Chapter 1
Medical Informatics as a Scientific Discipline

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Abstract This chapter will provide a brief history of the birth and development of medical informatics, followed by a description of its principal domains. These domains include bioinformatics, which relates to molecular and cellular aspects of medicine, clinical informatics, which deals with patient data and medical knowledge relating to the care of individual patients, and public health informatics, which brings together the tools, techniques and applications for reasoning at the population level. Links with other disciplines, including subdisciplines of computer sciences, biostatistics and biomedical engineering, have also been developed. There are many scientific societies for medical informatics, operating at the national, continental and international levels. These societies are presented here, together with the principal journals, scientific conferences and exhibitions in this field.

Keywords Medical informatics history • Biomedical informatics • Bioinformatics

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A. Venot et al. (eds.), *Medical Informatics, e-Health*, Health Informatics,
DOI 10.1007/978-2-8178-0478-1_1,

After reading this chapter you should:

- Be able to explain what medical informatics is
- Be aware of the principal links between medical informatics and various other areas, such as scientific computing, biostatistics and biomedical engineering
- Be able to cite the major associations and national and international scientific societies in the field of medical informatics
- Be aware of the principal journals and international conferences for researchers to communicate their results and know how to submit articles and communications
- Be aware of some of the research structures and sources of funding for research in medical informatics.

1.1 Development of Medical Informatics

Medical informatics has gradually established itself as a scientific discipline (Shortliffe and Blois 2006; Geissbuhler et al. 2011; McCray et al. 2011). In this chapter, we present a brief history of this discipline, its key areas and links with other disciplines. We describe the national and international societies for medical informatics and discuss the major journals and congresses in this field, together with sources of research funding.

Interest in the potential value of computers for use in medicine first emerged in the 1960s and has rapidly grown ever since. From the outset, the goals were ambitious, including the replacement of paper medical records with electronic records and assisting or replacing doctors in diagnostic or therapeutic procedures. Early in the development of medical informatics, a number of key issues, such as terminology systems in health, medical knowledge modelling and reasoning, rapidly emerged.

A new scientific community developed. In 1968, a working group of researchers working specifically in the field of medical informatics was established within the International Federation for Information Processing (IFIP). One of its notable participants was Professor François Grémy (Degoulet et al. 2005), a French researcher. This committee became increasingly powerful, leading to the creation in 1974 of an international scientific society, the International Medical Informatics Association (IMIA). In the same year, the IMIA organised the first world congress in this field, Medinfo.

This medical informatics community has organised itself into a network of national and international associations. It has set up its own conventions, journals and standards bodies. In academia, research laboratories have been set up, together with specific Masters and Ph.D. programmes.

This research has generated a number of commercial products. Software vendors and database editors in the domain of health have set up companies developing products for the computerisation of hospitals, medical and dental offices, imaging departments and pharmacies.

Public partners, firmly convinced of the potential value of information technology (IT) to improve health systems and reduce costs, have been established in many countries to provide research funding at the national or supranational level.

IT management rapidly developed to reach a certain level of maturity, but the computerisation of healthcare processes proved complex. After 50 years of development, IT has spread throughout the health sector, but further progress and active research are required (Haux 2010). For example:

- Electronic medical records are widespread, but their routine use in everyday practice remains difficult and time-consuming for health professionals. Physicians, for example, continue to input many patient data in natural language, whereas the use of terminology systems to code these data would make more elaborate functions possible.
- Many clinical studies have shown that diagnostic errors are often made in the various medical specialities. Much effort has been made over the last 30 years to develop software to assist doctors with diagnosis, but these programs do not yet seem to have come of age.
- Many physicians appreciate computer assistance in the drafting of prescriptions, because it reduces the risk of unsafe prescriptions, but handwritten prescriptions are still very abundant.

1.2 The Main Areas of Medical Informatics

Since the 2000s, the term "medical informatics", which was originally coined in Europe, has spread to the United States and around the world, to describe this discipline. However, the term "biomedical informatics" has recently gained in popularity, as it ensures that rapid developments in bioinformatics are not obscured (Shortliffe and Blois 2006). This term encompasses bioinformatics as a subdomain.

