Preface

The history of DNA sequencing and its use in biomedical research is brief but very impactful. First described in the mid-1970s, DNA sequencing emerged coincident with the field of molecular biology and has been used extensively to characterize the genomes of many species from viruses and bacteria to humans and plants. Once a cottage industry, it has rapidly grown to the industrial scale needed to map and decipher large, complex genomes like human with increasing accuracy and speed. The emergence of facile platforms that permit DNA sequencing to be carried out across a range of scale and throughput, as needed, to inform a multitude of research aims has largely been democratized. So-called next-generation sequencing, the topic of this book, has revolutionized biomedical research and increasingly is being employed as a diagnostic tool that is driving precision medicine across the spectrum of human diseases. The importance of this transition should not be overlooked and, indeed, was our impetus for planning the book around relevant topics described in the chapters. Genome sequencing as a diagnostic test will become routine in our lifetime, with NGS platforms already reaching practical clinical utility but with computational hurdles for robust analysis and scalable decision support remaining to be overcome.

As in any rapidly changing and evolving area of technology, it is quite difficult to represent the complete range of applications that have been facilitated by next-generation sequencing as these are rapidly emerging and evolving. Rather, we have largely focused on the basics of the technology and its application to biomedicine, emphasizing the translation from basic research to clinical diagnosis and its attendant difficulties, including the return of results. To this end, we thank the many authors who agreed to contribute their expertise and perspectives to written descriptions of next-generation sequencing, in spite of very busy schedules. We also thank Barbara Acosta for her patience and perseverance in seeing this book through from concept to completion. Finally, we thank readers of this book and hope that the breadth and scope of what is presented in these pages provides an informed view of how the initial concept of DNA sequencing is emerging as a component of evidence-based medicine.

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