GENETIC TEST:
Leigh / NARP Syndrome

FULL NAME:	Leigh / NARP Syndrome
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
METHOD CATEGORY:	Sequence analysis: entire coding region Mutation screening and sequence analysis of selected exons
METHOD TECHNIQUE:	Next Generation Sequencing (NGS)
RIZIV CODE:	565493-565504

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):	6 months
CREATED:	26 Aug 2019 - 11:36
CHANGED:	09 Mar 2023 - 16:00
URL:	https://laboguide.uzbrussel.be/laboguide#Analyses:Leigh&&&&&2897&COLLECTION&0

RELATED CONTENT

Related Diseases

- Hypertrophic cardiomyopathy with kidney anomalies due to mitochondrial DNA mutation
- Isolated cytochrome C oxidase deficiency
- Kearns-Sayre syndrome
- Leigh syndrome with cardiomyopathy
- Leigh syndrome with leukodystrophy
- Leigh syndrome with nephrotic syndrome
- MELAS
- MERRF
- Maternally-inherited diabetes and deafness
- Mitochondrial DNA-associated Leigh syndrome
- Mitochondrial DNA-related progressive external ophthalmoplegia
- Mitochondrial non-syndromic sensorineural deafness with susceptibility to aminoglycoside exposure
- NARP syndrome
- Rare mitochondrial non-syndromic sensorineural deafness

Related Laboratories

- Centrum Medische Genetica - UZ Brussel VUB

Related Analytes

- mitochondrially encoded ATP synthase membrane subunit 6
- mitochondrially encoded ATP synthase membrane subunit 8

- mitochondrially encoded cytochrome c oxidase I
- mitochondrially encoded cytochrome c oxidase II
- mitochondrially encoded cytochrome c oxidase III
- mitochondrially encoded cytochrome b
- mitochondrially encoded NADH:ubiquinone oxidoreductase core subunit 1
- mitochondrially encoded NADH:ubiquinone oxidoreductase core subunit 2
- mitochondrially encoded NADH:ubiquinone oxidoreductase core subunit 3
- mitochondrially encoded NADH:ubiquinone oxidoreductase core subunit 4
- mitochondrially encoded NADH:ubiquinone oxidoreductase core subunit 4L
- mitochondrially encoded NADH:ubiquinone oxidoreductase core subunit 5
- mitochondrially encoded NADH:ubiquinone oxidoreductase core subunit 6
- mitochondrially encoded 12S rRNA
- mitochondrially encoded 16S rRNA
- mitochondrially encoded tRNA-Ala (GCN)
- mitochondrially encoded tRNA-Cys (UGU/C)
- mitochondrially encoded tRNA-Asp (GAU/C)
- mitochondrially encoded tRNA-Glu (GAA/G)
- mitochondrially encoded tRNA-Phe (UUU/C)
- mitochondrially encoded tRNA-Gly (GGN)
- mitochondrially encoded tRNA-His (CAU/C)
- mitochondrially encoded tRNA-Ile (AUU/C)
- mitochondrially encoded tRNA-Lys (AAA/G)
- mitochondrially encoded tRNA-Leu (UUA/G) 1
- mitochondrially encoded tRNA-Leu (CUN) 2
- mitochondrially encoded tRNA-Met (AUA/G)
- mitochondrially encoded tRNA-Asn (AAU/C)
- mitochondrially encoded tRNA-Pro (CCN)
- mitochondrially encoded tRNA-Gln (CAA/G)
- mitochondrially encoded tRNA-Arg (CGN)
- mitochondrially encoded tRNA-Ser (UCN) 1
- mitochondrially encoded tRNA-Ser (AGU/C) 2
- mitochondrially encoded tRNA-Thr (ACN)
- mitochondrially encoded tRNA-Val (GUN)

- mitochondrially encoded tRNA-Trp (UGA/G)
- mitochondrially encoded tRNA-Tyr (UAU/C)

Related Gene Panels

- Leigh syndrome (mtDNA / 37 genes) - VUB