In this book, we have deliberately avoided including a chapter dealing explicitly with bioinformatics, although some concepts specific to this area are presented in Chap. 17. Our goal was to focus on the field of health, without including purely biological applications.

The diagram below outlines the major areas of biomedical informatics and its various microscopic and macroscopic levels (Fig. 1.1).

Bioinformatics deals with the molecular and cellular levels of medicine. Very sophisticated methods have been developed for the analysis of gene sequences. Bioinformatics gained prominence with the advent of functional genomics, proteomics, transcriptomics and high-throughput sequencing. Research is progressive and continually adding to our knowledge, and there is now a need for a rational

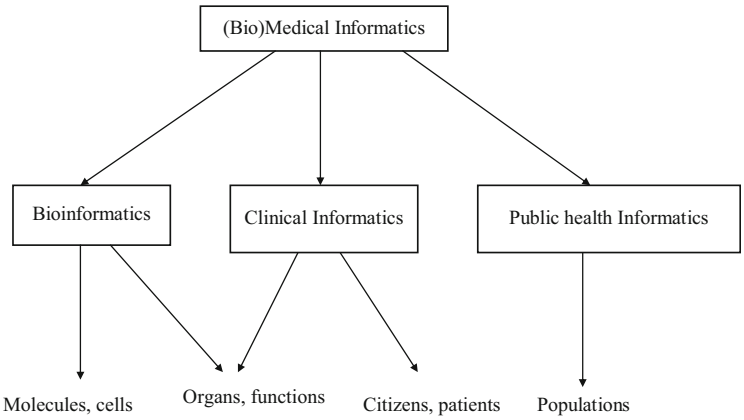

Fig. 1.1 (Bio)Medical informatics and its various subdomains

framework to explain large imbalances and their translation to the cellular or molecular level.

The principal applications in personalised medicine and health concern diagnosis and treatment. Functional genomics data from microarrays are useful for studies of the relationships between genes and for understanding the determinants and molecular mechanisms underlying the severity of a given disease in a given individual. They can also be used clinically, to select a drug treatment suitable for use in an individual with a particular genetic background (Kulikowski and Kulikowski 2009).

Clinical and bioclinical informatics deal with patient data and medical knowledge relating to the care of individual patients. Clinical informatics aims to provide methodological and technical solutions for data and knowledge representation, organisation, capture, storage, interrogation, interpretation, communication and use in practice (Mitchell et al. 2011). Computer support can be used in the presence of patients or remotely, through the use of telemedicine tools.

Specific methods are applied to populations, in a domain known as public health informatics. Public Health aims to develop educational, preventive, curative and social activities to improve the overall health of populations. If such actions are to be effective, they require the support of information systems designed with medical informatics methods. The WHO is now insisting that all states should consider the quality of their health information systems and seek to improve them.

Public health informatics brings together the tools, techniques and applications for reasoning not at an individual level, but at the population level. Tools for following cohorts, disease registries or vigilance systems have been derived from this approach. This is the case, for example, for pharmacovigilance, in which suspected adverse event reports for particular drugs are collected from various sites and efforts must then be made to group them on the basis of their similarities.

There is thus an important need for the aggregation of information, data mining and automatic alerts, to support decision-making with a potentially major impact on the health of the population.

1.2.1 Medical and Computer Sciences

Developments in medical informatics are based on methods and tools developed by computer scientists, although some of the problems encountered in the domain of health are original and lead to advances in computational methods.

The major areas of IT providing techniques widely used in medical informatics include:

Databases: the computerization of medical records is based on the use of database management systems (DBMS). One particular problem is posed by the need to develop and use specific terminology systems to represent and code the symptoms, clinical signs, disease and treatment of patients.

Artificial intelligence: the components of artificial intelligence are widely used in medical informatics. Decision-making is one of the core activities of physicians. It is difficult because the information on which decisions are based is plentiful but scattered and heterogeneous, and may be ambiguous or implicit, and because the mechanisms of medical reasoning may be complex. Knowledge engineering methods are used to model medical knowledge, to design inference engines for decision support systems and to develop health ontologies. Machine learning methods are used for medical data mining and language processing can be used for automatic structuring of the information (diseases, treatments) contained in the hospital and surgical reports.

