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Welcome to the 8th Edition of Health Informatics: Practical Guide. The goal of this textbook is to provide a comprehensive introduction to the field of health informatics (also known as biomedical and health informatics or just plain informatics) for students. The book can also serve as a current update for those who are already practitioners. As in any professional field, there is a knowledge base and history of scientific progress in informatics that informs current practice.

The impetus for this book came from Robert Hoyt, MD, who served as the lead Editor for the first seven editions. In preparing the 7th edition, Dr. Hoyt engaged me as a Co-Editor, with the plan for me to become the Editor of the 8th edition, and hopefully beyond. Dr. Hoyt and his wife Dr. Ann Yoshihashi have been great mentors in the process of putting a book like this together.

This book is aimed at those who wish to learn about the field of biomedical and health informatics (BMHI). The content is almost completely redone from previous editions and roughly follows the current content of a course I have taught for three decades to about 5000 students in an introductory graduate course at Oregon Health & Science University (OHSU), the American Medical Informatics Association (AMIA) 10x10 ("ten by ten") program, and a number of other derivative courses for medical students, graduate students, and health IT professionals. I have engaged a variety of other authors for many of the chapters to give additional expertise and perspective.

The contents of the book aim to give a practical and big-picture view of BMHI while providing copious references and additional readings for more detailed study. Chapter 1 introduces BMHI and its core terminology. Chapter 2 provides a brief history of BMHI, as work in the present is often better understood by the perspective of what was done in the past to get to where we currently are. Although BMHI is not centrally about technology, understanding of essential computing concepts is required, which is covered in Chapter 3.

Chapter 4 covers perhaps the most central application of BMHI, the electronic health record and the clinical data within it. Chapter 5 explores the one of the key challenges for BMHI, namely standardization of data and its interoperability across systems. As BMHI and all of health and healthcare are being impacted by data science and artificial intelligence, Chapter 6 covers the accomplishments and limitations of these areas. Chapter 7 explores the current status and future directions for clinical decision support. As a good deal of health information is in textual format, Chapter 8 reviews the applications and current state of the art of natural language processing. Chapter 9 explores the role of BMHI in the safety, quality, and value of healthcare.

Chapter 10 explores the importance and implementation of exchange of health information across business and other boundaries. As the implementation of the electronic health records is a complex process, Chapter 11 covers the selection and implementation of such systems. Chapter 12 reviews the applications telemedicine and telehealth, covering their tremendous growth in recent years. Chapter 13 explores the critical topics of privacy and security of health information.

The next few chapters move beyond the focus of direct patient care. Chapter 14 covers information retrieval (also known as search) systems that enable clinicians, researchers, and increasingly patients to find and apply knowledge for health-related decisions. Chapter 15 introduces clinical research informatics, focusing on how BMHI can improve clinical and translational research. With growing discovery of genomics and other omics, Chapter 16 explores how they are impacting health and healthcare.

These chapters are followed by those covering more focused areas of BMHI. The critical role of nursing and informatics in health and healthcare is covered in Chapter 17. The vital role of the patient or consumer and their interactions with BMHI is explored in Chapter 18. The equally critical role of BMHI in public health is described in Chapter 19. While not all introductions to BMHI include coverage of evidence-based medicine, Chapter 20 explores the application of the best evidence for making clinical decisions. Chapter 21 introduces the important role of imaging in BMHI. The final two chapters cover...
the essential areas of ethical issues in Chapter 22 and human-computer interaction in Chapter 23.

A major appreciation for this book goes to Managing Editor Kate Fultz Hollis, who did an excellent job editing chapters and working with all the authors. She has also done a tremendous job at Oregon Health & Science University (OHSU) in serving as a teaching assistant in many of my courses and a research assistant in many of my projects. I also appreciate all those authors listed above for completing their chapters in a timely manner. This effort and all of my other individual successes would not have occurred without the support of the faculty, students, and staff of the Department of Medical Informatics & Clinical Epidemiology at OHSU.

This book also would also not be possible without the support of my family, including my wife Sally, my daughter Becca, my daughter Alyssa and her husband AJ Sowles, and the various dogs (Otis, Pheo) and cats (Cappie, Rusty) across our family.

William Hersh
July, 2022
The context of health. The term medical informatics is purported to first have been used in 1974.(3) At present, informatics is most substantially used in the biomedical arena, although there are fields outside of medicine such as legal informatics, chemoinformatics, social informatics, and so forth.

The various areas within biomedical and health informatics are depicted in Figure 1. Sometimes narrower words appear in front of the term informatics. Other areas of biomedical and health informatics include:

- Bioinformatics – the application of informatics in cellular and molecular biology, often with a focus on genomics
- Imaging informatics – informatics with a focus on imaging, including the use of systems to store and retrieve images across all types of informatics
- Clinical informatics – informatics applied in healthcare settings
- The application of informatics focused on specific healthcare disciplines, such as nursing (nursing informatics), dentistry (dental informatics), pathology (pathology informatics), etc.
• **Consumer health informatics** – the field devoted to informatics from a consumer view, often with a focus on mobile health
• **Clinical research informatics** – the use of informatics to facilitate clinical research, with increasing emphasis on translational research that aims to accelerate research findings into clinical practice
• **Public health informatics** – the application of informatics in areas of public health, including surveillance, reporting, and health promotion

**Figure 1. Areas within biomedical and health informatics**

![Figure 1](https://informaticsprofessor.blogspot.com)

Shortliffe et al. describe the continuum of bioinformatics, clinical informatics, and public health informatics as a spectrum applied from cellular and molecular processes to people to populations, although there is considerable overlap, as exemplified through the use of genomics technologies to investigate public health outbreaks.(5) They define the field of health informatics as the confluence of clinical informatics and public health informatics.

Readers may have heard this field called different names, such as biomedical informatics, clinical informatics, and medical informatics. Some names for the field focus even more narrowly, such as medical specialties, e.g., pathology informatics, and other health professions, e.g., dental informatics and nursing informatics. Others focus on broader categories, such as imaging informatics or research informatics. This author has noted that the field has an “adjective problem,” in that there is variation in the adjective(s) preceding the word informatics. Another group of ambiguous terms includes the name for practitioners of HCIN, who are called informaticians by some and informaticists by others. However, all of these different names for the field to have more in common than in difference, and one definition from the American Medical Informatics Association (AMIA), the professional association for the field, can be applied to all of them: The interdisciplinary field that studies and pursues the effective uses of biomedical data, information, and knowledge for scientific inquiry, problem solving, decision making, motivated by efforts to improve human health.(6)

Some earlier definitions of BMHI included the storage, acquisition, and use of information,(7) the science of information applied to biomedicine – data plus meaning,(8) a paradigm shift in biomedicine from individual brains to systems of brains,(9) and the science of sociotechnical systems.(10) There have also been European and global perspectives.(11–13)

Friedman defines the field from the perspective of what informatics is and is not.(14) Informatics is cross-training, where basic informational sciences meet a biomedical application domain; relentless pursuit of assisting people; and a tower of achievement that includes model formulation, system development, system implementation, and study of their effects. Informatics is not scientists or clinicians tinkering with computers, analysis of large data sets per se, circumscribed roles related to deployment of electronic health records, the profession of health information management, or anything done using a computer. Friedman has also defined a fundamental theorem that depicts the goal of informatics, which is to use computing technologies to augment human cognitive abilities rather than replace them.(15)

This author has written a great deal on BMHI and aspects of it, including on a Web site,¹ in a blog,² and in a number of articles and chapters:

• Medical informatics: improving healthcare through information (16)
• Health care information technology: progress and barriers (17)
• Characterization of and changes in the profession (18)
• Many career opportunities (19)
• Reconciling definitions of terms (4)
• The informatics professional workforce (20)

BMHI is not only important for those who work in the field, but is a core competency of modern health professionals. According to a 2003 Institute of Medicine report, the modern healthcare professional must have competency in informatics as part of the larger goal of providing patient-centered care.(21) Critically important is that informatics competency is not just computer literacy, as the so-called Google generation does not necessarily have good information skills.(22) BMHI is also a core component of the learning health system.(23,24) Informatics is

¹ http://informatics.health/
² https://informaticsprofessor.blogspot.com
a core skill for all modern health-related endeavors. It is certainly as essential skill for healthcare professionals, who must have competence in the use of key applications for patient care, such as the electronic health record and its critical functions, such as clinical decision support and order entry.(25) The ability to find and apply knowledge is also a critical skill in modern times.(26)

Related Terminology

We can also gain more perspective about informatics by noting how the field is distinguished from related subjects. The term information technology (IT) generally refers to computers and related technology. Computer science is an academic discipline that focuses on the scientific aspects of computing and IT. A related discipline is management information systems, which is another field underlying IT that is usually taught in business schools. We sometimes see the use of the term health information technology (HIT or health IT), which generally refers to the health-related application of IT.

