DISEASE:
Gaucher disease type 2

NAME:	Gaucher disease type 2
DESCRIPTION:	Gaucher disease type 2 is the acute neurological form of Gaucher disease (GD; see this term). It is characterized by early-onset and severe neurological involvement of the brainstem, associated with an organomegaly and generally leading to death before the age of 2.
ORPHACODE:	77260
SYNONYMS:	Acute neuronopathic Gaucher disease Infantile cerebral Gaucher disease
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	GBA1
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Genetic Tests

- Enzymatic dosage Gaucher disease
- Gaucher disease diagnostic (GBA gene hot spot mutations - p.Asn409Ser; p.Leu483Pro; c.84dupG; c.115+1G>A;)
- Gaucher disease diagnostic (GBA gene sequencing)
- Neurodegeneration (gene panel)

Related Laboratories

- Centre de Génétique-Institut de Pathologie et de Génétique (IPG)
- Centrum Medische Genetica - UZ Brussel VUB

Related Analytes

- glucosylceramidase beta 1

Related Gene Panels

- Neurodegeneration (99 genes) - IPG

Source URL: http://gentest.healthdata.be/disease/2899