Networks: computer networks support data exchange, providing the user with easy access to large volumes of data increasing the availability of data for patient management. However, the use of networks generates specific problems in medicine relating to the standardisation of content in terms of the semantics of messages, data security and privacy and the need to respect the rights of patients.

Automatic processing and analysis of medical images: medical imaging devices generate digital images and methods of contrast enhancement, 2D or 3D reconstruction, segmentation and the automatic registration of images are widely used.

The methods used in geographic information systems are useful for the development of epidemiological surveillance systems (simulation and visualisation of epidemics of influenza or measles, for example).

1.2.2 Medical Informatics and Biostatistics

Biostatistics provides a methodological framework for clinical research, which involves humans and is designed to generate new information about diseases or to assess and compare new treatments or new diagnostic procedures. Biostatistics can be used to make inferences and to draw conclusions about populations from observations or measurements on samples of individuals. Medical informatics and biostatistics differ principally in that many of the developments in the domain of medical informatics involve no probabilistic modelling. However, these two disciplines are closely linked and complementary in many areas.

Some of the methods developed by biostatisticians are used in the development of decision support systems in medical informatics. A particular example of this is provided by the use of Bayes' theorem for computer-aided diagnosis. Indeed, purely statistical techniques make little use of medical knowledge and are far from the reasoning of physicians. Conversely, purely logical methods do not take into account a very important type of information: the prevalence of disease and the frequency of signs in disease. This probabilistic dimension, represented by Bayes' theorem, significantly enriches the reasoning process and much work in medical informatics is based on this approach. In addition, purely statistical data mining approaches can be used for the analysis of large collections of data, and computer-based methods derived from machine learning techniques may also be applied.

1.2.3 Medical Informatics and Biomedical Engineering

Biomedical engineering involves the application of engineering methods and techniques to the medical domain, with the aim of developing devices useful for disease diagnosis and non-pharmacological treatments. This field combines physics, computer science, medicine and biology. Digital images are processed to compensate for their imperfections and merged images are produced by several methods, for automatic comparisons of the same object at successive time points, to highlight certain features of the image and to calculate areas or quantitative parameters of clinical interest. Such treatments may be applied to static images or to sequences of images. Some devices developed especially for surgical robotics and interventional radiology are largely based on medical image processing methods. Biomedical engineering covers the area of signal processing (analysis of physiological signals or applications to help patients to continue living at home (e.g. fall detection)).

1.3 Scientific Societies in the Domain of Medical Informatics

Many scientific societies have been set up in the domain of medical informatics over the last 40 years. These associations are federated at the continental level and these continental organisations themselves belong to the IMIA (International Medical Informatics Association).

1.3.1 The IMIA and Its Federations

Six continental federations of national associations have been formed:

- APAMI: Asia Pacific Association for Medical Informatics http://www.apami.org/;
- EFMI: European Federation For Medical Informatics (http://www.helmholtz-muenchen.de/ibmi/efmi/);
- Helina: African Region (http://www.helina-online.org/);
- IMIA LAC: Regional Federation of Health Informatics for Latin America and the Caribbean (http://www.imia-medinfo.org/new2/node/159);
- IMIA North America;
- MEAHI: Middle East Association for Health Informatics http://www.imia-medinfo.org/new2/node/160.

More than two dozen working groups bring together representatives from particular domains (e.g. computing and genomic medicine, dental computing).

The IMIA (http://www.imia.org/) has several types of activities and products:

- The World Congress of Medical Informatics, MEDINFO, which is organised under the auspices of the IMIA, every 3 years;
- A "Yearbook of Medical Informatics", reproducing a series of articles of particular importance published in various journals during the course of the year.

1.3.2 The National Societies

Many countries have national medical informatics associations. In the US, the American Medical Informatics Association (AMIA) is particularly active and organises two annual conferences. (https://www.amia.org/).