Another set of related terms falls under health information management (HIM), which is the discipline historically focused on management of (paper) medical records (changing in current environment), with three main levels of practice:

- Registered Health Information Administrator (RHIA) – highest level, baccalaureate degree
- Registered Health Information Technologist (RHIT) – associate degree
- Certified Coding Specialist (CCS) – usually less then associate degree

Another term gaining use is digital health, which is a broad term for digital, i.e., IT-related, aspects of health and healthcare.(27) This term emanates from information and communications technology (ICT), which is essentially same as IT with added emphasis on telecommunications. Similar terms include eHealth, which is the use of ICT for health, and mHealth, which involves the use of mobile devices for health. There are growing efforts in digital health around in the world, from the United States3 to Australia4 to Asia.5

Another set of related terms are the “tele” terms. These include telemedicine, which is the provision of healthcare when participants separated by time and/or distance. A related term is telehealth, which adds the pursuit of health when separated by time and/or distance, although these terms are frequently used interchangeably. Sometimes the clinical discipline is added to the term, e.g., telepathology or teleradiology.

Another related term is health systems science (HSS), which is defined as a new science for medicine, different from basic and clinical science. There is a textbook (28) that includes chapter on clinical informatics,(29) and this subject is increasingly incorporated into medical education.(30)

There are some other terms that focus on the application of evidence to clinical practice, often using BMHI tools. Evidence-based medicine (EBM) is the application of the best scientific evidence in medical decision-making. (31) Evidence-based practice (EBP) is the application of EBM in patient care. Comparative effectiveness research (CER) is research that compares one or more diagnostic or treatment options to evaluate effectiveness, safety or outcomes (also called patient-centered outcomes research). Information retrieval (also known as search, part of larger knowledge management) is the field devoted to searching (mostly text, mostly knowledge-based information).(26)

There are also terms related to medical or clinical records. The electronic health record (EHR) refers to the patient’s health record in digital form and has mostly supplanted the older term, electronic medical record (EMR). The personal health record (PHR) refers to the personally controlled health record. An important function of EHRs and other clinical data is health information exchange (HIE), which refers to the exchange of health information across traditional business and other boundaries (a verb). The organization managing HIE used to be called a Regional Health Information Organization (RHIO), but is now called by some an HIE (a noun).

Informatics is also essential for modern biomedical research. Noted in the National Institutes of Health (NIH) Roadmap6 to accelerate biomedical research discovery is the following observation: Today’s biomedical researcher routinely generates ... billions of bytes of data. ... There is no way to manage these data by hand. Researchers need computer programs and other tools to evaluate, combine, and visualize these data. In some cases, these tools will benefit from the awesome strength of supercomputers or the combined power of many smaller machines. In other cases, these tools will be used on modern personal computers connected to data and resources in the cloud. There are important terms to know for informatics applied to clinical research. Translational research,

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3 https://www.fda.gov/medical-devices/digital-health-center-excellence
4 https://digitalhealth.org.au/
6 https://commonfund.nih.gov/bioinformatics/overview
classically, the translation of basic research into clinical applicability (“bench to bedside”), but also from controlled settings to community and population. There is increasing recognition that clinical research findings must “translate” into clinical care more quickly and efficiently, leading to US government investment in clinical and translational research through the NIH Clinical & Translational Science Award (CTSA) program. Translational bioinformatics is bioinformatics applied to health-related problems. Precision medicine is clinical care tailored to an individual’s characteristics, including their genome. It was previously called personalized medicine. The importance of informatics in research has led to a subdiscipline of clinical research informatics (CRI), which is the area of informatics applied to clinical research. The difference between information technology (IT) and informatics is very evident in this domain.

A growing field related to informatics is data science. Data science is “the science of learning from data; it studies the methods involved in the analysis and processing of data and proposes technology to improve methods in an evidence-based manner.” Data science includes the area of data analytics, which is “the extensive use of data, statistical and quantitative analysis, explanatory and predictive models, and fact-based management to drive decisions and actions.” Another well-used term is Big Data, referring to data that meets the categories of the four Vs: Volume, Velocity, Variety, and Variability. There are other related data science terms. Machine learning is ability of computer programs to learn without being explicitly programmed or, the use of computers to optimize a performance criterion using example data or past experience. The current most successful approach to machine learning involves use of neural networks; when using multiple layers, called deep learning. Another term used in this context is artificial intelligence (AI), which is actually an older term referring to information systems and algorithms capable of performing tasks associated with human intelligence.

How is informatics different from data science? Payne et al. describe a process cycle of going from biological, social, or technical processes with afferent and efferent loops of going to and from knowledge respectively. Data science and informatics focus on generating knowledge but informatics is more broadly engaged in the use of that knowledge in real world of health, healthcare, research and other areas.

Blum introduced the notion of a continuum from data to information to knowledge. According to this perspective, data are the raw material collected and stored. Information gives meaning and organization to the data. Knowledge provides understanding and applicability to new situations. Some add a fourth term to this continuum, which is wisdom, defined as knowing how to apply knowledge. Bernstam et al. more fully elaborate the different views of the continuum. They note that data consist of symbols, with a lack of uniformity. Information is then considered to be data that is processed to be useful or given meaning. Information answers questions such as who, what, when, and where. Knowledge then consists of justified or true belief and is applied to data and information to answer questions of how. Wisdom provides understanding of why questions.

**Biomedicine as an Information Science**

Medicine is commonly thought of as a life science, but some point to observations that may more appropriately call it an information science. Clinicians spend substantial amounts of their time gathering and analyzing information to make decisions based on it. Many studies over the years, even from the era before computers, found physicians spend great deal of time with information. A study from 1973 found that physicians in a general medicine outpatient clinic spent about 37.8% of their time charting, 5.3% consulting, 1.7% in other activities, and remaining 55.2% of time with patients. More recent studies have found that 14-39% of clinical work took place outside the exam room. It has also been seen that work related to the patient when the physician was not present consumed 15-23% of the work day. Similar observations have been seen in hospitals and emergency departments. Time studies of hospital and emergency physicians show that these physicians spent about 15-38% of their time in direct patient care and 50-67% of their time in indirect patient care, divided between reviewing results, performing documentation, and engaging in communication.

More recent studies in era of widespread EHR adoption have found even more physician time spend engaged with information, both those in training and those in practice. For the former, a study of interns found that they spent about 40% of their time interacting with computers. A study of residents in an academic teaching hospital noted about 50% of time spent with computers versus 10% time directly with patients.

In one survey of practicing general internists, about 60% reported “loss” of around 48 minutes per day due to EHR usage. Another study of physicians found they spent two hours of time doing EHR and desk time for every hour of direct patient time, plus an additional 1-2 hours per night. Two studies of primary care
Chapter 1: Introduction to Biomedical and Health Informatics

