

Book review

Hereditary hearing loss and its syndromes, 3rd edn

Edited by HV Toriello and SD Smith. Oxford: Oxford University Press, 2013. 756pp.

The third edition of this classic text, edited by Helga Toriello and Shelley Smith, is dedicated to Dr Robert Gorlin, the eminent clinical geneticist with whom this book has always been intrinsically associated, and his colleague Michael Cohen Jr.

Known in short as *Gorlin's hearing loss*, it is *the* authoritative text on genetic hearing loss, concentrating mainly but not exclusively on the syndromes in which hearing loss is a major part. It will be useful to clinical geneticists, ENT surgeons, audiovestibular physicians and paediatricians, as well as to specialist physicians as there are chapters dedicated to hearing loss associated with most body systems (musculoskeletal, nervous, integumentary, endocrine, cardiovascular, renal, etc). Each syndrome or association is described in the same way: a brief introduction followed by physical findings in relevant systems, the auditory and vestibular findings, laboratory findings, molecular studies, diagnosis and differential diagnosis, and a summary, followed by relevant references which may number from a handful for rare syndromes, to over 100 for the more common ones.

Given the enormous increase in knowledge in the field of genetic hearing loss since the last edition eight years ago, it is not surprising that the book is significantly larger than before. Much of this arises from the expanded sections for each disorder on 'Molecular findings/studies', rather than the description of lots of new syndromes. There are, however, two new chapters on 'Gene/environment interactions in acquired hearing loss' and 'Genetic diagnosis and gene discovery for hearing loss using massively parallel sequencing' as molecular genetics has become increasingly important in these areas.

In addition, there are expanded chapters on existing topics in which there have been significant advances, eg the chapter 'Genetic hearing loss with no associated abnormalities' (non-syndromic hearing loss) by Shelley Smith contains useful tables of all genes for non-syndromic dominant and recessive forms of hearing loss. These include brief details of age of onset, type of hearing loss and whether the vestibular system is involved. The tables are followed by a section on what is known about each gene/locus clinically (the audiological and vestibular findings), together with radiology/histology, molecular studies and relevant references, each gene/locus being presented in the same format. There are also small summary tables of genes that have been associated with particular clinical presentations or associations, such as genes causing progressive hearing loss, those presenting with

low-frequency hearing loss, dominant and common recessive mutations in *GJB2* (encoding the connexin 26 protein), and genes causing auditory neuropathy.

Another significantly longer chapter is that on 'Embryology of the mammalian ear'. A description of the morphological development of the ear is followed by a highly detailed account of the molecular basis of early morphogenesis, based on mouse studies, including a table spanning six pages, detailing the role of important genes, their expression pattern and timing at each stage of development, and defects in knockout mice. The level of detail is probably far beyond that needed by even a specialist clinician but, as a reference text, it is thorough.

All of the specialist chapters (hearing loss associated with particular body systems) have been updated with new references and clinical descriptions, and with a few new syndromes added to each. The chapter on eye disorders now includes the condition of 'X-linked retinitis pigmentosa with recurrent infections and hearing loss' and 'PHARC syndrome' (polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, cataract), and the brittle cornea syndrome is now defined separately from Ehlers–Danlos syndrome type 6, as a result of molecular genetic analysis demonstrating that different genes are responsible for each, even though there are phenotypic similarities. Furthermore, advances in molecular genetics mean that some conditions may now be subclassified at the molecular level; so each form of Alport's syndrome, X linked, recessive and dominant, is discussed individually, revealing some of the notable clinical differences.

Although a comprehensive reference text, there were one or two little things that might have made it easier to use. In the previous edition, the Contents included not only the individual chapter titles, but also a list of the syndromes described, together with the page reference for each. This is missing in the third edition, with the Contents detailing only each chapter title and the authors, together with the starting page number. Furthermore, within the chapters themselves, the publishers have preferred to use capital letters for each new section title, rather than bold lettering, which makes syndromes a little more difficult to find. It would also have been good to have included more new illustrations as many of the pictures are identical to those in the second edition. However, the book remains a comprehensive review of 'what it says on the tin'; it will be an invaluable reference text for all those whose work brings them into contact with patients with rare and complex forms of hearing loss. ■

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