In France, the AIM (originally known as the Association for Computing Applications in Medicine) has been in existence since 1968 (http://france-aim.org/).

Table 1.1 The main international journals in medical informatics

Name of the journal	Free access Y/N	Editor	Scientific domain
Artificial Intelligence in Medicine http://www.aiimjournal.com/	N	Elsevier	Theory and practice of artificial intelligence in medicine
BMC Medical Informatics and Decision Making www.biomedcentral.com/bmcmedinformdecismak	Y	BioMed Central	Information technologies and decision-making
International Journal of Medical Informatics www.elsevier.com/locate/ijmedinf/	N	Elsevier	Clinical informatics
Journal of Biomedical Informatics www.elsevier.com/locate/yjbin	N	Elsevier	New methodologies and techniques
Journal of the American Medical Informatics Association http://jamia.bmj.com/	Y (after 12 months)	BMJ group	Clinical informatics
Journal of Medical Internet Research http://www.jmir.org/	Y	Eysenbach	e-health, clinical applications of the Internet
Methods of Information in Medicine http://www.schattauer.de/en/magazine/subject-areas/journals-a-z/methods	N	Schattauer	Methodologies for the processing of health information

1.4 The Main Journals and Scientific Conferences in Medical Informatics

1.4.1 The Scientific Journals

Medical informatics researchers submit articles reporting the results of their studies to various international scientific journals (Fu et al. 2011; Spreckelsen et al. 2011). Table 1.1 provides the details of several of these journals (name, publisher, type of access and fields covered).

The websites of these journals provide recommendations and guidelines for the writing and submission of articles. The submitted articles are then sent to at least three "reviewers" competent in the domain, who analyse the article and provide their opinions and comments. The editor summarises the recommendations made by the reviewers and transmits his or her decision to the authors. The decision may be the rejection of the article, a request for modifications and improvements (the authors are asked to provide a covering letter with their resubmission, explaining how they have taken into account the reviewers' criticisms and suggestions), or direct acceptance with no modifications requested (very unusual). The review and resubmission process for an article may take between a few weeks and a few months.

1.4.2 The Conferences

Conferences are organised by national or supranational scientific societies, at various intervals. Some of these conventions are particularly interesting because the resulting articles (but not the posters) are indexed in Medline and can be found by searching the PubMed search engine developed by the National Library of Medicine.

These conferences include:

- MEDINFO: World Congress of Medical Informatics, held every 3 years under the auspices of the International Medical Informatics Association (IMIA). Articles are published in the series "Studies in Health Technology and Informatics";
- The annual conference of the AMIA (American Medical Informatics Association), held annually (in Washington or in another American city), bringing together researchers from the US and elsewhere.
- The MIE (Medical Informatics Europe) Congress, organised annually under the auspices of the European Federation for Medical Informatics (EFMI). Articles are published in the series "Studies in Health Technology and Informatics".

The submission process is similar to that for journal articles, with the conference website providing instructions for authors. Medical software is often presented and exhibited at these conferences. The exhibitions organised by the HIMSS (Healthcare Information and Management Systems Society http://www.himss.org) are particularly useful in this respect. HIMSS is represented in North America, Europe and Asia.

1.5 Structuring and Funding of Research in Medical Informatics

Most medical informatics research is carried out in university laboratories, but the medical software industry also plays a role in research and development. Medical informatics research is funded by national and supranational organisations.

- For the USA, the various funding bodies are listed on the site of the AMIA (https://www.amia.org/informatics/research/agencies.asp);
- The European Union annually publishes tenders for projects including research linking academic and industrial partners (http://ec.europa.eu/information_society/activities/health/research/index_en.htm);

1.6 For More Information

Internet search engine queries with the keywords “medical informatics history” return a series of links to articles tracing the history of the development of medical informatics.

The websites of the major associations and medical informatics journals and conferences provide tips for authors and information about submission procedures.

Exercises

Q1 Go to the European Union website and look for tenders in medical informatics for this year and next year.
What type of file should be created to respond to such tenders?

Q2 What are the principal medical informatics conferences scheduled for the next year? You should visit the websites of the major societies in this field to find the answer.

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