

## Understanding Genes

What are genes? What do genes do? These questions are not simple and straightforward to answer; at the same time, simplistic answers are quite prevalent and are taken for granted. This book aims to explain the origin of the gene concept, its various meanings both within and outside science, as well as to debunk the intuitive view of the existence of “genes for” characteristics and disease. Drawing on contemporary research in genetics and genomics, as well as on ideas from history of science, philosophy of science, psychology, and science education, it explains what genes are and what they can and cannot do. By presenting complex concepts and research in a comprehensible and rigorous manner, it examines the potential impact of research in genetics and genomics and how important genes actually are for our lives. *Understanding Genes* is an accessible and engaging introduction to genes for any interested reader.

Kostas Kampourakis is the author and editor of books about evolution, genetics, philosophy, and history of science, and the editor of the Cambridge University Press book series *Understanding Life*. He is a former editor-in-chief of the journal *Science & Education*, and the book series *Science: Philosophy, History and Education*. He is currently a researcher at the University of Geneva, where he also teaches at the Section of Biology and the University Institute for Teacher Education (<http://kampourakis.com>).

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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.

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KOSTAS KAMPOURAKIS  
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*“Understanding Genes is an essential guide to this important, complex, and sometimes incendiary topic. In his clear and balanced discussion, Kostas Kampourakis cuts through all the hype and misconception that often surround the debate about what genes are and what they do, and provides the most honest and careful discussion I have seen of how DNA participates in the processes that support life. In doing so, he reveals the real promise, limitations, and dilemmas of the current age of genomics.”*

Philip Ball, science writer and author of *How to Grow a Human*

*“Did you know that two blue-eyed people can have a brown-eyed child? Why calico cats are (almost) always female? It’s in the genes, but it’s not *all* in the genes. Kampourakis shows that, while genes are unquestionably important, fears of ‘designer babies’ are both overblown and misguided. Genes alone do not make you who you are. They are not the ultimate essence of life. *Understanding Genes* is simply the best book out there for students or anyone wanting a smart, thoughtful introduction to what genes are and do – and what they aren’t and don’t.”*

Nathaniel Comfort, Professor, Department of the History of Medicine,  
Johns Hopkins University

*“Kampourakis has produced a comprehensive but highly readable introduction to genetics and genomics. His take on the fallacy of genetic fatalism is a must-read for both geneticists and the casual reader . . . The role of genetics and genomics in society is treated comprehensively by Kampourakis. He has produced a very readable book with an important message about genetic fatalism – it doesn’t exist!”*

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

*“ . . . provides a plain, rich, and direct narrative of what a gene is and is not, with practical examples of how genes relate to our daily life . . . clearly identifies controversial views in [the] fields of genetics, genomics, cell and organismic biology, and clarifies them for the comprehension of the just initiated as well as the experienced reader.”*

Carlos Sonnenschein MD, Tufts University School of Medicine, Boston,  
MA, USA, and Centre Cavailles, École Normale Supérieure,  
Paris, France

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*“Understanding Genes is a remarkably clear, rigorous, and yet accessible review of the biological and social roles of genes. Building on a wide range of sources including history, biology, philosophy, and social studies, the book identifies a variety of gene concepts currently in use, illustrates their significance through a wealth of concrete examples, and discusses the relations between these different ways of understanding genes. By deftly combining conceptual analysis with empirical evidence, the book succeeds in comprehensively introducing this complex subject without oversimplifying. It is highly recommended to readers venturing in this domain for the first time, as well as to experts wishing to expand their perspective.”*

Sabina Leonelli, University of Exeter, UK

*“Genes – many people use the word, few understand its many meanings and how they changed over time: from tools to think with, to tools to trace ancestors with. This book guides the reader through the many transformations of this concept from conception to misconceptions, from Mendel to the media. We learn about genetics, genomics, and post-genomics, but also about the interactions between scientific and public understandings and the role of metaphor in spicing things up. Readers come to realize that genes are neither essences, nor things, nor actors; genes only work in context, and in collaboration with each other within an interactive genome. This makes it difficult to find easy solutions to medical problems, but it also means that genes don’t determine who we are. This book is more than a guide to understanding genes; it is essential reading for everyone interested in the role that genes play in science and culture.”*

Brigitte Nerlich, University of Nottingham, UK

*“In rigorous but uncomplicated prose, Kostas Kampourakis gives us a present we wish we could have received 100 years ago: a clear explanation of what genes do, what they do not do, what they are, and what they are not. In doing so, he teaches us salutary lessons in both the history and philosophy of science and in human psychology. At a time when our ability to manipulate nature is reaching new levels, Kampourakis provides a road map for understanding the relevance of genetics to our lives. This is a book everyone should read.”*

