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COLORADO
Assistance for those
SERVICES TO
with both vision
CHILDREN WITH
and hearing loss
DEAFBLINDNESS

Fact Sheet

Alport syndrome is a rare inherited condition that affects the kidney, inner ear (cochlea) and eye. In 1927 British Dr. Cecil A. Alport described three generations of a family with combinations of progressive kidney disease, hearing loss, and vision problems / impairment. The most common symptom among these individuals was the presence of blood in the urine (hematuria), which indicates abnormal kidney function. In the years following, accounts of many more families with the same symptoms were documented, which led to the condition being named Alport syndrome in 1961.

A genetic mutation or change in the Type IV collagen family of fibrous proteins that make up connective tissue causes Alport syndrome. Type IV collagen is a major part of important tissue structures called basement membranes that are present in all tissues including the kidney, inner ear, and eye. The results of this inherited mutation are progressive deterioration of the kidneys as scar tissue forms in the normal kidney structures (glomeruli and tubules). As the kidneys filter proteins out of the blood, these molecules damage the filtering system or glomeruli because of the abnormal collagen makeup. This process is known as fibrosis and it eventually leads to kidney failure or end-stage renal disease (ESRD). Because of the loss of kidney function, many people with Alport syndrome also develop high levels of protein in their urine (proteinuria) and high blood pressure.

Another symptom of Alport syndrome is sensorineural hearing loss caused by abnormalities of the inner ear of younger individuals. The hearing loss is not present at birth, but becomes evident by late childhood or early adolescence and, generally, occurs before the onset of kidney failure.

Eye abnormalities are another sign in affected individuals. The individual may have misshapen lenses in the eyes (anterior lenticonus) causing slow progressive loss of vision, which requires them to change the prescription of their glasses frequently. Anterior lenticonus may also lead to formation of cataracts. Abnormal coloration of the light-sensitive tissue at the back of the eye (retina), called dot-and-fleck retinopathy may be present. Recurrent corneal erosion may also occur in individuals with Alport syndrome. Those who experience this problem should take measures to protect their corneas from minor trauma, such as wearing goggles when riding a bike.

Alport Syndrome is estimated to affect around 1 in 5,000 people. Alport syndrome affects boys more often than girls because 80% of the time, the disease is passed on by a mutation of the X chromosome (called X-linked Alport Syndrome). Males only have one X-chromosome while females have two X chromosomes. In girls, the normal X chromosome buffers the effect of the

mutated gene. The other 20% of persons with Alport syndrome have the autosomal recessive or autosomal dominant form of the disease where males and females are equally affected.

Males with the X-linked form of Alport syndrome develop kidney failure by the teenage years or early adulthood, but the onset of kidney failure may not happen until 40 to 50 years of age. Most females with X-linked Alport syndrome do not progress to the kidney failure stage. However, as women with Alport syndrome age, the risk of kidney failure does increase. Boys and girls with the autosomal recessive form of Alport syndrome will develop kidney failure by their teens or young adult years. People with autosomal dominant Alport syndrome are usually well into middle age before kidney failure develops.

Approximately 80% of young males with X-linked Alport Syndrome develop hearing loss at some point in their lives, often by their teenage years. In young females with X-linked Alport syndrome, hearing loss is less frequent and happens later in life. Boys and girls with autosomal recessive Alport syndrome typically develop hearing loss in childhood. Individuals with autosomal dominant Alport syndrome develop hearing loss at a later age.

What are the Functional Vision and Hearing Implications? The eye abnormalities seldom lead to permanent vision loss and may be corrected by prescriptive lenses or surgery to remove cataracts. Hearing is not affected in all cases of Alport syndrome, and hearing aids often prove to be very effective. Children and youth with Alport syndrome should be closely followed by an eye care specialist and audiologists for their vision and hearing status and need for changing glasses, cataract surgery and/or amplification devices. These individuals should also be evaluated and monitored to determine their educational needs and whether they will qualify for special education service due to a need for specialized instruction.

Resource Information:

Alport Syndrome Foundation website. This site is designed for patients and families affected by Alport Syndrome, a genetic kidney disease. It is their goal to provide a central location for information about this disease, to create an opportunity to meet and talk to other families who are dealing with Alport syndrome, and to support research aimed at curing the disease. www.**alportsyndrome**.org

For more information about the CO Services for Children and Youth with Combined Vision and Hearing Loss Project contact:

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Fact Sheets from the Colorado Services to Children and Youth with Combined Vision and Hearing Loss Project are to be used by both families and professionals serving individuals with vision and hearing loss. The information applies to children, birth through 21 years of age. The purpose of the Fact Sheet is to give general information on a specific topic. More specific information for an individual student can be provided through personalized technical assistance available from the project. For more information call (303) 866-6681 or (303) 866-6605. Updated: 6/13