

Understanding Forensic DNA

Forensic DNA analysis plays a central role in the judicial system. A DNA sample can change the course of an investigation, with immense consequences. Because DNA typing is recognized as the epitome of forensic science, increasing public awareness in this area is vital. Through several cases, examples, and illustrations, this book explains the basic principles of forensic DNA typing, and how it integrates with law enforcement investigations and legal decisions. Written for a general readership, *Understanding Forensic DNA* explains both the power and the limitations of DNA analysis. This book dispels common misunderstandings regarding DNA analysis and shows how astounding match probabilities such as one-in-a-trillion are calculated, what they really mean, and why DNA alone never solves a case.

Suzanne Bell is Emeritus Professor of Forensic Science at West Virginia University, United States. She served on the National Commission on Forensic Science. In addition to numerous scientific publications and books, she has written multiple editions of *Forensic Science: An Introduction to Scientific and Investigative Techniques* (5th edition, CRC Press, 2019) and *Forensic Chemistry* (3rd edition, CRC Press, 2022).

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Cambridge University Press & Assessment
 978-1-316-51718-5 — Understanding Forensic DNA
 Suzanne Bell , John M. Butler
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CAMBRIDGE
UNIVERSITY PRESS

Cambridge University Press & Assessment
978-1-316-51718-5 — Understanding Forensic DNA

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CAMBRIDGE UNIVERSITY PRESS

University Printing House, Cambridge CB2 8BS, United Kingdom

One Liberty Plaza, 20th Floor, New York, NY 10006, USA

477 Williamstown Road, Port Melbourne, VIC 3207, Australia

314–321, 3rd Floor, Plot 3, Splendor Forum, Jasola District Centre,
New Delhi – 110025, India

103 Penang Road, #05–06/07, Visioncrest Commercial, Singapore 238467

Cambridge University Press is part of the University of Cambridge.

It furthers the University's mission by disseminating knowledge in the pursuit of education, learning, and research at the highest international levels of excellence.

www.cambridge.org

Information on this title: www.cambridge.org/9781316517185

DOI: 10.1017/9781009043311

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First published 2022

Printed in the United Kingdom by TJ Books Limited, Padstow Cornwall

A catalogue record for this publication is available from the British Library.

Library of Congress Cataloging-in-Publication Data

Names: Bell, Suzanne, author. | Butler, John M. (John Marshall), 1969– author.

Title: Understanding forensic DNA / Suzanne Bell, John M. Butler. Other titles: Understanding life series

Description: Cambridge, United Kingdom ; New York, NY : Cambridge University Press, 2022. | Series: Understanding life | Includes bibliographical references and index.

Identifiers: LCCN 2022022812 (print) | LCCN 2022022813 (ebook) | ISBN 9781316517185 (hardback) | ISBN 9781009044011 (paperback) | ISBN 9781009043311 (epub)

Subjects: MESH: Forensic Medicine – methods | Sequence Analysis, DNA – methods | DNA Fingerprinting

Classification: LCC RA1063.4 (print) | LCC RA1063.4 (ebook) | NLM W 700 | DDC 614.1–dc23/eng/20220701

LC record available at <https://lccn.loc.gov/2022022812>

LC ebook record available at <https://lccn.loc.gov/2022022813>

ISBN 978-1-316-51718-5 Hardback

ISBN 978-1-009-04401-1 Paperback

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“This is an excellent book, clearly and comprehensively explaining the scientific aspects of DNA and its analysis and the interpretation of DNA profiles used in forensic science. The sections on emerging issues and dispelling common misunderstandings are particularly useful. The book will be an invaluable guide for scientists and non-scientists alike, and the authors are to be congratulated for an outstanding text.”

Professor Niamh Nic Daéid, Director of the Leverhulme Research Centre for Forensic Science, University of Dundee, UK

“It is essential that the expectations of the public and legal professionals about forensic DNA technology are not based on crime-fiction TV shows, but rather on reality. The authors of this book explain very clearly what DNA can tell us, address existing misconceptions, and share exciting new developments. I enjoyed the real cases included in the book, all very interesting and informative. *Understanding Forensic DNA* is definitely a must-read for anyone who wants to become a DNA expert!”

