How is Mitochondrial Disease diagnosed?

Many symptoms of Mito are similar to those of other ailments - and symptoms may be erratic, making the journey to diagnosis difficult. Sometimes a specific diagnosis cannot be achieved. There is no easy test to detect Mitochondrial Disease. Clinical features and biochemical blood and urine tests can strongly suggest Mitochondrial Disease. DNA tests and/or muscle or liver biopsies are usually required to confirm it. Sometimes, because not a lot is known about the disease, a specific genetic diagnosis cannot be achieved so a diagnosis of Mito is made on the basis of a number of clinical, radiological and biochemical features. Our understanding of the genetic basis of Mito is growing all the time.

Is there any treatment for Mitochondrial Disease?

There are currently no medically approved treatments for most forms of Mito. Some people can respond very well to certain vitamins however for most, treatment is about diagnosis, care, support, symptom management, and genetic counselling. Care includes hydration, nutrition, adequate rest and vitamin supplements. Some people with Mito have certain triggers for deterioration, particularly stroke-like events. Maintaining good nutrition, avoiding sickness and certain medications can be factors that help people stay well. Living with Mitochondrial Disease can be very challenging and demanding physically, psychologically, and emotionally.

Don’t give up hope - don’t let the power go out!
A diagnosis of Mitochondrial Disease can, understandably, be devastating. There will be many questions and emotions. This resource has been collated to provide information and support for New Zealanders affected by Mito, along with their family and friends.

What are Mitochondria?

Mitochondria are found in every cell in our body; they are responsible for making energy and are known as the “powerhouse” or battery of the cell. Mitochondria are necessary for growth and development and allow the body to perform basic functions like talking, walking and moving.

What is Mitochondrial Disease?

Mitochondrial disease or “Mito” is caused when our mitochondria are faulty and therefore unable to produce enough energy. This can result in damaged cells and sometimes organs beginning to fail. It is a degenerative disease that usually targets the parts of our body that require the most energy such as the brain, heart and muscles, but any organ system at any age can be affected. At present, there is no cure for Mitochondrial Disease.

Don’t let the power go out

We all have mitochondria. They are the tiny parts of our cells that create energy for our body, allowing us to walk, talk and breathe. When they work correctly they are the POWER-HOUSE for our body, when they don’t work the body has a power failure and major organs begin to shut down. This power failure is called Mitochondrial Disease.

What are the signs and symptoms?

The severity of Mito symptoms differs from person to person and everyone’s journey is different. One hallmark of Mito is multi-organ disease. In babies in particular, the brain is invariably involved. Some common symptoms might be:

- Poor growth
- Neurological problems: stroke-like events
- Affecting specific parts of the brain
- Seizures
- Poor co-ordination
- Muscle weakness (hypotonia)
- Developmental delay or learning disability
- Poor eye movement (ophthalmoplegia) or neurological deafness
- Heart, liver or kidney disease or failure
- Adults may present with early onset diabetes and multiple miscarriages

A person with Mito may look “normal” but face great challenges and potential health issues due to their condition.

Why does it happen?

All of the proteins in our body are made from our DNA, our genetic material that makes us human. We have two lots of DNA in every cell. One is our nuclear DNA (nDNA) which we inherit from parents. There are over 20,000 genes in our nDNA, each gene has a different job to do. Over 1000 are involved in helping the mitochondria function. The second is mitochondrial DNA (mtDNA) which is passed down from our mother as only the egg contains mtDNA. This is a ring of DNA found in every mitochondria. Mitochondrial disease is always genetic. The inheritance is most commonly; due to a fault (mutation) in the nDNA of both parents (autosomal recessive); due to a mutation in the mtDNA that can be maternally inherited or it can occur for the first time in the affected individual (de novo); and it can be passed down from one affected parent (autosomal dominant).