of course, true for $n = 0, 1$. Now by Equation (5),

$$a_{n+2} = \frac{P_{n+2}}{P_{n+1}} = \left(1 - \frac{1}{N_n}\right) + \frac{P_n}{P_{n+1}} \frac{1}{2N_n} \geq 1 - \frac{1}{N_n} \geq \frac{1}{2},$$

the last inequality being a consequence of the fact that $N_n \geq 2$. Also

$$a_{n+2} = \left(1 - \frac{1}{N_n}\right) + \frac{1}{a_{n+1}} \frac{1}{2N_n} \leq \left(1 - \frac{1}{N_n}\right) + 2\frac{1}{2N_n} = 1,$$

thus proving the claim.

In view of the inequality $a_n \leq 1$, the sequence $(P_n)_{n\geq 0}$ is non-increasing in n, and, therefore has a limit, say, P_∞. Moreover, noting that $a_0 = a_1 = 1$, we have

$$P_\infty = \prod_{n=0}^{\infty} a_{n+2} = \prod_{n=0}^{\infty} \left[1 - \frac{1}{N_n}\left(1 - \frac{1}{2a_{n+1}}\right)\right]. \tag{6}$$

By the standard theory of infinite products,

$$P_\infty > 0 \quad \text{if and only if} \quad \sum_{n=0}^{\infty} \frac{1}{N_n}\left(1 - \frac{1}{2a_{n+1}}\right) < \infty.$$

We now claim that

$$\sum_{n=0}^{\infty} \frac{1}{N_n}\left(1 - \frac{1}{2a_{n+1}}\right) < \infty \quad \text{if and only if} \quad \sum_{n=0}^{\infty} \frac{1}{N_n} < \infty.$$

Denote the two series by S_1 and S_2 respectively. Since $1 - \frac{1}{2a_{n+1}} \leq \frac{1}{2}$ for all n, convergence of the series S_2 clearly implies that of S_1. Towards the other part, we have already noted that, convergence of the series S_1 implies $\prod_{n=0}^{\infty} a_{n+2} > 0$; in particular, we will have $a_n \to 1$ as $n \to \infty$. As a consequence, for all sufficiently large n, $a_n > 3/4$ and hence $1 - \frac{1}{2a_n} > 1/3$. The proof can now be completed easily. Thus, we have

Theorem 2.3: *Suppose that we have a finite bisexual diploid population evolving through random mating. Let N_n denote the effective population size and P_n the panmictic index for the nth generation. Then, $\lim_{n\to\infty} P_n$ always exists. Further,*

$$\lim_{n\to\infty} P_n > 0 \quad \textit{if and only if} \quad \sum_{n=0}^{\infty} \frac{1}{N_n} < \infty.$$

The moral of the story is that if the population size does not grow indefinitely large or if it grows indefinitely large but not at a sufficiently fast rate, then the entire population will eventually become autozygous. Clearly, the constant population size discussed in Section 2.5.1 is an extreme example of this. Other examples of asymptotic autozygosity would be (i) when the population size grows at a linear rate, that is, $N_n \sim an + b$, (ii) when $N_n \sim n \log n$. On the other hand, if the population explodes sufficiently fast, for instance when $N_n \sim n^{1+\epsilon}$ for some $\epsilon > 0$, the theorem says that, with positive probability, the eventual population will not be autozygous. The reader should contrast this with the Hardy-Weinberg law (Theorem 2.1), where the population is theoretically infinite. In the next chapter, we shall treat a more general situation when the population sizes are allowed to be random.

2.6 Exercises

1. A woman has a rare abnormality of the eyelids called *Ptosis* which makes it impossible for her to open her eyes completely. This condition has been found to depend on a single dominant gene P. The woman's father has ptosis but her mother has normal eyelids. Her father's mother had normal eyelids.

   (a) What are the probable genotypes of the woman, her father and her mother ?

   (b) What proportion of her children are expected to have ptosis if she marries a man with normal eyelids ?

2. In humans a type of *myopia* (an eye abnormality) is dependent on a dominant gene M. Summarize the expected results from a cross between a woman with myopia but heterozygous and a normal man.

3. In humans an abnormality of the intestines called *polyposis* is dependent on a dominant gene A and a nervous disorder called *Huttington's Chorea* is determined by a dominant gene H. Assume that these two genes have loci on different chromosomes. A man with genotype $Aahh$ married a woman with genotype $aaHh$. Indicate the proportion of their children that might be expected to have (i) each abnormality, (ii) neither, (iii) both ?

4. Dark hair (M) in humans is dominant over blonde hair (m). Freckles (F) are dominant over no freckles (f). If a blonde freckled man whose mother had no freckles marries a dark haired non-freckled woman whose mother was blonde, what proportion of their children will be dark haired and freckled? blonde and non-freckled?
   If the woman in this problem were homozygous recessive for both genes, how would this change the answer?
   If the parents were both dyhybrids $MmFf$, how would this change the answer? If the parents were also heterozygous for *brachydactly* (short fingers) caused by a dominant gene B with recessive allele b (on a different chromosome), what proportions of their children will be (i) blonde, freckled and short fingered, (ii) blonde, non-freckled with normal fingers?

5. A man known to be a victim of haemophilia — a blood disease caused by an X-linked recessive gene h — marries a normal woman whose father was known to be a haemophilic. What proportion of their sons may be expected to be haemophilic? What proportion of their children are expected to be haemophilic?
   If the man in the above problem were normal, how would this affect the answers to these two questions?

6. In humans two abnormal conditions — cataract in the eyes and excessive fragility in the bones — seem to depend on separate dominant genes

located on different autosomes. A man with cataract and normal bones whose father had normal eyes maries a woman free from cataract but with fragile bones. Her father had normal bones. What is the probability that their first child (i) will be free from both abnormalities? (ii) will have cataract but not fragile bones? (iii) will have fragile bones but no cataracts? (iv) will have both the abnormalities?

7. Suppose a man is colourblind — determined by an X-linked recessive gene c and has blonde hair (see Exercise 4). His wife has normal vision and dark hair but her father is colourblind and has blonde hair. What proportion of their children will have the same phenotype as the man? What proportion will have the genotype $MmCc$?

8. In short-horn cattles, the gene R for red coat colour is co-dominant with its allele r for white coat colour. The heterozygous are roans. A breeder has white, red and roan cows and bulls. What phenotypes might be expected from the following matings and in what proportions: Red $\otimes$ Red, Red $\otimes$ White, Red $\otimes$ Roan, Roan $\otimes$ Roan, Roan $\otimes$ White, White $\otimes$ White?

9. The coloured grain, purple or red, in maize is due to the presence of *aleurone* and it is suggested that it is controlled by four independent loci. The dominant genes at each locus are defined as follows.
C is the gene for colour and must be present for any colour to develop;
R is the gene for red aleurone and presence of CR exhibits red color;
P is the gene for purple and is effective only in the presence of CR;
(CRP exhibits purple , CRp exhibits red, others give white grain)
I is the gene which inhibits colour development.
(only $CRPi$ gives purple, $CRpi$ gives red and others give white)
Show that the offsprings of $CcRrPpIi \otimes CcRrPpIi$ have the phenotypic ratio 27 purple : 9 red : 220 white. Find the phenotypic ratios in the offsprings of (i) $CcRRPpii \otimes CcRRPpii$, (ii) $CcRRPpIi \otimes CcRRPpIi$ and (iii) $CCRrPpIi \otimes CCRrPpIi$.

10. Suppose that a certain trait is controlled by one gene with two alleles A, a with A dominant and a recessive. Suppose that a is lethal recessive and that the survival ratio of the genotypes are $AA : Aa : aa = 1 : \alpha : 0$ where $0 < \alpha < 1$. Suppose that initially $AA : Aa := h_1 : h_2$ where $h_1 + h_2 = 1$. Assume random mating.

    (a) Find the frequencies in the first generation .

    (b) What is the probability of occurrence of aa zygote in the first generation?

    (c) Suppose that the surviving individuals of the first generation are again subjected to random mating. Find the distribution of genotypes in the seond generation.

11. Two parents are selected from the population $uAA + 2vAa + waa$ at random. From their progeny two parents are selected and this process is repeated (sibmating). Find the probability that both parents of the k-th filial generation are of genotypes AA, for $k = 1, 2, 3$.

12. Let a be a recessive X-linked gene and suppose that a selection process makes mating of a males impossible. If the genotypic composition of females is $AA : Aa : aa = u : 2v : w$, show that for female descendents of the next generation the genotypic composition is $AA : Aa : aa = (u+v) : (v + w) : 0$. Conclude that the frequency of a genes among females is reduced by a factor of $\frac{1}{2}$.

13. Suppose that $AA : Aa : aa = u : 2v : w$ and by the time they become mature and become parents a fraction λ , $0 < \lambda < 1$ of aa is eliminated by a selection process. Show that for parents of the next generation, the proportions of A and a gametes are given respectively by

$$p = \frac{u + v}{1 - \lambda w}, \qquad q = \frac{v + (1 - \lambda)w}{1 - \lambda w}.$$

If p_n and q_n denote the proportions of A and a gametes in the parents of the n-th generation, then show that

$$p_{n+1} = \frac{p_n}{1 - \lambda q_n^2}, \qquad q_{n+1} = \frac{(1 - \lambda q_n)q_n}{1 - \lambda q_n^2}.$$

In problems 14–21, *random mating is assumed and in both males and females the genotypic ratios are* $AA : Aa : aa = p^2 : 2pq : q^2$.

14. Given that a man is of genotype Aa, show that the probability that his brother is of the same genotype is $\frac{1}{2}(1 + pq)$.

15. Number the genotypes AA, Aa, aa as $1, 2, 3$. Let p_{ik} be the conditional probability that an offspring is of genotype k, given that the father is of genotype i. Here the mother is assumed to be 1 or 2 or 3 with probabilities p^2, $2pq$, q^2 respectively. Calculate this matrix denoted by P.

16. Show that the conditional probability that the father is of genotype k given that the first offspring is of genotype i, is also p_{ik}.

17. Show that the conditional probability of a grandson (resp. grandfather) to be of genotype k, if it is known that the grandfather (resp. grandson) is of genotype i, is the (i, k)-th entry of the matrix P^2, denoted by $p_{ik}^{(2)}$.

18. Show that $p_{ik}^{(2)}$ is also the conditional probability that a man is of genotype k if it is known that a specified half-brother is of genotype i.

19. Let $p_{ik}^{(n)}$ be the probability that a descendant of the n-th generation is of genotype k, if a specified ancestor is of genotype i. Show by induction on n, that this matrix is given by

$$\begin{pmatrix} p^2+\frac{pq}{2^{n-1}} & 2pq+\frac{q(q-p)}{2^{n-1}} & q^2-\frac{q^2}{2^{n-1}} \\ p^2+\frac{p(q-p)}{2^n} & 2pq+\frac{1-4pq}{2^n} & q^2+\frac{q(p-q)}{2^n} \\ p^2-\frac{p^2}{2^{n-1}} & 2pq+\frac{p(p-q)}{2^{n-1}} & q^2+\frac{pq}{2^{n-1}} \end{pmatrix}.$$

20. Consider Exercise 18 for a full brother instead of a half-brother. Show that the corresponding matrix is

$$\begin{pmatrix} \frac{1+p^2}{4} & \frac{q(1+p)}{2} & \frac{q^2}{4} \\ \frac{p(1+p)}{4} & \frac{1+pq}{2} & \frac{q(1+q)}{4} \\ \frac{p^2}{4} & \frac{p(1+q)}{2} & \frac{(1+q)^2}{4} \end{pmatrix}.$$

21. Show that the degree of relationship between uncle and nephew is the same as that between grandfather and grandson, that is, the corresponding transition matrices are same.

22. Derive the following expressions for the coefficient of parentage.
Father $\otimes$ Daughter = Brother $\otimes$ Sister = $\frac{1}{4}$.
Uncle $\otimes$ Niece = Double first-cousins = $\frac{1}{8}$.
First-cousins = $\frac{1}{16}$. First-cousins once-removed = $\frac{1}{32}$.
Second-cousins = $\frac{1}{64}$. Second-cousins once-removed = $\frac{1}{128}$.
Third-cousins = $\frac{1}{256}$.
What is the formula for mth cousins n-removed ?
Recall that A and B are sibs, to be called 0th order cousins, if they have the same parents. A and B are mth order cousins if one parent of A and one parent of B are $(m-1)$th order cousins. Sometimes mth order cousins are also called mth order cousins 0-removed. A and B are called mth order cousins 1-removed if EITHER B and a parent of A are mth order cousins OR A and a parent of B are mth order cousins. In general, A and B are mth order cousins n-removed in case, EITHER B and an nth ancestor of A are mth order cousins OR A and an nth ancestor of B are mth order cousins.

23. *(Parent-Offspring Mating)* Consider the system where to get the n-th generation an individual of the $(n-1)$-th generation is mated with its parent in the $(n-2)$-th generation. Show that the Panmictic index P_n satisfies $P_{n+2} - \frac{1}{2}P_{n+1} - \frac{1}{4}P_n = 0$, just as in the sibmating.

24. *(Positive Assortative Mating)* Consider a population $d_0AA+h_0Aa+r_0aa$. Assume that A is dominant over a so that there are only two phenotypes.

Consider the system where the same phenotypes mate. Show that the first generation is $d_1 AA + h_1 Aa + r_1 aa$ where

$$d_1 = \frac{(d_0 + \frac{1}{2}h_0)^2}{d_0 + h_0}, \qquad h_1 = \frac{h_0(d_0 + \frac{1}{2}h_0)}{d_0 + h_0},$$

$$r_1 = \frac{h_0^2 + 4r_0(d_0 + h_0)}{4(d_0 + h_0)}.$$

If p_n and q_n are the frequencies of A and a genes in the n-th generation so that $p_0 = d_0 + \frac{1}{2}h_0$ and $q_0 = \frac{1}{2}h_0 + r_0$, show that $p_n = p_0$ for all n. Denote this by p . If h_n is the proportion of heterozygotes in the n-th generation, then show that

$$h_1 = \frac{2ph_0}{2p + h_0}, \quad h_n = \frac{2ph_0}{2p + nh_o}.$$

Assuming that the initial population is $p^2 AA + 2pqAa + q^2 aa$, show that the limiting population is $pAA + qaa$.

25. Consider an autosomal gene with two alleles A and a. Assume zygotic selection to operate with fitness coefficients σ, γ and η for AA, Aa and aa respectively. This means a proportion σ of AA zygotes survive to maturity and participate in producing offsprings of the next generation and similarly for other coefficients. Show that you can always take $\gamma = 1$, allowing the possibility of one or both of the other coefficients to be larger than one. This means the following. Consider the system with the above coefficients and the system with coefficients σ/γ, 1, and η/γ. Of course, random mating of the matured population is assumed for obtaining the next generation. Show that these two systems are equivalent for our purposes. Incidentally, you can start with initial (unmatured) population to be $p_0^2 AA + 2p_0 q_0 Aa + q_0^2 aa$.

26. In Section 2.4, we considered the above problem when both σ and η are smaller than one (heterozygotic advantage). As stated there, anything is theoretically possible. Some of our calculations do not really depend on the assumption of the two fitness coefficients being smaller than one. Do the analysis. First find out the equilibrium states. See if there are stable equilibria and if so, how many and which initial positions lead where.

27. An interesting situation arises when $\sigma \cdot \eta = 1$. (This means, in the unnormalized notation, $\gamma^2 = \sigma \cdot \eta$ or, in other words, γ is the geometric mean of the other two.) Of course, if both σ and η are one, then fitness is same for all and the Hardy-Wienberg equilibrium persists. Assume therefore, that one of them is smaller than 1 and naturally, the other is larger than 1. In this case, show that zygotic selection acts just like the gametic selection with fitness coefficients $\sqrt{\sigma}$ and $\sqrt{\eta}$ for the gametes A and a respectively. This means the following. Consider the present gene pool,

take fitness into account and look at the normalized matured genepool and use random union of gametes to arrive at the initial genotypic frequencies of the next generation. This will be the same as what you arrive at, if instead, you normalized the matured zygotic pool, as we did and proceed. Discuss this situation.

28. Now, do not normalize but keep the fitness coefficients as σ, γ and η as in problem 27. And initial population as $p_0^2 AA + 2p_0q_0Aa + q_0^2 aa$. The mean viability for the population is defined to be $\sigma p_0^2 + 2\gamma p_0 q_0 + \eta q_0^2$. If you selected one person at random and asked about his fitness then this is the answer you get (convince yourself). There is a theorem of P.A.G.Scheur and S.P.H.Mandel which says that the mean viability can not decrease from one generation to the next. Show this.

29. There is a general theorem due to L.E.Baum and J.A.Eagon. Let W be a symmetric $k \times k$ matrix with non-negative entries (like fitness matrix). Consider the quadratic form $Q(x) = x'Wx$, a homogeneous polynomial of degree 2 in the variables $x_1, x_2, \ldots, x_k$. Consider any point p in the gene-frequency space (corresponding to k alleles). Let p^* denote the point whose i-th coordinate is

$$p_i^* = p_i \frac{\partial Q}{\partial x_i}(p) \Big/ \sum_i p_i \frac{\partial Q}{\partial x_i}(p) .$$

Then show that $Q(p^*) > Q(p)$ unless $p^* = p$. Assuming this, deduce the result of the previous problem. (These and many other interesting results are in the book of A. W. F. Edwards.)

30. Describe Hardy-Wienberg equilibria for an autosomal gene with k alleles. Describe Hardy-Wienberg equilibria for an X-linked gene with k alleles.

31. There seems to be more symmetry in the genetic code than what meets the eye. A. J. Koch and J. Lehmann, based on suggestions of previous researchers, analyzed a particular DNA data sequence. Recall that there are four nucleotides A,T,C,G and each trinucleotide (triplet of nucleotides) codes an aminoacid or * . For each nucleotide, the data showed the following probabilities of its occurrence in the first or second or third place in the trinucleotide

base α	$p_1(\alpha)$	$p_2(\alpha)$	$p_3(\alpha)$
A	0.276	0.315	0.222
T	0.166	0.285	0.268
C	0.204	0.228	0.268
G	0.354	0.172	0.242

For a trinucleotide $\alpha\beta\gamma$ define $P(\alpha\beta\gamma) = p_1(\alpha) \cdot p_2(\beta) \cdot p_3(\gamma)$.
All the sixty trinucleotides — AAA, TTT, CCC, GGG are excluded — are decomposed into three classes $\mathcal{C}_0$, $\mathcal{C}_1$ and $\mathcal{C}_2$ as follows. Put $\alpha\beta\gamma$ in

$\mathcal{C}_0$ iff the chances of its occurrence are larger compared to its other two circular permutations, namely $\beta\gamma\alpha$ and $\gamma\alpha\beta$. That is, $\alpha\beta\gamma \in \mathcal{C}_0$ iff

$$P(\alpha\beta\gamma) > P(\beta\gamma\alpha) \text{ as well as } P(\alpha\beta\gamma) > P(\gamma\alpha\beta)$$

If $\alpha\beta\gamma \in \mathcal{C}_0$ then put $\beta\gamma\alpha \in \mathcal{C}_1$ and $\gamma\alpha\beta \in \mathcal{C}_2$.

Show that $\mathcal{C}_0$ consists of

A A T , A A C , A T T , A T C , A C T , C A C , C T T ,
C T C , G A A , G A T , G A C , G A G , G T A , G T T ,
G T C , G T G , G C A , G C T , G C C , G C G .

Show that these classes $\mathcal{C}_0$, $\mathcal{C}_1$ and $\mathcal{C}_2$ are disjoint.

Show that $\mathcal{C}_0$ is self-complementary whereas $\mathcal{C}_1$ and $\mathcal{C}_2$ are complementary to each other. Here complement means the following. For nucleotides, A and T are complementary, whereas C and G are complementary. For trinucleotides, RST is complementary to XYZ iff R and Z are complementary, S and Y are complementary and T and X are complementary. Note carefully the order used here in the definition.

2.7 References/Supplementary Readings

For more on the material discussed in this chapter the reader may consult the following books.

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Chapter 3

MARKOV MODELS IN GENETICS

3.1 Introduction

Markov chain models have been the most widely used ones in the study of random fluctuations in the genetic compositions of populations over generations. Besides being a convenient theoretical tool, Markov chains have provided rather satisfactory theoretical explanations to some observed long-run phenomena related to the genetic structure of populations.

In the previous chapter, we had already set the stage for discussing the fluctuations of genotypic frequencies over generations under various mating systems. For instance, under selfing and sibmating, we concluded that individuals become eventually homozygous (in fact, autozygous, even though that feature is not going to be highlighted in the sequel). However, the methods used there were not powerful enough and leave many questions unanswered. For instance, what will be the average number of generations needed to make the population homozygous? Also, if the alleles under consideration are A and a, say, then what are the chances of the population being eventually stabilized or *fixed* at the particular homozygous state, say, AA? It is precisely in this context, that Markov chain methods will be helpful. We shall be discussing an autosomal gene with two alleles A, a throughout. As was already evident, once the mating system is fixed, the structure of a generation depends entirely on the previous generation. In other words, the future evolution of the population depends only on the composition of the present generation and the past history is irrelevant. Mathematically speaking, this precisely is the same as saying that the evolution is markovian.

In this chapter, we discuss, in full detail, some of the fundamental and classical research done in this area. The most pioneering is, of course, the classical work of S. Wright and R. A. Fisher, now known as the *Wright-Fisher*

model. This is taken up in Section 3.4. This is preceeded by a discussion of two relatively simpler models, namely, selfing and sibmating, in Sections 3.2 and 3.3. Section 3.5 is entirely devoted to three models proposed by Moran — one for the haploids and two for diploids. This is followed, in Section 3.6, by a discussion of an interesting classical model due to Kimura for ageing. In Section 3.7, we return to the Wright-Fisher model and present some more recent ramifications of the model, allowing the population size to change from generation to generation. The final section, Section 3.8, contains an outline of the diffusion approximations for the Wright-Fisher and Moran models.

3.2 Selfing

For an autosomal gene with two alleles A and a, we have three genotypes AA, Aa and aa. Let us name them as the three states 1, 2 and 3. Let us consider selfing and follow a line of descent. Thus, if an individual is in state 1 or 3, then all its descendents will be in the same state. If, on the other hand, an individual is in state 2, then its descendent in the next generation will be in the states 1, 2 or 3 with probabilities 1/4, 1/2 and 1/4 respectively. In other words, we have a Markov chain $(X_n)_{n\geq 0}$ with states 1, 2, 3 and transition matrix

$$P = \begin{pmatrix} 1 & 0 & 0 \\ 1/4 & 1/2 & 1/4 \\ 0 & 0 & 1 \end{pmatrix}.$$

In this entire analysis, we assume, of course, that there are no mutations or fitness constraints. The states 1 and 3 are absorbing, whereas state 2 is transient. This matrix is simple enough to allow a complete analysis as follows. The matrix has eigen values 1, 1/2, and 1. The corresponding right eigen vectors are $(1,1,1)'$, $(0,1,0)'$ and $(1,2,3)'$, while the left eigen vectors are $(3/2,0,-1/2)$, $(-1/2,1,-1/2)$ and $(-1/2,0,1/2)$. Thus, P can be diagonalised as

$$P = \begin{pmatrix} 1 & 0 & 1 \\ 1 & 1 & 2 \\ 1 & 0 & 3 \end{pmatrix} \begin{pmatrix} 1 & 0 & 0 \\ 0 & 1/2 & 0 \\ 0 & 0 & 1 \end{pmatrix} \begin{pmatrix} 3/2 & 0 & -1/2 \\ -1/2 & 1 & -1/2 \\ -1/2 & 0 & 1/2 \end{pmatrix} = \Lambda D \Lambda^{-1}.$$

This makes it possible to explicitly calculate the n-step transition matrix $P^n = \Lambda D^n \Lambda^{-1}$, from which it easily follows that

$$p_{11}^{(n)} = p_{33}^{(n)} = 1; \quad p_{21}^{(n)} = \frac{1}{2}[1 - 2^{-n}],\ p_{22}^{(n)} = 2^{-n},\ p_{23}^{(n)} = \frac{1}{2}[1 - 2^{-n}];$$

From the above one easily gets $\alpha_{21} = P(X_n \text{ is eventually } 1 | X_0 = 2) = 1/2$ and $\alpha_{23} = P(X_n \text{ is eventually } 3 | X_0 = 2) = 1/2$. One can of course get all these by direct probabilistic calculations without bringing in the matrix P or P^n — the reader should do this as an exercise.

Thus starting from state 2, the absorption probabilities to the two states 1 and 3 are 1/2 each, as is expected from symmetry. Let T be the time till

absorption, that is, $T = n$ iff $X_n = 1$ or 3 but $X_{n-1} = 2$. Then we have $P(T = n \mid X_0 = 2) = (1/2)^{n-1} - (1/2)^n = (1/2)^n$, so that $E(T \mid X_0 = 2) = 2$. This says that, starting from state 2, the system takes two generations on an average to get absorbed in one of the two states 1 or 3. For the sake of completeness, we advise the reader to calculate the variance of T. The whole situation here is unusually simple and we actually have the exact distribution of T.

In the above, the Markov chain (X_n) models the genotype sequence of a line of descent under selfing. One can also get the structure of the population in the nth generation as follows. Suppose initially we have $\underline{p} = (p_1, p_2, p_3)$ as the proportions of various genotypes in the population. Clearly, the proportions in the nth generation are given by $\underline{p}P^n$ which is simple to explicitly evaluate in this case. Indeed, since

$$P^n = \begin{pmatrix} 1 & 0 & 0 \\ (1-2^{-n})/2 & 2^{-n} & (1-2^{-n})/2 \\ 0 & 0 & 1 \end{pmatrix},$$

one gets $\underline{p}P^n = (p_1 + (1-2^{-n})p_2/2,\ 2^{-n}p_2,\ p_3 + (1-2^{-n})p_2/2)$.

3.3 Sibmating

In case of selfing, an individual has only one parent and the transition from the genotype of the father to the genotype of the offspring is modelled by a Markov chain. But in sibmating, each offspring has two parents and hence the genotype of an individual depends on those of both its parents. It is therefore evident that simply the individual genotypic changes from generation to generation cannot form a Markov chain. To build a markovian model, we consider the evolution of genotypic pairs of sibs. In other words, we look at a line of descent of sibs as follows. Consider two sibs of a generation; from their offsprings, select two sibs at random; from their offsprings again select two at random and so on. For instance, if the present sibs are (Aa, Aa), then their offsprings consist of $\frac{1}{4}AA + \frac{1}{2}Aa + \frac{1}{4}aa$ in both males and females, so that if two independent choices are made — one from males and one from females — then the sibs so formed will be of type (AA, AA) with chance $1/16$, of type (AA, Aa) with chance $1/4$, etc. While considering genotypic pairs of sibs, we do not attach any importance to which one of the pairs is a male member and which is female. In other words, genotypic pair (AA, Aa) means that one of the sibs is AA and the other is Aa and do not ask which is which. Thus the state space of our Markov chain (X_n) is (AA, AA), (aa, aa), (AA, Aa), (aa, Aa), (AA, aa) and (Aa, Aa)

— numbered as 1,2,3,4,5 and 6 respectively. The transition matrix is

$$P = \begin{pmatrix} 1 & 0 & 0 & 0 & 0 & 0 \\ 0 & 1 & 0 & 0 & 0 & 0 \\ 1/4 & 0 & 1/2 & 0 & 0 & 1/4 \\ 0 & 1/4 & 0 & 1/2 & 0 & 1/4 \\ 0 & 0 & 0 & 0 & 0 & 1 \\ 1/16 & 1/16 & 1/4 & 1/4 & 1/8 & 1/4 \end{pmatrix} = \begin{pmatrix} I_{2\times 2} & O_{2\times 4} \\ R_{4\times 2} & Q_{4\times 4} \end{pmatrix}, \text{ say.}$$

States 1 and 2 are absorbing and the others are transient. We shall apply the fundamental matrix method to analyze this Markov chain. One can verify that the fundamental matrix, in the terminology of Section 0.8.3, is

$$N = (I - Q)^{-1} = \begin{pmatrix} 8/3 & 2/3 & 1/6 & 4/3 \\ 2/3 & 8/3 & 1/6 & 4/3 \\ 4/3 & 4/3 & 4/3 & 8/3 \\ 4/3 & 4/3 & 1/3 & 8/3 \end{pmatrix},$$

so that the vector of mean times till absorption is

$$\widetilde{m} = Ne = \begin{pmatrix} 29/6 \\ 29/6 \\ 20/3 \\ 17/3 \end{pmatrix},$$

and the absorption probabilities, in the terminology of Section 0.8.5, are

$$A = NR = \begin{pmatrix} 3/4 & 1/4 \\ 1/4 & 3/4 \\ 1/2 & 1/2 \\ 1/2 & 1/2 \end{pmatrix}.$$

Thus, if we start with sibs (AA, Aa), that is in state 3, then in their progeny, sibs will eventually be of types (AA, AA) or (aa, aa) with chances 3/4 and 1/4 respectively. Moreover it takes 29/6 (approximately 5) generations on an average, for this eventual fixation. Using Exercise 6, Section 0.8.3, the vector of variances of the absorption time in this case turns out to be

$$\widetilde{V} = \begin{pmatrix} 769/36 \\ 769/36 \\ 816/36 \\ 816/36 \end{pmatrix}.$$

A direct calculation shows that P has eigenvalues $\lambda_1 = \lambda_2 = 1$, $\lambda_3 = \frac{1+\sqrt{5}}{4}$, $\lambda_4 = \frac{1}{2}$ $\lambda_5 = 1/4$ and $\lambda_6 = \frac{1-\sqrt{5}}{4}$. Thus, following the general theory as discussed in Section 0.8.5, we conclude that the rate of absorption is given by $\frac{1+\sqrt{5}}{4}$. Of course this was already observed in Section 2.4.4 of the previous chapter.

We leave it to the reader to make a similar analysis with X-linked gene with two alleles A and a. The state space now consists of (AA, AY), (aa, aY), (AA, aY), (aa, AY), (Aa, AY) and (Aa, aY).

3.4 The Wright-Fisher Model

The Hardy-Wienberg law and its ramifications as discussed in the previous chapter have some serious limitations, namely, that the stochastic content in them is limited only to the randomness in the matings in a given generation. This stems from the tacit assumption of the population being potentially infinite. Such analysis, therefore, fails to capture the phenomenon of genetic evolution in finite populations, where the sampling fluctuations play a central role. This suggests adopting models that capture this component of randomness in evolution. This was already realized by Pearson (1904), Yule (1906) and Fisher (1911). The first such model was proposed by Fisher (1930) and Wright (1931). Since then this model, known as the Wright-Fisher model, has occupied the centre stage in mathematical models of genetics. We proceed to describe this model.