Dr Lourdes Prieto, Forensic Science Institute, Santiago de Compostela; General Headquarters of the Spanish Forensic Police, Madrid, Spain

“*Understanding Forensic DNA* is the perfect resource for anyone wishing to learn about forensic DNA analysis for the very first time. It is a quick read, ideal for the legal professional and for the student who is just beginning to learn about forensic science. Unlike many books of this type, it is written from a historical perspective that outlines how the discipline has evolved in a few short decades. The book thoroughly examines all existing and past technologies in the most comprehensive way and explains the strengths and limitations of each. One would be hard pressed to think of a relevant topic that has been left out. The case examples are instructive and in no way sensational. For someone who teaches forensic science at the college and graduate level, the book provided me with fresh ideas that I can use to improve my teaching of the subject.”

Lawrence Quarino, PhD, ABC-GKE, Cedar Crest College, Pennsylvania, USA

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“This is a go-to book to enhance anyone’s understanding of forensic DNA. It is of value to the general public, students of forensic sciences, forensic practitioners, and lawyers. It comprehensively covers all key aspects relating to forensic DNA and plenty of additional intriguing elements you may not have considered. There is a focus on the past, present, and future of forensic DNA with descriptions of key historical events. Complex issues are expertly described with helpful examples, tables, and illustrations. The authors provide a balanced view of the utilization and limitations of forensic DNA analyses, demystify common misunderstandings, and explain current concerns. Reading this book refreshed my understanding and brought new insights to my attention. Another outstanding contribution to forensic science by two exceptional authors.”

Roland van Oorschot, Forensic DNA researcher

“Suzanne Bell and John Butler take a journey through the history of forensic genetics. They showcase how this exciting field emerged from its precursor sciences, hematology and serology, and guide the reader through the most recent scientific developments and debates. This entertaining trip provides an unseen density of information and is accessible to a broad audience.”

Dr Walther Parson, Institute of Legal Medicine, Medical University of Innsbruck, Austria; PennState University, Pennsylvania, USA

“This book is full of information delivered in a very accessible way. It will be useful for the general public who want to inform themselves about DNA, and also for those scientists who want to be reminded of the historical basis or background of some techniques. It will be an aid to those a little daunted by the more advanced texts. The use of real-life examples throughout the text gives an excellent insight into the uses and limitations of forensic DNA. The basic facts about DNA and the techniques in routine use are understandably described in greater detail than the more modern techniques which are not so mature.”

Sheila Willis, Honorary Professor, University of Dundee, Fellow of the Leverhulme Research Centre for Forensic Science, UK; retired Director General, Forensic Science Ireland

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“A book that anyone with an interest in DNA profiling will really enjoy. After reading it, you will be well informed, and up to date on current developments. From the inception of biological identification methods to novel methodologies, all is well illustrated by reference to key cases and how the use of DNA profiling has changed forensic investigations. I will be recommending *Understanding Forensic DNA* as a key text to my undergraduates.”

Professor Adrian Linacre, OAM, Chair in Forensic DNA
Technologies, Flinders University, Adelaide, Australia

“This book has a very logical structure and touches on all aspects of forensic DNA analysis in an easy-to-understand fashion. Case examples illustrate specific challenges and make the subject matter come alive. I like that the authors include difficult concepts, for example stochastic effects, and relevant calculations, for instance for genotype frequencies and mixture ratios. Forensic DNA analysis is explained but not simplified.”

Mechthild Prinz, PhD, Professor John Jay College for Criminal
Justice, New York, USA

“Forensic DNA analysis contributes significantly to the provision of justice in many jurisdictions around the world. However, there are still misconceptions about what it really is. It is a technical and multidisciplinary subject encompassing biology, statistics, and the law. A book providing a clear and comprehensive description of forensic DNA analysis is very welcome, not only for public awareness but also for improving communication between those involved in this area.”