Table 1. HIMSS health information and technology job titles

<table>
<thead>
<tr>
<th>Analytics/Report Writer</th>
<th>Application Analyst</th>
</tr>
</thead>
<tbody>
<tr>
<td>Applied Data Scientist</td>
<td>Business Analyst</td>
</tr>
<tr>
<td>Chief Business Development Officer</td>
<td>Chief Digital Officer</td>
</tr>
<tr>
<td>Chief Experience Officer (CxO) (Chief Patient Experience Officer)</td>
<td>Clinical Information Specialist</td>
</tr>
<tr>
<td>Chief Medical Officer</td>
<td>Chief Medical Information Officer</td>
</tr>
<tr>
<td>Chief Nursing Informatics Officer</td>
<td>Chief Privacy Officer</td>
</tr>
<tr>
<td>Chief Risk Officer</td>
<td>Chief Technology Officer</td>
</tr>
<tr>
<td>Clinical Systems Analyst</td>
<td>Clinical Transformation Analyst</td>
</tr>
<tr>
<td>Compliance Professional</td>
<td>Cyber Architect</td>
</tr>
<tr>
<td>Data Analyst</td>
<td>Data Analyst / Health Plan</td>
</tr>
<tr>
<td>Data Architect</td>
<td>Database Administrator</td>
</tr>
<tr>
<td>Data Manager</td>
<td>Data Privacy Analyst</td>
</tr>
<tr>
<td>Data Security Analyst</td>
<td>Desktop/Field Services Technician</td>
</tr>
<tr>
<td>Director Of Clinical Information Systems</td>
<td>Document Integrity Specialist</td>
</tr>
<tr>
<td>Information Security Manager</td>
<td>Integration Analyst</td>
</tr>
<tr>
<td>IT Auditor and Risk Analyst</td>
<td>Nurse Informaticist</td>
</tr>
<tr>
<td>Privacy Specialist</td>
<td>Process Engineer/Process Improvement Specialist</td>
</tr>
<tr>
<td>Program/Project Manager</td>
<td>Quality Analyst</td>
</tr>
<tr>
<td>Revenue Cycle Analyst</td>
<td>Risk Professional</td>
</tr>
<tr>
<td>Sales, Solutions Specialist</td>
<td>Security Analyst</td>
</tr>
<tr>
<td>Senior Privacy Specialist</td>
<td>Senior Solutions Architect</td>
</tr>
<tr>
<td>Service Desk Analyst</td>
<td>Senior System Administrator</td>
</tr>
<tr>
<td>Senior Director of Business Intelligence</td>
<td>Software Developer</td>
</tr>
<tr>
<td>Systems Architect</td>
<td>Telemedicine/Telehealth Nurse Practitioner</td>
</tr>
<tr>
<td>Vice President, Chief Information Officer</td>
<td>Vice President, IT Operations</td>
</tr>
</tbody>
</table>

physicians found spending 50%(65) and 60%(66) of workday with EHR and other desktop medicine tasks.

While modern physicians likely spend too much time with their computers, often these studies do not answer what is the right amount of time.(67) Physician performance and safety are impacted by “information chaos.”(68) Epidemiology as well as all of the healthcare system have been labeled as “complex adaptive systems,” where small changes can be unpredictable and lead to new states of equilibrium.(69)

The Informatics Workforce

BMHI can also be understood by describing the activities of practitioners, researchers, and others who work in the field. One way to categorize the work of informatics is to consider the major activities of the professional workforce. One way to describe the workforce is to categorize its traditional groupings, at least in healthcare settings: (20)

- Information technology (IT) – usually with computer science or information systems background
- Health information management (HIM) – historical focus on medical records
- Health or clinical informatics (CI) – often but not always from healthcare backgrounds, performing implementation, analysis, training, etc.
- Others – librarians, managers, etc.

Informatics professionals also work in other settings such as academia, research institutions, industry, and government.

HIMSS recently collated a list of health information and technology job descriptions,7 with the different job titles listed in Table 1.

7 https://www.himss.org/resources/health-information-and-technology-job-descriptions
Another question is, what is the size of the HIT workforce? We actually know surprisingly little. Most studies assessing the workforce have been done in hospital settings, usually focused on the main groups:
- IT – studies using HIMSS Analytics Database
- HIM – using Bureau of Labor Statistics data
- CI – using ad hoc data

The ambiguity of definitions of health informatics results in confusion as to who works in the field and what they do. In the US, the Bureau of Labor Statistics maintains statistics about the workforce based on its Standard Occupational Classification (SOC) system. Many in the larger health information technology (HIT) community called for the 2018 update of the SOC to add a code for health informatics. Unfortunately this call was unheeded, and health informatics was lumped into a category of Health Information Analysts and Medical Registrars, under which “health informatics specialist” was considered an illustrative example. The lack of a specific SOC code for health informatics makes its workforce less defined and visible. This invisibility has been noted in other countries, such as Australia.

One former source of workforce data in the US was the HIMSS Analytics Database, although it was focused on larger HIT employment, a superset of those whose work in health informatics. Mainly a source of self-reported HIT systems usage by healthcare organizations, the HIMSS Analytics Database formerly captured FTE levels overall and for various job categories, such as management, project management, programming, operations, network administration, help desk, PC support, security, and EMR support. Unfortunately, the workforce data of this resource is no longer maintained.

The HIMSS Analytics Database was used for some workforce analyses. An analysis of data from 2007 found that the US HIT workforce size was estimated to be approximately 108,390. In addition, it was found that as levels of adoption of HIT applications increased within healthcare organizations, so did the amount of FTE. This led to an estimate that the workforce could grow in size to 149,174 if hospitals with lower levels of adoption reached the FTE per bed levels of those at higher levels, such as those incorporating functions such as clinical decision support and order entry. This led to US$118 million in funding for workforce development being included in the US$30B HITECH Act that provided incentives for EHR adoption across the country.

The paper analyzing the 2007 data looked at HIT FTE staffing, especially as it related to level of adoption, based on the well-known HIMSS Analytics Electronic Medical Record Adoption Model (EMRAM), a 0–7 scale that measures milestones of EHR adoption. This was, of course, before the HITECH Act, when a much smaller number of hospitals and health systems had adopted EHRs. Also around that time, there had been the publication of the first systematic review of evidence supporting benefit of healthcare IT, showing the value came mainly from use of clinical decision support (CDS) and computerized provider order entry (CPOE).

As such, the paper looked at the level of healthcare IT staffing by EMRAM stage, with a particular focus on what increase might be required to achieve the level of IT use associated with those evidence-based benefits.

Another finding from the study was that if EHR adoption were to increase to the level supported by evidence of improved care, namely EMRAM Stage 4 (use of CDS and CPOE), and FTE/Bed ratios remained the same for those hospitals, the size of the workforce would need to grow to 149,174. In other words, there was a need to increase the size of the healthcare IT workforce by 40,784 people.

Since 2007, EHR adoption has grown substantially in the US, to 96% of hospitals and 87% of office-based physicians and other clinicians. By 2014, one-quarter of US hospitals had reached EMRAM Stages 6 and 7. An updated study based on 2014 data assessed the impact of increased EHR adoption on the workforce. Although the FTE/Bed ratios in 2014 for different levels of EMRAM are similar to those in 2007 (with the exception of Stage 7, which no hospitals had reached in 2007), because of the advancing of hospitals to higher EMRAM stages beyond Stage 4, the total workforce increased in size from 2007. The study of 2014 data estimated that if all hospitals were to achieve Stage 6, an additional 19,852 healthcare IT FTE would be needed.

The second major group of the HIT workforce is HIM professionals. HIM is a field in transition with the transition from paper to electronic records, and with leadership of the field promoting advanced skills, degrees, and work. The professional association for the field, has developed a career map.

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8 https://bok.ahima.org/PdfView?oid=107412
9 https://brand.amia.org/m/599c75bffeda72b1/original/AMIA-Responds-to-Proposed-Updates-to-Standard-Occupational-Classification-Codes.pdf
10 https://www.himssanalytics.org/
11 https://www.himssanalytics.org/emram
14 https://my.ahima.org/careermap
Two parts of the HIT workforce are the only groups explicitly represented in US labor statistics, which are RHITs and CCSs. The SOC code Medical Records and Health Information Technicians includes both RHITs and CCSs but not RHIAs. Recent BLS statistics estimate about 416,400 employed in 2020, and estimated to increase by 37,100 by 2030 (about 9% growth).

The main reason for the lack of informatics workforce data is inadequate labor statistics. In particular, there are not specific codes in the US Standard Occupational Classification (SOC).\(^\text{15}\) There are some codes in the SOC related to informatics:

- 29-1000+ Health Diagnosing and Treating Practitioners
- 29-2071 Medical Records Specialists – Non-RHIA HIM: RHIT, CCS, and other coders
- 29-9021 Health Information Technologists and Medical Registrars – health informatics specialist is one of several examples but this category is much broader
- 11-911 Medical and Health Services Managers – has Chief Medical Information Officer as an example
- 15-1000+ Computer Specialists

CI professionals represent a growing distinct workforce within HIT. These are mostly individuals who bring skills at intersection of healthcare and IT (Hersh, 2010), although some note they are becoming harder to distinguish from IT (HealthITJobs.com, 2015 – defunct now) The primary skill gap in healthcare IT—specifically Clinical Systems (EMR/EHR)—is that most leaders are looking for employees with both hands-on clinical experience and technical expertise. Historically, these have been two very separate skill sets. One early estimate of need was a stated need for one physician and nurse in each US hospital (~10,000).\(^\text{76}\)

Another way to understand the professional work of those who work in informatics is to analyze job postings. An analysis done in 2020 of the job recruitment site, indeed.com identified 206 positions deemed to be seeking people for work in health and/or clinical informatics positions.\(^\text{77}\) Overall, about three-quarters of the postings were for clinical informaticians, with 62.1% of posts requiring a minimum of a bachelor’s education. RN licensure was required for about two-fifths of the posts, and only 7.3% required formal education in health informatics. The average experience overall was 1.6 years, with bachelor’s and master’s education levels increasing mean experience to 3.5 and 5.8 years respectively.

