DANIEL L. HARTL

Principles of Population Genetics

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POPULATION GENETICS

DANIEL L. HARTL WASHINGTON UNIVERSITY SAINT LOUIS

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PRINCIPLES OF POPULATION GENETICS

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Preface

This is a textbook of population genetics designed for junior-senior undergraduates and graduate students. Population genetics is an extraordinarily diverse field. It cuts across molecular biology, genetics, ecology, evolutionary biology, systematics, natural history, plant breeding, animal breeding, many areas of conservation and wildlife management, human genetics, sociology, anthropology, mathematics, and statistics. Because population genetics plays a special role in so many areas of biology, I have tried to summarize the principles of the field for the widest possible audience. No prerequisites in genetics or advanced mathematics are necessary for an understanding of the material in the book. The relevant background is provided in Chapter 1. The first half of Chapter 1 deals with the necessary background in genetics and molecular biology; it is intended as a summary for students whose prior training has been in statistics or mathematics, or for students in biology who have not yet studied genetics. This material also serves as a memory refresher for students who have studied genetics. The second half of Chapter 1 provides the relevant statistical and mathematical background; it is designed primarily for students in any of the biological or social sciences.

Chapters 2 through 4 concern what is usually regarded as the core of population genetics. The theme of these chapters is the origin, maintenance, and significance of genetic variation. Chapter 2 focuses on the detection and measurement of genetic variation, and on the organization of genetic variation as influenced by particular mating systems such as random mating or inbreeding. Chapter 3 deals with the evolutionary effects of mutation, migration, selection, and small population size. Chapter 4 considers traits that are influenced by alleles at many loci as well as by the effects of environment. Although particular attention is devoted to methods of artificial selection that are used for the genetic improvement of crop plants and domesticated animals, Chapter

4 also discusses how the principles of quantitative genetics can be applied to humans and natural populations of other species.

Chapter 5 is a synthesis of the elements of population genetics as they apply to actual, evolving populations in an ecological setting. The first part of the chapter is devoted to Sewall Wright's shifting balance theory of evolution and its alternatives. The second part deals with modes of selection that do not act directly on the individual but rather on the individual's relatives: group selection, interdeme selection, and kin selection. The third part illustrates the principles of population genetics in action by means of four particularly well studied examples — the evolution of antibiotic resistance in bacteria, of heavy-metal tolerance in plants, of industrial melanism in moths, and of Batesian mimicry in butterflies. Finally, Chapter 5 deals with the role of population genetics in understanding the origin of species.

Throughout the book, I have tried to emphasize the interplay between experimental observations and theoretical deductions. The experimental side of population genetics is wonderfully rich and diverse. But the literature of experimental population genetics is scattered among a variety of journals extending from those in plant and animal breeding to those in ecology and evolution. One motivation for writing the book was to bring this disparate literature together and emphasize its underlying unity. The theoretical side of population genetics is less imposing than might be expected. Especially at this introductory level, the mathematics involved in theoretical population genetics is mainly simple and straightforward algebra. In addition, the theoretical arguments in the book are developed step by step and are illustrated with numerous actual examples.

A number of special features of the book deserve mention. Throughout the book are a number of boxes that are set off from the main text in smaller type. These boxes serve several functions. A few provide background material in experimental or statistical methods. A few others show how certain equations stated in the text can be derived. Most of the boxes, however, take up where the text leaves off; they extend the material in the text and briefly discuss more specialized or advanced topics. Although a few boxes make use of elementary calculus, emphasis is on the meaning and application of the results, not on the mathematics. Moreover, all required mathematical formulas are provided as needed.

Use of the boxes is at the instructor's discretion and depends on the level and orientation of the course. None of the boxes is

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required for an understanding of the text. In this sense, the boxes are optional. However, an instructor may wish to assign certain boxes as required reading; the boxes in Chapter 4 would be particularly relevant in courses oriented toward plant or animal breeding, for example. Many of the boxes contain sufficiently important material to justify their requirement in courses at the graduate level.

