Mitochondrial Diseases - Nuclear Genes Only: Deletion/Duplication Panel

**Test Code:** MD300  
**Turnaround time:** 2 weeks  
**CPT Codes:** 81228 x1, 81404 x1

### Condition Description

Mitochondrial diseases are a group of disorders caused by mutations in either mitochondrial DNA (mtDNA) or nuclear genes (nDNA). Production of energy in mitochondria, by means of oxidative phosphorylation, strictly depends upon factors which are encoded both by the mtDNA and the nDNA. Respiratory chain complexes are formed, for the most part by subunits of nuclear origin, as are several indispensable complex-assembling proteins. Accurate replication and efficient maintenance of mtDNA are also essential for the respiratory chain to function properly.

Many metabolic processes, distinct from ATP production, are fulfilled in mitochondria: for instance, important steps of metal cation metabolism take place in the mitochondrial matrix. Furthermore, mitochondria actively fuse and divide, and move interacting with the cytoskeleton. All these functions require the expression of nDNA. Mitochondrial disorders caused by nDNA defects have been the object of increasing attention in the past few years, establishing themselves as an important and relatively prevalent group of pathologies, and challenging the relevance of disease caused by inherited mutations of mtDNA itself.

References:
- OMIM.
- GeneReviews.

### Genes

ATPAF2, BCS1L, COX10, COX15, COX4I1, COX4I2, COX6B1, FASTKD2, FOXRED1, LRPPRC, NDUFA1, NDUFA10, NDUFA11, NDUFA13, NDUFA2, NDUFA8, NDUFAF1, NDUFAF3, NDUFAF4, NDUFAF5, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS5, NDUFS6, NDUFS7, NDUFS8, NDUFS9, NDUFS10, NDUFS11, NDUFS12, NDUFS13, NDUFS14, NDUFS15, NDUFS16, NDUFS17, NDUFS18, NDUFS19, NDUFS20, NUBPL, SCO1, SCO2, SDHAF1, SDHAF2, SDHB, SDHC, SDHD, SURF1, TACO1, TMEM70, TTC19, UQCRB, UQCRQ

### Indications

This test is indicated for:
- Confirmation of a clinical diagnosis of mitochondrial diseases.

### Methodology

**Deletion/Duplication Analysis:** DNA isolated from peripheral blood is hybridized to a gene-targeted CGH array to detect deletions and duplications. The targeted CGH array has overlapping probes that cover the entire genomic region.

Please note that a "backbone" of probes across the entire genome are included on the array for analytical and quality control purposes. Rarely, off-target copy number variants causative of disease may be identified that may or may not be related to the patient's phenotype. Only known pathogenic off-target copy number variants will be reported. Off-target copy number variants of unknown clinical significance will not be reported.

**Detection**

**Deletion/Duplication Analysis:** Detection is limited to duplications and deletions. The CGH array will not detect point or intronic mutations. Results of molecular analysis must be interpreted in the context of the patient's clinical and/or biochemical phenotype.

### Specimen Requirements

Submit only 1 of the following specimen types

**Type: Whole Blood**

Specimen Requirements:

In EDTA (purple top) tube:
Infants (2 years): 3-5 ml  
Older Children & Adults: 5-10 ml.

Specimen Collection and Shipping: Ship sample at room temperature with overnight delivery.

**Type: Isolated DNA**

Specimen Requirements:

In microtainer: 10 ug

Isolation using the Qiagen™ Puregene kit for DNA extraction is recommended.

Specimen Collection and Shipping: Refrigerate until time of shipment in 100 ng/ul of TE buffer. Ship sample at room temperature with overnight delivery.

**Related Tests**

- Mitochondrial Diseases - Nuclear Genes Only: Sequencing Panel
- Mitochondrial Genome: Sequencing