Hearing impairment and vestibular function in patients with a pathogenic splice variant in the LHX3 gene

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Background

In northern Sweden we have found a group of nine patients that are homozygous for the same mutation in the LHX3 gene.

LHX3 is a gene coding for a LIM-homeodomain transcription factor important for the fetal development of several organs such as the pituitary gland, spinal motor neurons and the inner ear. The LHX3 gene is expressed in the hair cells of both cochlear and vestibular organs but there are no previous reports on the balance in patients with mutations in the LHX3 gene.

Aim

The aims of this study are to evaluate hearing impairment and progression in a long-time follow-up setting for patients with homozygous mutations in the LHX3 gene and to investigate if there is a vestibular impairment connected to this mutation.

Method

We have retrospectively studied hearing-tests from newborn until today and we have done vestibular testing in six patients.

Result

In this study we show for the first time that a mutation in the LHX3 gene affects the function of both cochlear and vestibular organs. We can also show that the hearing impairment for this cohort is progressing over time.

Discussion

These clinical findings are important to better understand the role of LHX3 in the development of the inner ear and it also have direct clinical implications in the treatment of patients with this syndrome.