Oren Harman, Senior Research Fellow at the Van Leer Jerusalem Institute and Chair of the Graduate Program in Science, Technology and Society, Bar Ilan University, and author of *The Man Who Invented the Chromosome*, *The Price of Altruism*, and *Evolutions: Fifteen Myths that Explain Our World*

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*“Understanding Genes* is the first book that provides an honest, nuanced, and full accounting of how genes operate in an organism that is accessible to a general reader. I have not seen in one volume such clear analysis of the ‘gene’ and its deconstruction from a primary cause to a ‘segment of DNA’ that is a necessary, but not sufficient, cause of different types of biochemical events. The book exhibits the expertise of an author whose breadth of knowledge of genetics, history and philosophy of science, and science education makes this book exceptionally valuable as a scientific antidote to the tide of popular oversimplifications and the trend in the scientific literature of genetic reductionism.”

Sheldon Krinsky, Lenore Stern Professor of Humanities & Social Sciences, and Adjunct Professor of Public Health & Community Medicine, Tufts University

*“If you are looking for a concise and up-to-date book on the role of genes (and the science of genes) in our society, look no further: *Understanding Genes* is an accessible, yet nuanced, account of how the concept of the gene has developed throughout history, how its cultural and social meanings have changed, and how genetic factors influence the expression of human behavior and diseases. It conveys not only the basics of genetic thinking, but also a sense for how our understanding of what genes are, and what they do, is always also a response to the big questions that society asks at any given time. I highly recommend this beautifully written book to students, journalists, researchers from other disciplines, and in fact anyone seeking to understand the role of genes – and of genetics – in our world.”*

Barbara Prainsack, University of Vienna, Austria

*“In *Understanding Genes*, Kostas Kampourakis draws on history and popular culture as well as the latest scientific research to help the beginning reader to grasp what genes are, why they are important, and how to give that importance its due without hype or hysteria. Anyone looking for an introduction to genetics that is both reliable and readable need look no further.”*

Gregory Radick, University of Leeds, UK

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“This excellent book is comprehensive, detailed, and amazingly informative, yet eminently readable; it’s a really lovely synthesis of the past half-century of thought about what genes are, what genes do, and why they – along with their contexts – are so extremely important. Kampourakis presents biological facts with a ‘systems’ perspective that remains unwaveringly attentive to the fact that genetic information is always embedded in a context, a context that renders developmental outcomes unpredictable from DNA sequence information alone. Because the book holds fast to this valuable perspective, it brilliantly and clearly makes the case that there are no such things as genes ‘for’ specific traits or diseases, despite what we might have gleaned from other media. By deploying wonderful new metaphors and unpacking older and potentially misleading ones, Kampourakis helps readers to avoid many of the misunderstandings that arise from various sources. Accurate and poised at the cutting edge, this primer is lucid enough to be accessible for the general public and students learning about genetics for the first time, but erudite enough for scientists interested in what we currently know about genes. *Understanding Genes* beautifully illustrates the shortcomings of the Human Genome Project, genome-wide association studies, and current personalized medicine and direct-to-consumer genetic tests, clarifying what we now understand and what we are still very much in the dark about. This book serves as an important antidote to the optimistic hype that has a lot of people believing that treatment programs based on individuals’ DNA are just around the corner. They are not, and this book explains why, making it a truly *important* read for everyone – or at least, everyone who has genes.”

David S. Moore, Pitzer College and Claremont Graduate University



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To my brother, Yiannis, and our mother, Evaggelia, who have  
always made me think hard about “nature” and “nurture”

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## Foreword

Following on from the success of his *Understanding Evolution* book, Kostas Kampourakis and I discussed the need for a book that addressed the common misconceptions surrounding genes in a comparable way. *Making Sense of Genes* resulted and was published in 2017, receiving universally positive feedback for its eloquent, multidisciplinary treatment of the fundamental questions: What are genes and what do they do? From the success of this book came an invitation for Kostas to speak at the Cambridge Science Festival; it was an honour to host him in Cambridge and, as we walked to the venue, share with him his book on display in the window of the Cambridge University Press bookshop.

Since then Kostas and I have developed the *Understanding Life* series together. Our vision for it is to provide concise, accessible guides to key topics, written by leading thinkers in the field and focusing on the common misconceptions and misunderstandings that are potential barriers to gaining a deeper understanding. Genes are, of course, an obvious topic for inclusion in this series, and hence this book arose, which updates and condenses the coverage in *Making Sense of Genes*, providing an invaluable introduction to the topic. Its discussion of the role of genetics and genomics in society, and their presentation in the media, are particularly timely.

It has been an enormously fulfilling, fun, and enjoyable experience working with Kostas on this book and on the series more broadly. I'm proud of what we have achieved with it and it makes an important

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contribution to the Press' mission, to publish and disseminate high-quality information to aid learning. I very much look forward to seeing how *Understanding Life* develops and grows in the future as I move on to new adventures.

