PEDIATRIC / ADULT

Complex symptoms and no diagnosis can be challenging for families and providers. Baylor Genetics offers mitochondrial testing designed to find the answer to your patient's various ailments allowing the best treatment options available.

BAYLOR GENETICS

MITO
Mitochondrial Testing

Gain insights into complex health conditions through Mitochondrial testing

Changes in hundreds of genes have been found to be associated with mitochondrial disorders.

Mitochondrial disorders often impact multiple organ systems, such as muscles, eyes, brain, or liver. These disorders, ranging from mild to severe, can be identified at any point during a patient's life. Doctors may suspect a mitochondrial disorder when three or more organ systems are involved and may recommend further testing.

Genes are sections of DNA that act like our body's instruction manual for how to grow and develop.

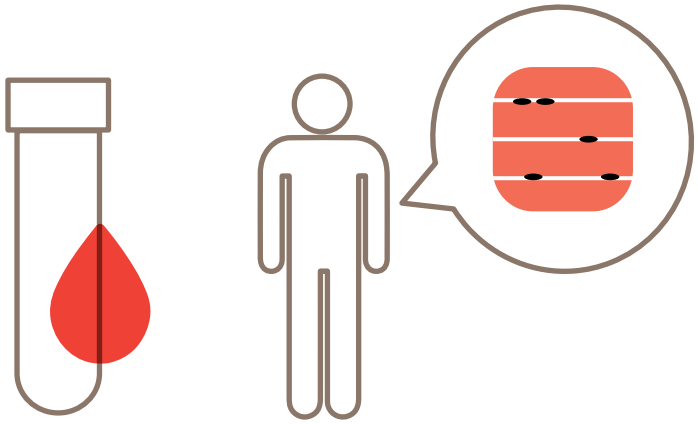
Scientists can study a person's genes and identify changes in DNA. Some of these changes can cause a gene to function improperly. Most DNA is found within the nucleus of the cell; however, a smaller amount of DNA is found within the cell's mitochondria. The genetic changes causing mitochondrial disorders can occur in either nuclear DNA (nDNA) or mitochondrial DNA (mtDNA).

Since mitochondrial disorders are genetic, they can be passed down from generation to generation. Disorders in nDNA can be inherited from the mother and/or the father. However, disorders due to defects in mtDNA can only be inherited from the mother. Some mitochondrial disorders caused by various mitochondrial deletions can occur in patients with no family history.

Common Symptoms

- ACUTE VISION LOSS IN SECOND DECADE
- GASTROINTESTINAL PSEUDO-OBSTRUCTION
- DROOPING EYELIDS (PTOSIS)
- ELEVATED LACTATE/PYRUVATE RATIO OR ALANINE/LYSINE RATIO
- EXERCISE INTOLERANCE
- HEARING LOSS, ESPECIALLY WITH DIABETES
- LIVER FAILURE
- MUSCLE WEAKNESS
- PARALYSIS OF THE MUSCLES THAT CONTROL EYE MOVEMENT
- STROKE-LIKE EPISODES

If a mitochondrial disorder is suspected based on clinical symptoms or family history, additional genetic testing may be recommended to confirm the diagnosis.



Changes in hundreds of genes have been found to be associated with mitochondrial disorders. Testing for these genetic changes can typically be performed on a blood sample. However, in some cases it's recommended to test mitochondrial DNA in affected tissue such as muscle to address issues of de novo mutation, mutation heteroplasmy, or mutation loss.

Baylor Genetics offers a wide variety of mitochondrial testing options. Choices range from deletion / duplication to next-generation sequencing with a variety of panels to choose from. Through mitochondrial testing, our team of experts focuses on finding the genetic cause of your patient's medical condition. Reaching a diagnosis can offer guidance for appropriate treatment options providing the family with greater peace of mind.

Mitochondrial and Metabolic Panels

MITOCHONDRIAL PANELS

TEST CODE	PANEL NAME
2000	MitoMet®Plus aCGH Analysis
2010	Advanced mtDNA Point Mutations and Deletions by Massively Parallel Sequencing (17 genes)
2055	Comprehensive mtDNA Analysis by Next Generation Sequencing (MitoNGS℠)
2085	Dual Genome Panel by Massively Parallel Sequencing (BCM_MitomeNGS℠)
2086	Mitome200 Nuclear Panel (164 nuclear genes)
2100	CoQ10 Deficiency Panel (5 genes)
2130	mtDNA Depletion/Integrity Panel (19 genes)
2140	Progressive External Ophthalmoplegia (PEO) Panel (10 genes)
2155	Mitochondrial Respiratory Chain Complex I Deficiency Panel (21 genes)
2160	Mitochondiral Respiratory Chain Complex II Deficiency Panel (5 genes)
2165	Mitochondiral Respiratory Chain Complex III Deficiency Panel (4 genes)
2170	Mitochondrial Respiratory Chain Complex IV Deficiency Panel (10 genes)
2175	Mitochondrial Respiratory Chain Complex V Deficiency Panel (3 genes)
2180	Mitochondrial Respiratory Chain Complex I – V Panel (43 genes)
2185	PDH & Mitochondrial Respiratory Chain Complex V Panel (9 genes)
20601	Leigh Disease Panel (82 nuclear genes)

