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FAQS

It can happen to **ANYONE** at **ANYTIME**

What is mitochondrial disease?

You may have only recently heard of mitochondrial disease, but you surely have heard of **Alzheimer's disease, Parkinson's disease, diabetes, ALS, cancer** and many other more commonly recognized chronic diseases. Research tells us that mitochondrial dysfunction is at the core of these more well-known diseases that affect millions of people globally.

Mitochondria are the **'power plants'** that provide our bodies with all of the energy we need to power our cells and organs in order to walk, talk, laugh, hear, digest food, and breathe.

Mitochondrial disease is a group of disorders caused by dysfunctional mitochondria. Each condition is the result of a genetic mutation, in either the **mitochondrial DNA (mtDNA)** or **nuclear DNA (nDNA)**, which results in changes in the genetic material of the mitochondria causing them to fail.

When someone has mitochondrial disease, it means that the power plants in their cells are not working properly. It also means that they could suffer a **'power failure'** in the organ systems that sustain life. This is a devastating disease and can be fatal.

How many people are affected?

- **1 in 4,000** people are estimated to have a primary mitochondrial disease
- At least **1 in 200 individuals harbor a mitochondrial mutation**, which could manifest into mitochondrial disease
- Due to complexity in diagnosis, it is estimated **only 10% of patients receive a diagnosis**
- Globally, **every 30 minutes**, a child is born with mitochondrial disease

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How does mitochondrial disease affect the body?

The parts of our bodies that need the most energy, such as the **heart, brain, muscles and lungs**, are the most impacted by mitochondrial disease.

Affected individuals may suffer strokes, seizures, reflux, severe vomiting, constipation, diarrhea, swallowing difficulties, blindness, deafness, heart and kidney problems, muscle failure, diabetes, lactic acidosis, immune system problems and liver disease.

What symptoms could an undiagnosed individual exhibit?

Symptoms span a broad spectrum in presentation. The disease may manifest in children and adults in a variety of ways. **Seizures, gastrointestinal problems, failure to thrive, chronic fatigue, heat/cold intolerance, poor muscle tone, delayed achievement of milestones, unable to fight typical childhood infections or repeated infections and fevers** without a known origin are all potential signs.

A 'red flag' for mitochondrial disease is when a child or adult is experiencing **issues involving 3 or more organ systems** or when a 'typical' disease exhibits atypical qualities. Even patients within the same family who have the same mitochondrial disease can have differences in symptoms, severity, and age of onset (start of symptoms).

Due to the complexity of the disease and diagnosis combined with few, physicians and experts specializing in the field, **many go undiagnosed for decades.**

When does the disease manifest?

The **disease can appear at any age**, but predominantly manifests before the age of 10. Many children and adults journey for years seeking a proper diagnosis of mitochondrial disease.

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What is the prognosis for these individuals?

Mitochondrial disease is a **chronic progressive illness** for which there is **no cure**. However progress is being made by Canadian researchers for more effective methods of diagnosis and treatment allowing some to live with the hope of slowing the progression of the disease until a cure is found.

Sadly some of the most severely affected children do not survive their teenage years and many adults see a once productive lifestyle turn to debilitation.

Are patients mis-diagnosed?

Around the world every 30 minutes, a child is born who will develop a mitochondrial disease by age 10. While exact numbers of Canadians suffering from mitochondrial disease are hard to determine because so many are **frequently misdiagnosed**, we do know that the disease is approaching the frequency of childhood cancers.

Mitochondrial disease is the **most common form of inborn errors of metabolism. Many patients are misdiagnosed** with atypical cerebral palsy, various seizure disorders, diseases of aging or even sometimes told they have no disease and are hypochondriacs. Still others aren't diagnosed until after death.

Can adults have mitochondrial disease?

Yes, **many adults are diagnosed** with adult-onset mitochondrial disease. Mitochondrial disease **can manifest at anytime** and is among the **most common adult form of inherited neurological disorders.** Some of these individuals have been ill their whole lives but have gone undiagnosed. Others have carried the genetic mutation that causes mitochondrial disease since birth but did not show any symptoms until a severe illness brought on the disease.

Adult mitochondrial disease patients are affected in a similar manner as children.

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What types of treatments and therapies are available?

At present, there are **no highly effective treatments**. Related symptoms may be managed with various medications.

Most physicians prescribe a **'mito cocktail'** of over the counter supplements that may include Coenzyme Q-10, L-Carnitine, Thiamine, Riboflavin, Vitamin C and Vitamin E to name a few. The supplements are often prescribed in high doses and may require a patient taking up to 50 different pills per day.

Exercise has been scientifically proven as one of the most beneficial therapies (as long as the patient can tolerate it). When exercising, you produce more energy on a biochemical level, which improves mitochondrial function and increases the number of healthy mitochondria in the cells.

Is there a cure?

No - but we are working on it!
There are a number of clinical trials underway.

What can I do?

MitoCanada is Canada's only patient advocacy organization for mitochondrial disease. We offer patient **support** and programs, **research** funding, **public education** and **awareness** campaigns, and **advocacy**. If you would like to help us towards a cure, you can **volunteer, participate** in an event**, host** your own event, advocate and **spread awareness** through your social media channels o**r make a donation.**

Please visit our website for more information and consider a gift today at **mitocanada.org**.