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Medical Genetics in the Clinical Practice of ORL

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Preface

The sequencing of the human genome, completed in 2003, laid the foundation for great advances in scientific knowledge and molecular and informational technologies. Because of the Human Genome Project, which took 20 centers around the world over 5 years to complete at a cost of USD 2.7 billion, an individual’s entire genome (all their genetic information) can today be sequenced for less than USD 10,000. The cost of whole genome sequencing and our understanding of the genome will continue to change exponentially, and individuals may soon have their whole genome sequenced as part of routine medical care.

There is almost no part of the clinical practice of otorhinolaryngology that is not touched by genetics. It has long been recognized that an immense number of genetic syndromes include hearing loss, craniofacial abnormalities, cochlear malformations, cleft lip/palate, and tumors of the head and neck. In recent years, the genetic causes of many of these syndromes and a number of other conditions including nonsyndromic hearing loss and chronic rhinosinusitis have been elucidated, resulting in an improved understanding of the developmental and biochemical processes involved, allowing the development of genetic tests to aid in diagnosis and risk assessment, and suggesting novel approaches for therapeutic intervention.

This book is written as a practical guide to medical genetics as it applies to the clinical practice of otorhinolaryngology. It describes recent advances in understanding the genetics of diseases of the head and neck, introduces emerging knowledge and trends, and provides resources that empower clinicians to incorporate genetics into clinical practice, thereby improving patient care.

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