At the end of each chapter is a set of fifteen problems designed to help students verify their understanding of the text. Solutions to the problems, which include the methods of solution as well as final answers, can be found in the back of the book. Problems are also provided as a test of mastery in each box; these, too, are solved in full at the back of the book. In writing the problems, no special effort has been made to avoid the need for numerical calculations, as most students these days have access to electronic calculators.

At the end of each chapter is a set of further readings. These are general sources, mainly books and review articles. All references cited in the text are listed in the bibliography at the end of the book. Although the bibliography contains over 600 references to the primary literature of population genetics, the number could easily have grown much larger. In order to keep the bibliography in manageable proportions, I have emphasized review articles (wherever possible) and recent literature. The bibliography is designed mainly as an entry point to the literature. Moreover, I have made no serious attempt to cite the first or "priority" paper on any subject, on the grounds that such distinctions are best left to historians of the science. It should be noted, however, that most of the ideas of modern population genetics are derived from the works of Sewall Wright, R. A. Fisher, and J. B. S. Haldane. While I have tried to be fair to original investigators in acknowledging their contributions, there are no doubt a few unintentional oversights. For these I apologize.

Writing a book is always grueling, at least it is for me. This task was made much easier by reviewers who offered expert guidance in matters of emphasis and balance and in correcting wrong or misleading statements. I thank James F. Crow, Franklin D. Enfield, Joseph Felsenstein, Bruce R. Levin, Rollin Richmond, Michael J. Simmons, and Edward O. Wilson, each of whom read all or major parts of the book. Carl W. May did a splendid job of collating all the reviewers' suggestions and offering numerous helpful comments of his own.

The material in the book formed the basis of a course in experimental population genetics that was offered while I was Visiting Professor at the University of Zürich in Switzerland. I am grateful to Professor Hans Burla for arranging the course, to the students for their advice and criticism, and especially to Hans Jungen and Elizabeth Hauschteck-Jungen for their generous hospitality during my stay in Zürich.

Rollin Richmond used a preliminary draft of the book for a course in population genetics at Indiana University, and I am gratified by the favorable student response. Stephen Rich of Purdue University read the final draft and rechecked the answers to all the chapter-end and box problems.

The final manuscript was expertly typed by Betty Gick, Pat Oswalt, and Carol Wigg. Natalie Brown and Stephen Rich helped in proofreading. Credit for the final appearance of the book should go to Sinauer Associates and to Joseph Vesely Production Services. A special thanks goes to Andy Sinauer for his advice and encouragement.

The people in my laboratory deserve plaudits for their continuing interest and patience while I struggled with the manuscript. My thanks to Daniel Dykhuizen, Doris Freeman, and Luanne Wolfgram for keeping the house in order, and to John Dunne, David Haymer, Paul Kaytes, Nizam Kettaneh, and Susan Wurster for their forebearance.

I want also to thank Dana Margaret and Theodore James Hartl for their unflagging curiosity and enthusiasm for the project. Their constant "When will it be finished?" and "When can we see it?" served as a needed prod.

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Genetic and Statistical Background

Population genetics is the study of how Mendel's laws and other genetic principles apply to entire populations. Such a study is essential to a proper understanding of evolution because, fundamentally, evolution is the result of progressive change in the genetic composition of a population. Population genetics thus seeks to understand and to predict the effects of such genetic phenomena as segregation, recombination, and mutation; at the same time, population genetics must take into account such ecological and evolutionary factors as population size, patterns of mating, geographic distribution of individuals, migration, and natural selection. The many genetic, ecological, and evolutionary factors that influence populations also interact and feed back, one upon the other. As might well be expected, to gain an understanding of such complex interactions is a formidable task.

