

## Frequently Asked Questions

### What is Mitochondrial Disease?

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

Studies have also linked mitochondrial dysfunction to autism spectrum disorders, cardiac issues and even some cancers.

Scientists believe that by focusing on mitochondrial dysfunction, they may be able to devise effective treatments and potential cures for mitochondrial disease and help the millions of people who suffer from the other above-mentioned diseases.

### What are Mitochondria?

All cells in the human body contain mitochondria. Mitochondria are the **'power plants'** that provide the body with all of the energy it needs to walk, talk, laugh, hear, digest food, and breathe. When a person has mitochondrial disease, the power plants in their cells are not working properly. It's like having a massive power outage within your body. In some cases, mitochondrial disease is fatal.

### When was mitochondrial disease first discovered?

The first case of mitochondrial disease was diagnosed in an adult in the 1960s and in the paediatric population in the 1980s.

### How does mitochondrial disease affect the body?

The parts of the body that need the most energy, such as the heart, brain, muscles and lungs, are the most affected by mitochondrial disease.

Individuals may suffer strokes, seizures, gastrointestinal problems (reflux, severe vomiting, constipation, diarrhea), swallowing difficulties, failure to thrive, blindness, deafness, heart and kidney problems, muscle failure, heat/cold intolerance, diabetes, lactic acidosis, immune system problems and liver disease.

### What symptoms could an undiagnosed individual exhibit?

A 'red flag' for mitochondrial disease is when a child or adult has issues with more than 3 organ systems or when a 'typical' disease exhibits atypical qualities.

### When does the disease manifest?

The disease can appear at any age. Many children and adults journey for years seeking a proper diagnosis of mitochondrial disease. Because there are few physicians and experts in the field, some people are often misdiagnosed.

### What is the prognosis for these individuals?

As more research dollars are raised to find more effective treatments and ultimately a cure, some of the affected children and adults are living fairly normal lives with mitochondrial disease.

At the opposite end of the spectrum, many are severely affected, and some children do not survive their teenage years. Adults may see a once productive lifestyle turn to debilitation.

### How many people are affected?

We know, around the world, that every 30 minutes, a child is born who will develop a mitochondrial disease by age 10. While exact numbers of children and adults suffering from mitochondrial disease are hard to determine because so many people who suffer from mitochondrial disease are frequently misdiagnosed, **we now know the disease is approaching the frequency of childhood cancers.**

Many are misdiagnosed with atypical cerebral palsy, various seizure disorders, childhood diseases and diseases of aging. 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 is among the most common adult form of inherited neurological disorders - affecting 1 in 4300. Some of these individuals have been ill their whole lives but went undiagnosed. Others have carried the genetic mutation that causes mitochondrial disease since birth but did not show any symptoms until a severe illness brought them on.

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

### What types of treatments are available?

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

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

### What is the cost of Mitochondrial Disease in Canada?

The overall disease burden is unknown but significant, resulting in substantial direct and indirect health care costs to the patient and society as a whole.

### Is there a cure?

Currently, no - but we are working on it.

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