Solving Problems in Genetics

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With 266 Figures

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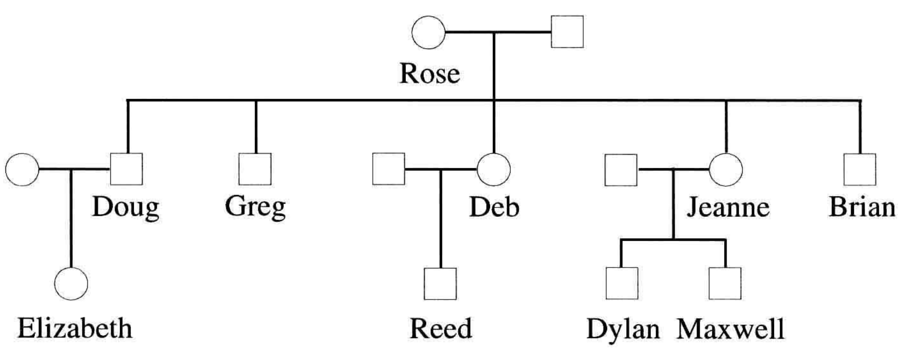
Dedication



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Preface

Genetics is largely an analytical science. Techniques of analysis are an important dimension of genetics. The development of the discipline has been staggering in recent years, but the basic mechanics of genetics should not be neglected. Many instructors of genetics agree that students tend to have difficulty conducting genetic analysis. The sole objective of this book is to make students more adept in the analysis of genetic problems. The book is meant to be supplemental and useful to students of genetics regardless of the textbook being used in their course. The supplemental, and often complemental, material will show applications of the concepts explained in genetics textbooks. Some instructors, however, may want to use the book as their textbook, which would depend upon the structure and nature of their courses. Students who learn how to manage genetics problems will better understand the basic principles of genetics and will know how to more effectively apply the information to other related genetic situations.

The book is intended to be used by undergraduate college students majoring in biology and studying introductory genetics. The biology curricula of many colleges and universities now follow their introductory genetics course with one or more additional courses that heavily focus on the many recent advances of molecular biology. This book has several chapters relating to molecular genetics, but the main objective is to present a broad coverage of genetics. Topics, therefore, concentrate on Mendelian relationships, transmission, cytogenetics, molecular, quantitative, and population genetics, with some additional areas such as maternal inheritance and human genetics. An attempt is made to present a balanced account of genetics.

The same format is used throughout the entire book. A brief introductory discussion is provided to set the stage for the ensuing genetic analysis. The discussion concentrates on genetic analysis, the interpretation of data, and the explanation of classic experiments. The text material is followed with a relevant sample and its solution. This format repeats itself throughout each of the chapters. Many related problems are presented at the end of each chapter with their solutions immediately following that section. The emphasis for both sample problems and those at the end of the chapters is "solutions" and not just "answers." Most textbooks, of course, provide problems and answers. Few will show the student how to actually solve the problem. Often the student does not have any idea concerning how the answer was derived. The strategy within this book is to provide the student with the essential steps and the reasoning involved in conducting genetic analysis. Although important as other facets of genetics, discussion and descriptive type questions are not included. Everything is a problem to be analyzed.

The book is not designed to be read rapidly; rather, it is meant to be studied in conjunction with the many excellent genetics textbooks available. The text part of the book contains very few if any embellishments. A deliberate attempt has been made to make complex concepts simple, and sometimes to make simple concepts complex. The ruling goal has been to aid the student in developing analytical ability. Numerous figures and diagrams are included to promote the goal of making problem solving as student friendly as possible.

In many cases, more than one technique exists to solve genetic problems. This book offers one variation in solving problems. Some topics and related problems are included simply for the sake of developing analytical ability; that is, they are deemed excellent for analysis purposes. Other topics are intentionally omitted because they are not conducive to the type of analysis preferred in the book. Much important genetics today centers upon the isolation, cloning, and sequencing of genes. As imperative as these topics are to a good genetics course, they do not always lend themselves well to good quantitative-type problems for students of introductory genetics. This book is not intended to be a comprehensive textbook touching upon everything that has happened in the field of genetics.

