Cambridge University Press 978-1-108-84198-6 — Understanding DNA Ancestry Sheldon Krimsky Frontmatter <u>More Information</u>

#### **Understanding DNA Ancestry**

DNA ancestry companies generate revenues in the region of \$1 billion each year, and the company 23andMe is said to have sold 10 million DNA ancestry kits to date. Although evidently popular, the science behind how DNA ancestry tests work is mystifying and difficult for the general public to interpret and understand. In this accessible and engaging book, Sheldon Krimsky, a leading researcher, investigates the methods that different companies use for DNA ancestry testing. He also discusses what the tests are used for, from their application in criminal investigations to discovering missing relatives. With a lack of transparency from companies in sharing their data, absent validation of methods by independent scientists, and currently no agreed-upon standards of accuracy, this book also examines the ethical issues behind genetic genealogy testing, including concerns surrounding data privacy and security. It demystifies the art and science of DNA ancestry testing for the general reader.

Sheldon Krimsky is Lenore Stern Professor of Humanities and Social Sciences and Adjunct Professor of Public Health and Community Medicine at Tufts University, Massachusetts, USA. His research has focused on the linkages between science/technology, ethics/values, and public policy. His areas of specialization include biomedical sciences, bioethics, science and technology studies, risk assessment and communication, social history of science, and environmental health. He is the author of over 200 articles and reviews, and author, coauthor, or editor of 16 books.

Cambridge University Press 978-1-108-84198-6 — Understanding DNA Ancestry Sheldon Krimsky Frontmatter <u>More Information</u>

The **Understanding Life** series is for anyone wanting an engaging and concise way into a key biological topic. Offering a multidisciplinary perspective, these accessible guides address common misconceptions and misunderstandings in a thoughtful way to help stimulate debate and encourage a more in-depth understanding. Written by leading thinkers in each field, these books are for anyone wanting an expert overview that will enable clearer thinking on each topic.

Series Editor: Kostas Kampourakis http://kampourakis.com

#### Published titles:

Understanding Evolution	Kostas Kampourakis	9781108746083
Understanding Coronavirus	Raul Rabadan	9781108826716
Understanding Development	Alessandro Minelli	9781108799232
Understanding Evo-Devo	Wallace Arthur	9781108819466
Understanding Genes	Kostas	9781108812825
-	Kampourakis	
Understanding DNA Ancestry	Sheldon Krimsky	9781108816038
Forthcoming:		
Understanding Intelligence	Ken Richardson	9781108940368
Understanding Metaphors in	Andrew S.	9781108940498
the Life Sciences	Reynolds	
Understanding Creationism	Glenn Branch	9781108927505
Understanding Species	John S. Wilkins	9781108987196
Understanding the Nature-	Eric Turkheimer	9781108958165
Nurture Debate		
Understanding How Science	Kevin McCain	9781108995504
Explains the World		
Understanding Cancer	Robin Hesketh	9781009005999
Understanding Forensic DNA	Suzanne Bell and	9781009044011
	John Butler	
Understanding Race	Rob DeSalle and	9781009055581
	Ian Tattersall	
Understanding Fertility	Gab Kovacs	9781009054164

Cambridge University Press 978-1-108-84198-6 — Understanding DNA Ancestry Sheldon Krimsky Frontmatter <u>More Information</u>

# **Understanding DNA Ancestry**

SHELDON KRIMSKY Tufts University, Massachusetts



Cambridge University Press 978-1-108-84198-6 — Understanding DNA Ancestry Sheldon Krimsky Frontmatter <u>More Information</u>

#### 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/9781108841986 DOI: 10.1017/9781108895651

© Sheldon Krimsky 2022

This publication is in copyright. Subject to statutory exception and to the provisions of relevant collective licensing agreements, no reproduction of any part may take place without the written permission of Cambridge University Press.

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.

ISBN 978-1-108-84198-6 Hardback ISBN 978-1-108-81603-8 Paperback

Cambridge University Press has no responsibility for the persistence or accuracy of URLs for external or third-party internet websites referred to in this publication and does not guarantee that any content on such websites is, or will remain, accurate or appropriate.

Cambridge University Press 978-1-108-84198-6 — Understanding DNA Ancestry Sheldon Krimsky Frontmatter <u>More Information</u>

> "Sheldon's book represents a much needed historical, technical, and ethical treatment of this rapidly evolving and growing industry. Understanding DNA Ancestry tackles a complex topic that many are fascinated by but few have the educational background to appreciate fully un-shepherded, and does so in a way that is accessible and easy to internalize by the very lay readers who have literally built the entire industry with their demand. His book is not only timely, but way, way overdue, and is special for many reasons. For one, this book comes from an author who is independent of the industry; the vast majority of white-paper, journal publications, editorials, and books do not, and often come with unfair biases and agendas. Secondly, and uniquely, the book covers the entire spectrum of societal, technical, and potentially personal implications of this new technology. With comprehensive scope, Sheldon brings you to appreciate fully the mechanics of the methods; the impact of this new technology on medicine, privacy, and other ethical considerations; their upsides and downsides, power and limitations; and how they may affect you personally - we come to appreciate the technology in its universal totality, including even its impact on Forensic Science, in terms not just of phenotyping (ascribing physical characteristics from crime-scene DNA), but of genetic genealogy, which is the ultimate evolution of the technology that almost all ignore, but that touches us all in terms of public safety. Sheldon's book is historically and technologically accurate, socially responsible, objective, inclusive in scope, and sensitive to personal risks and rewards. As such, this book is in my view instrumental for anyone considering a genetic ancestry test. If you are a lay consumer of genetic ancestry testing products, it has my highest recommendation for you."