Let us consider, as usual, an autosomal gene with two alleles A and a, so that, there are three genotypes AA, Aa and aa. We wish to study the evolution of genotypic frequencies in a given population. Ideally what we wish to do is the following. Suppose that initially there are N_1 males with composition $N_{11}AA + N_{12}Aa + N_{13}aa$ and N_2 females with composition $N_{21}AA + N_{22}Aa + N_{23}aa$. Let us assume random mating. For the kth generation we want to know the data $N_1^k = N_{11}^k AA + N_{12}^k Aa + N_{13}^k aa$ and $N_2^k = N_{21}^k AA + N_{22}^k Aa + N_{23}^k aa$. The problem in this generality is complicated and first we affect some simplifications.

Let us assume that for all k, $N_{1i}^k = N_{2i}^k$ for $i = 1, 2, 3$, that is, in all generations the genotypic frequencies are the same for both the sexes. Of course, this will imply that $N_1^k = N_2^k = N^k$, say. To put it differently, we consider unisex population, as for example, plants. Then the problem simplifies to describing the 3-tuple (N_1^k, N_2^k, N_3^k). As a further simplification, we assume that $N^k = N$ for all k, that is, variation in the total population size is also ruled out. This may look like a gross over-simplification, far removed from reality. However, it can be given the following interpretation. Imagine a 'real' population evolving in time with possibly changing size. But to facilitate calculations, we concentrate on N individuals randomly chosen from each generation. The cautious reader would of course realize that this is not a completely truthful interpretation. Truly speaking, the population is constrained to have N individuals — neither more nor less — in each generation, where N is fixed in advance. Under this simplification, it suffices to know how many AA and how many aa are there. Thus, the problem is reduced to describing the evolution of the pair (N_1^k, N_3^k) only.

Even after all these simplifications, the problem still remains quite intractable. Therefore, we are going to simplify it further. However, in the subsequent sections, we shall return to the problems described above. For the time being we decide to concentrate only on the variations in the gene frequencies rather than the genotypic frequencies. In any generation, the N individuals carry a total of $2N$ genes, some of which are A and the rest are a. Let X_k be

the number of A genes in the kth generation so that $2N - X_k$ is the number of a genes. We are going to study the evolution of X_k. Of course, this would have been perfectly alright if, to start with, we had a haploid population of $2N$ individuals, in which case there are only two genotypes A and a.

We now come to the specific hypothesis concerning how a generation gives rise to the next generation. We assume that the $2N$ genes of a generation are obtained by simply taking a random sample of size $2N$ with replacement from the $2N$ genes of the parent generation. This is the classical Wright-Fisher model. It is clear that for each n, X_n, the number of A genes in the nth generation, is a random variable taking values $0, 1, \ldots, 2N$. The above assumption really means firstly, that the conditional distribution of X_{n+1} given $X_0, X_1, \ldots, X_n$ depends only on X_n and secondly, that given $X_n = j$, X_{n+1} is distributed as the binomial variable $B(2N, \frac{j}{2N})$. In other words, the process $(X_n)_{n\geq 0}$ forms a Markov chain with state space $\{0, 1, \ldots, 2N\}$ and transition probabilities

$$p_{jk} = \binom{2N}{k} \theta_j^k (1-\theta_j)^{2N-k} \quad \text{for} \quad 0 \leq j,\ k \leq 2N,$$

$$\text{where} \quad \theta_j = \frac{j}{2N}.$$

For this chain, it is clear that the states 0 and $2N$ are absorbing while others are transient. Thus, no matter where we start, the chain eventually gets absorbed in one of the two absorbing states. Thus $X_\infty = \lim_{n\to\infty} X_n$ exists and takes the two values 0 and $2N$ with probability one.

The first important question that we would like to address is the following. Given that the number of A genes is i to start with (that is $X_0 = i$), what are the probabilities $b_0(i)$ and $b_{2N}(i)$ of the chain to be absorbed in the states 0 and $2N$ respectively. Actually, $b_0(i)$ and $b_{2N}(i)$ are nothing but $\alpha_{i,0}$ and $\alpha_{i,2N}$ in the notation of Section 0.8.3. Note that $b_0(i) = P(X_\infty = 0 \,|\, X_0 = i)$ and $b_{2N}(i) = P(X_\infty = 2N \,|\, X_0 = i)$. The usual fundamental matrix method is not of much help here (try and get convinced). Here is a beautiful alternative due to Feller.

Observe that the process $(X_n)_{n\geq 0}$ has the property that

$$E(X_{n+1} \,|\, X_n) = X_n \quad \text{for every } n.$$

Indeed, since the conditional distribution of X_{n+1}, given $X_n = j$, is $B(2N, \frac{j}{2N})$ we have $E(X_{n+1} \,|\, X_n = j) = 2N \cdot \frac{j}{2N} = j$. Because of the Markov property, the above equation is the same as

$$E(X_{n+1} \,|\, X_0, X_1, \ldots, X_n) = X_n \quad \text{for every } n.$$

Thus $(X_n)_{n\geq 0}$ is a martingale in the sense discussed in Section 0.7. In particular, for all n, $E(X_n \,|\, X_0 = i) = i$. Since the martingale is uniformly

bounded, it follows that $E(X_\infty | X_0 = i) = i$. But of course, $E(X_\infty | X_0 = i) = 0 \cdot b_0(i) + 2N \cdot b_{2N}(i)$. This yields

$$b_{2N}(i) = \frac{i}{2N} \quad \text{and} \quad b_0(i) = 1 - \frac{i}{2N}.$$

Thus, if initially there are i many A genes, then eventually the number of A genes will be 0 or $2N$ with probabilities $1 - \frac{i}{2N}$ and $\frac{i}{2N}$ respectively.

Having thus obtained the absorption probabilities, we now turn to the rate at which absorption takes place. One simple-minded way to get this would be to evaluate $p_{i,0}^{(n)}$ and $p_{i,2N}^{(n)}$ explicitly. Khazanie and McKean have done this, but unfortunately their analysis is complicated. Instead, we recall that it suffices to know the largest (in modulus) eigenvalue of the transition matrix, which is smaller than one in modulus. S. Wright and G. Malecot obtained approximations for this eigenvalue. But later, Feller gave an elegant method to get the exact expression for all the eigenvalues, which is what we discuss now. Define

$$\lambda_0 = 1 \quad \text{and,} \quad \text{for } 1 \leq r \leq 2N, \quad \lambda_r = 1\left(1 - \frac{1}{2N}\right)\cdots\left(1 - \frac{r-1}{2N}\right),$$

or equivalently

$$\lambda_r = \binom{2N}{r}\frac{r!}{(2n)^r} \quad \text{for} \quad 0 \leq r \leq 2N. \tag{1}$$

Note that, $\lambda_0 = \lambda_1 = 1 > \lambda_2 > \lambda_3 > \cdots > \lambda_{2N}$. We shall now show that these are precisely the eigenvalues of P from which it would follow that convergence takes place geometrically at the rate $\lambda_2 = (1 - \frac{1}{2N})$.

Consider the two linearly independent vectors $v_0 = (1, 1, \ldots, 1)'$ and $v_1 = (0, 1, \ldots, 2N)'$, each of order $2N + 1$. Since P is stochastic matrix, $Pv_0 = v_0$. Also, since for each j, the j-th row of P is the probability mass function of $B(2N, \frac{j}{2N})$ distribution, one sees that $Pv_1 = v_1$. Since there are two absorbing states, by the general theory of Section 0.8.5, P has 1 as an eigenvalue and the dimension of the eigenspace associated to this eigenvalue is 2. This takes care of λ_0 and λ_1 of Equation (1). To complete the proof we shall now exhibit, for each $r = 2, \ldots, 2N$, a non-null vector v_r such that $Pv_r = \lambda_r v_r$. To this end, we fix some notation. For any real number x and any integer $i \geq 1$, let $(x)_i$ denote the i-th factorial power of x, that is, $x(x-1)\cdot\cdots\cdot(x-i+1)$. We define $(x)_0 \equiv 1$. We first make the following observation.

Lemma 3.1: *Let $2 \leq r \leq 2N$ and $a_0, a_1, \ldots, a_r$ be numbers with $a_r \neq 0$. Then the vector $v = (x_0, x_1, \ldots, x_{2N})$ with $x_k = \sum_{i=0}^{r} a_i(k)_i$ is non-null.*

Proof: $P(x) = \sum_{i=0}^{r} a_i(x)_i$ is a polynomial in x of degree exactly $r \leq 2N$, and has at most r real zeros. Noting that $x_k = P(k)$, for $k = 0, 1, \ldots, 2N$, it follows that not all the x_k can be zero. ∎

Our proposed v_r is going to be of the same form as v of the lemma. Note that the vector of the lemma is completely determined by the numbers $a_0, \ldots, a_r$.

For a vector v of this type to satisfy $Pv = \lambda_r v$ it is necessary and sufficient to have, for $j = 0, 1, \ldots, 2N$,

$$\begin{aligned}
\lambda_r x_j &= \sum_{k=0}^{2N} p_{jk} x_k \\
&= \sum_{k=0}^{2N} p_{jk} \sum_{l=0}^{r} a_l (k)_l \\
&= \sum_{l=0}^{r} \sum_{k=l}^{2N} a_l (k)_l \binom{2N}{k} \theta_j^k (1-\theta_j)^{2N-k} \\
&= \sum_{l=0}^{r} a_l (2N)_l \theta_j^l ,
\end{aligned}$$

the last two equalities being consequences of the facts that $(k)_l = 0$ for $k < l$ and

$$\sum_{k=l}^{2N} \binom{2N-l}{k-l} \theta_j^{k-l} (1-\theta_j)^{2N-k} = 1.$$

Thus $Pv = \lambda_r v$ becomes equivalent to having

$$\sum_{l=0}^{r} \lambda_r \, a_l \, (j)_l = \sum_{l=0}^{r} a_l \, (2N)_l \, \theta_j^l \quad \text{for} \quad j = 0, 1, \ldots, 2N . \tag{2}$$

To proceed with the choice of the numbers $a_0, \ldots, a_r$, so that the above holds, we need the following basic fact.

Lemma 3.2: *For any integer $k \geq 0$, the polynomials $\{(x)_i \, , \, 0 \leq i \leq k\}$ form a basis for the vector space of all polynomials in x of degree less than or equal to k.*

The proof of Lemma 3.2 is easy and hence omitted. From this lemma, one obtains that for each m, $0 \leq m \leq 2N$, there exist constants $C_{m,0}$, $C_{m,1}$, $\ldots$, $C_{m,m}$ so that $\dfrac{x^m}{(2N)^m} \equiv \sum_{\nu=0}^{m} C_{m,\nu}(x)_\nu$. In particular, $\theta_j^h = \sum_{\nu=0}^{h} C_{h,\nu}(j)_\nu$, for every j. Comparing the coefficients of j^h on both sides we get $C_{h,h} = \frac{1}{(2N)^h}$, so that $(2N)_h C_{h,h} = \lambda_h$. Using this, Equation (2) is equivalent to

$$\lambda_r \sum_{l=0}^{r} a_l \, (j)_l = \sum_{l=0}^{r} a_l \, (2N)_l \sum_{\nu=0}^{l} C_{l,\nu} \, (j)_\nu ,$$

or to

$$\sum_{\nu=0}^{r} \lambda_r \, a_\nu \, (j)_\nu = \sum_{\nu=0}^{r} \left[\sum_{l=\nu}^{r} a_l \, (2N)_l \, C_{l,\nu} \right] (j)_\nu .$$

For this to hold, it suffices to ensure that

$$\lambda_r \, a_\nu = \sum_{l=\nu}^{r} a_l (2N)_l \, C_{l,\nu} \quad \text{for} \quad \nu = 0, 1, \ldots, r . \tag{3}$$

For $\nu = r$, this equation is trivially satisfied by any a_r in view of the fact that $(2N)_r\, C_{r,r} = \lambda_r$. We can and do take $a_r = 1$. For $\nu < r$, (3) is equivalent to

$$(\lambda_r - \lambda_\nu)\, a_\nu = \sum_{l=\nu+1}^{r} (2N)_l\, C_{l,\nu}\,,$$

that is,

$$a_\nu = \frac{1}{\lambda_r - \lambda_\nu} \sum_{l=\nu+1}^{r} a_l\, (2N)_l\, C_{l,\nu}\,.$$

Note that $\lambda_r \neq \lambda_\nu$, since $r \geq 2$ and $\nu < r$. This last equation is an equation prescribing a_ν in terms of $a_{\nu+1}, \ldots, a_r$. Having chosen $a_r = 1$, all the a_ν for $\nu < r$ can now be automatically determined. Thus we have proved the following theorem.

Theorem 3.3: *The numbers* $\{\lambda_r : 0 \leq r \leq 2N\}$ *as given in (1) are precisely the eigenvalues of the Wright-Fisher matrix* P.

In the terminology of genetics, absorption is called *fixation* or *homozygosity* and the rates of absorption are called the *rates of fixation.* In the Wright-Fisher model, we are selecting each generation as a random sample from the previous generation, but yet ultimately one gene is being fixed. This is what is called *genetic drift* and is attributed to the finiteness of the population and the consequential sampling fluctuations. To understand this statement, suppose we have a sequence $s_1 = \langle x_1, x_2, \ldots, x_M \rangle$ of length M where each x_i is an integer from $\{1, 2, \ldots, N\}$. One may think of s_1 as a sample (with replacement) of size M from a population having N distinct elements. Treating now s_1 as a population of size M, a simple random sample of size M is drawn to obtain another sequence s_2. With s_2 as the population of size M, we again draw a simple random sample of size M to get s_3. We continue this process. It should be emphasized here that at each stage of our sampling scheme, the M population units need not be distinct, since our samples are always drawn with replacement. As a consequence, if a particular symbol, say i, occurs n_i times in s_k then the chance of i being excluded from s_{k+1} can easily be seen to be $[1 - \frac{n_i}{M}]^M$. Thus a symbol with less number of repetitions in s_k is more likely to be excluded from s_{k+1}. The reader can convince himself that even if the original sequence s_1 consisted of M distinct symbols, sampling fluctuations force repetitions to occur sooner or later. This along with the previous observation explains why in the long run the process will stabilize at one symbol being repeated M times. A more precise formulation and justification of this phenomenon can be given using Markov chain terminology. It will be instructive for the reader to try.

We have discussed the simplest case of the Wright-Fisher model. The most important thing that happens in nature all the time is mutation. A realistic model should take this into account. Suppose we assume that there is a chance α_1 of A mutating to a and a chance α_2 of a mutating to A. Any particular gene can mutate at most once in passing from one generation to the next. Suppose

that there are j many A genes and $2N-j$ many a genes now. Select one of them at random and allow for possible mutation. What is the chance that it is A ? This can happen only when A is selected and mutation did not take place or a is selected and mutation did take place. The selection probabilities should thus be modified as,

$$\theta_j = \frac{j}{2N}(1-\alpha_1) + \left(1-\frac{j}{2N}\right)\alpha_2 .$$

One can now discuss the model incorporating mutation as follows. Fix mutation probabilities $0 < \alpha_1 < 1$ and $0 < \alpha_2 < 1$. Then $(X_n)_{n\geq 0}$ is a Markov chain with the conditional distribution of X_{n+1}, given $X_n = j$, being $B(2N, \theta_j)$ where θ_j now is as above. In practice, only small values of α_1 and α_2 are relevant. Let us assume therefore that $0 < \alpha_1 + \alpha_2 < 1$. Then the chain is irreducible and hence necessarily recurrent. One would like to know the stationary distribution. Also recall that since the chain is clearly aperiodic, the distribution of X_n is eventually going to be this stationary distribution, no matter how the chain started initially. If we derive the stationary distribution, this will give us the long-run proportion of A and a genes in the population, without any need for the knowledge of the origins of the population(!).

We end this section by pointing out a limitation of the discrete model of Wright-Fisher. For example, going back to the Wright-Fisher model without mutation, one quantity of interest would be the average time needed for fixation. In other words, denoting by T the first time the chain is absorbed, T is clearly a random variable. We would like to know the distribution of T, or at least, the expected value of T. Clearly

$$\begin{aligned} P(T = n \,|\, X_0 = i) &= P(X_n \in \{0, 2N\}, X_{n-1} \notin \{0, 2N\} \,|\, X_0 = i) \\ &= (p_{i,0}^{(n)} + p_{i,2N}^{(n)}) - (p_{i,0}^{(n-1)} + p_{i,2N}^{(n-1)}) . \end{aligned}$$

The above equation gives the exact distribution of T, given $X_0 = i$, from which it is theoretically possible to calculate its expected value. However the formulae for $p_{i,j}^{(n)}$ are complicated and do not yield a tractable and easily understandable expression for $E(T \,|\, X_0 = i)$. An alternative would be to look for at least an approximate expression for $E(T|X_0 = i)$, which is simple enough to understand. Such an approximation was indeed given by Feller and subsequently studied by Kimura. We will take a brief look of these developments in Section 3.8.

3.5 Moran's Models

In this section we consider what seems to be the second most significant work on Markov chain modelling in population genetics. The following models were proposed by P. A. P. Moran in 1958.

3.5.1 Haploid Model

The set-up is the same as in the Wright-Fisher model. We consider a haploid population with two genotypes A and a. Instead of forming new generation by sampling from the previous one, this model envisages to follow the population through its birth-death events. More precisely, a new generation is formed from the old one in the following way. One individual chosen at random from the existing population gives birth to an individual (naturally of the same genotype) and, at the same time, one randomly chosen individual of the old population dies. Note that this keeps the population size fixed. We denote it by M. As before, we denote by X_n, the number of A genes in the nth generation. Here, the nth generation means the population after n birth-death events. We want to study the Markov chain $(X_n)_{n\geq 0}$, which has state space $\{0, 1, \ldots, M\}$. If the present population has composition $iA + (M-i)a$, then the individual who dies is A with probability $\frac{i}{M}$ and is a with probability $1 - \frac{i}{M}$. At the same time, a new born is of type A with probability $\frac{i}{M}$ and a with probability $1 - \frac{i}{M}$. So the transition probabilities of the chain are given by

$$p_{i,i-1} = p_{i,i+1} = \frac{i}{M}(1 - \frac{i}{M}),$$

$$p_{i,i} = (\frac{i}{M})^2 + (1 - \frac{i}{M})^2 \quad \text{for} \quad 0 < i < M.$$

Of course, $p_{0,0} = p_{M,M} = 1$. Note that $p_{i,j} = 0$ if $j \notin \{i-1, i, i+1\}$.

Here again, 0 and M are the absorbing states and all others are transient. Also, from the transition probabilities, it is clear that $E(X_{n+1} \mid X_n = i) = i$. Thus, as in the Wright-Fisher model, $(X_n)_{n\geq 0}$ is a martingale and, as earlier, given $X_0 = i$, the chain will be absorbed in 0 with probability $1 - \frac{i}{M}$ and absorbed in M with probability $\frac{i}{M}$.

In this case again, we can write down all the eigenvalues of the transition matrix P. In fact, this matrix is much simpler than the Wright-Fisher matrix. We follow the method given in Watterson (1961) and prove the following two lemmas (attributed to E. J. Hannan). In the first lemma we use the usual convention that $\binom{i}{j}$ is zero for $j > i$.

Lemma 3.4: *Let R be the matrix with entries $R_{ij} = \binom{i}{j}$ for $0 \leq i, j \leq M$. Then R^{-1} is the matrix S with entries $S_{ij} = (-1)^{i+j}\binom{i}{j}$.*

Proof: The (i,j)-th entry of RS is $\sum_k \binom{i}{k}(-1)^{k+j}\binom{k}{j}$. Note that, if $i < j$, then $\binom{i}{k}\binom{k}{j} = 0$ for all k, while if $i = j$, then $\binom{i}{k}\binom{k}{j} = 0$ for all $k \neq i$ (why?). This proves that $(RS)_{ij} = 0$ if $i < j$ and $(RS)_{ij} = 1$ if $i = j$. Finally for $i > j$, $(RS)_{ij}$ equals

$$\binom{i}{j}(-1)^{j-i} \sum_{j\leq k\leq i} \binom{i-j}{i-k}(-1)^{i-k} = 0.$$

This completes the proof. ∎

Lemma 3.5: *The matrix $R^{-1}PR$ has non-zero entries only at the diagonal and the first super diagonal. More precisely, the matrix $R^{-1}PR = A$, say, has entries $a_{i,j}$ given by*

$$a_{i,i} = 1 - \frac{i(i-1)}{M^2}, \quad a_{i,i+1} = \frac{i(M-i)}{M^2} \quad \text{and} \quad a_{i,j} = 0 \quad \text{for } j \neq i, i+1.$$

Once this lemma is proved, it follows that the eigenvalues of the matrix P, same as those of $R^{-1}PR$, are given by

$$\lambda_i = 1 - \frac{i(i-1)}{M^2} \qquad i = 0, 1, \ldots, M.$$

Note that $\lambda_0 = \lambda_1 = 1$. Thus, the second largest (in modulus) eigenvalue after 1 is given by $\lambda_2 = 1 - \frac{2}{M^2}$. This gives the rate of approach to homozygosity, or fixation rate, in the same sense as before.

Proof of lemma 3.5: Since $P_{k,l} = 0$ for $l \notin \{k-1, k, k+1\}$, it is clear that the (i,j)-th element of SPR, with S as in Lemma 3.4, is

$$\sum_{k=0}^{M} S_{i,k}(P_{k,k-1}R_{k-1,j} + P_{k,k}R_{k,j} + P_{k,k+1}R_{k+1,j}).$$

Since $S_{i,k} = 0$ for $k > i$, and $R_{l,j} = 0$ for $j > l$, the above sum actually extends from $k = j-1$ to $k = i$. In particular, when $j - 1 > i$, it is zero. Thus $a_{i,j} = 0$ for $j > i+1$. In case $j = i+1$, there is only one term in the sum. In case $j = i$, the sum consists of two terms only. These easily reduce to the stated expressions for $a_{i,j}$. The only thing to be shown now is that for $j < i$, the sum above reduces to zero. If we substitute the values and use the fact that $\sum S_{i,k}R_{k,j}$ is zero for $j < i$, then we have to show that

$$\sum_{k=j-1}^{i} (-1)^{i+k} \binom{i}{k} \frac{k(M-k)}{M^2} \left[\binom{k-1}{j} - 2\binom{k}{j} + \binom{k+1}{j}\right] = 0.$$

This easily follows from the following three equations whose verification is left to the reader.

$$\binom{k-1}{j} - 2\binom{k}{j} + \binom{k+1}{j} = \frac{(k-1)!}{j!(k-j+1)!} j(j-1),$$

$$\sum_{k=0}^{i-j+1} (-1)^k \binom{i-j+1}{k} = 0,$$

$$\sum_{k=0}^{i-j+1} (-1)^{(k-1)} \binom{i-j+1}{k} (k-1) = 0.$$

This completes the proof. ∎

Next, we wish to know the time taken for absorption. The exact distribution, though complicated, has been evaluated by Watterson. Here we present his method to find the expected time for absorption. The fundamental matrix method applies here beautifully.

From the transition matrix P, we omit the rows and columns corresponding to the two absorbing states and get the $(M-1) \times (M-1)$ matrix Q and the objective is to find the matrix $N = (I-Q)^{-1}$. It is easy to see that $I - Q = DB$, where D is the diagonal matrix with diagonal entries $\frac{k(M-k)}{M^2}$ for $1 \leq k \leq (M-1)$, and the matrix $B = ((b_{i,j}))$ is given by

$$\begin{aligned} b_{i,j} &= 2 \quad \text{if} \quad i = j, \\ &= -1 \quad \text{if} \quad i = j-1 \text{ or } j+1, \\ &= 0 \quad \text{otherwise.} \end{aligned}$$

Moreover the rows of the matrix B^{-1} are given by

$$\left(\frac{1(M-i)}{M}, \frac{2(M-i)}{M}, \ldots, \frac{i(M-i)}{M}, \frac{i(M-i-1)}{M}, \ldots, \frac{i \cdot 2}{M}, \frac{i \cdot 1}{M} \right),$$

for $1 \leq i \leq M-1$. Thus, the fundamental matrix N, which equals $B^{-1}D^{-1}$, can be explicitly computed. In particular, $m_i = E(T \mid X_0 = i)$, which is the i-th row sum of N, turns out to be

$$m_i = (M-i) \sum_{j=1}^{i} \left(1 - \frac{j}{M}\right)^{-1} + i \sum_{j=1}^{M-i-1} \left(1 - \frac{j}{M}\right)^{-1}.$$

However this expression does not admit a nice closed form. In particular, its dependence on i is not very clearly revealed from the above expression. We now proceed to obtain a neater expression which approximates m_i. Rewrite the above expression as

$$m_i = M^2 \left(1 - \frac{i}{M}\right) \sum_{j=1}^{i} \frac{1}{M} \frac{1}{(1 - \frac{j}{M})} + M^2 \frac{i}{M} \sum_{j=1}^{M-i-1} \frac{1}{M} \frac{1}{(1 - \frac{j}{M})}.$$

We observe that the two sums appearing above can be viewed as appropriate Riemann sums and it is natural to approximate them by the corresponding Riemann integrals. Denoting the initial proportion (i/M) of A genes by p we thus obtain that m_i can be approximated, at least for large M, by

$$M^2(1-p) \int_0^p \frac{1}{1-x}\, dx + M^2 p \int_0^{1-p} \frac{1}{1-x}\, dx = M^2 \log\left[p^{-p}\,(1-p)^{-(1-p)}\right].$$

We can evaluate it for special values of p and in Section 3.8, we will use this to compare this model with the Wright-Fisher model. Note that the fundamental matrix method further enables us to evaluate the variance of the absorption time.

In this model also, we can incorporate mutation. This gives an ergodic aperiodic chain, whose stationary distribution can be explicitly evaluated. Some of these aspects are discussed in the exercises.

3.5.2 Diploid Unisex Model

We shall discuss diploid unisex population. As usual, we consider a gene with two alleles A and a. We fix the population size at N. At each instant, we should know the number of various genotypes present. Of course, the population size being fixed, it suffices to know the number of any two of the genotypes. Thus, if the population has the composition $AA : Aa : aa = k : (N-k-l) : l$, it is enough to know k and l. In other words, this is essentially a bivariate process. How does this process evolve? One individual is born whose genotype is determined by two independent choices made from the existing gamete pool and simultaneously one randomly selected individual from the existing population dies.

More precisely, let us denote by Y_n and Z_n the number of AA and aa individuals respectively, in the nth generation. Clearly Y_n and Z_n are random variables. We are interested in the bivariate process $X_n = (Y_n, Z_n)$. The model simply says that $(X_n)_{n\geq 0}$ is a Markov chain with state space $\{(k,l) : k \geq 0, l \geq 0, k+l \leq N\}$ and transition probabilities to be described shortly. Meanwhile, observe that the total number of states is $\frac{1}{2}(N+1)(N+2)$. Also, when the process is in the state (k,l), the gamete pool has the composition $A : a = (N+k-l) : (N-k+l)$. The transition probabilities from the state (k,l) to the various states are given in the following table:

State	Probability
$(k+1, l)$	$\frac{(N-k-l)(N+k-l)^2}{4N^3}$
$(k+1, l-1)$	$\frac{l(N+k-l)^2}{4N^3}$
$(k, l-1)$	$\frac{l[N^2-(k-l)^2]}{2N^3}$
$(k-1, l)$	$\frac{k[N^2-(k-l)^2]}{2N^3}$
$(k-1, l+1)$	$\frac{k(N+l-k)^2}{4N^3}$
$(k, l+1)$	$\frac{(N-k-l)(N+l-k)^2}{4N^3}$
(k, l)	$\frac{k(N+k-l)^2+l(N+l-k)^2+2(N-k-l)(N^2-(k-l)^2)}{4N^3}$

The reader can easily verify that from the state (k,l), transitions can occur only to the states listed above. As expected therefore, the second column of the above table adds to one. Note that for some boundary states like, $(k,l) = (N,0)$, ordered pairs such as $(N+1, 0)$ appear in the above list even though they are not elements of the state space. However, this poses no serious problem, because the corresponding entries for probabilities of such 'non-states' reduce to zero.

There are two absorbing states, namely, $(N,0)$ and $(0,N)$, while the other states are all transient. However, application of the fundamental matrix method does not turn out to be convenient for this absorbing chain. Here is what

we do to evaluate the absorption probabilities. Set $W_n = Y_n - Z_n$ and $\Delta W_{n+1} = W_{n+1} - W_n$, for $n \geq 0$. Then from the above table of transition probabilities, it can be easily seen that, given $X_n = (k,l)$, the random variable ΔW_{n+1} takes on the values $-2, -1, 1, 2$ and 0 with (conditional) probabilities

$$p_{-2} = \frac{k(N+l-k)^2}{4N^3}, \quad p_{-1} = \frac{(N-k+l)(N^2+3k^2-l^2-2kl)}{4N^3},$$

$$p_1 = \frac{(N+k-l)(N^2+3l^2-k^2-2kl)}{4N^3}, \quad p_2 = \frac{l(N+k-l)^2}{4N^3}, \text{ and}$$

$$p_0 = 1 - (p_2 + p_{-2} + p_1 + p_{-1})$$

respectively. A trite algebra shows that $p_1 + 2p_2 = p_{-1} + 2p_{-2}$. It follows that $E(\Delta W_{n+1} \mid X_n) = 0$, and consequently, $E(W_{n+1} \mid X_n) = W_n$. By the Markov property and the smoothing property of conditional expectations (Section 0.2), $(W_n)_{n\geq 0}$ is a bounded martingale.

Let $b_{(N,0)}(k,l)$ be the probability of eventual absorption in the state $(N,0)$ starting from (k,l). Then, conditional on $X_0 = (k,l)$, $W_\infty = \lim_n W_n$ takes the values N and $-N$ with probabilities $b_{(N,0)}(k,l)$ and $1 - b_{(N,0)}(k,l)$ respectively. But $E(W_\infty \mid X_0 = (k,l)) = W_0 = k - l$. One easily deduces that

$$b_{(N,0)}(k,l) = \frac{N+k-l}{2N} \quad \text{and} \quad b_{(0,N)}(k,l) = \frac{N-k+l}{2N}.$$

We shall now proceed to obtain the absorption rate or the rate of homozygosity. Unlike in the previous cases, the eigenvalues of the transition matrix are almost impossible to find (at least, they have not been found so far). In particular, we do not even know whether the transition matrix admits a spectral representation (in the sense discussed in Section 0.8.5). Therefore, we are not in a position to apply the method of identifying the second largest eigenvalue as the fixation rate. Moran solved the problem by an ingeneous method which we now present. In the sequel we exclude the trivial case $N = 1$ and assume that $N \geq 2$.