Dr Roberto Puch-Solis, Forensic Statistician, Leverhulme
Research Centre for Forensic Science, University of Dundee, UK

“The authors present an excellent overview of the history of forensic DNA analysis, starting with biological identification based on serology in the 1980s and moving on to the first application of DNA profiling by Alec Jeffreys to help solve a case in 1986. The chapters that follow discuss the progressive use of ever smaller fragments and quantities of DNA for identification. This evolution was accomplished by new discoveries about the human genome in conjunction with more sophisticated and sensitive analytical tools and techniques to process

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DNA. The book discusses the issues that are raised when new methods are applied to DNA evidence, ranging from increased sensitivity to interpretation of data and acceptance in the courtroom. The final chapters deal with cutting-edge technology and new issues, including the use of DNA to create phenotypic profiling, familial searching, genetic genealogy, behavioral profiling, and non-human applications.”

Mark Okuda, Evergreen Valley College, San Jose,
California, USA

“DNA has brought about a revolution in all fields of the life sciences, and in medicine, biotechnology, and pharmaceuticals. But, arguably, the public knows of DNA from its impact on ancestry and forensics. These are closely related, for in both cases DNA is the key to determining connections, whether between people or between evidence and perpetrator. Bell and Butler’s book is therefore welcome and timely, setting out in clear and readable language the basis of DNA-based forensics and how it is used in practice. Most importantly, it also makes clear the limitations of DNA forensics, both the technical and societal issues that it raises. The authors write that their goal is ‘to provide an overview of DNA methods, their use, and the current issues in the field.’ They have succeeded admirably.”

Professor Jan A. Witkowski, Cold Spring Harbor Laboratory,
New York, USA

“*Understanding Forensic DNA* provides an intriguing overview of one of the most innovative fields of applied sciences. Written by two compelling narrators with proven expertise in molecular and forensic genetics, this book not only covers the fascinating scientific background of current forensic DNA technologies but also delves into recent innovations such as investigative genetic genealogy that helped to identify the Golden State Killer. Similarly, other high-profile cases are used to illustrate the evidential power of a given technology. At the same time, a critical distance is maintained, such as when addressing cognitive bias or discussing the use of high-throughput genotyping that may threaten the privacy of suspects or witnesses. This book is worth reading not only for forensic science aficionados but also for

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DNA experts who want to discover state-of-the-art resources for education and training.”

Professor Peter M. Schneider, Institute of Legal Medicine,
Cologne, Germany

“Suzanne Bell and John Butler have put together an outstanding text on forensic DNA that the general public can understand. The reader can easily appreciate the impact and limitations of the use of DNA technology through the excellent descriptions and illustrations. This book should be on the shelf of every educator, student, scientist, lawyer, investigator, or anyone working in the criminal justice system who needs to understand the role of DNA in forensic science.”

Thomas A. Brettell, PhD, Professor of Chemistry, Cedar Crest
College, Pennsylvania, USA

“This easy-to-understand book is a guide I will reference again and again. If you want to truly understand how DNA is used in forensic science, this is the book for you!”

CeCe Moore, Chief Genetic Genealogist, Parabon

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To the unsung heroes in libraries everywhere – the research for this book would have been impossible without them. Also, to my co-author, colleague, and friend, Dr. John Butler, who has done more to make forensic science better than anyone I know.

Dr. Suzanne Bell

To Margaret Kline, my NIST colleague for many years in the DNA Technology and Applied Genetics Groups, who showed by example how to be a great scientist with her attention to detail and passion for excellence.

Dr. John M. Butler

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Foreword

“I go where the evidence takes me.” “Witnesses lie; the evidence does not lie.” Statements like these made a strong impression on me many years ago when I watched some episodes of one of those forensic investigation TV series. The message conveyed by these statements is that forensic evidence, especially forensic DNA evidence, is the most reliable kind, and it can guide an investigator toward the person who committed a crime. However, things are not as simple and straightforward as that. As Suzanne Bell and John Butler clearly explain in this brilliant book, forensic DNA evidence can lead neither to infallible, nor to definitive, conclusions. The reason for this is that as we do not have the DNA profiles of everyone in the world, we can only figure out the probabilities that someone could have or not have the profile found at a crime scene just by chance. We also need additional evidence to confirm that the evidence found at the scene is indeed related to the crime. This does not make forensic DNA analyses useless. However, as with any science-related endeavor, forensic DNA analysis has a certain potential as well as particular limitations. Suzanne Bell and John Butler have produced the best, most accessible, and most concise guide possible to the science of forensic DNA. Some of the most striking features of the present book are the detailed explanations of the methods used, which are accompanied by very informative and clear illustrations. Read this book, and the forensic investigation TV series may never look the same again.

