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Toward a New Conceptualization and Operationalization of Risk Perception Within the Genetic Counseling Domain

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The purpose of this paper is twofold. First, it provides an historical overview of studies of risk, risk perception, and decision making under risk within the genetic counseling domain. Second, it proposes an alternative conceptualization and operationalization for the study of risk perception. The conceptualization involves probability, adversity, incompleteness, and ambiguity. Prior studies of risk perception focus on the recurrence risk and operationalize risk perception by asking for interpretations of the magnitude of the probability of the outcome. Their focus is on the probability of a particular outcome. We formulate the problem in terms of a gamble and suggest that risk perception be operationalized in terms of the riskiness of the gamble. Our focus is on the riskiness of a decision option which entails two or more outcomes.

KEY WORDS: risk perception; decision making; risk; uncertainty; genetic counseling.

Study the past, if you would divine the future.
—Confucius

INTRODUCTION

Risk is almost universally conceptualized as a combination of probability and something adverse, unpleasant, or dangerous (Lowrance, 1976;

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Fishburn, 1984; Faden *et al.*, 1987; Vlek, 1987). In fact, the word *risk* denotes the possibility or probability of something adverse occurring (*The American Heritage Dictionary,* 1980). In many fields, including genetic counseling, risk is measured numerically, in terms of a probability (Fullwood and Hall, 1988; Edwards, 1989). The measure of risk in terms of a probability is a particular operationalization of the concept of risk. Since this measure must reflect the concept of risk, the probability implicitly refers to some adverse event, e.g., birth of child with genetic disorder or number of fatalities due to nuclear core meltdown. Within the genetic counseling domain the probability-based measure of risk is termed the occurrence or recurrence risk (Fraser, 1970). For simplification we will hereafter only refer to recurrence risks.

The determination and provision of recurrence risks is considered an essential element of genetic counseling (Pearn, 1973; Lippman-Hand and Fraser, 1979a). The importance of the recurrence risk stems from the impact it is believed to have on decision making, e.g., reproductive decision making (Lippman-Hand and Fraser, 1979a; Kessler and Levine, 1987). Since (1) genetic counseling is identified, in part, with the provision of recurrence risks, and (2) the recurrence risk is thought to be an important variable in decision making, many studies involving the recurrence risk have simultaneously been studies of decision making under risk and studies of the impact or effectiveness of genetic counseling.

Roberts' study (1962) was influential in defining the dual role played by the recurrence risk: as a means to study both decision making and the impact and effectiveness of genetic counseling. Specifically, he evaluated the relationship between the magnitude of the recurrence risk and subsequent reproductive decisions and used his findings to draw conclusions about the impact and effectiveness of genetic counseling. This study was later reported in full in Carter *et al.* (1971), a paper credited as the first published large-scale follow-up study of the impact and effectiveness of genetic counseling (Reed, 1977). This was followed by a number of other studies that attempted to evaluate genetic counseling from the relationship between the magnitude of the recurrence risk and subsequent reproductive decisions (Emery *et al.*, 1972; Leonard *et al.*, 1972; Sultz *et al.*, 1972; Emery *et al.*, 1973; Ives *et al.*, 1973; Hsia, 1974; Reynolds *et al.*, 1974; Hutton and Thompson, 1976; Godmilow and Hirschhorn, 1977; Emery *et al.*, 1979). All of these studies employed an objective characterization of the magnitude of a recurrence risk. The cornerstone of this characterization was the belief that the magnitude of the recurrence risk had an inherent meaning of high, medium, or low, which was invariant across observers and diseases.

Other studies, however, employed a subjective characterization of the magnitude of a recurrence risk. The term subjective replaces objective to

emphasize that the interpretation of the magnitude of a recurrence risk as high, medium, or low lies within the observer as opposed to the number. There are two types of studies which involve the subjective interpretation of a recurrence risk, or *risk perception,* as it is termed. The first simply replaces the objective with the subjective interpretation to study the relationship between the recurrence risk and subsequent decisions (e.g., Frets *et al.*, 1990a, b). Thus, risk perception, rather than the numeric recurrence risk, is used as a means to evaluate the variability of decisions and the impact and effectiveness of genetic counseling.

The second type of study focuses on explaining the variability noted in the subjective interpretations given to recurrence risks (Ekwo *et al.*, 1985; Chase *et al.*, 1986; Wertz *et al.*, 1986; Kessler and Levine, 1987; Shiloh and Sagi, 1989; Denayer *et al.*, 1992). In this context, risk perception, rather than the decision made, is the variable under study. These studies are conducted more for identifying factors and exploring underlying cognitive processes that result in an interpretation of a recurrence risk than for evaluating genetic counseling (although the ultimate purpose of understanding ways to improve or enhance genetic counseling remains). This branch of research also occasionally evaluates the role of risk perception in the decision making process (Lippman-Hand and Fraser 1979b, d).

