Common inherited genetic variant identified as frequent cause of deafness in adults

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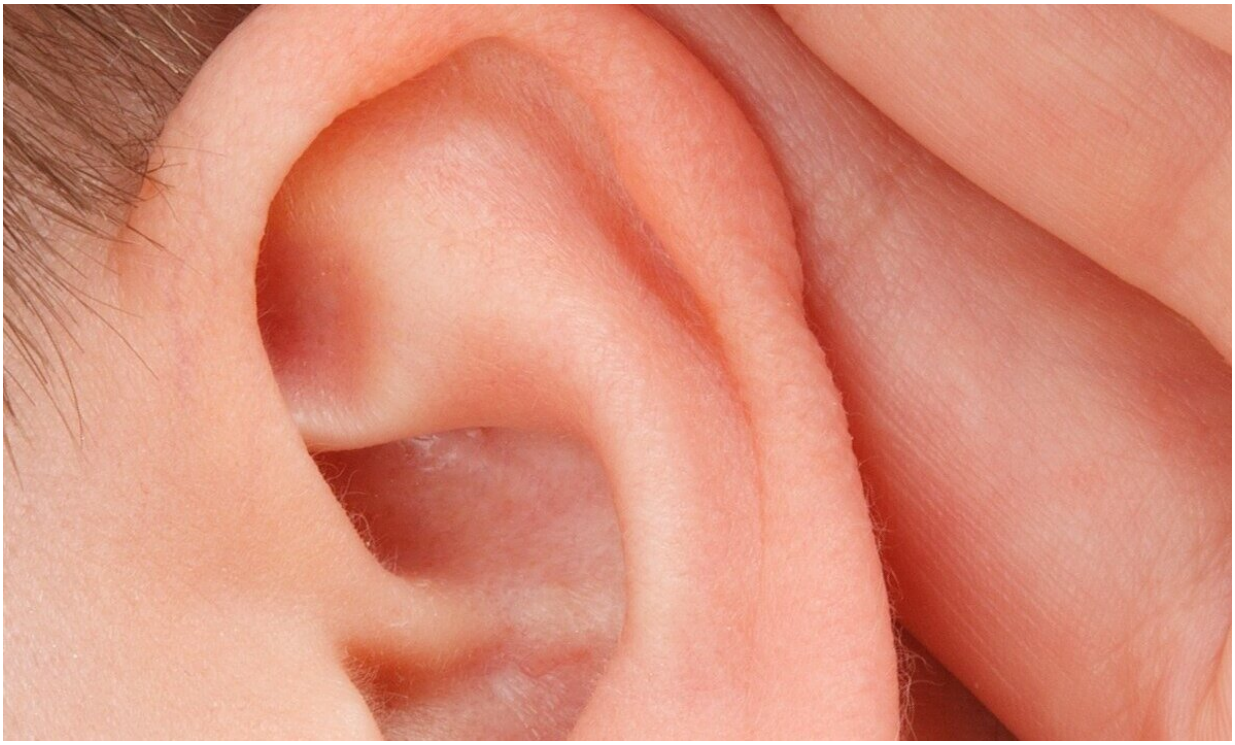

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A common inherited genetic variant is a frequent cause of deafness in adults, meaning that many thousands of people are potentially at risk, reveals research published online in the *Journal of Medical Genetics*.

Deafness in adults is known to be inherited. But, unlike childhood

deafness, the genetic causes largely remain a mystery, say the researchers, who suggest that their discovery makes it an ideal candidate for gene therapy.

Deafness is one of the most prevalent disabilities worldwide and has a major impact on quality of life. So far, 118 genes have been associated with the heritable form. Variants in these genes explain a large proportion of congenital and childhood deafness, but not adult deafness.

This is despite the fact that between 30% and 70% of hearing loss in adults is thought to be inherited.

The researchers had already discovered the chromosomal region involved in hearing loss in one family, but not the gene involved.

To explore this further, they carried out gene sequencing of this family among whom hearing loss in one or both ears had occurred as well as 11 other families (200 people in all).

Each family member had a general ear, nose and throat check and their hearing was tested in both ears.

The genetic sequencing in the first family revealed a missing section of the RIPOR2 gene in 20 of the 23 family members with confirmed hearing loss.

But this genetic variant was also found in three other family members aged 23, 40, and 51, who didn't yet have any hearing loss.

This prompted the researchers to carry out gene sequencing, and the same medical and hearing examinations, in a further 11 families affected by hearing loss.

The identical genetic variant was found in 39 of 40 family members with confirmed hearing loss as well as in two people aged 49 and 50 who weren't affected by hearing loss.

What's more, the RIPOR2 genetic variant was found in a further 18 out of 22,952 randomly selected people for whom no information on hearing loss was available.

Four family members with hearing loss didn't have the RIPOR2 genetic variant. Their deafness might have been associated with heavy smoking or genetic abnormalities other than that in RIPOR2, suggest the researchers.

While the particular manifestations of this genetically induced hearing loss varied, as did the age at which hearing problems began, its prevalence suggests that it is common (highly penetrant) and that many thousands of people might be at risk of deafness as a result, explain the researchers.

Based on their findings the researchers estimate that in The Netherlands alone the RIPOR2 genetic variant is likely present "in more than 13,000 individuals who are therefore at risk of developing [hearing loss] or who have developed [hearing loss] already due to this variant."

And they suggest that a further 30,000 people in northern Europe are likely to have this genetic variant and therefore be at risk of deafness.

"Because of the large number of subjects estimated to be at risk for [hearing loss] due to the c.1696_1707 del RIPOR2 variant, it is an attractive target for the development of a genetic therapy," they conclude.

More information: An RIPOR2 in-frame deletion is a frequent and

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highly penetrant cause of adult-onset hearing loss, *Journal of Medical Genetics*, DOI: 10.1136/jmedgenet-2020-106863

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