Alagille Syndrome: A Rare Cholestatic Liver Disease

Alagille syndrome (ALGS) is a rare genetic disorder caused by abnormalities in bile ducts which can lead to progressive liver disease.

Bile ducts carry bile (which helps to digest fats) from the liver to the gallbladder and small intestine. Malformed bile ducts cause the accumulation of bile acids in the liver (cholestasis) which leads to scarring and prevents the liver from working properly.[1]

Signs of ALGS typically begin by age 3 and may include[2]:

- Yellow skin or eyes, AKA jaundice
- Itchy skin, AKA pruritus
- Stunted growth
- Heart murmur
- Changes in size or shape of blood vessels
- Enlarged spleen
- Skin lesions
- White ring on the cornea
- Kidney disease
- Spinal growth changes
- Hard, yellow skin bumps, AKA xanthomas

Additional organs affected by ALGS include[2]:

- Liver
- Heart
- Kidneys
- Central nervous system

Children with ALGS experience[3]:

- Severe, unrelenting itch that is not relieved by scratching, also known as pruritus
- Disruptions in sleep and mood, resulting in impaired quality of life

Diagnosis includes[2]:

- Genetic testing
- Heart and blood vessel tests
- Eye exams
- Spine X-ray
- Abdominal ultrasound
- Kidney function tests
- Liver biopsy

The current treatment options for ALGS can include special diets, nutrition supplements and, in cases where the condition manifests into liver disease, a liver transplant[4]

There are fewer than **3,000** children with ALGS in the United States and Europe, respectively[5]

In patients who do not receive a liver transplant,

75% have continuous scratching, with

32% having destruction of skin, bleeding, or scarring[5]

References:

1. ghr.nlm.nih.gov/condition/alagille-syndrome **2.** hopkinsmedicine.org/health/conditions-and-diseases/alagille-syndrome **3.** ncbi.nlm.nih.gov/pmc/articles/PMC6110620/ **4.** ssmhealth.com/cardinal-glennon/pediatric-transplant/pediatric-liver-transplant/alagille-syndrome **5.** Mirum Pharma market research