GENETIC TEST:
Mitochondrial disorders, mitchondrial DNA based (Full sequencing of mtDNA genome)

FULL NAME:	Mitochondrial disorders, mitchondrial DNA based (Full sequencing of mtDNA genome)
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
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
TURNAROUND TIME (MAXIMUM):	6 months
CREATED:	26 Aug 2019 - 16:14
CHANGED:	28 Sep 2022 - 15:41
URL:	https://laboguide.uzbrussel.be/laboguide#Analyses:Mitochondriale%20dragers&&&&&…

RELATED CONTENT

Related Diseases

- Adult-onset chronic progressive external ophthalmoplegia with mitochondrial myopathy
- Autosomal dominant progressive external ophthalmoplegia
- Autosomal recessive progressive external ophthalmoplegia
- Isolated cytochrome C oxidase deficiency
- Kearns-Sayre syndrome
- Maternally-inherited diabetes and deafness
- Mitochondrial DNA depletion syndrome, encephalomyopathic form with methylmalonic aciduria
- Mitochondrial DNA depletion syndrome, encephalomyopathic form with renal tubulopathy
- Mitochondrial DNA depletion syndrome, encephalomyopathic form with variable craniofacial anomalies
- Mitochondrial DNA depletion syndrome, hepatocerebrorenal form
- Mitochondrial DNA depletion syndrome, myopathic form
- Mitochondrial non-syndromic sensorineural deafness with susceptibility to aminoglycoside exposure
- Pearson syndrome
- Proximal myopathy with focal depletion of mitochondria

Related Laboratories

- Centrum Medische Genetica - UZ Brussel VUB

Related Gene Panels

- mitochondrial disorders, mitochondrial DNA based / mtDNA resequencing - VUB

Source URL: http://gentest.healthdata.be/genetic_test/716