**Dr. Katrina Halliday**  
**Executive Publisher, Life Sciences**  
**Cambridge University Press**

## Preface: Genes, Science, and Science Fiction

Antonio and Marie Freeman follow a nurse who shows them the way to the geneticist's office. Marie carries their young son, Vincent. As soon as they sit down, a monitor turns on and they see four embryos, each consisting of a few cells. Next to another monitor across the room, the geneticist is sitting. He says: "Your extracted eggs, eh . . . Marie, have been fertilized with Antonio's sperm. After screening we are left, as you see, with two healthy boys and two very healthy girls. Naturally, no critical predispositions to any of the major heritable diseases." The geneticist stands up and approaches the couple. "All that remains is to select the most compatible candidate," he says as he sits down next to them. "First, we might as well decide on gender – have you given it any thought?"

"We would want Vincent to have a brother, you know, to play with," Marie says, looking at her son, who is playing on the ground with a ball-and-stick molecular model.

"Of course you would. Hello Vincent," the geneticist says, smiling at him. Vincent smiles back and shyly says "Hi."

The geneticist turns to the couple and continues: "You have specified hazel eyes, dark hair, and fair skin. I have taken the liberty of eradicating any potentially prejudicial conditions: premature baldness, myopia, alcoholism and addictive susceptibility, propensity for violence, obesity, etc."

"We didn't want any, I mean diseases yes, but . . ." Marie interrupts the geneticist, and she and Antonio look at each other. Antonio continues:

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“Right, we were just wondering if it’s good to just leave a few things to chance.”

The geneticist replies immediately: “You want to give your child the best possible start. Believe me, we have enough imperfection built in already. Your child doesn’t need any additional burdens. Keep in mind, this child is still you. Simply the best of you. You could conceive naturally a thousand times and never get such a result.”

This is a scene from the 1997 dystopian film *GATTACA*, which presents an oppressive society where sexual intercourse between a man and a woman is no longer the natural way of having a child. Instead, *in vitro* fertilization and genetic screening of the emerging embryos are invoked to ensure that only children with the desired characteristics and without any “prejudicial conditions” are brought to life. Vincent, who was not conceived through such a process, is considered to be genetically inferior. This is why Marie and Antonio decided to have a second child through *in vitro* fertilization and genetic screening.

*GATTACA* was a science fiction film that postulated a future society of genetic discrimination. At that time, the Human Genome Project was still under way and the potential for genetic screening was relatively limited – even though the expectations were great. Preimplantation genetic diagnosis was possible at the time, but it was usually used for particular genetic diseases in the cases of couples with a family history of those diseases (diagnosis is the search for conditions or specific alleles – different versions of the same gene – in people already considered as likely to have them, whereas screening refers to the search for conditions or specific alleles in the general, healthy, and asymptomatic population).

Nowadays, almost a quarter-century later, things seem to have changed. According to one estimate by Antonio Regalado, senior editor for biomedicine for the *MIT Technology Review*, more than 26 million people had taken a genetic test by the beginning of 2019. Margo Georgiadis, president and chief executive officer of Ancestry.com, has estimated that by early 2020, 30 million people had taken a DNA test. Even though in most of these cases the tests were related to ancestry, one could expect prospective parents to be interested in predictive genetic tests to select the healthiest, smartest,

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loveliest – or whatever – babies. Does this mean that we are entering a *GATTACA*-style era? How far are we from such possibilities? This might happen if indeed genes somehow cause conditions in an if-you-have-the-gene-then-you-will-have-the-condition manner. In that case, we could indeed screen embryos and select only those with the desired characteristics. If we came to know the “genes for” particular characteristics, then we could distinguish between individuals who carry them and those who do not. Whether this is or will become possible (or even plausible) requires understanding genes: what they are and are not, as well as what they can and cannot “do.”

The aim of the present book is to counter misperceptions about genes, and help readers to acquire a better understanding of them. Along the way I show that the importance of genes has often been exaggerated. Of course, I am not going to argue that genes are not important – indeed, they are! But it is one thing to say that genes are important for what we are or do, and another that they matter more than anything else. Genes have been presented as autonomous entities that contain all the necessary information to determine characteristics and are capable of making use of it. They have thus been described as the “essence” of life, as the absolute “determinants” of characteristics and disease, and therefore as providing the ultimate explanations for all biological phenomena because the latter can be “reduced” to the gene level and thus be explained.

Therefore, there exist at least three misunderstandings about genes:

*Genetic essentialism*: Genes are fixed entities that are transferred unchanged across generations, and that are the essence of what we are by specifying characteristics from which their existence can be inferred.

*Genetic determinism*: Genes invariably determine characteristics, so that the outcomes are just a little, or not at all, affected by changes in the environment, or by the different environments in which individuals live.