Electronic health record support, training, and experience with other clinical systems were the most sought-after skills.

Despite the lack of measurement of the workforce, there is one development in the US that points to its relevance, and that is professional certification. The first formal certification was developed for physicians, with clinical informatics designated as a subspecialty of all specialties.\(^\text{78}\) This was done through the formal board-certification mechanism. The core content of the subspecialty was outlined in an analysis by Gardner et al. organized around four domains: (79)

1. Fundamentals
2. Clinical Decision Making and Care Process Improvement
3. Health Information Systems
4. Leading and Managing Change

The American Medical Informatics Association (AMIA) has also developed the Advanced Health Informatics Certification (AHIC)\(^\text{16}\), which certifies other professionals who work in health informatics.\(^\text{80,81}\) AMIA has also developed a set of master’s-level competencies for health informatics professionals based on a foundation of health science, social and behavioral sciences, and information science and technology.\(^\text{82}\) These competencies are used in master’s-level program accreditation being spearheaded by the Council on Accreditation of Health Informatics & Information Management (CAHIIM).\(^\text{17}\)

### Knowledge, Skills, and Competencies

In an attempt to define the work of health informatics professionals, AMIA also commissioned two workforce analyses that interviewed those working in the field to catalog their knowledge, skills, and tasks. This was done for both physicians in the clinical informatics subspecialty\(^\text{83}\) as well as others working in health informatics.\(^\text{84}\) One interesting finding of these independent analyses was a comparable set of high-level domains of work, which are shown in Table 2.

An integration of the workforce analyses led by Silverman and Gadd defines well the work of informatics practice. Both analyses describe the first domain of fundamental knowledge and skills, which include a common vocabulary, basic knowledge across all informatics domains, and understanding of the environment(s) in which the workforce functions. Depending on where an individual works, this may include consumer health, health care, public health, or research settings.

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\(^\text{15}\) https://www.bls.gov/soc/home.htm

\(^\text{16}\) https://www.amia.org/ahic

\(^\text{17}\) https://www.cahiim.org/accreditation/health-informatics
Although not explicitly mentioned in the overall descriptions of this domain (but covered in the details of practice) are the ability to identify and minimize biases in data and mitigate their impact as well as to implement and evaluate machine learning and artificial intelligence applications in all health-related settings.

The final domain reflects the sociotechnical aspects of informatics, with required abilities in leadership, professionalism, and transformation. Informatics practice should be able to build support and create alignment for informatics best practices as well as lead informatics initiatives and innovation through collaboration and stakeholder engagement across organizations and systems.

Other research has noted that competency in informatics is a requirement of all healthcare professionals for whom use of informatics is essential to clinical practice. Hersh et al. defined such competencies for medical students, although they noted they really applied to all health care professionals. Table 3 lists the high-level competencies for medical students. Another effort to define competencies for the health care workforce is the TIGER Initiative.

The second domain differs somewhat between the HI and CIS analyses but can be integrated into an overall focus on enhancing health decision-making and improving health care delivery and outcomes. Informatics practice should support and enhance decision-making by clinicians, patients, policy makers, researchers, and public health professionals. It must also analyze existing health processes and identify ways that health data and health information systems (HIS) can enable improved outcomes. Informatics work should also evaluate the impact of HIS on professional practice as well as pursue discovery and innovation. More clinically, informatics practice should be able to develop, implement, evaluate, monitor, and maintain clinical decision support while also supporting innovation in the health system through informatics tools and processes.

The third domain of each analysis can be combined into an overall category of health and enterprise information systems. Informatics practice should include planning, developing or acquiring, implementing, maintaining, and evaluating HIS that are integrated with existing information technology systems across the continuum of care. This should include the clinical, consumer, and public health domains and address security, privacy, and safety considerations. This domain should also include the development, curation, and maintenance of institutional knowledge repositories, also while addressing security, privacy, and safety considerations.

A critical domain is the new addition to the previous four domains of the Gardner et al. analysis, which can be integrated as data governance, management, and analytics. Practice should include establishing and maintaining data governance structures, policies, and processes. The workforce should be able to acquire and manage health-related data to ensure their quality and meaning across settings and to utilize them for analysis that supports individual and population health while driving innovation. It is also critical to incorporate information from emerging data sources and derive insights to optimize clinical and business decision-making.

Table 3 lists the high-level competencies for medical students. Another effort to define competencies for the health care workforce is the TIGER Initiative. Informatics Leaders

What do we know about informatics leaders? Most healthcare organizations have an individual who oversees all IT and is typically called the Chief Information Officer (CIO). A survey of CIOs SSI-Search survey found that most see their role as strategic more than operational leaders in healthcare organizations. Their most common bachelor’s degrees were in fields such as computer science, IT, business, and management. About 74% have master’s degrees, most commonly MBA and other business but also MHA, MPH, and others. They viewed their most important skills looking forward to be data analytics and clinical experience.

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Table 2. Domains of practice for health informatics (84) and the clinical informatics subspecialty.

<table>
<thead>
<tr>
<th>Domain</th>
<th>Health Informatics</th>
<th>Clinical Informatics Subspecialty</th>
</tr>
</thead>
<tbody>
<tr>
<td>1. Foundational Knowledge</td>
<td>Fundamental Knowledge and Skills</td>
<td>Improving Care Delivery and Outcomes</td>
</tr>
<tr>
<td>2. Enhancing Health Decision-making, Processes, and Outcomes Tasks</td>
<td>Improving Care Delivery and Outcomes</td>
<td>Enterprise Information Systems</td>
</tr>
<tr>
<td>3. Health Information Systems (HIS) Tasks</td>
<td>Enterprise Information Systems</td>
<td>Data Governance and Data Analytics</td>
</tr>
<tr>
<td>4. Data Governance, Management, and Analytics Tasks</td>
<td>Data Governance and Data Analytics</td>
<td>Leadership and Professionalism</td>
</tr>
<tr>
<td>5. Leadership, Professionalism, Strategy, and Transformation Tasks</td>
<td>Leadership and Professionalism</td>
<td></td>
</tr>
</tbody>
</table>

---

There are also IT leaders who focus on clinical information systems and fall under the category of Chief Clinical Informatics Officers (CCIOs).(86) Most large and many smaller healthcare organizations have a Chief Medical Information (or Informatics) Officer (CMIO) who is typically a physician. Growing numbers of healthcare organizations have a Chief Nursing Informatics Officer (CNIO). It is less common for other clinical areas (e.g., Dental, Pharmacy) to have a titled leader but all areas of healthcare have a need for critical skill sets and training in informatics. Hospitals and healthcare organizations increasingly creating operational clinical informatics departments. These may be separate from IT (and CIO) and usually have clinical leadership, often the CMIO. They increasingly incorporate HIM departments.

Some analysis has been done of the CMIO role. The position is now an important part of healthcare organizations, serving as the liaison between clinicians and IT as well as the executive informatician.(87) One early analysis of five CMIOs found that leadership, communication, and consensus-building were among most important skills, they desired to be part of senior physician executive team, and did not want to be seen as just “techie” doctors.(88) More information and insights comes from a “survival guide”(89) and annual surveys from the recruiting firm, WittKieffer (Chicago, IL).

The most recent WittKieffer-AMDIS annual physician survey was completed by 161 physician informatics leaders.(90) The survey found that CMIO was still the most prominent title for physician informatics leaders but that there was growing use of the title, Chief Health Information Officer (CHIO). Some were titled, Director, Medical Informatics and there were others in associate roles of all of these titles. The majority worked in large healthcare delivery organizations, e.g., multi-hospital systems or integrated delivery networks. Over half continued to work part-time clinically at up to 25% time, with the remainder split between no clinical practice or >25% clinical practice. CMIOs typically reported to the CIO (44%), CMO (32%), CEO (8%), or others (16%). Three-fourths had direct reports, i.e., led teams. Almost three-fourths were board-certified in clinical informatics, a number that has been increasing over time. However, only 28% had formal informatics training, with little change over time.

