DISEASE:
Neonatal ichthyosis-sclerosing cholangitis syndrome

NAME:	Neonatal ichthyosis-sclerosing cholangitis syndrome
DESCRIPTION:	Neonatal ichthyosis-sclerosing cholangitis (NISCH syndrome) is a very rare complex ichthyosis syndrome characterized by scalp hypotrichosis, scarring alopecia, ichthyosis and sclerosing cholangitis.
ORPHACODE:	59303
SYNONYMS:	IHSC Ichthyosis-hypotrichosis-sclerosing cholangitis syndrome NISCH syndrome
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	CLDN1
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Genetic Tests

- Cholestasis (gene panel)
- Ichthyosis (gene panel)

Related Laboratories

- Centre de Génétique Médicale UCL
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- claudin 1

Related Gene Panels

- Cholestasis (40 genes) - UCL
- Ichthyosis and erythroderma (98 genes) - KUL

Source URL: http://gentest.healthdata.be/disease/2829