Introduction to Conservation Genetics

The biological diversity of the planet is being rapidly depleted due to the direct and indirect consequences of human activity. As the size of animal and plant populations decreases, loss of genetic diversity reduces their ability to adapt to changes in the environment, with inbreeding and reduced fitness inevitable consequences for most species. This textbook provides a clear and comprehensive introduction to genetic principles and practices involved in conservation. Topics covered include:

- evolutionary genetics of natural populations
- loss of genetic diversity in small populations
- inbreeding and loss of fitness
- population fragmentation
- resolving taxonomic uncertainties
- genetic management of threatened species
- contributions of molecular genetics to conservation.

The text is presented in an easy-to-follow format, with main points and terms clearly highlighted. Each chapter concludes with a concise summary, which, together with worked examples and problems and answers, illuminates the key principles covered. Text boxes containing interesting case studies and other additional information enrich the content throughout, and over 100 beautiful pen-and-ink drawings help bring the material to life.

Written for advanced undergraduate and graduate students studying conservation, this book will be equally useful to practising conservation biologists and wildlife managers needing an accessible introduction to this important field.

The authors comprise a team with a range of skills and experience that make them uniquely qualified to put together the first teaching text on conservation genetics:

DICK FRANKHAM is Professor of Biology at Macquarie University, Sydney, Australia. He began his career in quantitative genetics, achieving international recognition for his work on *Drosophila* before turning to conservation genetics in the early 1990s. He has made a significant contribution to the establishment and advancement of the field and has become one of the major figures in the discipline.

JON BALLOU is Population Manager at the Smithsonian Institution's National Zoological Park in Washington DC, USA and an adjunct member of the faculty at the University of Maryland. His career has focused on developing the science underlying the practical management of small populations of endangered or threatened species, both captive and wild. The results of his studies have been instrumental in highlighting the key role played by genetics in wildlife conservation and management.

DAVID BRISCOE is Associate Professor of Biology at Macquarie University, Sydney, Australia where he has been a close collaborator with Dick Frankham on *Drosophila* research, as well as working with others on rock wallabies, velvet worms and slime molds. An outstanding communicator, his inspirational teaching enthuses students at all levels and reaches beyond the academic sphere through television appearances and popular books such as *Biodiversity: Australia's Living Wealth* to which he contributed.

Introduction to Conservation Genetics

Richard Frankham,
Macquarie University, Sydney

Jonathan D. Ballou
Smithsonian Institution, Washington, DC

and David A. Briscoe
Macquarie University, Sydney

Line drawings by
Karina H. McInness
Inkbyte, Melbourne

CAMBRIDGE
UNIVERSITY PRESS

PUBLISHED BY THE PRESS SYNDICATE OF THE UNIVERSITY OF CAMBRIDGE
The Pitt Building, Trumpington Street, Cambridge, United Kingdom

CAMBRIDGE UNIVERSITY PRESS
The Edinburgh Building, Cambridge CB2 2RU, UK
40 West 20th Street, New York, NY 1011–4211, USA
477 Williamstown Road, Port Melbourne, VIC 3207, Australia
Ruiz de Alarcón 13, 28014 Madrid, Spain
Dock House, The Waterfront, Cape Town 8001, South Africa

http://www.cambridge.org

First published 2002
Fourth printing 2004

Printed in the United Kingdom at the University Press, Cambridge

Typeface 9.5/12pt Swift *System* QuarkXPress® [SE]

A catalogue record for this book is available from the British Library

Library of Congress Cataloguing in Publication data

Frankham, Richard, 1942–
Introduction to conservation genetics/Richard Frankham, Jonathan D. Ballou, and David A. Briscoe; line drawings by Karina H. McInness.
p. cm.
Includes bibliographical references and index.
ISBN 0 521 63014 2 – ISBN 0 521 63985 9 (pb.)
1. Germplasm resources. I. Briscoe, David A. (David Anthony), 1947– II. Ballou, J. D. (Jonathan D.) III. Title.

