GENETIC TEST:
Leber hereditary optic neuropathy (LHON) (hot spot mutations - p.Ala53Thr in MT-ND1; p.Arg340His in MT-ND4; p.Met64Val in MT-ND6)

FULL NAME:	Leber hereditary optic neuropathy (LHON) (hot spot mutations - p.Ala53Thr in MT-ND1; p.Arg340His in MT-ND4; p.Met64Val in MT-ND6)
TEST TYPE:	Clinical
TEST SPECIALTY:	Molecular Genetics
TEST PURPOSE:	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
METHOD CATEGORY:	Targeted variant analysis

METHOD TECHNIQUE:	PCR based technique
RIZIV CODE:	565456-565460
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 - 11:43
CHANGED:	09 Mar 2023 - 15:59
URL:	https://laboguide.uzbrussel.be/laboguide#Analyses:Leber

RELATED CONTENT

Related Diseases

- Leber hereditary optic neuropathy

Related Laboratories

- Centrum Medische Genetica - UZ Brussel VUB

Related Analytes

- mitochondrially encoded NADH:ubiquinone oxidoreductase core subunit 1
- mitochondrially encoded NADH:ubiquinone oxidoreductase core subunit 4
- mitochondrially encoded NADH:ubiquinone oxidoreductase core subunit 6

Related Gene Panels

- LHON (3 genes) - VUB