Preface to the Fourth Edition

The world of biomedical research and health care has changed remarkably in the 25 years since the first edition of this book was undertaken. So too has the world of computing and communications and thus the underlying scientific issues that sit at the intersections among biomedical science, patient care, public health, and information technology. It is no longer necessary to argue that it has become impossible to practice modern medicine, or to conduct modern biological research, without information technologies. Since the initiation of the human genome project two decades ago, life scientists have been generating data at a rate that defies traditional methods for information management and data analysis. Health professionals also are constantly reminded that a large percentage of their activities relates to information management—for example, obtaining and recording information about patients, consulting colleagues, reading and assessing the scientific literature, planning diagnostic procedures, devising strategies for patient care, interpreting results of laboratory and radiologic studies, or conducting case-based and population-based research. It is complexity and uncertainty, plus society's overriding concern for patient well-being, and the resulting need for optimal decision making, that set medicine and health apart from many other information-intensive fields. Our desire to provide the best possible health and health care for our society gives a special significance to the effective organization and management of the huge bodies of data with which health professionals and biomedical researchers must deal. It also suggests the need for specialized approaches and for skilled scientists who are knowledgeable about human biology, clinical care, information technologies, and the scientific issues that drive the effective use of such technologies in the biomedical context.

Information Management in Biomedicine

The clinical and research influence of biomedical-computing systems is remarkably broad. Clinical information systems, which provide communication and information-management functions, are now installed in essentially all healthcare institutions. Physicians can search entire drug indexes in a few seconds, using the information provided by a computer program to anticipate harmful side effects or drug interactions. Electrocardiograms (ECGs) are typically analyzed initially by computer programs, and similar techniques are being applied for interpretation of pulmonary-function tests and a variety of

laboratory and radiologic abnormalities. Devices with embedded processors routinely monitor patients and provide warnings in critical-care settings, such as the intensive-care unit (ICU) or the operating room. Both biomedical researchers and clinicians regularly use computer programs to search the medical literature, and modern clinical research would be severely hampered without computer-based data-storage techniques and statistical analysis systems. Advanced decision-support tools also are emerging from research laboratories, are being integrated with patient-care systems, and are beginning to have a profound effect on the way medicine is practiced.

Despite this extensive use of computers in healthcare settings and biomedical research, and a resulting expansion of interest in learning more about biomedical computing, many life scientists, health-science students, and professionals have found it difficult to obtain a comprehensive and rigorous, but nontechnical, overview of the field. Both practitioners and basic scientists are recognizing that thorough preparation for their professional futures requires that they gain an understanding of the state of the art in biomedical computing, of the current and future capabilities *and* limitations of the technology, and of the way in which such developments fit within the scientific, social, and financial context of biomedicine and our healthcare system. In turn, the future of the biomedical computing field will be largely determined by how well health professionals and biomedical scientists are prepared to guide and to capitalize upon the discipline's development. This book is intended to meet this growing need for such well-equipped professionals. The first edition appeared in 1990 (published by Addison-Wesley) and was used extensively in courses on medical informatics throughout the world. It was updated with a second edition (published by Springer) in 2000, responding to the remarkable changes that occurred during the 1990s, most notably the introduction of the World Wide Web and its impact on adoption and acceptance of the Internet. The third edition (again published by Springer) appeared in 2006, reflecting the ongoing rapid evolution of both technology and health- and biomedically-related applications, plus the emerging government recognition of the key role that health information technology would need to play in promoting quality, safety, and efficiency in patient care. With that edition the title of the book was changed from *Medical Informatics* to *Biomedical Informatics*, reflecting (as is discussed in Chap. 1) both the increasing breadth of the basic discipline and the evolving new name for academic units, societies, research programs, and publications in the field. Like the first three editions, this new version provides a conceptual framework for learning about the science that underlies applications of computing and communications technology in biomedicine and health care, for understanding the state of the art in computer applications in clinical care and biology, for critiquing existing systems, and for anticipating future directions that the field may take.

In many respects, this new edition is very different from its predecessors, however. Most importantly, it reflects the remarkable changes in computing and communications that continue to occur, most notably in communications, networking, and health information technology policy, and the exploding interest in the role that information technology must play in systems integration and the melding of genomics with innovations in clinical practice and

treatment. In addition, new chapters have been introduced, one (healthcare financing) was eliminated, while others have been revamped. We have introduced new chapters on the health information infrastructure, consumer health informatics, telemedicine, translational bioinformatics, clinical research informatics, and health information technology policy. Most of the previous chapters have undergone extensive revisions. Those readers who are familiar with the first three editions will find that the organization and philosophy are unchanged, but the content is either new or extensively updated.[1]

This book differs from other introductions to the field in its broad coverage and in its emphasis on the field's conceptual underpinnings rather than on technical details. Our book presumes no health- or computer-science background, but it does assume that you are interested in a comprehensive summary of the field that stresses the underlying concepts, and that introduces technical details only to the extent that they are necessary to meet the principal goal. It thus differs from an impressive early text in the field (Ledley 1965) that emphasized technical details but did not dwell on the broader social and clinical context in which biomedical computing systems are developed and implemented.