The unchanging core of all of the cited studies is the use of the numeric measure of risk, the recurrence risk, as the primary means to measure the magnitude of the risk faced by a couple. However, the objective and subjective characterizations of the magnitude of the recurrence risk represent fundamentally different perspectives: whether the interpretation is inherent to the number or uniquely defined by the person, respectively. Interestingly, these characterizations fall into two relatively distinct periods: 1962–1979 (objective) and 1979–current (subjective). A closer examination of these periods reveals not only differences in the characterization of the magnitude of the recurrence risk, but also in the role ascribed to burden of disease in evaluating decisions, and the desired impact or effect of genetic counseling on decision making. When evaluated from an historical perspective, the changes noted from the former to the latter period reflect an evolution in the goals of genetic counseling, which in turn impacted on the study of risk perception and decision making under risk.

Although studies of the underlying determinants of risk perception provide insight into the mental processes involved in interpreting the magnitude of recurrence risks, we believe that the conceptualization and operationalization of risk perception are fundamentally limited. A goal of this paper is to begin to articulate an alternative formulation for the study of risk perception. To this end we present an historical account of studies of risk perception and decision making under risk within the genetic counsel-

ing domain. This perspective will provide a deeper understanding for the way in which these topics were formulated and studied, for their evolution over time, and for an alternative formulation for studying risk perception.

1962–1979

Between 1962 and 1979 the objective characterization of the magnitude of the recurrence risk provided the means to capture the magnitude of "the risk," or the amount of risk entailed in a decision (Roberts, 1962; Carter *et al.*, 1971; Emery *et al.*, 1972; Leonard *et al.*, 1972; Sultz *et al.*, 1972; Emery *et al.*, 1973; Ives *et al.*, 1973; Hsia, 1974; Reynolds *et al.*, 1974; Hutton and Thompson, 1976; Emery, 1977; Godmilow and Hirschhorn, 1977; Emery *et al.*, 1979). Recurrence risks were classified as high, medium, or low, and the classification was viewed as an inherent and immutable characteristic of each recurrence risk. The assumption of invariance across observers and diseases was not questioned.

With some minor variation between researchers, high and low risks were generally those for which the risk of recurrence was greater than and less than 10%, respectively. Roberts (1962) was the first to employ this particular classification in an empirical study, however, it was suggested at least as early as 1957 at the Hereditary Counseling Symposium by Dr. William J. Schull (later published in the *Eugenics Quarterly,* 1958). There was an occasional comment about the apparent arbitrariness of this particular classification which invoked a 10% cut-off point between low and high risks (Emery *et al.*, 1972), but alternative classifications were not suggested nor employed in studies.

The fact that these researchers recruited the numeric measure of risk as a means to assess the magnitude of "the risk" of having a child with a genetic disorder is consistent with findings by Slovic *et al.* (1979) that experts, such as scientists, rely on the mathematical, statistical definition of risk as probability when rating the magnitude of a risk. Moreover, large probabilities are equated with high risks and small probabilities are equated with low risks. The direct correlation between size of probability and size of risk, regardless of what the probability is referring to, is consistent with an objective characterization of the magnitude of the recurrence risk. Although Pearn discussed the concept of a subjective interpretation of the recurrence risk in 1973, the objective characterization dominated during this period and resulted in a failure to recognize that subjects might legitimately create different classifications (for examples of this phenomenon see: Ives *et al.*, 1973; Hsia, 1974; Reynolds *et al.*, 1974; Godmilow and Hirschhorn, 1977).

It was generally accepted during this period that genetic counseling should promote rational decision making (Lippman-Hand and Fraser, 1979c, d; Kessler, 1980). Rational decisions were understood among professionals to be those based on recurrence risks and the consensus in the literature was that high recurrence risks would deter further child-bearing while low recurrence risks would encourage it (Roberts, 1962; Carter *et al.*, 1971; Lippman-Hand and Fraser, 1979b; Lubs, 1979; Sorenson, 1993). The desired impact of genetic counseling, coupled with the prevailing belief that couples *would* make reproductive decisions based on the recurrence risk (Sorenson, 1993), provided one means to measure the impact and effectiveness of genetic counseling, i.e., in terms of the rationality or irrationality of subsequent reproductive decisions (Lippman-Hand and Fraser, 1979c).

These studies were not entirely limited to the study of the impact of recurrence risks on decision making. Early on in this period, burden of the disease was noted to have an impact on reproductive decisions. Leonard *et al.* (1972) concluded that burden of disease was more important to decision making than the recurrence risk. This conclusion was based on the finding that the majority of respondents identified the disease as the primary reason for their reproductive decision. This study was influential in separating the recurrence risk of a genetic disease from the burden of that disease. Its effect on the studies of this period was to establish the recurrence risk and burden of disease as separate variables, each having a direct impact on the decision. Figure 1 depicts the structure and underlying assumptions of these studies.

