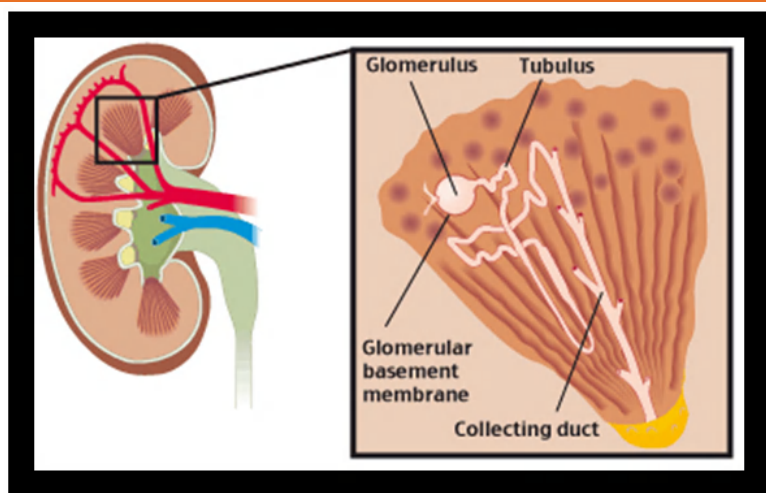


## ALPORT SYNDROME



[http://www.socialstyrelsen.se/rarediseases/Bilder/Alports\\_syndrom\\_eng.gif](http://www.socialstyrelsen.se/rarediseases/Bilder/Alports_syndrom_eng.gif)

Alport Syndrome is an inherited disease that primarily affects the glomeruli, the tiny tufts of capillaries in the kidneys that filter wastes from the blood. The disease was first described by an English doctor named A. Cecil Alport. Alport Syndrome is caused by changes in genes (mutations) that affect type IV collagen, a protein that is important to the normal structure and function of glomeruli. The earliest symptom of the disease is blood in the urine (hematuria).

### What are the symptoms?

Alport Syndrome always affects the kidneys. Many people with Alport Syndrome also have hearing problems and abnormalities of the eyes, because the type IV collagen proteins are important to the normal structure and function of the inner ear and the eye..

**Kidneys.** The central feature of the disease is the presence of blood in the urine (hematuria). Boys with X-linked Alport Syndrome develop hematuria in infancy, and it is always present. The great majority of girls with X-linked Alport Syndrome also have hematuria, but it may come and go. The hematuria of Alport Syndrome is usually microscopic, meaning it can only be detected with a microscope or a urine dipstick. Sometimes children with Alport Syndrome have brown, pink or red urine (gross hematuria) for several days, brought on by a cold or the flu. This gross hematuria will go away on its own and while it may be frightening, it is not harmful.

As boys with Alport Syndrome grow, they begin to show other signs of kidney disease, including protein in the urine and high blood pressure. These symptoms are often present by the time the boys are teen-agers. Girls with Alport Syndrome usually do not have protein in the urine and high blood pressure until much later in life, but occasionally these symptoms appear in teen-aged girls with Alport Syndrome.

**Ears.** Deafness is another important feature of Alport Syndrome. About 80% of boys with Alport Syndrome will develop deafness at some point in their lives, often by the time they are teenagers. The deafness affects both ears. Fortunately, hearing aids are usually very effective in these people. Girls with Alport Syndrome may also develop deafness, but less frequently than boys, and usually later in life. Kidney transplantation does not improve the deafness of Alport Syndrome.

**Eyes.** About 15% of men with Alport Syndrome have an abnormality in the shape of the lens called anterior lenticonus. People with anterior lenticonus may have some problems with their vision, and may develop cataracts.

### How serious is Alport Syndrome?

Alport Syndrome causes progressive kidney damage. The glomeruli and other normal kidney structures such as tubules are gradually replaced by scar tissue, leading to kidney failure. All boys with Alport Syndrome, regardless of the genetic type, eventually develop kidney failure. These boys often need dialysis or transplantation during their teen-age 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 X-linked Alport Syndrome do not develop kidney failure. However, as woman with Alport Syndrome get older the possibility of kidney failure increases.

All boys and girls with autosomal recessive Alport Syndrome develop kidney failure, usually by their teens or young adult years. People with autosomal dominant Alport Syndrome are usually well into middle age before kidney failure develops.

### How is Alport Syndrome diagnosed?

Currently, diagnosis of Alport Syndrome relies on careful evaluation of the patient's clinical features, family history and results of tissue biopsies. Alport Syndrome produces unique changes in the walls of the blood vessels of the glomeruli that can be detected by electron microscopy of kidney biopsy material. Kidney biopsies can also be tested for the presence or absence of the type IV collagen alpha-3, alpha-4 and alpha-5 chains. This information is often very helpful in confirming a suspected diagnosis of Alport Syndrome. An alternative diagnostic procedure is skin biopsy. The type IV collagen alpha-5 chain is normally present in the skin. In most men with the X-linked form of Alport Syndrome the alpha-5 chain is completely missing from the skin.

### 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 the best treatment when end-stage kidney failure is approaching.

Medical researchers are very interested in understanding why people with Alport Syndrome develop kidney failure, and in developing treatments that can slow or prevent the development of kidney failure. Several treatment approaches are being tested in animals with Alport Syndrome.

### How is Alport Syndrome inherited?

Type IV collagen is actually a family of six proteins, or chains, that are known as alpha-1, alpha-2, alpha-3, alpha-4, alpha-5 and alpha-6. Mutations that affect the alpha-3, alpha-4 or alpha-5 type IV collagen chains can cause Alport Syndrome

There are three genetic types of Alport Syndrome

**X-linked.** This is the most common form of Alport Syndrome. About 80% of the people with this disease have the X-linked type. Boys with this type are severely affected and always develop kidney failure sometime in their lives. Girls with this type usually have milder symptoms than boys, but they can develop kidney failure. The rest of the people with Alport Syndrome have either the **autosomal recessive type** which affects 15 percent, or the **autosomal dominant type**, which affects 5 percent.

### What is the outlook for Alport Syndrome?

Researchers recently isolated the gene responsible for Alport Syndrome. The research disclosed three mutations of the gene. This discovery not only will permit more precise diagnosis of Alport Syndrome, but it opens the possibility for future gene therapy for this disorder.

Reference:

<http://www.kidney.org>