Preface

The opportunity to edit this book coincided with a huge explosion in human genetics. The sequencing and analysis of an enormous amount of individual human genomes has made it possible to identify genes and genetic/genomic variants that exhibit associations with specific phenotypes. An impressive amount of genetic variation has already been underscored, yet the picture is becoming ever more complex. Which of these variants will have phenotypic consequences? Are the variant effects constitutive or do they surface only in response to specific environmental challenges? What is the contribution of single genes versus that of genomic makeup? Currently the main challenges are to discern correlative versus causative roles in genetic variation and to understand the physiological consequences of the identified variants. Concomitant with the explosion of genomic information came a revolution in the availability of CRISPR/Cas technologies, which make gene and genomic editing much easier than before.

Within this book we wanted to describe the world of genetic modeling, which is so crucial to the understanding of human genetics. The introductory chapter was conceived to give an idea of the current state of knowledge in human genetics, exemplifying the impressive genetic/genomic variability and complexity. This is followed by five chapters that describe specific models, going from smaller to bigger in size as we did not want to assign them any order of relevance. Every model organism—not just the ones described in this book but also others omitted because of a lack of space and time—can provide specific and unique answers, and should be thanked and respected for that. Within each of the chapters, the main idea was to describe not only the importance and the history behind each of the models but also to mention tools and strategies that could be used. The next two chapters describe methodology and tools that can be employed with any model organism, providing generalizable information. The final chapter aims to summarize the current situation and to discuss future challenges and directions within this field. The overall goal of this book is to present available modeling methods currently used to delineate phenotype-causative genes through

rigorous design and testing, and to make researchers enthusiastic about the field of human genetics and organism modeling.

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We had fun imagining, organizing, and writing this book. We hope you find it enjoyable, interesting, and helpful.