
Preface

Genetic epidemiology is the study of the role of genes and environments on markers of health and disease risk in populations. It emerged as a mainstream discipline in the early 1980s, arising from firm foundations laid by mathematical population genetics, clinical genetics, and statistical epidemiology. Though genetic epidemiology attempts to identify the many components of risk attributable to genes, environments, and interactions between these two factors, the course of the research towards this goal can follow many diverse paths. In the last few years, the success of genome-wide association studies in their identification of hundreds of disease susceptibility loci has brought this specialist field to the forefront of biomedical research.

Advances in molecular genetics will soon offer affordable means to measure or observe study participant's genetic material at the sequence level as well as more detailed functional data, such as gene expression. It is evident that genetic epidemiology projects increasingly require long-term collaboration between bioinformaticians, geneticists, clinicians, statisticians, and epidemiologists. As with any field that is making rapid advances, technologies, and methodologies are both developed and superseded quickly. However, in spite of the rapid changes in techniques, much of the basic language, models, and principles have remained the same.

Interdisciplinary research requires good communication and understanding across the participating disciplines, and this book aims to provide a basic framework for this communication suited to newcomers to the field as well as experienced researchers and graduate level students. Statistical methods are applied in a wide range of disciplines, and this is one subject area that is well catered for by existing text books, particularly at the introductory level. This book assumes a basic level of competence with regard to statistical and probabilistic reasoning, so readers lacking confidence in this respect are guided towards more introductory texts [1–3].

Section 1 consists of three chapters covering the very basics of modern molecular genetics, the terminology and models frequently employed in genetic epidemiology, and an introduction to epidemiology. This section concisely presents most of the language and key concepts that are required to understand the more specific topics discussed in the subsequent sections.

Principles of genetic linkage analysis are outlined in Sect. 2. Section 3 contains five chapters that cover genetic association studies, including an overview chapter of genomic resources available through the Web.

Sections 4 and 5 contain some more specialist topics and three case studies where many of the concepts introduced in earlier chapters are illustrated with interpreted examples.

Those wanting more detail on how to apply statistical reasoning or how to use the necessary computational methods can move onto the more advanced range of textbooks which each have their own perspective. There are a good number of texts specifically for genetic epidemiology [4–10]. For many years, Jurg Ott's "The Analysis of Human Genetic Linkage" [11] was the key source for researchers in gene mapping, though more recent

books also cover these methods [12–15]. The founder disciplines, population, and quantitative genetics, are well covered by many textbooks, and we refer to just a selection of these [16–20]. Statistical modellers and graduate researchers find the handbook of statistical genetics [21] and the encyclopaedia of biostatistics and genetic epidemiology comprehensive source material [22]. In addition to printed texts, there are extensive ranges of educational material available online. Resources supporting education in genetics are particularly well developed [23–25].

I thank sincerely all those who have helped to bring this book together, particularly the co-authors. The content and emphasis of this book has been strongly influenced by both colleagues on the academic staff and students of the MSc in Genetic Epidemiology that was lead by Professor Chris Cannings at the University of Sheffield.

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