GENE Seeker

You know the risk of your child being a carrier of a genetic disease.

Autosomal dominant non-syndromic sensorineural deafness type DFNA

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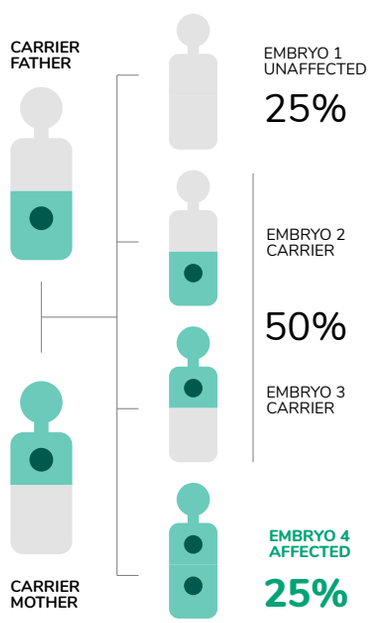
What is Autosomal dominant non-syndromic sensorineural deafness type DFNA?

Deafness is the most frequent form of sensorial deficit. In the vast majority of cases, the deafness is termed nonsyndromic or isolated and the hearing loss is the only clinical anomaly reported. In developed counties, 60-80% of cases of early-onset hearing loss are of genetic origin.

The majority of cases presenting at birth concern perceptive deafness (with a neurosensory origin associated with the inner ear) rather than conductive deafness (anomalies in the amplification of sound waves between the middle ear - tympanum and auditory ossicles - and the outer ear). Autosomal dominant forms are characterised by very early onset and bilateral hearing loss with varying degrees of severity (ranging from mild to profound). No malformations of the inner ear can be detected by CT scan. Mutations in the PDS gene are responsible for 7% of cases of childhood deafness. In these cases, the deafness is marked by early-onset, usually bilateral (but sometimes asymmetric) hearing loss with autosomal recessive transmission. This form of deafness is always associated with malformations of the inner ear that can be detected by CT scan. In rare cases, thyroid gland disease may also be present. For the autosomal dominant forms of deafness, mutations in the COCH gene result in progressive postlingual deafness associated with severe attacks of vertigo and subjective tinnitus. This form of deafness should be distinguished from Meniere's disease (see this term). Autosomal dominant mutations in the WFS1 gene cause either a form of hearing loss affecting mainly low frequency sounds or deafness associated with optic atrophy.



What is the next step if I'm a carrier of Autosomal dominant non-syndromic sensorineural deafness type DFNA?

If you are found to be a carrier of Autosomal dominant non-syndromic sensorineural deafness type DFNA, it is important that your partner be tested for the same genetic disorder.

What if my partner is not a carrier?

If your partner's test for Autosomal dominant non-syndromic sensorineural deafness type DFNA is negative, the chance to have an affected child is low. However there is currently no test able to detect all existing mutations, so there is always a residual risk that the person who has done the test is a carrier of other less frequent mutations.

What if both me and my partner are carriers of Autosomal dominant non-syndromic sensorineural deafness type DFNA?

When both parents are carriers of Autosomal dominant non-syndromic sensorineural deafness type DFNA the probability of having a child with Autosomal dominant non-syndromic sensorineural deafness type DFNA is 25%.

We recommend that you discuss your results with your doctor or genetic counselor in order to know more about reproductive options.

If both you and your partner are carriers, speak with your doctor or genetic counselor about reproductive options.

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