USHER SYNDROME

Division of Human Genetics
University of Cape Town

WHAT IS USHER SYNDROME?

Usher Syndrome is an inherited condition characterized by hearing impairment and progressive vision loss. Balance may also be affected. Symptoms vary from person to person and progress at different rates.

Hearing loss in Usher Syndrome is due to a genetic mutation (fault) affecting nerve cells in the cochlea, a sound-transmitting structure of the inner ear.

The vision loss is due to Retinitis Pigmentosa (RP), a degenerative condition of the retina, and usually appears during adolescence or early adulthood. The retina is a delicate tissue in 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 impulses that transfer messages to the brain. In both cases, hearing and vision loss are caused by the same genetic change, or fault.

WHAT ARE THE SYMPTOMS?

There are at least three different forms 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), new-born children 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.

HOW IS USHER SYNDROME INHERITED?

Usher Syndrome is an autosomal recessive conditions which means that an affected child has inherited two faulty Usher Syndrome genes, one from each parent. In other words, each parent has at least one faulty Usher Syndrome gene, which has been passed on to the affected child. (Refer to Autosomal Recessive fact sheet 10). A person with only one copy of the gene is a “carrier” and rarely has any symptoms.

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 this genetic condition. Researchers have found numerous genetic variations causing Usher Syndrome, allowing for the designation of a variety of subtypes (i.e., 1A, 1B, IC, 1D, 1E, 1F, 1G, 2A, 2B, 2C and 3A).

Oxford Biomedica have just started the USHTAT Phase l / ll gene therapy trial to replace the defective gene in Usher Type 1b. The gene involved in this type of Usher Syndrome is the MYO7A gene. This 18-person study began in Oregon USA early in 2012 and a second part of the trial will begin in Paris in 2013. The study is scheduled to run through to December 2014. Evaluating safety is the primary goal of the trial, though investigators will also be looking at the treatment’s effect on the retina and vision.

Usher patients will also benefit from research looking into other kinds of therapy for RP including artificial retinal implants, Optogenetics and stem cell treatments.

GENETIC COUNSELLING

Genetic counsellors are excellent resources for discussing heritability, risk assessment and genetic management, family planning, genetic testing, and other related issues. Prior to genetic testing a visit to a genetic counsellor is required. Genetic counselling is generally funded by medical aids.

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TESTING

Worldwide, it is the leading cause of combined deafness and blindness. Approximately 30% of people with RP report some degree of hearing loss, and about half of them are diagnosed with Usher Syndrome. Genetic testing is available through Retina South Africa to help people define their condition and the risk of other family members or future offspring being affected.

NUTRITION

Patients with Usher Syndrome are advised to take a specially formulated supplement- Retina Plus, which may reduce the rate of vision loss. This supplement contains Lutein, Zeaxanthin, Alpha Lipoic Acid and L- Glutathione. A healthy diet rich in leafy green vegetables and other fruit and vegetables is also recommended. Smoking, over-exposure to bright sunlight and all artificial colorants, flavourings and preservatives should also be avoided.

Please discuss any supplementation with your eye specialist.

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 the understanding and treatment of other forms of RP will directly benefit people with Usher Syndrome, and vice versa.

SOUTH AFRICAN RESEARCH

The University of Cape Town, in partnership with Retina South Africa, is conducting a genetics research project to investigate the genes causing retinal degeneration in South African families. In addition, a gene-testing service is offered by Retina South Africa. A co-payment plan is available to members of RSA who wish to find theirspecific genetic mutation. Only people who have a genetic diagnosis will be eligible for upcoming gene-specific therapy trials and treatments.

Who do I contact for more information regarding testing?

Division of Human Genetics Molecular Laboratory:

Prof. Jacquie Greenberg

(021) 406-6299

Genetic Nurses:

(021) 406-6304

Information supplied by Retina South Africa

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