Genetics in Otorhinolaryngology
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47 figures, 2 in color, and 15 tables, 2000
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Molecular genetics has been developing rapidly, and the molecular basis of many diseases has been discovered. Identification of the relevant genes has led to more precise diagnosis and has provided clinicians and researchers with important clues as to the identity of previously unsuspected molecules whose function is critical for the development and maintenance of the disease. This includes diseases in the field of otorhinolaryngology. A knowledge of genetics has already become important for the otolarinolaryngologist when examining patients at their outpatient clinic. Research is in progress to identify the genes for deafness and craniofacial anomalies as well as the development of novel therapeutic modalities, including gene therapy for head and neck cancer.

Otorhinolaryngologists have been active participants and have made significant contributions to advancing clinical as well as basic scientific knowledge in the molecular characterization of the above diseases. In many cases, the suspicions of an otorhinolaryngologist have led to the identification of underlying genetic abnormalities and clues toward making a diagnosis have been identified. Many patients have highlighted the need for otolarinolaryngologists to be constantly alert to the possibility of diagnosing hereditary disorders and to obtain genetic consultations for a complete evaluation. However, genetics, and particularly molecular genetics, remains an unfamiliar subject for most otolarinolaryngologists. Therefore, we present a volume on this subject in this Karger series.

We have organized this volume around the molecular analysis of diseases as well as emphasizing the clinical significance of this molecular analysis in the field of otorhinolaryngology. The purpose of the volume is to explain to general otolarinolaryngologists how genetics can be useful to them and to their patients, and to summarize the information about genetic diseases that is relevant to their clinical activity.
In this volume, we do not include a detailed description of the basic methodology of genetic studies. You will find information about the positions of the various loci on chromosomes, and, in cases where the gene has been identified, details of the gene and the effects of the mutations. The clinical aspects of diseases are described with attention to particularly distinguishing features that may be diagnostic and to any variability observed in the phenotype. We focus on how this information will be useful to the clinician and to the patient, particularly with reference to enabling accurate genetic counseling, with a list of references if more details are needed.

A basic and general review of genetics, which is requisite for readers to be able to understand the content of this volume, is presented in the first part of the volume. More pages are allocated to hearing impairment than to other aspects of otolaryngology because great progress has been made over the last few years in identifying the genes associated with hearing impairment. One chapter focuses on for the mouse model for hearing impairment because the mouse is the most useful animal system for advancing research in hereditary hearing impairment.

The application of molecular analysis to head and neck carcinoma has been one of the fundamental breakthroughs in understanding the cell biology of the carcinoma. In relation to these breakthroughs, two chapters are devoted to a discussion of tumor suppressor genes and oncogenes.

Understanding of the molecular basis of the disease will lead to develop new modality in treating patients with that disease. Although it seems unlikely that gene therapy will be clinically useful for a long time, gene therapy, ultimately, is expected to become the treatment of choice for curing patients with diseases related to genetic abnormalities. One chapter is therefore devoted to gene therapy.

The field of molecular genetics has rapidly evolved from basic scientific research to having diagnostic and therapeutic clinical applications. Today’s laboratory discoveries will undoubtedly lead to major advances in the way clinicians diagnose and treat diseases. We do hope that this volume can review the molecular and clinical features of different types of genetic disorders facing the otorhinolaryngologist and can create a productive interplay between geneticists and otorhinolaryngologists.

Finally, I thank Prof. Karen P. Steel for her great editorial work as an editor and all contributors for their commitment to this volume.

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Preface