# Belgian Genetic Tests database

## DISEASE:
## Isolated neonatal sclerosing cholangitis

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| **NAME:** | Isolated neonatal sclerosing cholangitis |
| **DESCRIPTION:** | Isolated neonatal sclerosing cholangitis is a rare, genetic, biliary tract disease characterized by severe neonatal-onset cholangiopathy with patent bile ducts and absence of ichthyosiform skin lesions. Patients present with jaundice, acholic stools, hepatosplenomegaly and high serum gamma-glutamyltransferase activity. Liver histology shows portal fibrosis, ductular proliferation, hepatocellular metallothionein deposits, and intralobular bile-pigment accumulations. Some patients may also have renal disease. |
| **ORPHACODE:** | 480556 |
| **XREF(S):** | Orphanet |
| **ANALYTE(S):** | DCDC2 |
| **CREATED:** | 13 May 2019 - 01:02 |
| **CHANGED:** | 22 Jun 2023 - 16:14 |

# RELATED CONTENT

## Related Genetic Tests

- Cholestasis (gene panel)
- Ciliopathy (gene panel)
- Ciliopathy / polycystic kidney and liver diseases / ADTKD/ nephronophtisis / Bardet-Biedl syndromes and kidney cancers (gene panel)
- Hepatorenal disorders (gene panel)

## Related Laboratories

- Centre de Génétique Médicale UCL
- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Medische Genetica - UZ Gent

## Related Analytes

- doublecortin domain containing 2

## Related Gene Panels

- Cholestasis (40 genes) - UCL
- Ciliopathy (120 genes) - UGent
- Ciliopathy, polycystic kidney and liver diseases, ADTKD, nephronophtisis, Bardet-Biedl syndromes and kidney cancers (146 genes) - IPG