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Principles of genetics in hearing loss

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Hereditary hearing loss ranks among the most common disorders in man and means a major burden for the affected individual, his family, and the society in general. Whereas the congenital and childhood forms of inherited hearing impairment are most often transmitted in a Mendelian pattern, i. e. are caused by a genetic alteration of a single gene in the respective family, the late-onset forms of hereditary hearing loss are multifactorial diseases caused by a combination of environmental risk factors together with genetic susceptibility variants in several genes. The correct diagnosis of a distinct form of hereditary hearing loss is complicated by an extraordinary genetic (locus) heterogeneity and a pronounced clinical variability. Nevertheless, the molecular elucidation of a large number of monogenic forms of hearing loss (both syndromic and non-syndromic) enabled us to improve the diagnosis and genetic counselling of affected families. Furthermore, these results led to a very much improved understanding of the molecular physiology of hearing and the pathophysiology of deafness. In contrast, the molecular nature of susceptibility genes in human presbyacusis and other complex inherited forms of hearing impairment is still elusive. However, significant improvements in genotyping technologies and our knowledge about the human genome and its variability now enables us to comprehensively study the genetic factors underlying these common disorders and will certainly be successful in the nearer future.