Hereditary Hearing Loss and Its Syndromes (Oxford Monographs on Medical Genetics)

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Genetic deafness affects 1 in 1000 children, and over the last decade several dozen of the responsible genes have been identified. This unique textbook aims to assist clinicians dealing with deaf patients and families by critically reviewing all relevant published material on genetics, pathology, clinical presentation, diagnosis, and laboratory findings. Thoroughly revised, the Second Edition has been updated throughout and includes a new chapter on hearing loss with cardiovascular disorders. It continues with the successful formula of presenting separate chapters on deafness associated with findings in specific body systems. Careful attention to cross referencing between chapters means that the multifaceted clinical presentations of distinct conditions are highlighted. These clinical variations are complemented by excellent clinical photographs, audiograms, figures from essential laboratory or other investigations, and comprehensive reference lists. Gene mutations that cause deafness are highlighted throughout the text, both in chapters dealing with syndromes and in a vastly expanded chapter focusing specifically on nonsyndromic forms of deafness. Thus, the new edition reflects all the progress on the molecular understanding of deafness made in recent years and integrates these findings into clinical practice. It also makes an important contribution to the cataloguing of new syndromes that have emerged in recent years, such as HIDS and X-linked maxillofacial dysostosis.

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