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BOOK REVIEW

M.T. Dorak: Genetic association studies: background, conduct, analysis, interpretation

Garland Science, £52, ISBN 978-0-8153-4463-6

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This book aims to provide an introduction to genetic association studies to those new to the field. It would be particularly suitable for postgraduate students and early postdoctoral researchers seeking to learn the necessary background to this type of study. Although written from the point of view of an epidemiologist, it covers a range of fields.

The first three chapters provide an excellent introduction to the molecular, medical and population genetics needed to understand association studies. Topics covered include the structure of DNA, chromosomes and genes, mutation and its consequences for protein structure and gene expression, genetic disorders and their modes of transmission, and the spread of genetic variation through the population. The complex topic of linkage disequilibrium is explained clearly and in some detail, although more time could have been given to the discussion of population structure.

Chapters 4–6 cover the basics of epidemiology and statistics, including the important issues of confounding, types of study and the relationship between genotypic and environmental effects. The statistics chapter is brief but gives an overview of the statistical concepts involved in performing an association study.

Chapter 7 covers the different genotyping technologies available and their uses, while chapters 8 and 9 cover the difference between candidate gene studies and genome-wide association studies (GWAS), how these studies are designed and conducted, and how their results are analysed. Although candidate gene studies were a prevalent form of association study in the past, they are now much less common than GWAS methods, so possibly less space could have been devoted to their discussion.

Chapter 10 covers the basics of bioinformatics tools that help the interpretation of GWAS results and gives the details of many useful programs and websites. Chapter 11 rounds out the book by looking at the current and future utility and successes of association studies.

The book has many strong points. The text is clearly written, covering the basics of the multiple disciplines that underpin association studies without assuming too much knowledge of the subject, but also without going into excessive detail. Well annotated references and reading lists are provided if more information is needed. The chapters follow a natural organisation, with explanations of the underlying genetics and medical information explained before the technology, statistics and study design of genotyping are introduced.

The book has few weaknesses. Some areas might have been covered in slightly more depth. The discussion of stranding in the first chapter is brief and does not explain the difference between the positive and negative strands fully. As stranding issues are a common cause of problems when working across multiple datasets, it would have been useful to give more information on them. Risk profile scores are relatively common analyses, but are only mentioned in the context of predictive medical applications; more details on these would have been useful. The treatment of genotype imputation is similarly brief, although this may be too technical a topic for an introductory text. Lastly, controlling for population stratification is a particularly important part of GWAS quality control, especially in case/control studies. A more detailed example of the use of principal component analysis for this would have been helpful and interesting.

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However, these omissions are minor, and do not detract from the book's high quality. In general, this book is particularly suitable for anyone new to association analyses, and may also be a useful reference tool for those with more experience.