Belgian Genetic Tests database

DISEASE:
Tay-Sachs disease, B variant, infantile form

NAME:	Tay-Sachs disease, B variant, infantile form
ORPHACODE:	309178
SYNONYMS:	GM2 gangliosidosis, B variant, infantile form Hexosaminidase A deficiency, infantile form
XREF(S):	Orphanet ICD-10
ANALYTE(S):	HEXA
CREATED:	13 May 2019 - 01:02
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Source URL: http://gentest.healthdata.be/disease/2089

RELATED CONTENT

Related Genetic Tests

- GM2-gangliosidosis / Tay-Sachs syndrome diagnostic (HEXA gene hot spot mutations - c.1274_1277dupTATC, c.1421+1G>C and c.805G>A (p.Gly269Ser))
- Lysosomal Storage Disease (gene panel)
- Tay Sachs disease (hot spot mutations - c.1274_1277dupTATC, c.1421+1G>C and c.805G>A (p.Gly269Ser))

Related Laboratories

- Centrum Medische Genetica - UZ Antwerpen
- Centrum Medische Genetica - UZ Brussel VUB

Related Analytes

- hexosaminidase subunit alpha

Related Gene Panels

- Lysomal Storage (64 genes) - VUB