

GENETIC TEST:

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

FULL NAME:	Gaucher disease diagnostic (GBA gene hot spot mutations - p.Asn409Ser; p.Leu483Pro; c.84dupG; c.115+1G>A;)
TEST TYPE:	Clinical
TEST SPECIALTY:	Molecular Genetics
TEST PURPOSE:	Carrier diagnosis, Mutation confirmation, Post-natal 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

EQA:	• 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 - 09:31
CHANGED:	09 Mar 2023 - 15:53
URL:	https://laboguide.uzbrussel.be/laboguide#Analyses:Ziekte%20van%20Gaucher&&&&99...

Source URL: http://gentest.healthdata.be/genetic_test/675

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