



Alport Syndrome Vocabulary

ACE Inhibitors + ARBs

The current standard of care for Alport syndrome is angiotensin-converting enzyme (ACE) inhibitors or angiotensin receptor blockers (ARBs). ACE and ARB medications are proven to slow the spill of protein into the urine, slowing the scarring of the kidneys. ACE and ARB medications help moderate the body's fluid balance and blood pressure.

Albuminuria

An abnormal excretion rate of albumin (a type of protein) in the urine. Albumin leakage from the kidneys must be monitored closely by your physician. Microalbuminuria refers to the excretion of 30–299 mg of albumin every 24 hours. As the amount of albumin in your urine increases, the terms “albuminuria” or “proteinuria” are used to describe the condition.

Aldosterone inhibitors

These medications block the action of aldosterone. They are sometimes prescribed to those affected by Alport syndrome as a means of controlling blood pressure.

Anemia

Condition in which the body lacks an adequate amount of healthy red blood cells. Decline in renal function of Alport patients can result in this condition.

Bilateral sensorineural hearing loss

This type of hearing loss involves hearing impairment in both ears and can be part of an Alport syndrome patient's experience, but not always. Some individuals experience unilateral hearing loss (only in one ear) or their hearing may not be affected.

Benign variant or likely benign variant

You may see one of these terms listed in your genetic testing results, indicating that a variant (change) in your COL4A3, COL4A4, and/or COL4A5 gene(s) should not cause symptoms of progressive renal disease nor any other manifestations of Alport syndrome.

Biopsy

Medical procedure in which a needle is inserted through the skin to retrieve a tissue sample. Alport patients may undergo a biopsy for initial diagnosis or for check-up during disease progression or post-transplantation.

COL4A variants

Variants (changes) in the genes COL4A3, COL4A4, and COL4A5 are what cause the tissue damage seen in Alport syndrome. Identification of these variants through genetic testing

can lead to an Alport syndrome diagnosis. Type IV collagen is found throughout the body, including in the kidneys, ears, eyes, and aorta.

Creatinine

Chemical waste product from the metabolism of creatine. A creatinine blood test is used to indicate renal function. This will be one of the most commonly referred to pieces of information on your lab results as an Alport syndrome patient. The average creatinine range for adults with normal renal function is 0.5 to 1.2 mg/dL. Creatinine levels can fluctuate due to medications, hydration, high blood pressure, and other factors.

eGFR (estimate glomerular filtration rate)

GFR is an index of overall kidney function that is calculated using your creatinine level, age, birth sex, and other factors. eGFR is presented as a number ranging from 0–120, with 90+ representing normal renal function. A GFR of 15 or less represents stage 5 kidney disease, the final stage of kidney failure when dialysis or transplantation are needed to support your kidney function.

Hematuria

The presence of blood in the urine. Microhematuria refers to blood in the urine that is only visible under a microscope. For many patients, microhematuria is the first physical symptom of Alport syndrome. Microhematuria can be detected via a simple urinalysis.

Hemodialysis (HD)

A form of dialysis in which the blood is cleaned with the assistance of a dialyzer machine. Blood exits the body into the machine where it is cleaned of waste by a special filter and then returned to the body. This procedure is most often performed at a clinic multiple times a week, but can also be done at home with adequate training and supervision.

Hyperkalemia

Condition in which the body retains excess potassium in the blood. Hyperkalemia is often a complication of kidney disease and/or a side effect of ACE inhibitor use. As ACE inhibitors are part of the current standard treatment for Alport syndrome, many patients face this issue and are required to reduce potassium intake in their daily diet and/or take certain medications to help the body excrete excess potassium.

Hypertension

Commonly known as high blood pressure, this refers to a condition of constantly elevated blood pressure. Hypertension negatively affects the kidneys and cardiovascular system over time.

Immunosuppressant medications

These medications, taken by almost all organ transplant recipients, suppress the strength of the recipient's immune system in order to prevent organ rejection.

Pathogenic variant or likely pathogenic variant

You may see one of these terms listed in your genetic testing results, indicating that a variant (change) in your COL4A3, COL4A4, and/or COL4A5 gene(s) may cause symptoms of progressive renal disease. Additional disease symptoms such as hearing loss and

specific eye conditions can also occur as type IV collagen is also found throughout the body, including in the ears, eyes, aorta, lungs, and more.

Peritoneal dialysis (PD)

With this type of dialysis, the abdominal cavity is filled with a sterile solution called dialysate, which collects waste products and excess body fluid. These are then removed from the body via an abdominal catheter. PD can be performed manually or automatically (with use of an electronic machine called a cycler). PD is generally performed in the home, with daily treatment times specific to each patient.

Phosphorus

A mineral component of your bones, teeth, and cell membranes. It is required in your body for making proteins and the growth and repair of cells and tissue. Individuals with declining kidney function are required to limit their intake of phosphorus, which, if unmoderated, can lead to bone and cardiac issues and an increased risk of death. Phosphate binders are prescribed to help reduce the body's absorption of phosphorus.

Phosphorus binders

These pills are taken with meals and snacks to prevent the body from absorbing phosphorus from the foods eaten. Binders are important for patients on dialysis and awaiting transplantation.

Potassium

A major intracellular electrolyte in the body that controls the electrical conductivity of the heart, acid-base balance, and helps regulate blood pressure. Potassium is limited in the renal diet to prevent heart abnormalities, as kidneys with declining function cannot properly regulate potassium levels in the body. Often potassium binder medications are prescribed for patients with hyperkalemia (high blood potassium) in late stages of kidney disease to help the body get rid of potassium.

Preeclampsia

Disorder of pregnancy affecting a portion of females with Alport syndrome and occurs after 20 weeks of gestation, consisting of new onset high blood pressure (> 140/90 mmHg) and proteinuria (protein in the urine).

Proteinuria

Protein detected in the urine. Proteinuria is an early indicator for diagnosis and progression of Alport syndrome.

Red blood cell (RBC) count

Found on routine lab work, an RBC count refers to the number of red blood cells in your body. Kidney disease results in a decreased RBC count. RBCs serve the body by transporting oxygen to blood cells and carbon dioxide to the lungs.

Registry

Scientifically and uniformly collected health data from individuals with the same condition, which helps advance research and clinical trials for a specific disease. All U.S. patients are strongly encouraged to join the ASF Alport Patient Registry.

Rejection

Process in which the body's immune system attacks a graft (transplanted organ such as a kidney). Immunosuppressant medication is taken to prevent the likelihood of rejection.

Tinnitus

Formal term for a distinct ringing in the ears. Subjective tinnitus means it is only internally heard by the affected individual. The ringing can be temporary or permanent and can range in intensity of volume on an individual basis. While there is no cure for tinnitus, coping mechanisms, including hearing aids, help mask the unwanted noise. Tinnitus is a challenge faced by many individuals living with hearing loss caused by Alport syndrome.

Urine protein creatinine ratio (UPCR)

This test helps assess kidney function by measuring the amount of protein and creatinine in the urine.

Variant of uncertain significance (VUS)

VUS is a genetic variant that has been identified through genetic testing but whose significance to the function or health of the patient is not known. When a genetic test result shows a VUS, it is recommended that patients ask their genetic counselor and physician(s) if any further steps are needed to support their care.

White blood cell (WBC) count

A WBC count refers to the number of white blood cells in the body. A high WBC count can be indicative of an infection, while a low WBC count can put patients at risk for not being able to effectively fight off infection.