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Hypoplastic pancreas-intestinal atresiahypoplastic gallbladder syndrome

INSFRM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base.

Hypoplastic pancreas-intestinal atresia-hypoplastic gallbladder syndrome.

ORPHA:293864

Hypoplastic pancreas-intestinal atresia-hypoplastic gallbladder syndrome is a rare, potentially fatal, genetic, visceral malformation syndrome characterized by neonatal diabetes, hypoplastic or annular pancreas, duodenal and jejunal atresia, as well as gallbladder aplasia or hypoplasia. Patients typically present intrauterine growth restriction, failure to thrive, malnutrition, intestinal malrotation, malabsorption, conjugated hyperbilirubinemia, acholia and infections. Cardiac anomalies may also be associated.

Qeios ID: M6LWI6 · https://doi.org/10.32388/M6LWI6