CENTOGENE
THE RARE DISEASE COMPANY

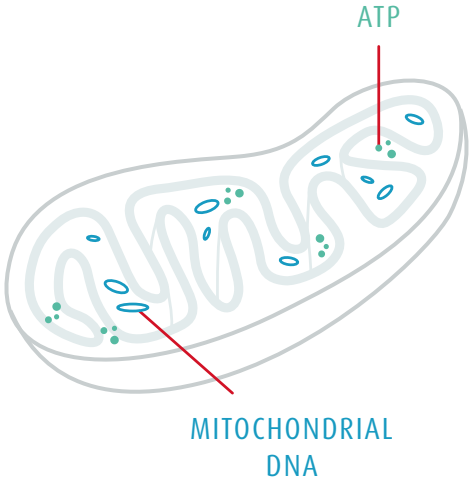
CentoMito®

MITOCHONDRIAL DISEASES

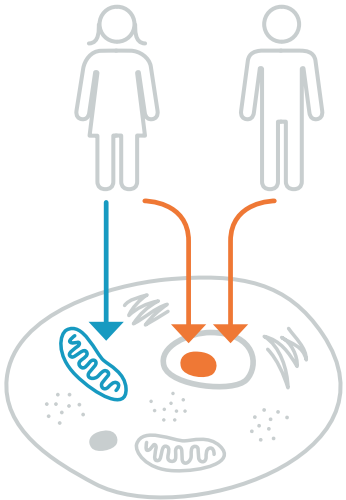
What are mitochondria?

Every cell in the body has small structures called mitochondria that act as "powerhouses". Mitochondria are responsible for producing almost 90% of the energy (in the form of ATP molecules) needed by cells via respiration and also regulate cellular metabolism.

Cells contain hundreds of mitochondria, and each mitochondrion contains several copies of mitochondrial DNA, which contains the information for making mitochondrial proteins. Cells that require more energy tend to have more mitochondria (eg. brain, muscles, heart).



