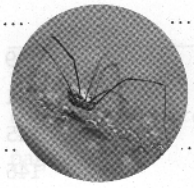


Contents

4 Replication

Endless copies

5 Genome

Accident and design

6 Gene

Making an organism

11 Phylogeny

Tree of life

page 329

12 Hypotheses

Seeing the wood for the trees

page 369

13 Rates

Tempo and mode

page 403

14 Dates

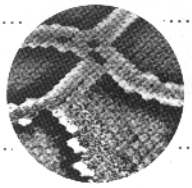
Telling the time

page 438

Coda: You are a scientist

What do I do now?

page 484



Preface

This book is designed to give a from-the-ground-up introduction to the use of molecular phylogenetic analysis in evolution and ecology. It describes how you can use analysis of DNA sequence data to reconstruct evolutionary history and processes. To do this, you need to have a basic understanding of molecular evolution: how mutations occur that change the DNA sequence, how some of these mutations become substitutions that create differences between populations or species, and how we can use the patterns of substitutions to reconstruct evolutionary history. The aims of this book, and the features it contains, are covered more fully in Chapter 1. Here is a brief summary with references to pages where each point is discussed in more detail.

Who is this book for?

This book is aimed at biology students who need to learn how to use DNA sequence analysis to understand evolution and ecology. The book assumes no more than a basic grounding in biology and genetics, so should be suitable for entry-level undergraduate students (see Chapter 1, 'Who is this book for?', page 11). However, this book may also be useful to bioinformaticians with a background in maths, statistics, or computation, who need to learn evolutionary and genetic principles in order to interpret patterns in biological data.

What is in this book?

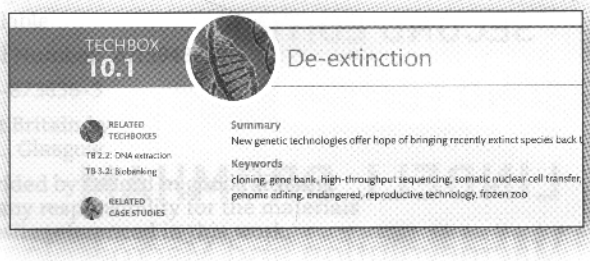
This book is designed to be read in a variety of ways, depending on your interests or background knowledge (see Chapter 1, 'How to use this book', page 13). To facilitate you getting what you need from this book, each chapter is divided into several sections.

The main text provides the background to key issues in molecular evolution and phylogenetics, and should be accessible to entry-level students with no previous knowledge of the subject.

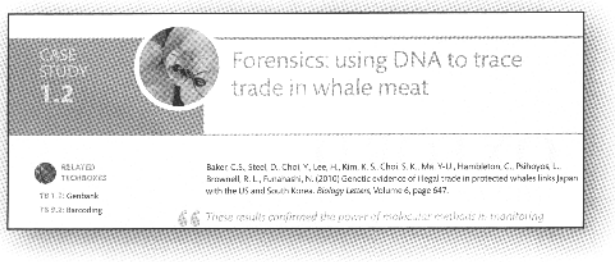
- ***Points to remember*** provides a bullet-point summary of the key issues covered in the chapter, as a revision aid.
- ***Ideas for discussion*** draw on the subjects covered in the chapter, but asks you to step beyond the material covered to think more broadly about the concepts.
- ***Sequences used in this chapter*** gives the Genbank description and accession number for any sequence used for illustrative purposes in the chapter.
- ***Examples used in this chapter*** is not a comprehensive bibliography but the publication references of specific examples mentioned in the text.

There are three kinds of boxes, which provide optional additions to the main text, as follows.

TechBoxes go into more detail on specific methods or ideas. The language is more technical and they may be challenging to entry-level students, but more informative for students with some previous knowledge of molecular genetic analysis.



Case Studies illustrate recent scientific research related to the principles covered in the chapter, cross-referenced to TechBoxes that explain the techniques used. There will be many terms and concepts that will be unfamiliar to beginners, so they may be better suited to more advanced students. There are three ***'Check your understanding'*** questions which are based on material from the box. The ***'What do you think?'*** section aims to get you to think more deeply about the implications of the topics discussed in the box, and could be used for class discussions. ***'Delve deeper'*** encourages you to go beyond the material presented in the case study and do some independent research on a related topic. ***References*** cited in the box are listed at the end, as are cross-references to other useful boxes.



Heroes of the Genetic Revolution illustrate scientists who have worked in the area.


HEROES OF THE GENETIC REVOLUTION 4

Rosalind Franklin

"As a scientist Miss Franklin was distinguished by extreme clarity and perfection in ev she undertook. Her photographs are among the most beautiful x-ray photographs of substance ever taken."

