



We are committed to working with the entire mitochondrial disease community to increase access to genetic testing, accelerate diagnosis, enhance medical management, and advance clinical research to bring more therapies to patients.

If you are a patient

Note: Please contact a mitochondrial disease care provider, who can determine eligibility and order this test, if appropriate. For more information, assistance in finding a provider, and general support, contact info@umdf.org



Mitochondrial disease is estimated to affect 1 in every 5,000 people.

UNITED MITOCHONDRIAL DISEASE FOUNDATION

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NO-COST GENETIC TESTING FOR SUSPECTED MITOCHONDRIAL DISEASE

A no-cost next generation sequencing of select nuclear DNA mutations and full mitochondrial DNA sequencing for people with suspected mitochondrial disease.

Supported by:



The patient journey to diagnosis of a Primary Mitochondrial Disease (PMD) can be long and frustrating and sometimes ends without a genetic confirmation of a specific mitochondrial disease.

This program is designed to provide clinicians with an opportunity to order no-cost genetic testing for patients who have a suspected PMD based on expert evaluation, who may not otherwise be able to receive a genetic test (due to cost or insurance coverage). Our aim is to facilitate a diagnosis to aid in optimal clinical care and potential participation in clinical trials.

Eligibility Criteria:

- Clinician and patient must be based in the USA.
- Patient must have a clinical presentation consistent with PMD.
- Patient has not had whole exome or whole genome sequencing in the past.
- Patient does not have insurance or their insurance does not cover costs for such tests.

We ask that you ensure all insurance options have been exhausted prior to submitting a test request. Since many other disorders have clinical symptoms that overlap with PMD, and testing capacity is limited, please use discretion before ordering.

About The Test

MNG Laboratories

Comprehensive Cellular Energetics Defects

(320 Gene NGS Panel and Copy Number Analysis + Full mtDNA Sequencing)

Specimen: Blood/Buccal Cells

How To Get Started

1. Visit umdf.org/genetic-testing-clinicians/
2. Download and review the test request form (TRF) and eligibility criteria to determine the patient meets all requirements.
3. Order the test kit, collect the specimen and return the kit to the lab -- along with a fully completed TRF and patient's consent form.
4. The genetic test will be processed by MNG Labs. Expect results within a month of full submission.

Mitochondrial Disease:

A Complex, Multi-Systemic Collection of Rare Diseases with a combination of any of these symptoms:



Brain

developmental delays, dementia, migraines, autistic features, seizure, stroke, atypical cerebral palsy, learning disabilities



Nerves

fainting, zero reflexes, heat/cold intolerance, pain



Kidneys

renal tube failure



Liver

low blood sugar, liver failure



Eyes

vision loss, ptosis, optic atrophy, strabismus, ophthalmoplegia, retinitis pigmentosa



Muscles

weakness/failure, cramping, reflux, vomiting, constipation, diarrhea, hypotonia, dystonia



Pancreas

diabetes, pancreatic failure, parathyroid failure



Heart

defects, blockage, cardiomyopathy



Ears

hearing loss



Systemic

failure to gain weight, fatigue, short stature, unexplained vomiting, respiratory problems