WHAT YOU SHOULD KNOW ABOUT USHER SYNDROME

WHAT IS USHER SYNDROME?

Usher syndrome is an inherited disease causing combined hearing loss and vision loss. The vision loss is due to retinitis pigmentosa (RP), a degenerative condition of the retina.

Researchers estimate that as many as 25,000 people in the U.S. have Usher syndrome. Worldwide, it is the leading cause of combined deafness and blindness. About 30 percent of people with RP report some degree of hearing loss, and about half of them are diagnosed with Usher syndrome.

THERE ARE THREE GENERAL CATEGORIES OF USHER SYNDROME:

Type 1 (USH1):
- Usually born with severe hearing loss.
- Experience problems with balance.
- The first signs of RP usually appear in early adolescence.

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.

Type 3 (USH3):
- Usually born with good or only mild impairment of hearing.
- Progressive hearing and vision loss, starting around puberty.

WHAT TO EXPECT WITH USHER SYNDROME:

- Effects both vision and hearing.
- Often diagnosed in childhood or adolescence.
- Can also cause problems with balance.
- Progression varies from person to person.

HOW IS USHER SYNDROME INHERITED?

Usher syndrome is autosomal recessive. That means it is genetically passed when both parents (unaffected carriers) have one gene for the syndrome paired with one normal gene.

There is a 25% chance of their child inheriting the two Usher syndrome genes.

GENETIC TESTING

Genetic testing helps people definitively diagnose 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.

The retina is a thin piece of tissue lining the back of the eye. Photoreceptor cells — also known as rods and cones — are responsible for converting light into electrical signals.

Hearing loss in Usher syndrome occurs, because the gene mutations affecting the retina also affect the cochlea, a sound-transmitting structure of the inner ear.

To learn more about living with Usher syndrome, finding a specialist, genetic testing or clinical trials, visit FightingBlindness.org or call 888-332-3667.