

**DISEASE:**

**Tay-Sachs disease, B variant, infantile form**

<b>NAME:</b>	Tay-Sachs disease, B variant, infantile form
<b>ORPHACODE:</b>	309178
<b>SYNOMYS:</b>	GM2 gangliosidosis, B variant, infantile form Hexosaminidase A deficiency, infantile form
<b>XREF(S):</b>	<a href="#">Orphanet</a> <a href="#">ICD-10</a>
<b>ANALYTE(S):</b>	<a href="#">HEXA</a>
<b>CREATED:</b>	13 May 2019 - 01:02
<b>CHANGED:</b>	22 Jun 2023 - 16:14

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Source URL: <http://gentest.healthdata.be/disease/2089>

## RELATED CONTENT

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### 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

- [Lysosomal Storage \(64 genes\) - VUB](#)