Mitochondrial Diseases

Mitochondria are small structures that make energy in most of the cells in the body by combining oxygen with the nutrients we take in from food. When mitochondria do not work right, our cells and organs do not have enough energy to work correctly and the unused oxygen and nutrients cause damage to the body. The heart, brain, and muscles need the most energy to work and are the most affected by mitochondrial disease. The most common symptoms include heart defects, cardiomyopathy, seizures, headaches, developmental delay, muscle weakness, and cramping. Some people with mitochondrial disease may also develop symptoms such as diabetes, kidney problems, low blood sugar, liver problems, hearing loss, and vision problems. Mitochondrial disease is variable; some people may have many of these symptoms while others may have only one of these problems.

WHAT CAUSES MITOCHONDRIAL DISEASES?

Mitochondrial diseases are caused by pathogenic (disease-causing) variants in genes that are needed to build mitochondria and keep them functioning. These variants can happen in two types of genes - those in the nucleus of our cells, where most of our genetic material (DNA) is housed, or in special genes that are in the mitochondria themselves. Since there are many genes involved in making sure mitochondria work, these conditions can be passed through families in many ways and members of the same family may experience different symptoms.

ASSOCIATED CONDITIONS

Mitochondrial disease may cause problems with one organ or may impact many parts of the body. Some patterns of symptoms have been given more specific names. Several examples of conditions associated with mitochondrial disease include but are not limited to:

- **Leigh Syndrome**
- **MERRF (myoclinic epilepsy with ragged-red fibers)**
- **MELAS (mitochondrial encephalomyopathy with lactic acidosis and stroke-like episodes)**
- **CPEO (Chronic progressive external ophthalmoplegia)**
- **LHON (Leber hereditary optic neuropathy)**
- **NARP (Neurogenic weakness with ataxia and retinitis pigmentosa)**
- **Pearson syndrome**
- **KSS (Kearns-Sayre syndrome)**

WHO IS THIS TEST FOR?

This panel may be appropriate for anyone who has a personal or family history of symptoms of mitochondrial disease, including muscle weakness or cramping, cardiomyopathy, eye problems (ptosis, external ophthalmoplegia, optic atrophy), hearing loss, and/or deafness.

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BENEFITS OF GENETIC TESTING

Genetic testing for mitochondrial diseases can:

- Establish or confirm the appropriate diagnosis
- Identify risks for additional health-related symptoms
- Assist in modifying lifestyle changes, including diet and exercise
- Result in more personalized treatment and symptom management
- Inform family members about their own risk factors
- Connect patients to relevant resources & support
- Provide options for family planning

RELATED PANELS

Visit Fulgent website for most up-to-date list

Mito Comprehensive

Mitochondrial Genome NGS Panel

Nuclear Mito-NGS Panel

mtDNA Depletion Syndrome NGS Panel

Comprehensive Metabolism NGS Panel

TEST SPECIFICATIONS

Acceptable Sample Requirements

- Blood, two 4-mL EDTA tubes, lavender top
- Extracted DNA, 3 µg in EB buffer
- Buccal swab or saliva

Turnaround Time 3-5 weeks

Coverage ≥96% at 20x

Reporting

VUS, likely pathogenic, and pathogenic variants

Customization

Customizable gene list, VUS opt-out

GET CONNECTED

United Mitochondrial Disease Foundation - umdf.org

Muscular Dystrophy Association - mda.org/disease/mitochondrial-myopathies

REFERENCES

- Chinnery PF. Primary Mitochondrial Disorders Overview. 2000 Jun 8 [Updated 2021 Jul 29]. In: Adam MP, Ardinger HH, Pagon RA, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2021. Available from: https://www.ncbi.nlm.nih.gov/books/NBK1224/
- MedlinePlus [Internet]. Bethesda (MD): National Library of Medicine (US); [updated 2021 Nov 3]. Mitochondrial Diseases; [updated 2021 Oct 27; reviewed 2016 Aug 23; cited 2021 Nov 8]; Available from: https://medlineplus.gov/mitochondrialdiseases.html#.

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A Patient's Guide to Genetic Testing

What does a genetic test check for?

Genetic testing checks the order of one's DNA sequence (coded by the letters A, T, G, C) in specific genes linked to genetic conditions. Letters that were added, missing, or changed, are known as variants and can sometimes be harmful to one's health, increasing the risk for a genetic condition.

What are the potential results?

There are three possible results from genetic testing:

Positive

A pathogenic/likely pathogenic variant is detected in one's DNA. This type of variant is known to increase one's risk of a genetic condition. Identifying the specific gene involved can help confirm a diagnosis, inform screening and management, and reveal risk factors for an individual and/or their family.

Negative

No variation known to be associated with a genetic condition was detected in one's DNA . While a result may not show an increased risk for the condition(s) tested for, one can still be at risk for disease, especially if there is a family history.

Variant of Uncertain Significance (VUS)

A variant was detected in one's DNA, however, not enough information is known about this variant to determine whether or not it is known cause the condition(s) tested for. More research is needed to better understand this variant.

What about family members?

Children, siblings, and parents of individuals who have a variant(s) identified in genetic testing could carry the same variant(s) and benefit from testing. Regardless of whether or not a variant was identified, individuals can still be at an increased risk for a genetic condition, especially with a family history.

Do genetic test results affect health insurance or employment?

No, the Genetic Information Nondiscrimination Act (GINA) was signed into law in 2008.
It protects individuals from discrimination by an employer or a health insurance company based on genetic testing results and genetic information. GINA does not protect against life and disability insurance discrimination. For more information on GINA, go to www.ginahelp.org.

Where can I learn more?

Medline Plus/Genetics Home Reference - medlineplus.gov/genetics/understanding
National Society of Genetic Counselors - nsgc.org
Fulgent Genetics - fulgentgenetics.com/products/carrierscreening/learning.html

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