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BOOK REVIEW

Hereditary hearing loss: how to keep track at the extreme end of genetic and phenotypic variability

Hereditary Hearing Loss and Its Syndromes Third Edition
Edited by: Helga V Toriello, Shelley D Smith
ISBN: 9780199731961
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Of all monogenic conditions, hearing loss is of unparalleled genetic heterogeneity – maybe with the only exception of retinal degeneration. The hereditary hearing loss homepage (http://hereditaryhearingloss.org) currently lists about 100 genes for non-syndromic deafness (the term comprehensively used for all degrees of hearing impairment in the scientific literature), and, in addition, deafness is part of more than 400 genetic syndromes. The pace at which novel causative genes are being identified has accelerated significantly since genomewide mapping, and, recently, next-generation sequencing technologies have become part of geneticists' analytical toolbox. With the current speed of discovery, relying on the latest review article in a human genetics journal is probably the best way for clinical geneticists to stay up-to-date in genetics of deafness. Heavy textbooks can only be updated every few years, and, consequently, they may appear old fashioned. But does this apply to the novel edition of Hereditary Hearing Loss and its Syndromes (3rd edition)?

No, I don't think so. The book is a very valuable resource for clinical geneticists for several reasons:

- *The overall concept of the book*: The chapters cover all aspects, from history, epidemiology, embryology, genetic counseling and clinical investigation to the comprehensive description of genetics and clinical presentations of isolated and syndromic hearing loss. Chapters on syndromes are arranged according to the other affected organs, making it easy to navigate to differential diagnoses that may fit to a patient.
- Although targeted single-gene testing will rapidly become the exception because of next-generation sequencing, aligning genotyping results with phenotypes will remain necessary in many cases to make the correct diagnosis. Consequently, phenotype characteristics are also pointed out for the many (at first glance clinically indistinguishable) genetic subtypes of non-syndromic hearing loss, for example, regarding age of onset, progression, affected frequencies and severity.
- Rare syndromes represent a major diagnostic challenge. Many of them comprise hearing loss, and the book assists effectively here. Syndromes are being described in great detail, and images support these descriptions where needed, making syndromic forms of deafness accessible to the reader.

The few small drawbacks concern two aspects that otherwise represent strengths of the book – the images and the comprehensive listing of genetic loci with their phenotypic characteristics. The resolution of some images is a bit poor, and this becomes critical where images are too small to recognize details on X-ray pictures, for example. Non-syndromic genes and loci with their clinical features are being listed not only in tables, but also in the running text. In a future edition, I would recommend to reduce these redundancies and display the information in tables only. This would save several pages and be more reader-friendly.

However, despite these minor points of criticism, this book is definitely a worthwhile investment not only for clinical geneticists but also for students and researchers, offering an excellent, comprehensive and detailed overview on this fascinating section of medical genetics. It is a great reference book for quickly obtaining well-presented information on any form of deafness, and at the same time its richness of detail makes it an inspiring reading for experts in the field.

CONFLICT OF INTEREST

The author declares no conflict of interest.

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