Tony N. Frudakis, Ph.D., Forensic Scientist, Albuquerque Police Department DNA Laboratory, and Founder DNAPrint Genomics, Inc. (1999)

Cambridge University Press 978-1-108-84198-6 — Understanding DNA Ancestry Sheldon Krimsky Frontmatter <u>More Information</u>

> "This book has it all - science and technology, history, ethics, law, and interesting stories of genealogy. It is classic Krimsky - a truly scholarly endeavor made incredibly approachable. Krimsky goes into sufficient depth to empower the reader with the background necessary to appreciate and understand DNA ancestry fully. The book is comprehensive, describing the key discoveries leading to the modern science of ancestry, including the history and development of the multiple generations of technologies used to achieve the resolution of understanding we have today. Applications of the technology's uses and misuses are covered, as well as privacy and ethical considerations. Krimsky is a terrific storyteller of individual cases, where people found out they weren't who they thought they were. He remains in the background throughout as a balanced and unbiased observer. A most interesting and timely book that will inform, entertain, and empower the millions who have had or are considering a consumer DNA test."

> > David R. Walt, Harvard Medical School

"How do private companies, like Ancestry.com and 23&me, use DNA to determine your ancestry? How does this differ from forensic DNA used by police and the FBI? And should we be worried about some or all of this? In this compelling book, Sheldon Krimsky provides clear, informative, and nuanced answers to all these questions and more."

> Naomi Oreskes, Henry Charles Lea Professor of the History of Science and Affiliated Professor of Earth and Planetary Sciences, Harvard University

"Professor Sheldon Krimsky's book *Understanding DNA Ancestry* introduces readers to the vast panoply of complications that can face those who are interested in the sources of their ancestors. Such readers may be surprised that results

Cambridge University Press 978-1-108-84198-6 — Understanding DNA Ancestry Sheldon Krimsky Frontmatter <u>More Information</u>

> from different genetic ancestry companies can vary in their conclusions about the results from company to company. Test-takers may sometimes confront unexpected surprises they had never suspected about co-existing family members, or discover that an assumed relationship may not be genetically based. They will also learn that some ancestry companies help the FBI in identifying criminals using those companies' DNA sequencing collections. What is more, the eager readers may find out that they are learning more than they knew about genetics. Entering these new studies raises questions about whether supposed classical races of peoples have a genetic basis at all.

> Professor Krimsky has a long career in preparing detailed collections that have been important to fields of science."

Jonathan Beckwith, Professor Emeritus, Harvard Medical School, Department of Microbiology (and a geneticist)

"In the last three years, there has been a sharp surge in genetic ancestry testing, not only in the US, but across the globe. There are now more than seventy companies promoting an array of such offerings, which range from recreational to medical to forensic uses and claims. Sheldon Krimsky has provided an illuminating social history of these developments, with lucid prose that explains the uses and limits of such testing. But *caveat emptor*: many consumers will be dismayed to learn that the seductive lure and broad claims commonly outstrip the capacity of these tests to provide clear and replicable results."

Troy Duster, Chancellor's Professor Emeritus, University of California, Berkeley

Cambridge University Press 978-1-108-84198-6 — Understanding DNA Ancestry Sheldon Krimsky Frontmatter <u>More Information</u>

> "Sheldon Krimsky has written an illuminating description in amazingly simple language on a most difficult but essential subject – ancestry.

> Krimsky's explanations of how DNA ancestry works and what it means for modern society are essential contributions to how we, as humans, understand our own variation. Understanding these important aspects of our variation is critical to our worldview and the place of our species in the modern world."

> > Robert DeSalle, American Museum of Natural History, New York

Cambridge University Press 978-1-108-84198-6 — Understanding DNA Ancestry Sheldon Krimsky Frontmatter <u>More Information</u>

For Eliot Krimsky, Lisa Benger, and Siona Rose Krimsky

Cambridge University Press 978-1-108-84198-6 — Understanding DNA Ancestry Sheldon Krimsky Frontmatter <u>More Information</u>

