GENETICS OF DEAFNESS: FROM FUNDAMENTAL RESEARCH TO CLINICAL APPLICATION

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Genes play an important role in the aetiology of deafness in children. Mutation in a single gene is responsible for deafness in a majority of children and many of these genes have been discovered over the last two decades since the development of positional cloning in genetics.

Many of these genes are responsible for syndromic deafness, where the hearing impairment occurs with additional clinical abnormalities. Nonsyndromic deafness however is more frequent than syndromic forms and makes up for 70% of deaf children. More than 40 genes for nonsyndromic deafness have been discovered over the last 13 years. All these results have lead to a better understanding of physiology of hearing.

An application of these results is genetic testing. DNA diagnostics can provide information about whether the hearing loss is caused by syndromic or non-syndromic deafness. It can also help in determining future evolution of the hearing loss as well and if the hearing loss is progressive. This knowledge can influence the therapeutic options including the choice of hearing aids or cochlear implants. It can also help relieve parents when the cause of hearing loss is defined.

This progress in genetics has however some limitations, since genetic testing for deafness in children currently gives a positive result in less than 25% of cases. Currently, only few genes can be tested with the technology available. However, new sequencing technologies are emerging and they hold great promise for use in DNA diagnostics in the near future.