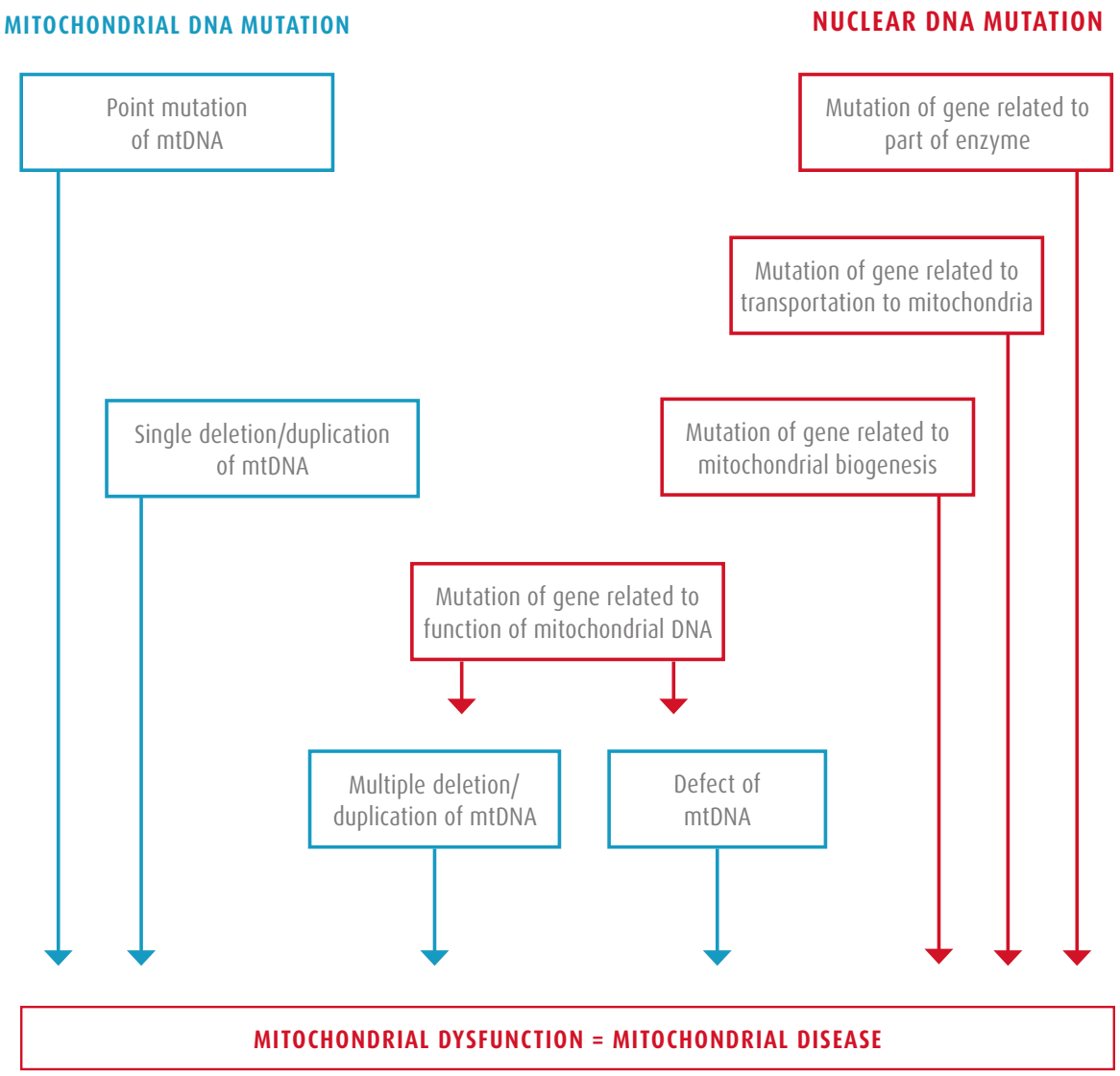
Mitochondrial disease and inheritance



Mitochondrial disorders affect 1 in 4,000 people and occur when mitochondria fail to produce enough energy for the cell's requirements. Mitochondrial failure causes cell death and can turn in organ failure if many of the cells in any organ die. They can be caused by genetic mutations in the mitochondrial genome or in nuclear genes encoding proteins that function inside mitochondria. When different mutations are present in all mitochondria in the same organism, this state is called 'homoplasmy' and when it is present in some mitochondria but not others, it's called 'heteroplasmy'. We use complex new techniques able to identify levels of heteroplasmy in the patients affected with mitochondrial disorders.

Mitochondrial mutations are only transmitted through the mother while nuclear gene mutations are inherited from either parent or both parents. Changes in either the mitochondrial or nuclear DNA instructions for making proteins can lead to very similar disease symptoms. Diseases caused by nuclear genes are predominantly inherited from both parents in an autosomal recessive way, whereas diseases caused by mitochondrial genes can only be inherited from the mother.