## Contents

	Foreword	page xv
	Acknowledgments	xvi
1	Introduction	1
	Familial and Historical Genealogy	2
	The Birth of Genome Sequencing	2
	Goal for the Book	3
	The Varieties of Ancestry	4
	Structure of the Book	6
2	The Business of DNA Ancestry	8
	The First DNA Ancestry Company	8
	Growth of the Digital Ancestry Sector	9
	Ancestry and Health	11
	Expanding the Types of Genetic Tests	13
	Behind Consumer Interest in Ancestry Tests	14
3	What Our Genomes Tell Us about the Geographical Origins	
	and Movements of Early Human Populations	17
	A Hypothetical Case of Isolated Populations	18
	The Evolution of Homo sapiens	20
	Genetic Diversity in the Human Population	22
4	The Science behind DNA Ancestry Testing	26
	Allele Frequencies and Gene Variants	26

Cambridge University Press 978-1-108-84198-6 — Understanding DNA Ancestry Sheldon Krimsky Frontmatter <u>More Information</u>

#### xii CONTENTS

	Patents on Inferring Ancestry from DNA Ancestry Markers	27 29
	Validation of Ancestry Inference	33
	Reference Panels	34
	Allele Frequencies in Populations	35
	Stages for Inferring Ancestry	36
5	Ancestry Informative Markers	41
	Forensic Applications of Genetic Markers	42
	From Forensic to Ancestry Markers	43
	Mitochondrial DNA Markers	44
	Y Chromosome Ancestral Markers	45
	Autosomal SNP Ancestry Markers	46
6	Ancestry DNA Population Reference Panels	50
	Proprietary Reference Panels	51
	Validation of Reference Panels	55
7	Comparing a Donor's DNA to Reference Panel Populations	60
	DNA Variants	60
	Population-Specific Variants	63
	Population Frequencies of Variants and FST Statistics	66
8	Probing Your DNA	73
	Analysis of Customer DNA and Microarrays	74
	Optical Fibers and DNA Probes	76
9	Forensic Applications of Ancestry DNA Results	79
	Discovery of DNA Identification Method	79
	The Grim Sleeper	80
	The Golden State Killer	81
	Identity by Descent	83
	Phenotyping	86
10	Privacy, Personal Identity, and Legal Issues	88
	Ownership of One's Cells and Genetic Information	88
	Ancestry Companies and the Privacy of Genetic Data	90

#### Cambridge University Press 978-1-108-84198-6 — Understanding DNA Ancestry Sheldon Krimsky Frontmatter <u>More Information</u>

	CONTENTS	Xiii
Advertising Claims		92
Forensic Applications and Privacy		93
Personal Identity through DNA Tests		98
Race and DNA Ancestry		102
11 Discovering Unknown, Missing, or Mistaken Relatives		106
DNA and Paternity Claims		106
Hidden Family Secrets		110
Family Ethnicity: Hidden Secrets		112
Secret Sperm		113
Breaking the Pure Ethnic Descent		115
12 Accuracy, Consistency, and Validation of DNA Ancestry Test	s	118
Comparing Different Company Ancestry Results		118
DNA Markers and Ancestry Regions		120
Ancestry Tests with Twins		121
Self-Declared Ancestry vs. Genomic Ancestry		124
13 Conclusion		126
Summary of Common Misunderstandings		129
References		131
Index		146

Cambridge University Press 978-1-108-84198-6 — Understanding DNA Ancestry Sheldon Krimsky Frontmatter <u>More Information</u>

## Foreword

Who am I? Where do I come from? The answers to these questions might seem self-evident to many people. Being aware of the history of their ancestors, their genealogical ancestry, they are able to talk about their ancestry, their ultimate "roots," and their resulting ethnicity/nationality. Others are still asking these questions, perhaps because they lack information about their genealogical ancestry. To find answers, they often turn to DNA tests that are supposed to provide information about their genetic ancestry. Thus, people hope, based on their presumed genetic ancestry, to be able to make inferences about their genealogical ancestry. However, this is not as simple as it may seem. Currently, millions of people have taken DNA ancestry tests, some hoping to find such answers, others just doing it as a recreational activity. But all these seem to take the validity and the utility of these tests for granted, often overlooking the fact that these tests are based on specific assumptions and have important limitations. The present book by Sheldon Krimsky does a lot at the same time: It diligently explains the science of DNA ancestry testing, it responsibly considers its limitations and pitfalls, and it sensitively discusses the related ethical and legal issues. If you are thinking about taking a DNA ancestry test, or if you are simply interested in this topic, then this fascinating book will guide you through the various issues at stake. It is a tour-de-force that provides an informed and pragmatic view of DNA ancestry testing that would be useful to anyone interested in really understanding this topic.

#### Kostas Kampourakis, Series Editor

Cambridge University Press 978-1-108-84198-6 — Understanding DNA Ancestry Sheldon Krimsky Frontmatter <u>More Information</u>

## Acknowledgments

I wish to thank David Walt, Tony Frudakis, and Michael Carson for reading selected chapters and providing feedback, John M. Coffin for clarifying some historical points about the first bacteria and viruses fully sequenced, and Bennett Greenspan, CEO of Family Tree DNA for agreeing to an interview. I am indebted to the excellent editors at Cambridge University Press: Kostas Kampourakis and Katrina Halliday.