To motivate the idea, let us recall that if P admits a spectral representation, that is, $P = L^{-1}DL$ as in Equation (32) of Section 0.8.5, then for any function f on the state space

$$E_i(f(X_n)) = a_{i,0} + a_{i,1} + \sum_{r\geq 2} a_{i,r}\lambda_r^n, \tag{4}$$

where $1 = \lambda_0 = \lambda_1 > \lambda_2 > \lambda_3 > \cdots$ are the eigenvalues of P and, for $r \geq 0$, $a_{i,r} = \sum_j (L^{-1})_{ir} L_{rj} f(j)$. If f could be so chosen that $a_{i,2} \neq 0$, then from the above equation itself, we could have got hold of the absorption rate.

Moran turned the tables around. By purely probabilistic arguments, he found a relation of the form (4) for a suitably chosen function f and was able to derive the absorption rate directly from that relation. One of the crucial advantages of his method is that one has to only find the eigenvalues of a 2×2 matrix rather than the huge matrix P.

Let us consider the 2×2 matrix $B = \begin{pmatrix} 1 - \frac{1}{N} & \frac{1}{2N} \\ \frac{1}{N^2} & 1 - \frac{3}{2N^2} \end{pmatrix}$, which has two distinct eigenvalues given by $\mu_1 = 1 - \frac{1}{2N} - \frac{3}{4N^2} - \frac{1}{2N}\sqrt{1 - \frac{1}{N} + \frac{9}{4N^2}}$ and $\mu_2 = 1 - \frac{1}{2N} - \frac{3}{4N^2} + \frac{1}{2N}\sqrt{1 - \frac{1}{N} + \frac{9}{4N^2}}$. From $N \geq 2$, one gets that $0 < \mu_1 < \mu_2 < 1$.

Consider now the function g on our state space $S = \{(k,l) : 0 \leq k, l, k+l \leq N\}$, defined as $g(k,l) = (k+l)/N$. Then we have

Theorem 3.6: *For any transient state i, there exist numbers a_i and b_i with $b_i > 0$ such that*

$$E_i(g(X_n)) = 1 - a_i\mu_1^n - b_i\mu_2^n \,. \tag{5}$$

To get on with the main idea, we shall postpone the proof of this theorem. Instead, let us now show that μ_2 is the rate of absorption. Following the general theory, we continue to denote the set of transient states by S_T. Noting that $g(s) = 1$ for any absorbing state s, it follows that

$$1 - E_i(g(X_n)) = \sum_{s \in S_T} [1 - g(s)] p_{is}^{(n)} \,.$$

Thus from Equation (5) we have

$$\sum_{s \in S_T} [1 - g(s)] p_{is}^{(n)} = a_i\mu_1^n + b_i\mu_2^n \,,$$

so that

$$\lim_{n \to \infty} \frac{\sum_{s \in S_T} [1 - g(s)] p_{is}^{(n)}}{\mu_2^n} = b_i \,. \tag{6}$$

We shall now show that

$$0 < \limsup_{n \to \infty} \frac{\sum_{s \in S_T} p_{is}^{(n)}}{\mu_2^n} < \infty, \tag{7}$$

which, by definition, implies that μ_2 is the rate of homozygosity. We first claim that

$$0 \leq \limsup_{n} \frac{[1 - g(s)] p_{is}^{(n)}}{\mu_2^n} < \infty \quad \text{for every} \quad s \in S_T \,. \tag{8}$$

Indeed, the first inequality follows from the fact that $g(s) \leq 1$ for each s. On the other hand, if for some $s_0 \in S_T$ the limsup in (8) equals ∞, then

$$\limsup_n \frac{\sum_{s \in S_T}[1 - g(s)]p_{is}^{(n)}}{\mu_2^n} = \infty,$$

by the non-negativity of the summands above, thus contradicting (6). Our next claim is that

$$\limsup_n \frac{[1 - g(s)]p_{is}^{(n)}}{\mu_2^n} > 0 \quad \text{for some} \quad s \in S_T\,. \tag{9}$$

Indeed, if this were zero for all $s \in S_T$, then

$$\limsup_n \frac{\sum_{s \in S_T}[1 - g(s)]p_{is}^{(n)}}{\mu_2^n} = 0,$$

contradicting (6) again. Clearly, for any s as in (9), we must have $g(s) < 1$, whence it follows that for such an s

$$0 < \limsup_n \frac{p_{is}^{(n)}}{\mu_2^n} < \infty\,. \tag{10}$$

The first inequality of (7) is an immediate consequence of the first inequality of (10). To prove the other inequality of (7), we show now that for every $s \in S_T$,

$$\limsup_n \frac{p_{is}^{(n)}}{\mu_2^n} < \infty\,.$$

If $g(s) < 1$, then this follows directly from the second inequality of (8). Suppose now that $s \in S_T$ and $g(s) = 1$. For any such s, there is a $t \in S_T$ with $g(t) < 1$, such that $p_{st} > 0$. Since

$$p_{it}^{(n+1)} \geq p_{is}^{(n)} p_{st},$$

one obtains

$$\frac{p_{is}^{(n)}}{\mu_2^n} \leq \frac{p_{it}^{(n+1)}}{\mu_2^{n+1}} \frac{\mu_2}{p_{st}} \quad \text{for each } n\,.$$

The previous case now gives the desired inequality. The proof of (7) is now complete.

We now return to proving the theorem. Fixing $i \in S_T$, we denote $E_i(g(X_n))$ by u_n. The idea behind getting the relation (5) is as follows. We introduce an 'auxiliary' sequence v_n and get a recurrence relation for the vector sequence (u_n, v_n). The Equation (5) is obtained simply by solving this recurrence relation.

Proof of Theorem 3.6: Fix $i \in S_T$ and denote by u_n the quantity $E_i(g(X_n))$

as above. Define $v_n = [E_i(Y_n - Z_n)^2]/N^2$. A direct computation using the transition matrix now shows that

$$\begin{pmatrix} u_{n+1} \\ v_{n+1} \end{pmatrix} = B \begin{pmatrix} u_n \\ v_n \end{pmatrix} + \begin{pmatrix} 1/2N \\ 1/2N^2 \end{pmatrix}.$$

Using the above equation recursively one obtains

$$\begin{pmatrix} u_n \\ v_n \end{pmatrix} = B^n \begin{pmatrix} u_0 \\ v_0 \end{pmatrix} + [B^{n-1} + B^{n-2} + \cdots + I] \begin{pmatrix} 1/2N \\ 1/2N^2 \end{pmatrix}.$$

Noting that both the eigenvalues of B are different from one, so that $I - B$ is invertible, it follows that

$$\begin{pmatrix} u_n \\ v_n \end{pmatrix} = B^n \begin{pmatrix} u_0 \\ v_0 \end{pmatrix} + (I - B^n)(I - B)^{-1} \begin{pmatrix} 1/2N \\ 1/2N^2 \end{pmatrix}.$$

Thus, if we denote,

$$\widetilde{b} = \begin{pmatrix} u_0 \\ v_0 \end{pmatrix} - (I - B)^{-1} \begin{pmatrix} 1/2N \\ 1/2N^2 \end{pmatrix}, \; \widetilde{c} = (I - B)^{-1} \begin{pmatrix} 1/2N \\ 1/2N^2 \end{pmatrix},$$

then, we have

$$\begin{pmatrix} u_n \\ v_n \end{pmatrix} = B^n \widetilde{b} + \widetilde{c}.$$

Since μ_1 and μ_2 are distinct eigenvalues of B, we can diagonalize B as

$$B = L \begin{pmatrix} \mu_1 & 0 \\ 0 & \mu_2 \end{pmatrix} R,$$

with $LR = RL = I$. Observe that

$$B^n = L \begin{pmatrix} \mu_1^n & 0 \\ 0 & \mu_2^n \end{pmatrix} R.$$

Using this, one gets

$$\begin{pmatrix} u_n \\ v_n \end{pmatrix} = L \begin{pmatrix} \mu_1^n & 0 \\ 0 & \mu_2^n \end{pmatrix} R\widetilde{b} + \widetilde{c}.$$

Considering the first row of this equation one obtains

$$u_n = \alpha + \beta\mu_1^n + \gamma\mu_2^n, \tag{11}$$

for appropriate constants α, β and γ. Since both μ_1 and μ_2 are less than one it follows that α is actually $\lim_{n\to\infty} u_n$. Recall however that the chain being absorbing, $g(X_n) \longrightarrow 1$ with probability one, so that by the bounded convergence theorem, $\lim_{n\to\infty} u_n = 1$. Thus

$$\alpha = 1 \quad \text{and} \quad u_n = 1 + \beta\mu_1^n + \gamma\mu_2^n.$$

We now show that $\gamma < 0$. As a first step, we argue that γ can not be strictly positive. Indeed, if $\gamma > 0$, then $\gamma\mu_2^n = |\gamma\mu_2^n| > |\beta\mu_1^n|$, for all sufficiently large n. Here we are making use of the fact that $0 < \mu_1 < \mu_2$. From (11), this will imply that $u_n > 1$ for suficiently large n, which contradicts the fact that $u_n = E_i(g(X_n)) \leq 1$ for all n.
Next we argue that $\gamma = 0$ is impossible. Indeed, if γ were zero, then $u_n = 1 + \beta\mu_1^n$, where of course $\beta \neq 0$, because u_n cannot be identically 1. Observe also that u_n is a rational number, because it is the expectation of a random variable which takes finitely many rational values with rational probabilities. Since $u_0 = 1 + \beta$, it follows that β is also rational. Since $u_1 = 1 + \beta\mu_1$, it would follow that μ_1 has to be a rational number. However, since for every $N \geq 2$, $4N^2 - 4N + 9$ is not a perfect square (left as an exercise), μ_1 can not be rational.
Having thus proved that γ of (11) is strictly negative, the proof is now complete in view of the fact that (5) is the same as (11) after making appropriate changes in the notation and making the dependence of β and γ on i explicit. ∎

3.5.3 Diploid Bisexual Model

We shall now briefly explain the diploid bisexual model of Moran. We shall fix the population size at N_1 for males and at N_2 for females in each generation. As earlier, we consider an autosomal gene with two alleles A and a. If the population structure is $AA : Aa : aa = k : (N_1 - k - l) : l$ in males and $AA : Aa : aa = r : (N_2 - r - s) : s$ in females, then the state of the system is specified by 4-tuples (k, l, r, s). In other words, the state space is

$$\{(k, l, r, s) : k, l, r, s \geq 0\,;\, k + l \leq N_1\,;\, r + s \leq N_2\}\,.$$

When the population is in the state (k, l, r, s), then the proportion of A gametes is $\dfrac{N_1 + k - l}{2N_1}$ in males and $\dfrac{N_2 + r - s}{2N_2}$ in females. The system evolves as follows. A new individual is born. Simultaneously, a randomly selected individual from the old population dies. To preserve the population sizes in males as well as in females, we assume that the sex of the new born is the same as that of the deceased. The genotype of the new born is determined by choosing at random one gamete from the male population and one from the female population. There are again two fixation states, namely, $(N_1, 0, N_2, 0)$ and $(0, N_1, 0, N_2)$. All other states are transient.

The detailed analysis of this chain is slightly complicated and hence not included here. However, it is worth noting that the rate of fixation has been analysed by Moran. He showed that it is approximately $\left(1 - \dfrac{1}{4N_1N_2}\right)$ for large values of N_1 and N_2.

3.6 Kimura's Model

The model to be considered in this section is different from the earlier ones, both in its objective and in its analysis. The previous models aimed at studying the evolution of the aggregate population with respect to a fixed locus. In other words, it was a macro analysis. By contrast, the present model is going to be a micro-model. We still concentrate on one locus, but instead of considering the entire population, we focus our attention on one particular cell and study its evolution through cell divisions. More precisely, if an aberration enters the cell at some stage, a natural question is how this aberration propagates through subsequent generations via cell divisions. An understanding of such a question requires a little digression into cell mechanics.

To fix ideas, let us consider the locus responsible for the manufacture of haemoglobin. The message to manufacture haemoglobin is stored like a telegraphic code at this locus. However, nature tries to be cautious rather than economical. So the same code is stored in various subparts at this locus. In other words, the gene has certain subparts each having the same code. This is to ensure that the proper transmission of the code from any one part is enough to do the job. Let us assume that the gene has N subparts, say, $P_1, P_2, \ldots, P_N$, where the same code is stored.

From now on, we shall think of $P_1, P_2, \ldots, P_N$ themselves as N repetitions of the same code. Suppose that for some reason, one of them, say P_1, suddenly appears in a mutant form, that is, as a useless code, say B. These N parts now are $B, P_2, \ldots, P_N$. Suppose that the cell now undergoes the process of cell division. Recall that during the cell division, it is not the chromosome, but the chromatin content that duplicates and goes on to form sister chromatids. The chromatin content corresponding to our locus is now $B, P_2, \ldots, P_N$, so that when it doubles we have $B, B, P_2, P_2, \ldots, P_N, P_N$. According to Mendelian hypothesis, the two sister chromatids should contain $B, P_2, \ldots, P_N$ each. However, all these take place in the miniature factory of the cell. Moreover, even though we are focussing our attention on one single locus of one chromosome, it should be borne in mind that during the actual cell division the entire chromatin content gets doubled and subsequently gets realigned to form sister chromosomes. Strict obeyance of the Mendelian hypothesis means that each daughter cell is a true replica of the mother cell even upto the subpart levels. As a consequence, the level of functioning of the different genes at the different loci would remain unaltered. In other words, each daughter cell would be functionally as efficient as the mother cell. However real life experience seems to contradict it. For example, with ageing, a definite deterioration in the functioning of the cells is observed. Kimura looked for an explanation of this phenomenon by suggesting a slight deviation from the Mendelian hypothesis. More precisely, returning to the particular locus considered earlier, his suggestion is that the $2N$ subparts $B, B, P_2, P_2, \ldots, P_N, P_N$ are distributed at random into two subgroups of N parts each to form the sister chromosomes, rather than realigning in such a way as to form exact replicas $(B, P_2, \ldots, P_N)$ in each sister chromosome.

Assuming this model for cell division, what we wish to study can be precisely stated as follows. We start with a cell with configuration $(B, P_2, \ldots, P_N)$ at a locus as before. This B, as explained earlier, appeared as a mutant and useless subpart. We allow cell divisions to take place to form successive generations. Let X_n denote the number of subparts with the label B, in a typical nth generation cell. Clearly $(X_n)_{n\geq 0}$ is a stochastic process and this is the subject of our study. Of course, $X_0 = 1$ and the state space of the process is $\{0, 1, \ldots, N\}$. It is left to the reader to convince herself that $(X_n)_{n\geq 0}$ is a Markov chain with transition probabilities

$$p_{i,j} = \binom{2i}{j}\binom{2N-2i}{N-j} \Big/ \binom{2N}{N} \quad \text{for} \quad 0 \leq i, j \leq N \ .$$

It is also clear that the states 0 and N are absorbing and the remaining states are all transient. To calculate the absorption probabilities, once again observe that $(X_n)_{n\geq 0}$ is a martingale and hence the absorption probabilities are $\alpha_{i,N} = \frac{i}{N}$ and $\alpha_{i,0} = 1 - \frac{i}{N}$. Since we have started with one mutant subpart, that is $X_0 = 1$, the probability that eventually the cell has all subparts consisting of B and hence ceases to function altogether is $1/N$.

The present transition matrix is simple and all the eigenvalues can be evaluated by the same Fellerian argument used in the Wright-Fisher model. The eigenvalues are

$$\lambda_r = 2^r \binom{2N-r}{N-r} \Big/ \binom{2N}{N}, \quad \text{for } r = 0, 1, \ldots . N \ .$$

To see this, first observe the following. If $(x)_r$ denotes, as earlier, the factorial power, namely, $x(x-1)\cdots(x-r+1)$, then we know from the hypergeometric probabilities, that

$$\sum_0^N p_{i,k}(k)_r = (2i)_r \binom{2N-r}{N-r} \Big/ \binom{2N}{N} = \frac{(2i)_r}{2^r}\lambda_r \ .$$

Consider now the basis of R^{N+1} given by

$$e_0 = (1, 1, \ldots, 1)' \quad \text{and} \quad e_i = (0, 1^i, 2^i, \ldots, N^i)', \ \text{for } 1 \leq i \leq N.$$

Then for the transition matrix P, Pe_r is a linear combination of e_r, e_{r-1}, ..., e_0. Further, the coefficient of e_r in that linear combination is λ_r. Thus, with respect to the above basis, P is lower triangular with diagonal entries $\lambda_0, \lambda_1, \ldots, \lambda_N$. Since the eigenvalues do not depend on the choice of the basis, this shows that λ_r, $0 \leq r \leq N$, are the eigenvalues.

Noting that $\lambda_0 = \lambda_1 = 1$ and $\lambda_2 = (2N-2)/(2N-1) > \lambda_3 > \cdots > \lambda_N$, it follows that the rate of absorption is $1 - \frac{1}{2N-1}$. However, in this case, the two absorbing states are not on an equal footing. The state N corresponds to death of the cell as far as this locus is concerned, while state 0 corresponds to the

complete disappearance of the mutant subpart. The quantity of real interest, therefore, is the probability of getting absorbed in the state N at time n, that is,

$$d_N^n = P(X_n = N \text{ and } X_{n-1} \neq N \,|\, X_0 = 1)\,.$$

The values of d_N^n can be evaluated for small values of N (see Exercise 8). Kimura has given continuous approximations for large values of N using hypergeometric functions. An extension of the above idea for n-ploid organisms with N sets of chromosomes has also been considered by Kimura.

3.7 W-F Model with Varying Generation Sizes

In this section, we shall discuss a modification of the Wright-Fisher model which allows the population size to change from generation to generation. As in the classical Wright-Fisher model, we still consider a haploid population with two alleles A and a. However unlike in the classical case, here we will allow the size of the nth generation, $2N_n$, to possibly depend on n. But of course, the stochastic mechanism of formation of new generation from the previous one is still the same, namely, by drawing a simple random sample, with replacement, of an appropriate size.

3.7.1 Deterministic Generation Sizes

Let $(N_n)_{n\geq 0}$ be a fixed sequence of positive integers with N_n denoting the size of the n-th generation. As before, denote by X_n, the number of A genes in the n-th generation. The conditional distribution of X_{n+1}, given $X_n = j$, is assumed to be Binomial with parameters $(2N_{n+1}, \frac{j}{2N_n})$ irrespective of the past history of the evolution. Thus the sequence $(X_n)_{n\geq 0}$ is assumed to have the Markov property. However, we cannot call it a Markov chain in our sense, because the transition probabilities depend also on n. Of course if $N_n = N$ for each n, as in the classical case, then we have a traditional Markov chain. In this special case, $(X_n)_{n\geq 0}$ was indeed seen to be an absorbing chain so that fixation occurs with probability one.

The question that we address here is what happens in the case of varying population sizes. Our previous experience with a similar situation, namely, Malecot's Theorem of Chapter 2, tempts us to conjecture that probability of eventual fixation is one if and only if $\sum \frac{1}{N_n} = \infty$. We show that this is indeed the case.

At this point, of course, the notion of fixation should be clearly understood. We do not have an absorbing chain in the usual sense. Indeed, the process here is not even a time-homogeneous Markov chain, as in Section 0.8. In other words, although markovian property holds, but the transition probabilities are not the same over generations. Therefore, we cannot as such use the terminology and theory of absorption as discussed in Section 0.8. The usual and natural interpretation of fixation is that of reaching a stage where an entire generation

consists of only one kind of individuals, say, either all a or all A. This is the same as saying that for some n, X_n becomes equal to 0 or $2N_n$. Notice that if this actually happens in some generation, then the same phenomenon persists with probability one in all subsequent generations. It turns out to be convenient to work with the proportion rather than the absolute number of A genes, that is, to work with the random variables $Y_n = \frac{X_n}{2N_n}$ rathr than X_n. Fixation in the above sense then reduces to Y_n becoming 0 or 1 eventually. This being too stringent in the context of varying sizes, we will take a broader view of fixation. Instead of requiring that $Y_n = 0$ or 1 for some n, we would simply ask whether $Y_n \longrightarrow 0$ or 1. Of course, a pertinent question is whether Y_n converges at all. We will argue that $Y = \lim_n Y_n$ exists with probability one. The precise meaning of fixation that we use is in terms of this random variable Y as stated above. We say that *fixation occurs* if and only if $Y = 0$ or 1.

Theorem 3.7:
(a) $(Y_n)_{n\geq 0}$ *is a martingale and* $Y = \lim_n Y_n$ *exists with probability one.*
(b) *Assume that* $P(0 < Y_0 < 1) > 0$. *Then* $P(Y = 0 \text{ or } 1) = 1$ *iff* $\sum \frac{1}{N_n} = \infty$.

Proof: (a) Since the conditional distribution of X_{n+1}, given $X_0, X_1, \ldots, X_n$, is $B(2N_{n+1}, \frac{X_n}{2N_n})$, we have $E(X_{n+1} \mid X_0, \ldots, X_n) = 2N_{n+1}\frac{X_n}{2N_n}$. It follows that (Y_n) is a martingale. Since $0 \leq Y_n \leq 1$, the martingale convergence theorem implies that $Y = \lim_n Y_n$ exists with probability one.
(b) It is easy to see that for $Z \sim B(m,p)$, $E\left(\frac{Z}{m}\left(1 - \frac{Z}{m}\right)\right) = p(1-p)(1-\frac{1}{m})$. Therefore,

$$E(Y_{n+1}(1 - Y_{n+1}) \mid X_0, \ldots, X_n) = Y_n(1 - Y_n)\left(1 - \frac{1}{2N_{n+1}}\right),$$

and hence by smoothing property

$$E(Y_{n+1}(1 - Y_{n+1})) = \left(1 - \frac{1}{2N_{n+1}}\right) E(Y_n(1 - Y_n)).$$

It follows that

$$E(Y_n(1 - Y_n)) = E(Y_0(1 - Y_0)) \prod_1^n \left(1 - \frac{1}{2N_k}\right).$$

By the Dominated Convergence Theorem now, one gets

$$E(Y(1 - Y)) = E(Y_0(1 - Y_0)) \prod_{n=1}^{\infty} \left(1 - \frac{1}{2N_n}\right).$$

The hypothesis implies that $E(Y_0(1 - Y_0)) > 0$. Therefore $E(Y(1 - Y)) = 0$ if and only if $\prod(1 - \frac{1}{2N_n}) = 0$, or equivalently, if and only if $\sum \frac{1}{N_n} = \infty$. The proof is completed by noting that, since $0 \leq Y \leq 1$ with probability one, the condition $E(Y(1 - Y)) = 0$ is equivalent to $P(Y = 0 \text{ or } 1) = 1$. ∎

The hypothesis $P(0 < Y_0 < 1) > 0$ simply means that the initial state is 'transient' with positive probability. In the classical case, namely, $N_n = N$ for each n, the infinite series of Theorem 3.7(b) is clearly divergent and hence $P(Y = 0 \text{ or } 1) = 1$. Of course, in this special case $Y = 0$ or 1 if and only if the Y_n are eventually 0 or 1. Thus $P(Y_n = 0 \text{ or } 1 \text{ eventually }) = 1$. This is consistent with what we otherwise derived earlier for the fixed generation size model. The main contention of (b) is that for polymorphism to occur with positive probability in the 'limiting' population, it is necessary and sufficient for the generation sizes to explode at a sufficiently fast rate. The remarks made after Malecot's Theorem in Chapter 2 are equally relevant here.

3.7.2 Random Generation Sizes

One can get a little more ambitious and consider the case when the N_n are themselves random rather than a fixed sequence of numbers. Thus now the X_n as well as the N_n are random variables. We shall prove results analogous to the previous section. The main ideas are borrowed from C. C. Heyde and E. Seneta. Even though the N_n are random, our results do not require any stipulation on the distribution of these random variables. However the evolution of the process $(X_n)_{n\geq 0}$ is driven by a stochastic mechanism analogous to that of the previous section. To be precise, we assume that given $(X_0, N_0), (X_1, N_1), \ldots, (X_n, N_n)$ and N_{n+1}, the conditional distribution of X_{n+1} is again $B(2N_{n+1}, \frac{X_n}{2N_n})$. As before, denoting $Y_n = \frac{X_n}{2N_n}$, it follows that

$$E(Y_{n+1} \mid X_i, i \leq n \; ; \; N_i, i \leq n+1) = Y_n, \tag{12}$$

and,

$$E(Y_{n+1}(1-Y_{n+1}) \mid X_i, i \leq n \; ; \; N_i, i \leq n+1) = \left(1 - \frac{1}{2N_{n+1}}\right) Y_n(1-Y_n)\,. \tag{13}$$

Thus $(Y_n)_{n\geq 0}$ is a martingale and by the martingale convergence theorem, $Y = \lim_n Y_n$ exists with probability one. We continue to say that *fixation occurs* if and only if $Y = 1$ or 0 with probability one, or equivalently $E(Y(1-Y)) = 0$. A possible generalization of Theorem 3.7(b) would say that $P(Y = 0 \text{ or } 1) = 1$ if and only if $P(\sum \frac{1}{N_n} = \infty) = 1$. We first show that the *if* part of the statement is indeed correct.

Theorem 3.8:

(a) *Let* $U_n = Y_n(1-Y_n) + \sum_{k=0}^{n-1} \frac{1}{2N_{k+1}} Y_k(1-Y_k)$. *Then* $(U_n)_{n\geq 0}$ *is a martingale.*

(b) *The series* $\sum_0^\infty \frac{1}{N_{n+1}} Y_n(1-Y_n)$ *converges with probability one.*

Proof: (a) Using Equation (13),

$$\begin{aligned} E(U_n \mid X_i, i \le n-1; N_i, i \le n) &= (1 - \tfrac{1}{2N_n})Y_{n-1}(1-Y_{n-1}) \\ &\quad + \textstyle\sum_{k=0}^{n-1} \frac{1}{2N_{k+1}} Y_k(1-Y_k) \\ &= Y_{n-1}(1-Y_{n-1}) + \textstyle\sum_{k=0}^{n-2} \frac{1}{2N_{k+1}} Y_k(1-Y_k) \\ &= U_{n-1}\,. \end{aligned}$$

This shows that $(U_n)_{n\ge 0}$ is a martingale.
(b) Since the U_n are all non-negative, by the martingale convergence theorem, U_n converges with probability one. Since $Y_n(1-Y_n)$ also converges with probability one, we conclude that the series $\sum_{k=0}^{\infty} \frac{1}{2N_{n+1}} Y_n(1-Y_n)$ converges with probability one. ∎

Theorem 3.9: *If* $P(\sum \frac{1}{N_n} = \infty) = 1$, *then* $P(Y = 0 \text{ or } 1) = 1$.

Proof: Theorem 3.8(b) implies that whenever $\lim_n Y_n(1-Y_n) > 0$, the series $\sum \frac{1}{N_n}$ has to converge. Thus, if $P(\sum \frac{1}{N_n} = \infty) = 1$, then $\lim_n Y_n(1-Y_n) = 0$ with probability one, completing the proof. ∎

Remark: The proof of Theorem 3.9 shows something stronger. It actually shows that with probability one the occurrence of the event $(\sum \frac{1}{N_n} = \infty)$ implies the occurrence of the event $(Y = 0 \text{ or } 1)$. More precisely,

$$P\left(\sum \frac{1}{N_n} = \infty \text{ and } 0 < Y < 1\right) = 0\,.$$

Whether the converse holds is not known. Here are however a couple of results in that direction.

Theorem 3.10: *Assume that* $P(0 < Y_0 < 1) > 0$.
(a) *If for each* N, $\frac{1}{N_n} < \alpha_n$ *where* (α_n) *is a sequence of real numbers with* $\sum \alpha_n < \infty$, *then* $P(0 < Y < 1) > 0$.
(b) *If for each* n, N_{n+1} *is independent of* (X_n, N_n) *and* $\sum_0^{\infty} E(\frac{1}{N_n}) < \infty$, *then* $P(0 < Y < 1) > 0$.

Proof: (a) Taking expectations on both sides of Equation (13) and using the fact that $1 - \frac{1}{2N_{n+1}} \le 1$, we get

$$E(Y_{n+1}(1-Y_{n+1})) \le E(Y_n(1-Y_n)) \quad \text{for all } n\,. \tag{14}$$

From the definition of U_n, we get that, for any $n < m$,

$$U_m - U_n - [Y_m(1-Y_m) - Y_n(1-Y_n)] = \sum_{k=n}^{m-1} \frac{1}{2N_{k+1}} Y_k(1-Y_k)\,.$$

Taking expectations on both sides and using that $(U_n)_{n\ge 0}$ is a martingale, we get

$$E[Y_n(1-Y_n)] - E[Y_m(1-Y_m)] = \sum_{k=n}^{m-1} E\left[\frac{1}{2N_{k+1}} Y_k(1-Y_k)\right]\,.$$

Letting $m \to \infty$ and using Dominated Convergence Theorem, one obtains

$$E[Y_n(1-Y_n)] - E[Y(1-Y)] = \sum_{k=n}^{\infty} E\left[\frac{1}{2N_{k+1}} Y_k(1-Y_k)\right]. \qquad (15)$$

Under the hypothesis, the right side of the above equation is

$$\leq \sum_{k=n}^{\infty} \frac{1}{2}\alpha_{k+1} E(Y_k(1-Y_k)) \quad \leq E(Y_n(1-Y_n)) \sum_{k=n}^{\infty} \frac{1}{2}\alpha_{k+1},$$

where the second inequality is a consequence of (14). It is easy to see that the assumption $P(0 < Y_0 < 1) > 0$ implies $E(Y_n(1-Y_n)) > 0$ for all n. Since $\sum \alpha_n < \infty$, by choosing n so that $\sum_{k=n}^{\infty} \alpha_{k+1} < 2$, one gets that for such n, the right side of (15) is strictly smaller than $E(Y_n(1-Y_n))$. It follows that $E(Y(1-Y)) > 0$, thus completing the proof of (a).
(b) We notice that the right side of (15) equals

$$\sum_{k=n}^{\infty} E\left(\frac{1}{2N_{k+1}}\right) E(Y_k(1-Y_k)) \quad \leq E[Y_n(1-Y_n)] \sum_{k=n}^{\infty} E\left(\frac{1}{2N_{k+1}}\right).$$

Again working with an n such that $\sum_{k=n}^{\infty} E(\frac{1}{2N_{k+1}}) < 1$, the proof can be completed as before. ∎

3.8 Diffusion Approximations

We shall return to the Wright-Fisher model of Section 3.4. Recall that the W-F model is a model for the evolution of gene frequencies for a gene with two alleles in a haploid population, whose size is constrained to be always $2N$. More precisely, if the two alleles are named as A and a, and if X_n denotes the number of A genes in the nth generation, then X_n is a Markov chain with state space $\{0, 1, \ldots, 2N\}$ and transition probabilities

$$p_{i,j} = \binom{2N}{i} \left(\frac{i}{2N}\right)^j \left(1 - \frac{i}{2N}\right)^{2N-j}.$$

We know that this is an absorbing chain, with 0 and $2N$ being the absorbing states. Further we know that given $X_0 = i$, the probability of eventual absorption at $2N$ is $\frac{i}{2N}$, while the rate of absorption is $1 - \frac{1}{2N}$. However, we still do not have any formula for the mean absorption time or the variance of the absorption time. While in principle these are completely determined from the transition matrix, in practice it is extremely difficult to find nice formulae. It turns out however, that one can get relatively simple approximations for these quantities. The main idea is to approximate the underlying Markov chain with

an appropriate diffusion on $[0,1]$ and then evaluate these quantities for the diffusion process by the techniques outlined in Section 0.9.2.

