

Preface

Medicine and health care are currently faced with a significant rise in their complexity. This is partly due to the progress made during the past three decades in the fundamental biological understanding of the causes of health and disease at the molecular, (sub)cellular, and organ level. It is also partly caused by the increased specialization of both biomedical research and clinical practice, and greater involvement of policy makers in health care to control costs. Promises made by biomedical researchers that their research results will have clinical impact, e.g., that cancer can be cured by immune therapy, have also increased expectations from society about what healthcare is able to deliver. However, it is rarely the case that a discovery at the molecular level has immediate consequences for the diagnosis and treatment of patients.

A major problem is that the progress made by the basic sciences increases the quantity of information that one has to deal with when making decisions at the level of the patient or health care in general. An additional problem is that this information arises from research at different levels: from the molecular level, via the subcellular level, at one end of the spectrum, to the patient and health-care level at the other end. How to bridge these different levels is currently unclear although it has given rise to the creation of yet another field: translational medicine.

However, although there are huge differences in the techniques and methods used by biomedical researchers, there is now an increasing tendency to share research results in terms of formal knowledge representation methods, such as ontologies, statistical models, network models, and mathematical models. As there is an urgent need for health-care professionals to make better decisions, computer-based support using this knowledge is now becoming increasingly important. It may also be the only way to integrate research results from the different parts of the spectrum of biomedical and clinical research.

Exploitation of knowledge technologies in biomedicine and health care, ranging from biological ontologies to computerized clinical practice guidelines, has been used as a solution to the aforementioned issues. However, it has been difficult to integrate knowledge from different levels, even when concerning a single disease.

Many different formal representations are being used at the output of biomedical research. Probabilistic methods, such as Bayesian networks, have proved themselves useful for problems where uncertainty is important, such as medical decision making and prognostics, but also in biology. Logic plays a key role as a basis for medical ontologies, but also in the formalization of important medical concepts such as diagnostics. Differential equations are popular for describing the dynamics of biological processes at the molecular level. These methods can be extended with all kinds of semantic concepts, such as space, in the biomedical domain. Space is an important concept when developing probabilistic models of, e.g., the spread of infectious disease, either in the hospital or in the community at large. Reasoning with time is already provided by differential equations, but can also be done in other formalisms, such as

probability theory and logic. Temporal reasoning is important in the context of personalized health care.

The aim of the book *Foundations of Biomedical Knowledge Representation* is to shed light on developments in knowledge representation at different levels of biomedical application, ranging from human biology to clinical guidelines, and using different techniques, from probability theory and differential equations to logic. While there is interdisciplinary cooperation between the different fields, there is a clear need for understanding the relationships of representation and reasoning among the different communities.

What the book will certainly make clear is that since the end of the 1970s, when knowledge representation and reasoning in the biomedical field became a separate area of research, huge progress has been made in the development of methods and tools that are finally able to have an impact on the way medicine is being practiced.

We wish to thank all the contributors to this book for their dedication in creating a truly outstanding account of modern methods in biomedical knowledge representation and reasoning.

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