ALAGILLE SYNDROME (ALGS)

WHAT IS ALGS?

ALGS is a rare cholestatic liver and multi-organ syndrome that impacts more than 7,000 people in the U.S.[1]

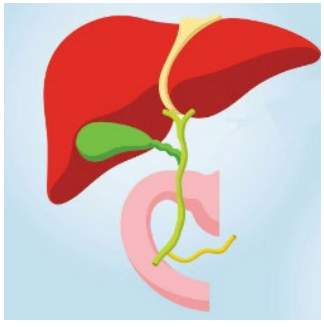
Almost 95% of ALGS cases are caused by changes in the JAG1 gene.[2]

60%

of people living with ALGS have no family history of the condition.[3]

ALGS and the liver

Bile is a liquid produced in the liver that helps rid the body of toxins. People with ALGS have too few bile ducts, causing bile to accumulate and ultimately lead to liver disease.



ALGS leads to the inability to absorb essential nutrients in the liver, causing deficiencies that can interfere with normal development and have other potentially serious consequences.

HOW IS IT DIAGNOSED?

ALGS is different from person to person.

To be diagnosed with ALGS, a person must have a reduction in the number of bile ducts, in addition to at least three of five major diagnostic criteria:[4]

- Cholestasis of the liver
- Characteristic facial features
- Vertebral abnormalities
- Eye abnormalities
- Heart defects

Genetic testing can also confirm diagnosis, if the JAG1 or NOTCH2 mutation is present, but is not necessary. Family history may also be a key indicator to confirm a diagnosis.[2]

Although the disease can emerge at any time, the symptoms often appear early in life and can cause delayed or slowed growth. Neonatal cholestasis affects 85% of people with ALGS, and 74% of those deal with the most difficult daily symptom of ALGS, pruritus, otherwise known as itching.[3]

ALGS can lead to serious health complications, related to the:

HEART

LIVER

KIDNEYS

BLOOD VESSELS

LIVING WITH ALGS

Early intervention can help preserve liver health.

The symptoms of ALGS vary widely and can include yellowing of skin and eyes, pale colored stools, itching, vitamin deficiencies, short stature, delayed growth, and heart murmurs.

For more support information and resources, visit:
Alagille Syndrome Alliance

PRURITUS

Many people experience chronic, severe itching (pruritus) as the most difficult day-to-day syndrome. This can make it very hard to sleep, engage in daily routines, focus in school or at work, cause physical discomfort or pain, and lead to bruising and cuts, especially for young children.

Treatments focus on alleviating the impact of symptoms, including reducing the amount of bile, addressing related conditions, or compensating for vitamins and nutrients. In severe cases, a liver transplant may be required.

References

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4. https://www.chop.edu/conditions-diseases/alagille-syndrome