To mimic the Markov chain by a diffusion process on $[0,1]$, it is first of all necessary to change the state space of the discrete chain so as to take values in $[0,1]$ only. This is easily achieved by considering the fraction rather than the number of A genes. This would give a Markov chain on the state space $\{0, \frac{1}{2N}, \frac{2}{2N}, \ldots, \frac{2N-1}{2N}, 1\}$ and transition probabilities

$$\widetilde{p}_{\frac{i}{2N},\frac{j}{2N}} = p_{i,j}$$

We shall denote this Markov chain by $\widetilde{X}_n$, which is still a discrete time chain. To proceed further, we observe a simple lemma.

Lemma 3.11: *For each $N \geq 1$, let $X_N \sim B(2N, \theta_N)$. Let $Y_N = \frac{X_N}{2N} - \theta_N$. Assume that $\theta_N \to \theta$ $(0 < \theta < 1)$, as $N \to \infty$. Then,*

$$\begin{aligned} E(Y_N) &= 0, \\ V(Y_N) &= \tfrac{1}{2N}\theta(1-\theta) + o(\tfrac{1}{2N}), \\ E|Y_N|^k &= o(\tfrac{1}{2N}) \quad \textit{for any } k \geq 3\,. \end{aligned}$$

Proof: The first two are simple consequences of the properties of Binomial distribution. For the third, we first show that for $k \geq 1$, $E|X_N - 2N\theta_N|^k \leq C_k N^{k/2}$ for some constant C_k. Denoting $(U_i)_{1 \leq i \leq 2N}$ to be the mean zero i.i.d random variables taking values $1-\theta_N$ and $-\theta_N$ with probabilities θ_N and $1-\theta_N$ respectively, it is clear that $E(X_N - 2N\theta_N)^{2k} = E(\sum U_i)^{2k}$. Using independence and mean zero property, it is easy to see that the only terms in the expansion of $(\sum U_i)^{2k}$ having non-zero expectation are those, where a U_i appears with either power zero or power at least two. Now using the identicality of distributions, the reader can easily verify that

$$\begin{aligned} E(X_N - 2N\theta_N)^{2k} &\leq \tbinom{2N}{k} E(U_1 + \cdots + U_k)^{2k} \\ &\leq \tbinom{2N}{k} k^{2k+1} E(U_1^{2k}) \leq (2N)^k \tfrac{k^{2k+1}}{4\,k!}\,. \end{aligned}$$

Therefore,

$$E|X_N - 2N\theta_N|^k \leq \sqrt{E(X_N - 2N\theta_N)^{2k}} \leq \sqrt{(2N)^k \frac{k^{2k+1}}{4\,k!}} = C_k N^{k/2}\,.$$

It now follows that for $k \geq 3$,

$$2N E|Y_N|^k = (2N)^{1-k} E|X_N - 2N\theta_N|^k \leq 2^{1-k} C_k N^{1-\frac{k}{2}} \to 0 \text{ as } N \to \infty\,.$$

∎

Getting back to our discrete markov chain $\widetilde{X}_n$, denote the increment in one step, namely, $\widetilde{X}_{n+1} - \widetilde{X}_n$ by $\Delta\widetilde{X}_{n+1}$. Let $0 < x < 1$. It follows from Lemma

3.11 that

$$\begin{aligned} E\left(\Delta\widetilde{X}_{n+1} \mid \widetilde{X}_n = \tfrac{[2Nx]}{2N}\right) &= 0, \\ V\left(\Delta\widetilde{X}_{n+1} \mid \widetilde{X}_n = \tfrac{[2Nx]}{2N}\right) &= \tfrac{1}{2N}x(1-x) + o(\tfrac{1}{2N}), \\ E\left(|\Delta\widetilde{X}_{n+1}|^k \mid \widetilde{X}_n = \tfrac{[2Nx]}{2N}\right) &= o(\tfrac{1}{2N}) \quad \text{for } k \geq 3\,. \end{aligned}$$

These equations look somewhat similar to the Equations (38)–(40) of Section 0.9.2, of course, with some obvious discrepancies. To remove these discrepancies and to get an exact match we need to do something more. The idea behind going from number of A genes to the fraction was to scale the state space of the chain to bring it down to the unit interval. The next thing we need is to use a different time scale also. The right scaling of the time that does the job is given by the following. Roughly what was one unit of time in the original chain will now become $1/2N$ units of time in the new time scale, or equivalently, one time unit in the new scale corresponds to $2N$ generations for our original population model. To be precise, we define a new discrete time process X'_t, indexed not by integers, but by t of the form $\frac{n}{2N}$, $n \geq 0$ as $X'_{\frac{n}{2N}} = \widetilde{X}_n$. Now denoting $t = \frac{n}{2N}$, and $h = \frac{1}{2N}$, the above equations can be rewritten as

$$\begin{aligned} E\left(X'_{t+h} - X'_t \mid X'_t = \tfrac{[2Nx]}{2N}\right) &= 0, \\ E\left(|X'_{t+h} - X'_t|^2 \mid X'_t = \tfrac{[2Nx]}{2N}\right) &= x(1-x)h + o(h), \\ E\left(|X'_{t+h} - X'_t|^k \mid X'_t = \tfrac{[2Nx]}{2N}\right) &= o(h) \quad \text{for } k \geq 3\,. \end{aligned}$$

These equations suggest that our Markov chain, after appropriate scaling in both time and space, should approximate — at least for large N — a continuous time Markov process (Z_t) with state space [0,1] satisfying Equations (38)–(40) of Section 0.9.2, with $a(x) = 0$ and $b(x) = x(1-x)$. By this we mean that, if we consider the diffusion process $(Z_t)_{t\geq 0}$ with $a(x)$ and $b(x)$ as above and with 0 and 1 as absorbing states, then the Markov chain $\widetilde{X}_n$ is approximately same as the chain obtained by observing this continuous process at time points $\{\frac{n}{2N}, n \geq 0\}$. That this is indeed true can be proved rigorously. Interested reader may look up the book by Ethier & Kurtz.

From the theory outlined in Section 0.9.2, the expected time of absorption for the diffusion process (Z_t) starting from the state x is given by the equation

$$T(x) = -2\int_0^x [y(1-y)]^{-1}(x-y)dy \;+\; 2x\int_0^1 [y(1-y)]^{-1}(1-y)dy\,.$$

Note that $\psi \equiv 1$ here. Trite calculation now yields

$$T(x) = -2[x\log x \;+\; (1-x)\log(1-x)]\,.$$

In view of the fact that $(Z_t)_{t\geq 0}$ is continuous time approximation for the discrete time chain (X'_t) and that (X'_t) is nothing but an appropriately scaled (with regard to time and space) modification of the original chain (X_n), we conclude the following. Wright-Fisher chain starting with i many A genes is expected to take approximately

$$t^*(i) \quad = \quad -4N\left[\frac{i}{2N}\log\frac{i}{2N} + (1-\frac{i}{2N})\log(1-\frac{i}{2N})\right]$$

generations for fixation. This is a reasonably good approximation, which is simple to compute. For example starting with 1 (or $2N-1$) A genes, it is expected to take approximately $2+2\log 2N$ generations to get absorbed; while, if initially half the genes are of one type, it takes approximately $4N\log 2 \sim 2.8N$ generations. It should not be surprising that, in case of equal initial frequencies, it takes a very long time for absorption. This is in confirmity with the fact that the second largest eigenvalue of the transition matrix is $1-\frac{1}{2N}$ which is very close to unity.

The same technique can be used to get simple approximation to the mean absorption time in Moran model too. We shall briefly outline the steps. Denoting by (X_n), the Markov chain of the Moran model with $2N$ haploids and denoting $\widetilde{X}_n = \frac{X_n}{2N}$ we obtain

$$\begin{aligned} E(\Delta\widetilde{X}_{n+1} \,|\, \widetilde{X}_n = \tfrac{i}{2N}) &= 0, \\ V(\Delta\widetilde{X}_{n+1} \,|\, \widetilde{X}_n = \tfrac{i}{2N}) &= \tfrac{1}{2N^2}\tfrac{i}{2N}(1-\tfrac{i}{2N}), \\ E(|\Delta\widetilde{X}_{n+1}|^k \,|\, \widetilde{X}_n = \tfrac{i}{2N}) &= o(\tfrac{1}{2N^2}) \quad \text{for } k \geq 3. \end{aligned}$$

As in the case of the W-F model, we now need to introduce an appropriate scaling of time. The obvious choice is to consider the process (X'_t) indexed by t of the form $\frac{n}{2N^2}$, for $n \geq 0$, defined as $X'_{\frac{n}{2N^2}} = \widetilde{X}_n$. It is clear then that the same diffusion process considered for the Wright-Fisher model (with $a(x)=0$ and $b(x)=x(1-x)$) is a continuous time approximation to the discrete time process (X'_t) in the sense described earlier.

It follows therefore that the Moran model starting with i many A genes is expected to take approximately

$$t^{**}(i) = -4N^2\left[\frac{i}{2N}\log\frac{i}{2N} + (1-\frac{i}{2N})\log(1-\frac{i}{2N})\right]$$

generations for fixation. Comparing the expressions for $t^*(i)$ and $t^{**}(i)$, we see that

$$t^{**} \quad = \quad Nt^* \tag{16}$$

In other words it takes N times longer in the Moran model to reach fixation than in the Wright-Fisher model.

Of course in the W-F model, in each generation $2N$ individuals die and $2N$ new individuals are born, whereas in the Moran model, only one individual dies

and a new one born in passing from one generation to the next. Thus roughly speaking, $2N$ birth-death events of the Moran model should correspond to one generation in the Wright-Fisher model. Thus one would expect t^{**} to equal $2Nt^*$. The discrepancy between this and Equation (16) is just a factor of 2, which also can be explained. However this is somewhat involved and hence we omit here.

3.9 Exercises

1. Verify the details of Selfing model. Show that P has eigenvalues 1 and $1/2$. Show that (i) $(1,1,1)'$ and $(1,2,3)'$ are two right eigenvectors associated to the eigenvalue 1 and $(0,1,0)'$ is a right eigenvector associated to the eigenvalue $1/2$; (ii) $(3/2,0,-1/2)$ and $(-1/2,0,1/2)$ are two left eigenvectors associated to the eigenvalue 1 and $(-1/2,1,-1/2)$ is a left eigenvector associated to the eigenvalue $1/2$. Calculate $p_{2,i}^{(n)}$ for $i = 1,2,3$ and the absorption probabilities $b_{2,1}$ and $b_{2,3}$. Here 1, 2 and 3 represent the states AA, Aa and aa respectively.

2. Verify the details of Sibmating matrix. Show that the eigenvalues are given by $\lambda_1 = \lambda_2 = 1$, $\lambda_3 = \frac{1+\sqrt{5}}{4}$, $\lambda_4 = \frac{1}{2}$, $\lambda_5 = \frac{1}{4}$ and $\lambda_6 = \frac{1-\sqrt{5}}{4}$. Thus the rate of absorption is given by $\frac{1+\sqrt{5}}{4}$. Show that the fundamental matrix is given by

$$N = \begin{pmatrix} 8/3 & 2/3 & 1/6 & 4/3 \\ 2/3 & 8/3 & 1/6 & 4/3 \\ 4/3 & 4/3 & 4/3 & 8/3 \\ 4/3 & 4/3 & 1/3 & 8/3 \end{pmatrix}.$$

   Calculate the absorption times and the absorption probabilities. Can you give a heuristic justification for your answers?

3. In the usual Sibmating model, two parents are selected at random. Let us continue sibmating, but, following Kemeny and Snell, change the procedure as follows. Select one offspring who is then allowed to select a mate. Assume that A is dominant and a is recessive. In the selection, assume that the odds ratio is $\alpha : 1$ for an offspring for picking someone with a different phenotype to picking someone of the same phenotype. In other words, an offspring selects someone like itself with probability $1/(\alpha+1)$. Of course, we also assume that an individual in the phenotype given by AA or Aa can be any one of the two with equal probabilities.

   (a) Show that with appropriate naming of the states the transition matrix takes the form

$$\begin{pmatrix} 1 & 0 & 0 & 0 & 0 & 0 \\ 0 & 1 & 0 & 0 & 0 & 0 \\ \frac{1}{4} & 0 & \frac{1}{2} & 0 & 0 & \frac{1}{4} \\ 0 & \frac{1}{2(\alpha+1)} & 0 & \frac{\alpha}{(\alpha+1)} & 0 & \frac{1}{2(\alpha+1)} \\ 0 & 0 & 0 & 0 & 0 & 1 \\ \frac{1}{4(\alpha+3)} & \frac{1}{4(3\alpha+1)} & \frac{1}{(\alpha+3)} & \frac{2\alpha(\alpha+1)}{(\alpha+3)(3\alpha+1)} & \frac{\alpha(\alpha+1)}{(\alpha+3)(3\alpha+1)} & \frac{1}{(\alpha+3)} \end{pmatrix}.$$

(b) Show that the fundamental matrix N is $\frac{1}{(2\alpha+1)(\alpha+3)} \times$

$$\begin{pmatrix} 4(\alpha^2+5\alpha+2) & 2\alpha(\alpha+1)^2 & \alpha(\alpha+1) & (3\alpha+1)(\alpha+3) \\ 2(3\alpha+1) & (4\alpha^2+9\alpha+3)(\alpha+1) & \alpha(\alpha+1) & (3\alpha+1)(\alpha+3) \\ 4(3\alpha+1) & 4\alpha(\alpha+1)^2 & 4\alpha^2+9\alpha+3 & 2(3\alpha+1)(\alpha+3) \\ 4(3\alpha+1) & 4\alpha(\alpha+1)^2 & 2\alpha(\alpha+1) & 2(3\alpha+1)(\alpha+3) \end{pmatrix}.$$

(c) Show that the average time for absorption m is given by

$$m = \frac{1}{(2\alpha+1)(\alpha+3)} \begin{pmatrix} 2\alpha^3+12\alpha^2+33\alpha+11 \\ 4\alpha^3+17\alpha^2+29\alpha+8 \\ 4\alpha^3+18\alpha^2+45\alpha+13 \\ 4\alpha^3+16\alpha^2+38\alpha+10 \end{pmatrix}.$$

(d) Show that the absorption probabilities B are given by

$$B = \frac{1}{4(2\alpha+1)(\alpha+3)} \begin{pmatrix} 4\alpha^2+23\alpha+9 & 4\alpha^2+5\alpha+3 \\ 9\alpha+3 & 8\alpha^2+19\alpha+9 \\ 18\alpha+6 & 8\alpha^2+10\alpha+6 \\ 18\alpha+6 & 8\alpha^2+10\alpha+6 \end{pmatrix}.$$

(e) Calculate m for $\alpha = 0, 1, 2$.

(f) Show that for $\alpha > 1$, the probability of absorption in $aa \times aa$ increases with α. The reason is that a large α favours the recessive strain a. Explain why this is so.

(g) Show that as α decreases from 1, the probability of absorption in $AA \times AA$ increases until it reaches a maximum at $\alpha = 1/3$ and then decreases and at $\alpha = 0$ the absorption probability is the same as at $\alpha = 1$.

(h) Deduce that if, mating takes place only between the same phenotypes, then the probabilities of absorption are the same as for random mating. Of course, time for absorption should be much less here. Show this.

4. A square matrix A with non-negative entries is called *regular* if some power of A has strictly positive entries. Such a matrix A has an eigenvalue $\lambda > 0$, which is simple, and all other eigenvalues of A are strictly smaller than λ in modulus. Moreover, associated to λ there is a right eigenvector with strictly positive entries. Also, λ is the only eigenvalue which has a right eigenvector with strictly positive entries. This is known as *Perron-Frobenius Theorem.* Read a proof of this.

5. In the Wright-Fisher model for N diploids, show that

$$E(f(X_{n+1}) \,|\, X_n) = \left(1 - \frac{1}{2N}\right) f(X_n),$$

where f is the function $f(k) = k(2N - k)$ for $0 \leq k \leq 2N$.
Use this to conclude that $1 - \frac{1}{2N}$ is the second largest (in modulus) eigenvalue after 1.

6. Consider the Wright-Fisher model with mutation where $A \to a$ with chance α and $a \to A$ with chance β. As usual, X_n denotes the number of A genes in the n-th generation.

   (a) Argue that $(X_n)_{n\geq 0}$ is a markov chain with state space $\{0, 1, \ldots, 2N\}$ and transition probabilities given by

$$p_{j,k} = \binom{2N}{k} p_j^k q_j^{2N-k}.$$

   where $p_j = \frac{j}{2N}(1-\alpha) + (1 - \frac{j}{2N})\beta$ and $q_j = 1 - p_j$.

   (b) If $\alpha + \beta = 1$ then show that (X_n) is an i.i.d. sequence.

   (c) If $\alpha + \beta = 2$ then what happens?
   Assume from now on that $\alpha, \beta > 0$ and $\alpha + \beta < 1$.

   (d) Show that the eigenvalues of the transition matrix are

$$\lambda_r = (1 - \alpha - \beta)^r \binom{2N}{r} \frac{r!}{(2N)^r} \quad \text{for} \quad 0 \leq r \leq 2N.$$

   (e) Show that (X_n) is an aperiodic irreducible markov chain.

   The stationary distribution is difficult to obtain. But some idea of it can be got as follows. Let $Y_n = \frac{X_n}{2N}$ denote the proportion of A genes in the n-th generation. Assume any initial distribution for the chain.

   (f) Show that $E(Y_{n+1}) = (1 - \alpha)E(Y_n) + \beta[1 - E(Y_n)]$.
   Deduce that $\lim_{n\to\infty} E(Y_n) = \frac{\beta}{\alpha+\beta}$.

   (g) Show that

$$V(Y_{n+1} | Y_n) = \frac{1}{2N}[(1-\alpha)Y_n + \beta(1-Y_n)][1 - (1-\alpha)Y_n - \beta(1-Y_n)].$$

(h) Deduce that

$$\lim_{n\to\infty} E(Y_n^2)\left[1-\left(1-\frac{1}{2N}\right)(1-\alpha-\beta)^2\right] =$$

$$\beta^2\left(\frac{2-\alpha-\beta}{\alpha+\beta}\right)+\frac{\beta(1-2\alpha)(1-\alpha-\beta)}{2N(\alpha+\beta)}+\frac{\beta(1-\beta)}{2N}.$$

(i) Conclude that $\lim_{n\to\infty} V(Y_n)$ is of the order of $\frac{1}{2N}$.

7. Consider the Haploid model of Moran with mutation, where $A \to a$ with chance α and $a \to A$ with chance β. Assume that, $\alpha, \beta > 0$ and $\alpha+\beta < 1$. Let X_n denote the number of A genes in the n-th generation.

(a) Show that $(X_n)_{n\geq 0}$ is an aperiodic markov chain with the state space $\{0, 1, \ldots, M\}$ and transition probabilities given by

$$p_{j,j-1} = \frac{j}{M}q_j,\ p_{j,j+1} = \left(1-\frac{j}{M}\right)p_j,\ p_{j,j} = \frac{j}{M}p_j + \left(1-\frac{j}{M}\right)q_j,$$

where $p_j = (1-\alpha)\frac{j}{M} + \beta(1-\frac{j}{M})$ and $q_j = 1-p_j$.
Let $\pi = (\pi_0, \ldots, \pi_M)$ be the stationary distribution.

(b) Show that

$$\begin{aligned}\pi_k &= \pi_{k-1}\frac{M-k+1}{kq_k}p_{k-1}\\ &= \pi_0\frac{M(M-1)\cdots(M-k+1)}{k(k-1)\cdots 1}\frac{p_0p_1\cdots p_{k-1}}{q_1q_2\cdots q_k}\end{aligned}$$

and hence evaluate π_k, $0 \leq k \leq M$.

(c) Prove the following by induction on M.
If $u_j = x + jy$, $v_j = 1 - x - jy$ for two real numbers x, y and if S_M is defined by

$$S_M(x,y) = \prod_1^M v_i + \binom{M}{1}u_0\prod_2^M v_i + \binom{M}{2}u_0u_1\prod_3^M v_i + \prod_0^{M-1} u_i,$$

then show that

$$S_M(x,y) = (1-y)(1-2y)\cdots(1-My).$$

(d) Use the above with $x = \beta$ and $y = \frac{1-\alpha-\beta}{M}$ to show that

$$1+\sum_{k=1}^M\binom{M}{k}\frac{p_0\cdots p_{k-1}}{q_1\cdots q_k} = \frac{1}{q_1\cdots q_M}\left[\sum_{i=0}^M\binom{M}{i}\prod_{j<i}p_j\prod_{j>i}q_j\right]$$

$$= \frac{\left(1-\frac{1-\alpha-\beta}{M}\right)\left(1-\frac{2(1-\alpha-\beta)}{M}\right)\cdots\left(1-\frac{M(1-\alpha-\beta)}{M}\right)}{\left(1-\beta-\frac{1-\alpha-\beta}{M}\right)\left(1-\beta-\frac{2(1-\alpha-\beta)}{M}\right)\cdots\left(1-\beta-\frac{M(1-\alpha-\beta)}{M}\right)}.$$

(e) Deduce that

$$\pi_0 = \frac{\Gamma\left(\frac{M(1-\beta)}{1-\alpha-\beta}\right) \quad \Gamma\left(\frac{M(\alpha+\beta)}{1-\alpha-\beta}\right)}{\Gamma\left(\frac{M}{1-\alpha-\beta}\right) \quad \Gamma\left(\frac{M\alpha}{1-\alpha-\beta}\right)}.$$

(f) Show that

$$\pi_k = \pi_0 \frac{\Gamma(M+1)\Gamma\left(\frac{M\beta}{1-\alpha-\beta}+k\right)\Gamma\left(\frac{M(1-\beta)}{1-\alpha-\beta}-k\right)}{\Gamma(k+1)\Gamma(M-k+1)\Gamma\left(\frac{M\beta}{1-\alpha-\beta}\right)\Gamma\left(\frac{M(1-\beta)}{1-\alpha-\beta}\right)}.$$

(g) Now suppose that $M \to \infty$, $\alpha \to 0$ and $\beta \to 0$ in such a way that $M\alpha \to a$ and $M\beta \to b$ for some $a > 0$, $b > 0$. Then show that

$$M\pi_k \sim \frac{\Gamma(a+b)}{\Gamma(a)\Gamma(b)}(1-x)^{a-1}x^{b-1}$$

where $x = k/M$. [see Section 4.3.4 for the definition of '$\sim$'; it is given just before Lemma 4.5.] Thus for a large population, that is, when M is large, the stationary distribution for the relative frequency of the genes can be approximated by a Beta distribution.

8. Consider Kimura's model for n subunits of which one is in mutant form.
(a) Show that for n=2, 4, the transition matrices are given by

$$\begin{pmatrix} 1 & 0 & 0 \\ 1/6 & 4/6 & 1/6 \\ 0 & 0 & 1 \end{pmatrix}$$

and

$$\begin{pmatrix} 1 & 0 & 0 & 0 & 0 \\ 15/70 & 40/70 & 15/70 & 0 & 0 \\ 1/70 & 16/70 & 36/70 & 16/70 & 1/70 \\ 0 & 0 & 15/70 & 40/70 & 15/70 \\ 0 & 0 & 0 & 0 & 1 \end{pmatrix}.$$

(b) Let d_n^t be the probability that a gene with all parts mutant appears for the first time in the t-th generation. Show that

$$d_2^t = \frac{1}{6}\left(\frac{2}{3}\right)^{t-1}, \quad d_4^t = \frac{1}{3080}\left[195\left(\frac{6}{7}\right)^{t-1} - 330\left(\frac{4}{7}\right)^{t-1} + 135\left(\frac{16}{70}\right)^{t-1}\right].$$

9. The idea of this problem is to extend the method of Feller and unify the techniques of evaluating the eigenvalues. This follows S.Karlin and J.Mcgregor.
Let $f(s) = \sum_0^\infty a_i s^i$ be a p.g.f. Let $N \geq 2$ be a fixed integer.

(a) Define

$$P_{ij} = \frac{\text{coefficient of } s^j t^{N-j} \text{ in } f^i(s) f^{N-i}(t)}{\text{coefficient of } \omega^N \text{ in } f^N(\omega)} \quad \text{for} \quad 0 \le i, j \le N .$$

Show that $P = ((P_{ij}))$ is a transition matrix. Here, of course, we make the assumption that the denominator above is non-zero. What does this mean in terms of f?

(b) The Markov chain corresponding to P is called the induced Markov chain of a direct product branching process. The reason is the following. Imagine two independent branching processes corresponding to two kinds of objects, say A and a, each having p.g.f. f. What is the conditional probability of having j objects of type A in the next generation given that there are i objects of type A in the present generation *and* in each generation the total number of objects is N. Verify that the answer is precisely P_{ij}.

(c) Let $f(s) = e^{s-1}$. Show that you get the Wright-Fisher matrix.

(d) Let $f(s) = (q + ps)^2$, where $0 < p < 1$. Show that you get the Kimura matrix.

(e) Let $f(s) = q^\alpha/(1 - ps)^\alpha$, where $0 < p < 1$ and $\alpha > 0$. Show that

$$P_{ij} = \frac{\binom{i\alpha+j-1}{j}\binom{N\alpha-i\alpha-j-1}{N-j}}{\binom{N\alpha+N-1}{N}} .$$

This generating function arises in the growth of heterogeneous populations as follows. Suppose that in a large geographical area, the progeny follows a branching process with progeny distribution $P(\lambda)$. The parameter λ depends on the sub-area and is assumed to have a distribution with density $g(\lambda) = (q/p)^\alpha \lambda^{\alpha-1} e^{-q\lambda/p}/\Gamma(\alpha)$, $\lambda > 0$. Then the compound distribution of the progeny is f. Verify this.

(f) Show that

$$P_{ij} = \frac{\text{coefficient of } s^j \omega^N \text{ in } f^i(s\omega) f^{N-i}(\omega)}{\text{coefficient of } \omega^N \text{ in } f^N(\omega)} .$$

Let G_i be the p.g.f. of P_{ij}, $0 \le j \le N$, that is, $G_i(s) = \sum_{j=0}^N P_{i,j} s^j$. Show that

$$G_i(s) = \frac{\text{coefficient of } \omega^N \text{ in } f^i(s\omega) f^{N-i}(\omega)}{\text{coefficient of } \omega^N \text{ in } f^N(\omega)} .$$

(g) Show that $G_i'(1) = i\lambda_1$, where

$$\lambda_1 = \frac{\text{coefficient of } \omega^{N-1} \text{ in } f^{N-1}(\omega) f'(\omega)}{\text{coefficient of } \omega^N \text{ in } f^N(\omega)} .$$

Conclude that λ_1 is an eigenvalue of P with $(0, 1, \ldots N)'$ as a right eigenvector.

(h) Let $\lambda_0 = 1$ and for $1 \leq r \leq N$

$$\lambda_r = \frac{\text{coefficient of } \omega^{N-r} \text{ in } f^{N-r}(\omega)[f'(\omega)]^r}{\text{coefficient of } \omega^N \text{ in } f^N(\omega)}.$$

By induction, show that for $r = 0, 1, \ldots, N$,

$$\sum_{j=0}^{N} P_{i,j} j^r = \lambda_r i^r + u_{r-1}(i),$$

where u_{r-1} is a polynomial in i of degree smaller than r.

(i) Let v_0 be the vector with all entries 1 and for $1 \leq k \leq N$, let v_k be the column vector with entries $0^k, 1^k, \ldots, N^k$. Show that these form a basis of R^{N+1} and with respect to this basis, P is upper triangular with diagonal entries λ_r, $0 \leq r \leq N$. [Karlin and Mcgregor also show that if $a_0 \cdot a_1 \cdot a_2 > 0$, then $1 = \lambda_0 = \lambda_1 > \lambda_2 > \cdots > \lambda_r > 0$.]

(j) Going to (c) above, deduce that the Wright-Fisher eigenvalues are

$$\lambda_r = N!/[(N-r)!N^r].$$

(k) Going to (d) above, deduce that the Kimura eigenvalues are

$$\lambda_r = \alpha^r \binom{N\alpha - r}{N - r} \Big/ \binom{N\alpha}{N}.$$

(l) Going to (e) above, deduce that the eigenvalues are

$$\lambda_r = \alpha^r \binom{\alpha N + N - 1}{N - r} \Big/ \binom{\alpha N + N - 1}{N}.$$

10. The idea is to discuss, following W. J. Ewens, the haploid model of Moran taking selection into account.
Assume that A and a produce offsprings with relative proportions μ_1 and μ_2 respectively. Thus a randomly chosen individual dies and is replaced by an individual who is A or a with probabilities $\mu_1 i/[\mu_1 i + \mu_2(M-i)]$ and $\mu_2(M-i)/[\mu_1 i + \mu_2(M-i)]$ respectively, where i is the number of A individuals before the death event.

