



# Patient Guide

**By Patients for Patients**

Developed by the Emerging Leadership Council and  
Medical Advisory Committee of Alport Syndrome Foundation

## What is Alport syndrome?

Alport syndrome is a rare, genetic condition that is passed from parent to child. It can also occur spontaneously, meaning that no one else in the family has been affected before.

Alport syndrome affects the kidneys and may progress to kidney failure regardless of sex assigned at birth. It also may cause hearing loss and/or changes to the eyes.

Alport syndrome uniquely impacts individuals based on a number of factors, such as genetics. The signs, symptoms, and rates of disease progression can vary greatly from person to person. Alport syndrome is considered a type of chronic kidney disease.

## What causes Alport syndrome?

Alport syndrome is caused by genetic mutations in a person's DNA. Individuals with Alport syndrome typically have defects in their COL4A3, COL4A4, or COL4A5 genes (or a combination of these).

The mutation causes a protein called type IV collagen to form incorrectly. Type IV collagen is a building block of the basement membranes found in the kidneys, inner ears, and eyes. Because these tissues don't have the proteins they need, the affected organs may begin to lose function over time.

## Who does Alport syndrome affect?

Alport syndrome can occur in people of all ages, sexes, and ethnic backgrounds. Recent research suggests it's likely that more people have Alport syndrome than previously thought, thanks to increased awareness and genetic testing. However, it is still considered a rare disease, meaning it affects less than 200,000 people in the United States.

The main symptoms of Alport syndrome and their similarity to other more common kidney diseases can make it difficult to diagnose. For example, blood (hematuria) and protein (proteinuria) in the urine are classic symptoms of several chronic kidney diseases besides Alport syndrome.

Because Alport syndrome is rare, receiving an [incorrect initial diagnosis](#) is common. Collecting a full patient history — including all of the patient's symptoms and family history — can help health care providers make an accurate diagnosis.

## How is Alport syndrome diagnosed?

For individuals with a "suspected diagnosis," there are many important reasons to gain a confirmed diagnosis: earlier access to [treatment](#), ability to identify other family members who may be at risk, clinical trial participation, and more. Currently, non-invasive and affordable diagnostic options are available for patients and families.

No-charge/minimum-charge genetic testing (based on insurance coverage) has recently become available to more patients and can be done with a simple blood test, cheek swab, or saliva sample. Information on how to obtain genetic testing can be found on ASF's [Genetics Overview page](#).

For many years, the most common way of [diagnosing Alport syndrome](#) was through a kidney biopsy. In a kidney biopsy, a small sample of tissue is taken using a needle that is placed through the skin and into the kidney. The sample is then examined under a microscope by experts. This is typically an outpatient procedure.

An increasingly less common method for diagnosis is with a skin biopsy, which can detect X-linked Alport syndrome (the most prevalent genetic type). Because type IV collagen is also found in the skin, its presence on a skin biopsy points to the same mutation in the kidneys.

## What are the most common symptoms of Alport syndrome?

The primary symptoms for many patients with Alport syndrome are blood in the urine (hematuria), protein in the urine (proteinuria), and high blood pressure (hypertension).

Most of the time, blood in the urine is microscopic and invisible to the naked eye. However, blood in the urine can occasionally become visible, and urine looks reddish brown or tea colored. This often happens with a cold/flu/virus/infection and goes away when the individual recovers.

If there is protein in the urine, it is also usually microscopic. One visible sign you may notice is that urine is very foamy when freshly passed.

As Alport syndrome progresses, high blood pressure can increase and cause swelling that's often seen in the ankles or calves.

Many patients also experience [hearing loss](#) and [eye abnormalities](#) due to the collagen defect. These symptoms can appear before or after the kidney symptoms are detected. No studies have yet been conducted to truly understand the prevalence of hearing loss and/or eye conditions.

## What are the less common effects of Alport syndrome?

There are less common, but documented ways Alport syndrome can affect individuals. While type IV collagen is mainly found in the kidneys, eyes, and ears, it is also found in lesser amounts in other parts of the body, such as the heart, lungs, and esophagus.

A small group of individuals experience noncancerous tumors of the smooth muscle (leiomyomas) found in the throat, windpipes, and genital tract. This condition is called diffuse leiomyomatosis. Diffuse leiomyomatosis can cause breathing, swallowing, or reproductive difficulties. Some patients with X-linked Alport syndrome may be at risk to develop leiomyomas, although this is rare.

There are both self-reported and documented cases of aortic aneurysm experienced by Alport syndrome patients of both sexes. The aorta is a large blood vessel that carries blood from the heart to the rest of the body. Aortic aneurysm can lead to life-threatening consequences if not detected and properly treated.

## What kinds of health care providers are most helpful in treating Alport syndrome?

It is very important for all individuals with Alport syndrome to have regular visits with certain medical specialists, such as:

- Nephrologists: Meeting regularly with a nephrologist allows for early treatment with the goal of prolonging kidney function. They can also help guide decisions around dialysis and transplant if that should become necessary.
- Renal dietitians: Renal dietitians work with patients in all stages of chronic kidney disease. They provide [nutritional education](#) and monitoring, and help create healthy meal plans based on an individual's lab values, needs, and preferences.
- Audiologists and ophthalmologists: Hearing specialists (audiologists) and eye specialists (ophthalmologists) can provide early assessment and intervention for hearing loss and eye conditions. Getting a baseline of hearing and eye health is helpful to detect any changes early.
- Additionally, heart specialists (cardiologists), lung specialists (pulmonologists), digestive specialists (gastroenterologists), and female reproductive specialists (OB/GYNs) may also be needed to evaluate and/or treat some of the less common symptoms associated with Alport syndrome.