*Genetic reductionism*: Genes provide the ultimate explanation for characteristics, and so the best approach to explain these is by studying phenomena at the level of genes.

These definitions help us distinguish between three important properties usually attributed to genes: (1) that they are fixed essences that specify who



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we are; (2) that they alone determine characteristics notwithstanding the environment; and (3) that they best explain the presence of characteristics. Collectively, these ideas can be described as *genetic fatalism*. In the present book, I explain why genetic fatalism is wrong.

A central feature of the present book is that it is mostly about human characteristics and disease. When this is not the case, it is usually about phenomena of relevance to human life. I must note that this is not due to any anthropocentrism on my part. Quite the contrary, I believe that we are not anything special in this world, or at least that we are not any more special than any other organism that lives in it. Nevertheless, I thought that the book would be more interesting and comprehensible to readers if I discussed phenomena about, or relevant to, human life. This approach is biased, of course, because it overlooks important aspects of life on earth. I hope that readers will find this biased-toward-humans book interesting and didactic. But they should also keep its bias in mind and avoid unwarranted generalizations from the mostly medical-centered and human-focused research presented in this book.

Some basic nomenclature: Gene and protein symbols in humans are written with uppercase letters. However, gene symbols are written with italicized characters, whereas protein symbols are written with regular characters (the protein called “hemoglobin A” is written HBA, whereas the respective gene is written *HBA*). For the purpose of consistency, most gene symbols and gene names in this book are derived from the Human Genome Organization Gene Nomenclature Committee website ([www.genenames.org](http://www.genenames.org)).

A note about historical periods: There are several ways to distinguish between the various periods of research related to genes and genomes. In many cases, and for many reasons, some periods may actually overlap for a significant amount of time. However, simply for convenience, in the present book I consider four distinct historical periods:

1. the period until the coining of the gene concept in 1909, which I describe as the pre-genetics era (even though the term “genetics” was coined in 1906);
2. the period from 1909 until the initiation of the Human Genome Project in 1990, which I describe as genetics because the focus of research was

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- mostly on genes – even though the Human Genome Project itself initially focused on individual genes. This period can be roughly divided into two periods, classical genetics (1900s to 1950s) and molecular genetics (1950s to 1990s);
3. the period from 1990 to 2013, when the findings of the ENCODE project and of genome-wide association studies made clear the vast complexity of genomes, which I describe as genomics; and
  4. the period from 2013 to today, which I describe as postgenomics.

I must note that I do not claim that this is how the history of genetics and genomics should be perceived or organized. I only want to note that there has been a shift of focus from genes to genomes, as well as that it took us a whole century – the century of the gene, as historian Evelyn Fox Keller described it – to realize the complexities of heredity.

The present book is intended for anyone who wants an accessible but rigorous introduction to genes. It provides a concise overview of contemporary genomics research and concepts. This research advances at an extremely fast pace, and I am sure that now that you are reading this book, there exist new articles and books that I could have considered. This was already the case when I was working on this new 2021 edition of the book, less than four years after the publication of the original 2017 edition. Nevertheless, I am confident that the main points of the present book and its conceptual foundations will remain unchanged for many years to come. Let us now begin our quest to understand genes.

## Acknowledgments

There are many people I would like to thank because they made writing this book possible in various ways. But there is no-one else that deserves to be acknowledged more in this case than Katrina Halliday, executive publisher for the life sciences at Cambridge University Press. Neither this book as you see it nor the book series to which it belongs would have existed without the insight and support of Katrina. The first edition of the present book, published in 2017, was very well received and was commended for its quality and readability (see excerpts from and links to the reviews at <http://kampourakis.com/making-sense-of-genes>). Yet, that was still an academic book. Thanks to Katrina, we now have this revised and updated, but also concise, version that I hope you will appreciate.

I am grateful to Bruno J. Strasser and Andreas Müller, who support my work and research at the University of Geneva. My interest in human genetics goes back in time to when, as an MSc student, I had the opportunity to work at the laboratory of Emmanouil Kanavakis at the University of Athens, whom I thank for that opportunity. While writing this book, I have been very fortunate to benefit from the thoughtful feedback of several scholars: Garland Allen, John Avise, Sheldon Krinsky, Alessandro Minelli, David Moore, Staffan Müller-Wille, John Parrington, Giorgos Patrinos, Erik Peterson, Anya Plutynski, Gregory Radick, Andrew Reynolds, Carlos Sonnenschein, Eric Turkheimer, and Tobias Uller. I thank them all for their valuable comments and suggestions. I owe special thanks to Nathaniel Comfort, whose comments were extremely useful in clarifying the main argument of this book. Writing this book has also benefited from discussions during an older collaboration with

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Last but not least, there are always those to whom I owe a lot: my family. I dedicate this book to my brother and our mother because the striking differences and similarities among us have always made me think hard about “nature” and “nurture.”