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# **Alport Syndrome Signs and Symptoms**

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Alport Syndrome is an inherited condition, which can lead to chronic kidney disease (CKD). In CKD, a patient's kidneys progressively do not work as well as they should and do not effectively filter out wastes from the body. It is the second highest cause of CKD after polycystic kidney disease, where fluid filled sacs develop in the kidneys and impair kidney function. The syndrome affects the kidneys by attacking the glomeruli (capillary blood vessels) that help with the filtering process.

There are several symptoms and signs of Alport Syndrome which are described below.

#### Hematuria

Hematuria, which is blood in urine, is a key symptom of the disease. There can be evidence of this in patients from their childhood or as a young adult. It can often be identified at a time when the patient has some other mild illness, such as a cough or cold. Hematuria can be present as trace amounts, which are barely visible or in higher concentration, making the urine pink, red or dark in colour.

When it is detected in a young patient, it tends not to be constant but is present when triggered by other health issues.

#### **Proteinuria**

Another key symptom of the disease is proteinuria, which is protein in the blood. With this, protein which is present in the blood passes through into the urine. This should not normally happen and is a sign that the kidneys are not functioning well. There are no major visible signs apart from possible frothing of the urine. Proteinuria is usually detected with a dipstick or laboratory tests.

With CKD, the most common protein that medical staff find in the patient's urine is albumin. A high rate of protein deposits in the urine, such as 2-3 g a day is a clear warning sign.

Patients can reach chronic renal failure by the time they are 25 years old. However, if they do receive a transplant during the progression of their disease and the patient may no longer have of the symptoms of Alport syndrome. There can also be a chance that the kidney is rejected.

#### **Blood Pressure**

Alongside the other symptoms of kidney failure, the patient can also develop high blood pressure. It can be caused by extra fluid circulating in the blood vessels that causes them to stretch and become less effective or by vessels that are not as flexible. High blood pressure can damage the blood vessels in the kidneys so that they are not removing wastes from the body. Rises in blood pressure in patients with Alport Syndrome can often be detected by the time they reach the age of 15 years.

### **Eyesight Symptoms**

The shape of the eye lens can change over a period of time with Alport syndrome. It may gradually become coneshaped resulting in short-sightedness (myopia). In extreme cases, the patient may need to have their eye lenses replaced.

Another symptom is abnormal coloration of the tissues in the retina. This can present as white flecks near the macula and periphery of the retina. However, the patient's vision is not affected with this.

## **Hearing Loss**

Hearing problems usually manifest themselves in the patient as sensorineural hearing loss, which is not present at birth but develops later in childhood or early in adolescence. Hearing loss can become more obvious in some patients around the age of 10.

The hair cells in the cochlea or the nerves involved in hearing are damaged in this type of hearing loss. With the condition, the person is less likely to be able to hear high tones. The hearing loss continually progresses to cause

significant damage that needs to be corrected with a hearing aid.

Reviewed by Yolanda Smith, BPharm

#### References

- US National Library of Medicine on Alport Syndrome: <a href="https://ghr.nlm.nih.gov/condition/alport-syndrome#synonyms">https://ghr.nlm.nih.gov/condition/alport-syndrome#synonyms</a>
- Kidney Research UK on Alport Syndrom: <a href="https://www.kidneyresearchuk.org/health-information/alports-syndrome">https://www.kidneyresearchuk.org/health-information/alports-syndrome</a>
- National Institute of Diabetes and Digestive Kidney Diseases on hematuria: <a href="https://www.niddk.nih.gov/health-information/health-topics/urologic-disease/hematuria-blood-in-the-urine/Pages/facts.aspx">https://www.niddk.nih.gov/health-information/health-topics/urologic-disease/hematuria-blood-in-the-urine/Pages/facts.aspx</a>
- Kidney Research UK on proteinuria: https://www.kidneyresearchuk.org/health-information/proteinuria
- Hearing Link on sensorineural hearing loss: <a href="http://www.hearinglink.org/your-hearing/types-causes-of-hearing-loss/">http://www.hearinglink.org/your-hearing/types-causes-of-hearing-loss/</a>/

#### **Further Reading**

- What is Alport Syndrome?
- How is Alport Syndrome Diagnosed?
- How is Alport Syndrome Treated?
- Alport Syndrome Genetics and Inheritance
- Alport Syndrome Pathophysiology

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