Abstract

Molecular Genetics Aspects of Hereditary Deafness

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Background: The genetic basis of hereditary deafness (HD) has undergone a dramatic transformation in the past 15 years. In the postgenomic era extensive research leading to the discovery of many genes essential for hearing was performed.

Aim: The aim of study the discovery of different type of gene mutations of hereditary deafness.

Material and Methods: Linkage analysis, gene sequencing and whole exome sequencing with array analysis using OtoChip will enhance.

Results: Hereditary deafness is present in two forms - non-syndromic (70%) and syndromic (30%). Today more than 100 gene loci have been linked to nonsyndromic HD – dominant (DFNA), recessive (DFNB), X-linked forms (DFN) and some in the mtDNA. Over 60 genes are associated with syndromic HD like the most frequent syndromes of Waardenburg, Alport, Usher, and Pendred. Taking in to account the exceptional genetic heterogeneity of HD it is not surprising that the genes found so far encode a large variety of proteins with different functions in the inner ear: connected to the structure and function of cochlear hair cells (7 types of myosin, otoferlin, cadherin, actin, stereocilin, harmonin, K and other ion channels, etc; proteins expressed in non-sensorial cells (connexins - 26, 30 and 31, pendrin, otoancorin, claudin14, etc.); proteins of tectorial membrane (collagen XI, alfa tectorine).

Conclusion: The discovery of different type of gene mutations by linkage analysis, gene sequencing and whole exome sequencing with array analysis using OtoChip will enhance undoubtably the diagnostic capabilities, genetic counseling, screening and therapy of patients with HD in the future.

Key words: Hereditary Deafness, Nonsyndromic HD, Syndromic HD, Otochip Will Enhance, Genetic Counseling