

New research increases knowledge of hereditary hearing loss

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Danish biologist Kirsten M. Sanggaard has written a Ph.D. thesis about hereditary hearing loss. Through interviews with ten families with a history of hereditary hearing loss, she identified some of the gene changes that cause hearing impairment.

We spoke to Ms. Sanggaard about the project, which was undertaken at the University of Copenhagen and supported by Widex.

How important a role does heredity play in hearing impairment?

Hearing loss is the most common sensory defect in human beings. One in 1000 newborn is reported to be hearing impaired. 2-3% in the age group from 40-50 and approx. 30% of those above 70 years have hearing loss exceeding 40 dB.

50% of all cases of hearing loss or deafness are caused by a hereditary change of the genes associated with hearing.

Moreover, there are more than 400 different hereditary syndromes, of which hearing impairment forms part. Today we know of more than 30 genes that may cause syndromic hearing impairment.

How far has research progressed in this field?

In 1994 not a single one of the genes that may cause hearing impairment or deafness had been identified. Today, however, we know of 37 different genes.

We know that there are as many as 129 areas in our chromosomes that contain a gene that may result in impaired hearing. But as these areas contain many genes, it is not always possible to isolate the specific gene causing the hearing impairment.

What result did you arrive at in your Ph.D. thesis?

Six of the families had BOR syndrome. In five of these, I succeeded in detecting changes in two different genes, EYA1 and SIX1, which contribute to the development of the inner ear and the kidneys. A consequence of changes in these genes is that the ears and kidneys do not develop as they should at the embryonic stage.

One family suffered from Helweg-Larsen syndrome. There I located the area on chromosome no. 1 where the gene causing the defect was.

The last three families had isolated hearing loss, and in these cases the results were less conclusive. In one of the families I identified a change in the gene for myosin VI. Myosin VI is found in the sensory cells in the inner ear. If these sensory cells do not function normally, we cannot perceive sound and consequently our hearing is impaired.

Why is it important to increase our knowledge about hereditary hearing loss?

Today, only 6% of all children at the age of 0-6 months with congenital hearing loss are identified and at the age of around one year, only 27%. These figures need to improve. Therefore, newborn screening - including a special sound test to detect any hearing impairment - has recently been introduced.

The more we know about the genes causing hearing loss and the type and degree of loss produced, the better the possibility of preparing the child and its parents for a life with hearing loss or deafness. The hope is that in the long run, a chip can be developed that makes it possible to examine many genes associated with hearing loss simultaneously by means of a simple blood test. And maybe, it will eventually be possible to design new treatment methods for deafness and hearing loss.

Facts about the BOR and Helweg-Larsen syndromes

BOR (Branchio-Oto-Renal) and Helweg-Larsen syndromes are examples of disorders in which hearing loss is a feature. This type of hearing loss is called syndromic as it is associated with specific traits additional to hearing impairment. Today we know of more than 400 different syndromes of which hearing impairment is a feature, and more than 30 genes that may cause syndromic hearing loss.

In addition to hearing impairment, patients with the BOR syndrome may suffer from kidney abnormalities and branchial fistulas (tracts that appear on the lower portion of the neck and may appear as a pit or bulge). BOR syndrome accounts for 2% of all hearing losses.

The clinical manifestations for patients with Helweg-Larsen syndrome include the inability to sweat due to the reduced amount of sweat glands. The condition can be life threatening, but fortunately the syndrome is rare.