Usher Syndrome

Usher syndrome is an *inherited* (passed on from parents) condition. It causes a serious hearing loss, usually present at birth or shortly thereafter. It also has an advancing (*progressive*) vision loss. This is caused by *retinitis pigmentosa* (RP). RP is a group of inherited diseases. They cause night-blindness and side (*peripheral*) vision loss through the progressive breaking down (*degeneration*) of the retina. That is the light-sensitive tissue at the back of the eye that is needed for vision.

TYPES OF USHER SYNDROME - TYPE 1, TYPE 2 AND TYPE 3

- Individuals with Usher syndrome **type 1** are nearly or completely deaf. They experience problems with balance from a young age. They usually begin to show signs of RP in early adolescence.
- Individuals with Usher syndrome **type 2** experience moderate to severe hearing impairment. They have normal balance, and show symptoms of RP later in adolescence.
- Individuals with Usher syndrome **type 3** are born with normal hearing. But they develop RP and then progressive hearing loss.

HOW IS USHER SYNDROME INHERITED?

The Usher syndrome types are inherited as an *autosomal recessive trait*. This means that an affected person receives one abnormal gene from each of his or her parents. A person who inherits a gene from only one parent will be a carrier. But he/she will not develop the disease. A person with Usher syndrome must pass on one disease gene to each of his or her children. But unless the person has children with another carrier of Usher genes, the individual's children are not at risk for developing the disease. Currently we cannot reasonably test everyone for carrier status. But this may change in the years ahead.

HOW IS USHER SYNDROME DIAGNOSED?

Since individuals with Usher syndrome have both hearing and visual problems (*symptoms*), testing of both systems is performed. This testing includes:

- Visual function tests: visual fields and electroretinogram (ERG).
- A retinal examination.
- Hearing tests.
- Balance tests for all patients age ten years and older.
- Some of the genes that cause Usher syndrome have been identified. But the *diagnosis* (learning what is wrong) is still based on visual (*ocula*r) and clinical testing.

IS GENETIC TESTING FOR USHER SYNDROME AVAILABLE?

At this time, genetic testing for Usher syndrome is done only as part of research projects. This is due to many factors. Usher syndrome is not caused by only one gene. Finding the genes is an important advance in the fight against Usher syndrome. Further study is required to characterize these genes, and learn how the mutated genes cause Usher syndrome. Additional genes that cause Usher syndrome also need to be identified. Several researchers throughout the world are working on this. Findings from this research may one day allow treatments for Usher syndrome to be developed.

RESOURCES

The information provided in this Resource Guide was developed by the National Eye Institute (NEI) to help patients and their families in searching for general information about Usher syndrome. An eye care professional who has examined the patient's eyes and is familiar with his or her medical history is the best person to answer specific questions.

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