GENETIC TEST:
Myoclonic epilepsy associated with ragged-red fibers (MERFF) (hot spot mutation - m.8344A>G) (1st tier)

FULL NAME:	Myoclonic epilepsy associated with ragged-red fibers (MERFF) (hot spot mutation - m.8344A>G) (1st tier)
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, Skin biopsy, Liver biopsy, Muscle biopsy, Amniotic fluid, Chorionic villi, Cell culture, Skin fibroblasts

METHOD CATEGORY:	Targeted variant analysis
METHOD TECHNIQUE:	Next Generation Sequencing (NGS)
RIZIV CODE:	565390-565401
ACCREDITATION (ISO 15189):	2021-10-07 / 2026-06-14
EQA:	• Mitochondrial disorders (including POLG), • Mitochondrial disorders (including POLG), • Mitochondrial DNA (mtDNA) Metabolic Disorders, • Mitochondrial DNA (mtDNA) Metabolic Disorders, • Mitochondrial DNA (mtDNA) Metabolic Disorders , • Mitochondrial DNA (mtDNA) Metabolic Disorders, • Mitochondrial DNA (mtDNA) Metabolic Disorders, • Mitochondrial DNA (mtDNA) Metabolic Disorders
TURNAROUND TIME (MAXIMUM):	3 months (10 working days for prenatal diagnosis)
CREATED:	07 Aug 2019 - 15:08
CHANGED:	09 Mar 2023 - 16:17
URL:	https://laboguide.uzbrussel.be/laboguide#Analyses:MERF

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