FOUNDATION FIGHTING BLINDNESS

USHER SYNDROME

What is Usher syndrome?

Usher syndrome is an inherited condition characterized by progressive vision loss and hearing impairment. The vision loss is due to retinitis pigmentosa (RP), a degenerative condition of the retina, and usually appears during adolescence or early adulthood. Balance may also be affected in people with Usher syndrome. Symptoms and disease progression vary from person to person.

There are three general categories of Usher syndrome. People with Usher syndrome type 1 (USH1) are usually born with severe hearing loss and experience problems with balance. The first signs of RP — night blindness and loss of peripheral vision — usually appear in early adolescence.

In Usher syndrome type 2 (USH2), newborns have moderate to severe hearing impairment. Symptoms of RP typically start shortly after adolescence. Visual problems progress less rapidly than in Usher type 1, and hearing loss usually remains stable.

A rarer, third type of Usher syndrome (USH3) was documented in 1995. Children with USH3 are usually born with good or only mild impairment of hearing. Their hearing and vision loss is progressive, starting around puberty. Balance may also be affected.

Usher syndrome is further subdivide into types, based on the mutated gene causing the disease. For example, people with Usher syndrome type 1B have mutations in the MY07A gene.

Hearing loss in Usher syndrome is due to a genetic mutation affecting nerve cells in the cochlea, a sound-transmitting structure of the inner ear. The same genetic

defect also adversely affects photoreceptor cells in the retina, leading to vision loss. The retina is a thin layer of tissue lining the back of the eye composed of light-sensing photoreceptor cells. These cells — also known as rods and cones — are responsible for converting light into electrical signals that the brain interprets as vision.

How is Usher syndrome inherited?

Usher syndrome is passed from parents to their offspring through an autosomal recessive inheritance pattern. In this type of inheritance, two copies of a mutated gene, one from each parent, are required for the child to be affected. A person with only one copy of the gene is a "carrier" and rarely has any symptoms.

Researchers estimate that as many as 50,000 people in the U.S. have Usher syndrome. Worldwide, it is the leading cause of combined deafness and blindness. Approximately 30 percent of people with RP report some degree of hearing loss, and about half of them are diagnosed with Usher syndrome.

Genetic testing is available to help people define their condition and the risk of other family members or future offspring being affected. A genetic diagnosis can also help a person qualify for a clinical trial. Genetic counselors are excellent resources for discussing inheritability, family planning, genetic testing and other related issues.

What treatments are available?

While there are no treatments for Usher syndrome, intensive research is underway to discover the causes of, and treatments for, all types of the disease.

In addition to nutritional therapies, researchers are working toward a variety of potential treatments for Usher syndrome, including gene therapies, stem cell treatments, and drugs.

For more information on research and clinical trials for Usher

syndrome, refer to the Foundation publication *Usher Syndrome: Research Advances.*

Are there any related diseases?
Other conditions, some of which are also inherited, can result in deafness and deaf-blindness, but are not related to Usher syndrome. However, the RP associated with Usher syndrome shares most of its characteristics with other forms of RP. Researchers expect that advances in understanding and treating other forms of RP will directly benefit people with Usher syndrome, and vice versa.

Extensive information on research and clinical trials for Usher syndrome, as well as low vision resources, are available at www.FightBlindness.org