Springer Handbook of
Auditory Research

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Genetics of Auditory Disorders

With 40 Illustrations

Springer

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Cover illustration: G-banded human male karyotype (46,XY), consisting of the normal complement of 46 chromosomes, including one X and one Y. This figure appears on p. 94 of the text.

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Series Preface

The *Springer Handbook of Auditory Research* presents a series of comprehensive and synthetic reviews of the fundamental topics in modern auditory research. The volumes are aimed at all individuals with interests in hearing research including advanced graduate students, post-doctoral researchers, and clinical investigators. The volumes are intended to introduce new investigators to important aspects of hearing science and to help established investigators to better understand the fundamental theories and data in fields of hearing that they may not normally follow closely.

Each volume is intended to present a particular topic comprehensively, and each chapter will serve as a synthetic overview and guide to the literature. As such, the chapters present neither exhaustive data reviews nor original research that has not yet appeared in peer-reviewed journals. The volumes focus on topics that have developed a solid data and conceptual foundation rather than on those for which a literature is only beginning to develop. New research areas will be covered on a timely basis in the series as they begin to mature.

Each volume in the series consists of five to eight substantial chapters on a particular topic. In some cases, the topics will be ones of traditional interest for which there is a substantial body of data and theory, such as auditory neuroanatomy (Vol. 1) and neurophysiology (Vol. 2). Other volumes in the series will deal with topics that have begun to mature more recently, such as development, plasticity, and computational models of neural processing. In many cases, the series editors will be joined by a co-editor having special expertise in the topic of the volume.

Richard R. Fay, Chicago, IL
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Preface

The purpose of this volume in the *Springer Handbook of Auditory Research* series is to inform the reader about the many clinical forms of genetic hearing loss, the mutations that are responsible, and the functions of the proteins that are encoded by the mutant genes. This volume treats with equal importance the basic principles of genetics, the methods and techniques used in human molecular genetic studies, and the examples of how scientific results can be applied in patient care. An overview of the basic principles of genetics is provided in Chapter 1 (Keats and Berlin), together with citations to other chapters that expand on the topic. In addition, Chapter 1 ends with an extensive glossary of genetic terms with which some readers may be unfamiliar. Chapters 2 (Avraham and Hasson) and 5 (Giersch and Morton) describe the structure of genes and chromosomes, while Chapters 3 (Mueller, Van Camp, and Lench) and 5 explain the methodologies and the genomic tools used to find the chromosomal locations and identify disease genes. In Chapter 4, Nance and Pandya discuss the application of the laws of population genetics and models of genetic epidemiology to hearing loss, while Chapter 8 (Steel, Erven, and Kiernan) clarifies the relevance of mouse models to advancing understanding of hearing loss in humans. The clinical and genetic heterogeneity of genetic hearing loss are delineated in Chapters 6 (Griffith and Friedman) and 7 (Fischel-Ghodsian), with Chapter 6 providing an in-depth coverage of autosomal and X-linked forms of hearing loss and Chapter 7 concentrating on hearing loss due to mitochondrial DNA mutations. Finally, Chapter 9 (Arnos and Oerlich) illustrates the role of the genetic counselor in communicating knowledge about hearing loss to the patient, thus translating the findings of medical science into clinical care.

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