GENETIC TEST:
Pompe disease, Glycogen storage disease II (GAA gene)

FULL NAME:	Pompe disease, Glycogen storage disease II (GAA gene)
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:	Sequence analysis: entire coding region Mutation screening and sequence analysis of selected exons
METHOD TECHNIQUE:	Bi-directional Sanger Sequence analysis
RIZIV CODE:	565471-565482
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:	27 Aug 2019 - 14:50
CHANGED:	09 Mar 2023 - 16:24
URL:	https://laboguide.uzbrussel.be/laboguide#Analyses:Ziekte%20van%20Pompe&&&&&1008…

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