Orderable - MD Mitochondrial Panel

Turnaround Time: 4-6 weeks  STAT: 4 weeks

Alternate Name(s):
- mtDNA sequencing
- LHON
- MELAS
- MERF

Specimen:
Whole blood-2 x 4 mL Lavender EDTA top Vacutainer tube

Collection Information:
Sample may be transported at room temperature. Alternative tissues may be accepted after consult with laboratory.

Reference Ranges:
See report

Interpretive Comments:
The human mitochondrial DNA (mtDNA) encodes 37 genes coding for two rRNAs, 22 tRNAs and 13 polypeptides within its 16,569 bp. The mtDNA-encoded polypeptides are all subunits of enzyme complexes of the oxidative phosphorylation system. Disease phenotypes resulting from mitochondrial mutations may appear as distinct syndromes, such as Kearns-Sayre syndrome (KSS), Leber’s Hereditary Optic Neuropathy (LHON), mitochondrial encephalomyopathy with lactic acidosis and stroke-like episodes (MELAS), chronic progressive external ophthalmoplegia (CPEO), myoclonic epilepsy with ragged-red fibers (MERRF), neurogenic weakness with ataxia and retinitis pigmentosa (NARP) or Leigh syndrome (LS). More frequently, the clinical presentation is much more heterogeneous. Some common symptoms include ptosis, external ophthalmoplegia, proximal myopathy, exercise intolerance, cardiomyopathy, sensorineural deafness, migraine, stroke-like episodes, pigmentary retinopathy,
Mitochondrial encoded: NC_012920.1: MT-TY, MT-TW, MT-TV, MT-TT, MT-TS2, MT-TS1, MT-TR, MT-TQ, MT-TP, MT-TN, MT-TM, MT-TL2, MT-TL1, MT-TK, MT-TI, MT-TH, MT-TG, MT-TF, MT-TE, MT-TD, MT-TA, MT-RNR2, MT-RNR1, MT-ND6, MT-ND5, MT-ND4L, MT-ND4, MT-ND3, MT-ND2, MT-ND1, MT-CYB, MT-CO3, MT-CO2, MT-CO1, MT-TC, MT-ATP8, MT-ATP6
Nuclear encoded: APTX(NM_0011952248.1), DGUOK(NM_080916.2), DNA2(NM_001080449.2), FBXL4(NM_001278716.1), GFER(NM_005262.2), MGME1(NM_052865.3), MPV17(NM_002437.4), OPA1(NM_130837.2), OPA3(isoformA & B: NM_001017989.2 & NM_025136.3), POLG(NM_001126131.1), POLG2(NM_007215.3), RRM2B(NM_001172477.1 & NM_015713.4), SLC25A4(NM_001151.3), SPG7(isoform1 & 2:NM_003119.2 & NM_199367.2 (x10)), SUCLA2(NM_003850.2), SUCLG1(NM_003849.3), TK2(NM_004614.4), TWNK(C10orf2)(NM_021830.4), TYMP(NM_001257989.1)

Storage and Shipment:
Samples may be stored and shipped at room temperature

Comments:
Mitochondrial encoded: NC_012920.1: MT-TY, MT-TW, MT-TV, MT-TT, MT-TS2, MT-TS1, MT-TR, MT-TQ, MT-TP, MT-TN, MT-TM, MT-TL2, MT-TL1, MT-TK, MT-TI, MT-TH, MT-TG, MT-TF, MT-TE, MT-TD, MT-TA, MT-RNR2, MT-RNR1, MT-ND6, MT-ND5, MT-ND4L, MT-ND4, MT-ND3, MT-ND2, MT-ND1, MT-CYB, MT-CO3, MT-CO2, MT-CO1, MT-TC, MT-ATP8, MT-ATP6
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Storage and Shipment:
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MITOCHONDRIAL GENOME SEQUENCING AND DEPLETIONS/INTEGRITY

PMID: 27376475, 28818680).

Test Schedule:
As required, Monday to Friday 0800-1600 hours