Most researchers during this time either had an intuitive grasp of the potential importance of burden on decision making or else paid heed to the conclusions of Leonard *et al.* (1972) such that they attempted to control for burden by evaluating a single disease entity (Emery *et al.*, 1972; Hutton and Thompson, 1976), including only serious diseases (Carter *et al.*, 1971; Emery *et al.*, 1973, 1979), or by comparing against a control diseased group (Sultz *et al.*, 1972). A few researchers did none of the above and enlisted the notion of burden in *post hoc* explanations of their results (Ives *et al.*, 1973; Hsia, 1974; Reynolds *et al.*, 1974). Controlling for burden enabled the focus to remain on the relationship between the recurrence risk and the reproductive decision. This formulation of the decision making model reveals the researchers' assumption "that the probability of having an affected child [would] have a predictable influence on reproductive choices, with risk and deterrence from further childbearing being directly related" (Lippman-Hand and Fraser, 1979d, pp. 328-329).

However, the studies of this period were unable to demonstrate a clear relationship between the magnitude of the recurrence risk and subsequent reproductive decisions. This was one of several findings that undermined the prevailing belief that couples would make rational reproductive decisions

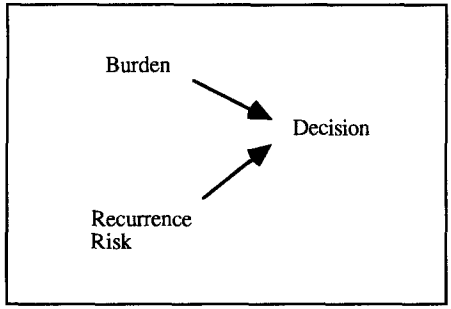

Fig. 1. Structure and assumptions of studies of decision making under risk (1962–1979).

when made aware of the recurrence risk, calling this goal of genetic counseling into question. Among other things, the apparent unimportance of the recurrence risk on decision making, the reluctance of counselees to use prevention technology, the discrepancy between counselees' intentions and actions, and an increased awareness of the psychosocial ramifications of genetic disease on counselees and their families helped to reshape the definition and goals of genetic counseling (Kessler, 1980). By the end of the 1970s the belief that couples should make the *best* decision for their situation fully supplanted the belief that they should make *rational* decisions. Genetic counseling evolved into a service that included an emphasis on education and supportive counseling, a bi-directional communication process, and a greater awareness of the psychosocial aspects of genetic disease and decisions related to it (Kessler, 1980). This evolution reflected a growing appreciation of the many diverse issues raised in genetic counseling and the importance of the counselees' point of view with regard to these issues (Levine, 1979). Subsequent studies of the impact of genetic counseling on decision making began to incorporate the counselees' point of view.

1979–Current

The year 1979 witnessed the incorporation of the counselees' perspective in the study of the recurrence risk and reproductive decision making, as exemplified by Lippman-Hand and Fraser's (1979a–d) four seminal articles. These authors challenged the prescriptive assumptions of the rational decision making model and proposed an alternative decision making model based on a descriptive study of *how* people arrived at reproductive

decisions. The intent of the study was to identify cognitive strategies, or heuristics, employed by at risk couples to arrive at reproductive decisions.

Based on a content analysis of semi-structured interviews with individuals or couples who had received genetic counseling, the authors concluded, among other things, that counselees interpreted the recurrence risk in semiquantitative terms as being high, medium, or low. Lippman-Hand and Fraser (1979b) suggested that this conversion represented a cognitive process whereby rate information, "the frequency with which something happens" (p. 56), was converted into risks — "their interpretation of this information and attitudes to it" (p. 56). However, it appeared that an interpretation of rate information as high, medium, or low was not useful for making a decision about whether or not to conceive a pregnancy.

The authors noted that subjects also tended to interpret the recurrence risk in binary form, "it either will happen or will not happen" (Lippman-Hand and Fraser, 1979b, p. 51). They suggested that this binary interpretation of the recurrence risk was the result of a heuristic employed to simplify complicated probabilistic information so that the decision making process could move forward. This simplification allowed subjects to focus on the consequences of the undesired outcome through the construction of worst case scenarios. Subjects would then evaluate their response to that scenario. Tolerance or intolerance of the scenario was usually associated with a decision to proceed with, or stop, procreation, respectively. Binary interpretation of the recurrence risk and scenario construction were proposed as cognitive strategies used for decision-making purposes.

Lippman-Hand and Fraser (1979b) described three basic categories of consequences on which subjects focused in their decision making deliberations. The first category, *ambiguity,* referred to the lack of a one-to-one correspondence between a diagnosis and the level of health and functioning it would permit. In other words, the variable prognosis inherent to many genetic diseases precluded perfect knowledge of what an affected child would be like if born. The second category involved the burden associated with the *need* to make a decision. Prior to the birth of their affected child most couples did not spend a great deal of time contemplating a desired pregnancy. However knowledge that it could happen again carried with it the burden of decision making. The third category was characterized as the *desire to normalize* their status as parents. This was particularly prominent among couples whose first born was affected.

