Alport syndrome

Alport syndrome is an inherited disease that affects a part of the kidney called the glomerulus. The glomerulus is like a spaghetti strainer: it keeps the good things in (the “spaghetti”: red blood cells, proteins are the good things) and lets the waste out (the “water the spaghetti was boiled in”: extra water, extra salt, broken down medication particles). The waste comes out as urine. In Alport syndrome, the “strainer” (glomerulus) doesn’t work like it should. Because of this, red blood cells will be in the urine.

What are the types of Alport syndrome?

There are three types, which relate to which chromosome the gene is on:
--X-linked recessive
--Autosomal recessive
--Autosomal dominant -- this form tends to be more mild than the other two, and cause problems later in life.

What are the symptoms of Alport syndrome?

In addition to kidney problems, many people with Alport syndrome also have hearing problems and eye problems. Symptoms may include:

- Blood in the urine (hematuria), the most common and earliest sign of Alport syndrome
- Protein in the urine (proteinuria)
- High blood pressure
- Swelling in the legs, ankle, feet and around the eyes
- Hearing loss
- Vision loss

These signs and symptoms may be different, based on age, sex and the genetic type of Alport syndrome. For example, hearing and vision problems tend to be more common in boys than girls.

Kidneys. The most common symptom of Alport syndrome is the presence of blood in the urine (hematuria). Most boys with Alport syndrome start having hematuria as babies, and it is always present after that. Most girls with Alport syndrome also have hematuria, but it may come and go. The hematuria of Alport syndrome is usually “microscopic”, meaning it can not be seen with the naked eye. It can only be detected with a microscope or a urine dipstick. On the other hand, some children with Alport syndrome have brown, pink or red urine (“gross” hematuria) for several days, brought on by a cold or the flu. This blood in the urine will go away on its own. While it may be frightening, it is not harmful.

As boys with Alport syndrome grow, they begin to show other signs (symptoms) of kidney disease. These include protein in the urine and high blood pressure. These symptoms are often present by the time the boys are teenagers. Girls with Alport syndrome usually do not have protein in the urine and high blood pressure until much later in life, but sometimes these symptoms appear in teenaged girls with Alport syndrome.

Ears. Most boys with Alport syndrome will develop progressive (worsening) hearing loss in both ears at some point in their lives, often by the time they are teenagers. Fortunately, hearing aids are usually very effective with this type of hearing loss. Girls with Alport syndrome may also develop hearing loss, but less often than boys, and usually later in life.

Eyes. About 15% of boys with Alport syndrome have an abnormality in the shape of the lens of their eyes called anterior lenticonus. People with anterior lenticonus may have some problems with their vision, and some develop cataracts. Yearly visits with an eye doctor are needed to monitor for eye disease in boys.

How serious is Alport syndrome?

Over time, Alport syndrome causes worsening kidney damage. Most boys with Alport syndrome eventually develop kidney failure. They may need dialysis or a kidney transplant during their teenage or young adult years, but kidney failure can occur as late as 40-50 years of age in some men with Alport syndrome. Most girls with Alport syndrome do not develop kidney failure, but some might.

The risk and timing of kidney failure depends on which genetic type of Alports you have. Talk with your kidney doctor about the possibility of genetic testing to find out which type of Alports you have.

How is Alport syndrome diagnosed?

Currently, the diagnosis of Alport syndrome is based on thinking carefully about the patient's symptoms and their family medical history. Hearing and vision should also be tested. The evaluation can also include a blood test, urine tests, and a kidney biopsy. Occasionally, a genetic test can be used to confirm the diagnosis and help identify the genetic type of Alport syndrome.

How is Alport syndrome treated?

Currently there is no specific treatment for Alport syndrome. The same treatments that are used in people with high blood pressure and other symptoms of kidney disease are used in people with Alport syndrome. Kidney transplantation is usually very successful in people with Alport syndrome, and is considered the best treatment when kidney failure is approaching.

Where Can I go for more information?

http://www.unckidneycenter.org
https://www.alportsyndrome.org/index.html