Overview and Guide to Use of This book

This book is written as a text so that it can be used in formal courses, but we have adopted a broad view of the population for whom it is intended. Thus, it may be used not only by students of medicine and of the other health professions, but also as an introductory text by future biomedical informatics professionals, as well as for self-study and for reference by practitioners. The book is probably too detailed for use in a 2- or 3-day continuing-education course, although it could be introduced as a reference for further independent study.

Our principal goal in writing this text is to teach *concepts* in biomedical informatics—the study of biomedical information and its use in decision making—and to illustrate them in the context of descriptions of representative systems that are in use today or that taught us lessons in the past. As you will see, biomedical informatics is more than the study of computers in biomedicine, and we have organized the book to emphasize that point. Chapter 1 first sets the stage for the rest of the book by providing a glimpse of the future, defining important terms and concepts, describing the content of the field, explaining the connections between biomedical informatics and related disciplines, and discussing the forces that have influenced research in biomedical informatics and its integration into clinical practice and biological research.

[1] As with the first three editions, this book has tended to draw both its examples and it contributors from North America. There is excellent work in other parts of the world as well, although variations in healthcare systems, and especially financing, do tend to change the way in which systems evolve from one country to the next. The basic concepts are identical, however, so the book is intended to be useful in educational programs in other parts of the world as well.

Broad issues regarding the nature of data, information, and knowledge pervade all areas of application, as do concepts related to optimal decision making. Chapters 2 and 3 focus on these topics but mention computers only in passing. They serve as the foundation for all that follows. Chapter 4 on cognitive science issues enhances the discussions in Chaps. 2 and 3, pointing out that decision making and behavior are deeply rooted in the ways in which information is processed by the human mind. Key concepts underlying system design, human-computer interaction, patient safety, educational technology, and decision making are introduced in this chapter.

Chapters 5 and 6 introduce the central notions of computer architectures and software engineering that are important for understanding the applications described later. Also included is a discussion of computer-system design, with explanations of important issues for you to consider when you read about specific applications and systems throughout the remainder of this book.

Chapter 7 summarizes the issues of standards development, focusing in particular on data exchange and issues related to sharing of clinical data. This important and rapidly evolving topic warrants inclusion given the evolution of the health information exchange, institutional system integration challenges, and the increasingly central role of standards in enabling clinical systems to have their desired influence on healthcare practices.

Chapter 8 addresses a topic of increasing practical relevance in both the clinical and biological worlds: natural language understanding and the processing of biomedical texts. The importance of these methods is clear when one considers the amount of information contained in free-text dictated notes or in the published biomedical literature. Even with efforts to encourage structured data entry in clinical systems, there will likely always be an important role for techniques that allow computer systems to extract meaning from natural language documents.

Chapter 9 is a comprehensive introduction to the conceptual underpinnings of biomedical and clinical image capture, analysis, interpretation and use. This overview of the basic issues and imaging modalities serves as background for Chap. 20, which deals with imaging applications issues, highlighted in the world of radiological imaging and image management (e.g., in picture archiving and communication systems).

Chapter 10 addresses the key legal and ethical issues that have arisen when health information systems are considered. Then, in Chap. 11, the challenges associated with technology assessment and with the evaluation of clinical information systems are introduced.

Chapters 12–26 (which include several new chapters in this edition) survey many of the key biomedical areas in which computers are being used. Each chapter explains the conceptual and organizational issues in building that type of system, reviews the pertinent history, and examines the barriers to successful implementations.

Chapter 27 is a new chapter in the fourth edition, providing a summary of the rapidly evolving policy issues related to health information technology. Although the emphasis is on US government policy, there is some discussion of issues that clearly generalize both to states (in the US) and to other countries. The book concludes in Chap. 28 with a look to the future—a vision of how

informatics concepts, computers, and advanced communication devices one day may pervade every aspect of biomedical research and clinical practice.