The post-genetic counseling design of the study by Lippman-Hand and Fraser (1979a–d) precluded an analysis of the effect of genetic counseling on decision making or risk perception. However in that same year, Lubs (1979) evaluated the impact of genetic counseling on the subjective

interpretations of the recurrence risk, or risk perception. The before-after design served to demonstrate that genetic counseling could alter risk perception.

A different aspect of the recurrence risk, the extent to which it is unequivocally known for an observable abnormality, was explored in 1979 by Black. Interestingly, Black used the term *ambiguity* to describe the situation when a counselee is provided more than one recurrence risk because the exact etiology of an abnormality, such as mental retardation, is unknown. Although this is a different use of the term ambiguity than that employed by Lippman-Hand and Fraser (1979b), in both cases it reflects the growing appreciation for the complexities and limitations of genetics knowledge for use in reproductive decision making.

In many ways the four articles by Lippman-Hand and Fraser (1979a–d) revolutionized the study of the recurrence risk and decision making within the genetic counseling domain. Most importantly they legitimized the counselees' perspective by focusing on the process used to make a decision rather than on what decisions should be made. In doing this, they introduced information processing theories of cognitive psychology as a means to identify and understand the mental strategies that people use to process information when formulating judgments and making decisions. Finally, they legitimized the counselee-based interpretation of the recurrence risk. This has since become viewed as risk *perception,* reflecting the change from the objective to the subjective interpretation of the recurrence risk. Many subsequent studies of decision making have incorporated risk perception as an explanatory variable (e.g., Frets *et al.*, 1990a, b) and Frets and Niermeijer (1990) concluded that the counselees' interpretation of the magnitude of the recurrence risk explains more variability in subsequent reproductive decision making than does its actual magnitude.

The demonstration that the same recurrence risk was interpreted in very different ways (e.g., Swerts, 1987; Somer *et al.*, 1988) prompted questions about the underlying determinants of an individual's interpretation of a recurrence risk. This explains the emergence of empirical studies of risk perception during this period (Ekwo *et al.*, 1985; Chase *et al.*, 1986; Wertz *et al.*, 1986; Kessler and Levine, 1987; Shiloh and Sagi, 1989; Denayer *et al.*, 1992). Despite variations in these studies, they all asked subjects for their own interpretation of the recurrence risk number. Researchers regarded the counselee-based interpretation of a probability as *the* way to measure risk perception and explored reasons for the variability in the interpretations. These studies can be classified into three categories: (1) those that focused on identifying background variables to explain differences in risk perception (Ekwo *et al.*, 1985; Denayer *et al.*, 1992), (2) those that focused on framing effects or cognitive heuristics to explain dif-

ferences in risk perception (Kessler and Levine, 1987; Shiloh and Sagi, 1989), and (3) those that did both (Chase *et al.*, 1986; Wertz *et al.*, 1986). Figure 2 depicts the structure and underlying assumptions of these studies.

Note that four out of the six empirical studies of risk perception documented in this article (Chase *et al.*, 1986; Wertz *et al.*, 1986; Kessler and Levine, 1987; Shiloh and Sagi, 1989) enlisted information-processing theories to identify the strategies used by people to formulate their judgments of the magnitude of the recurrence risk. This approach was probably influenced by the work of Lippman-Hand and Fraser (1979a–d) and reflects a heightened interest in understanding how counselees process information (e.g., recurrence risk) provided during genetic counseling, and how such processing affects or explains subsequent judgments and decisions. Thus, the goal of informed decision making influenced the choice of theories enlisted for explanation and understanding of the impact and effectiveness of genetic counseling.

Ekwo *et al.* (1985) focused on the relationship between the objective recurrence risk, as reported by the subjects, and its subjective interpretation. They found no demonstrable relationship between (in)accuracy of recall and risk perception. Other variables, such as age and a ranking of the perceived burden of caring for children with five hypothetical congenital malformations were also found to be unrelated to subjective estimates of risk. Similarly, Denayer *et al.* (1992) found no demonstrable relationship between a set of background variables (age, sex, educational level, religious convictions, number of children, wanting more children, health situation of the proband, frequency of contacts with proband, and knowledge about the genetic transmission) and risk perception.

Wertz *et al.* (1986) explored the effects of the cognitive heuristics of availability, representativeness, and anchoring (described by Tversky and Kahneman, 1974), on counselees' judgments of the likelihood to have a child with a genetic disorder. Wertz *et al.*, (1986) defined availability as

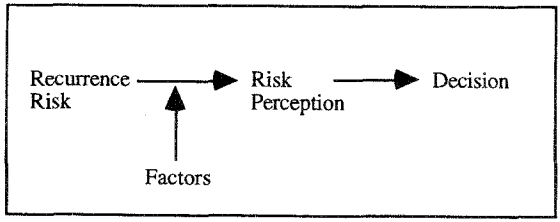

Fig. 2. Structure and assumptions of studies of risk perception (1979–current).

