METHODS IN MOLECULAR BIOLOGY

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Genetic Epidemiology

Methods and Protocols

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Preface

Genetic epidemiology is a relatively new field of epidemiology that studies the role of genetic factors in health and diseases and has witnessed some exciting findings in our quest to understand the nature of genetic inheritance. It is an amalgam of methods and approaches applied in traditional epidemiology, statistics, genetics, and bioinformatics and it brings together several scientific disciplines. In the last few years, scientists have been able to map thousands of genetic variants contributing to complex diseases helping to unravel the genetic causes of diseases on a population scale.

This book is a broad overview written at a level that should be accessible to a wide range of interested scientists including epidemiologists, genetic statisticians, human geneticists, clinicians, and bioinformaticians. I hope that this book will be also helpful for graduate students pursuing research in related fields. Some chapters of the book assume a basic level of competence with regard to statistic and probabilistic reasoning; however it was written and edited having in mind that a noncompetent reader will be able to follow, if not all, most of the text. For many scientists, genetic epidemiology is too convoluted to understand; however I hope to persuade the reader that this view is not correct. My goal was to provide a unifying overview of a fast-moving research while providing a description in some depth of the techniques and data that are helping us to understand our genome and how it is related to mainly complex diseases.

Chapter 1 provides an introduction to basic terms of epidemiology whereas Chapter 2 introduces the reader to the key principles of genetic epidemiology including genetic models of inheritance and associations. The next three chapters describe the process of quality control (Chapter 3), the analysis and the detection of common (Chapter 4) and rare variation (Chapter 5) whereas Chapter 6 outlines state-of-the-art meta-analyses approaches for the synthesis of such data. Chapter 7 outlines methods for detecting both gene-gene and gene-environment interactions as well as approaches for increasing statistical power.

The next seven chapters cover novel, state-of-the-art methods that go beyond the conventional approaches for the detection of common variation including analysis in the HLA region (Chapter 8), novel family-based approaches (Chapter 9), approaches for polygenic traits (Chapter 10), multivariate methods for meta-analysis of genetic associations and meta-analysis of gene expression data (Chapters 11 and 12). Chapter 13 covers the rapidly evolving method of Mendelian Randomization that is used for the estimation of causal effects of an exposure on an outcome, whereas computational methods for the analysis of Copy Number Variation are presented in Chapter 14. We conclude in the last two chapters by assessing the functional role of the identified variants (Chapter 15) and the challenges we are facing to use human genetics to identify and validate novel drug targets (Chapter 16).

I thank sincerely all those who have helped to bring this book together and I am grateful to the coauthors who accepted my invitation and contributed to this book, devoting valuable time and effort.

Ioannina, Greece *Evangelos Evangelou*

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