(a) Show that the transition probabilities of the corresponding Markov chain with state space $\{0, 1, \ldots, M\}$ are

$$p_{i,i-1} = \mu_2 i(M-i)/M[\mu_1 i + \mu_2(M-i)] = \pi_i, \text{ say,}$$

$$p_{i,i+1} = \mu_1 i(M-i)/M[\mu_1 i + \mu_2(M-i)] = \eta_i, \text{ say,}$$

$$\text{and} \quad p_{i,i} = 1 - \pi_i - \eta_i.$$

(b) Show that the probability of absorption in 0 given $X_0 = k$ is

$$(\alpha^{M-k} - 1)/(\alpha^M - 1)$$

where $\alpha = \mu_1/\mu_2$.
From now on we fix an initial state k. We wish to find the mean time for absorption given $X_0 = k$.

(c) Define an "amended chain" by putting $\tilde{p}_{0,k} = \tilde{p}_{M,k} = 1$. For $i \neq 0, M$, $\tilde{p}_{i,j}$ is the same as $p_{i,j}$. Thus this amended chain is like the original one except that whenever the original reaches an absorbing state then we make it start afresh again from k. Show that this amended chain is irreducible and aperiodic. Let $\lambda = (\lambda_0, \lambda_1, \ldots, \lambda_M)$ be the unique stationary initial distribution.

(d) Show that $\lambda_1 = \lambda_0/\pi_1$ and $\lambda_2 = \lambda_0(1+\alpha)/\pi_2$.

(e) Show that for $1 \leq i \leq k-3$, λ_i satisfies

$$-\eta_i\lambda_i + (\eta_{i+1} + \pi_{i+1})\lambda_{i+1} - \pi_{i+2}\lambda_{i+2} = 0.$$

Put $\xi_i = \pi_i\lambda_i$ so that $\eta_i\lambda_i = \alpha\xi_i$. Show that

$$-\alpha\xi_i + (\alpha+1)\xi_{i+1} - \xi_{i+2} = 0,$$

$$\xi_1 = \lambda_0 \quad \text{and} \quad \xi_2 = \lambda_0(1+\alpha).$$

Deduce that, for $1 \leq i \leq k-1$, $\lambda_i = \dfrac{\lambda_0(\alpha^i - 1)}{\pi_i(\alpha - 1)}$.

(f) Show that $\lambda_{M-1} = \lambda_M/\eta_{M-1}$ and $\lambda_{M-2} = \lambda_M(1 + \frac{1}{\alpha})/\eta_{M-2}$.

(g) Put $\xi_i = \eta_i\lambda_i$ and show that, for $k+1 \leq i \leq M-1$,

$$-\xi_i + (1 + \frac{1}{\alpha})\xi_{i+1} - \frac{1}{\alpha}\xi_{i+2} = 0.$$

Deduce that, for $k+1 \leq i \leq M-1$, $\lambda_i = \dfrac{\lambda_M(\alpha^M - \alpha^i)}{\eta_i(\alpha^M - \alpha^{M-1})}$.

(h) There is one more equation from $\lambda\tilde{P} = \lambda$ to be used now. Use it and show that

$$\lambda_k = \frac{\lambda_0 + \lambda_M + \lambda_0\left(\dfrac{\alpha^k - \alpha}{\alpha - 1}\right) + \lambda_M\left(\dfrac{\alpha^{M-1} - \alpha^k}{\alpha^M - \alpha^{M-1}}\right)}{\pi_k + \eta_k}.$$

(i) Show that

$$\frac{\lambda_0}{\lambda_M} = \frac{\text{Probability of absorption at 0 in the original chain}}{\text{Probability of absorption at } M \text{ in the original chain}}$$

(j) Show that the mean time until absorption, say m_k, for the original chain is $\sum_{i=1}^{M-1} \lambda_i/(\lambda_0 + \lambda_M)$.

(k) Show that

$$m_k = \frac{\alpha^{M-k}-1}{\alpha^M-1}\sum_{i=1}^{k}\frac{\alpha^i-1}{\pi_i(\alpha-1)} + \frac{\alpha^M-\alpha^{M-k}}{\alpha^M-1}\sum_{i=k+1}^{M-1}\frac{\alpha^M-\alpha^i}{\eta_i(\alpha^M-\alpha^{M-1})}.$$

(l) If μ_1 approaches μ_2, so that α approaches 1 – that is, there is no selective advantage – verify that the above formula does indeed give the result of Watterson.

11. Consider as usual an autosomal gene with two alleles A and a. The genotypes thus are AA, Aa and aa. Define a process as follows. X_0 is arbitrary. X_n is genotype of an offspring whose father is X_{n-1} and mother is AA, Aa or aa with probabilities p^2, $2pq$ and q^2 respectively. Show that we have here an aperiodic Markov chain with transition matrix P having rows $(p,q,0)$; $(p/2,1/2,q/2)$ and $(0,p,q)$ respectively. Show that the stationary distribution is given by $\pi = (p^2, 2pq, q^2)$. Show that this chain is *reversible*, that is, $\pi_i P_{ij} = \pi_j P_{ji}$ for all i and j. Interpret reversibility and compare with Exercises 15–18 of Chapter 2. Show that P^n is given by the following matrix.

$$e\Pi' + \frac{1}{2^{n-1}}\begin{pmatrix} pq & q(q-p) & -q^2 \\ p(q-p)/2 & (1-4pq)/2 & q(p-q)/2 \\ -p^2 & p(p-q) & pq \end{pmatrix}.$$

12. The idea of this problem is to find a formula for the expected absorption time in the Wright-Fisher model. This follows R. G. Khazanie and H. E. Mckean. This will also rework some of the other quantities that we already know.
Notation: $M = 2N$; $p_i = i/M$; $p_{i,j} = \binom{M}{j} p_i^j (1-p_i)^{(M-j)}$; P is the transition matrix $(p_{i,j})$; $(x)_0 = 1$; and for $n \geq 1$, $(x)_n = x\,(x-1)\cdots(x-n+1)$; $\Delta f(x) = f(x+1) - f(x)$; $c_{r,n} = \Delta^n x^r/n!\,\big|_{x=0}$ also written as $\Delta^n 0^r/n!$.

(a) Show that $\Delta\,(x)_n = n\,(x)_{n-1}$.

(b) Show that $x^r = \sum_{n=0}^{r} c_{r,n}\,(x)_n$.

(c) Show that $\Delta^n\,0^r = \sum_{j=0}^{n}(-1)^j\binom{n}{j}(n-j)^r$.

(d) Deduce that $x^r = \sum_{n=0}^{r} a_{r,n}(x)_n$, where, for $0 \leq n \leq r$,

$$a_{r,n} = \sum_{j=0}^{n}(-1)^j\frac{(n-j)^{r-1}}{(n-j-1)!j!}.$$

Note that $a_{r,r} = 1$.
These numbers $a_{r,n}$ are called Stirling numbers of the second kind.

(e) Show that $\sum_{j=0}^{M} p_{k,j}\, j^r = \sum_{s=1}^{r} a_{r,s}(M)_s(\frac{k}{M})$.

(f) Show that $\sum_{j=0}^{M} p_{i,j}^{(n+1)}\, j^r = \sum_{s=1}^{r} a_{r,s}\frac{(M)_s}{M^s} \sum_{k=0}^{M} p_{i,k}^{(n)} k^s$.

(g) Put

$$\mu_{n,r,i} = E(X_n^r \,|\, X_0 = i),$$

$$e_{n,i} = \text{ the column vector } (\mu_{n,r,i})_{1\le r\le M}.$$

Let C be the lower triangular matrix

$$C = \begin{pmatrix} a_{1,1}\lambda_1 & 0 & 0 & 0 & \cdots & 0 \\ a_{2,1}\lambda_1 & a_{2,2}\lambda_2 & 0 & 0 & \cdots & 0 \\ a_{3,1}\lambda_1 & a_{3,2}\lambda_2 & a_{3,3}\lambda_3 & 0 & \cdots & 0 \\ \vdots & \vdots & \vdots & \vdots & \ddots & \vdots \\ a_{M,1}\lambda_1 & a_{M,2}\lambda_2 & 0 & 0 & \cdots & a_{M,M}\lambda_M \end{pmatrix},$$

where $\lambda_s = (M)_s/M^s$, $a_{r,s} = \sum_{i=0}^{s-1} \frac{(-1)^i (s-i)^{r-1}}{(s-i-1)!\,i!}$ for $1 \le s \le r$.

Show that $e_{0,i}$ is the column vector $(i, i^2, \ldots, i^M)'$.

Show that $e_{n+1,i} = Ce_{n,i}$.

Deduce that $e_{n-1,i} = C^{n-1}e_{0,i}$.

(h) Show that $\lambda_1, \lambda_2, \ldots, \lambda_M$ are distinct and they are precisely the eigenvalues of C.

Show that corresponding to λ_s, there is a right eigenvector of the form

$$R_s = (0, 0, \ldots, 1, u_{s,s+1}, u_{s,s+2}, \ldots, u_{s,M})'$$

and a left eigenvector of the form

$$L_s = (v_{s,1}, v_{s,2}, \ldots, v_{s,s-1}, 1, 0, \ldots, 0)$$

and describe an algorithm to obtain these vectors.

Show that $R_1 = (1, M, M^2, \ldots, M^{M-1})'$.

(i) Show that

$$e_{n-1,i} = R_1 L_1 e_{0,i} + \sum_{s=2}^{M} \lambda_s^{n-1} R_s L_s e_{0,i}.$$

(j) Show that

$$\mu_{n-1,j+t,i} = 1 \quad \text{if } j = t = 0,$$

$$\mu_{n-1,j+t,i} = iM^{j+t-1} + \sum_{s=2}^{M} \sum_{\beta=1}^{s} \lambda_s^{n-1} v_{s,\beta} u_{s,j+t} i^{\beta} \quad \text{if } j + t > 0.$$

(k) Show that

$$p_{i,j}^{(n)} = \sum_{t=0}^{M-j} (-1)^j \binom{M}{j} \binom{M-j}{t} M^{-t-j} \mu_{n-1,j+t,i} .$$

(l) Show that for $0 < j < M$,

$$p_{i,j}^{(n)} = \binom{M}{j} \sum_{t=0}^{M-j} (-1)^t \binom{M-j}{t} M^{-t-j} \sum_{s=2}^{M} \sum_{\beta=1}^{s} \lambda_s^{n-1} v_{s,\beta} u_{s,j+t} \, i^\beta .$$

Show that

$$p_{i,M}^{(n)} = \frac{i}{M} + M^{-M} \sum_{s=2}^{M} \sum_{\beta=1}^{s} \lambda_s^{n-1} v_{s,\beta} u_{s,M} \, i^\beta .$$

Show that

$$p_{i,0}^{(n)} = 1 - \frac{i}{M} + \sum_{t=1}^{M} (-1)^t \binom{M}{t} M^{-t} \sum_{s=2}^{M} \sum_{\beta=1}^{s} \lambda_s^{n-1} v_{s,\beta} u_{s,t} \, i^\beta .$$

(m) Show that

$$\lim_n p_{i,0}^{(n)} = 1 - \frac{i}{M}, \qquad \lim_n p_{i,M}^{(n)} = \frac{i}{M},$$
$$\lim_n p_{i,j}^{(n)} = 0 \quad \text{for} \quad 1 \le j \le M-1 .$$

(n) Show that $p_{i,j} = p_{M-i,M-j}$.
Hence deduce that $p_{i,j}^{(n)} = p_{M-i,M-j}^{(n)}$.

(o) Let T be the time spent before absorption. Clearly

$$P(T = 0 \,|\, X_0 = 0) = 1, \qquad P(T = 0 \,|\, X_0 = M) = 1.$$

So from now on $1 \le i \le M-1$. Show that

$$P(T = 1 \,|\, X_0 = i) = p_{i,0} + p_{i,M},$$
$$\begin{aligned} P(T = n \,|\, X_0 = i) &= p_{i,M}^{(n)} - p_{i,M}^{(n-1)} + p_{M-i,M}^{(n)} - p_{M-i,M}^{(n-1)} \\ &= M^{-M} \textstyle\sum_{s=2}^{M} \sum_{\beta=1}^{s} \lambda_s^{n-2} c(i,s,\beta), \end{aligned}$$

where

$$c(i,s,\beta) = (\lambda_s - 1)[i^\beta + (M-i)^\beta] u_{s,M} v_{s,\beta} .$$

(p) Show that $G_i(z)$, the p.g.f. of T when $X_0 = i$, is given by

$$\left[(1 - \frac{i}{M})^M + (\frac{i}{M})^M \right] z + M^{-M} \sum_{s=2}^{M} \sum_{\beta=1}^{s} c(i,s,\beta) \frac{z^2}{1 - z\lambda_s}$$

for $|z| < \frac{M}{M-1}$.

(q) Show that $E(T \mid X_0 = i)$ is given by

$$m_i = (1 - \frac{i}{M})^M + (\frac{i}{M})^M + M^{-M} \sum_{s=2}^{M} \sum_{\beta=1}^{s} c(i, s, \beta) \frac{2 - \lambda_s}{(1 - \lambda_s)^2}.$$

3.10 References/Supplementary Readings

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Chapter 4

MODELS IN EPIDEMICS

4.1 Generalities

One of the important areas of real-life applications of stochastic processes is in epidemiology, more specifically, in analyzing the spread of epidemics. Roughly speaking, the situation that we want to look at is as follows. There is a group of individuals, all mixing homogeneously together. Due to some reason, one or more of them contracted an infectious disease. They are the individuals *initially infected.* After a certain period of time, called the *latent period*, the infected become *infectious.* This means that they are now capable of passing on the infection to other individuals in the population to be called *susceptibles.* This leads to new infections. Thus, some of the susceptibles move to the infected group and this continues. Simultaneously, as time passes there is also what is called *removal* of infectious from circulation. Such removals in reality may take place by way of death or by way of detection and quarantine. Of course, removal may not always mean that the concerned individuals are actually taken out of the population. For example, an infectious person may have been cured and has become immune. Thus, as far as the epidemic is concerned, they are as good as removed.

In the next few sections, we shall model such phenomena mathematically. In the light of the different models, we would like to investigate how the epidemic progresses in terms of the changes in the number of infected and susceptibles. A quantity of vital interest is the total size of the epidemic, to be explained later. The rate at which the infection spreads is called the *rate of infection*, whereas the rate at which the infectious individuals get removed is called the *rate of removal.* If the infection rate is too small compared to the removal rate, then one intuitively feels that the epidemic should not build up. An important class of theorems in epidemiology known as the *Threshold Theorems* are aimed at justifying this mathematically. One distinctive feature of this chapter is that unlike the previous chapters where the models were discrete in time, here we have an evolution taking place in continuous time. As a result, in Sections 4.2

and 4.3, we will use continuous time Markov chains.

Needless to say that the description given above conforms to the real world meaning of epidemics. However the same picture obtains if we wish to study the spread of a disease across the cells of a single individual.

4.2 Simple Epidemic

For the sake of simplicity, we first consider the case when there is no latent period and also there are no removals. The first assumption means that an individual becomes infectious as soon as he receives the infection. In the absence of removals, an infectious individual remains in circulation forever. In this case, it is intuitively clear that infection would continue to spread until all are infected.

4.2.1 Deterministic Model

We start with a deterministic model first. We consider a population of $n+1$ individuals in which initially, that is, at time $t=0$, there are n susceptibles and 1 infectious. We denote by $x(t)$ and $y(t)$, the number of susceptibles and the number of infected individuals respectively at time t. Of course, it is clear that for every t, $x(t)+y(t)=n+1$. Also $x(0)=n$ and $y(0)=1$. In view of the fact that $y(t)=n+1-x(t)$, it suffices to describe $x(t)$. The central step in modelling the process $x(t)$ involves deciding on the mechanism governing the evolution of $x(t)$. More precisely, suppose that at some time instant, say t, we have $x(t)=a$. This means that at time t there are a susceptibles and $n+1-a$ infected in the population. The question is how $x(t)$ should change in a small time interval, say Δt. This, of course, means how many new infections take place during the time period $(t, t+\Delta t)$. This is where we bring in our modelling assumptions. First of all, the number of new infections should be proportional to the duration of the interval, namely, Δt. Indeed, one does feel that the number of new infections in a given time interval should be large or small depending on whether the interval is large or small. Secondly, the number of new infections should be proportional to the possible number of contacts between the infected and the susceptibles. Since at time t we have a susceptibles and $n+1-a$ infected the possible number of contacts (that is, pairings) between these two groups is $a(n+1-a)$. Our modelling assumption, therefore, reduces to speculating that $x(t+\Delta t)-x(t) \sim -\beta x(t)[n+1-x(t)]\Delta t$. Here β is a positive constant, frequently referred to as the infection rate. Dividing by Δt and taking the limit as $\Delta t \to 0$ the above amounts to

$$x'(t) = -\beta x(t)[n+1-x(t)]\,. \tag{1}$$

This is our precise mathematical assumption regarding the evolution of $x(t)$. An initiated reader would, of course, raise objections at this point. The fact is that $x(t)$ denotes the number of susceptibles at time t and is hence integer

valued. Let alone being differentiable, $x(t)$ cannot even be continuous unless it is a constant function of t. Here is one way to make sense out of Equation (1). Assume that the population size $(n+1)$ is large and consider the proportion $\bar{x}(t) = x(t)/(n+1)$ of susceptibles rather than $x(t)$ itself. In that case $\bar{x}(t)$ can be regarded as taking values in the continuum $[0,1]$ (at least approximately). Our modelling assumptions can now be summarized in a genuine differential equation for $\bar{x}(t)$, obtained as follows. As seen earlier, for small Δt,

$$\frac{x(t+\Delta t) - x(t)}{\Delta t} \sim -\beta x(t)(n+1-x(t)),$$

that is,

$$\frac{\bar{x}(t+\Delta t) - \bar{x}(t)}{\Delta t} \sim -\beta(n+1)\bar{x}(t)(1-\bar{x}(t),$$

which on taking limit as $\Delta t \downarrow 0$ yields

$$\bar{x}'(t) = -\beta(n+1)\bar{x}(t)[1-\bar{x}(t)]\,.$$

Of course, all this is just a matter of mathematical precision and should be viewed as a way of rationalizing Equation (1). In any case you should remember that Equation (1) itself reflects only an approximation for the actual state of affairs.

Returning now to Equation (1), it can be written as

$$\frac{x'(t)}{x(t)} + \frac{x'(t)}{n+1-x(t)} = -\beta(n+1)\,.$$

By integrating and using the initial condition $x(0) = n$, one obtains

$$x(t) = \frac{n(n+1)}{n + e^{\beta(n+1)t}}\,.$$

It follows that

$$y(t) = \frac{(n+1)e^{\beta(n+1)t}}{n + e^{\beta(n+1)t}}\,.$$

The rate at which the infections accrue is given by

$$\begin{aligned}\omega(t) &= y'(t) = -x'(t)\\ &= \beta x(t)[n+1-x(t)]\\ &= \beta n(n+1)^2 \frac{e^{\beta(n+1)t}}{[n+e^{\beta(n+1)t}]^2}\,.\end{aligned}$$

This is of considerable interest in epidemiology and the graph of $\omega(t)$ is called the *epidemic curve*. The above epidemic curve starts at $\omega(0) = \beta n$, increases until it reaches a peak at time $t_0 = \log n/[\beta(n+1)]$ and then gradually dies down. The time point t_0 is usually of some interest. In our model, the number of susceptibles and the number of infected become almost equal at time t_0.

The fact that $\lim_{t\to\infty} x(t) = 0$ is only natural because in this model the entire population is clearly going to be infected eventually. This completes our discussion of the deterministic model. After all, the main purpose of this model is to motivate a more realistic model, namely, a stochastic one.

4.2.2 Simple Stochastic Epidemic

In the stochastic model we do not say that in the small time-interval t to $t + \Delta t$, a certain number of new infections is sure to take place. Instead, we introduce a chance mechanism for the number of new infections. As before, let us imagine a population of size $n + 1$ with n susceptibles and 1 infected initially. Let X_t and Y_t denote the number of susceptibles and the number of infected, respectively, at time t. Here X_t and Y_t are going to be random variables. Of course $X_t + Y_t = n + 1$, for all t ; $X_0 = n$ and $Y_0 = 1$. Now we come to the main assumption. Given that at time t, $X_t = a$ and $Y_t = n+1-a$, we assume that during the time interval t to $t+\Delta t$, the probability of exactly one new infection is $\beta a(n + 1 - a)\Delta t + o(\Delta t)$ and that of no new infection is $1 - \beta a(n+1-a)\Delta t + o(\Delta t)$. This of course implies that the probability of two or more new infections during the period t to $t + \Delta t$ is $o(\Delta t)$. Thus $(X_t)_{t\geq 0}$ is a continuous time pure death chain starting at $X_0 = n$ (see the concluding paragraph of Section 0.9.1). The death rates are given by

$$\mu_i = \beta i(n + 1 - i), \qquad i = 0, 1, \ldots, n\,.$$

Denoting, $p_r(t) = P[X_t = r]$, the usual Kolmogorov equations are

$$p_r'(t) = -\beta r(n + 1 - r)p_r(t) + \beta(r + 1)(n - r)p_{r+1}(t) \quad \text{for } 0 \leq r < n,$$
$$\text{and} \quad p_n'(t) = -\beta n p_n(t)\,. \tag{2}$$

Of course $p_n(0) = 1$ where as $p_r(0) = 0$ for $0 \leq r < n$.

As we shall see later, it is possible to solve the above equations successively for $p_n, p_{n-1}, \ldots, p_0$. But the formulae are too complicated to give an insight into the phenomenon. Of course, since $(X_t)_{t\geq 0}$ is a pure death chain with death rates μ_i given in (1), the general theory tells us that the first new infection takes place after a random time distributed as $\mathcal{E}xp(\mu_n)$, the next infection occurs after a further random time with distribution $\mathcal{E}xp(\mu_{n-1})$, and so on. Finally, the process comes to a halt when all are infected. This happens in a finite time with probability one. Denoting by T the total duration, it is clear that T is the sum of n independent exponential random variables with parameters $\mu_n, \ldots, \mu_1$. One can find the exact distribution of T. However, we shall be

content with noting that

$$\begin{aligned} E(T) &= \frac{1}{\beta}\sum_{i=1}^{n}\frac{1}{i(n+1-i)} \\ &= \frac{1}{\beta(n+1)}\sum_{i=1}^{n}\left[\frac{1}{i}+\frac{1}{n+1-i}\right] \\ &= \frac{2}{\beta(n+1)}\sum_{i=1}^{n}\frac{1}{i}\,. \end{aligned}$$

But,

$$\sum_{i=1}^{n}\frac{1}{i} = \sum_{i=1}^{n}\frac{1}{i}\int_{i}^{i+1}dx \geq \sum_{i=1}^{n}\int_{i}^{i+1}\frac{1}{x}\,dx = \log(n+1)$$

and,

$$\sum_{i=1}^{n}\frac{1}{i} = 1+\sum_{i=2}^{n}\frac{1}{i}\int_{i-1}^{i}dx \leq 1+\sum_{i=2}^{n}\int_{i-1}^{i}\frac{1}{x}\,dx = 1+\log n\,.$$

Thus,

$$\frac{2}{\beta}\frac{\log(n+1)}{n+1} \leq E(T) \leq \frac{2}{\beta}\frac{1+\log n}{n+1}\,.$$

That is, $E(T) = O(\log n/n)$. Incidentally, this also shows that $E(T) \downarrow 0$ as $n \to \infty$. This seems to contradict one's first intuition that large populations should take longer to reach complete infection. However, one should not forget that larger population implies increased death rate also.

Returning to the Kolmogorov Equations (2), we now indicate how one can go about solving them and also what the nature of the expressions for $p_r(t)$ is. Firstly, it is immediate that

$$p_n(t) = e^{-\beta n t}\,.$$

This can also be directly seen from the fact that $p_n(t)$ is the probability that upto and including time t, no new infection has taken place and that the time till the first new infection is exponentially distributed with parameter βn. Next, the Equations (2) can be rewritten as

$$p_r'(t) + \mu_r p_r(t) = \mu_{r+1}p_{r+1}(t) \quad \text{for } 0 \leq r < n\,.$$

Multiplying both sides by $e^{\mu_r t}$ one obtains

$$\frac{d}{dt}\left[p_r(t)e^{\mu_r t}\right] = \mu_{r+1}e^{\mu_r t}p_{r+1}(t)\,.$$

Solving this one obtains the recurrence relation

$$p_r(t) = e^{-\mu_r t}\int_0^t \mu_{r+1}e^{\mu_r s}p_{r+1}(s)\,ds\,. \tag{3}$$

Thus explicit expressions for the $p_r(t)$, $0 \leq r < n$ can be obtained. Since the actual formulae are quite complicated and do not seem to reveal much, we shall be content with giving the following partial result.

Proposition 4.1: *For each* $r > \frac{n}{2}$, $p_r(t)$ *is a linear combination of the functions* $e^{-\mu_i t}$, $r \leq i \leq n$.

Proof: Clearly the assertion is true for $r = n$. Let $n > r > \frac{n}{2}$ and $p_{r+1}(t) = \sum_{i=r+1}^{n} C_{r+1,i} e^{-\mu_i t}$. We shall show that $p_r(t) = \sum_{i=r}^{n} C_{r,i} e^{-\mu_i t}$. This will complete the proof. Using (3) and our hypothesis concerning $p_{r+1}(t)$,

$$p_r(t) = e^{-\mu_r t} \int_0^t \sum_{i=r+1}^{n} \mu_{r+1} C_{r+1,i} \quad e^{(\mu_r - \mu_i)s} \, ds \, .$$

Observe that $\mu_i = \mu_r$ if and only if $i = r$ or $i = n + 1 - r$. It follows that if $r > n/2$, then $\mu_i \neq \mu_r$ for every $i > r$. As a consequence,

$$\begin{aligned} p_r(t) &= \sum_{i=r+1}^{n} \mu_{r+1} \frac{C_{r+1,i}}{\mu_r - \mu_i} [e^{-\mu_i t} - e^{-\mu_r t}] \\ &= \sum_{i=r}^{n} C_{r,i} e^{-\mu_i t} \, , \end{aligned}$$

where

$$C_{r,r} = - \sum_{i=r+1}^{n} \frac{\mu_{r+1} C_{r+1,i}}{\mu_r - \mu_i}$$

and for $i > r$,

$$C_{r,i} = \frac{\mu_{r+1} C_{r+1,i}}{\mu_r - \mu_i} \, .$$

∎

Remark: The condition that $r > n/2$ was crucially used in the above proof. For $r \leq n/2$, $p_r(t)$ is not a linear combination of the above type. Extra complications crop up due to the fact that for any $r \leq n/2$ there is indeed an $i > r$, namely $i = n + 1 - r$, such that $\mu_i = \mu_r$. This gives rise to terms involving $te^{-\mu_r t}$ also, thus making explicit expression for $p_r(t)$ more complicated.

4.3 General Epidemic

We now consider a slight generalization of the earlier model in that, we allow removals. Of course, there is still no latent period. Thus a person infected becomes infectious instantly and remains so until he is removed from the population. This phenomenon is referred to as *General Epidemic*. As in the case of simple epidemic, here also we first consider a deterministic model and then a stochastic one.

4.3.1 Deterministic Model

This deterministic model was proposed in 1927 by W. O. Kermack and A.G. McKendrick. Here is the model. We have a population of n individuals and initially a certain number of them are infected; the rest are naturally susceptibles. As time passes, new infections take place and also some infected individuals are removed. Thus at any point of time, the population consists of three groups of individuals — susceptibles, infected and removed. Let $x(t), y(t)$ and $z(t)$ denote the number of individuals in these three groups respectively at time t. Clearly $x(t) + y(t) + z(t) = n$, for all t. We assume that $x(0) = x_0 > 0, y(0) = y_0 > 0$ and $z(0) = 0$. We assume that the number of new infections in time interval $(t, t + \Delta t)$ depends only on the number of susceptibles and the number of infected at time t, but not on the number of individuals removed till time t. This stands to reason because new infections arise out of contacts between the susceptibles and the infected.

As in the simple epidemic model, we postulate that the actual number of new infections during $(t, t + \Delta t)$ is $\beta x(t)y(t)\Delta t$. The rationale behind this postulate has already been explained in Section 4.2.1. Regarding removals, we assume that the number of individuals removed during $(t, t+\Delta t)$ depends only on the number of infected at time t and neither on the number of susceptibles at time t nor on the number of individuals removed till time t. This last assumption may be a little unrealistic in some situations. For example, one can very well have a situation where the health authorities have limited resources and try to put a check on the new removals. However, for the sake of simplicity, we rule out such a possibility. Following the same idea as in the case of new infections, we postulate that the number of individuals removed during $(t, t + \Delta t)$ is $\gamma y(t)\Delta t$ where γ is again a positive constant like β. The constant β is still called the *infection rate*, while γ is called the *removal rate*. The quantity $\rho = \gamma/\beta$ will play an important role in our analysis and is usually referred to as the *relative removal rate*.

Our postulates above lead to the following differential equations :

$$\begin{aligned} x'(t) &= -\beta x(t)y(t), \\ y'(t) &= \beta x(t)y(t) - \gamma y(t), \\ z'(t) &= \gamma y(t), \end{aligned} \tag{4}$$

with the initial conditions $x(0) = x_0, y(0) = y_0, z(0) = 0$.

The Equations (4) are known as the *Kermack-McKendrick Equations*, or, simply as *KK Equations*. The Equations (4) reveal that the functions $x(t), y(t)$ and $z(t)$ have derivatives upto any order. For example, the differentiability of $x(t)$ and $y(t)$ implies the differentiability of x' in view of the first equation in (4).

The first equation of (4) implies that $x'(t) \leq 0$ for all t, so that $x(t)$ is a non-increasing function. Similarly, from the third equation, it follows that $z(t)$ is a non-decreasing function. We shall now solve for x in terms of z. Using the

third equation of (4) in the first, we get

$$x'(t) = -\frac{1}{\rho}\, z'(t)x(t)\,.$$

Let $t_0 = \sup\{t : x(t) > 0\}$. From the continuity of $x(t)$ and the fact that $x(0) = x_0 > 0$, we conclude that $t_0 > 0$. It could however be infinite. We shall now proceed to argue that t_0 is indeed infinite. First note that, by monotonicity of x, we have $x(t) > 0$ for all $t < t_0$. Therefore on the interval $(0, t_0)$

$$\frac{x'(t)}{x(t)} = -\frac{1}{\rho} z'(t), \quad \text{that is,} \quad \frac{d}{dt} \log x(t) = -\frac{1}{\rho} z'(t)\,.$$

This yields the simple solution

$$x(t) = x_0 e^{-z(t)/\rho} \quad \text{for} \quad t \in (0, t_0)\,.$$

If t_0 were finite then the continuity of x and z would imply that

$$x(t_0) = x_0 e^{-z(t_0)/\rho} > x_0 e^{-n/\rho} > 0\,.$$

But by definition of t_0, we must have $x(t_0) = 0$, if t_0 is finite. This contradiction shows that t_0 is indeed infinite. Thus we have

$$x(t) = x_0 e^{-z(t)/\rho} \quad \text{for all} \quad t \geq 0\,. \tag{5}$$

First observe that, x and z being monotone, both the limits $x_\infty = \lim_{t\to\infty} x(t)$ and $z_\infty = \lim_{t\to\infty} z(t)$ exist. Moreover, from (5), we get that $x_\infty = x_0 e^{-z_\infty/\rho}$. Clearly, $z_\infty \leq n$, so that, $x_\infty \geq x_0 e^{-n/\rho} > 0$. Since $x(t) + y(t) + z(t) = n$ for each t, it follows that $y_\infty = \lim_{t\to\infty} y(t)$ also exists.