"the ease with which instances of the risked event can be brought to mind" (p. 255), representativeness as "the degree to which an individual outcome (in the context of genetic counseling, a particular child with a particular birth defect) is regarded as representative or stereotypical of all outcomes" (pp. 254-255), and anchoring as occurring "when the person has a starting point or partial computation on which to base an interpretation of risk, for example, a prior belief that a risk is high or low" (p. 255). Although five variables explained 28.7% of the variation of pessimistic interpretations of the recurrence risk, the numeric recurrence risk accounted for the majority of this variation. The presence of an affected child and the degree of intellectual impairment of that child also explained a small amount of the variation. Describing these latter two variables as examples of the availability heuristic, the authors concluded that this heuristic played a role in determining risk perception. They also concluded that counselees employed the anchoring heuristic when interpreting the recurrence risk after genetic counseling.

Chase *et al.* (1986), Kessler and Levine (1987), and Shiloh and Sagi (1989) evaluated information processing in terms of framing effects. They recognized that probabilistic information can be presented, or framed, in various ways and that subjective interpretations of probabilities might vary with presentation format. Kessler and Levine (1987) and Shiloh and Sagi (1989) documented changes in the subjective interpretations of recurrence risks with different presentation formats. Kessler and Levine (1987) went one step further and also attempted to document and explain these effects via specific cognitive information processing strategies. Chase *et al.* (1986) did not note changes in risk perception with changes in presentation format; they also found no relationship between an opinion of the justifiability of abortion and risk perception.

In the studies of this more recent period, burden of disease was viewed as a variable that might affect and mold risk perception, thus as a potential determinant of risk perception. Ekwo *et al.* (1985) and Wertz *et al.* (1986) explicitly assessed burden of disease from the counselee's and expert's perspective, respectively, and evaluated the relationship between burden and interpretation of the recurrence risk. As noted above, Ekwo *et al.* (1985) did not demonstrate a relationship between these two variables. However, Wertz *et al.* (1986) found that burden, as measured by degree of intellectual impairment, accounted for a very small amount (less than 1%) of the explained variation in risk perception. It might be argued that Denayer *et al.* (1992) evaluated burden as the health situation of the proband; no relationship was demonstrated between this variable and risk perception. The studies of framing effects (Chase *et al.*, 1985; Kessler and Levine, 1987; Shiloh and Sagi, 1989) attempted to control for burden in

order to minimize its effect on risk perception so that the impact of changes in the framing of probabilistic information on risk perception could be assessed.

The risk perception studies that emerged during this period may be summarized in the following way: (1) they equate risk to probability, (2) they regard risk perception as the interpretation of that probability, (3) they involve a search for variables (e.g., numeric recurrence risk, burden of disease, age) that might influence or explain the interpretation of the recurrence risk, and/or (4) they regard information-processing heuristics as the basis for explanations of how these variables affect risk perception. Taken together, the conclusions of these studies are mixed. However, it might be generally concluded that the studies of framing of probabilistic information have been more successful in explaining the variability in risk perception than the studies assessing its relationship with other variables.

We posit that there is a fundamental limitation of all of these risk perception studies. This limitation is the equation of risk with probability, resulting in the view that risk perception is how people interpret the recurrence risk. This view is highly limiting when we recall that the universal conceptualization of risk involves both probability and adversity. We suggest that the findings of these empirical studies of risk perception support the idea that more research needs to be done to understand how counselees interpret *probabilities.* However they do not provide evidence that *risk perception* is what is actually being studied.

TOWARD A NEW CONCEPTUALIZATION AND OPERATIONALIZATION OF RISK PERCEPTION

The purpose of this section is to begin to articulate an alternative formulation for the study of risk perception. Since the study of risk perception presupposes a decision situation, the way in which risk perception is studied depends a great deal on the general model of decision making. We will discuss three basic ingredients to the study of risk perception: (1) the decision making model, (2) the conceptualization of risk perception, and (3) the factors contributing to risk perception. These three points are addressed below.

Decision Making Model. In the earliest studies the recurrence risk was invoked as the primary means to study decision making. As depicted in Fig. 1, these studies focused on the relationship between the probability of the implicitly adverse outcome and a decision. This formulation naturally gave rise to studies of risk perception that focused on the subjective interpretations of the recurrence risk and reasons for these interpretations.

An alternative formulation views decision making in terms of a gamble (e.g., von Neumann and Morgenstern, 1947; Kahneman and Tversky, 1979; Lopes, 1987) because the actual outcome of a decision is unknown at the time the decision is made. For example, many couples seen by genetic counselors ultimately must choose between conceiving a pregnancy or not conceiving a pregnancy. The decision to conceive a pregnancy could result either in a child without an abnormality or in a child with an abnormality; however, at the moment of decision making, the actual outcome is unknown. The alternative course of action, the decision not to conceive, maintains the status quo and provides a sure outcome (you know that you will not have a child).

