Preface

Modern medical studies have shown that human disease results from genetic and/or environmental causes. With our increasing ability to control infectious and nutritional diseases worldwide, genetic diseases increasingly impact upon human health and life, both physically and psychologically. Genetic diseases (or genetic disorders) are frequently old diseases with newly understood causes, these being one or more abnormalities in an individual's genome, and they may present from early embryonic development to postnatal life. Cancers fall into the genetic disease category, because of acquired somatic mutations produced by genomic instability, and also inherited gene mutations, as occurs in about 5-10% of cases. Studies have also demonstrated that genetic defects are the major known cause of pregnancy loss in developed nations, and almost half of all spontaneous abortions (miscarriages) involve a chromosomally abnormal fetus. It will be rare for any family to be entirely free of genetic disease, and genetic diseases therefore impose a huge burden upon individuals, families and societies.

Thanks to the Human Genome Project, researchers have begun to understand the blueprint of the human genome by learning more about the structures and functions of genes and proteins. Data collected on the human genome sequence, combined with newly-developed techniques for screening DNA, have resulted in an unprecedented boom in medical research, and an abundance of discoveries linking genetic variants to an assortment of diseases including various cancers. Studies have been advancing rapidly, revealing the pathological basis of genetic diseases, which allows the development of accurate and specific tests for disease diagnosis, and the eventual translation of research knowledge to clinical therapies. These approaches have had a major impact on the fields of medicine, biotechnology and the life sciences, and will continue to do so for many years to come. With the application of new technologies for genetic disease studies, the concept and practice of medicine is profoundly changing, as more and more diseases are classified as genetic diseases. In the past, studies aimed to determine if a disease was caused by genetic factors, but today it is rare to eliminate all genetic factors from any given disease.

Genome-wide association studies represent a promising way to study complex, common diseases in which many genetic variations contribute to a person's risk. This approach has already identified single nucleotide polymorphisms (SNPs) related to complex conditions including diabetes, heart abnormalities, Parkinson's disease, and Crohn's disease. Next generation sequencing technology has also been remarkably successful in the identification of causes of genetic diseases using whole-genome, whole-exome, and transcriptome sequencing. However, from a diagnostic perspective, we are still looking for an ideal technology for genetic disease testing with the features of specificity, sensitivity, accuracy, reliability, high throughput capacity, reproducibility, low cost, and ease of operation. Furthermore, despite our vastly improved knowledge of human genetic variations, studying associations between genetic disease genotype and phenotype still remains a major challenge.

This special edition brings together many of the challenges of genetic disease studies, with six review articles of non-cancer genetics topics, and five review articles in cancer genetics. We hope that these reviews will encourage the further translation of research to clinical practice, including new or improved genetic disease therapies.

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