# DISEASE:
# Congenital bile acid synthesis defect type 1

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| **NAME:** | Congenital bile acid synthesis defect type 1 |
| **DESCRIPTION:** | Congenital bile acid synthesis defect type 1 (BAS defect type 1) is the most common anomaly of bile acid synthesis (see this term) characterized by variable manifestations of progressive cholestatic liver disease, and fat malabsorption. |
| **ORPHACODE:** | 79301 |
| **SYNONYMS:** | 3-beta-hydroxy-delta-5-C27-steroid oxidoreductase deficiency<br>BASD1 |
| **XREF(S):** | Orphanet<br>MeSH<br>OMIM<br>ICD-10 |
| **ANALYTE(S):** | HSD3B7 |
| **CREATED:** | 13 May 2019 - 01:02 |
| **CHANGED:** | 22 Jun 2023 - 16:14 |

# RELATED CONTENT

## Related Genetic Tests

- Bile Acid Synthesis Congenital Defect (gene panel)
- Cholestasis (gene panel)

## Related Laboratories

- Centre de Génétique Médicale UCL

## Related Analytes

- hydroxy-delta-5-steroid dehydrogenase, 3 beta- and steroid delta-isomerase 7

## Related Gene Panels

- Cholestasis (40 genes) - UCL