GENETIC TEST:
GM2-gangliosidosis / Tay-Sachs syndrome diagnostic (HEXA gene hot spot mutations - c.1274_1277dupTATC, c.1421+1G>C and c.805G>A (p.Gly269Ser))

FULL NAME:	GM2-gangliosidosis / Tay-Sachs syndrome diagnostic (HEXA gene 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, Mutation confirmation, Post-natal Diagnosis, Pre-implantation genetic diagnosis, Prenatal diagnosis
SPECIMEN:	Peripheral (whole) blood on EDTA, Amniotic fluid, Chorionic villi, Cell culture
METHOD CATEGORY:	Targeted variant analysis
METHOD TECHNIQUE:	Bi-directional Sanger Sequence analysis
RIZIV CODE:	565456-565460

ACCREDITATION (ISO 15189):	2021-10-07 / 2026-06-14
EQA:	• DNA Sequencing - Sanger, • DNA Sequencing - Sanger, • DNA Sequencing - Sanger, • DNA Sequencing - Sanger, • DNA Sequencing - Sanger, • DNA Sequencing - Sanger, • DNA Sequencing - Sanger , • DNA Sequencing - Sanger
TURNAROUND TIME (MAXIMUM):	3 months (10 working days for prenatal diagnosis)
CREATED:	26 Aug 2019 - 10:01
CHANGED:	09 Mar 2023 - 15:53
URL:	https://laboguide.uzbrussel.be/laboguide#Analyses:Tay-sachs%20dragerscreening&&…

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
- Tay-Sachs disease, B1 variant

Related Laboratories

- Centrum Medische Genetica - UZ Brussel VUB

Related Analytes

- hexosaminidase subunit alpha