

Mitochondrial DNA Mutation Panel

Background

This is a panel test for the most common mitochondrial DNA point mutations associated with the mitochondrial disorders MELAS, MERRF, NARP, Leigh syndrome, and LHON. The mutation detection rate is estimated at >90% for MELAS, MERRF, and LHON. Portions of the mitochondrial genome are analyzed to determine if any of these mutations are present: A3243G, A3252G, C3256T, T3271C, T3291C, G3460A, A8344G, T8356C, G8363A, T8851C, T8993G, T8993C, G11778A, T14484C.

The cost of the panel is \$601. If a family is already known to carry a specific mutation, a test for a single specified mutation can also be ordered for \$300.

This testing does not screen for large mitochondrial deletions or duplications, which are the most common cause of Kearns-Sayre (KS) syndrome and progressive external ophthalmoplegia (PEO). If KS or PEO is suspected, a separate test for large deletions and rearrangements should be ordered. This test also does not screen for rare mitochondrial point mutations or for mutations of nuclear genes that may affect mitochondrial oxidative phosphorylation.

Indications for Testing

Diagnostic testing in symptomatic individual or at-risk relative

Ordering

1. Obtain blood sample.
2. Fill out a Clinical Lab Request - Genetics for each patient.
(available at <http://depts.washington.edu/labweb/Divisions/MolDiag/MolDiagGen/index.htm>).
3. Call Laboratory Medicine Community Services at (206)598-6066 to arrange the best method of shipment.

Sample Requirements and Specimen Handling

Whole blood - EDTA (purple top) - adults - 5 mL, children - 1-3 mL.
Samples should be received within 72 hours of collection.
Samples may be refrigerated until shipped.
Heparin (green top) tubes are not acceptable.

Test Frequency and Reporting

Performed weekly, results in 1-2 weeks

References

1. Smeitink J, van den Heuvel L, DiMauro S. The genetics and pathology of oxidative phosphorylation. *Nature reviews genetics*. 2001;2:342-352.
2. DiMauro S, Hirano M. Mitochondrial encephalomyopathies: an update. *Neuromuscular Disorders* 2005;15:276-286.