Non-syndromic Hearing Loss & Deafness (COL11A2)

Non-syndromic hearing loss can be caused by mutations in a number of genes, including the COL11A2 gene (referred to as DFNA13 hearing loss). This gene encodes for part of Type XI collagen. Type XI collagen plays a vital role in the inner ear, thus mutations in COL11A2 can lead to poor hearing, as the collagen fibrils in the ear lack their normal structure. Patients with this non-progressive deficiency find it particularly difficult to hear mid-level frequencies, while retaining the ability to detect low and high frequencies.

Initial studies focused on two families, one in the USA and one in the Netherlands. It is not yet possible to determine the prevalence of hearing loss due to COL11A2, although it seems to be rare. It is not clear whether any ethnic group is particularly affected. The condition is autosomal dominant which normally is inherited from at least one affected parent.

Sources

McGuirt, W.T. *et al.* (1999), “Mutations in COL11A2 cause non-syndromic hearing loss (DFNA13),” Nature Genetics, 23, 413-419.
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