Indeed, the role of the CMIO or CHIO is seen as evolving from implementation to optimization of the EHR, along with providing strategic leadership in AI, population health, and patient engagement.(91) Another workforce of importance is the nursing informatics workforce. Most of the data comes from HIMSS, which publishes a report on the nursing informatics workforce every few years.(92) According to this report, over two-thirds of nursing informatics professionals work for a hospital or multi-facility health system. There has been a significant change in reporting structure, with more nurse informaticians reporting to Nursing, Quality, and Operations over the historical trend of reporting to IT. There also continues to be uptake in the CNIO or Senior Nursing Informatics Officer title, with 41% reporting that their organization had that formal role. There are more who have master’s degrees in informatics and obtain nursing informatics certification. Finally, one recent article discussed how the role of CNIOs has been evolving from tactical to strategic concerns.(93)

Table 3. Competencies in clinical informatics for medical education (25)

<table>
<thead>
<tr>
<th>No.</th>
<th>Competency Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>1.</td>
<td>Find, search, and apply knowledge-based information to patient care and other clinical tasks</td>
</tr>
<tr>
<td>2.</td>
<td>Effectively read from, and write to, the electronic health record for patient care and other clinical activities</td>
</tr>
<tr>
<td>3.</td>
<td>Use and guide implementation of clinical decision support (CDS)</td>
</tr>
<tr>
<td>4.</td>
<td>Provide care using population health management approaches</td>
</tr>
<tr>
<td>5.</td>
<td>Protect patient privacy and security</td>
</tr>
<tr>
<td>6.</td>
<td>Use information technology to improve patient safety</td>
</tr>
<tr>
<td>7.</td>
<td>Engage in quality measurement selection and improvement</td>
</tr>
<tr>
<td>8.</td>
<td>Use health information exchange (HIE) to identify and access patient information across clinical settings</td>
</tr>
<tr>
<td>9.</td>
<td>Engage patients to improve their health and care delivery through personal health records and patient portals</td>
</tr>
<tr>
<td>10.</td>
<td>Maintain professionalism through use of information technology tools</td>
</tr>
<tr>
<td>11.</td>
<td>Provide clinical care via telemedicine and refer patients as indicated</td>
</tr>
<tr>
<td>12.</td>
<td>Apply personalized/precision medicine</td>
</tr>
<tr>
<td>13.</td>
<td>Participate in practice-based clinical and translational research</td>
</tr>
<tr>
<td>14.</td>
<td>Apply machine learning applications in clinical care</td>
</tr>
</tbody>
</table>
Professional Certification

Should there be certification of HIT or informatics professionals? Professional certification has been present in nursing and HIM for decades. Certification in nursing informatics is administered by the American Nurses Credentialing Center and requires a combination of a minimum of a bachelor’s degree in nursing along with practice experience.19

HIM has a variety of certifications administered by the American Health Information Management Association (AHMIA).20 The two main certifications require formal education:

- Registered Health Information Administrator (RHIA) – baccalaureate degree
- Registered Health Information Technician (RHIT) – associate degree

Other certifications are more focused and require a base education or certification plus passing a competency exam:

- Certified Coding Specialist/Associate (CCS, CCA, CCS-P)
- Certified Health Data Analyst (CHDA)
- Certified in Healthcare Privacy and Security (CHPS)
- Clinical Documentation Improvement Professional (CDIP)

A more recent certification has been implemented for physicians using the formal board certification process. Clinical informatics has been designated as a subspecialty of all medical specialties. This requires a physician to be certified in one of the 23 primary medical specialties, such as internal medicine, family medicine, pathology, and radiology. The vision and plan for the subspecialty was published by AMIA in 2009 and included core knowledge content (79) and fellowship training requirements.94 The subspecialty was formally approved by the American Board of Medical Specialties (ABMS) in 2011.78

As with all new medical specialties and subspecialties, initial certification was allowed via a “grandfathering” process, where those with experience working in the field would be eligible to take the board certification exam. After this period the only pathway to board certification would be through a fellowship accredited by the Accreditation Council for Graduate Medical Education (ACGME).22 The certification is administered by the American Board of Preventive Medicine (ABPM)23 for all specialties except pathology, whose certification is administered by the American Board of Pathology (ABP).24

Certification has been offered annually since 2013, and there are now over 2000 physicians who are board-certified.95 There are now over 50 fellowship programs, a list of which is maintained by AMIA.25 Early experiences in fellowship programs were described by Longhurst et al.96 Clinical informatics fellows have been found to make important contributions to healthcare systems, such as in their COVID-19 responses.97 A number of innovations in programs are being pursued, such as combined fellowships with other subspecialties.98

One of the downsides to the in-person fellowship requirement is its generally being limited to those early in career who can pursue such training before establishing careers and/or families. This author has called for more flexible training opportunities to pursue board certification.99

Of course, physicians are not the only professionals who work in health informatics. After several years of planning, AMIA implemented a professional certification for the rest of the informatics field, i.e., moving beyond just board certification for eligible physicians.100 The AMIA Health Informatics Certification (AHIC, formerly Advanced Health Informatics Certification)26 is open to all who have a master’s or doctoral degree in health informatics or a related discipline, the certification process is not conferred upon initial completion of one’s education. Rather, individuals also need to have completed qualifying work experience to be eligible for certification. This is different from some fields, such as medicine, including the clinical informatics subspecialty, where one takes the board certification exam shortly after completing formal training. There are a number of healthcare disciplines that require significant work experience for certification, such as some of the advanced certifications offered by the ANCC noted above. The content of the certification exam will be based on the health informatics workforce analysis described earlier.84

Those who are certified will be designated as ACHIP, the AMIA Certified Health Informatics Professional. The qualifications for AHIC have two tracks of eligibility.

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19 https://www.nursingworld.org/our-certifications/informatics-nurse/
20 https://www.ahima.org/certification
21 https://www.abms.org/
22 https://www.acgme.org/
23 https://www.theabpm.org/become-certified/subspecialties/clinical-informatics/
24 https://www.abpath.org/index.php/to-become-certified/requirements-for-certification?id=40
25 https://amia.org/careers-certifications/clinical-informatics-subspecialty
26 https://amia.org/careers-certifications/amia-health-informatics-certification-ahic
Chapter 1: Introduction to Biomedical and Health Informatics

Track 1 is for those who have a graduate degree in a health informatics-related area, e.g., health informatics, biomedical informatics, nursing informatics, public health informatics, translational bioinformatics, etc. Track 2 is for those who have a graduate degree in a related field, e.g., health professions such as nursing, pharmacy, and medicine, and other fields such as computer science and public health. The work time required for those in Track 1 is 50-100% work time over the last four of six years or 20-49% time over the last six of eight years. The work time required for those in Track 2 is 50-100% work time over the last six of eight years or 20-49% time over the last eight of 10 years.

There are additional HIT certifications offered by HIMSS. These include the original Certified Professional in Healthcare Information & Management Systems (CPHIMS), the entry level to CPHIMS Certified Associate in Healthcare Information & Management Systems (CAHIMS), and the more recent Certified Professional in Digital Health Transformation Strategy (CPDHTS).

In addition to those who work in informatics professionally are others who use informatics in their health or healthcare-related work or activities. The informatics competencies for physicians and medical students were first addressed by the AAMC Medical School Objectives Project. Patients need knowledge and skills in informatics too, as 58% of US adults look online for health information and 35% attempt to diagnose illness in that manner. There have also been defined competencies in informatics for clinical and translational scientists, next-generation research scientists, and nurses.

Informatics also plays an important role in health professions education. Safran noted that “informatics training for clinicians is more important than hardware and software.” Fridsma stated that health informatics is a “required skill for 21st century clinicians.” There have been publications around competencies, curricula, and challenges for medical education.

Resources for Informatics

There are many resources for BMHI. This section will describe specialty associations, textbooks (listed in Additional Readings), journals, conferences, Web sites, blogs, podcasts, and educational opportunities.

The premiere professional association is the American Medical Informatics Association (AMIA). The mission of AMIA is to advance the informatics profession relating to health and disease. To this end it advances the use of health information and communications technology in clinical care and clinical research, personal health management, public health/population, and translational science with the ultimate objective of improving health.

