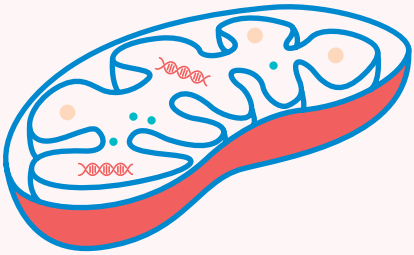
SingHealth DukeNUS
ACADEMIC MEDICAL CENTRE

Genomic
Medicine Centre

Mitochondrial Diseases

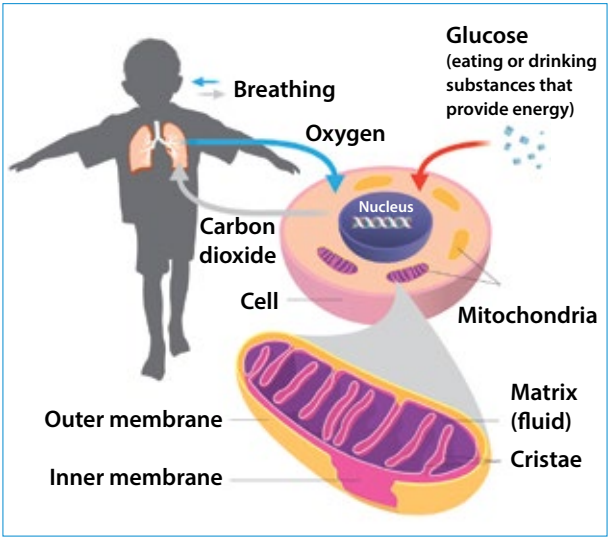


SingHealth

What are mitochondria?

Mitochondria are present in nearly every single cell of our body. Although most of our DNA is kept in the nucleus of each cell, mitochondria have their own set of DNA (mtDNA).

Mitochondria are often considered the 'power battery' of our body as their main function is to make energy from oxygen and food. They produce about 90% of the energy that our body needs to function.



What are mitochondrial diseases?

Mitochondrial diseases (or in short, mito) are a group of chronic genetic disorders which occur when mitochondria fail to produce sufficient energy for our body to function properly.

Therefore, it usually affects body parts that require high levels of energy – which are most of our organs that are used daily.

How common are mitochondrial diseases?

It is estimated that around **one in 5,000 people** are affected by mito.

What are the features seen in mitochondrial diseases?

One hallmark of mito is that it is a **multiorgan disease**.

Most cells in the body have mitochondria. The areas of the body that rely on mitochondria the most are the brain, spinal cord, heart, muscles, kidneys, stomach, intestines and endocrine glands (e.g., thyroid and pancreas). This is because these tissues have a large percentage of mitochondria.

With mito, these organs may not function properly, leading to the following symptoms:

Muscle weakness or 'heaviness'

Vision or hearing problems

Liver or kidney disease

Diabetes mellitus

Gastrointestinal problems

Short stature and poor weight gain

Brain problems such as seizures and stroke-like episodes

People with mito can develop fatigue very suddenly. They may be unable to tolerate mild exercise. They may also take longer than usual to recover from a minor infection like a cold.

Types of mitochondrial diseases

Here are some of the more common mitochondrial syndromes:

1 **Mitochondrial encephalopathy, lactic acidosis, and stroke-like episodes (MELAS)**

MELAS usually starts in childhood to early adulthood. The hallmark of this syndrome is the 'MELAS attack': **stroke-like episodes with seizures**.

Other clinical features include exercise intolerance, short stature, migraine-type headaches, inability to move the eyes, hearing loss, diabetes, digestive problems, heart problems, kidney disease and muscle weakness.

2 **Chronic progressive external ophthalmoplegia (CPEO)**

CPEO is characterised by weakness of the eye muscles which may cause droopy eyelids and reduced ability to move the eyes. This often occurs in conjunction with other mitochondrial syndromes.

3 **Kearns-Sayre syndrome (KSS)**

KSS usually starts before age 20. Common symptoms are a gradual inability to move the eyes (CPEO), difficulty seeing in the dark, heart problems and uncoordinated limb movements. Other symptoms may include hearing problems, kidney disease, diabetes or short stature.

4 **Leigh syndrome: subacute necrotising encephalomyopathy**

Leigh syndrome usually starts in infancy and progression can be fast or slow.