QH75.A1 F73 2002
333.95′34–dc21 2001035028

ISBN 0 521 63014 2 hardback
ISBN 0 521 63985 9 paperback

Contents

Preface

The World Conservation Union (IUCN), the primary international conservation body, recognizes the crucial need to conserve genetic diversity as one of the three fundamental levels of biodiversity. This book provides the conceptual background for understanding the importance of genetic diversity in avoidance of species extinctions.

Conservation genetics is the theory and practice of genetics in the preservation of species as dynamic entities capable of evolving to cope with environmental change to minimize their risk of extinction

Conservation genetics encompasses the following activities:

- genetic management of small populations to maximize retention of genetic diversity and minimize inbreeding,
- resolution of taxonomic uncertainties and delineation of management units, and
- the use of molecular genetic analyses in forensics and to understand species' biology.

Purpose of the book

We have endeavoured to make this book appealing to a wide readership. However it is primarily directed towards those encountering the discipline for the first time, either through formal coursework or by self-instruction.

This book is intended to provide an accessible introduction to conservation genetics with an emphasis on general principles

Conservation genetics is a relatively young discipline. While it is founded on more than a century of advances in evolutionary theory, including population genetics, quantitative genetics and plant and animal breeding, it has developed its own unique attributes, specialist journals, etc. In particular, conservation genetics focuses on processes within small and fragmented populations and on practical approaches to minimize deleterious effects within them. It has implications for organizations and individuals with very different immediate concerns. These include zoo staff undertaking captive breeding programs, wildlife biologists and ecologists, planners and managers of National Parks, water catchments and local government areas, foresters and farmers. Perhaps of most importance to the future, conservation genetics is of concern to a growing body of undergraduate and postgraduate students, to whom will fall much of the onus of implementing practical measures. Their enthusiasm was a major stimulus to our preparing this volume.

We have endeavoured to make *Introduction to Conservation Genetics* as accessible as possible to this broad array of readers. At the time we began, there were a number of excellent and scholarly texts on population, quantitative and evolutionary genetics and conservation biology, but no introductory textbook on conservation genetics. We have placed emphasis on general principles, rather than on detailed experimental procedures which can be found in specialist books, journals and conference proceedings. We have assumed a basic knowledge of Mendelian genetics and simple statistics. Conservation genetics is a quantitative discipline as its strength lies in its predictions. We have restricted most use of mathematics to simple algebra to make it accessible to a wide audience.

We trust that colleagues will find this material suitable for a full tertiary course on conservation genetics. At the same time, we hope that it will satisfy the needs of evolutionary geneticists and evolutionary ecologists seeking conservation examples to enthuse their students. Finally, we have endeavoured to create an easily accessible and formalized reference book for both professional conservation geneticists and a wider readership.

Précis of contents

This book provides a broad coverage of all strands of conservation genetics

We have encompassed all of the major facets that comprise conservation genetics, from the impacts of inbreeding and loss of genetic diversity, through taxonomic uncertainties and genetic management of threatened species, to the use of molecular genetic analysis in forensics and resolution of critical aspects of species' biology. We conclude by exploring connections between conservation genetics and the wider field of conservation biology.

Chapter 1 provides an overview of the contemporary conservation context and the reasons why genetic theory and information are crucial in management of endangered species. **Chapter 2** explores the central issues in the application of genetics to conservation biology. Inbreeding reduces reproductive potential and survival and, thereby, increases extinction risk in the short term, while loss of genetic diversity reduces the long-term capacity of species to evolve in response to environmental changes.

We have divided the book into three subsequent sections; **Section I** describes the evolutionary genetics of natural populations, **Section II** explores the genetic consequences of reduced population size, and **Section III** focuses on applications of genetic principles to management of threatened and endangered species in wild, semi-wild and captive situations. The relationships of genetics with broader issues in conservation biology conclude this section.