In this alternative formulation, risk perception is studied with respect to a particular decision option (see Fig. 3), rather than in terms of the interpretation of the probability of a particular outcome (see Fig. 4) as previously formulated. Perceived riskiness of a gamble addresses the issue of being at risk for having a child with an abnormality, the existential state identified by Lippman-Hand and Fraser (1979b) as the key issue to be resolved in decision making. This formulation for the study of risk perception has recently been theoretically and empirically explored by Weber (1988) and Weber and Bottom (1989) using simple gambles with monetary outcomes.

Conceptualization of Risk Perception. In order to conceptualize risk perception we must first return to the concept of risk. Recall that risk is conceptualized as a combination of probability and adversity. Thus, it seems plausible that risk *perception* should at least involve these two aspects. It is important to emphasize here that probability is actually a specific operationalization of the concept of uncertainty. Thus it is more accurate to conceptualize risk as a combination of uncertainty and adversity (Kaplan and Garrick, 1981), the perception of which is believed to influence sub-

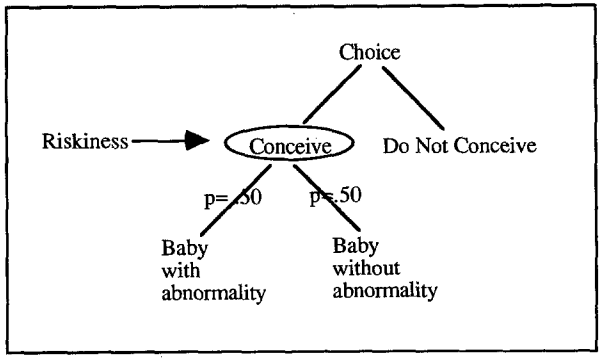

Fig. 3. Alternative formulation for studying risk perception.

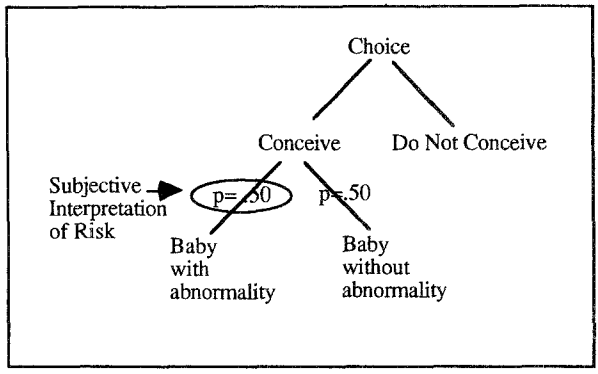

Fig. 4. Relationship between the way risk perception has been studied and our proposed formulation.

sequent decisions (see Fig. 5). For all intents and purposes probability is *the* measure of uncertainty in use today, so our discussion will honor this slightly limited view of uncertainty. However the limitations of this measure will become apparent in subsequent discussion.

Probability is inherent to the transmission of a genetic disorder from one generation to another; thus this aspect of risk, so long as it is recognized, is always present. The second aspect of risk, adversity, is *not* so readily apparent. Unlike probability, adversity is not inherent to a genetic disorder. Someone must attach meaning to a genetic disorder and form a judgment of adversity (Pearn, 1973). This judgment may arise from experience with, and information about, the genetic disorder, and the judgment process may be complicated by the variable expression of a disease. Therefore, in order for the possibility of having a baby with a genetic disorder to be perceived as *risky* (as opposed to *"chancy"*), someone must determine that this event is adverse. The terms risk and chance are differentiated on the basis of a judgment of adversity or burden of the outcome. When we

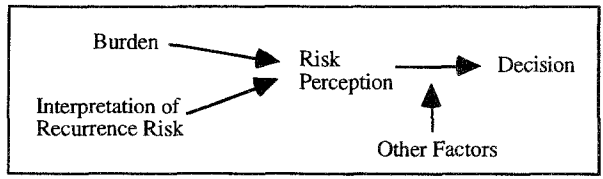

Fig. 5. Proposed structure of components of risk perception and their relationship to decision making.

say that there is a 25% probability, or chance, of something occurring we usually mean to imply non-adversity (or at least neutrality); when we say that there is a 25% risk of something occurring we usually imply that the outcome is adverse. Thus the vocabulary we choose to think about or communicate that probability reflects our beliefs about what constitutes adversity.

Because the concept of risk is dependent on a judgment of adversity, it will always be based partly on perception. However the individual(s) whose judgment(s) regarding the presence or absence of adversity are *relevant* are those for whom the decision is relevant. Thus, our beliefs about who assumes the role of the key decision maker dictates whose judgment of adversity is relevant.

One could argue as well that risk belongs entirely to the realm of perception. This view arises from the more generalized conceptualization of risk as a combination of uncertainty and adversity. The potential for the recurrence of an event may not always be measurable with a uniquely specifiable probability (cf. Black, 1979), however the uncertainty of the situation remains, and if it is also judged as adverse, the situation may still be considered risky.