The information and accompanying problems tend to build upon each other; that is, analytical methods learned in early parts of the book are often subsequently necessary in later analyses. Pedagogically, this adds an element of recall. Some of the problems are substantial challenges. Instructors may or may not assign them, but such problems are sometimes useful for those students who are not adverse to a good challenge. Instructors can, of course, be selective with regard to all of the problems. Some topics may not even be used in their course. Statistical analysis is described at the point of necessity throughout the book. Additionally, Chapter 12 is highly statistical. Statistical analyses are very important in the biological sciences and certainly have a solid place in genetics. They constitute a powerful set of tools, and the discipline lends itself extremely well to many facets of genetics. Still, many instructors may choose to use only part of Chapter 12 or completely omit it. Altogether, the book contains 115 sample problems and 317 end of the chapter problems.

Acknowledgments are always in order for such an undertaking. I particularly want to thank Dr. Clare Korte for her many hours of proofreading of the manuscript and Kate Finley for handling the secretarial details between author and publisher. Patti Reick proved to be a competent illustrator with a good eye for detail. Dr. Robin Smith, Executive Editor for Life Sciences at Springer-Verlag New York, Inc., made the project fun and relatively painless. Also, thanks always must be given to the reviewers who are so good at detecting what you have overlooked. And lastly, thanks to the many biology students over the years who were subjected to many of these problems. They could never resist an opportunity to inform me of errors and ambiguities.

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Contents

1 Mendelism

Introduction

One of the most remarkable features of living things is that they can reproduce. The phenomenon whereby biological traits are transmitted from one generation to another is called heredity. The physical and molecular organization underlying the hereditary process is the major focus of the scientific discipline known as genetics. Geneticists study the continuity of life, overall variation in organisms, specific traits due to heredity, and the mode of inheritance.

Today, much is known about the continuity of life. Reproduction is basic to life, survival, and the heredity of an organism. Two organisms of opposite mating types can produce a fertilized egg, called a zygote, and this initial cell is capable of growing and developing into a tremendously complicated system, another living organism. We know that the hereditary information involved in reproduction is particulate. These particles are now called genes. Their composition, transmission, and role in molding characteristics of organisms are being elucidated at a rapid pace. The methods used to gain hereditary information often involve quantitative relationships, calculations, and problem-solving approaches.

Information about hereditary particles and how they are transmitted from parents to offspring began with the Austrian monk, Gregor Mendel. In very few instances will one find the origin of a particular discipline so firmly linked to one individual as is genetics to Gregor Mendel (1822–1884). The 1866 publication, *Experiments on Plant Hybridization*, is certainly a classic scientific contribution. Following the rediscovery of the publication in 1900, the conclusions Mendel derived from his plant experiments became the foundation for the science of genetics. Mendelism, therefore, refers to that aspect of inheritance that describes the transmission of biological traits from parents to offspring and subsequent generations.

To review, Mendel perceived (1) that inheritance is due to various kinds of factors that are particulate; (2) that the factors are present in pairs; (3) that the pairs of factors segregate from each other during the reproductive process; (4) that the factors are restored to pairs in the offspring; and (5) that the members of two different pairs of factors tend to assort independently from each other. From those ideas, two basic statements have been synthesized that have become known as Mendel's Laws of Inheritance.

1. Mendel's First Law—the principle of segregation. Members of a pair of factors segregate from each other during the formation of sex cells in an individual.
2. Mendel's Second Law—the principle of independent assortment. Members of different pairs of factors assort independently of each other during the formation of sex cells in an individual.

Basic Probability: Methods for Analyzing Mendelian Genetics

An essential ingredient in the Mendelian hypothesis is that the choice of which allele passes to a gamete is made at random; hence, two alleles of a pair enter any gamete with equal probability. Also, the gametes of two different mating types are combined in the same random manner through the process of fertilization. One of the assumptions in Mendelian inheritance is that during fertilization male gametes combine with female gametes independently of any alleles that they carry. Fixed probabilities, then, can be applied to such events as meiosis and fertilization when conducting genetic analysis.