Section I (Chapters 3–9) covers essential background material in evolutionary genetics. **Chapter 3** deals with the extent of genetic diversity and methods for measuring it. Special attention is paid to comparisons of genetic diversity in endangered versus non-endangered species. **Chapters 4** and **5** describe methods and parameters used to characterize genetic diversity. As major genetic concerns in conservation biology are centred on reproduction and survival in the short term (the effects of inbreeding) and the long term (evolutionary potential and speciation), we have placed considerable emphasis on quantitative (continuously varying) characters, as reproductive fitness is such a character (**Chapter 5**). Molecular measures of genetic diversity, for which vast data sets have accumulated, have a disturbingly limited ability to predict quantitative genetic variation. The paramount importance placed on the functional significance of genetic diversity distinguishes conservation genetics from the related field of molecular ecology, where selectively neutral variation is frequently favoured. **Chapters 6** and **7** introduce factors affecting the amount and evolution of genetic diversity in large populations. The same processes in small populations, including species of conservation concern, are detailed in **Chapter 8**. Chance (stochastic) effects have a much greater impact on the fate of genetic diversity in small, endangered populations than in very large populations, where natural selection has far greater influence. Since conservation genetics focuses on retention of evolutionary potential, **Chapter 9** examines the maintenance of genetic diversity.

Having established the basic principles, Section II concentrates on the genetic implications of population size reduction, loss of genetic diversity (**Chapter 10**), the deleterious consequences of inbreeding on reproduction and survival (inbreeding depression) (**Chapters 11** and **12**), and the genetic effects of population fragmentation (**Chapter 13**). The section concludes with consideration of the population size required to maintain the genetic viability of a population (**Chapter 14**).

Section III explores practical issues, genetic resolution of taxonomic uncertainties and delineation of management units (**Chapter 15**), the genetic management of wild (**Chapter 16**) and captive **(Chapter 17)** populations, and reintroduction **(Chapter 18)**. **Chapter 19** addresses the developing use of molecular genetic analyses in forensics and resolution of cryptic aspects of species biology. **Chapter 20** expands to a broader picture, the integration of genetic, ecological and demographic factors in conservation biology. In particular, we explore the concepts of population viability analysis (PVA) using computer simulations. The final component, **Take home messages** presents a brief summary of the contents of the book, followed by a **Glossary**.

Introduction to Conservation Genetics concentrates on naturally outbreeding species of plants and animals, with lesser attention to self-fertilizing plants. Microbes have not been included, as little conservation effort has been directed towards them.

We have used examples from threatened species wherever possible. However, most conceptual issues in conservation genetics have been resolved using laboratory and domesticated species, non-threatened but related species, or by combined analyses of data sets (typically small) from many species (meta-analyses). Endangered species are clearly unsuitable for experimentation.

Format

The book is profusely illustrated to make it visually attractive and to tap the emotional commitment that many feel to conservation. To highlight significant points and make it easy to revise, the **main points** of each chapter are given in a box at the start of the chapter along with **Terms** used in the chapter and a **Summary** is given at the end of each chapter. Within chapters, the **main points** of each section are highlighted in small boxes. Much of the information is presented in figures, as we find that biology students respond better to those than to information in text or tables. In many figures, the message is highlighted in italics. Numerous examples and case studies have been used to illustrate the application of theory to real world conservation applications. These have been chosen to be motivating and informative to our audience. Case studies are given in **Boxes** throughout the book. Boxes are also used

Extensive effort has been made to motivate readers by making the book attractive, interesting, informative and easy to follow to provide additional or more difficult information in a way that does not impede the flow of information for those who wish to skip such detail.

We are deeply indebted to Karina McInnes, whose elegant drawings add immeasurably to our words.

The text and format have been trialled on four cohorts of final-year undergraduate students at Macquarie University and extensively refined in response to their comments, and those from many colleagues.

The order of topics both within and across chapters has been designed to motivate students

The order of topics throughout the book, and within chapters, is based on our teaching experience. We have chosen to introduce practical conservation issues as early as possible, with the details of parameter estimation etc. provided later. We hope that readers will find it more stimulating to appreciate *why* a parameter is important, before understanding *how* it is logically or mathematically derived. As an example, Chapter 2 directly addresses the relationship between genetics and extinction, and provides an overview of much of the later material, prior to a detailed treatment of inbreeding (Chapters 11 and 12).

