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Genetics of deafness, B. Vona, T. Haaf, editors (Karger, Basel, Switzerland) 2016. 146 pages. Price: US$ 186.00 / CHF 158.00 / EUR 148.00

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Hearing Impairment is the most common sensory deficit in humans affecting more than 360 million people worldwide. Approximately 2-3 of every 1000 newborns are born hearing challenged and the prevalence of hearing loss burden is highest in South Asia, Asia Pacific and subsaharan Africa with great social and economic impact. The consequences include delayed or poor speech and language development, educational and occupational disadvantage, social isolation and stigmatisation. It is evident that deafness is genetically determined or at least genetically predisposed. Many of the deafness genes have been cloned and their protein products identified. Discovery of new candidate genes and mutations will have a profound impact on the therapeutic strategies of hearing impairment.

This book is a good compilation of some of the best research works in the field of deafness. The summary at the end of each chapter carries all the key points and the relevant bibliography allows the reader to probe further into the background information provided in each chapter.

The first chapter, "Genetics and Deafness: A View from the Inside" highlights the bioethical issues that arise with the advancement of genetic technologies. Not all hard of hearing individuals want to correct their hearing, *i.e.*, some accept sign language and consider hearing loss as a difference rather than a disability. This deaf culture is a global culture created only out of necessity, hearing challenged adults have the right to either adhere to this deaf culture or join the hearing community with aided hearing. The proxy decision making by parents of children born deaf may also be a contributing factor in this. Because of neural plasticity, earlier hearing restoration in congenitally deaf children with cochlear implants always offers better outcomes. It is unethical to delay implantation in a prelingually deaf child since late implantation is doomed to failure from the start. Restoration of hearing in deaf children will certainly expand their educational and employment opportunities in a future that requires Auditory Verbal skills especially in underdeveloped and developing countries where there are not much deaf community privileges. So, cochlear implants or any other surgical or genetic techniques that offer a bright and open future for a deaf child are arguably not only morally permissible but should be morally encouraged world over.

Presbyacusis is a highly prevalent public health issue that is poorly addressed in many parts of the world. Hearing and communication as rightly pointed out in the second chapter are integral to successful ageing and hearing healthcare interventions should not only focus on the hearing aid devices but address the key issues like lack of self-motivation, lack of family support and social stigma. Genetic research on age-related hearing loss can pave the way for prevention and targeted treatment in future.

Audiological assessment of hearing loss has been discussed in the next chapter. As mentioned, it should involve a battery of tests since no single test is conclusive and each test is complementary to another. Newborn hearing screening does not identify auditory neuropathy spectrum disorder or children with risk of late-onset hearing loss. Molecular genetic testing is an indispensable adjunct to audiological assessment which will provide the opportunity to identify infants at risk of late onset or progressive hearing loss. Electrically evoked auditory brain stem response and Real Ear Measurement (REM) for hearing aid trial should have also been included in the summary of audiological procedures. Apart from cochlear implants, bone bridge implants and auditory brain stem implants are very

promising advancements and deserve a special mention among the available auditory prostheses.

The introduction of genomic analysis into next generation new born screening will definitely enable more accurate diagnosis of hearing loss, more precise assessment of prognosis after implantation and early identification of syndromes even before clinical features become evident, as discussed in chapter 4. Additionally, appropriate genetic counselling in terms of recurrence risk will also have a significant public health impact globally.

A multidisciplinary team approach (including otolaryngologist, audiologist, audioverbal therapist, paediatrician, ophthalmologist, cardiologist, neurologist and geneticists) is indicated in the management of hearing loss especially in children to overcome the diagnostic challenges as discussed in chapter 5. The advent of next-generation sequencing (NGS) techniques has revolutionized the identification of novel mutations and candidate genes but the cost of genetic tests seems to be the major limiting factor in common clinical practice. Hopefully in future, with reduction in the costs, genetic testing will become increasingly available as a routine aid in the assessment of a hearing challenged child. Also deafness screening panels need constant upgradation. However, the NGS techniques are limited by the role of epigenetics and whole-exome sequencing is necessary to thoroughly evaluate intergenic and intragenic regions.

The understanding of the role of genetic modifiers especially the suppressors will help us to understand the complex auditory process and aid in the development of targeted therapy for hearing loss as discussed in chapter 7. There are several animal models employed in auditory research but mouse models are the most common ones as detailed in chapter 9, since all auditory structures present in the humans exist in the mouse. Hence, correction of hearing loss by gene delivery using viral vectors in mouse models is the most appropriate and crucial step in the search for human deafness cure. As mentioned in chapter 10, in recent years Zebrafish is being used as a model to study human deafness and regeneration, because its genome has been fully sequenced and hence targeted gene inactivation is possible. It combines rapid and accessible embryogenesis with a host of genetic and genomic tools for systematic gene discovery and analysis. Vestibular and auditory defects have been detected in adult animals, making the Zebrafish a useful system to tackle the genetic causes of late onset deafness and vestibular disease.

Application of gene replacement therapy in humans can alleviate the disease at the molecular level as discussed in chapter 11. Successful outcomes of cochlear gene therapy in animal models holds the promise of hearing restoration in humans reversing the adverse gene effects. With further refinement and improved methods of gene delivery with a better delineation of viral vectors and with additional improvements in hair cell regeneration, successful gene therapy in humans is well within our reach.

Overall, this is a comprehensive and exhaustive compendium of current knowledge on the subject of "Genetics of Deafness". It is well referenced with tabulations, illustrations and detailed descriptions of experimental evidence. The book will become an important reference tool for anyone with a research intent in the field of deafness. It is an anthology of current perspectives and opens up new avenues for future research. If one has to point out to deficiencies in the book it could only be to recommend enlarging the scope of the book by adding chapters in future editions on allied topics like, genetics of noise-induced hearing loss, autoimmune inner ear diseases and otosclerosis.

Overall, this book will provide a stimulating and fascinating intellectual journey for the reader elevating the level of the reader's knowledge profusely and is recommended for every academic department of Otolaryngology.

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