Bernal, J. D. (1958) 'Dr Rosalind E. Franklin: Obituary' Nature, Volume 182, page 154

EXAMPLE PUBLICATIONS

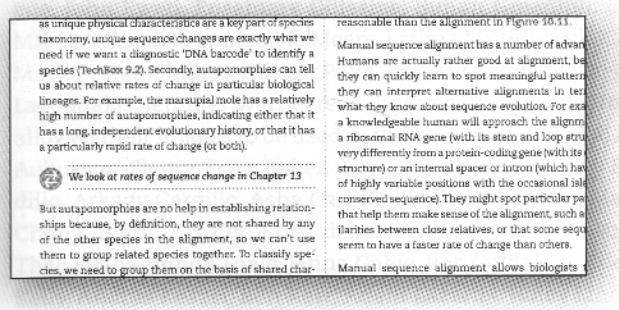
Franklin, R. E., Gosling, R. G. (1953) Molecular configuration in sodium thymonucleate. Nature, Volume 171, page 740.

Franklin, R. E. (1955) Structure of tobacco mosaic virus. Nature, Volume 175, page 379.

Rosalind Franklin (Figure 4.28) was a dedic scientist from the time she was a teenager, st physics and chemistry at school, then gradu from Newnham College at Cambridge Univ 1941 (seven years before Cambridge finally awarding degrees to female students). Altho started a research position in the physical ch

Other features of the text

Cross-references to information in other chapters, to help you navigate through the book and build up a knowledge base.


as unique physical characteristics are a key part of species taxonomy, unique sequence changes are exactly what we need if we want a diagnostic 'DNA barcode' to identify a species (TechBox 9.2). Secondly, autapomorphies can tell us about relative rates of change in particular biological lineages. For example, the marsupial mole has a relatively high number of autapomorphies, indicating either that it has a long, independent evolutionary history, or that it has a particularly rapid rate of change (or both).

We look at rates of sequence change in Chapter 13

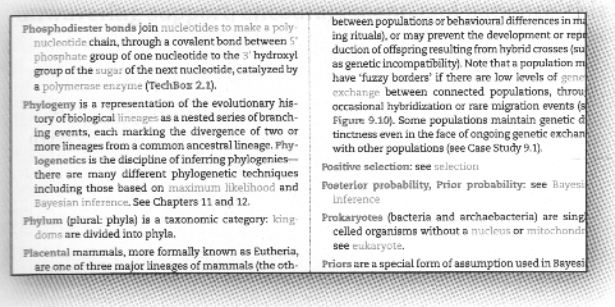
But autapomorphies are no help in establishing relationships because, by definition, they are not shared by any of the other species in the alignment, so we can't use them to group related species together. To classify species, we need to group them on the basis of shared char-

reasonable than the alignment in Figure 16.11.

Manual sequence alignment has a number of advan Humans are actually rather good at alignment, be they can quickly learn to spot meaningful patterns they can interpret alternative alignments in ter what they know about sequence evolution. For exa a knowledgeable human will approach the alignm a ribosomal RNA gene (with its stem and loop stru very differently from a protein-coding gene (with its structure) or an internal spacer or intron (which ha of highly variable positions with the occasional isla conserved sequence). They might spot particular pa that help them make sense of the alignment, such a ilarities between close relatives, or that some sequ seem to have a faster rate of change than others.

Manual sequence alignment allows biologists

Glossary contains some basic biological terms and explains some tricky concepts in evolution and genetics.


Phosphodiester bonds join nucleotides to make a polynucleotide chain, through a covalent bond between 5' phosphate group of one nucleotide to the 3' hydroxyl group of the sugar of the next nucleotide, catalyzed by a polymerase enzyme (TechBox 2.1).

Phylogeny is a representation of the evolutionary history of biological lineages as a nested series of branching events, each marking the divergence of two or more lineages from a common ancestral lineage. Phylogenetics is the discipline of inferring phylogenies—there are many different phylogenetic techniques including those based on maximum likelihood and Bayesian inference. See Chapters 11 and 12.

Phylum (plural: phyla) is a taxonomic category: kingdoms are divided into phyla.

Placental mammals, more formally known as Eutheria, are one of three major lineages of mammals (the oth-

between populations or behavioural differences in m ing rituals), or may prevent the development or repr duction of offspring resulting from hybrid crosses (su as genetic incompatibility). Note that a population m have 'fuzzy borders' if there are low levels of gene exchange between connected populations, throu occasional hybridization or rare migration events (s Figure 9.10). Some populations maintain genetic d tinctness even in the face of ongoing genetic exchan with other populations (see Case Study 9.1).

