

GENETIC TEST:
Chronic progressive external ophthalmoplegia (CPEO) (Full sequencing of mtDNA genome)

FULL NAME:	Chronic progressive external ophthalmoplegia (CPEO) (Full sequencing of mtDNA genome)
TEST TYPE:	Clinical
TEST SPECIALTY:	Molecular Genetics
TEST PURPOSE:	Carrier diagnosis, Mutation confirmation, Post-natal 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 Deletion/duplication analysis

METHOD TECHNIQUE:	Next Generation Sequencing (NGS) Southern blot
RIZIV CODE:	565493-565504
EQA:	<ul style="list-style-type: none"> • 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 (10 working days for prenatal diagnosis)
CREATED:	23 Aug 2019 - 21:53
CHANGED:	01 Mar 2023 - 16:31
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