We shall now show that $y_\infty = 0$. These have the following epidemiological interpretation. After a sufficiently long time has elapsed and a stable state is reached no infected individual remains in circulation and the population still retains a positive number of uninfected people. This, of course, is a consequence of the dynamics embodied in Equations (4). Reality may not always follow Equations (4). Turning to the second Equation in (4) let us rewrite it as

$$y'(t) = \beta y(t)\,[\,x(t) - \rho\,]\,.$$

If $x(t) \geq \rho$ for all t, then $y(t)$ would be non-decreasing throughout. In particular $y(t) \geq y_0 > 0$ for all t, so that, $z'(t) \geq \gamma y_0$ for all t. But this would mean that $z(t) \geq \gamma y_0 t$ for all t, contradicting the fact that $z(t)$ is bounded by n. Thus there exists a finite time point $t_1 \geq 0$, such that on $[t_1, \infty)$, $x(t) \leq \rho$. As a consequence, y is non-increasing on $[t_1, \infty)$ and, in particular, $y(t) \geq y_\infty$. From the third Equation in (4), it follows that if $t > t_1$, then $z(t) \geq z(t_1) + \gamma y_\infty (t - t_1)$. Now $z(t) \leq n$ for all t would force y_∞ to be zero.

Recalling the definition of t_1 in the above paragraph, it is clear that $t_1 = 0$ or $t_1 > 0$ according as $x_0 \leq \rho$ or $x_0 > \rho$. We now bring to the fore the fact that

these two cases are indeed different in terms of their epidemiological manifestations. Let us first discuss the case $x_0 \leq \rho$. In this case, as observed above, $y(t)$ is a non-increasing function throughout. This means that the number of infected in circulation keeps on decreasing as time passes. In epidemiological terms, one says that the epidemic never really builds up. This should not however be construed as saying that there are no new infections. This only means that the removal rate is sufficiently high compared to the infection rate, so as to keep the number of infected individuals in circulation going down.

The more interesting case is when $x_0 > \rho$. In this case, as observed above, $y(t)$ increases upto a positive time instant t_1 and then decreases. That is, initially the epidemic does build up and reaches a peak at time instant t_1, after which it gradually subsides. In this case, it is interesting to get an idea of the size of the epidemic. A good measure of the size could be $(x_0 - x_\infty)$, which is precisely the total number of individuals who got infected during the course of the epidemic. Of course, we know that

$$x_\infty = x_0\, e^{-z_\infty/\rho}\,.$$

Using the fact that $y_\infty = 0$, so that $z_\infty = n - x_\infty$, we obtain

$$x_\infty = x_0\, e^{-(n-x_\infty)/\rho},$$

or equivalently,

$$n - x_\infty = n - x_0\, e^{-(n-x_\infty)/\rho}\,.$$

Denoting $n - x_\infty$ by u and setting $g(u) = n - x_0 e^{-u/\rho}$, we get

$$u = g(u)\,. \tag{6}$$

Given n, x_0 and ρ, the above is an equation in u, whose solution would give us $n - x_\infty$ or equivalently x_∞. Let us first point out that the equation (6) has a unique positive solution. Indeed, setting $h(u) = g(u) - u$, we observe that

$$h'(u) = g'(u) - 1 = \frac{x_0}{\rho} e^{-u/\rho} - 1\,.$$

Let $u_0 = \rho \log \dfrac{x_0}{\rho}$. Since $x_0 > \rho$, we have $u_0 > 0$. Further, it is easy to see that $h'(u) > 0$ on $[0, u_0)$ and $h'(u) < 0$ on (u_0, ∞). Consequently, h is strictly increasing on $[0, u_0)$ and is strictly decreasing on (u_0, ∞). Noting that $h(0) = n - x_0 = y_0 > 0$ and $\lim\limits_{u\to\infty} h(u) = -\infty$, it can be easily concluded that $h(u) = 0$ has a unique positive solution or equivalently that (6) has a unique positive solution. However, computing the exact value of the solution is difficult, perhaps impossible. There is no standard method of solving equations of the form $u = g(u)$, where g is an exponential function. So the next best thing is to replace g by an approximating polynomial and solve the resulting equation to get an approximate solution. The classical finite Taylor expansion of g would be an ideal choice for such an approximation. Following Kermack

and McKendrick, we use the second order Taylor polynomial. More precisely, we replace $g(u) = n - x_0 e^{-(u/\rho)}$ by

$$n - x_0 \left[1 - \frac{u}{\rho} + \frac{u^2}{2\rho^2}\right] = (n - x_0) + x_0 \frac{u}{\rho} - x_0 \frac{u^2}{2\rho^2} ,$$

so that Equation (6) takes the form

$$u = (n - x_0) + x_0 \frac{u}{\rho} - \frac{x_0}{2\rho^2} u^2 .$$

or equivalently,

$$\frac{x_0}{2\rho^2} u^2 + \left(1 - \frac{x_0}{\rho}\right) u - y_0 = 0 . \tag{7}$$

In view of $x_0 > \rho$, this quadratic equation is easily seen to have only one positive solution.
However, if we also assume that y_0 is small enough and can be neglected from (7), we get a simple formula for this unique positive solution, namely,

$$u^* = \frac{2\rho}{x_0}(x_0 - \rho) .$$

We can utilize this simple form of u^* to get a quantitative idea of the spread of the epidemic. Noting that u^* was obtained as an approximation for $n - x_\infty$ and that $n = x_0 + y_0$ we have

$$\begin{aligned} x_\infty &\sim n - \frac{2\rho}{x_0}(x_0 - \rho) \\ &= x_0 - 2(x_0 - \rho)\frac{\rho}{x_0} + y_0 \\ &\geq x_0 - 2(x_0 - \rho) \qquad [\text{ since } x_0 > \rho, \quad y_0 > 0] \\ &= \rho - (x_0 - \rho), \end{aligned} \tag{8}$$

that is, approximately, $\rho - x_\infty \leq x_0 - \rho$.

Thus, we are lead to the following conclusion. If the initial number of susceptibles exceeds ρ, then the epidemic certainly builds up. However, after the epidemic has died out, the final number of susceptibles can go only as far below ρ as the initial number was above ρ. Noting that $x_0 - x_\infty$ gives the total number of new infections during the course of the epidemic, the above observation really says that this number is approximately no more than $2(x_0 - \rho)$. We summarize our observations in the following theorem:

Theorem 4.2:
(a) *We always have,*

$$\lim_{t\to\infty} y(t) = 0 \text{ and } \lim_{t\to\infty} x(t) \geq x_0\, e^{-(n/\rho)} > 0 .$$

(b) *If $x_0 \leq \rho$, then $y(t)$ continuously decreases in t. Thus, as long as $x_0 \leq \rho$,*

the epidemic does not build up.
(c) *If $x_0 > \rho$, then $y(t)$ initially increases with t, reaches a peak and then gradually decreases. Thus the epidemic does build up. If it is further assumed that y_0 is negligible, then $2(x_0 - \rho)$ is an approximate upper bound for the number of people infected in the course of the epidemic.*

Thus, ρ acts as a threshold value for the initial number of susceptibles in order for the epidemic to build up or not. For this reason, parts (b) and (c) of Theorem 4.2 are referred to as the *Kermack-McKendrick Threshold Theorem.* Going back to (8), it is clear that if y_0 is negligibly small and x_0 is only marginally above the threshold value ρ, so that $\frac{x_0}{\rho} \sim 1$, then one can safely say that $x_\infty \sim \rho - (x_0 - \rho)$. Very often the existing literature states this approximate equality as part of the threshold theorem rather than the approximate inequality we stated in part (c). This is alright as long as the assumptions $y_0 \sim 0, \frac{x_0}{\rho} \sim 1$ are kept in mind.

We now turn to the assumption that y_0 is small. This amounts to saying that initially there is only a trace of the infection in the population. This is not altogether unjustified — and in fact quite natural — for the following reason. In the study of continuous time epidemic model, it is only natural to take the time origin as the time point when the infection first surfaced in the population. Granted that, the assumption of y_0 being small is only logical because most infections start by traces.

Before ending the section, we would like to take up a curious point. One of the key steps in getting an approximate solution of the equation $u = g(u)$ was to replace $g(u)$ by an appropriate Taylor polynomial. In deriving the Threshold Theorem, the second order polynomial was used. The natural question is: why not start with the first order polynomial? Here is an argument. Using the first order polynomial would lead to the equation $u = y_0 + \frac{x_0}{\rho}u$. In case $y_0 \sim 0$ as we have been assuming throughout, this equation almost reduces to $u = \frac{x_0}{\rho}u$. This is of course no good. If $\frac{x_0}{\rho} \not\sim 1$, this equation admits no solution other than zero, whereas if $\frac{x_0}{\rho} \sim 1$, we end up with too many solutions ! Going in the other direction, it may be worthwhile to try and see what one obtains by approximating $g(u)$ by a third order polynomial.

4.3.2 General Stochastic Epidemic

We start with a population consisting initially of a susceptibles and b infected persons. For any time instant t, X_t will denote the number of susceptibles at time t, Y_t the number of infected in circulation at time t and Z_t the number of persons removed till time t. We shall assume now that the spread of the epidemic is governed by a chance mechanism, so that X_t, Y_t and Z_t are random variables. Our object of study is the evolution of the process $(X_t, Y_t, Z_t)_{t \geq 0}$.

The initial conditions, as stated already, are $X_0 = a, Y_0 = b$ and $Z_0 = 0$. It is clear that, for all t, $X_t + Y_t + Z_t = a + b$. Thus, studying the two-dimensional process $(X_t, Y_t)_{t\geq 0}$ suffices. We now describe the chance mechanism. The idea is the same as in the case of simple stochastic epidemic. Given $X_t = x$ and $Y_t = y$, we assume that during a small time interval $(t, t + \Delta t)$, there will be one new infection with probability $\beta xy\Delta t + o(\Delta t)$ and no new infection with probability $1 - \beta xy\Delta t + o(\Delta t)$. This, of course, means that the probability of two or more new infections during $(t, t+\Delta t)$ is $o(\Delta t)$. Regarding removals, our assumption is that during the same time interval, there will be one removal with probability $\gamma y\Delta t + o(\Delta t)$ and no removals with probability $1 - \gamma y\Delta t + o(\Delta t)$. Further, the two events, namely, that of infection and that of a removal during such small time intervals, are assumed to be independent. This description clearly entails that $(X_t, Y_t)_{t\geq 0}$ is a bivariate continuous time Markov chain with state space

$$S = \{(r, s) : r, s \text{ non-negative integers}, r \leq a, r + s \leq a + b\}.$$

It is also clear, by considering the embedded discrete chain, that this is an absorbing chain with the states $\{(r, 0) : 0 \leq r \leq a\}$ being the absorbing states and all others transient. In particular, $Y_\infty = \lim_{t\to\infty} Y_t = 0$ with probability 1, and $X_\infty = \lim_{t\to\infty} X_t$ exists. In line with the deterministic case, the random variable $(X_0 - X_\infty)$ would denote the size of the epidemic. For $(r, s) \in S$, let

$$p_{r,s}(t) = P(X_t = r \text{ and } Y_t = s).$$

It is convenient to have $p_{r,s}$ defined for $(r, s) \notin S$ also, by simply adopting the convention that, for $(r, s) \notin S$, $p_{r,s}(t) = 0$ for all t. We then have

$$\begin{aligned} p_{r,s}(t + \Delta t) &= p_{r+1,s-1}(t)\beta(r + 1)(s - 1)\Delta t[1 - \gamma(s - 1)\Delta t] \\ &\quad + p_{r,s+1}(t)\gamma(s + 1)\Delta t[1 - \beta r(s + 1)\Delta t] \\ &\quad + p_{r,s}(t)[1 - \beta rs\Delta t][1 - \gamma s\Delta t] \\ &\quad + p_{r+1,s}(t)[\beta(r + 1)s\Delta t][\gamma s\Delta t] + o(\Delta t). \end{aligned}$$

This gives us the Kolmogorov equations

$$\frac{dp_{r,s}(t)}{dt} = \beta(r+1)(s-1)p_{r+1,s-1}(t) + \gamma(s+1)p_{r,s+1}(t) - s(\beta r + \gamma)p_{r,s}(t). \quad (9)$$

In particular,

$$\frac{dp_{a,b}(t)}{dt} = -b(\beta a + \gamma)p_{a,b}(t).$$

Using the initial condition $p_{a,b}(0) = 1$, we get

$$p_{a,b}(t) = e^{-b(\beta a+\gamma)t}.$$

For $r = a, s = b - 1$, the Equation (9) becomes

$$\frac{dp_{a,b-1}(t)}{dt} = \gamma b p_{a,b}(t) - (b - 1)(\beta a + \gamma)p_{a,b-1}(t),$$

or equivalently,

$$\frac{dp_{a,b-1}(t)}{dt} + (b-1)(\beta a + \gamma)p_{a,b-1}(t) = \gamma b e^{-b(\beta a+\gamma)t}.$$

Using the initial condition $p_{a,b-1}(0) = 0$, the solution can easily be seen to be

$$p_{a,b-1}(t) = \frac{\gamma b}{\beta a + \gamma} e^{-(b-1)(\beta a+\gamma)t}[1 - e^{-(\beta a+\gamma)t}].$$

Thus the Kolmogorov equations (9) can be successively solved, using the initial condition $p_{r,s}(0) = 0$ for $(r,s) \neq (a,b)$, to get $p_{r,s}(t)$ for all r, s. However, explicit formulae turn out to be extremely complicated and fail to give any insight into the state of affairs. Nevertheless, several people had attempted to get $p_{r,s}(t)$ explicitly by different methods. We briefly illustrate two such attempts here. For further details the reader may consult the book of Bailey.

The first one, due to Siskind, converts the system of differential equations (9) to a single partial differential equation. The idea is to look at the joint p.g.f. of (X_t, Y_t) defined as

$$F(t,u,v) = \sum_{r,s} p_{r,s}(t) u^r v^s .$$

The equations (9) lead to the following partial differential equation for F:

$$\frac{\partial F}{dt} = \beta(v^2 - uv)\frac{\partial^2 F}{\partial u \partial v} + \gamma(1-v)\frac{\partial F}{\partial v},$$

with the initial condition

$$F(0,u,v) = u^a v^b .$$

Siskind solved this explicitly and derived formulae for the functions $p_{r,s}(t)$.

The second one, due to Gani, looks at the Laplace transforms of the functions $p_{r,s}(t)$. Recall that for any bounded continuous function $p(t)$ on $[0,\infty)$, its *Laplace Transform* is the function $q(\lambda)$ on $(0,\infty)$, defined by

$$q(\lambda) = \int_0^\infty e^{-\lambda t} p(t)\, dt$$

Recall further that q determines p uniquely. The idea of Gani in considering the Laplace transforms $q_{r,s}(\lambda)$ of $p_{r,s}(t)$ was to convert the system of equations (9) into a system of recurrence relations for the functions $q_{r,s}$ given by,

$$(\lambda + \beta rs + \gamma s)q_{r,s} = \beta(r+1)(s-1)q_{r+1,s-1} + \gamma(s+1)q_{r,s+1}, \qquad (10)$$

for $(r,s) \neq (a,b)$, with the initial condition

$$(\lambda + \beta ab + \gamma b)q_{a,b} \equiv 1 .$$

These recurrence relations may not be difficult to solve. But in order to get back to the functions $p_{r,s}$, one still faces the problem of inverting the Laplace transforms $q_{r,s}$, which is a difficult one. However, there is one probabilistic question that can be answered without having to go for inversion. For example, what is the probability that the total size of the epidemic, not including the initial number of infected, is k for a given $0 < k < a$? In other words, we are interested in the quantity $P_k = \lim_{t\to\infty} p_{a-k,0}(t)$. Under suitable conditions, it is easy to verify that, if $q(\lambda)$ is the Laplace transform of $p(t)$, then

$$\lim_{t\to\infty} p(t) = \lim_{\lambda\to 0} \lambda q(\lambda)\,.$$

Thus, $P_k = \lim_{\lambda\to 0} \lambda\, q_{a-k,0}(\lambda)$. Using the recurrence relations (10), one can reduce this to

$$P_k = \gamma \lim_{\lambda\to 0} q_{a-k,1}(\lambda)\,.$$

Thus, knowledge of $q_{a-k,1}$ would give us P_k for each k.

4.3.3 A Closer Analysis: Threshold Theorems

We now turn to what are regarded as two fundamental theorems in Markov models for epidemics — the so called *"Threshold Theorems"*. Two quantities that are of interest in understanding the extent of the epidemic are its duration and size. To make the definitions of these quantities precise, let us turn to the Markov process $(X_t, Y_t)_{t\geq 0}$ and observe the following salient features. Recall that the process has a finite state space given by

$$S = \{(r,s) : r, s \text{ non-negative integers }\,;\; r \leq a;\; r+s \leq a+b\}\,.$$

As mentioned earlier, the states $\{(r,0) : r \leq a\}$ are precisely the absorbing states for the process. Also, the set of states $\{(0,s) : s \leq a+b\}$ forms a closed set and once the process hits this set, it then evolves like a death chain in the second coordinate, getting ultimately absorbed at $(0,0)$. Let us denote by F the union of the above two sets of states. Clearly, with probability one, the chain hits the set F in a finite amount of time. Once the set F is entered, no new infections are possible and therefore the epidemic can be thought of as having ended for all practical purposes. It is natural therefore, to regard the time τ needed to enter F as the duration of the epidemic. One would like to draw conclusions about the distribution, and in particular, the expected value of τ. To the best of our knowledge, the existing literature does not contain any non-trivial information on this. On the contrary, much emphasis has been given to what may be called the *size of the epidemic.* From the definition of τ, it is clear that $X(\tau)$ gives the eventual number of susceptibles left in the population. In other words, $X(\tau) = \lim_{t\to\infty} X(t)$. The size of the epidemic is clearly given by the random variable

$$W = X(0) - \lim_{t\to\infty} X(t) = a - X(\tau)\,.$$

One would like to obtain the distribution of the random variable W. Following the general theory of finite state Markov processes, the process (X_t, Y_t) evolves as follows. Given that at time instant t the process is in a non-absorbing state, it waits there for an exponential time and then makes a jump. From a non-absorbing state (r, s), jumps are possible to the states $(r-1, s+1)$ and $(r, s-1)$ with probabilities $p_r = \beta rs/(\beta rs + \gamma s) = r/(r+\rho)$ and $q_r = \rho/(r+\rho)$ respectively. Here, ρ is as defined in the deterministic case, namely, $\rho = \gamma/\beta$. A moment's reflection shows that while τ is the sum of all these waiting times starting from the beginning till the chain hits F, the random variable W has nothing to do with the waiting times. In order to get the distribution of W, it is therefore sufficient to keep track of only the states visited by the chain at its successive jumps. In other words, the distribution of W depends on (X_t, Y_t) only through the embedded Markov chain, as discussed in Section 0.9.1. Let us note that the embedded chain here can be described as follows.

Let $\tau_0 \equiv 0$ and $(\tau_n, n \geq 1)$ be the successive jump times of the process (X_t, Y_t). For $n \geq 0$, let $U_n = X_{\tau_n}$ and $V_n = Y_{\tau_n}$. Then $(U_n, V_n)_{n \geq 0}$ is the embedded chain with state space S. The transition probabilities are as follows. From a state (r, s) with $s \neq 0$, transition can take place to $(r-1, s+1)$ with probability $p_r = r/(r+\rho)$ and to $(r, s-1)$ with probability $q_r = \rho/(r+\rho)$. The states $(r, 0)$ are absorbing. Now note that, $\lim_n U_n = \lim_{t \to \infty} X_t$, so that, $W = a - \lim_n U_n$. This is precisely what we meant, when we said earlier that W depends only on the embedded chain. This was observed by Foster [1955] and was beautifully exploited by him and later, by Rajarshi [1981] to get the exact distribution of the random variable W.

We start with some notation. For a non-negative integer w let A_w denote the set of all sequences $\widetilde{\alpha} = (\alpha_0, \alpha_1, \ldots, \alpha_w)$ of length $w+1$, where the α_i are non-negative integers satisfying

i) $\alpha_w > 1$,

ii) for $j < w$, $\alpha_0 + \alpha_1 + \ldots + \alpha_j < b + j$,

iii) $\alpha_0 + \alpha_1 + \ldots + \alpha_w = b + w$.

Theorem 4.3 (Foster): *For* $0 \leq w \leq a$,

$$P(W = w) = \prod_{l=0}^{w-1} p_{a-l} \sum_{\widetilde{\alpha} \in A_w} \prod_{j=0}^{w} q_{a-j}^{\alpha_j} .$$

Proof: We can view the state space S of the chain $(U_n, V_n)_{n \geq 0}$ as the set of lattice points (r, s) — that is, points with integer coordinates — in the xy plane. The evolution of the chain can then be regarded as the motion of a particle through these lattice points. From a point (r, s) with $s > 0$ the particle moves either one step vertically downwards to $(r, s-1)$ or one step diagonally northwest (that is, up and left) to $(r-1, s+1)$. The probabilities of these two types of transitions are q_r and p_r respectively. Once the particle hits the x-axis it halts. On the other hand once it hits the y-axis, then only the vertically

downward transitions are allowed until it reaches the origin. Viewed this way, the event $(W = w)$ means that the particle starting from (a, b) hits the x-axis precisely at the point $(a - w, 0)$.
Observe that for this to happen, the particle has to make exactly w many northwest transitions, with the first coordinate reducing by 1 after each such transition, until it finally becomes $a - w$. Note that, each of these transitions would result in an increase in the second coordinate by 1, so that the particle has to make $b + w$ many vertically downward transitions in order to reach the x-axis. Starting from (a, b), denote by α_0, the number of downward transitions before the first diagonal transition. In general, for $1 \leq j < w$, let α_j be the number of downward transitions between the j-th and $(j+1)$th diagonal steps. Finally α_w is the number of downward transitions after the w-th diagonal step. Clearly α_j, for $0 \leq j \leq w$, are non-negative integers. Moreover, their sum is $b + w$. The fact that the particle does not hit x-axis before making the w-th diagonal transition implies that, for each $j < w$, $\alpha_0 + \alpha_1 + \cdots + \alpha_j < b + j$. In particular, $\alpha_0 + \alpha_1 + \cdots + \alpha_{w-1} < b + w - 1$, implying that $\alpha_w > 1$. Thus the sequence $\widetilde{\alpha} = (\alpha_0, \alpha_1, \ldots, \alpha_w) \in A_w$. Conversely, any $\widetilde{\alpha} \in A_w$ is a possible choice for the number of vertical motions in between the successive diagonal ones, so that the required event $(W = w)$ occurs. For any $\widetilde{\alpha} \in A_w$, the probability of making the transitions as prescribed by $\widetilde{\alpha}$ is $\prod_{l=0}^{w-1} p_{a-l} \prod_{j=0}^{w} q_{a-j}^{\alpha_j}$. Here the fact that the transition probabilities from any state (r, s) depend only on the first coordinate r is important. The proof is now complete. ∎

For an estimate of the above probability later, we need the following lemma, as in Rajarshi [1981].

Lemma 4.4: *The number of elements in the set A_w is* $\dfrac{b}{b+2w}\binom{b+2w}{b+w}$.

Proof: From Chapter 0.8.1, $\dfrac{b}{b+2w}\binom{b+2w}{b+w}$ is precisely the number of paths of a random walk starting at $(0, 0)$ and ending at $(b + 2w, b)$, which lie strictly above the horizontal axis. The proof will be completed by establishing a one-one correspondence between such paths and elements of A_w. Here is the correspondence. Consider such a path of random walk. First of all, the path would have w many downward steps and $b + w$ many upward steps. Let α_0 be the number of upward steps after the w-th, that is, the final downward transition. Let α_1 be the number of upward steps between the $(w - 1)$th and w-th downward motions. In general, α_j will denote the number of upward steps between the $(w - j)$th and $(w - j + 1)$th downward motions. Finally, α_w is the number of upward steps before the first downward transition. This defines a sequence $\widetilde{\alpha} = (\alpha_0, \alpha_1, \ldots, \alpha_w)$ of non-negative integers of length $w + 1$. We now show that $\widetilde{\alpha} \in A_w$. Since $\alpha_0 + \alpha_1 + \cdots + \alpha_w$ gives the total number of upward movements, it is clear that this sum is $b + w$. Now let $0 \leq j < w$. To show that $\alpha_0 + \alpha_1 + \cdots + \alpha_j < b + j$, it suffices to prove that $\alpha_{j+1} + \cdots + \alpha_w > w - j$. From the definition of the α_j, it should be clear that $\alpha_{j+1} + \cdots + \alpha_w$ is pre-

cisely the total number of upward motions before the $(w-j)$th downward motion. Since the path lies strictly above the horizontal axis we must have $\alpha_{j+1} + \cdots + \alpha_w > w - j$. In particular, $j = w - 1$ gives $\alpha_w > 1$. In fact, this is also directly obvious because α_w is the number of upward steps before the first downward step. Conversely, given $\widetilde{\alpha} \in A_w$, consider the path which makes α_w many upward transitions starting from $(0,0)$, then makes a downward transition followed by α_{w-1} many upward transitions, and so on. It is easy to see that this gives a path of the required type for the random walk. ∎

The above two results lead to the following simple estimate of the distribution of W, for large values of a. For stating this we need the following notation. For two functions $h(a)$ and $g(a)$, we will write $h(a) \sim g(a)$ as $a \to \infty$ to mean that $\lim_{a\to\infty}[h(a)/g(a)] = 1$. It is easy to see that, $h_1(a) \sim g_1(a)$ and $h_2(a) \sim g_2(a)$ as $a \to \infty$ imply that $h_1(a) + h_2(a) \sim g_1(a) + g_2(a)$ as $a \to \infty$.

Lemma 4.5: *For each* $w \geq 0$,

$$P(W = w) \sim \frac{b}{2w+b}\binom{2w+b}{w+b} p_a^w q_a^{b+w} \qquad as \quad a \to \infty\,.$$

Proof: For each $l = 0, 1, \ldots, w-1$,

$$\frac{p_{a-l}}{p_a} = \frac{a-l}{a-l+\rho}.\frac{a+\rho}{a} \to 1 \quad \text{as } a \to \infty\,.$$

For each $j = 0, 1, \ldots, w$,

$$\frac{q_{a-j}}{q_a} = \frac{\rho}{a-j+\rho}.\frac{a+\rho}{\rho} \to 1 \quad \text{as } a \to \infty\,.$$

It follows that for a fixed w and an $\widetilde{\alpha} \in A_w$ we have,

$$\prod_{l=0}^{w-1} p_{a-l} \prod_{j=0}^{w} q_{a-j}^{\alpha_j} \sim p_a^w q_a^{b+w}\,.$$

By summing over $\widetilde{\alpha} \in A_w$ and using Theorem 4.3, we get

$$P(W = w) \sim |\, A_w \,|\, p_a^w q_a^{b+w} \quad \text{as} \quad a \to \infty,$$

whence the assertion follows by using Lemma 4.4. ∎

We now present two theorems due to Williams (1971) and Whittle (1955), known as the *Threshold Theorems* for the General Stochastic Epidemic. The common theme of both the theorems is to identify ρ as a threshold quantity to determine whether the epidemic builds up or not. The following lemma will play a crucial role in the proofs of the threshold theorems. This is due to Williams. However, his proof is non-probabilistic and uses certain power series expansion. We give a probabilistic proof that uses transience of random walk.

Lemma 4.6: *For* $0 \leq p < 1$ *and* $q = 1 - p$,

$$\sum_{w=0}^{\infty} \frac{b}{2w+b}\binom{2w+b}{w+b} q^w p^{b+w} = \min\left\{\left(\frac{p}{q}\right)^b, 1\right\}.$$

Proof: It suffices to show that if $p \geq q$, then

$$\sum_{w=0}^{\infty} \frac{b}{2w+b}\binom{2w+b}{w+b} q^w p^{b+w} = 1.$$

First recall that $\frac{b}{2w+b}\binom{2w+b}{w+b}$ is precisely the number of paths of a random walk starting at the origin and reaching the state b in $(2w+b)$ many steps without hitting the x-axis. But by reversing the motion, this would be the same as the number of paths starting at state b and reaching the origin for the first time in $(2w+b)$ many steps, or equivalently (by shifting the x-axis), starting at the origin and reaching state $-b$ for the first time in $(2w+b)$ many steps. Thus if we consider a random walk with probability q (p respectively) of upward (downward respectively) transitions, then the summand is just the probability that such a random walk starting from the origin reaches $-b$ for the first time in $(2w+b)$ many steps. Since for different w, the above events are disjoint, summing over w gives us the probability that such a random walk starting from the origin ever reaches $-b$. By Exercise 2(vi) of Section 0.8.1, this probability is one whenever $q \leq p$ (Do not forget that here q is the probability of upward transition). ∎

Let us now present the two threshold theorems mentioned above in the way that we understand them. Both the threshold theorems are statements concerning the extent of the epidemic for large values of the initial number of susceptibles a. To be precise they both talk about the limiting probabilities as $a \to \infty$. Since we are varying a, it makes sense to allow ρ also to possibly vary with a. This dependence is going to be made explicit by using the notation ρ_a. In what follows we consider limits of certain probabilites as the pair (a, ρ_a) varies in such a way that $a \to \infty$ and ρ_a/a converges to a limit, say, δ. Since a is varying, the probabilities associated to the corresponding epidemic model will also vary. We will make it explicit by writing P_a for such probabilities. Note that we are keeping b, the initial number of infected individuals, fixed.

