

MITOXPANDED PANEL

Mitochondrial disorders are a clinically and genetically heterogeneous group of disorders caused by dysfunction of the mitochondrial respiratory chain. Mitochondrial disorders may affect a single organ, but many involve multiple organ systems particularly those that are highly dependent on aerobic metabolism (brain, skeletal muscle, heart, kidney and endocrine system).

Features:

- A comprehensive and dynamic gene list. Currently, the test offers sequencing analysis of approximately 1800 nuclear genes, which includes:
 - Genes currently associated with nuclear mitochondrial disorders
 - Genes associated with phenotypes that overlap significantly with mitochondrial disorders
- Utilizes a Trio approach (includes concurrent analysis of the affected proband and both parents) in order to increase the likelihood of identifying a definitive genetic explanation for mitochondrial disease
- This test is also available to order with concurrent mtDNA testing, as needed. This testing option is available using test code 554
- TAT: 6 weeks

Why is this Test Useful?

More than a thousand nuclear genes have been associated with primary mitochondrial disorders or disorders that have a significant phenotypic overlap with primary mitochondrial disorders. With whole exome sequencing, this list of genes continues to grow, underscoring the need for a broad and flexible molecular test such as MitoXpanded.

Reporting Strategies:

- Phenotype driven test reports, which will be more succinct than our current nuclear testing options
- Reported Variants:
 - Pathogenic or likely pathogenic variants in genes associated with the patient's phenotype
 - Variants of uncertain significance (VUS) in genes that are possibly associated with patient's phenotype
- Incidental findings will not be reported

Possible Outcomes of Genetic Testing

There are four possible outcomes of genetic testing: positive (pathogenic variant), likely pathogenic variant, variant of uncertain significance (VUS), and negative. Genetic counseling is recommended prior to genetic testing to understand the benefits and limitations of testing.

A **positive** result indicates that a pathogenic variant (harmful change) was identified in a specific gene and the risk to develop a particular disease is increased.

A **likely pathogenic variant** result indicates that there is a change, or genetic variant, in a specific gene that is likely pathogenic (harmful). For this type of result, there is significant, but not conclusive, evidence that there is a risk to develop a particular disease.

A **variant of uncertain significance** (VUS) indicates that the effect of the variant cannot be clearly established. A VUS is a change in a gene that has never been seen before or because of conflicting or incomplete information in the medical literature, its association with disease is unknown.

A **negative** result means that no reportable changes were identified.

Sample Submission

Genetic testing can be performed on blood, oral rinse or extracted DNA samples. GeneDx test kits are available to ordering providers, and include sample collection items (such as mouthwash, collection tubes), the necessary sample submission paperwork, and a self-addressed return shipping label. Additionally, all test requisition forms are available for download from the GeneDx website: www.genedx.com/forms

Please note that all testing must be performed under the guidance of a healthcare provider. For more information on the sample submission process, please visit our website: www.genedx.com/supplies or email us at: zebras@genedx.com.

Testing must be performed under the guidance of a healthcare provider.

1. Download the Mitochondrial/Metabolic test requisition form from the GeneDx website: www.genedx.com/forms
2. Complete all the forms with required information
3. Ship completed forms along with patient sample to the following address:

**Accessions
GeneDx
207 Perry Parkway
Gaithersburg, MD 20877**

4. Accepted sample types include blood, buccal swabs, oral rinse or extracted DNA.

To place an order for shipping kits, please visit our website: genedx.com/supplies or email us at: zebras@genedx.com



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