The Study of Computer Applications in Biomedicine

The actual and potential uses of computers in health care and biomedicine form a remarkably broad and complex topic. However, just as you do not need to understand how a telephone or an ATM machine works to make good use of it and to tell when it is functioning poorly, we believe that technical biomedical-computing skills are not needed by health workers and life scientists who wish simply to become effective users of evolving information technologies. On the other hand, such technical skills are of course necessary for individuals with career commitment to developing information systems for biomedical and health environments. Thus, this book will neither teach you to be a programmer, nor show you how to fix a broken computer (although it might motivate you to learn how to do both). It also will not tell you about every important biomedical-computing system or application; we shall use an extensive bibliography to direct you to a wealth of literature where review articles and individual project reports can be found. We describe specific systems only as examples that can provide you with an understanding of the conceptual and organizational issues to be addressed in building systems for such uses. Examples also help to reveal the remaining barriers to successful implementations. Some of the application systems described in the book are well established, even in the commercial marketplace. Others are just beginning to be used broadly in biomedical settings. Several are still largely confined to the research laboratory.

Because we wish to emphasize the concepts underlying this field, we generally limit the discussion of technical implementation details. The computer-science issues can be learned from other courses and other textbooks. One exception, however, is our emphasis on the details of decision science as they relate to biomedical problem solving (Chaps. 3 and 22). These topics generally are not presented in computer-science courses, yet they play a central role in the intelligent use of biomedical data and knowledge. Sections on medical decision making and computer-assisted decision support accordingly include more technical detail than you will find in other chapters.

All chapters include an annotated list of Suggested Readings to which you can turn if you have a particular interest in a topic, and there is a comprehensive Bibliography, drawn from the individual chapters, at the end of the book. We use **boldface** print to indicate the key terms of each chapter; the definitions of these terms are included in the Glossary at the end of the book. Because many of the issues in biomedical informatics are conceptual, we have included Questions for Discussion at the end of each chapter. You will quickly discover that most of these questions do not have "right" answers. They are intended to illuminate key issues in the field and to motivate you to examine additional readings and new areas of research.

It is inherently limiting to learn about computer applications solely by reading about them. We accordingly encourage you to complement your

studies by seeing real systems in use—ideally by using them yourself. Your understanding of system limitations and of what *you* would do to improve a biomedical-computing system will be greatly enhanced if you have had personal experience with representative applications. Be aggressive in seeking opportunities to observe and use working systems.

In a field that is changing as rapidly as biomedical informatics is, it is difficult ever to feel that you have knowledge that is completely current. However, the conceptual basis for study changes much more slowly than do the detailed technological issues. Thus, the lessons you learn from this volume will provide you with a foundation on which you can continue to build in the years ahead.

The Need for a Course in Biomedical Informatics

A suggestion that new courses are needed in the curricula for students of the health professions is generally not met with enthusiasm. If anything, educators and students have been clamoring for *reduced* lecture time, for more emphasis on small group sessions, and for more free time for problem solving and reflection. A 1984 national survey by the Association of American Medical Colleges found that both medical students and their educators severely criticized the traditional emphasis on lectures and memorization. Yet the analysis of a panel on the General Professional Education of the Physician (GPEP) (Association of American Medical Colleges 1984) and several subsequent studies and reports have specifically identified biomedical informatics, including computer applications, as an area in which new educational opportunities need to be developed so that physicians and other health professionals will be better prepared for clinical practice. The AAMC recommended the formation of new academic units in biomedical informatics in our medical schools, and subsequent studies and reports have continued to stress the importance of the field and the need for its inclusion in the educational environments of health professionals.

The reason for this strong recommendation is clear: *The practice of medicine is inextricably entwined with the management of information*. In the past, practitioners handled medical information through resources such as the nearest hospital or medical-school library; personal collections of books, journals, and reprints; files of patient records; consultation with colleagues; manual office bookkeeping; and (all-too-often flawed) memorization. Although these techniques continue to be variably valuable, information technology is offering new methods for finding, filing, and sorting information: online bibliographic-retrieval systems, including full-text publications; personal computers, laptops, tablets, and smart phones, with database software to maintain personal information and commonly used references; office-practice and clinical information systems to capture, communicate, and preserve key elements of the health record; information retrieval and consultation systems to provide assistance when an answer to a question is needed rapidly; practice-management systems to integrate billing and receivable functions with other aspects of office or clinic organization; and other online information resources that help to reduce the

pressure to memorize in a field that defies total mastery of all but its narrowest aspects. With such a pervasive and inevitable role for computers in clinical practice, and with a growing failure of traditional techniques to deal with the rapidly increasing information-management needs of practitioners, it has become obvious to many people that an essential topic has emerged for study in schools that train medical and other health professionals.