Kostas Kampourakis, Series Editor

Preface

Forensic DNA typing has leaped into the mainstream of criminal justice over the last few decades. DNA analysis is described as “the gold standard,” and for a good reason. DNA evidence has revolutionized biological identification and given the justice system one of its most powerful tools. As with all such techniques, however, analytical power can lead to unexpected problems and consequences. The science and technology of DNA typing have outpaced general understanding, leaving policymakers scrambling to catch up. We will explore these aspects of modern forensic DNA testing methods.

Forensic DNA analysis plays a central role in the judicial system and the life-changing decisions it renders. We will introduce many such cases in the pages to come. This book is intended to explain the basic principles of forensic DNA typing and how it integrates with law enforcement investigations and legal decisions. Because DNA typing is recognized as the epitome of forensic science, increasing public awareness and understanding in this area is vital. Many misconceptions regarding DNA persist, and DNA testing is not infallible. Accordingly, we will explore the power of the technology as well as important pitfalls and limitations. A DNA sample can change the course of an investigation, with immense consequences for one or more people. The more the public understands forensic DNA typing, the more likely it will be utilized to best effect. We can accomplish this admittedly ambitious goal by clearing common misconceptions.

We will set the stage with a brief overview of biological identification, which began with blood typing techniques developed in the early 1900s. These methods led to the use of probability and statistical approaches that are still

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used today in DNA typing. The first application of DNA in a criminal case was in 1986 in England, in which a double murderer was brought to justice. We will see how DNA evidence, as powerful as it is, does not solve cases by itself. DNA results are always part of an extensive investigation.

Following the background information, we will move on to current DNA profiling methods and practices. What used to take days or weeks can now be completed in a few hours. Techniques have steadily improved such that DNA residues the size of the period at the end of this sentence can be typed. The ability to detect DNA transferred by touching has been a boon but also a problem. Even minute amounts of DNA, which may or may not be relevant to a case, can be detected on almost any surface. Finding someone's DNA on an item does not tell you when, how, or why it got there. Sorting through complex mixtures from "touch evidence" remains a significant challenge facing forensic DNA. The last two chapters will delve into emerging technologies and current issues, including investigative genetic genealogy.

Our goal is to provide an overview of DNA methods, their use, and the current issues in the field. This book is not intended to be a standalone textbook, nor is it a primary resource for legal or investigative purposes. If, as a reader, you wish to explore these topics further, the books mentioned in the *Acknowledgments* and the *References and Further Reading* provide a good starting point.

Acknowledgments

The foundation of this work is the three-book series by John Butler:

Butler, J. M. (2010). *Fundamentals of Forensic DNA Typing*. San Diego, CA: Academic Press/Elsevier.

Butler, J. M. (2012). *Advanced Topics in Forensic DNA Typing: Methodology*. San Diego, CA: Academic Press/Elsevier.

Butler, J. M. (2015). *Advanced Topics in Forensic DNA Typing: Interpretation*. San Diego, CA: Academic Press/Elsevier.

Selected figures and tables from these are graciously provided as non-copyrighted materials by Dr. Butler and the National Institute of Standards and Technology (NIST), US Department of Commerce.

Dr. Suzanne Bell

I owe a debt to the staff at NIST who reviewed the book and provided feedback and guidance. Also, thanks to the editors and staff of Cambridge University Press and the series editor, Kostas Kampourakis, for allowing us to add a book to the wonderful *Understanding Life* series.

Dr. John M. Butler

I appreciate the opportunity to work with Suzanne Bell on a book intended for the general public.

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