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Defining Usher Syndrome and Looking for Signs

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Usher Syndrome is the name given to a group of genetic conditions involving both hear- ing loss and retinitis pigmentosa, a progressive, degenerative eye disease. There are three types of Usher syndrome described in the chart that follows. Some people with Usher syndrome are profoundly deaf and some are hard-of-hearing. Some may be born hearing or hard-of-hearing and lose their hearing later. One group has significant bal- ance problems in addition to vision and hearing loss. There are ten genes already identi- fied that cause Usher syndrome and there may be more.

Does a child inherit Usher syndrome from his or her parents? The Usher Syndromes are all autosomal (not sex linked) recessive conditions. This means that both parents are asymptomatic carriers (having no symptoms of Usher syndrome). Neither parent knows that he or she is a carrier of this condition until they produce a child with Usher syndrome. Both parents must be carriers; each pregnancy of this couple carries a one in four chance that the child born will have Usher syndrome. If only one parent is a carrier, then the child born cannot have Usher syndrome. Parents find out they are car- riers of Usher syndrome only when their child is diagnosed. Typically they have never heard of the condition before and are offered little information. Consequently the diagno- sis, which means that their deaf or hard-of-hearing child may well become blind, is a terrible and overwhelming shock.

How common is Usher syndrome? Among children who are born profoundly deaf, 3 to 6 percent can be expected to have Usher syndrome, Type 1 (see note 1). Among children ....

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children born hard of hearing, Usher syndrome, Type 2, probably occurs at the same rate (see note 2). Vision screenings for retinitis pigmentosa are not routinely performed in the school setting, resulting in an under-identification of students with Usher syndrome.

Types of Usher Syndrome. There are three types of Usher Syndrome. Usher Syndrome, Type I, born profoundly deaf, retinitis pigmentosa (RP), and balance problems. Night blindness in infancy or early childhood. Can be legally blind by early adulthood. Seven different genes have been identified so far that cause Usher syndrome Type I. Usher IA through Usher 1F. Usher Syndrome, Type II has moderate stable hearing loss, RP, and normal balance; blind spots by late childhood or teens. Can be legally blind by early adulthood. There are at least 2 genes that cause Usher Syndrome, Type II, Usher IIA and Usher IIB. Usher Syndrome, Type III has progressive hearing loss, RP, and the status of the balance system is still being determined, night blindness in childhood or teens, can be legally blind by early to mid adulthood. There are at least 2 genes that cause Usher Syndrome, Type III, Usher IIIA and Usher IIIB. More genes are still being identified.

What things might be noticed first about a child with Usher syndrome? Although many students with Usher syndrome may be unaware that they have a visual loss, they do realize, and often at an early age, that they have difficulties with vision that other kids don't seem to have. A common first sign of Usher syndrome is the inability to see clearly at night. Parents or teachers may notice a number of things: that the child trips or bumps into objects; has problems walking around the home in dim light; is uncomfortable with unfamiliar steps or curbs; and may have a heightened fear of the dark. A second sign is the inability to see peripherally (above, below, and to the sides) under any lighting conditions. This is often described as tunnel vision. A student with Usher syndrome may not notice another student waving hello from the side, an obstacle on the floor, a step or curb, a street pole off to the side of one shoulder, etc.

Another sign is difficulty adjusting to changes in lighting: entering or leaving a darkened movie theater; entering or leaving a building on a bright sunny day.
Additional signs are problems in maintaining balance, blind spots (scotoma), sensitivity to glare (under snow, bright light or sun conditions), and problems with clarity of central vision (acuity).
To summarize, the common signs of Usher syndrome are:

- Night blindness
- Peripheral vision loss
- Difficulties adapting to lighting changes
- Blind spots
- Glare discomfort (often related to developing)
- Balance problems
- Acuity problem

Why do students with Usher syndrome become night blind? All students with Usher syndrome have hearing loss and retinitis pigmentosa (RP). In the typical course of RP, blood vessels constrict decreasing the flow of vital nutrients to the peripheral retina. The retina of the human eye contains two types of light-receptor cells, rods and cones. Cones allow us to see clearly under daylight conditions and give us our color vision. Rods allow us to see clearly under evening light conditions, and give vision only in shades of gray. The loss of night vision is caused by the deterioration of rods. The peripheral part of the retina is where there the highest concentration of rods exists. As these cells die, the ability to see clearly in the evening decreases. This is what most students with Usher syndrome experience first, the loss of night vision, or what is commonly called night blindness.

Is there a cure for retinitis pigmentosa? At this time (June 1997) there is no cure for the degenerative eye disease known as retinitis pigmentosa. Research is moving rapidly and focuses on two main areas: genetic research and transplantation of retinal tissue.

Note 1. *Kimberling W. J., Möller, C. (1995), Clinical and Molecular Genetics of Usher Syndrome, Journal of the American Academy of Audiology, 6, 63-72 (1995)*
Note 2. *Personal communication with Sandra Davenport, MD*

Joe Cioffi and Ilene Miner presented at the Usher Syndrome Screening Workshop sponsored by Connections on April 6-7 at Maryland School for the Deaf. With their assistance, and that of our trainer Llewellyn Jones, we have established a Statewide Team to conduct screenings for Usher Syndrome in the Local School Systems. The Screening Team will be available beginning in the Fall of 2006. If you are interested in scheduling free screenings for students in your school system, please contact Donna Riccobono at 301-405-0482 or donnaric@umd.edu.