GENETIC TEST:
Tay Sachs disease (hot spot mutations - c.1274_1277dupTATC, c.1421+1G>C and c.805G>A (p.Gly269Ser))

FULL NAME:	Tay Sachs disease (hot spot mutations - c.1274_1277dupTATC, c.1421+1G>C and c.805G>A (p.Gly269Ser))
TEST TYPE:	Clinical
TEST SPECIALTY:	Molecular Genetics
TEST PURPOSE:	Carrier diagnosis
SPECIMEN:	Peripheral (whole) blood on EDTA
METHOD CATEGORY:	Targeted variant analysis
METHOD TECHNIQUE:	Single nucleotide extension
RIZIV CODE:	565390-565401
TURNAROUND TIME (MAXIMUM):	1 month
CREATED:	21 Aug 2019 - 18:06
CHANGED:	22 Jun 2022 - 12:25
URL:	https://labogidsmedgen.uza.be/analyses/tay-sachs-ziekte-van

Source URL: http://gentest.healthdata.be/genetic_test/567

RELATED CONTENT

Related Diseases

- Tay-Sachs disease, B variant, adult form
- Tay-Sachs disease, B variant, infantile form
- Tay-Sachs disease, B variant, juvenile form

Related Laboratories

- Centrum Medische Genetica - UZ Antwerpen

Related Analytes

- hexosaminidase subunit alpha

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