Another critical organization for BMHI is the US National Library of Medicine (NLM). One of the institutes and centers of the National Institutes of Health (NIH), the NLM is both the world’s medical library as well as a leading funded of research and training in BMHI. After being led for three decades by Dr. Donald Lindberg, its Director since 2016 has been Dr. Patricia Brennan.

There are other professional organizations devoted to related areas of BMHI and HIT:
- Healthcare Information and Management Systems Society (HIMSS)
- American Health Information Management Association (AHIMA)
- Association of Medical Directors of Information Systems (AMDIS)
- Alliance for Nursing Informatics (ANI)
- Public Health Informatics Institute (PHII)
- International Society for Computational Biology (ISCB)
- Society for Imaging Informatics in Medicine (SIIM)
- Association for Computing Machinery (ACM)
- Medical Library Association (MLA)
- American Medical Association (AMA)
- American Nurses Association (ANA)
- Association of American Medical Colleges (AAMC)
- American College of Physicians (ACP)

Resources

27 https://www.himss.org/resources-certification/overview
28 https://www.amia.org/
• American Academy of Family Physicians (AAFP)\textsuperscript{43}

As with any scientific discipline, an important source of information is scientific journals, which publish the leading research in the field. Prominent informatics journals include:

• Journals of AMIA
  ◦ Journal of the American Medical Informatics Association (AMIA)
  ◦ JAMIA Open
• Methods of Information in Medicine (MIM)
• International Journal of Medical Informatics (IJMI)
• Journal of Medical Internet Research (JMIR)
• JMIR Medical Informatics
• Journal of Biomedical Informatics (JBI)
• Applied Clinical Informatics (ACI)
  ◦ ACI Open
• BMJ Health & Care Informatics
• Bioinformatics
• Journal of Digital Imaging (JDI)
• Biomed Central (BMC)
  ◦ BMC Medical Informatics and Decision Making
  ◦ BMC Bioinformatics

Another important source of information is scientific conferences, where research as well as panels, tutorials, and workshops on the state-of-the-art of the field are presented:

• AMIA meetings
  ◦ Annual Symposium
  ◦ Informatics Summit
  ◦ Clinical Informatics Conference
• MedInfo (biennial meeting of IMIA)
• HIMSS, national meeting and local chapters
• AMDIS Physician-Computer Connection
• Pacific Symposium on Biocomputing (PSB)
• International Society for Computational Biology (ISCB)

A great deal of information can be found on Web sites. Particularly authoritative Web sites include those from the US government:

• Office of the National Coordinator for Health IT (ONC)\textsuperscript{44} – including their 2020-2025 strategic plan\textsuperscript{45}
• National Library of Medicine\textsuperscript{46} – including their 2017-2027 strategic plan\textsuperscript{47}
• ONC Health IT Playbook\textsuperscript{48}
• ONC HIT curriculum\textsuperscript{49}
• Agency for Healthcare Research & Quality (AHRQ) Digital Healthcare Research\textsuperscript{50}
• AHRQ US Health Information Knowledgebase\textsuperscript{51}

Some other Web sites of note include:

• HealthIT Answers\textsuperscript{52}
• Clinfowiki\textsuperscript{53}

One can also keep up to date from various email lists, including those from organizations such as AMIA, HIMSS, AMDIS, etc. Others include:

• HISTalk\textsuperscript{54}
• HIT Strategist\textsuperscript{55}

There are a number of blogs:

• Mayo Clinic Platform\textsuperscript{56} (formerly Geek Doctor from John Halamka)\textsuperscript{57}
• Healthcare Standards (Keith Boone)\textsuperscript{58}
• Health IT Buzz (ONC)\textsuperscript{59}
• Informatics Professor (William Hersh)\textsuperscript{60}
• CMIO Perspective (Dirk Stanley)\textsuperscript{61}
• From the Mezzanine (Patricia Brennan)\textsuperscript{62}

One can also keep up via podcasts:

• AMIA
  ◦ Women in AMIA For Your Informatics
  ◦ AMIA Clinical Informatics Fellows (ACIF) Go-Live\textsuperscript{63}
• Kevin Johnson, Informatics in the Round\textsuperscript{64}
• Jason Moore, Biomedical Informatics Roundtable\textsuperscript{65}
• Healthcare Interoperability\textsuperscript{66}

\textsuperscript{43} https://www.aafp.org/
\textsuperscript{44} https://www.healthit.gov/
\textsuperscript{45} https://www.healthit.gov/topic/2020-2025-federal-health-it-strategic-plan
\textsuperscript{46} https://www.nlm.nih.gov/
\textsuperscript{48} https://www.healthit.gov/playbook/
\textsuperscript{49} https://www.healthit.gov/topic/health-it-resources/health-it-curriculum-resources-educators
\textsuperscript{50} https://digital.ahrq.gov/
\textsuperscript{51} https://ushik.ahrq.gov/mdr/portals
\textsuperscript{52} https://www.healthitanswers.net/
\textsuperscript{53} http://clinfowiki.org
\textsuperscript{54} https://histalk2.com/
\textsuperscript{55} https://www.modernhealthcare.com/topic/healthcare-it-strategist
\textsuperscript{56} https://mayoclinicplatform.org/blog/
\textsuperscript{57} https://geekdoctor.blogspot.com/
\textsuperscript{58} https://motorcycleguy.blogspot.com/
\textsuperscript{59} https://www.healthit.gov/buzz-blog
\textsuperscript{60} https://informaticsprofessor.blogspot.com/
\textsuperscript{61} https://www.dirkstanley.com/
\textsuperscript{62} https://nlmdirector.nlm.nih.gov/
\textsuperscript{63} https://amia.org/news-publications/podcasts/for-your-informatics
\textsuperscript{64} https://amia.org/news-publications/podcasts/acif-go-live
\textsuperscript{65} https://kevinjohnsonmd.podbean.com/
\textsuperscript{66} http://bmipodcast.org/
\textsuperscript{67} https://podcasts.apple.com/us/podcast/exploring-healthcare-interoperability/id1570910011
There are many opportunities for education and training in BMHI and other flavors of informatics. It must be remembered that informatics is an inter-disciplinary field. As such, there are many programs with diverse curricula. Programs come in many flavors: clinical, biomedical, health, bio-, nursing, etc. Education historically focused on academics but expanded to meet needs and opportunities for practitioners and users. There has been a particularly large growth in applied master's degree programs with about 75-80 in US.(112) A major funder of research training is the NLM, which funds programs to train future researchers at doctoral (PhD) and postdoctoral levels at 16 universities.(113) Funding for education is also available from other sources, including institutions funding clinical informatics subspecialty fellowships. A catalog of US informatics programs is maintained on the AMIA Web site.69

Another well-known pathway for BMHI training is the AMIA 10x10 (“ten by ten”) program.70 The 10x10 program started when Dr. Charles Safran, who was then Chairman of AMIA, stated a need to train one physician and one nurse in informatics in the United States from each of the 6000 hospitals in the country. As this author already had an online graduate-level informatics course, he stated an aim to train 10,000 individuals in informatics by the year 2010 and launched the 10x10 course with AMIA in 2005.71 By 2022, over 3000 people have completed the OHSU offering of the 10x10 course.

Career pathways in informatics have diverse inputs and outputs.(4) As seen in Figure 2, people enter informatics study from a variety of academic backgrounds, go through education with others of diverse backgrounds, and then emerge into jobs that reflect their initial studies followed by informatics education. Figure 3 shows a cardinal rule (formula) of informatics education is that you emerge from education based on your prior knowledge and what you learn in your informatics program.

CONCLUSION

These are exciting times for BMHI, with many opportunities in a wide variety of settings. Attention must also be paid to the professional practice and education of informaticians. But the main focus of the field must be how to optimally use information and technology properly to advance human health.