Positive selection: see selection

Posterior probability, Prior probability: see Bayesi inference

Prokaryotes (bacteria and archaebacteria) are sing celled organisms without a nucleus or mitochondri see eukaryote.

Priors are a special form of assumption used in Bayesi

Figure legends sometimes have interesting facts not directly related to the subject of the chapter, because that's what makes biology fun.

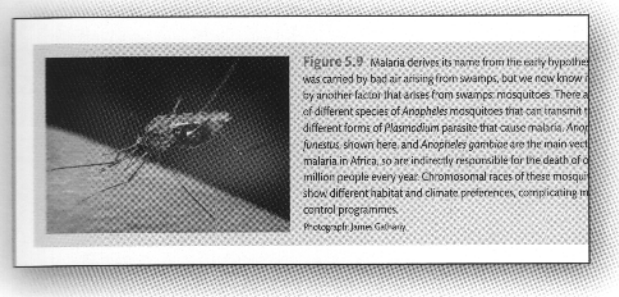


Figure 5.9 Malaria derives its name from the early hypothes was carried by bad air arising from swamps, but we now know by another factor that arises from swamps: mosquitoes. There a of different species of *Anopheles* mosquitoes that can transmit different forms of *Plasmodium* parasite that cause malaria. *Anop funestus*, shown here, and *Anopheles gambiae* are the main vect malaria in Africa, so are indirectly responsible for the death of million people every year. Chromosomal races of these mosqui show different habitat and climate preferences, complicating m control programmes.

Photograph: James Gathany.

What is not in this book?

Instructions for computer programs: The aim of this book is to give you an understanding of the evolutionary principles which are the basis of molecular phylogenetic analysis, rather than to give you training in the use of particular programs. The reason that this book sticks to general principles and avoids specific methods is that new methods are constantly being developed. Any programs or methods I describe today are likely to be out of date within a few years. But by giving you the general principles underlying those methods, I hope to equip you with the intellectual tools you need to learn how to use particular methods, and to adapt your analysis when new methods become available.

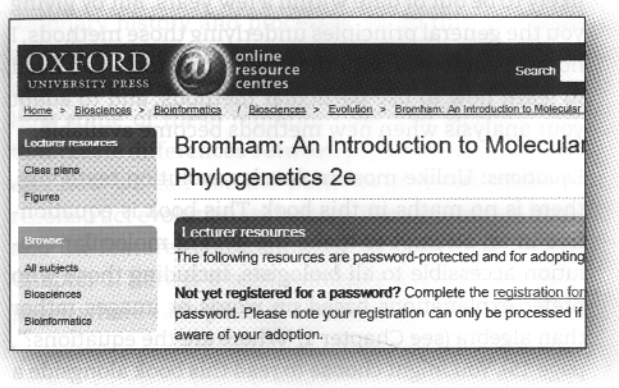
Equations: Unlike most molecular evolution textbooks, there is no maths in this book. This book is equation-free in an attempt to make the field of molecular evolution accessible to all biologists, including those who prefer explanations based on words or images rather than algebra (see Chapter 1, 'Where are the equations?', page 15). You might choose to read this book alongside a traditional molecular evolution text which will provide all the population genetic equations for the principles discussed here.

What has changed since the first edition?

The first edition of this book was titled *Reading the Story in DNA: a beginner's guide to molecular evolution*. This new edition contains some of the same material, but has been considerably expanded, updated, and remodelled in response to feedback from readers. The book has increased from 8 chapters to 14, which allows the material to be divided more evenly into key concepts and may make the book better fit a semester teaching structure. There are more features designed to help students and teachers come to grips with the material, such as summary points at the end of chapters, and discussion questions in the chapters and case studies. There are now nearly twice as many case studies, and many of the previous case studies have been replaced with more recent examples. I have discovered that the problem with writing a book based on molecular genetics is that the field changes so fast that much of the first edition had been superseded only five years after its publication. For example, high-throughput (next-gen) sequencing, which took up only a paragraph in the first edition, is on its way to eclipsing Sanger sequencing as the method of choice in molecular phylogenetic analysis, so the chapters and case studies have been updated

to reflect this fundamental change in data generation and analysis. It's inevitable that by the time this edition is in print, the field will have moved on even further. The best approach is to use this book as a jumping-off point for an examination of the most recent techniques in the scientific literature.

What is in the Online Resource Centre?



For lecturers: downloadable figures, tutorial exercises, practical projects.

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