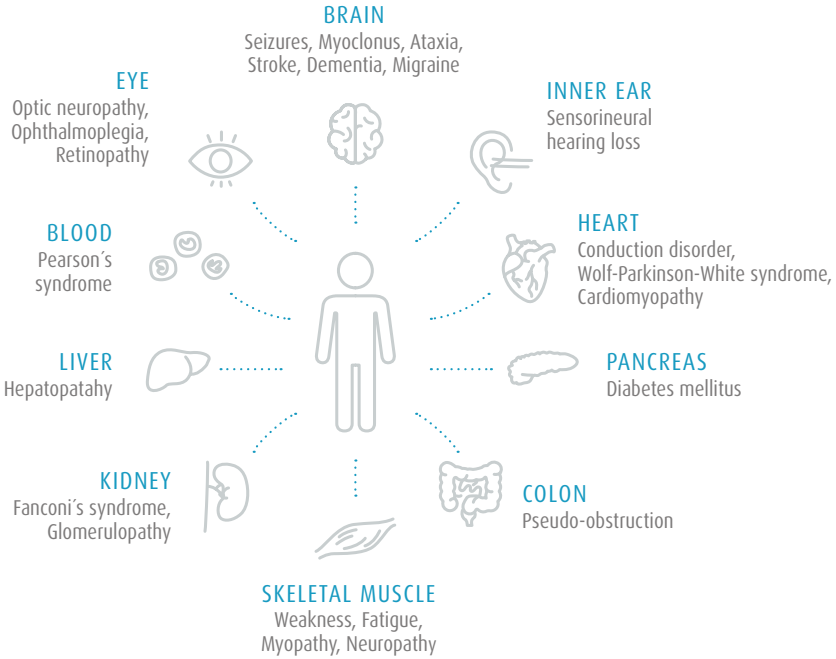
Causes of mitochondrial disorders



mtDNA = Mitochondrial DNA

Symptoms of mitochondrial disorders

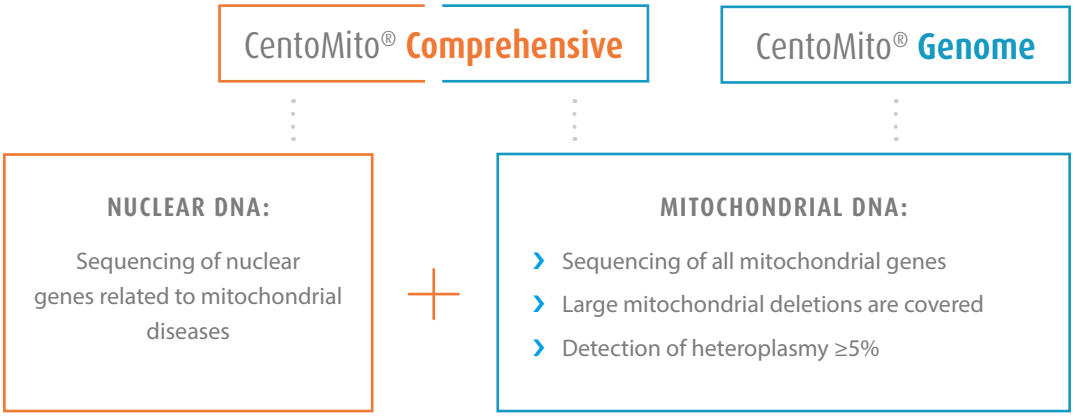
Mitochondrial disorders occur when mitochondria do not function properly, resulting in low or no energy production in cells. As all cells require energy for growth, maintenance and performing their respective functions, this dysfunction leads to multisystemic disease and various organs in the body are affected. The major symptoms caused by mitochondrial dysfunction are described below.



Who should be tested for mitochondrial diseases?

- Individuals with clinical symptoms characteristic of a specific mitochondrial disorder
- Individuals with any progressive multisystem disorder of unknown etiology
- Individuals with multiple complex neurologic features or a single neurological symptom with other system involvement
- Children presenting with lactic acidosis

Testing at CENTOGENE for mitochondrial disorders:



SAMPLE REQUIREMENTS:

- ≥1µg DNA or
- ≥1ml EDTA Blood or
- ≥1 CentoCard® or
- ≥50mg of snap frozen affected tissue (muscle/liver biopsy)

COVERAGE: Mitochondrial genes: >= 97% >=200x covered
Nuclear genes: ≥99.5% covered ≥20x

TAT: 25 business days

Please note:
We accept DNA isolated from blood or affected tissue

Common syndromes and disorders covered

CentoMito® Comprehensive
Chronic progressive external ophthalmoplegia, Kearns-Sayre syndrome, Leigh's syndrome and maternally inherited, Leigh's syndrome, Mitochondrial disorders, Mitochondrial encephalomyopathy, lactic acidosis and stroke-like episodes, Myoclonus epilepsy with ragged red fibers, Myogastrointestinal encephalomyopathy, NARP, Neonatal mitochondrial hepatopathies, Pearson syndrome

CentoMito® Genome
Chronic progressive external ophthalmoplegia, Kearns-Sayre syndrome, Leber hereditary optic neuropathy, Leigh-like syndrome, Leigh syndrome, Mitochondrial disorders, NARP

Please visit our website
for more information:

www.centogene.com

CONTACT DETAILS:

CENTOGENE GmbH
Am Strande 7
18055 Rostock
Germany

CENTOGENE GmbH is a subsidary of CENTOGENE N.V.

customer.support@centogene.com
+49 (0)381 80 113 - 416
+49 (0)381 80 113 - 401

CLIA #99D2049715

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