ADDITIONAL READINGS

In addition to this textbook are a number of the prominent general textbooks:

Shortliffe and Cimino (eds.), *Biomedical Informatics: Computer Applications in Health Care and Biomedicine (5th Edition)*, Springer, 2021

Braunstein, *Contemporary Health Informatics*. Chicago, IL, AHIMA Press, 2016

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69 https://amia.org/careers-certifications/informatics-academic-programs
70 https://amia.org/education-events/amia-10x10-virtual-courses
71 https://dmice.ohsu.edu/hersh/10x10.html
Davis, Foundations of Health Information Management (5th Edition), Elsevier, 2019
There are also many specialty textbooks that cover specific areas of informatics or related areas:
Hersh, Information Retrieval: A Biomedical and Health Perspective (4th Edition), Springer, 2020
Bui and Taira (eds.), Medical Imaging Informatics, Springer, 2010
Reddy and Agarwal (eds.), Healthcare Data Analytics, Chapman & Hall, 2015
Sayles, Introduction to Information Systems for Health Information Technology (2nd Edition), 2018
Lesk, Introduction to Bioinformatics (5th Edition), Oxford University Press, 2019
Richesson and Andrews (eds.), Clinical Research Informatics (2nd Edition), Springer, 2019
Magnuson and Dixon (eds.), Public Health Informatics and Information Systems (3rd Edition), Springer, 2020
Skochelak et al. (eds.), Health Systems Science (2nd Edition), Elsevier, 2020
Saba and McCormick (eds.). Essentials of Nursing Informatics (7th Edition), McGraw-Hill, 2021
Branstetter, Practical Imaging Informatics: Foundations and Applications for Medical Imaging, Springer, 2021
Friedman, Wyatt, and Ash, Evaluation Methods in Biomedical and Health Informatics (3rd Edition), Springer, 2022
Braunstein, Health Informatics on FHIR: How HL7’s API is Transforming Healthcare (2nd Edition), Springer, 2022

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Chapter 1: Introduction to Biomedical and Health Informatics


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A Short History of Biomedical and Health Informatics

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LEARNING OBJECTIVES
After reading this chapter, the reader will be able to:
1. Describe key historical events and trends for biomedical and health informatics
2. Discuss current accomplishments and challenges in the context of history of the field

INTRODUCTION

The goal of this chapter is to provide a short historical overview of the biomedical and health informatics field. Such history establishes the context of many current developments in the field, including early work in artificial intelligence, electronic health records, and access to information. We will also cover advancements in genomics and bioinformatics, the important Institute of Medicine reports, and the HITECH Act. We will also discuss some backlash toward informatics and some recalibration that has occurred. Finally, we will highlight some of the impacts of COVID-19 and then look at some key challenges ahead.

ORIGINS

A 1959 paper by Ledley and Lusted is often attributed as the scientific origin of the field. (1) This paper aimed to model and understand physician reasoning in medical diagnosis and incorporated three elements. First was symbolic logic, the notion of representing concepts in patient diagnosis, such as the findings of evaluating the patient, the test results and diagnoses made. These were then combined with probabilities, giving the likelihood of outcomes, such as the likelihood of a diagnosis based on those concepts or symbols. The third element of this process was based on value theory, which would quantify the complexity of values that go into medical decision making. This paper led to early attempts at computer-based decision making in medicine in the 1960s, and a citation analysis of this paper done in the 1980s showed that it had significant influence on the field.(2)

The Ledley and Lusted paper gave rise to what might be described as the first era of artificial intelligence (AI) in healthcare. The focus of early systems was on developing handcrafted knowledge bases that would define either the rules or other algorithms that would aim to provide artificial intelligence of medical decision making. Another well-known early paper came from Warner et al., who developed a mathematical model for diagnosing congenital heart disease.(3) This system predicted diagnosis with the highest conditional probability given a set of symptoms of the patient.

Another early line of work came from Weed, who developed so-called problem-knowledge couplers that aimed to connect patient findings and diagnoses.(4) From these emerged the so-called expert systems, which was a major part of the work of early AI. Expert systems were computer programs that were designed to mimic human expertise. Early work focused on rule-based systems, developing large numbers of rules that would guide the computer to make a diagnosis. This work was the PhD dissertation of Shortliffe, one of the early
leaders in the field, (5) and led to subsequent work by other researchers in trying to build systems that could mimic human expertise in diagnosis and treatment. (6) Another early approach was the development of expert systems that used scoring algorithms. (7,8) These systems had disease profiles of patients’ findings, test results, and their likelihood in diseases. The systems would be used to generate a list of diseases with the highest-ranking disease being the most likely diagnosis.

All of these systems, whether scoring-based or rule-based, were very limited in the clinical areas that they covered. In addition, the computers of the time were slow and cumbersome to use. Most of them used text-based interfaces and were not easy to use or integrate in clinical practice. Miller, who was involved in the development of Internist-1, (8) also noted that these systems were attempting to act like a Greek Oracle, to be the purveyor of truth, as opposed to being tools that would help physicians make better diagnoses. (9) Miller called for the demise of the Greek Oracle approach to AI. At the same time, the first era of artificial intelligence, including beyond medicine, was winding down and entering what some called the “AI winter.” What did come out of these early systems was more focused clinical decision support, the rules we see in electronic health record systems in modern times, and less of the Greek Oracle all-knowing approach. (10)

**EARLY ELECTRONIC HEALTH RECORD SYSTEMS**

This era also saw some of the early electronic health record (EHR) systems, which were, for the most part, text-based and slow and cumbersome to use, but did achieve widespread use, at least in the institutions in which they were developed. The COSTAR system was developed at Massachusetts General Hospital and made its way into a local HMO, Harvard Community Health Plan. (11) COSTAR was built using the MUMPS programming language, which is still used in the VA and Epic systems. (12) There were also EHR systems such as HELP from the University of Utah, (13) TMR from Duke, (14) and the Regenstrief system at Indiana, (15) which were used mostly locally at these institutions. There was an early system developed at El Camino Hospital in Southern California that became the Technicon and later TRS system. (16) Of all these systems, the only one that’s still in significant, widespread use is the VistA system developed by the Veterans Administration. On top of VistA sits CPRS, or Computerized Patient Records System. (17) VistA is used across the entire VA system to this day, although is being phased out and replaced by the Cerner EHR system (as of 2021).

**ROLE OF THE NATIONAL LIBRARY OF MEDICINE**

Another important organization, both in the history and the current activity of the informatics field, is the U.S. National Library of Medicine (NLM), (1) which is part of the National Institutes of Health (NIH). (2) NLM played a critical role in early informatics research, database development, and education and training. The NLM was led for over 30 years by the late Dr. Donald Lindberg, (18,19) who stepped down in 2016 and passed the torch to Dr. Patricia Brennan. (20) The NLM played a critical role in the early information retrieval systems, initially for accessing bibliographic databases, but later moving on to full text and other kinds of knowledge resources. The NLM was a longtime publisher of Index Medicus, which was a paper-based method for accessing biomedical literature consisting of large catalogs on the shelves of most medical libraries. NLM also pioneered the development of early electronic systems for accessing biomedical literature. This started with the ELHILL system, (21) which had a command-line interface, then the NLM moved to PC-based systems with the development of the Grateful Med system, (22) and then moved to a Web-based system in PubMed that most use now to search the biomedical literature. (3) NLM has also been a leader in developing standards for terminology (23) and has also been a funder of research as well as training grants and other types of education in informatics. (24)

**SEQUENCING THE HUMAN GENOME**

Another important historical development for informatics was the sequencing of the human genome. This gave rise to many of the techniques of bioinformatics and much of the expanded work that bioinformatics specialists currently do. The Human Genome Project was started in 1988, an effort to sequence the entire human genome. It took over a decade, and in 2001, the NIH published the first draft of the human genome (25) simultaneously with an effort that started later from a private company called Celera Genomics. (26) The Human Genome Project was deemed to be completed in 2003, (27) although the

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1 https://www.nlm.nih.gov/
2 https://www.nih.gov/
Entire sequence was not elucidated until recently. (28) Since the initial sequencing, the project has been applied to more and more humans, which has greatly increased understanding of the variation as well as complexity of the human genome.

There continue to be advances in bioinformatics. There is next-generation sequencing, which enables highly accurate and inexpensive sequencing of genomes, including the human genome. (29) There are other biomolecular technologies, such as measurement of gene expression, in particular, how much the messenger RNA is expressed from the DNA in living organisms. (30) There are also advances in understanding of the structure and function of proteins.

Related advances have been to demystify other so-called -omes and -omics. There is proteomics, which studies protein structure and function. There is transcriptomics, which studies the expression or transcription of DNA. There has been an explosion of knowledge around the microbiome, which is all of the microorganisms that inhabit larger living organisms. There has also been characterization of the phenotype, which is the manifestation of characteristics and diseases and so forth of living organisms, which can then be mapped to the genotype. In the case of humans, this takes us full circle from clinical data to the original genome and phenotype.