Each chapter has been designed to provide instructors with material suitable for one lecture, with additional information for independent study

In presenting material, we have aimed for a balance between that necessary for student lectures, and a comprehensive coverage for advanced students and conservation professionals. The material in each chapter is more than adequate for a single lecture, allowing instructors to choose what they wish to emphasize in their course. However the material in each chapter should not prove overwhelming to their students. Some topics are too extensive for a single lecture. We have therefore divided ‘Evolution in large populations’ into two chapters. We have also allowed some repetition of material, as this is inevitable if different chapters are to be comprehensible on a ‘stand-alone’ basis.

Worked examples and problems with solutions are provided

Everyone who has taught genetics recognizes that mastery of the discipline comes through active participation in problem-solving, rather than passive absorption of facts. Worked **Examples** are given within the text for most equations presented. **Problem** questions are posed at the end of each chapter, together with **Problem answers** and **Revision problems** at the end of the book.

Named species are used in many problem questions, to make them more realistic. These are usually fictitious problems, but reflect situations similar to those that have, or reasonably might have, occurred in the named species. Real data are referenced where used.

Practical exercises are suggested for many chapters

Practical exercises are suggested at the end of chapters covering topics where laboratory exercises are relevant. Most of these have been trialled in our own teaching and are frequently computer exercises, using readily available software. These have proved to be particularly valuable in illuminating the relationship between inbreeding and extinction (Chapter 2), evolutionary genetics of large and small populations (Chapters 6 and 8), maintenance of genetic diversity (Chapter 9), loss of genetic diversity in small populations (Chapter 10) and the use of population viability analysis in management of threatened species

(Chapter 20). Suggestions for molecular genetics practicals are given for Chapters 3, 15 and 19.

Referencing is not intended to be exhaustive, nor to quote primary papers. The references given to reviews and recent papers are sufficient to gain access to the most significant literature. Space does not permit direct reference to many other excellent studies by our colleagues. An annotated list of **General references**, relevant to many chapters, is given at the end of Chapter 1. Readers seeking further detail on specific topics will find an annotated list of suggested **Further reading** at the end of each chapter. We have also included a sprinkling of related books written for popular audiences. These may serve as an introduction to some of the, often controversial, characters involved in conservation biology, and the passions that motivate their work. In the interests of balance, referencing and data presentation are more extensive for contentious topics.

For clarity and brevity, referencing is mainly restricted to reviews and recent papers

As most of the principles of conservation genetics apply equally to different eukaryotic species, we primarily use common names in the text. Genus and species names in the **Index** are cross-referenced to common names.

Controversies

The development of conservation genetics has been driven by what many consider to be a global environmental crisis – 'the sixth extinction'. As a consequence, many other dimensions, economic, political, social, ethical and emotional, impact upon the field. The fate of species, populations and habitats are in the balance. We have flagged these controversies and attempted to provide a balanced, up-to-date view, based upon information available in mid-2000. Where feasible, we have consulted experts to corroborate facts and interpretations. Inevitably, some readers will disagree with some of our views, but we trust that they will accept that alternative interpretations are honestly given. New data will alter perspectives in some cases. For example, the controversial red wolf and northern spotted owl scenarios have changed during the time we were writing the book. We hope that readers find the book as stimulating to read as we found it to write, but not as tiring! Feedback, constructive criticism and suggestions will be deeply appreciated (email: rfrankha@Rna.bio.mq.edu.au).

We will maintain a web site to post updated information, corrections, etc. (http://consgen.mq.edu.au/).

Acknowledgments

Our entries into conservation genetics were initiated by Kathy Ralls of the Smithsonian National Zoo, Washington, DC. Subsequently we have received much-needed support and encouragement from many colleagues, especially from Kathy Ralls, Georgina Mace, Bob Lacy, Rob Fleischer, Stephen O'Brien, Michael Soulé and Ulie Seal. We owe a substantial intellectual debt to Douglas Falconer, author of *Introduction to Quantitative Genetics*. RF and DAB trained using this textbook, and its successive editions have subsequently been major reference sources for us. DAB is particularly appreciative of the mentorship and friendship freely given, over 25 years, by Douglas and his colleagues in The Institute of Animal Genetics, Edinburgh. Not surprisingly, we used Falconer's crisp but scholarly texts as models in our preparation of this book. RF thanks Stuart Barker for his highly influential roles as undergraduate lecturer, PhD supervisor, collaborator and mentor.