Risk is either present or absent, regardless of whether it resides partly or entirely in the realm of perception. It is composed of the presence and awareness of a probability or uncertainty and a judgment of adversity. Risk perception always resides totally in the realm of perception and it serves as a measure of gradation, of how much risk is present. Risk perception is composed of an interpretation of the magnitude of the probability and a judgment of the level of adversity. At the least, a study of risk perception should address both an interpretation of the magnitude of the probability of occurrence of an outcome and a judgment of the level of its adversity.

Let us look closely at the difference between our view of risk and risk perception and the more traditional view within the genetic counseling literature. We suggest that risk is like a 0-1 variable; it either is absent or present, respectively. Risk will be absent (take on a 0 measure) if: (1) there is no uncertainty associated with the outcome of a decision, in other words, probability equal 0 or 1 for that outcome, or (2) the outcome of a decision is not judged to be adverse. Risk will be present (take on a 1 measure) when there is uncertainty associated with the outcome of a decision *and* that outcome is judged to be adverse. The *presence* of both uncertainty and adversity is the relevant and necessary aspect of our view of risk, not the *amount.* Risk perception serves as the individual-specific measure of the amount of risk present in a decision situation.

On the other hand, the view of risk generally employed within the genetic counseling literature is a measure of both presence *and* amount.

This is because the concept of risk has been reduced to a probability and as such it is able to provide a sense of whether and how much risk is present. As a probability, risk is a continuous variable which ranges from 0 to 1. A value of 0 suggests that no risk is present, values increasing from 0 to 0.999, suggest that risk is present and increasing in magnitude, and a value of 1 suggests that risk is absolute. Value judgments of the outcome are implicit within the term risk. We do not favor this view of risk because it conflates probability with risk and mutes the value judgments associated with the outcome. Our view, however, maintains the integrity of the concept of risk as a combination of probability and adversity.

Factors Contributing to Risk Perception. We postulate that several factors contribute to the perceived riskiness of a gamble. Drawing directly from our conceptualization of risk perception, we postulate that both the interpretation of the magnitude of the probability and a judgment of the level of adversity associated with having a child with an abnormality drive the overall perceived riskiness of the gamble. However, logically speaking, the judgment of the level of adversity associated with having a child with an abnormality is really the result of judgments of the adversity of a myriad of consequences, or potential events, which make up that abnormality. The actual consequences will differ with each genetic disorder, although general categories such as medical, psychosocial, and financial will generally be common to many genetic disorders. Each consequence may have a different influence on the overall judgment of adversity. Therefore, it is important to organize the consequences associated with an outcome as a hierarchy of events. We call this hierarchy an event structure and an example event structure for achondroplasia is shown in Fig. 6.

The primary event in the event structure depicted in Fig. 6 is the birth of a child with achondroplasia; consequences, or secondary events,

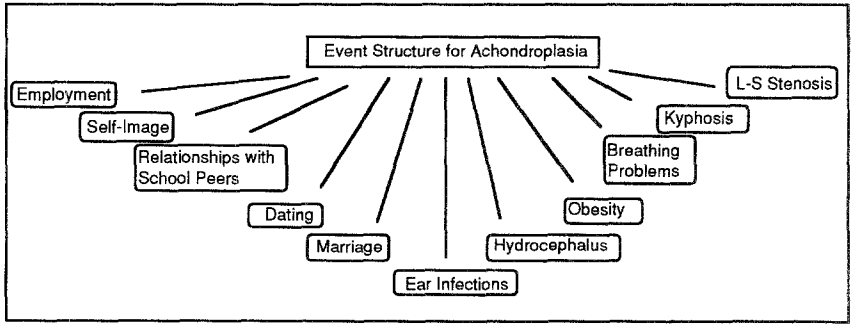

Fig. 6. Example of an event structure for achondroplasia.

emanate from the primary event. However, as noted in Lippman-Hand and Fraser (1979b), there is generally no one-to-one correspondence between a particular disorder and the actual manifestations of the consequences. This suggests that each consequence is also associated with some probability of occurrence. We postulate that the interpretations of the adversities and the magnitudes of the probabilities of occurrence of each of these consequences affect the perceived riskiness of the gamble.

Drawing from cognitive psychology, it is possible that one's knowledge base plays a role in interpreting the magnitudes of the various probabilities and formulating judgments of adversity. Knowledge base refers to the information, personal experiences, and beliefs that one has at one's disposal for problem-solving, or decision-making activities (Klayman and Schoemaker, 1993). People have different knowledge bases and value systems and consequently may perceive the same event structure for a specific genetics problem situation differently. Lippman-Hand and Fraser (1979b) recognized that the knowledge base provided the foundation for formulating the scenarios to be used for decision-making purposes; Ekwo *et al.* (1985) also recognized that perceptions are formed from the knowledge base.