What is less clear is how the subject should be taught, and to what extent it should be left for postgraduate education. We believe that topics in biomedical informatics are best taught and learned in the context of health-science training, which allows concepts from both the health sciences and informatics science to be integrated. Biomedical-computing novices are likely to have only limited opportunities for intensive study of the material once their health-professional training has been completed.

The format of biomedical informatics education is certain to evolve as faculty members are hired to develop it at more health-science schools, and as the emphasis on lectures as the primary teaching method continues to diminish. Computers will be used increasingly as teaching tools and as devices for communication, problem solving, and data sharing among students and faculty. In the meantime, key content in biomedical informatics will likely be taught largely in the classroom setting. This book is designed to be used in that kind of traditional course, although the Questions for Discussion also could be used to focus conversation in small seminars and working groups. As resources improve in schools and academic medical centers, integration of biomedical informatics topics into clinical experiences also will become more common. The eventual goal should be to provide instruction in biomedical informatics whenever this field is most relevant to the topic the student is studying. This aim requires educational opportunities throughout the years of formal training, supplemented by continuing-education programs after graduation.

The goal of integrating biomedicine and biomedical informatics is to provide a mechanism for increasing the sophistication of health professionals, so that they know and understand the available resources. They also should be familiar with biomedical computing's successes and failures, its research frontiers and its limitations, so that they can avoid repeating the mistakes of the past. Study of biomedical informatics also should improve their skills in information management and problem solving. With a suitable integration of hands-on computer experience, computer-based learning, courses in clinical problem solving, and study of the material in this volume, health-science students will be well prepared to make effective use of computer-based tools and information management in healthcare delivery.

The Need for Specialists in Biomedical Informatics

As mentioned, this book also is intended to be used as an introductory text in programs of study for people who intend to make their professional careers in biomedical informatics. If we have persuaded you that a course in biomedical

informatics is needed, then the requirement for trained faculty to teach the courses will be obvious. Some people might argue, however, that a course on this subject could be taught by a computer scientist who had an interest in biomedical computing, or by a physician or biologist who had taken a few computing courses. Indeed, in the past, most teaching—and research—has been undertaken by faculty trained primarily in one of the fields and later drawn to the other. Today, however, schools have come to realize the need for professionals trained specifically at the interfaces among biomedicine, biomedical informatics, and related disciplines such as computer science, statistics, cognitive science, health economics, and medical ethics. This book outlines a first course for students training for careers in the biomedical informatics field. We specifically address the need for an educational experience in which computing and information-science concepts are synthesized with biomedical issues regarding research, training, and clinical practice. It is the *integration* of the related disciplines that traditionally has been lacking in the educational opportunities available to students with career interests in biomedical informatics. If schools are to establish such courses and training programs (and there are growing numbers of examples of each), they clearly need educators who have a broad familiarity with the field and who can develop curricula for students of the health professions as well as of informatics itself.

The increasing introduction of computing techniques into biomedical environments will require that well-trained individuals be available not only to teach students, but also to design, develop, select, and manage the biomedical-computing systems of tomorrow. There is a wide range of context-dependent computing issues that people can appreciate only by working on problems defined by the healthcare setting and its constraints. The field's development has been hampered because there are relatively few trained personnel to design research programs, to carry out the experimental and developmental activities, and to provide academic leadership in biomedical informatics. A frequently cited problem is the difficulty a health professional (or a biologist) and a technically trained computer scientist experience when they try to communicate with one another. The vocabularies of the two fields are complex and have little overlap, and there is a process of acculturation to biomedicine that is difficult for computer scientists to appreciate through distant observation. Thus, interdisciplinary research and development projects are more likely to be successful when they are led by people who can effectively bridge the biomedical and computing fields. Such professionals often can facilitate sensitive communication among program personnel whose backgrounds and training differ substantially.

It is exciting to be working in a field that is maturing and that is having a beneficial effect on society. There is ample opportunity remaining for innovation as new technologies evolve and fundamental computing problems succumb to the creativity and hard work of our colleagues. In light of the

increasing sophistication and specialization required in computer science in general, it is hardly surprising that a new discipline should arise at that field's interface with biomedicine. This book is dedicated to clarifying the definition and to nurturing the effectiveness of that discipline: biomedical informatics.

