Abstract EFAS/DGA 2007

Genotypes and phenotypes of various nonsyndromic hearing losses of genetic origin

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Objectives:
Nonsyndromic hearing loss of genetic origin is one of the most abundant human sensory disorders, and can be found in 1 out of 1000 newborns. The phenotype varies from moderate hearing loss to almost complete deafness. Most of the phenotypical alterations can not be attributed to one single mutation. There is a high probability that most of these disorders are polygenic.

Methods:
We collect blood from patients with probable hereditary hearing loss, or deafness. We carry out allele-specific PCR (AS-PCR) reactions on each sample. We have analysed our samples looking for 35delG mutation in the GJB2 (Cx26) gene. Since then we are in the process of screening our population of patients with Denaturing High Performance Liquid Chromatography (DHPLC), which enables us to screen for 43 genetic regions in total.

Results:
We found numerous patients with 35delG mutations, both heterozygous and homozygous forms. Most of these mutations have different phenoypes. Other genetic profiles we get from the mutation screening are being compared to the audiological findings of the examined patient.

Conclusions:
Our plan is to develop a method that can detect the risk of hearing loss of genetic origin with high probability, at best after birth, from only a little blood sample. This goal is very hard to reach, considering the high number of genes involved in the physiology of hearing. Still, our experiments can get us closer to the desired objective.