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Genetics in Diabetes

Type 2 Diabetes and Related Traits

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Preface

In the 1960s, the American Geneticist J.V. Neel referred to diabetes as the 'geneticists nightmare' owing to the high probability that the phenotype was heterogeneous, not clearly defined with a variable age of onset and a strong environmental influence. In the 1970s, the distinction between autoimmune (type 1 diabetes) and non-autoimmune (type 2 diabetes) was made clarifying a major cause of the disease heterogeneity. Through the 1990s, the discovery of the genes involved in mendelian forms of diabetes demonstrated the enormous power of human genetics to uncover fundamental insights into glucose homeostasis and to inform on treatment and prognosis for patients with particular genetic subtypes of diabetes. The genetic basis of type 2 diabetes, however, remained elusive.

In recent years, the field of human genetics discovery has been revolutionised by publically funded initiatives such as the Human Genome Project, HapMap and 1000 Genomes projects. These, in tandem with technological advances such as array genotyping and next-generation sequencing, have enabled genome-wide studies of genetic variation in previously unimaginable sample numbers. This has in turn led to an explosion in the number of genetic loci robustly implicated in type 2 diabetes risk.

In writing this book, we have called upon a number of our colleagues who, over the years, have been part of highly collaborative international efforts to advance our understanding of the genetic basis of type 2 diabetes and related traits. We are indebted to them for agreeing to help us with capturing this journey. They have described the huge progress that has been made, whilst at the same time outlining the substantial challenges that lie ahead if we are to fully capitalise on the these discoveries and translate our improved understanding of the genetic basis of type 2 diabetes into advances in clinical care.

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