ALAGILLE SYNDROME:
A RARE 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 impact the functioning of several organs and cause the accumulation of bile acids in the liver (cholestasis) which leads to inflammation and injury, and prevents the liver from working properly.[1]

Signs of ALGS typically begin during infancy and symptoms attributed to cholestasis include:[2]

- Yellow skin or eyes *AKA jaundice*
- Itchy skin *AKA pruritus*
- Stunted growth
- Disfiguring cholesterol deposits under the skin *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

Approximately **1 in every 30,000** children are born with ALGS in the United States and Europe[1,5]

Pruritus affects up to **88%** of ALGS patients.[3]

Only **24%** of ALGS patients make it to adulthood with their native liver.[6]

Management options can include special diets, nutrition supplements, antipruritic agents, and, in some cases where the condition manifests into liver disease, a liver transplant.[4]

References:

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