



♥ PEDIATRIC / ADULT

For your patients with neurological and multi-systemic symptoms, mitochondrial testing can help get you an answer.

BAYLOR
GENETICS

nDNA & mtDNA
Nuclear & Mitochondrial
DNA Testing

Gain additional insights into your patients with complex clinical presentations with mitochondrial testing

What is Mitochondrial testing?

Mitochondrial testing can help identify genetic changes causing mitochondrial disorders in either the nuclear DNA (nDNA) or mitochondrial DNA (mtDNA).

Mitochondrial disorders may result from genetic changes in nDNA or mtDNA. Genetic changes in nDNA may be inherited from one or both parents. Genetic changes in mtDNA are only inherited from an individual's mother. Changes in mtDNA might be new to the affected patient (de novo) or inherited from their mother who has a change in some mitochondria but not enough altered mtDNA to be affected herself.

Who should consider Mitochondrial testing?

Mitochondrial disorders can be identified at any point in a patient's life. Multiple systems are often impacted by these disorders - the systems affected and severity of symptoms can vary greatly even between other affected family members.

Common Symptoms

- Ataxia
- Stroke or stroke-like episodes
- Seizures
- Dementia
- Neuropathy
- Myopathy and cardiomyopathy
- Developmental delay
- Vision and hearing deficits
- Diabetes and other endocrine symptoms
- Gastrointestinal symptoms, such as pseudo-obstruction and dysmotility
- Laboratory abnormalities, such as elevated lactate: pyruvate ratio

Mitochondrial Testing Options

TEST CODE	TEST NAME	TEST DESCRIPTION
2055	MitoNGS SM Comprehensive mtDNA Analysis by Massively Parallel Sequencing	Next Generation Sequencing (NGS) of the entire mtDNA with heteroplasmy determination. This test will detect disorders attributed to sequencing variants, gross deletions/duplications, and multiple deletions in the mtDNA.
2086	MitomeNGS SM Nuclear DNA Panel by Massively Parallel Sequencing	This panel analyzes 164 nuclear genes associated with mitochondrial disorders using massively parallel sequencing.
2085	MitomeNGS SM Dual Genome Panel by Massively Parallel Sequencing	This test combines BCM-MitomeNGS SM (164 nuclear genes) testing and 37 mitochondrial genes involved in mitochondrial disorders using massively parallel sequencing.
1532	Whole Exome Sequencing + MitoNGS SM Comprehensive mtDNA Analysis	Whole Exome Sequencing (WES) analyzes a patient's exome –about 22,000 genes and the protein-coding regions of DNA. When combining MitoNGS SM with WES, Baylor Genetics can detect genetic alterations in both the nuclear and mitochondrial genomes.
2130	MitomeNGS SM mtDNA Depletion/Integrity Panel by Massively Parallel Sequencing	19 nuclear-encoded genes involved in the biosynthesis of the mitochondrial genome and the maintenance of mtDNA integrity are analyzed using massively parallel sequencing.
2000	MitoMet [®] Plus aCGH Analysis	This microarray contains 180,000 oligonucleotide (short DNA molecules) probes targeted to both mitochondrial and nuclear genes involved in mitochondrial and metabolic-related diseases.

For a complete list of mitochondrial tests, please visit baylorgenetics.com.