Disease characteristics include vomiting, uncoordinated limb movements, muscle weakness, muscle floppiness or spasticity, seizures, feeding and speech difficulties, hearing loss, abnormal eye movements, visual loss and motor and intellectual regression.

Can mitochondrial diseases be cured or treated?

Unfortunately, most mito do not have curative treatments.

Some patients with certain mito respond well to vitamins and supplements, but the mainstay of treatment would be maintaining good health to prevent worsening of the condition. This would involve good nutrition, regular mild-to-moderate activity and avoidance of infections.

When should I seek medical attention?

If you feel severe lethargy, weakness, numbness or confusion, please seek treatment as soon as possible. Monitor your health more frequently when ill, even with minor infections like a cold.

What causes mitochondrial diseases?

Mito can be caused by changes in either the mtDNA or nuclear DNA.

- Mito caused by mutations in **mtDNA** are inherited (passed down) from mothers. Adult-onset mito are often due to mutations in mtDNA.
- Mito caused by mutations in **nuclear DNA** may be inherited from either parent. Childhood-onset mito are often due to mutations in nuclear DNA.

Mito can affect an individual at any age and symptoms vary widely among individuals.

How are mitochondrial diseases diagnosed?

There is no easy test that can be used to diagnose mito. The diagnosis is made based on a combination of the following:

A **full clinical evaluation** with blood and urine tests looking for features suggestive of mito

DNA tests, which are often required

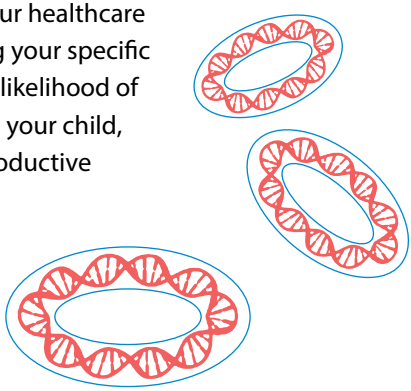
Muscle/liver/tissue biopsies, which may occasionally be needed

Coming up with a diagnosis can be difficult because many symptoms of mito can mimic that of other diseases. Clinical and blood tests may also be inconclusive and difficult to interpret. However, our understanding of mito has been growing with time.

How likely will my child have mitochondrial diseases if I do?

Mito which involve mtDNA are passed down from mothers to their children, while mito which involve nuclear DNA can be passed down from either parent.

Please speak to your healthcare provider regarding your specific condition and the likelihood of passing it down to your child, as well as the reproductive options available.



Mitochondrial diseases are lifelong conditions. Should you require financial assistance or emotional support, please approach your doctor for referral to a medical social worker.

Acknowledgements

Genetics Service, KK Women's and Children's Hospital

Dr Kaavya Narasimhalu,
Department of Neurology (SGH Campus),
National Neuroscience Institute

Dr Chen Zhiyong,
Department of Neurology (TTSH Campus),
National Neuroscience Institute

Information contributed by:

SingHealth DukeNUS ACADEMIC MEDICAL CENTRE

Genomic Medicine Centre

KK Women's and Children's Hospital
6294 4050
www.kkh.com.sg

Singapore General Hospital
6321 4377
www.sgh.com.sg

Changi General Hospital
6850 3333
www.cgh.com.sg

Sengkang General Hospital
6930 6000
www.skh.com.sg

National Cancer Centre Singapore
6436 8088
www.nccs.com.sg

National Heart Centre Singapore
6704 2000
www.nhcs.com.sg

National Neuroscience Institute
6321 4377 (SGH Campus)
6330 6363 (TTSH Campus)
www.nni.com.sg

Singapore National Eye Centre
6227 7266
www.snec.com.sg

Find out more about the Centre at:
www.singhealth.com.sg/genomic-medicine-centre

SingHealth
www.singhealth.com.sg

SingHealth Hospitals

Singapore General Hospital | Changi General Hospital | Sengkang General Hospital | KK Women's and Children's Hospital

National Specialty Centres

National Cancer Centre Singapore | National Dental Centre Singapore | National Heart Centre Singapore | National Neuroscience Institute | Singapore National Eye Centre

SingHealth Community and Primary Care

Polyclinics SingHealth | SingHealth Community Hospitals

Reg. No.: 200002698Z
Information correct as at February 2023

Disclaimer: *The information provided does not replace information from your healthcare professional. Please consult your healthcare professional for more information.*

Scan for a digital copy