Alagille Syndrome

National Cancer Institute

Source

National Cancer Institute. Alagille Syndrome. NCI Thesaurus. Code C35139.

An autosomal dominant genetic syndrome caused by mutations in the JAG1 gene. It is characterized by cholestatic jaundice in infancy, hepatosplenomegaly, distinctive facial features (prominent forehead, elongated nose, and pointed chin), cardiac murmurs, bone malformations, and sometimes mild mental retardation.