## What other types of support may be helpful?

- Geneticists and genetic counselors: These health care providers specialize in interpreting genetic test results and can explain how [Alport genetic mutations](#) affect individual patients. They can also answer questions about who else in a family may be at risk of having Alport syndrome and how the genetic variant is inherited or passed down in families for the purpose of family planning.
- Mental health professionals: Clinical psychologists, clinical psychiatrists, mental health counselors, licensed clinical social workers, and pastoral counselors are all types of mental health professionals that can help individuals cope with the emotional toll of having a life-long condition.
- Educational specialists: Families with children who have Alport-related hearing loss are advised to work with their school system (K–12). An Individualized Education Plan (IEP) can be created that specifically includes evaluations over time and individualized accommodations to support successful learning. Often, these accommodations change as the student's hearing loss progresses. Hearing loss is covered by the [Americans with Disabilities Act](#) and ensures that students have access to often simple, but critical, modifications at school that make their learning environment suitable.

## How is Alport syndrome treated?

There is currently no cure for Alport Syndrome and no FDA-approved therapies to treat this specific condition. However, there are [medications](#) that can help reduce some of the renal symptoms and prolong kidney function.

Medications that are used to treat high blood pressure are often prescribed, even if patients don't have high blood pressure. These drugs are known as ACE inhibitors (such as lisinopril or ramipril) and ARB medications (such as losartan). These drugs have been shown to slow the leak of protein into the urine and slow the decline of kidney function. Other medications, such as diuretics, may be used later in the course of the disease to help manage retention of fluids that cause swelling.

Current research, including active [patient clinical trials](#), seeks to better understand the disease and find additional treatment options. If patients reach end-stage renal failure, when their kidneys can no longer meet the demands of the body, dialysis can help do the work of the kidneys until a transplant can be performed. ASF's website includes information about [dialysis options](#) and preparing for [transplant](#).

At this time, there are no treatments available to repair or prevent Alport-related hearing loss. However, many Alport patients note that daily use of hearing aids can be very helpful in social, educational, and work environments.

## What are the common side effects associated with treatment?

Chronic kidney disease itself, as well as some of the medications prescribed for Alport syndrome, can cause high potassium (hyperkalemia). It is usually treated with a [low potassium diet](#) (cutting out potatoes, tomatoes, avocados, etc.) and/or potassium-binding medications to help rid the body of excess potassium.

Keeping potassium under control is critical. High potassium levels can go undetected and cause serious consequences, such as stroke or heart attack.

Another common side effect of treatment is low blood pressure (hypotension), which can cause fatigue, drowsiness, dizziness, and fainting. ACE inhibitors can also cause a dry cough after you take the medication.

If patients experience dizzy spells, fainting, or a dry cough, contacting their doctor is advised. Switching to a different, similar medication or taking medications, such as ACE/ARBs, at a different time of day may be helpful.

## Can living with Alport syndrome affect emotional and mental health?

Living with a rare, genetic condition such as Alport syndrome can affect one's emotional well-being. Many individuals with Alport syndrome have reported experiencing emotions such as:

- guilt in unknowingly passing a genetic disease on to a child
- anxiety about an unknown future of disease progression
- frustration over the fatigue that can come with kidney disease and/or hearing loss
- coping with a rare disease that is unseen by others
- frustration when visiting health care providers who are not familiar with Alport syndrome

It is important for patients and their families to know they are not alone in these feelings, experiences, and concerns. Open communication with a health care team is essential in finding a treatment plan that optimizes the health of the patient as defined by the patient.

We also encourage patients to [connect with others living with Alport syndrome](#) via our dedicated Facebook support group, at our annual Alport Connect meetings, and through webinars or virtual meetings organized by ASF. These resources offer encouragement, education, and support. ASF can also connect individuals or families to one another any time just by emailing or calling staff members.

# Can individuals delay the effects of Alport syndrome?

The best ways to delay the effects of Alport syndrome are to be informed about the disease, find a nephrologist who is familiar with Alport syndrome (if possible), and develop an open line of communication with health care providers.

Additionally, it is important to maintain healthy lifestyle habits, take medications as prescribed, follow dietary recommendations as set by a nephrologist or renal dietitian, recognize emotional and mental health effects, and seek support when needed. Alport patients, and all those with chronic kidney disease, should always consult with their nephrologist regarding the use of over-the-counter (OTC) drugs, as some widely used medications can [adversely affect the kidneys or ears](#).

Because Alport syndrome is rare, many providers do not have a great deal of experience in treating this condition and its complex set of symptoms. If new symptoms or challenges arise, it's important that patients share them with their medical provider(s).

Often, informed patients are one of the most helpful resources in educating providers about their experiences, symptoms, and even about new research or clinical trial opportunities. Asking questions and motivating providers to do their own research can help ensure the needs of patients are met. Self-advocacy is key in ensuring the best possible outcomes. Alport Syndrome Foundation is dedicated to providing peer support and engagement for all individuals with Alport syndrome and their families. For connection opportunities and/or general inquiries, email ASF: [info@alportsyndrome.org](mailto:info@alportsyndrome.org).

## About Alport Syndrome Foundation

Alport syndrome is a non-profit organization led by and dedicated to the Alport community of patients and families. Our mission is to improve the lives of people living with Alport syndrome through education, advocacy, awareness, and investment in research.

### Resources for patients and families:

- Educational Materials
- Network of Support
- Patient and Family Meetings
- Webinars
- Patient Advocacy
- Legislative Advocacy
- Awareness Efforts
- Research Investment

### Resources for medical professionals:

- CME/CNE Online (free)
- Diagnosis Information
- Recommended Treatment Guidelines
- Current Research
- Clinical Trial Updates
- Genetics Guide