Our book could not have been written without the efforts of the students, staff and collaborators in the RF–DAB laboratory. Suzanne Borlase carried out the first experimental modelling of problems in conservation genetics using *Drosophila*. She has been followed by many others, especially Margaret Montgomery and Lynn Woodworth who with Edwin Lowe managed the pedigreed populations for the MVP experiment that have been used in so many studies, including the tests for mutational accumulation done by Dean Gilligan. Roderick Nurthen, in his quiet and efficient way, has supervised all our electrophoretic work, while Phillip England developed microsatellites and supervised their use. Barry Brook and Julian O'Grady conducted computer simulation studies on inbreeding and extinction and on the predictive accuracy of population viability analysis. David Reed completed two important meta-analyses and kept the 'Flyfarm' running while this book was in its final throes. Jennifer Mickelberg took care of business at the Zoo while JDB was in Australia completing the book

The support of our home institutions is gratefully acknowledged. They have made it possible for us to be involved in researching the field and writing this book. The research work by RF and DAB was made possible by Australian Research Council and Macquarie University research grants. RF acknowledges the hospitality of the Smithsonian National Zoological Park during 1997 when the first two drafts were written. JDB also gratefully acknowledges the Smithsonian National Zoological Park for providing a sabbatical to Macquarie University to finalize the preparation of this book. We thank Alan Crowden from Cambridge University Press for his advice and assistance during the writing of the book and to Jayne Aldhouse, Shana Coates, Anna Hodson and Maria Murphy for facilitating the path to publication.

This book could not have been completed without the continued support and forbearance of our wives Annette Lindsay, Vanessa Ballou and Helen Briscoe, and families. Annette and Vanessa (plus Lara and Grace) spent extended periods away from their home countries to facilitate its completion. Vanessa sorted and filed our copious reference collection while Annette corrected the final draft of the book.

We thank the students in the Conservation and Evolutionary Genetics course at Macquarie University in 1998–2001. Their comments, criticisms and suggestions did much to improve the book, especially those from A. Corson, A. Gibberson, H. Ferguson, E. Laxton, H. Macklin and R. Suwito. We are grateful to L. Bingaman-Lackey, D. Cooper, N. Flesness, T. Foose, J. Groombridge, S. Haig, C. Lynch, S. Medina, P. Pearce-Kelly and M. Whalley for supplying information, and to M. Eldridge, R. Fleischer, J. Howard, T. Madsen, B. Pukazhenthi,

I. Saccheri, M. Sun, P. Sunnucks, R. Vrijenhoek, A. Young and G. Zegers for supplying material for illustrations. The book was improved greatly by comments on individual sections and chapters from A. Beattie, L. Beheregaray, M. Burgman, D. Charlesworth, S. Haig, L. Mills (and his class of students at University of Montana), S. O'Brien, K. Ralls, I. Saccheri, P. Sunnucks, A. Taylor, B. Walsh, R. Wayne and A. Young. S. Barker, B. Brook, M. Eldridge, B. Latter, J. O'Grady and D. Reed generously provided detailed comments on the whole text. We apologize to those whose assistance we have omitted to record. We have not followed all of their suggestions and some disagree with our conclusions on controversial issues. Any errors and omissions that remain are ours.

Copyright acknowledgments

We are grateful to the following for kind permission to reproduce copyright material:

The Cambridge University Press for the elephant seal illustrations in Box 8.2 and Ex. 10.5 and the chimpanzee image in Table 3.4 from figures 3–6 and 1–10, respectively, in Austin, C. R. and R. V. Short (1984) *The Evolution of Reproduction* (illustrated by J. R. Fuller); The Kluwer Academic Publishers for: Fig. 19.13 from figure 1 in Lens, L., P. Galbusera, T. Brooks, E. Waiyaki and T. Schenck (1998) Highly skewed sex ratios in the critically endangered Taita thrush as revealed by CHD genes. *Biodiversity and Conservation* 7: 869–873 and Fig. 3.3 from figure 2 page 10 from Avise, J. *Molecular Markers, Natural History and Evolution*; The Oxford University Press for: the figure in Box 3.1 from figure 3 in Gilbert, D. A., C. Packer, A. E. Pusey, J. C. Stephens and S. J. O'Brien (1991) Analytical DNA fingerprinting in lions: parentage, genetic diversity, and kinship. *Journal of Heredity* 82: 378–386; the frontispiece Chapter 19 from figure 1 in Harry, J. L. and D. A. Briscoe. 1988. Multiple paternity in the loggerhead turtle (*Caretta caretta*). *Journal of Heredity* 79: 96–99; the frontispiece Chapter 6 from Plate 8.2 in Kettlewell, B (1973) *The Evolution of Melanism*; Fig. 19.11 from Fritsch, P. and L. H. Rieseberg (1966) The use of random amplified polymorphic DNA in conservation genetics, in *Molecular Genetic Approaches in Conservation*, ed. T. B. Smith and R. K. Wayne, copyright 1996 by Oxford University Press, Inc. Used by permission of Oxford University Press. Inc.; Fig. 7.4 from Map 5 in Mourant, A. E., A. C. Kopéc and Domaniewsha-Sobczak, K. (1976) *The Distribution of the Human Blood Groups and Other Polymorphisms*, Oxford University Press, London; The MIT Press for Fig. 10.2 from figure 2 in Foose, T. J. (1986) Riders of the last ark, in *The Last Extinction*, ed. Kaufman, L. and K. Mallory; The Social Contract Press for Fig. 1.3 from the maps in Tanton, J. H. (1994) End of the migration epoch? *The Social Contract*, 4(3): 162–176 (for critiques of this essay see *The Social Contract* 5(1): 28–47. Note: These texts are available online); The Center for Applied Studies in Forestry for the map in Box 13.1 and the Chapter 16 frontispiece from figure 2 in James, F (1995) The status of the red-cockaded woodpecker and the prospect for recovery, in Kulhavy, D. L., R. G. Hooper and R. Costa, *Red-cockaded Woodpecker: Recovery, Ecology and Management*. Center for Applied Studies, Stephen F. Austin State University, Nacogdoches, TX; CSIRO Publishing for Fig. 15.3 from figure 2 in Johnston, P. G., R. J. Davey and J. H. Seebeck (1984) Chromosome homologies in *Potoroos tridactylus* and *P. longipes* based on G-banding patterns. *Australian Journal of Zoology* 32: 319–324; John Wiley and Sons, Inc. for Fig. 8.6 from figure 1 in Hedrick, P., P. S. Miller, E. Geffen and R. Wayne (1997) Genetic evaluation of the three captive Mexican wolf lineages. *Zoo Biology* 16: 47–69; The Carnegie Institute of Washington for Fig. 5.2 from figures 19, 23 and 25 in Clausen, J., D. D. Keck and W. M. Hiesey (1940) Experimental studies on the nature of species. I. Effect of varied environments on Western North American Plants. *Carnegie Institution of Washington Publications No. 520*. Carnegie Institute, Washington, DC; Blackwell Publishers Ltd, for Fig. 6.4 from figure 1 in Kettlewell, H. B. D. (1958) A survey of the frequencies of *Biston betularia* (L.) (Lep.) and its melanic forms in Great Britain. *Heredity* 12: 551–572; The Evolution Society for the map in Box 12.1 from Vrijenhoek, R. C., Pfeiler, E. and J. Wetherington (1992) Balancing selection in a desert stream-dwelling fish, *Peociliopsis monacha*. *Evolution* 46: 1642–1657; and the National Academy of Sciences for Fig. 19.9 from Bowen, B. W., F. A. Abreu-Grobois, G. H. Balazs, N. Kamenzaki, C. J. Limpus and R. J. Ferl (1995) Trans-Pacific migrations of the loggerhead turtle (*Caretta caretta*) demonstrated with mitochondrial DNA markers. *Proceedings of the National Academy of Sciences, USA* 92: 3731–3734.