PREFACE

Wilhelm Johannsen introduced the conceptual distinction between genotype and phenotype in 1911, and since then biologists have tried to disentangle the fascinating connection between genotypes and phenotypes, that is how nucleotide changes produce observable changes in living organisms. The elucidation of the actual genes and mutations responsible for observable differences between individuals and species started in the 1980s and has revolutionized the study of evolution. A wide array of powerful methods have been developed in recent years to find the genes and the mutations responsible for evolutionary changes so that empirical data on the "loci of evolution" are accumulating at a fast pace. The field of evolutionary genetics has moved from pure theoretical computations of changes in allele frequencies in populations to assessments of the actual mutations that occurred in populations and that cause observable changes in phenotype, bringing up new ideas and new questions. This volume aims to provide an overview of current knowledge and ongoing research on the genes and the mutations responsible for phenotypic evolution.

Chapter 1 highlights the fact that the term "gene" can have two different meanings in biology research papers and tries to clarify the distinction between both, to avoid confusion and misconceptions. Both concepts of "gene" are still extremely used and useful in today's research and are part of two relevant frameworks for explaining the biological world. Chapters 2–4 examine the entire set of genes and mutations (about a hundred of mutations/genes for each chapter) that have been identified so far as responsible for particular phenotypes, *Drosophila* pigmentation patterns (Chapter 2), Crop seed and fruit retention (Chapter 3), and biotic interactions involving *Arabidopsis thaliana* (Chapter 4). These chapters are particularly representative of the current state of evolutionary genetics research. Drosophila pigmentation (Chapter 2) is a premier model that has led to important insights on the precise genetic modifications responsible for phenotypic evolution and on the predominance of *cis*-regulatory changes. Chapter 3 points out that domestication generally involves the repeated evolution of similar traits in various species, with causal mutations often affecting the same genes. Collecting information on the genes causing domestication traits can thus help to define candidate genes for genetic improvement and domestication of other plant species. Chapter 4 will make readers realize that *A. thaliana* is, besides humans maybe, the Eukaryotic species whose genetic basis of

phenotypic variation is best understood. Examination of the genetics underlying interactions between *A. thaliana* and other species reveals that integrating multiple genetic effects within a natural environment and at the level of ecosystems is far from simple and poses interesting challenges.

Very little is known about the genetic basis of behavioral evolution. Indeed, most evolutionary changes in phenotype whose causal genes have been identified so far are morphological and physiological. Chapter 5 examines whether the concept of genetic toolkit, which derives from morphological studies, may also apply to behavioral evolution. Chapter 6 manipulates two important concepts in contemporary biology, gene networks and novelties, and argues that the mutations underlying the appearance of a novel trait are likely to affect particular positions within gene networks. Chapters 7 and 8 illustrate the fact that genes are not the unique factors that should be taken into account when trying to understand how phenotypes evolve. Chapter 7 shows that the shape and diversification of epithelial tissues result from a complex interplay between genes, signaling pathways, and forces. Chapter 8 examines precisely, with clear-cut and well-chosen examples, the multiple interactions and feedbacks between genes, organisms, and environment at various levels and timescales, which together lead to phenotypic changes. The last chapter provides a comprehensive overview of the genes and mutations that are currently known to cause phenotypic diversity in actual human populations and in our recent past history.

This volume is being published at a time when many research laboratories are focusing on the evolution of diverse phenotypic traits and trying to dissect their genetic basis. While these ongoing studies might look, to the eyes of certain biologists, quite repetitive and unoriginal, this volume provides a good material to reflect on our current state of research and to try to detect new future explanatory frameworks and ideas to be discovered from our examination of the genes and the mutations responsible for evolution.

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