

Contents

<i>List of contributors</i>	page ix
<i>Forewords by Stephen W. Scherer and Peter M. Visscher</i>	xix
<i>Preface</i>	xxiii
<i>List of abbreviations</i>	xxvi
Part I. Genome-wide association studies	1
1. Introduction to genome-wide association studies and personalized medicine	3
Krishnarao Appasani and Raghu K. Appasani	
2. GWAS: a milestone in the road from genotypes to phenotypes	12
Urko M. Marigorta, Juan Antonio Rodriguez and Arcadi Navarro	
3. Introduction to statistical methods in genome-wide association studies	26
Can Yang, Cong Li, Dongjun Chung, Mengjie Chen, Joel Gelernter and Hongyu Zhao	
4. GWAS replicability across time and space	53
Urko M. Marigorta, Juan Antonio Rodriguez and Arcadi Navarro	
Part II. Genome-wide studies in disease biology	67
5. Genome-wide association studies of body mass index	69
Tuomas O. Kilpeläinen	
6. Identification of myocardial infarction-susceptible genes and their functional analyses	79
Kouichi Ozaki and Toshihiro Tanaka	
7. Admixture mapping for disease gene discovery	89
Randall C. Johnson, Cheryl A. Winkler and Meredith Yeager	
8. Genome-wide association analysis in schizophrenia	106
Sven Stringer, Dorien H. Nieman, René S. Kahn, and Eske M. Derks	
9. Epigenome-wide association studies in neurodevelopmental disorders	123
Takeo Kubota, Kunio Miyake and Takae Hirasawa	

Part III. Single nucleotide polymorphisms, copy number variants, haplotypes and eQTLs	137
10. Finding SNPs that affect microRNA regulation in disease-associated genomic regions	139
Laurent F. Thomas and Pål Sætrom	
11. From linkage to complex associations: the role of <i>GABRA2</i> as a risk factor for alcohol use	151
Sandra M. Villafuerte, Elisa M. Trucco and Margit Burmeister	
12. Copy number variation in monozygous twins	168
Erwin Brosens, K.G. Snoek, D. Veenma, H. Eussen, D. Tibboel, and A. de Klein	
13. Haplotypes of CpG-related SNPs and associations with DNA methylation patterns	193
Yiyi Ma, Caren E. Smith, Yu-Chi Lee, Laurence D. Parnell, Chao-Qiang Lai and José M. Ordovás	
14. eQTL mapping	208
Mengjie Chen, Can Yang, Cong Li and Hongyu Zhao	
Part IV. Next-generation sequencing technology and pharmaco-genomics	229
15. Next-generation sequencing for rare diseases	231
Elena Bosch and Ferran Casals	
16. Next-generation sequencing for complex disorders	243
Ferran Casals and Elena Bosch	
17. Chromosomal breakpoints in breast cancer co-localize with differentially methylated regions	255
Man-Hung Eric Tang, Vinay Varadan, Sitharthan Kamalakaran, Michael Q. Zhang, James Hicks and Nevenka Dimitrova	
18. Signaling network analysis of genomic alterations predicts breast cancer drug targets	269
Naif Zaman and Edwin Wang	
19. Pharmacogenetic studies in pediatric acute myeloid leukemia	281
Neha S. Bhise, Lata Chauhan and Jatinder Kaur Lamba	
20. Pharmaco-genomics of antiretroviral drugs	297
Chonlaphat Sukasem, Apichaya Puangpetch and Sadeep Medhasi	
Part V. Population genetics and personalized medicine	313
21. Population stratification and its implications: lessons from genome-wide studies	315
Sheikh Nizamuddin, Rakesh Tamang and Kumarasamy Thangaraj	
22. How to solve genetic disease on a population scale	341
Barry Merriman	
23. Economics of personalized medicine	366
Katherine Payne and Martin Eden	
<i>Index</i>	383

The colour plates are to be found between pages 192 and 193