

Preface

Like most academic disciplines, anthropological genetics experienced a long period of development and maturation, characterized by "hybridization" between physical (biological) anthropology, human genetics, and molecular biology. The first acknowledgment, of which I'm aware, of anthropological genetics as a distinct academic field is in an article title ("Assumption and Fact in Anthropological Genetics") by Derek Roberts in 1965. Thus, we have just recently passed the 50-year anniversary of the "emergence" of the discipline. The real foundation of the field, however, is usually pinned to Crawford and Workman's seminal volume, *Method and Theory in Anthropological Genetics*, published 45 years ago this spring.

Initially, the field was primarily focused on assessing gene frequency differences between populations, identifying evolutionary forces that resulted in population differences as measured by genetic distance statistics, and linking patterns of genetic diversity and population structure to underlying demographic processes – what today might be characterized as genetic histories of regional populations. The scope of anthropological genetics has expanded rapidly in recent decades, encompassing studies in the genetics of disease and health, primate genetics and behavior, phylogenetics and molecular evolution, the genetics of quantitative variation, as well as the evolution and genetics of human adaptation to diverse environments. Anthropological genetics has also made significant contributions to archeology and the study of prehistory via the developing field of ancient DNA (aDNA) analysis. The dramatic growth of anthropological genetics is evidenced by the large scientific literature that now characterizes the field, spanning several disciplinary journals as well as numerous books focused on specific issues and/or analytical approaches. Given this growth, and the notable advances in genetic, molecular, and statistical methods that have dramatically influenced the field in recent years, a new synthetic treatment of the scope of anthropological genetics is appropriate and timely.

The following chapters are designed to cover the scope of research now routinely conducted under the rubric of anthropological genetics. **Part 1** of the volume sets the stage for what follows by providing context for the development of the field and current best practices. Chapter 1 provides the background for the development of the discipline, from its earliest efforts at gene frequency descriptions in select populations to the modern genomic approaches that characterize human evolutionary genetics, by one of the founders and promoters of the field, M.H. Crawford. As genetic and genomic analyses have become more powerful, risks to personal and community identities have become of greater concern should there be breaches of confidentiality or misuse of individual genomic profiles. In addition, consultation with identifiable communities that are often the subject of such study has taken on primary importance. Increasingly, investigators partner with participant communities as research collaborators, attempt to carefully identify both community and individual risks and benefits of the proposed research, and solicit input and advice from the community on acceptable questions and approaches. Accordingly, Garrison and Bardill (in Chapter 2) explore the ethical issues attendant to the rapidly expanding use of genomics in ancestry testing, at both the individual and population levels, while Malhi (Chapter 3) draws on his own experience in human population genetics and community consultation to explicate ethical issues in community-based research programs.

Part 2 of the volume explores topics in macroevolution and phylogenetics, topics that were not really emphasized in the early years of the field, but that have become increasingly important and prominent with the development of newer genomic and bioinformatic tools. A fundamental tenet of molecular evolution is that genomes evolve at a constant rate. But not all genomes, or components of genomes, evolve at the same rate. Knowing how to calibrate the various molecular clocks is essential for using molecular data to date evolutionary events, a topic summarized by Cox in Chapter 4. Use of the "molecular clock" is central to the review of primate genomics and phylogenetics by Schmitt and Gokcumen (in Chapter 5), who discuss the variation in mutation rates and evolutionary histories of different primate lineages. In addition, this chapter highlights the enhanced resolution afforded by next-generation sequencing approaches, from new insights into the relationship between functional genomic and phenotypic variation to the role of new genomic methods in field primatology for study of behavioral and ecological variation. As in so many cases, the newer genomic approaches utilized in primate studies permit an examination of the demographic history of both species and local populations.

Use of molecular dating methods is also central to Williams' (Chapter 6) summary of hominin phylogenetic diversity. The role of molecular genomic approaches in addressing questions of human evolution and diversity is a recent adjunct to the traditional methods of paleoanthropology, and such approaches have been made possible by advances in molecular methods via aDNA analyses. Integrating separate streams of knowledge from different fields to focus on common questions is

always a major challenge in science. Von Cramon-Taubadel takes on the task by assessing the correspondence between molecular diversity, geography, ecology, and quantitative phenotypic variation in modern humans, illustrating how the methods are both powerful and broadly applicable to questions of subspecific diversity and history (Chapter 7). Completing this section of the book, Miró-Herrans (Chapter 8) demonstrates the importance of demographic processes in structuring genetic variability in time and space. She illustrates how simulation is used to reconstruct and test alternative demographic scenarios and, thus, human evolutionary history using approximate Bayesian computational methods.

The five chapters in **Part 3** treat topics that are nearer to the core of anthropological genetics at its inception, with one possible exception. Relethford provides a review of traditional population structure and population history approaches, and clarifies their differences (Chapter 9). He brings those traditions into the present with updated methods and examples from the study of mate choice, population and cultural subdivision, and isolation by distance. Importantly, he shows how such local analyses are related to broader questions and analyses of both regional and global human population history. Similarly, Cabana examines the evolutionary roles of migration (gene flow) and genetic drift in structuring the patterns of genetic variation observed in human populations (Chapter 10). She discusses clearly the importance of population size in evaluating such effects in the context of classical population genetic models. This chapter also introduces analyses of aDNA from archeological samples and the concomitant use of simulation to test hypotheses of ancestry in regional population studies. Taking an explicitly geographic perspective, Kivisild (Chapter 11) reviews the patterns of genetic and genomic variation observed in Eurasian populations and explores what they tell us about how areas of the region were colonized. In particular, this chapter emphasizes the role of environmental change in structuring genomic variation over time, in this case the importance of climate change during the Last Glacial Maximum, and the shift to a more settled agrarian lifeway during the Neolithic. In a similar vein, Bisso-Machado and Fagundes (Chapter 12) examine patterns of genetic and genomic diversity in the Western Hemisphere and describe alternative models of human dispersal into the hemisphere that can be tested by both archeology and genetics. Finally, in a contribution highlighting recent innovations in methods, Raff (Chapter 13) explores the utility of aDNA analyses in contributing to bioarchaeological studies. In so doing, the chapter links to many other contributions in the volume by touching on the importance of community consultation and ethical precepts in aDNA research, the role of these methods in assessing identity, contributions to paleopathology, and reconstructing regional population histories.

