What is DFNA9 ?

Text accompanying the video

What is DFNA9 ?

DFNA9 is a very progressive and dominant hereditary disease (also called Cochlear Mutation) that takes away your hearing and your balance.

« What is it exactly ? »

The first 3 letters DFN come from the word « deafness »

The letter A refers to « autosomal dominant transmission » which means that the disease is inherited (for more information, see below).

The addition of the figure 9 indicates chronologically when the disease has been recognized by means of several studies.

As a summary, DFNA9 is a hereditary disease that is found in several areas in the world.

DFNA9 is caused by a mutation in the COCH gene.

Until today, different types of mutations in the COCH gene have been identified, each mutation demonstrating another pathology.

It is difficult to say how many people did get the DFNA9 diagnosis on a worldwide basis.

According to more than 80 scientific studies, it seems that most people with DFNA9 can be found in Belgium and the Netherlands.

In 1998, a common DFNA9 mutation, with the name p.Pro1Ser (abbreviated P51S), in the COCH gene was identified. By consequence, most probably all DFNA9 patients have the same ancestor with a gene defect.

The first signs of the disease usually start at the age of 40.

Generally, this hearing loss is assisiated with serious balance issues and vertigo.

The hearing loss and the deterioration of the vestibular system happen asynchronous.

Finally, the hearing and the balance are completely lost.

« What is autosomal dominant transmission ? »

Certains pathologies are inherited “autosomal dominant”.

That means that the person inherits one normal copy of the gene and one defect copy of the gene, but the defect copy of the gene will mutate and will be dominant over the normal copy.

Therefore, these people will finally develop the disease.

Autosomal means that transmission equally applies to boys and girls.

Certains inherited dominant pathologies are already visible at birth.. Others, like DNFA9, are visible later.

A parent having DFNA9 can either transmit the normal COCH gene or the defect copy to its child. Each child has by consequence 50% chance to inherit the defect gene and so develop the disease.

Equally the child has a 50% chance to receive the normal gene. In that case, the child won't be a carrier of the pathology and won't be able to transmit the disease to its children.

Currently, there is no solution to stop or to heal DFNA9. However, different ideas and possibilities are examined in order to develop a genetic therapy in order to stop the pathology.