If judgments are based on one's knowledge base, then the amount and consistency of the information, personal experiences, and beliefs in that knowledge base become of interest. At issue is the amount of "uncertainty" associated with interpretations of the magnitude of probabilities and judgments of adversity of the events in the event structure and the effect that this uncertainty has on an individual's perceived riskiness of the gamble. We postulate that there are at least two different, but related sources of this uncertainty: *incompleteness* and *ambiguity.*

Incompleteness is elicited in two situations. The first is with respect to the comprehensiveness of the event structure. The less knowledge one has about a primary event (the birth of a child with an abnormality), the less-well delineated and therefore complete, the event structure will be. The interpretation of an incomplete event structure, implying unknown events with unknown probabilities and adversities, may affect the judgment of riskiness of the gamble. Second, incompleteness refers to how *much* is perceived to be known about the probabilities and descriptions of those events that are known.

Ambiguity refers to how *well* the probabilities and descriptions of the known events are perceived to be captured and measured and how well these events are consistently represented. Ambiguity arises from sources such as conflict, unreliability, lack of clarity, or vagueness of the probabilities and descriptions of known events for a particular decision situation. One source of ambiguity is the lack of understanding or adequacy of the

model used to depict the generation of the event. This is the ambiguity described by Black (1979). Another source is some perceived imprecision of the measures used to capture the probability or the description of the event (inaccuracy, unreliability); yet another source is conflicting estimates of the probability or conflicting descriptions of the event. When taken in the context of the primary event, this latter source of ambiguity reflects that described by Lippman-Hand and Fraser (1979b).

Finally, the formulation of the decision in terms of a gamble, as depicted in Fig. 3, reveals the potential importance and possible impact of the alternative outcome of the gamble, a baby without an abnormality, on risk perception. Perceived riskiness of the gamble may represent a composite of perceptions of the previously discussed dimensions (probability, adversity, incompleteness, and ambiguity) of both possible outcomes.

Although there are many key differences between our formulation for the study of risk perception and that previously used in the literature, our view actually expands upon and incorporates the previous work on risk perception. We suggest that the previous work did not capture risk perception, however, it provided useful information for thinking about the interpretations of probabilities — one aspect of our conceptualization of risk perception. Moreover, the previous emphasis on cognitive information-processing theories is entirely appropriate given the emphasis within genetic counseling on dissemination of information and its goal of informed decision making. Of course, there are other levels at which genetic counseling, risk perception, and decision making can be evaluated (e.g., sociological, affective) and these will serve to create a holistic understanding of the impact and effectiveness of genetic counseling.

CONCLUSION

The empirical studies of risk perception to date have been instrumental in identifying several important issues for future empirical studies of risk perception within the field of genetic counseling. First, is the interpretation of a probability an appropriate way to conceptualize risk perception? This question can best be answered by clarifying one's conceptualization of risk perception. Bearing in mind that the conceptualization of risk perception used by previous researchers reflects a reduction of risk to its statistical, numeric definition, then this conceptualization is appropriate. However, this approach has yielded mixed results and has not been generally successful in identifying variables that explain interpretations of the

recurrence risk. We believe that viewing risk perception as the interpretation of a recurrence risk is fundamentally limited.

We described an alternative conceptualization of risk perception in this paper that views risk as a combination of probability and adversity. In addition, we proposed that the study of risk perception be formulated in terms of the riskiness of a particular decision option. This formulation also employs the notion of an event structure which realistically represents the myriad of consequences associated with an outcome and the ambiguity inherent in most genetic diagnoses. Moreover, we propose that notions of incompleteness and ambiguity, reflecting a higher state of uncertainty, may be important to risk perception. These notions are foreshadowed in research conducted by Black (1979), Lippman-Hand and Fraser (1979b), and a discussion by Vlek (1987). This alternative formulation warrants serious consideration and much more research into these topics is necessary.

Second, how is risk perception explained? In other words, what is the theory about why people perceive risk the way that they do? The explanations employed thus far lie in the information processing branch of cognitive psychology. Is this an appropriate domain from which to draw explanations for the empirical data on risk perception within the genetic counseling domain? This is an extremely important issue and one for which there is as yet no clear answer (Humphreys and Berkeley, 1987; Vlek, 1987). Given the current emphasis of genetic counseling on dissemination of information and informed decision making the domain of cognitive psychology seems highly relevant. Much more theoretical debate and empirical study is warranted.

Third, what is the role played by risk perception in the final decision? The increasing acceptance of risk perception as an important component of decision making accounts for its inclusion as an explanatory, or independent, variable in studies which attempt to explain or predict decisions (Lippman-Hand and Fraser, 1979d; Evers-Kiebooms, 1987; Frets *et al.*, 1990a, b). A clearer understanding of the role of risk perception should aid in a clearer understanding of the decision-making process. However, additional research aimed at clarifying the role of risk perception within the decision making process is necessary.

The purpose of this discussion has been to assess the origins of the studies of risk perception, expose their fundamental limitation, and to introduce an alternative approach to the study of risk perception. Although our ideas require further refinement and validation, we believe this discussion provides a good "first approximation." We hope this article initiates a healthy debate about the conceptualization and operationalization of risk perception.

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