Ideally, one would wish to know how to describe the types and frequencies of genes in a population, to explain how the population's genetic composition came to be the way it is, and to predict how the population would change as a result of natural selection or as a result of artificial selection applied by a plant or animal breeder. Population geneticists are a long way from being able to do these things as well as they might be done, but in the 80 or so years since the formal study of population genetics was inaugurated, considerable progress has been made. Indeed, a balanced view of the recent history of biology would not permit the brilliant and relatively rapid success of molecular biologists to completely overshadow the slow, steady progress of population geneticists against their own problems, problems that can be particularly stubborn because they are often so subtle and sometimes maddeningly complex.

Nevertheless, it is appropriate to point out that certain central problems in population genetics have been solved by use of relatively straightforward approaches and have yielded relatively simple principles; these principles have applications and implications for topics that range all the

way from medicine to ecology, from anthropology to plant and animal breeding. This book is my attempt to summarize these principles.

The Genetic Background of Population Genetics

The first half of this chapter is intended as a memory refresher for students in any of the biological sciences and as a brief summary of genetics for students in mathematics, statistics, or the social sciences. Because major aspects of population genetics deal with the implications of Mendel's laws as they affect whole populations, we may do well to begin with Mendel — Gregor Johann Mendel (1822-1884), amateur plant breeder, who spent most of his adult life as a priest and later abbot in a monastery in the city of Brünn, Moravia, now Brno, Czechoslovakia. A few key terms must first be defined, however.

GENE is a general term meaning, loosely, that physical entity transmitted during the reproductive process that influences hereditary traits among the offspring. Genes influence such human traits as hair color, eye color, skin color, height, weight, and intelligence, although most of these traits are also influenced more or less strongly by environment. Genes also determine the nature of such proteins as hemoglobin, which carries oxygen in the red blood cells, or insulin, which is important in maintaining glucose balance in the blood. Genes can exist in different forms or states. For example, a gene for hemoglobin may exist in a normal form or in any one of a number of forms that produce hemoglobin molecules that are more or less abnormal. These alternative forms of a gene are called ALLELES.

From a biochemical point of view, a gene corresponds to a specific sequence of constituents (called nucleotides) along a molecule of DNA (deoxyribonucleic acid) — DNA is the genetic material. Different sequences of nucleotides that may occur in a gene, therefore, represent alleles. (See Figure 1

and the section on molecular genetics later in this chapter for further detail.)

Mendel studied seven traits in the garden pea *Pisum sativum*. He self-fertilized plants for several years until he obtained strains that "bred true"; that is, until he obtained strains that were genetically stable in the sense that the offspring in any generation strongly resembled each other and their parents with respect to each trait considered. For example, Mendel obtained a strain that bred true for *round* seeds; crosses between individuals of this strain always yielded plants with *round* seeds. He also obtained a strain that bred true for *wrinkled* seeds, and crosses within this strain always yielded plants with *wrinkled* seeds. Among the seven traits for which Mendel obtained true-breeding strains that exhibited contrasting appearances were, as mentioned, seed shape (*round* versus *wrinkled*), color of the seed (actually the color of the cotyledon, the first leaf of the embryo; *yellow* versus *green*), and seedpod shape (*smooth* versus *constricted*).

With the true breeding strains that he had developed, Mendel began his monumental series of breeding experiments that culminated in 1866 with publication of "Experiments on plant hybrids" in the *Proceedings of the Brünn Natural History Society*. (The original paper is in German, but an excellent English translation is available in Stern and Sherwood, 1966.) Mendel's paper was almost completely ignored until 1900, when Hugo de Vries in Amsterdam, Carl Correns in Tübingen, and Erich von Seysenegg Tschermak in Vienna independently rediscovered Mendel's findings and finally uncovered his original publication.

DOMINANCE

Now, when Mendel crossed plants from the true-breeding strain having *round* seeds with plants from the true-breeding strain having *wrinkled* seeds, he obtained a startling result: the offspring or HYBRID seeds were *round*. Similarly, crosses between the *yellow*-seed strain and the *green*-seed strain yielded *yellow* seeds; crosses between the *smooth*-pod and *constricted*-pod strains gave rise to plants with *smooth* pods, and so on for all seven traits. These results can be summa-

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