There are also many data resources now available from a wide variety of organizations. Most prominent among these, the NLM’s National Center for Biotechnology Information (NCBI). There is a journal, Nucleic Acids Research, that has an annual database issue, with updates on many of the key data sources. These advances are manifested in precision medicine, where there is aim for diagnosis and treatment based on highly precise molecular components of organisms, including gene variants, the microbiome, and more. (31,32) The year 2021 marked the 20-year anniversary of the sequencing of the human genome, and a couple of retrospectives noted both the accomplishments of this effort but also elucidated a number of challenges still ahead, both in science and policy. (33,34)

INTERNET AND WORLD WIDE WEB

Another important development for the informatics field was the emergence of the Internet and World Wide Web. (35) The Internet emerged in the 1980s as a confluence of a number of different computer networks that enabled much more widespread communication. The initial use of the Internet focused on sharing information, things like file transfer and email. Of course, the Internet became much more prominent in society with the development of the World Wide Web, an application that ran on the Internet that enabled the widespread dissemination of information literally across the globe. The Internet saw an initial boom and bust in the dotcom era of the 1990s, but subsequently developed more sustainable business models. The Internet and Web are now ubiquitous around the world, whether accessed through wired (also called broadband connections) or wireless (through Wi-Fi and cellular) connectivity.

INSTITUTE OF MEDICINE

Also important in the history of informatics has been the thought leadership and vision provided by various reports from the Institute of Medicine (IOM), which is now called the National Academy of Medicine (NAM). The reports in the 1990s and early 21st century were critical and high profile for BMHI. The first IOM report on an informatics topic was the Computer-Based Patient Record, published initially in 1991 and subsequently updated in 1997. (36) This report noted that paper records of the time were illegible, inefficient and error prone, and called for the computer-based record being vital to modern healthcare. Another important IOM report was For the Record, which was the first report to raise visibility about issues of privacy and security of EHRs. (37) This report noted that the benefits of electronic health information could be compromised by its inadequate protection, and this report informed the emerging HIPAA privacy and security legislation that emerged in the late 1990s. Another important report was Networking Health, (38) which noted the emergence of the Internet and other computer networks recognizing their value and also pointing out that we didn’t need, as some had advocated, a separate Internet for health. Probably the most high-profile IOM report was To Err is Human or the so-called errors report. (39) This report brought to light the fact that medical errors are common and are really a systems problem as opposed to being problems of any one or any number of individuals. This gave rise to the patient safety movement.

The initial IOM reports identifying the problems were then followed by reports that looked at solutions. One of these was Crossing the Quality Chasm, (40) which set out to define a set of aims and rules for high-quality 21st century healthcare. The aims advocated for high-quality care were that healthcare be:

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5 https://academic.oup.com/nar
• Safe – avoid injuries from care intended to help
• Effective – provide service based on scientific knowledge and avoid care unlikely to benefit
• Patient-centered – care respectful of patients’ preferences, needs, and values
• Timely – reduce waits and delays in care
• Efficient – avoid waste of equipment, supplies, and energy
• Equitable – provide care that does not vary based on personal characteristics

Some more recent IOM reports that have been highly influential include the reports around the learning health system, the idea that healthcare should learn from what it does, in particular, collecting data, looking at outcomes, and acting accordingly.(41) We should strive to build a learning health system that would feature transparency of data and information, rewarding outcomes and value, and not just rewarding clinicians and hospitals for the volume of care provided.(42) We should also promptly identify errors and correct them. The IOM was also early in noting that health IT systems could not only be beneficial, but could also introduce error and cause harm, and advocated for means to design and properly use IT systems to avoid harm and provide benefit.(43)

HEALTH INFORMATION TECHNOLOGY FOR CLINICAL AND ECONOMIC HEALTH (HITECH) ACT

A watershed event for BMHI was the Health Information Technology for Clinical and Economic Health (HITECH) Act. In the late 1990s and into the new century, there was a growing amount of research supporting the value of the EHR and CDS for improving the quality and safety of care. In fact, President George W. Bush mentioned the potential value for health IT in his State of the Union addresses in the years between 2004 and 2007. Then in 2008, the Great Recession occurred, and there was a significant economic downturn leading to the need for an economic stimulus package. This was right after the election of President Barack Obama and one of his early actions in office was to lead the passage of the American Recovery and Reinvestment Act (ARRA). ARRA included the HITECH Act that allocated about $30 billion for incentives for the adoption and the “meaningful use” of the EHR. There was additional funding of about $2 billion for investment in health information exchange, regional extension centers, workforce development, and informatics research.(44–46)

A combination of advances in bioinformatics and adoption of EHRs and other sources of data becoming available gave rise to the so-called second era of AI.(47,48) AI has been reinvigorated largely on the success of machine learning, which has occurred because of the increased availability of data, more powerful computers, and advances in the science, particularly in the use of so-called deep learning.(49) Rather than humans building rules and scoring algorithms, machine learning systems learn from data. This has led to successes in image interpretation, natural language processing, and other tasks where computers have become highly effective at learning from growing bodies of data. A retrospective from Shortliffe, who was heavily involved in the first era of AI, provides some retrospectives from that era.(50) AI is also now drawing attention from leading policy bodies, such as a recent report from the National Academy of Medicine.(51)

SOME BACKLASH

There have also been areas of backlash in BMHI, particularly with clinicians and EHRs. The current systems that most clinicians use slow down their work. (52,53) The healthcare system and the organizations within it oftentimes prioritize non-clinical aspects of care with the EHR and all of these have been shown to lead to clinician burnout.(54) At the same time, there has been criticism over the lack of adoption of standards and interoperability, with the HITECH investment leading to systems that did not talk well to each other.(55) There are also concerns that go beyond healthcare, such as privacy and security in our modern world.(56)

Some of the backlash and other problems related to the HITECH Act were addressed in a comprehensive piece of legislation that was passed in 2016 called the 21st Century Cures Act.(57,58) Some viewed the health IT portion of the legislation as a recalibration or correction of some of the limitations in the original HITECH Act. (59) For example, many criticized the HITECH Act for paying insufficient attention to data interoperability. (60) In between the passage of the HITECH Act and later in the 2010s decade was the emergence of the app framework known as Substitutable Medical Apps, Reusable Technology (SMART).(61,62) The 21st Century Cures Act mandated the development of interoperability rules and an app framework, with the SMART on FHIR framework was chosen.(61) Another concern that arose out of the HITECH Act was the development of information blocking, sometimes by EHR vendors and other times by healthcare organizations.(63,64) As such, there were rules mandated to be developed in the 21st Century Cures Act that would prohibit information blocking.
**COVID-19 PANDEMIC**

Informatics, like many other areas of healthcare and many other areas of society, was profoundly impacted by the COVID-19 pandemic. One specific impact related to informatics is that US health information systems, especially its public health information systems, have not been up to the task of tracking the pandemic or our efforts to control it. (65) Another significant impact arose from the need for healthcare to become more virtual. This led to the relaxation of various kinds of rules around digital healthcare. For example, rules from the HIPAA Security rule that previously prohibited common communications platforms that were viewed as less secure, such as video conferencing applications like Zoom, FaceTime on iPhones, and others. (66) HIPAA security rules were relaxed at the onset of the pandemic to allow these channels to be used for patient care. This led to the rapid expansion of use of telemedicine and telehealth. (67) There were also some delays in the compliance dates for the 21st Century Cures rule to make them more achievable by most healthcare organizations. (68)

Another big impact related to informatics came from advances in “open science,” where scientific information and data are made more widely accessible. These are beneficial in getting science out more quickly to other scientists and the larger world, but in the case of COVID-19 resulted in the publication of suboptimal science with inadequate peer review and so-called fake news. (69) Some of the open science advances, like preprints of journal articles, where authors can post their articles before peer review, were abused. Somewhat related were the growing political attacks on science and scientists, especially with regards to public health measures for COVID-19. (70)

**CONCLUSION**

Historical perspectives can often inform the present by providing a context for the world as it is now. It also allows us to learn from the mistakes, and successes, of the past. Clearly, informatics must improve the usability of systems in clinical care, especially the EHR. It must provide access to data, information and knowledge for the betterment of people’s health and the healthcare they receive. We need to learn from the data we are collecting while protecting its privacy and security and must also find ways to integrate the new AI into healthcare professions and activities.

**ADDITIONAL READINGS**


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