Part 4 of the volume contains chapters that survey how we examine patterns in human variation, with particular reference to the role of natural selection. Many of the topics covered here relate to the evolution of specific traits or phenotypes that are of long-standing interest in anthropology and human evolutionary

biology, and they are brought up-to-date with modern analytical methods in the chapters included. Weight and Harpending (Chapter 14) provide a historical perspective on the debate between the biometrician and Mendelian schools in accounting for patterned variation, and argue that this continues in some ways in molecular versus organismal biological approaches. Having set this context, these authors continue to explore recent research on how we think about tempo and mode in human evolution. Taking a finer grained look at human adaptability, Bigham (Chapter 15) reviews the current evidence for the role of natural selection in permitting humans to adapt to extreme environments, permitting our species to successfully disperse to all continents except Antarctica. Evidence for natural selection is amassed from populations that occupy environments from high-altitude locales in the Andes, the Himalayas, and Ethiopia to the extreme cold of the Arctic. In each of these environmental contexts, the physiological responses and health effects are examined. In Chapter 16, Veilleux reviews the evidence for selection on sensory polymorphisms and, therefore, its role in human dietary adaptation. In all mammals, including primates, sensory systems mediate the interactions of an organism with the external environment. Among primates, particularly humans, the evolution of color vision and taste sensitivities affects diet and general foraging strategies. Variation in opsin genes (responsible for color vision) and taste receptor genes in humans is reviewed in this chapter, along with an examination of how adaptive dietary shifts may have influenced the evolution of these sensory polymorphisms.

Human skin color has a long, and sometimes notorious, history in anthropology. In Chapter 17, Norton examines the evidence for the operation of natural selection on the geographically structured variation in human skin pigmentation. The correlation between ultraviolet radiation and degree of skin pigmentation strongly implicates natural selection in the evolution of skin color. As noted by Norton in this chapter, most studies to date have been conducted on European populations, with much less attention given to the genetic architecture and evolution of pigmentation in other geographic areas. She encourages future research to address this deficiency in our understanding of the evolution of human skin color. The role of genetics in growth trajectories that produce such common phenotypes as stature and body mass index across the life course is the subject of Chapter 18 by Czerwinski, Demerath, and Towne. Historically, genetic studies in growth and development have been few, but recent developments in analytical techniques in statistical genetics have reversed this trend. Accordingly, the authors of this chapter provide an up-to-date review of the genetic epidemiology of normal human growth and development. Concluding this section, Non and Thayer (Chapter 19) remind us that not all variation derives from genetic differences among individuals. Epigenetic markers in the genome affect gene function and expression and serve as one link between the environment and the genome. Epigenetic markers can enhance human plasticity across environments without alteration in the underlying DNA sequence. Although conceptually epigenetics as

an area of inquiry is decades old, recent methods of analysis and the recognition that both the external environment and the genome itself affect epigenetic variation have stimulated an explosion of research. To address this relatively new area of analysis, this chapter illustrates the rapid pace of discovery and evolution of molecular methods, their effect on the discipline, and our constantly evolving knowledge base.

The final section of the volume, **Part 5**, contains two chapters relating to anthropological genetic research on the evolution of health and disease. Knapp and Torosin (in Chapter 20) review the evolution of immune response in the primate order. Summarizing work on the evolution of both the innate and adaptive immune systems, the chapter clearly describes the role of different cell types in mounting immune responses in each system. The chapter particularly focuses on the major histocompatibility complex (MHC) and its role in immune response. Finally, VanSteelandt and Stone (Chapter 21) treat the related area of the evolution of human pathogens and how such studies inform a newer perspective in evolutionary medicine. These authors explore human-pathogen co-evolution and summarize the ways in which pathogens have evolved strategies for avoiding a host's immune response. Through examples and case studies, this chapter describes research on how evolution has shaped patterns of human health and disease and how such work links anthropological genetics with evolutionary medicine.

The chapters contained herein survey a remarkably diverse array of modern methods and results in contemporary research on nonhuman primate and human evolutionary genetics. Almost all chapters highlight, either explicitly or implicitly, the importance of the continual evolution of genetic analytical methods and the importance of population demography in providing critical context for genetic and genomic studies of human evolution and adaptation. Similarly, many of the chapters relate core topics to historical, or continuing, debates in anthropology and genetics, for example the relative importance of selection versus drift in structuring variation in population, or quantitative versus Mendelian approaches to the study of phenotypic diversity. Collectively, I hope these chapters, by top researchers in the relevant specialties, provide a snapshot of the continuing growth of anthropological genetics and the field's role in explicating the evolution of our species.

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Lawrence, Kansas, May 2018