New York, NY
Bethesda, MD

Edward H. Shortliffe
James J. Cimino
October 2013

Acknowledgments

In the 1980s, when I was based at Stanford University, I conferred with colleagues Larry Fagan and Gio Wiederhold and we decided to compile the first comprehensive textbook on what was then called medical informatics. As it turned out, none of us predicted the enormity of the task we were about to undertake. Our challenge was to create a multi-authored textbook that captured the collective expertise of leaders in the field yet was cohesive in content and style. The concept for the book first developed in 1982. We had begun to teach a course on computer applications in health care at Stanford's School of Medicine and had quickly determined that there was no comprehensive introductory text on the subject. Despite several published collections of research descriptions and subject reviews, none had been developed with the needs of a rigorous introductory course in mind.

The thought of writing a textbook was daunting due to the diversity of topics. None of us felt that he was sufficiently expert in the full range of important subjects for us to write the book ourselves. Yet we wanted to avoid putting together a collection of disconnected chapters containing assorted subject reviews. Thus, we decided to solicit contributions from leaders in the respective fields to be represented but to provide organizational guidelines in advance for each chapter. We also urged contributors to avoid writing subject reviews but, instead, to focus on the key conceptual topics in their field and to pick a handful of examples to illustrate their didactic points.

As the draft chapters began to come in, we realized that major editing would be required if we were to achieve our goals of cohesiveness and a uniform orientation across all the chapters. We were thus delighted when, in 1987, Leslie Perreault, a graduate of our training program, assumed responsibility for reworking the individual chapters to make an integral whole and for bringing the project to completion. The final product, published in 1990, was the result of many compromises, heavy editing, detailed rewriting, and numerous iterations. We were gratified by the positive response to the book when it finally appeared, and especially by the students of biomedical informatics who have often come to us at scientific meetings and told us about their appreciation of the book.

As the 1990s progressed, however, we began to realize that, despite our emphasis on basic concepts in the field (rather than a survey of existing systems), the volume was beginning to show its age. A great deal had changed since the initial chapters were written, and it became clear that a new edition would be required. The original editors discussed the project and decided that we should redesign the book, solicit updated chapters, and publish a new edition. Leslie Perreault by this time was a busy Director at First Consulting

Group in New York City and would not have as much time to devote to the project as she had when we did the first edition. With trepidation, in light of our knowledge of the work that would be involved, we embarked on the new project.

As before, the chapter authors did a marvelous job, trying to meet our deadlines, putting up with editing changes that were designed to bring a uniform style to the book, and contributing excellent chapters that nicely reflected the changes in the field in the preceding decade.

No sooner had the second edition appeared in print than we started to get inquiries about when the next update would appear. We began to realize that the maintenance of a textbook in a field such as biomedical informatics was nearly a constant, ongoing process. By this time I had moved to Columbia University and the initial group of editors had largely disbanded to take on other responsibilities, with Leslie Perreault no longer available. Accordingly, as plans for a third edition began to take shape, my Columbia colleague Jim Cimino joined me as the new associate editor, whereas Drs. Fagan, Wiederhold, and Perreault continued to be involved as chapter authors. Once again the authors did their best to try to meet our deadlines as the third edition took shape. This time we added several chapters, attempting to cover additional key topics that readers and authors had identified as being necessary enhancements to the earlier editions. We were once again extremely appreciative of all the authors' commitment and for the excellence of their work on behalf of the book and the field.

Predictably, it was only a short time after the publication of the third edition that we began to get queries about a fourth edition. We resisted for a year or two but it became clear that the third edition was becoming rapidly stale in some key areas and that there were new topics that were not in the book and needed to be added. With that in mind we, in consultation with Grant Weston from Springer's offices in London, agreed to embark on a fourth edition. Progress was slowed by my professional moves (to Phoenix, Arizona, then Houston, Texas, and then back to New York) with a very busy three-year stint as President and CEO of the American Medical Informatics Association. Similarly, Jim Cimino left Columbia to assume new responsibilities at the NIH Clinical Center in Bethesda, MD. With several new chapters in mind, and the need to change authors of some of the existing chapters due to retirements (this too will happen, even in a young field like informatics!), we began working on the fourth edition, finally completing the effort in early 2013.

The completed fourth edition reflects the work and support of many people in addition to the editors and chapter authors. Particular gratitude is owed to Maureen Alexander, our developmental editor whose rigorous attention to detail was crucial given the size and the complexity of the undertaking. At Springer we have been delighted to work on this edition with Grant Weston, who has been extremely supportive despite our missed deadlines. And I want to offer my sincere personal thanks to Jim Cimino, who has been a superb and talented collaborator in this effort for the last two editions. Without his hard work and expertise, we would still be struggling to complete the massive editing job associated with this now very long manuscript.

New York, NY Edward H. Shortliffe