ETC/COPY NUMBER ANALYSIS

TEST CODE	PANEL NAME
3200	Mitochondrial Respiratory Chain Enzyme Analysis (ETC) - Skeletal Muscle
3210	Mitochondrial Respiratory Chain Enzyme Analysis (ETC) - Skin Fibroblasts
3700	Mitochondrial DNA Content (qPCR) Analysis - Skeletal Muscle
3720	Mitochondrial DNA Content (qPCR) Analysis - Liver

METABOLIC PANELS

TEST CODE	PANEL NAME
2095	Fatty Acid Oxidation Deficiency Panel (20 genes)
2100	CoQ10 Deficiency Panel (5 genes)
2105	Cholestasis Panel (7 genes)
2110	UCD and Hyperammonemia Panel (8 genes)
2120	Cobalamin Metabolism Panel + Severe MTHFR Deficiency by Massively Parallel Sequencing (20 genes)
2125	Glycogen Storage Disease (GSD) Comprehensive Panel (23 genes)
2126	Glycogen Storage Disease (GSD) Muscle Panel (13 genes)
2127	Glycogen Storage Disease (GSD) Liver Panel (13 genes)
2300	Myopathy/Rhabdomyolysis Panel (25 genes)
2345	Trifunctional Protein Deficiency Panel (2 genes)
2347	Propionic Acidemia Panel (2 genes)
2349	Multiple Acyl-CoA Dehydrogenase Deficiency (MADD) Panel (3 genes)
3780	Methylmalonic Acidemia Panel (3 genes)
3782	3-Methylcrotonyl-CoA Carboxylase (3MCC) Panel (2 genes)
5095	Congenital Disorders of Glycosylation (CDG) Panel (36 genes)
5270	Proximal Urea Cycle Disorders Panel (3 genes)
5405	Hemochromatosis Panel by Sanger Sequencing (5 genes)
21700	Hyperinsulinism Panel (8 genes)
21900	Maturity Onset Diabetes of the Young (MODY) Panel (25 genes)
22100	Peroxisomal Disorders Panel (22 genes)
32870	Maple Syrup Urine Disease (MSUD) Panel (4 genes)

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

Specimen Requirements

TYPE	REQUIREMENTS	SHIPPING CONDITIONS
BLOOD	Draw blood in an EDTA (purple-top) tube(s) and send 3-5 cc (Adults/ Children) and 3-5 cc (Infant<2yrs).	Ship at room temperature in an insulated container by overnight courier.
CORD BLOOD	1-2 cc for Cord Blood. Ensure properly labeled. Also send 3 cc of maternal blood in properly labeled EDTA tube for MCC studies at no charge as needed.	Ship at room temperature in an insulated container by overnight courier.
CULTURED SKIN FIBROBLAST	Send three (3) T25 flasks at approximately 60-80% confluence.	Ship at ambient temperature in an insulated container by overnight courier.
FRESH FROZEN TISSUE	50mg for Tissue	Fresh Tissue should be flash frozen in liquid nitrogen at collection with no media added, stored at -80°C, and shipped by overnight courier on 3-5 lbs of dry ice.
LIVER	50mg for Liver	Liver should be flash frozen in liquid nitrogen at collection with no media added, stored at -80°C, and shipped by overnight courier on 3-5 lbs of dry ice.
PURIFIED DNA	Send at least 5ug of purified DNA (minimal concentration of 50ng/uL; A260/A280 of ~1.7).	Ship at room temperature in an insulated container by overnight courier. Do not heat or freeze.
SALIVA	Collected with Oragene DNA Self-Collection Kit.	Ship at room temperature in an insulated container by overnight courier. Do not heat or freeze.
SKELETAL MUSCLE	150mg for ETC 150mg for CoQ 10mg for mtDNA	Skeletal Muscle should be flash frozen in liquid nitrogen at collection with no media added, stored at -80°C, and shipped by overnight courier on 3-5 lbs of dry ice.

Please go to baylorgenetics.com or call the lab at 1.800.411.4363 to discuss the sample requirements.

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