JAG1
Alagille Syndrome

Mode of Inheritance	• **Autosomal dominant** • **Variable expressivity and incomplete penetrance**
Renal Phenotype	• **Variable, including: hypodysplasia, increased echogenicity, medullary cystic disease, vesicoureteral reflux, renal artery stenosis, and other structural anomalies**
Extra-renal Manifestations	• **Characteristic facies: broad forehead, triangular face** • **Eye disease (most common is posterior embryotoxon)** • **Cardiac defects** • **Liver disease (e.g. cholestasis, biliary atresia)** • **Vertebral anomalies (e.g. butterfly vertebrae, hemivertebrae)** • **Vascular disease (e.g. moya moya, coarctation of the aorta)** • **Increased risk for hepatocellular carcinoma and papillary thyroid carcinoma** • **Mild developmental delay or learning disabilities**
Pre-Transplant Management	• **Screening and management of extra-renal manifestations**
Transplant Considerations	• **Careful screening of potential living related donors (inherited in an autosomal dominant manner)** • **Consideration of a combined liver-kidney transplant**
Post-Transplant Management	• **Low risk of disease recurrence**

OMIM: https://www.omim.org/entry/118450