Probability (p) is equated to proportional frequency, that is, the number of times that some event will likely occur over the range of possible occurrences. If the event cannot occur, the probability is 0. If the event must occur, the probability is 1. For those events with an element of uncertainty, the probability of their occurrence is a value that lies between 0 and 1.

Using the basic rules for elementary probability theory, combinations of events in Mendelian genetics can be calculated. The two types of combinations most often encountered are those events given as (1) mutually exclusive and (2) independent. Mutually exclusive means that the occurrence of one event excludes the possibility of the others. Whenever such alternative possibilities exist for the satisfaction of a genetic problem, the individual probabilities are combined by addition. For example, the probability of obtaining the dominant phenotype from a *Gg* × *Gg* cross is 3/4. This result is due to the addition of 1/4 (the probability for GG) and 1/2 (the probability for *Gg*). Two or more events are deemed to be independent if the occurrence or the nonoccurrence of any one of them does not affect the probable occurrence of any of the others. The combined probability in this case is the product of the individual probabilities. Since the genotype of any particular progeny will be the result of combining the alleles of the gametes from the two parents, the proportions of the different genotypes will be the product of the individual gamete probabilities. For example, in the cross *Gg* × *Gg*,

$$\text{Maternal } Gg \rightarrow \text{gametes } 1/2\ G \text{ and } 1/2\ g$$
$$\text{Paternal } Gg \rightarrow \text{gametes } 1/2\ G \text{ and } 1/2\ g$$

The probabilities of the genotypes are derived by multiplying the individual gamete probabilities in all combinations, maternal with paternal.

$$\begin{aligned} 1/2\ \mathrm{G} \times 1/2\ \mathrm{G} &= 1/4\ GG \\ 1/2\ \mathrm{G} \times 1/2\ \mathrm{g} &= 1/4\ Gg \\ 1/2\ \mathrm{g} \times 1/2\ \mathrm{G} &= 1/4\ Gg \\ 1/2\ \mathrm{g} \times 1/2\ \mathrm{g} &= 1/4\ gg \\ \text{and: } 1/4\ Gg + 1/4\ Gg &= 1/2\ Gg \end{aligned}$$

Mendelian genetics is heavily based upon probability concept because of the randomness involved in its mechanisms. Questions about the expected progeny from a cross in which both parental genotypes are known can be calculated by analyzing each of the parental gene pairs separately. Thereafter, the product rule of probabilities or the summation of probabilities can be applied, dependent upon the information being sought. Probabilities relative to gametes or zygotes, one locus or many loci, and one offspring or many offspring can be calculated in this manner. The need for large, laborious Punnett squares can be eliminated if desired.

Sample Problems: Consider a cross between two organisms in which the genotypes of five different loci are known as follows, with all of the gene pairs segregating independently from each other. A number of questions can be asked about this cross, and a sampling of such questions follows.

female male
Aa Bb CC Dd EE (×) *AA Bb Cc Dd Ee*

(a) What is the probability that any particular gamete from the female parent will be *AbCdE*?

Solution: *Aa Bb CC Dd EE* will transmit *A* with p = 1/2; *b* with p = 1/2; *C* with p = 1; *d* with p = 1/2; and *E* with p = 1. Therefore, using the product rule of probability, p = 1/2 × 1/2 × 1 × 1/2 × 1 = 1/8.

(b) What is the probability that any particular zygote will be *EE*?

Solution: The female parent with *EE* gives *E* with p = 1 and the male parent with *Ee* gives *E* with p = 1/2. Therefore, p = 1 × 1/2 = 1/2.

(c) What is the probability that any particular zygote will be *Aa bb Cc dd EE*?

Solution: Analyze each locus separately.

Aa × *AA* results in *Aa* with p = 1/2
Bb × *Bb* results in *bb* with p = 1/4
CC × *Cc* results in *Cc* with p = 1/2
Dd × *Dd* results in *dd* with p = 1/4
EE × *Ee* results in *EE* with p = 1/2

Next, apply the product rule as follows:

p = 1/2 × 1/4 × 1/2 × 1/4 × 1/2 = 1/128