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The Genetics of Hearing Loss in Childhood

Cremers, C.W.R.J.

Radboud University Nijmegen Medical Centre, Department of Otorhinolaryngology, 6500 HB Nijmegen

In the last decades the so called acquired cases of profound childhood deafness like bacterial meningitis, rubella embryopathy and bloodgroup antagonism have become less frequent, so the relative incidence of genetic causes has increased to over 50%.

In early childhood the genetic causes have been split up in syndromic and non-syndromic. About 450 genetic syndromes with hearing impairment as a feature have been reported. About 10 of these 450 are expected to be diagnosed more frequently. The non-syndromic ones are nominated as DFNA, DFNB, DFN in case gene-linkage has been reached. DFN means deafness and the addition A means autosomal dominant and B autosomal recessive inheritance. Without any addition DFN refers to X-linked hearing impairment. Hereditary non-syndromic profound childhood deafness has usually an autosomal recessive inheritance. DFNB1 (Cx26) is by far the most frequent with a frequency between 10 to 50% in the population depending of the geographic area. Mutationanalysis is in most centres available. Mutations of DFNB2 (MyO7A) may be traced with the micro-array for USH IB (MyO7A). Mutations for DFNB4 like Pendred Syndrome and EVA-Syndrome may be asked for in case of widened vestibular aquaducts on CT scanning of the petrous bones.

Autosomal dominant inherited non-syndromic hearing impairment in childhood (DFNA) is usually moderate and may be progressive. Low frequency hearing impairment or midfrequency hearing impairment may guide to a specific DFNA-type and so the opportunity for mutation analysis. The heterogenety of hereditary hearing impairment in childhood is so large (DFNB 1-67, DFNA 1-54), that in case the clinical presentation is still non-symdromic, specific screening tests for mutation analysis will be needed. Therefore a micro-array for Usher sydnrome (deaf blindness), in early childhood still presenting as non-syndromic, has been made available. In due time this may become available for DFNB-types excluded DFNB1.

Mutation analysis in children for a presumed DFNB-etiology starts with a search for DFNB1 (Cx26) and in addition in case CT-scanning has shown widened vestbular aquaducts with a search for mutations of DFNB 4 and Pendred-EVA-syndrome. In case the family history suggests a DFNA-type specific audometric configuration may guide to a specific mutation analysis to be asked for.