Usher Syndrome

Usher syndrome is an inherited disorder that is characterized by (1) moderate to profound hearing impairment which is present at birth or shortly thereafter, and (2) progressive vision loss due to retinitis pigmentosa (RP). The condition is named after a British ophthalmologist, C.H. Usher, who in 1914 described several cases in which the link between congenital deafness and RP was stressed. RP is a disorder of the sensory cells of the retina of the eye that includes the rods that control night vision, and cones that control day vision and color vision. In RP, the rods deteriorate first, then the cones. The rod cells which lie on the periphery of the retina lose function and fail to transmit information about changes in light levels and shapes of objects on the periphery. So the first symptom to appear in RP is night blindness; this may also be coupled with difficulty in adapting to bright light or rapidly changing light conditions. In RP the extent and speed at which vision deteriorates is extremely variable. In some individuals the breakdown of the retina is so marked that at the age of six they require special education for the visually impaired. Others may have no severe restriction of vision until they reach their thirties, and it is not uncommon for some to retain their reading vision well into old age.

Clinical Description

There are three known types of Usher syndrome. Individuals with Usher Type I are born profoundly (completely) deaf and experience problems with balance due to absent vestibular function. Because of the balance problems, babies have a hard time learning to crawl and walk. They may use a 5-point crawl with their heads down on the ground. Walking may be delayed, and they often enjoy twirling and spinning since they do not get dizzy. Individuals usually begin to exhibit the first signs of RP: night blindness and loss of peripheral vision. Individuals with Usher Type II experience moderate to severe hearing impairment at birth, but they do not have balance problems. Symptoms of RP develop later in adolescence. With Usher Type III, hearing loss and vision loss due to RP are both progressive.

Genetics

Usher is transmitted genetically by an autosomal recessive gene. Autosomal implies that the gene is not sex linked and so both males and females are equally affected by the condition. In this type of inheritance, two copies of an Usher syndrome gene, one from each parent, are required for a person to have the syndrome. An individual who has only one copy of the gene, is called a carrier and will have no symptoms of the disorder. The child with Usher syndrome has two recessive genes for the trait and will transmit these genes to their offspring.

Visual Adaptations and Other Considerations

Some useful adaptations for individuals with Usher syndrome include: high illumination with no glare, absorptive lenses, infra-red viewing devices, prism glasses to increase visual field, and closed circuit television for maximum contrast. Some educational considerations include: observing lighting conditions and adapting requirements to them, teaching organized search patterns using a “grid” pattern to aid the student in locating objects or visual targets, seating considerations for field loss, glare considerations, and length of time to complete assignments.

Contact Information

National Information Clearinghouse on Children Who Are Deaf-Blind at DB-Link, Teaching Research Division, Western Oregon University, 343 North Monmouth Ave., Monmouth OR 97361, (800) 438-9376, www.tr.wou.edu/dblink/usherfulltext.htm.

The Foundation Fighting Blindness, Executive Plaze 1, Suite 800, 11350 McCormick Road, Hunt Valley, MD 21030-1014, (888) 394-3937, www.blindness.org.

Additional Web sites: www.deafblind.com/usher.html, www.nidch.nih.gov/health/pubs_hb/usher.htm