Theorem 4.7 (Williams' Threshold Theorem): *If* (a, ρ_a) *vary in such a way that* $a \to \infty$ *and* $\frac{a}{\rho_a} \to \delta$, *then*

$$\lim_{M\to\infty} \lim_{a\to\infty} P_a(W \leq M) \quad = \quad \min(\delta^b, 1).$$

Proof: By Theorem 4.5, for each w,

$$\lim_{a\to\infty} P_a(W = w) = \frac{b}{2w+b}\binom{2w+b}{w+b}\left(\frac{1}{1+\delta}\right)^w \left(\frac{\delta}{1+\delta}\right)^{b+w},$$

so that for any M,

$$\lim_{a\to\infty} P_a(W \le M) = \sum_{w=0}^{M} \frac{b}{2w+b}\binom{2w+b}{w+b}\left(\frac{1}{1+\delta}\right)^w\left(\frac{\delta}{1+\delta}\right)^{b+w}.$$

Lemma 4.6 now completes the proof. ∎

The quantity $\lim_{a\to\infty} P_a(W \le M)$ can be interpreted as the probability of having an epidemic of size at most M, for large values of a. Taking now the limit of that probability as $M \to \infty$ could therefore have the interpretation of being the probability of a finite epidemic for large values of a. Thus, Theorem 4.7 says that, if $\delta \ge 1$ then the epidemic is surely of finite size, while for $\delta < 1$ the probability of a finite epidemic is δ^b, which is strictly smaller than one. Indeed, this is how the Threshold Theorem is stated in the literature. There is one little subtlety, namely, instead of stating the result in terms of $\delta = \lim_a(\rho_a/a)$, the standard practice is to simply say that for large values of a, the probability of a finite epidemic equals one if $\rho_a \ge a$, while it equals $(\rho_a/a)^b$ if $\rho_a < a$. This is what Theorem 4.7 may be argued to say, provided the inequalities and equalities are all interpreted properly. For example, $\rho_a < a$ should be interpreted as $\lim_a(\rho_a/a) < 1$ (or more generally, for the present purpose, $\limsup_a(\rho_a/a) < 1$).

We shall now proceed to Whittle's Threshold Theorem. This deals with the probability, for large values of a, of the epidemic not exceeding a certain proportion of the initial number of susceptibles a. More specifically, for fixed x, $0 < x < 1$, we consider the probability $P_a(W \le xa)$. Whittle's Threshold Theorem attempts to get two-sided bounds for these probabilities, at least for large values of a.

Getting an asymptotic lower bound is not difficult. Let us assume as before that the parameters (a, ρ_a) vary in such a way that $a \to \infty$ and $(\rho_a/a) \to \delta$. Denoting $\pi_x^a = P_a(W \le xa)$, we show that

$$\lim_{a\to\infty} \pi_x^a \quad \ge \quad \min\left\{\delta^b, 1\right\}. \tag{11}$$

To see this, fix any n and observe that for large a, we have $xa > n$, so that

$$\pi_x^a \quad \ge \quad \sum_{w=0}^{n} P_a(W = w).$$

Using Lemma 4.5,

$$\lim_{a\to\infty} \pi_x^a \quad \ge \quad \sum_{w=0}^{n} \frac{b}{2w+b}\binom{2w+b}{w+b}\left(\frac{1}{1+\delta}\right)^w\left(\frac{\delta}{1+\delta}\right)^{b+w}.$$

The above inequality being true for all n, Lemma 4.6 yields the inequality (11).

Inequality (11) constitutes only one half of Whittle's Threshold Theorem and is often stated as

$$P(W \leq xa) \quad \geq \quad \min\left\{\left(\frac{\rho}{a}\right)^b, 1\right\} \quad \text{for large values of } a\,.$$

This is true as long as it is properly interpreted as discussed after the statement of Theorem 4.7 above.

The other half of Whittle's Theorem, which seeks an upper bound, is based on the use of a *comparison technique* which is interesting in its own right and is described below.

Our epidemic process (X_t, Y_t) is a Markov process starting from (a, b) and having transition mechanism determined by the parameters p_r. Consider now another chain evolving in the same manner, but with a different transition mechanism determined by parameters p'_r. To avoid complication, we use the same notation (X_t, Y_t) for this new process also. The difference in transition mechanism is indicated by using P' for probabilities of the new chain. If $p_r \geq p'_r$ for every r, then it is natural (why?) to expect that the random variable X_τ is stochastically larger under P' than under P, that is,

$$P'(X_\tau \geq k) \quad \geq \quad P(X_\tau \geq k) \quad \text{for each } k\,. \tag{12}$$

We shall show that this indeed is the case. But, for the present, we assume this and proceed to complete the remaining half of Whittle's theorem.
Recall that the parameters for our epidemic process are defined as

$$p_r \quad = \quad \frac{r}{r+\rho_a} \quad \text{for} \quad 0 \leq r \leq a\,.$$

Let us define p'_r for $0 \leq r \leq a$, as

$$\begin{aligned} p'_r \quad &= \quad p_r \qquad\qquad\quad \text{for} \quad r < (1-x)a \\ &= \quad \frac{(1-x)a}{(1-x)a+\rho_a} \qquad \text{for} \quad r \geq (1-x)a\,. \end{aligned}$$

Clearly, for every r, $p_r \geq p'_r$, so that by (12)

$$P(X_\tau \geq (1-x)a) \quad \leq \quad P'(X_\tau \geq (1-x)a)\,.$$

In view of the fact that $W = a - X_\tau$, the above inequality is the same as

$$P(W \leq xa) \quad \leq \quad P'(W \leq xa)\,.$$

Invoking the arguments of Theorem 4.3 and using Lemma 4.4, the right hand side can easily be seen to equal

$$\sum_{w=0}^{xa} \frac{b}{2w+b}\binom{2w+b}{w+b}\left(\frac{(1-x)a}{(1-x)a+\rho_a}\right)^w\left(\frac{\rho_a}{(1-x)a+\rho_a}\right)^{b+w},$$

which by Lemma 4.6 is clearly bounded above by $\min\left\{\left(\frac{\rho_a}{(1-x)a}\right)^b, 1\right\}$. We conclude that

$$P(W \leq xa) \quad \leq \quad \min\left\{\left(\frac{\rho_a}{(1-x)a}\right)^b, 1\right\}.$$

We have thus proved

Theorem 4.8 (Whittle's Threshold Theorem): *For any x, $0 < x < 1$, and for large values of a,*

$$\min\left\{\left(\frac{\rho_a}{a}\right)^b, 1\right\} \quad \leq P(W \leq xa) \leq \quad \min\left\{\left(\frac{\rho_a}{(1-x)a}\right)^b, 1\right\}.$$

Although we have stated the result in the way it is usually done, the reader should note that, the right hand side inequality is actually valid for all a, whereas the left hand side is valid only in the limit, that is, in the sense discussed earlier. It may be noted that a comparison technique, similar to the one used above, can be used also to get a lower bound valid for all a. Indeed, one can show (left as an exercise) that

$$P(W \leq xa) \quad \geq \quad \sum_{w=0}^{xa} \frac{b}{2w+b}\binom{2w+b}{w+b}\left(\frac{a}{a+\rho_a}\right)^w\left(\frac{\rho_a}{a+\rho_a}\right)^{b+w}$$

for all a and all x, $0 < x < 1$. It is tempting to claim that the right hand side of the above inequality is approximately $\min\left\{\left(\frac{\rho_a}{a}\right)^b, 1\right\}$, for large a, in view of Lemma 4.6. One may then erraneously claim that $\min\left\{\left(\frac{\rho_a}{a}\right)^b, 1\right\}$ is an actual lower bound for $P(W \leq xa)$ for all large a. In fact, the standard literature seems to make that claim. We wish we could justify this, thus avoiding interpretation through limits.

We now get back to our claim (12). Since comparison technique is an important and useful technique in the context of Markov chains, we will prove a slightly more general result. First, let us introduce some notation.

For any pair of integers $a \geq 1$, $b \geq 1$, and any a-tuple $\theta = (\theta_1, \ldots, \theta_a)$, with $0 \leq \theta_r \leq 1$ for all r, let P_θ denote the probability law of the Markov chain starting from (a, b) having state space

$$S = \{(r, s) : r, s \text{ non-negative integers}; \quad r \leq a; \quad r + s \leq a + b\}$$

and evolving in the following manner. State (r, s) is absorbing unless both r and s are strictly positive. From a non-absorbing state (r, s), the chain moves to $(r-1, s+1)$ with probability θ_r and moves to $(r, s-1)$ with probability $1 - \theta_r$. We will denote this process by $(U_n^{a,b}, V_n^{a,b})$. It is left as an exercise for

the reader to verify that we have a Markov chain on a finite state space for which every non-absorbing state is transient and hence it is an absorbing chain. Let T be the time till absorption. Thus $P_\theta(T < \infty) = 1$. Our objective would be to get a stochastic comparison of the random variable $U_T^{a,b}$, to be denoted by $Z^{a,b}$, for various a-tuples θ. The relevance of this in our context stems from the fact that, with $\theta_r = r/(r+\rho)$ for $1 \leq r \leq a$, the chain (U_n, V_n) is just the embedded chain associated with our epidemic process (X_t, Y_t) stopped at time τ. In particular the random variable X_τ and $Z^{a,b}$ are identical. We want to prove

Theorem 4.9: *If θ and θ' are two a-tuples with $\theta'_r \geq \theta_r$ for all r, then $Z^{a,b}$ is stochastically larger under P_θ than under $P_{\theta'}$, that is, for all k,*

$$P_\theta(Z^{a,b} \geq k) \quad \geq \quad P_{\theta'}(Z^{a,b} \geq k)\,.$$

To prove the theorem, we need a series of lemmas.

Lemma 4.10: *Let $w_1, \ldots, w_n$ and $v_1, \ldots, v_n$ be non-negative numbers such that for $1 \leq j \leq n$, $\sum_{i \leq j} w_i \leq \sum_{i \leq j} v_i$. Then, for any sequence of numbers $c_1 \geq c_2 \geq \cdots \geq c_n \geq 0$, one has $\sum_{i \leq n} c_i w_i \leq \sum_{i \leq n} c_i v_i$.*

Proof: Note that the hypothesis implies that for each $j = 1, \ldots, n-1$, the inequality

$$(c_j - c_{j+1}) \sum_{i \leq j} w_i \leq (c_j - c_{j+1}) \sum_{i \leq j} v_i$$

holds. Putting $c_{n+1} = 0$, the same inequality is seen to hold for $j = n$ also. Adding these n inequalities yields the desired result. ∎

Lemma 4.11: *For any θ, the probability $P_\theta(Z^{a,b} \leq k)$ is non-decreasing in b.*

Proof: We shall show that $P_\theta(Z^{a,b+1} \leq k) \geq P_\theta(Z^{a,b} \leq k)$. Suppose that $s \leq k$ and α is a path from (a, b) hitting the x-axis for the first time at $(s, 0)$. Let α^* be the path obtained by adding one to the second co-ordinate of all points of the path α. Clearly α^* is a path from $(a, b+1)$ and hitting the horizontal line $y = 1$ for the first time at the point $(s, 1)$. Let η be the hitting time of the line $y = 1$. The correspondence $\alpha \longleftrightarrow \alpha^*$ and the fact that the two paths α, α^* have the same probabilities (because the transition probabilities from any state depend only on the first coordinate of the state and we have not disturbed the first coordinates of points in α to get α^*) can be put together to deduce that $P_\theta(U_T^{a,b} \leq k) = P_\theta(U_\eta^{a,b+1} \leq k)$. However from the dynamics of the process it is clear that the event $(U_\eta^{a,b+1} \leq k)$ implies $(U_T^{a,b+1} \leq k)$. It now follows that $P_\theta(U_T^{a,b+1} \leq k) \geq P_\theta(U_T^{a,b} \leq k)$, as was to be shown. ∎

Lemma 4.12: *Let θ and θ' be two a-tuples such that $\theta'_a \geq \theta_a$, while $\theta'_r = \theta_r$, for all $r < a$. Then $Z^{a,b}$ is stochastically larger under P_θ than under $P_{\theta'}$.*

Proof: Let $k \leq a - 1$. We prove

$$P_\theta(Z^{a,b} \leq k) \quad \leq \quad P_{\theta'}(Z^{a,b} \leq k)\,.$$

Let η be the hitting time of the vertical line $x = a - 1$. Note that the event $(Z^{a,b} \leq k)$ implies that $\eta < \infty$. Indeed, $\eta \leq b$ and hence

$$P_\theta(Z^{a,b} \leq k) \quad = \quad \sum_{i=1}^{b} P_\theta(Z^{a,b} \leq k \,|\, \eta = i) P_\theta(\eta = i)\,.$$

Using the Markov property, the conditional probability $P_\theta(Z^{a,b} \leq k \,|\, \eta = i)$ is the same as the probability $P_\theta(Z^{a-1,b-i+2} \leq k)$, so that

$$P_\theta(Z^{a,b} \leq k) = \sum_i P_\theta(Z^{a-1,b-i+2} \leq k) P_\theta(\eta = i)\,.$$

Analogously,

$$P_{\theta'}(Z^{a,b} \leq k) = \sum_i P_{\theta'}(Z^{a-1,b-i+2} \leq k) P_{\theta'}(\eta = i)\,.$$

Since $\theta'_r = \theta_r$, for $r \leq a - 1$, it is clear that for every i,

$$P_\theta(Z^{a-1,b-i+2} \leq k) = P_{\theta'}(Z^{a-1,b-i+2} \leq k) = c_i, \quad \text{say.}$$

Lemma 4.11 gives that c_i is non-increasing in i. Putting $w_i = P_\theta(\eta = i)$ and $v_i = P_{\theta'}(\eta = i)$ for $i \leq b$, we complete the proof simply by showing that the hypothesis of Lemma 4.10 holds. Observe that $w_i = (1 - \theta_a)^{i-1}\theta_a$ and $v_i = (1 - \theta'_a)^{i-1}\theta'_a$, so that for any j, $\sum_{i \leq j} w_i = 1 - (1 - \theta_a)^{j+1}$ and $\sum_{i \leq j} v_i = 1 - (1 - \theta'_a)^{j+1}$. From the hypothesis that $\theta'_a \geq \theta_a$, it follows that $\sum_{i \leq j} w_i \leq \sum_{i \leq j} v_i$ holds for all j. ∎

Lemma 4.13: *Let $1 \leq m \leq a$. Suppose θ and θ' are such that $\theta'_m \geq \theta_m$ while $\theta'_r = \theta_r$ for all $r \neq m$. Then $Z^{a,b}$ is stochastically larger under P_θ than under $P_{\theta'}$.*

Proof: In view of Lemma 4.12, we need only consider $m < a$. Observe that for $k \geq m$, the hitting time η of the vertical line $x = k$ has the same distribution under both P_θ and $P_{\theta'}$. In view of

$$P_\theta(Z^{a,b} \leq k) \quad = \quad P_\theta(\eta \leq b - a + k - 1)$$

and similar equality under $P_{\theta'}$, it follows that

$$P_\theta(Z^{a,b} \leq k) \quad = \quad P_{\theta'}(Z^{a,b} \leq k) \quad \text{for all} \quad k \geq m.$$

We now consider $k \leq m - 1$ and show

$$P_\theta(Z^{a,b} \leq k) \quad \leq \quad P_{\theta'}(Z^{a,b} \leq k)\,.$$

Let us now denote η to be the hitting time of the vertical line $x = m - 1$. By the same argument as used in Lemma 4.12, one sees that

$$P_\theta(Z^{a,b} \leq k) \quad = \quad \sum_{i=1}^{b+2(a-m)} P_\theta(Z^{m-1,b-i+2a-2m+2} \leq k) P_\theta(\eta = i)$$

and

$$P_{\theta'}(Z^{ab} \leq k) \quad = \quad \sum_{i=1}^{b+2(a-m)} P_{\theta'}(Z^{m-1,b-i+2a-2m+2} \leq k) P_{\theta'}(\eta = i)\,.$$

Let $w_i = P_\theta(\eta = i)$ and $v_i = P_{\theta'}(\eta = i)$ for $i \leq b + 2(a-m)$. As in the proof of Lemma 4.12, we get the desired result once we show that for every j, $\sum_{i \leq j} w_i \leq \sum_{i \leq j} v_i$, that is, $P_\theta(\eta \leq j) \leq P_{\theta'}(\eta \leq j)$. This can perhaps be seen directly but here is a trite method.
Let $\bar{\eta}$ be the hitting time of the vertical line $x = m$. Noting that $\bar{\eta}$ has the same distribution under P_θ and $P_{\theta'}$, it suffices to show that for every $l \leq j-1$, $P_\theta(\eta \leq j \,|\, \bar{\eta} = l) \leq P_{\theta'}(\eta \leq j \,|\, \bar{\eta} = l)$. Using the Markov property, one sees that $P_\theta(\eta \leq j \,|\, \bar{\eta} = l) = 1 - (1 - \theta_m)^{j-l}$, while $P_{\theta'}(\eta \leq j \,|\, \bar{\eta} = l) = 1 - (1 - \theta'_m)^{j-l}$, from which the required inequalities follow. The proof is now complete. ∎

Proof of Theorem 4.9: Define $a+1$ many a-tuples, $\theta^0, \theta^1, \ldots, \theta^a$ by

$$\begin{aligned} \theta_i^m &= \theta'_i \quad \text{for} \quad i \geq a - m + 1 \\ &= \theta_i \quad \text{for} \quad i \leq a - m\,. \end{aligned}$$

Note that for any $0 \leq m \leq a-1$, we have $\theta^m_{a-m} \leq \theta^{m+1}_{a-m}$ and $\theta^m_r = \theta^{m+1}_r$ for all $r \neq a - m$. It follows from Lemma 4.13 that

$$P_{\theta^m}(Z^{a,b} \leq k) \quad \leq \quad P_{\theta^{m+1}}(Z^{a,b} \leq k)$$

for all k and all m with $0 \leq m \leq a-1$. Noticing that $\theta^0 = \theta$ and $\theta^a = \theta'$ the proof is complete. ∎

4.4 Spread in Households: Chain Binomial Models

The models discussed so far study the spread of an epidemic in a community at large. In this section, we take up the question of how an infectious disease spreads in a particular household. We shall discuss two stochastic models to describe this phenomenon — one is due to M. Greenwood and the other due to J. Reed and W.H. Frost.

Suppose that in a household, some individuals got infected by a contagious disease. This puts the other members of the household at the risk of catching the disease. Of course, in reality there is a fixed period of incubation and it is only after that period, that the infected individuals become infectious. The disease now spreads through contacts between the infected and uninfected individuals. However, not every such contact is likely to result in a new infection. Thus, there is a chance factor arising out of both the possibility of contact as well as a contact resulting in an infection. Specification of this chance factor is what would constitute a stochastic model. Before going into the details of the models, we describe the common setup.

We assume that there are K individuals in a household and initially s_0 of them are infected. We denote by r_0 the initial number of uninfected individuals, that is, $r_0 = K - s_0$. We assume that the incubation period is one time unit. To simplify matters, we also assume that the infected individuals remain infectious only for an instant of time at the end of the incubation period. This is indeed a simplifying assumption. However, in reality the period of infectiousness may often be very small, for example, they may perhaps be quarantined or even be cured and become immune. Let s_1 denote the number of new infections at time 1. The number of uninfected in circulation now is $r_1 = r_0 - s_1$. In general, let s_n be the number of persons who got infected at time n and $r_n = r_{n-1} - s_n$ be the resulting number of uninfected in circulation. It is to be noted that, at time n, the persons who can pass on the infection are precisely those who became newly infected at time $n-1$. Also, at any point of time the persons who are susceptible are only those who have not been infected so far. Clearly, as soon as $s_n = 0$, there will be no more new infections and the epidemic will come to a halt. Of course $r_{n+1} = 0$ would also guarantee this (perhaps not in a desirable way).

4.4.1 Greenwood Model

According to the model proposed by M. Greenwood, the probability of a susceptible coming in contact with the group of infectious persons and getting himself infected is assumed to be a constant p, $0 < p < 1$. Moreover the fates of different susceptibles are assumed to be stochastically independent. Clearly, these assumptions lead to a binomial distribution for the number of new infections at time n. More precisely, if at time $(n-1)$, there are s_{n-1} newly-infected persons (with $s_{n-1} > 0$) and if r_{n-1} denotes the number of susceptibles, then the probability of s_n new infections at time n is

$$\binom{r_{n-1}}{s_n} p^{s_n} (1-p)^{r_{n-1}-s_n}, \qquad \text{for } s_n = 0, 1, \ldots . r_{n-1} .$$

In case $s_{n-1} = 0$, then $s_n = 0$ and hence $r_n = r_{n-1}$. Note that in case $s_{n-1} > 0$, its actual value has no relevance in the distribution of the number of new infections at time n. This is one of the important features of this model.

Denote by S_n and R_n, the number of new infections and the number of susceptibles respectively at time n. Thus S_n and R_n are random variables. Also $(R_n, S_n)_{n \geq 0}$ is a Markov chain. The state space of this process is

$$\{(i,j) : i \geq 0,\ j \geq 0,\ i + j \leq K\} .$$

This is an absorbing chain and the absorbing states are precisely the states $\{(i,0) : i \leq K\}$. The transition probabilities are given by

$$\begin{aligned} P_{(i,j)(i',j')} &= \binom{i}{j'} p^{j'} (1-p)^{i'} \quad \text{for } 0 \leq j' \leq i \text{ and } i' = i - j' \quad \text{if} \quad j > 0 \\ &= \delta_{(i,j)(i',j')} \qquad\qquad\qquad\qquad\qquad\qquad\qquad\quad \text{if} \quad j = 0 . \end{aligned}$$

Note that, for $j > 0$, $P_{(i,j),(i',j')}$ does not depend on j. This enables us to replace the original bivariate chain by an appropriately stopped univariate chain as follows.

Consider the Markov chain $(X_n)_{n\geq 0}$ with state space $\{0, 1, \ldots, K\}$, initial state $X_0 = r_0$, and transition probabilities

$$P(X_{n+1} = j \mid X_n = i) = \binom{i}{j} p^{i-j}(1-p)^j \quad \text{for} \quad j = 0, 1, \ldots, i\,.$$

Let T be the stopping time defined by

$$T = \min\{n : X_n = X_{n-1}\},$$

that is, T is the first time the chain did not move from its previous state. Let (Y_n) be the process (X_n) stopped at T, that is,

$$\begin{aligned} Y_n &= X_n \quad \text{if} \quad n \leq T \\ &= X_T \quad \text{if} \quad n > T\,. \end{aligned}$$

It is to be noted that $(Y_n)_{n\geq 0}$ is no longer a Markov chain, as we are going to see. A moment's reflection shows that the process $(Y_n)_{n\geq 0}$ is precisely $(R_n)_{n\geq 0}$ of the Greenwood Model; just recall that $S_n = 0$ is same as saying that $R_n = R_{n-1}$. And of course, $(R_n)_{n\geq 0}$ itself is not a Markov chain. In the new formulation, the random variable T is clearly seen to represent the duration of the epidemic. The rest of this section is devoted to finding the distribution of T.

More generally, let $(X_n)_{n\geq 0}$ be a Markov chain with state space $\{0, 1, \ldots, K\}$ and an arbitrary transition matrix $P = ((p_{ij}))$. We only assume that the diagonal entries of P are positive. For this Markov chain, we want to find the distribution of the stopping time T defined as

$$T = \min\{n : X_n = X_{n-1}\}\,.$$

The analysis that follows is due to J. Gani and M. Jerwood. Let Q denote the diagonal matrix with diagonal entries same as those of P and let $R = P - Q$. Clearly, R has all its off-diagonal elements same as those of P, while all its diagonal entries are zero. Using this notation it is now easy to see that

$$P(T = n, X_n = j \mid X_0 = i) = R_{ij}^{n-1} p_{jj} \quad \text{for each } j\,, \tag{13}$$

so that

$$P(T = n \mid X_0 = i) = e_i' R^{n-1} Q e, \tag{14}$$

where e is the column vector with all entries one and e_i' is the row vector with i-th entry one and all other entries zero. Recall that R is a matrix with non-negative entries and having each row sum strictly less than one. Elementary matrix theory shows that $(I - R)$ is invertible and $(I - R)^{-1} = \sum_{n=0}^{\infty} R^n$. The facts that $Q = P - R$ and $Pe = e$ can now be used to deduce that

$\sum_{n=1}^{\infty} e_i' R^{n-1} Qe = 1$. In view of (14), we have thus proved that T is finite with probability one. We now go on to find the p.g.f. of T. Throughout, we assume that we are starting from a fixed initial state i and denote by $g_i(\theta)$, the corresponding p.g.f. of T. Thus

$$g_i(\theta) = \sum_{n=1}^{\infty} \theta^n e_i' R^{n-1} Qe = e_i'(I - \theta R)^{-1}(\theta Q)e\,.$$

In view of its similarity with the p.g.f. of the standard geometric distribution, the distribution of T has been called a *Markov Geometric Distribution* by Gani and Jerwood. The moments of T — in particular its expectation and variance — can now be easily obtained by successive differentiation of the p.g.f. For example, it turns out that

$$E(T \,|\, X_0 = i) = e_i'(I - R)^{-1}e\,.$$

Turning back to (13), it can also be written as

$$P(T = n \text{ and } X_T = j \,|\, X_0 = i) = R_{ij}^{n-1} p_{jj}\,.$$

Thus we actually have the joint distribution of (T, X_T). One can use this to find the marginal distribution of X_T, in particular the expected value and variance of X_T.

Let us now return to the Greenwood Model. This is a special case where,

$$P = \begin{pmatrix} 1 & 0 & 0 & \dots & 0 \\ p & q & 0 & \dots & . \\ p^2 & 2pq & q^2 & \dots & . \\ \vdots & \vdots & \vdots & \vdots\vdots\vdots & \vdots \\ p^K & \binom{K}{1} p^{K-1} q & \binom{K}{2} p^{K-2} q^2 & \dots & q^K \end{pmatrix}.$$

Note that in this case R^K is the zero matrix, implying that $T \leq K$ with probability one. This is understandable, because the longest possible duration of the epidemic happens when one new person gets infected each day. However this does not make explicit computations all that easy (compare R^2 and R^3!).

4.4.2 Reed-Frost Model

The model proposed by J. Reed and W.H. Frost differs from the earlier one in that here the probability of one particular susceptible coming in contact with one particular infectious person and getting infected is denoted by p. Of course the usual assumption of independence of interaction between different individuals is retained. It follows that, with the same notations r_n, s_n as in the Greenwood Model, the probability of any one of the r_{n-1} susceptibles getting infected at time n is $1 - (1-p)^{s_{n-1}}$, so that the probability of s_n many new infections at time n is given by

$$\binom{r_{n-1}}{s_n} [1 - (1-p)^{s_{n-1}}]^{s_n} (1-p)^{s_{n-1}(r_{n-1} - s_n)} \quad \text{for} \quad s_n = 0, 1, \dots, r_{n-1}\,.$$

Note that in case $s_{n-1} = 0$, the above formula automatically implies that $s_n = 0$. For detailed analysis of the Reed-Frost Model, interested reader can consult Von Bahr & Martin Lof (1980) and F. Ball (1983).

4.5 Spatial Spread: Neyman-Scott Model

In the earlier models, the geographic location of the epidemic was fixed and the temporal spread was under study. In this section, we describe a model proposed by J. Neyman and E. L. Scott for the spread of epidemic over a geographical area.

The geographical area under consideration is called the *habitat*, denoted by H. Mathematically, H could be any subset of the Euclidean plane R^2, preferably open. However, to simplify matters, we take our habitat to be all of R^2. As before, the incubation period is assumed to be one time unit and the period of infectiousness is contracted to a single point. It is reasonable to assume that an infectious person at a particular location in the habitat infects only susceptibles at that location. However, it is equally reasonable that an infectious person at a crowded location is likely to infect more people than at a desolate location. This dependence of infectivity on the location should be captured in the model. Another feature of the proposed model is that it tries to capture the mobility factor also. In other words, it takes into account the fact that an individual infected at a particular location may move to another location by the time he becomes infectious. Indeed, that is how the infection spreads over the habitat. Mathematically, this will involve introducing two parameters, one for the spread of infection and the other for the mobility of the infected individual. This is done as follows.

For every $u \in H$, we have a probability distribution γ_u on non-negative integers, representing the distribution of the number of individuals infected by one infectious person at the location u. We denote the p.g.f. of γ_u by $g(\cdot \mid u)$. We emphasize the dependence of γ_u on u as mentioned earlier. To take care of the mobility factor we have, for every $u \in H$, a probability density function $f_u(\cdot)$ on H. This has the following interpretation. A person infected at u at time k moves to a region $S \subset H$ at time $(k+1)$ with probability $\int_S f_u(x)\,dx$. Our model assumes that different individuals act independently. That is, first of all, the number of individuals infected by different infectious persons are independent random variables, no matter where the infectious persons are located. Secondly, given any set of infected individuals at time k, no matter where they are located, the places where they move to at time $k+1$ are independent random variables. The dependence of $f_u(\cdot)$ on the location u has an easy explanation. A person infected at home is not very likely to move away from home by the time he becomes infectious; whereas, if one catches infection when he is on board a train, he is quite likely to move far away.

We shall now see how to describe mathematically the temporal spread of the epidemic over the entire habitat. To fix ideas, we start with one infectious

person at time 0 at location u. This infectious person will infect a certain number, say X_1^u , of individuals at location u. From what has been said earlier, X_1^u is a random variable with distribution γ_u and p.g.f. $g(\cdot\,|\,u)$. By the time these persons become infectious, they would have moved to various locations — each person, independently of the others, choosing a random location given by the probability density $f_u(\cdot)$. Each of them would now infect people in their respective new locations, who would in turn move to different locations by the time they become infectious, and so on. This is how the infection would spread over the habitat with time. Let X_n^u denote the number of infectious people in the entire habitat at time n. The dependence on u comes from the fact that we started initially with one infected person at the location u. We want to study the distribution of X_n^u for $n \geq 1$. Let us denote the p.g.f. of X_n^u by $G_n(\cdot\,|\,u)$. Thus clearly

$$G_1(\cdot\,|\,u) = g(\cdot\,|\,u)\,. \tag{15}$$

To get the p.g.f. of X_2^u, we argue as follows. Consider the i-th person infected by the initial infectious and let Y_i denote the number of individuals infected by him at time 1. It should be clear that Y_i has p.g.f. given by

$$H_1(t\,|\,u) = \int g(t\,|\,x) f_u(x)\,dx\,. \tag{16}$$

This is because, given that the i-th individual has moved to location x at time 1, the conditional p.g.f. of the number of individuals infected by him is $g(\cdot\,|\,x)$, so that the unconditional p.g.f. would indeed be H_1 as given. It should be noted here that to make sense of the integral in (16), some assumptions on g as a function of x are needed. [For example, assuming that g varies continuously with x would do. A reader familiar with the Lebesgue Integration Theory would quickly see that measurability of g in the x variable is all that is needed.]

