DISEASE:
Sialuria

NAME:	Sialuria
DESCRIPTION:	An extremely rare metabolic disorder described in fewer than 10 patients to date and characterized by variable signs and symptoms, mostly in infancy, including transient failure to thrive, slightly prolonged neonatal jaundice, equivocal or mild hepatomegaly, microcytic anemia, frequent upper respiratory infections, gastroenteritis, dehydration and flat and coarse facies. Learning difficulties and seizures may occur in childhood.
ORPHACODE:	3166
SYNONYMS:	Sialuria, French type
XREF(S):	Orphanet OMIM MeSH MedDRA ICD-10
ANALYTE(S):	GNE
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

Source URL: http://gentest.healthdata.be/disease/406

RELATED CONTENT

Related Genetic Tests

- Congenital disorders of glycosylation (79 genes)
- Trombosis - Hemostasis (gene panel)

Related Laboratories

- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- glucosamine (UDP-N-acetyl)-2-epimerase/N-acetylmannosamine kinase

Related Gene Panels

- Congenital disorders of glycosylation (79 genes) - KUL
- Trombosis - Hemostasis (107 genes) - KUL