
Human Population Genomics

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Editors

Human Population Genomics

Introduction to Essential Concepts
and Applications

 Springer

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*To Walter and James
And
To Hannah and Milla*

Preface

Human population genomics is an exciting and rapidly changing field. Over the past decade, there has been a deluge of genetic variation data from the entire genome of individuals from many populations. These data have allowed us an unprecedented look at human history and how natural selection has impacted humans during this journey. Simultaneously, there have been increased efforts to determine how genetic variation affects complex traits in humans. Due to technological and methodological advances, progress has been made at determining the architecture of complex traits.

Because the field is so rapidly evolving, it can become difficult for newcomers to gain footing in the morass of technical concepts of population genetics, complex trait genetics, and data analysis. This is unfortunate as human population genomics brings together researchers from disparate areas of expertise, including computer science, statistics, medicine, genetics, evolution, and anthropology.

This book represents our modest attempt to make the highly technical and contemporary aspects of the field of human population genomics more accessible to researchers in all these groups. We wanted to start with the basics, but then include more advanced and current research. Thus, we hope that this book can serve as a gateway to modern human population genetics research for those new to the field. We hope that seasoned practitioners will also find it as a useful reference on their shelf.

The book has three parts. The first part provides an introduction to essential concepts in population genetics. These chapters will be useful for learning some of the more arcane, but important, concepts in population genetics, which are relevant for any organism. The second part covers the genetics of complex traits in humans. The third part of the book focuses on applying these techniques and concepts to genetic variation data to learn about demographic history and natural selection in humans.

In Chap. 1, Wakeley provides an introduction to one of the most useful tools in population genetics, the coalescent. The coalescent is used ubiquitously throughout population genetics, and an understanding of it will help develop intuition about how evolutionary forces impact genetic variation. In Chap. 2, Slatkin provides a discussion of linkage disequilibrium. While linkage disequilibrium may have originally been an esoteric theoretical topic with little practical value, it has become an essential phenomenon for associating genetic variation with complex traits. In Chap. 3, Skoglund and colleagues discuss tools used to measure and quantify human

population structure. While understanding human population structure is valuable in itself, it is vital to properly design association studies for complex traits. In Chap. 4, Enard provides an introduction to natural selection in humans and contemporary statistical methods used to detect positive selection.

Chapters 5–7 discuss medical genetic and association studies between genetic variants and complex traits. In Chap. 5, Graff and Witte discuss statistical methods used to associate genetic variants with complex phenotypes. This chapter provides an introduction to genome-wide association studies (GWAS) which have become a mainstay in human genetics. In Chap. 6, Thompson describes an exciting approach of using identity by descent (IBD) mapping for implicating genetic variants in disease risk. In Chap. 7, Voight provides a summary of some of the lessons that the human genetics community has learned from the early GWAS.

Lastly, Chaps. 8 and 9 provide an overview of human evolutionary genetics. In Chap. 8, Wall summarizes what we have learned about human history for the analysis of genome-wide genetic variation data. In Chap. 9, Emery and Akey provide an overview of what we have learned about how natural selection has shaped genetic variation across the human genome. They discuss some specific examples of human adaptations and what these selection scans tell us about human evolution.

We wish to thank the authors of these chapters as well as the editors at Springer for their continued patience with us. This project took nearly a decade to complete, and we are grateful for their support and helping us eventually complete the book.

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Berkeley, CA
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