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Human evolutionary genetics: origins, peoples, and disease (2004)

M.A. Jobling, M.E. Hurles and C. Tyler-Smith: Garland Science, ISBN 0-8153-4885-7, paperback

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The authors of *Human Evolutionary Genetics: Origins, Peoples, and Disease* have achieved no mean feat—a textbook introduction to an interdisciplinary field. The book covers the basics not only of molecular genetics, genomics, and statistical population genetics, but also a range of topics in evolutionary anthropology, from the fossil record and linguistics to the spread of agriculture and the peopling of the Americas. They also illustrate applications of evolutionary genetics to medical and forensic science. This accomplishment is couched in direct expository prose.

The book is divided into six sections, beginning with a straightforward articulation of the central role for genetic data in understanding human history. Several nice features make their appearances right from the start. First, the authors are very generous with figures and graphs, about one per page. In later chapters, these illustrations are supplemented by data tables from the primary literature; these allow readers more directly to evaluate the support for various claims. Second, the authors provide ample up-to-date literature citations in the body of the text, with full reference lists at the end of each chapter; these serve as an indictment of the opacity and inconvenience of most other textbooks. Third, scattered through the book are opinion boxes, viz., short pieces penned by a diverse cast of distinguished experts. These permit the presentation of contentious ideas (of which human evolutionary genetics has no shortage), while not forcing the book's authors to stray from their even-handed treatment.

Section two introduces the genome and then details the molecular nature of human polymorphism and the methods for its discovery. The authors do a good job clarifying the diverse mechanics of mutation, both for single-nucleotide polymorphisms (SNPs) and for tandem repeats. Substantial space is devoted to the pitfalls of studying

ancient human DNA, and readers will be impressed with the challenges facing even the most rigorous efforts to make use of this valuable resource. The explanation of regulatory DNA, which in some accounts constitutes the majority of functional DNA in the genome, is superficial, but the study of regulatory DNA is still in its infancy and the authors of this otherwise up-to-date book can hardly be faulted. The only worrisome detail in section two (and indeed the book) is the authors' incomplete treatment of SNP ascertainment bias. It is true, as the authors note, that SNPs discovered in one population are a biased sample of the SNPs segregating in a different population. However, it is also true that they are a biased sample of the SNPs segregating in the original population. The size of an SNP ascertainment panel strongly influences the allele frequency spectrum in the population at large, and so evolutionary population genetic analyses, which often employ allele frequency spectra to test evolutionary models, must correct for this insidious bias.

Section three gives an excellent high-level introduction to statistical population genetics. Although the authors are stingy with the mathematical details, they provide plenty of references for those wishing to perform the analyses described. An appealing feature is the inclusion of website addresses for many software resources.

At heart, this is a book about human history, and not about methods. Consequently, sections four and five, which show the wealth of knowledge of our species that has been gained from the study of genetic variation, constitute the core of the volume. Beginning with comparative population biology of the great apes and moving through human origins and on to the colonization of the globe and subsequent migration and admixture, the authors do an exceptional job incorporating diverse threads of evidence into a coherent presentation. Contentious issues are explained in patient detail (perhaps too much detail in the case of the migration of Neolithic farmers into Europe). The authors emphasize the unique virtues of studies of mitochondrial, Y-chromosomal, and autosomal data, and these virtues are exemplified

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repeatedly. I particularly enjoyed the discussion of insights into human evolution from genetic studies of domesticated animals and plants.

By virtue of their backgrounds, the authors focus on the inference of demographic history from genetic data. As a result, they generally discuss natural selection as a confounding influence, the reverse of the perspective of evolutionary functional geneticists who consider demographic history an obstacle to the discovery of the action of natural selection. In the last section of the book, the authors adopt this functional genetics viewpoint, and they detail many of the well-documented examples of natural selection operating in human populations past and present. For those of us with an interest in bringing genetics to bear on the evolution of phenotypes, this section was very satisfying, although the box explaining quantitative genetics makes the common mistake of conflating components of variance with modes of gene action. Section six also includes a remarkably reasonable discussion of the future of human evolution. Throughout, the authors avoid the easy allure of just-so story telling.

Medical geneticists will be attracted to section six's valuable discussion of the implications of evolutionary genetics for the study of the genetics of human disease. I was pleased to find an account of the evolutionary models underlying the HapMap project. Future editions of this book would do well to expand this section, better to convince medical geneticists of the importance and relevance of the work by their evolutionary colleagues. As data on comparative genomics accumulate and as SNP and linkage disequilibrium maps grow ever more detailed, evolutionary population genetics will play a more and more central role in both the genetics of complex disease and pharmacogenetics.

I strongly recommend *Human Evolutionary Genetics* as an undergraduate textbook. At the same time, I recommend this book to any readers with an interest in human evolution or human genetics. It would also be of value to evolutionary biologists unfamiliar with the data from what is rapidly becoming the best model organism for evolutionary genetics and to medical geneticists who may not realize that an evolutionary perspective will serve them well.