Since the Y_i are i.i.d. and $X_2^u = \sum_{i=1}^{X_1^u} Y_i$, it follows that

$$G_2(\cdot\,|\,u) = g(H_1(\cdot\,|\,u)\,|\,u)\,.$$

Proceeding in an analogous manner we can deduce that, for every $n \geq 1$,

$$H_n(\cdot\,|\,u) = \int G_n(\cdot\,|\,x) f_u(x)\,dx, \tag{17}$$

$$G_{n+1}(\cdot\,|\,u) = g(H_n(\cdot\,|\,u)\,|\,u)\,. \tag{18}$$

Note that even if we are interested only in $G_n(\cdot\,|\,u)$ for the specified initial location u, we have to compute $G_n(\cdot|x)$ for all x in order to get $H_n(\cdot|u)$, which is required for the subsequent $G_{n+1}(\cdot\,|\,u)$. Having started with one infectious individual at time 0 at location u, we have obtained the p.g.f. of the number of infectious individuals in the habitat at time n to be $G_n(\cdot\,|\,u)$. Along the way, we came across another sequence of functions which are also p.g.f.s, namely the $H_n(\cdot|u)$. The reader would naturally wonder as to which stochastic process they

correspond to. Well, instead of starting with one *infectious* person, suppose we had started with one *infected* person at time 0 at location u. Let us now consider the number of infected individuals — say Z_n^u, for $n \geq 1$ — in the entire habitat at successive time points. It is then clear that Z_n^u has p.g.f. $H_n(\cdot \mid u)$. Neyman and Scott describe X_n^u (Z_n^u respectively) as the n-th generation of an epidemic started by an infectious (infected, respectively) at location u at time 0. Let us denote the expected values of X_n^u and Z_n^u by α_n^u and β_n^u respectively. Using Equations (15)–(18) one gets

$$\alpha_1^u = m(u), \tag{19}$$

$$\beta_1^u = \int m(x) f_u(x)\, dx, \tag{20}$$

$$\alpha_{n+1}^u = m(u)\beta_n^u, \tag{21}$$

$$\beta_{n+1}^u = \int \alpha_{n+1}^x f_u(x)\, dx, \tag{22}$$

where $m(u)$ denotes the mean of the distribution γ_u. It is worth noting that, in order for the above formulae to be true, it is not necessary to assume that $m(x)$ is finite for each x.

We next discuss the problem of extinction of the epidemic from the habitat. We say that the epidemic, originating at u, is *heading for an extinction*, if X_n^u converges to zero in probability. Since the X_n^u are integer valued, this is equivalent to saying that $P[X_n^u = 0] \longrightarrow 1$ as $n \to \infty$. Here is a first positive result in this direction.

Theorem 4.14: *If* $\sup_x m(x) < 1$, *then for every* u, *the epidemic originating at* u *heads for extinction.*

Proof: Denote $\sup_x m(x)$ by c. Then clearly for any u, $\alpha_1^u \leq c$ by (19) and $\beta_1^u \leq c$ by (20). Using induction and the relations (21) and (22), one easily obtains that, for every u in H and every $n \geq 1$, $\alpha_n^u \leq c^n$ and $\beta_n^u \leq c^n$. In particular if $c < 1$, then $\alpha_n^u \to 0$. That is, $E(X_n^u) \to 0$ as $n \to \infty$. Note that

$$E(X_n^u) = \sum_{k=1}^{\infty} kP(X_n^u = k) \geq \sum_{k=1}^{\infty} P(X_n^u = k) = 1 - P(X_n^u = 0)\,.$$

It follows that $P(X_n^u = 0) \to 1$ as $n \to \infty$. ∎

We admit that the hypothesis that $\sup_x m(x) < 1$ in the above Theorem 4.14 is a strong one. However, it should be noted that, first of all, nothing is assumed about the mobility distributions. Secondly, the conclusion of the theorem is also very strong in the sense that the convergence is uniform over u, that is, $\sup_u P(X_n^u > 0) \to 0$. The next theorem on extinction has a slightly weaker hypothesis.

Theorem 4.15: *If* $\sup_x \beta_1^x < 1$, *then for every* u, *such that* $m(u) < \infty$, *an*

epidemic originating at u heads for extinction.

Proof: Denote $\sup_x \beta_1^x$ by c. It is easy to see by induction that, for every $n \geq 1$, $\alpha_n^u \leq m(u)c^{n-1}$. Therefore, if $c < 1$ and $m(u) < \infty$, then $\alpha_n^u \to 0$. The proof is now completed as earlier. ∎

The reader may note that under the hypothesis of Theorem 4.14, one surely has $\sup_x \beta_1^x < 1$ and, of course, for every u, $m(u) < 1$. After the above two theorems, which assert that under certain conditions the epidemic heads for extinction, we now go to a result describing when an epidemic does not.

Theorem 4.16: *If $H_1(0 \mid u) = 0$ for every u, then for every u such that $\gamma_u(\{0\}) < 1$, an epidemic originating at u does not head for extinction.*

Proof: We first show that $H_n(0 \mid u) = 0$, for every u and every n. Indeed, $G_2(0 \mid u) = g(0 \mid u)$, by Equation(18) and the hypothesis. But this, in turn, implies that $H_2(0 \mid u) = 0$ for all u, by Equation (17) and the hypothesis. Induction will now do the job. In particular, for every u and every n, one has $P(X_n^u = 0) = G_n(0 \mid u) = g(0 \mid u) = \gamma_u(\{0\})$, independent of n. It follows that if $\gamma_u(\{0\}) < 1$, then X_n^u does not converge to zero in probability. ∎

We shall discuss one more problem related to this model. Can it so happen that an epidemic originating at some locations will head for extinction, whereas an epidemic originating at others will not? We show that this cannot happen unless there are deserts or unless the mobility is curtailed. A desert means a location where an infectious person can not infect anybody else. More precisely, we say that a point $u \in H$ is a *desert*, if the distribution γ_u is concentrated on the singleton $\{0\}$. Also we say that there is *full mobility* in the habitat if for every u, the density $f_u(\cdot)$ is strictly positive everywhere. This means that an infected person from any location can move to any other location with positive probability by the time he becomes infectious. We are now ready to state the main result.

Theorem 4.17: *Assume that there are no deserts and that there is full mobility. Then, an epidemic originating at u will head for extinction, either for all $u \in H$ or for no $u \in H$.*

For the proof, we need a little lemma on integrals. We simply state it without proof, just because the proof needs Lebesgue integration theory, something that we are not assuming from the reader. However, those who are familiar with this theory will quickly agree that the result is indeed elementary. For those who are not, here is a motivation: suppose $\{a_n\}$ is a non-negative sequence and you are told that for some strictly positive sequence $\{p_n\}$, the series $\sum a_n p_n = 0$. It trivially follows that a_n must equal zero for all n. Analogously, suppose that $a(x)$ is a non-negative function of a real variable and you are told that the integral $\int a(x)p(x)\,dx = 0$ for some strictly positive function p. Now of course we cannot say that $a(x)$ must equal zero for all x. However what the lemma asserts is that $a(x)$ is almost zero.

Lemma 4.18: *Let $a(x)$ be a non-negative function on the real line such that*

for some strictly positive function $p(x)$, $\int a(x)p(x)\,dx = 0$. *Then for every function* $q(x)$, $\int a(x)q(x)\,dx = 0$.

Proof of Theorem 4.17: We start by showing that $G_n(0 \mid u)$ increases with n, for every u. This does not require any of the hypotheses of the theorem. We prove by induction. First note that $G_1(0 \mid u) = g(0 \mid u)$ and $H_1(0 \mid u) = \int g(0 \mid x) f_u(x)\,dx \geq 0$, so that

$$G_2(0|u) = g(H_1(0|u)|u) \quad \geq \quad g(0|u) = G_1(0|u)\,.$$

The inequality is a consequence of the fact that $g(\cdot \mid u)$ is a p.g.f. and hence non-decreasing. Assuming now that $G_n(0|x) \geq G_{n-1}(0|x)$ for all x, Equations (17) and (18) can be used to show that $G_{n+1}(0|x) \geq G_n(0|x)$.
As a consequence, for every x, $\lim_n G_n(0|x)$ exists, to be denoted by $G_\infty(0|x)$. From Equation (17), it follows that for every x, $H_n(0|x)$ is also non-decreasing with n and hence has a limit, say $H_\infty(0|x)$. Further, the Equation (17) and the Monotone Convergence Theorem [see Exercise 4, Section 0.4] give

$$H_\infty(0|u) = \int G_\infty(0|x) f_u(x)\,dx\,. \tag{23}$$

Again Equation (18) and the continuity of g_u gives

$$G_\infty(0|u) = g(H_\infty(0|u)|u)\,. \tag{24}$$

To prove the theorem now, suppose that for some $u_0 \in H$, an epidemic starting at u_0 heads for extinction, that is $G_\infty(0|u_0) = 1$. We show that, under the hypotheses of the theorem, $G_\infty(0 \mid u) = 1$ for all u, that is, an epidemic starting at any u heads for extinction. First observe that the hypothesis that there are no deserts, implies in particular, that $\gamma_{u_0}(\{0\}) < 1$. This, in turn, implies that $g(\cdot|u_0)$ is strictly increasing and, hence $g(t|u_0) = 1$ if and only if $t = 1$. Therefore, Equation (24) implies that $H_\infty(0|u_0) = 1$. In view of Equation (23), this means that $\int [1 - G_\infty(0|x)] f_{u_0}(x)\,dx = 0$. Now invoke Lemma 4.18, with $a(x) = 1 - G_\infty(0|x)$ and $p(x) = f_{u_0}(x)$, to deduce that for any u, $\int [1 - G_\infty(0|x)] f_u(x)\,dx = 0$, that is, $H_\infty(0|u) = 1$. This implies, by Equation (24), that $G_\infty(0|u) = 1$, as was to be proved. ∎

So far, we have been considering the spread of the epidemic over the entire habitat. However, in practice one may be more interested in the spread of the epidemic over certain pockets of the habitat. More precisely, let $R_1, R_2, \ldots, R_k$ be k disjoint sub-regions of the habitat. For any $u \in H$, let X_{ni}^u, for $1 \leq i \leq k$, denote the number of infectious persons in the region R_i at time n, for an epidemic starting at u. Note that we do not demand that $\bigcup_{i=1}^k R_i = H$; also, we allow for the possibility that $u \notin \bigcup_{i=1}^k R_i$. We may be interested in the distribution of the vector process $\widetilde{X}_n^u = (X_{n1}^u, \ldots, X_{nk}^u)$, in particular its asymptotic properties. The methods of this section enable us to discuss these issues as well. We discuss some of them in the exercises.

4.6 Exercises

1. Consider the simple deterministic epidemic. Sometimes the following function is interpreted as the frequency function of the time of occurrence of a new infection.

$$\frac{\omega(t)}{n} = -\frac{1}{n}\frac{dx}{dt} = \beta(n+1)^2 \frac{e^{\beta(n+1)t}}{[n+e^{\beta(n+1)t}]^2}.$$

Show that this is indeed the probability density of a non-negative random variable with mean $\log(n+1)/\beta n$.

2. Consider the simple stochastic epidemic with $\beta = 1$. Let q_r be the Laplace transform of p_r, that is ,

$$q_r(\lambda) = \int_0^\infty e^{-\lambda t} p_r(t)\, dt \quad \text{for} \quad \lambda > 0.$$

Show that

$$q_r = \frac{(r+1)(n-r)}{\lambda + r(n+1-r)} q_{r+1} \quad \text{for} \quad 0 \le r < n,$$

$$q_n = 1/(\lambda + n).$$

Hence deduce that

$$q_r = \frac{n!(n-r)!}{r!} \prod_{j=r}^{n} \frac{1}{\lambda + j(n-j+1)}.$$

3. Consider the simple stochastic epidemic. Let $F(x,t)$ be the generating function of $p_r(t)$ for $0 \le r \le n$, that is,

$$F(x,t) = \sum_{r=0}^{n} p_r(t) x^r.$$

Show that $F(x,0) = x^n$ and

$$\frac{\partial F}{\partial t} = \beta(1-x)\left(n\frac{\partial F}{\partial x} - x\frac{\partial^2 F}{\partial x^2}\right).$$

4. In the Chain-Binomial models, let $P(s_0, s_1, \ldots, s_n)$ denote the probability $P(S_0 = s_0, S_1 = s_1, \ldots, S_n = s_n)$.
Show that in the Greenwood model

$$P(s_0, s_1, \ldots, s_n) = \frac{r_0!}{s_1! s_2! \cdots s_n! r_{n+1}!} p^{\sum_1^n s_i} q^{\sum_1^{n+1} r_j}$$

and in the Reed-Frost model

$$P(s_0, s_1, \ldots, s_n) = \frac{r_0!}{s_1! s_2! \cdots s_n! r_{n+1}!} \; q^{\sum_0^n s_j r_{j+1}} \prod_0^{n-1} (1 - q^{s_i})^{s_{i+1}} .$$

5. Let $P(n, j, a)$ be the probability that a household of size n will have a total of j cases when there are a initial cases. Show that for the Greenwood model

$$P(n, j, a) = \sum_{k=1}^{j-a} \binom{n-a}{k} p^k q^{n-a-k} P(n-a, j-a, k) .$$

Show that for the Reed-Frost model, $P(n, a, a) = q^{a(n-a)}$, and

$$P(n, j, a) = \sum_{k=1}^{j-a} \binom{n-a}{k} (1 - q^a)^k q^{a(n-a-k)} P(n-a, j-a, k) .$$

Hence deduce that in the Reed-Frost model,

$$P(n, j, a) = \binom{n-a}{j-a} q^{j(n-j)} P(j, j, a) .$$

6. The idea is to describe, following R. Bartoszynski, a branching process model of epidemics. Here is the set-up.
(i) Every infected individual passes through a period of illness of $X + Y$ days, that is, a period of incubation of X days followed by a period of infectiousness of Y days. (X, Y) has joint p.g.f. $F(s, t) = \sum_{m=0}^{\infty} \sum_{n=1}^{\infty} p_{m,n} s^m t^n$. It is to be noted that Y is at least one. (ii) During the illness period of $X + Y$ days, a person may be detected and automatically isolated. The probability of getting detected on a day is $(1 - \alpha)$ during the incubation period and $(1 - \beta)$ during the infectious period. Here $0 < \alpha, \beta \leq 1$. (iii) During the Y days of infectiousness an undetected individual makes a certain number of contacts with the susceptibles. The number of contacts for different days are i.i.d with p.g.f. $R(t) = \sum_{k=0}^{\infty} r_k t^k$. (iv) Each contact of a susceptible with an infectious, independent of other contacts, leads to infection with probability γ where $0 < \gamma \leq 1$. (v) The events described above are independent for different individuals.
The interpretations of (i)–(v) are as follows. F describes the nature of the disease; R describes the social and environmental conditions like mobility etc.; α and β describe the efficiency of the health services in detecting the cases; γ measures the individual resistance via immunization programs of the health services.

(a) Put $q_{m,n} = \sum_{k=n+1}^{\infty} p_{m,k}$, $\quad Q(s, t) = \sum_{m,n=0}^{\infty} q_{m,n} s^m t^n$.
Show that $\sum_{m=0}^{\infty} q_{m,0} s^m = F(s, 1)$.
Show that for $|s| \leq 1$ and $|t| < 1$, $Q(s, t) = \dfrac{F(s, 1) - F(s, t)}{1 - t}$.

(b) Put ω_n = the probability that an infected individual remains undetected and infectious for exactly n days. Show that

$$\omega_n = \sum_{m=0}^{\infty} p_{m,n}\alpha^m\beta^n + (1-\beta)q_{m,n}\alpha^m\beta^n \quad \text{for} \quad n \geq 1,$$

$$\omega_0 = 1 - \sum_{1}^{\infty} \omega_n = 1 - \beta F(\alpha, 1)\,.$$

(c) Show that the p.g.f. of the number of persons infected by a single individual during one day of his infectiousness (when undetected) is given by $R(1-\gamma+\gamma t)$.

(d) Let $D(s) = \frac{\partial}{\partial t}F(s,t)\,|_{t=1} = \sum\sum np_{m,n}s^m$.
Show that $F(\alpha, 1)$ is the probability that an infective remains undetected during the whole incubation period.
Show that $D(\alpha)/F(\alpha, 1)$ is the expected length of infectious period for those who remain undetected during the incubation period.

(e) Let $G(t)$ be the p.g.f. of the number of individuals infected by a single infective. Show that

$$\begin{aligned} G(t) &= \sum_{n=0}^{\infty} \omega_n R^n(1-\gamma+\gamma t) \\ &= 1 - F(\alpha,1) + F(\alpha, \beta R(1-\gamma+\gamma t)) \\ &\quad +(1-\beta)\frac{F(\alpha,1) - F(\alpha, \beta R(1-\gamma+\gamma t))}{1-\beta R(1-\gamma+\gamma t)}\,. \end{aligned}$$

If $R'(1) = \gamma$ with $0 < \gamma < \infty$, then show that $G'(1) = \gamma r D(\alpha)$ in case $\beta = 1$, and, $= \gamma r \frac{\beta}{1-\beta}[F(\alpha,1) - F(\alpha,\beta)]$, in case $\beta < 1$.

(f) For $0 < x \leq 1$, put $h_1(x) = F(\alpha, x)$ and $h_2(x) = F(\alpha,1) - \frac{1}{\gamma r}\frac{1-x}{x}$.
Show that $h_1' > 0$, $h_2' > 0$, $h_1'' \geq 0$ and $h_2'' < 0$.
Show that $x = 1$ is a root of $h_1 = h_2$, and, if $h_1'(1) \leq h_2'(1)$, then this is the only root.
Show that if $h_1'(1) > h_2'(1)$ then there is one more root $x < 1$ of the equation $h_1 = h_2$.

(g) Consider the n-th generation of infected individuals as follows. $Z_0 = 1$ and for $n \geq 1$, Z_n = the number of persons infected by the Z_{n-1} persons of the $(n-1)$-th generation. Show that (Z_n) is a branching process with progeny generating function $G(t)$ as given above. Define $\beta^*(\alpha,\gamma)$ as the smallest positive root of

$$\gamma\, r\, x\, [F(\alpha, x) - F(\alpha, 1)] + 1 - x = 0\,.$$

Show that

$$P(\lim_n Z_n = 0) = 1 \quad \text{iff} \quad \beta \leq \beta^*(\alpha,\gamma)\,.$$

[There are generalizations to the case when there are N zones and there is mobility for people from one zone to another.]

7. The idea is to describe a simple mathematical model for muscle movements. This is due to S. W. Greenhouse.

Phenomenon: A muscle fibril consists of alternating thick and thin filaments. During shortening and stretching they slide along each other. While this happens, certain chemical interactions occur at the molecular level. There are sites on the thin filament. Concentrate on one site now. The site may be occupied by a molecule or may be vacant. There are positions on the thick filament which are alternately 'release' and 'load' positions. A release position can pick up a molecule and a load position can give a molecule. Suppose that 1,3,5,... are release positions and 2,4,6,... are load positions. Imagine a site now at 0 and sliding along the positions. Wish to know whether a site is filled or vacant at time n, and time is counted in units of positions crossed by the site. Note that if the site is filled and arrives at a load position, then nothing happens and it passes on to the next position. Similarly, if the site is vacant and arrives at a release position then nothing happens and it passes on to the next position.

In real life, positions on the thick filament are only release positions and moreover, a site may pickup a molecule at any point between two release positions. Further, during muscular contractions, the thin filament – and hence, the sites – move with varying velocity. The slower the speed, the greater the interaction and the parameters α and β given below change with n. This is a simplified treatment.

Mathematically, $X_0, X_1, \ldots$ is a sequence of random variables each taking values 0 (vacant site) and 1 (filled site). X_0 is the initial position and X_n its state after n interactions. The two matrices A and B given below represent the probabilities of transition from X_{2r} to X_{2r+1} and from X_{2r+1} to X_{2r+2} respectively.

$$A = \begin{pmatrix} 1 & 0 \\ \alpha & 1-\alpha \end{pmatrix} \quad \text{and} \quad B = \begin{pmatrix} 1-\beta & \beta \\ 0 & 1 \end{pmatrix}.$$

Here $0 < \alpha < 1$ and $0 < \beta < 1$.
For $i = 0, 1$ and $n \geq 1$, let $p_n^i = P(X_n = 1 \,|\, X_0 = i)$.
Show that for even integers n,

$$p_n^0 = \beta \, \frac{1 - [(1-\alpha)(1-\beta)]^{n/2}}{1 - (1-\alpha)(1-\beta)},$$

$$p_n^1 = [(1-\alpha)(1-\beta)]^{n/2} + \beta \, \frac{1 - [(1-\alpha)(1-\beta)]^{n/2}}{1 - (1-\alpha)(1-\beta)},$$

and for odd integers n,

$$p_n^0 = \beta \, (1-\alpha) \, \frac{1 - [(1-\alpha)(1-\beta)]^{(n-1)/2}}{1 - (1-\alpha)(1-\beta)},$$

$$p_n^1 = (1-\alpha)[(1-\alpha)(1-\beta)]^{(n-1)/2} + \beta\,(1-\alpha)\,\frac{1-[(1-\alpha)(1-\beta)]^{(n-1)/2}}{1-(1-\alpha)(1-\beta)}.$$

8. The idea is to discuss a model for Leukemia proposed by I. A. Chow.
Phenomenon: The disease starts with anaemia and thrombocytopenia. This is followed by the appearance of immature leukemic cells which replace the normal mature leukocutes. As the disease advances the number of immature granulocytes increases while the number of normal cells (called polymorphonuclear granulocytes, abbreviated as PMNG) decreases. This is attributed to the fact that the abnormal immature cells have a long intravascular life and capacity for mitotic subdivision compared with normal cells. The incapability of the abnormal immature cells in phagocytosis makes the patient very susceptible to infection or haemmorhage leading to death.
Notation: Let $\lambda > 0$ and $\mu > 0$ be two numbers. m = maximum number of PMNG one can have. $X(t)$= Number of PMNG at time t. $X(0)$= initial number of PMNG at time 0 , say = n_0.
Modelling Assumptions: The probability that PMNG will decrease by 1 during $(t, t+\Delta t)$ given that there are n at time t is $n\mu\Delta t + o(\Delta t)$.
The probability that PMNG will increase by one during $(t, t+\Delta t)$ given that there are n at time t is $(m-n)\lambda\Delta t + o(\Delta t)$.
The probability that PMNG will not undergo any change during $(t, t+\Delta t)$ given that there are n at time t is $1-[n\mu+(m-n)\lambda]\Delta t + o(\Delta t)$.
Define $p_n(t) = P(X_t = n \,|\, X_0 = n_0)$ for $0 \le n \le m$, and $= 0$ for other values of n. Set $G(t,s) = \sum_{n=0}^{m} p_n(t)s^n$, the p.g.f. of X_t.

(a) Show that

$$\frac{dp_n(t)}{dt} = \lambda\,(m+n-1)\,p_{n-1} + \mu\,(n+1)\,p_{n+1} - [n\mu + (m-n)\lambda]\,p_n\,.$$

(b) Show that

$$\frac{\partial}{\partial t}G(t,s) = (1-s)\,(\mu+\lambda s)\,\frac{\partial}{\partial s}G(t,s) - (1-s)\,\lambda\,m\,G(t,s).$$

$$G(0,s) \quad = \quad s^{n_0}\,.$$

(c) Show that

$$G(t,s) \quad = \quad \left[1 - \frac{\lambda+\mu e^{-(\lambda+\mu)t}}{\lambda+\mu} + \frac{\lambda+\mu e^{-(\lambda+\mu)t}}{\lambda+\mu}s\right]^{n_0}$$

$$\times \left[1 - \lambda\frac{1-e^{-(\lambda+\mu)t}}{\lambda+\mu} + \lambda\frac{1-e^{-(\lambda+\mu)t}}{\lambda+\mu}s\right]^{m-n_0}.$$

(d) Show that X_t is the sum of two independent random variables, say, X_t^1 and X_t^2, where

$$X_t^1 \sim B(n_0, \alpha) \quad \text{with} \quad \alpha = \frac{\lambda+\mu e^{-(\lambda+\mu)t}}{\lambda+\mu},$$

$$X_t^2 \sim B(m - n_0, \beta) \quad \text{with} \quad \beta = \lambda \frac{1 - e^{-(\lambda+\mu)t}}{\lambda + \mu}.$$

This can be interpreted as follows. The PMNG at time t is made up of two kinds. First, there are those of the initial n_0 which are still surviving. Second, there are those that are liberated at some time $\tau < t$ and are still surviving at time t.

(e) Show that

$$E(X_t) = n_0 e^{-(\lambda+\mu)t} + m\lambda \frac{1 - e^{-(\lambda+\mu)t}}{\lambda + \mu}.$$

$$V(X_t) = n_0 \frac{\mu - \lambda}{\mu + \lambda} e^{-(\lambda+\mu)t} \left[1 - e^{-(\lambda+\mu)t}\right] + m\lambda \frac{1 - e^{-(\lambda+\mu)t}}{\lambda + \mu} \frac{\mu + \lambda e^{-(\lambda+\mu)t}}{\lambda + \mu}.$$

(f) Show that

$$p_0(t) = \left[\mu \frac{1 - e^{-(\lambda+\mu)t}}{\mu + \lambda e^{-(\lambda+\mu)t}}\right]^{n_0} \left[\frac{\mu + \lambda e^{-(\lambda+\mu)t}}{\lambda + \mu}\right]^m.$$

(g) Show that

$$p_0(\infty) = \lim_{t\to\infty} p_0(t) = \left(\frac{\mu}{\lambda + \mu}\right)^m.$$

(h) Assume that the volume of blood, say v units, is large and also the PMNG at time t is large. What is usually observed is Y_t, the density of PMNG, that is, the number of PMNG in unit volume of blood, at time t. Theoretically speaking, any of the X_t cells has a chance $1/v$ of appearing in the unit volume taken for the PMNG count. So it is believed that, given $X_t = n$, Y_t is Poisson with parameter n/v. In other words,

$$P(Y_t = n' \mid X_t = n) = \frac{(n/v)^{n'} e^{-(n/v)}}{n'!} \quad \text{for} \quad n' \geq 0.$$

Show that the conditional p.g.f. of Y_t given $X_t = n$ is $e^{-n(1-s)/v}$.

(i) If $H(t, s)$ is the unconditional p.g.f. of Y_t, then show that

$$H(t, s) = H_1(t, s) \cdot H_2(t, s),$$

where

$$H_1(t, s) = \left[1 - \frac{\lambda + \mu e^{-(\lambda+\mu)t}}{\lambda + \mu} + \frac{\lambda + \mu e^{-(\lambda+\mu)t}}{\lambda + \mu} e^{-(1-s)/v}\right]^{n_0},$$

$$H_2(t, s) = \left[1 - \lambda \frac{1 - e^{-(\lambda+\mu)t}}{\lambda + \mu} + \lambda \frac{1 - e^{-(\lambda+\mu)t}}{\lambda + \mu} e^{-(1-s)/v}\right]^{m-n_0}.$$

Conclude that Y, just like X, is the sum of two independent random variables.

(j) Show that

$$E(Y_t) = (n_0/v)\, e^{-(\lambda+\mu)t} + (m/v)\,\lambda\, \frac{1-e^{-(\lambda+\mu)t}}{\lambda+\mu},$$

$$\begin{aligned} V(Y_t) &= (n_0/v)\left[1 - (m/v)\,\frac{\lambda(1-e^{-(\lambda+\mu)t}}{\lambda+\mu}\right] \\ &\quad +(1/v)\,\frac{\mu-\lambda}{\mu+\lambda} e^{-(\lambda+\mu)t}\left[1-e^{-(\lambda+\mu)t}\right] \\ &\quad +(m/v)\,\lambda\,\frac{1-e^{-(\lambda+\mu)t}}{\lambda+\mu}\left[1 + (1/v)\frac{\mu+\lambda e^{-(\lambda+\mu)t}}{\lambda+\mu}\right]. \end{aligned}$$

(k) The parts (i) and (j) above are useful in estimating the parameters and making predictions. Chow considers these also.
In practice (n_0/v) and (m/v) are not observable and they are replaced by n_0', the initial PMNG density, and m', upper limit of the observed PMNG density.
If the patient is under treatment then the chances of a PMNG liberation from bone-marrow into the blood stream depends on the time instant t itself, apart from depending on the actual number at that time. In other words, λ is not a constant but a function of t. Similarly μ also is a function of t. These are denoted by $\lambda(t)$ and $\mu(t)$ respectively.
From now on this is what is assumed and m, X_0, X_t, n_0, p_n and $G(t,s)$ are as defined earlier.

(l) Argue that G satisfies a similar equation as earlier except that the numbers λ and μ are now functions of t.

(m) Show that

$$G(t,s) = [G_1(t,s)]^{n_o}\ [G_2(t,s)]^{m-n_0},$$

where

$$\begin{aligned} G_1(t,s) &= 1 - \left\{1 + \int_0^t \lambda(\tau)\, e^{R(\tau)}\, d\tau\right\} e^{-R(t)} \\ &\quad + \left\{1 + \int_0^t \lambda(\tau)\, e^{R(\tau)}\, d\tau\right\} e^{-R(t)}\, s, \end{aligned}$$

$$G_2(t,s) = 1 - \int_0^t \lambda(\tau)\, e^{R(\tau)}\, d\tau\, e^{-R(t)} + \int_0^t \lambda(\tau)\, e^{R(\tau)}\, d\tau\, e^{-R(t)}\, s$$

and

$$R(t) = \int_0^t [\lambda(\tau) + \mu(\tau)]\, d\tau\,.$$

(n) Show that

$$E(X_t) = n_0\, e^{-R(t)} + m\, e^{-R(t)} \int_0^t \lambda(\tau)\, e^{R(\tau)}\, d\tau$$

and

$$V(X_t) = n_0 e^{-R(t)} \left[1 - e^{-R(t)}\{1 + 2\int_0^t \lambda(\tau)\, e^{R(\tau)}\, d\tau\}\right] + m\, e^{-R(t)} \int_0^t \lambda(\tau)\, e^{R(\tau)}\, d\tau \left[1 - e^{-R(t)} \int_0^t \lambda(\tau)\, e^{R(\tau)}\, d\tau\right].$$

4.7 References/Supplementary Readings

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