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Exploring Mitochondrial Disease

A Guide from Diagnosis to Management



Exploring Mitochondrial Disease: A Guide from Diagnosis to Management

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Understanding Mito

The journey to diagnosis can be a long and often worrisome process. When a diagnosis is finally made, it can bring on feelings of fear and concern. A diagnosis can also provide relief in now knowing the reason behind unexplained symptoms.

Mitochondrial disease affects each person differently, so ensuring you have access to a supportive healthcare team and reliable information about symptoms, treatment, and their impact means you can navigate the path ahead from an informed position.

MitoCanada has created this resource, guided by the Mitochondrial Care Network's 2019 Newly Diagnosed publication, to provide you with credible information and resources you can trust. It is a tool to help you navigate your diagnosis or a future diagnosis and support you in your conversations with your healthcare team, family, friends and colleagues.

Mito Facts

- We make about 2 billion mitochondria every second of our lives
- Adults have about 100,000 trillion mitochondria
- The lifespan of a mitochondria is about 100 days

Why Mitochondria Matter

Mitochondria are present in almost every cell in the human body. Creating more than 90% of our required energy, our mitochondria transform the food we eat and the oxygen we breathe into essential energy called ATP (adenosine triphosphate). This energy keeps our hearts beating, lungs breathing, brains thinking, and bodies moving; we simply cannot survive without our mitochondria.

When mitochondria don't function properly, they don't produce the energy we need. Mitochondrial disease commonly affects energy-intensive systems such as the heart, liver, kidneys, brain, muscles, digestive tract, eyes and ears.

What is mitochondrial disease? Mitochondrial disease (mito) refers to a group of disorders that result when mitochondria cannot produce enough energy or an individual doesn't have enough healthy mitochondria in their bodies.

How common is mitochondrial disease?

An estimated
1 in 4,000
people have mito



It affects males and
females **equally**



Mito can be present
from **birth through to**
adulthood



What is Primary Mitochondrial Disease and Secondary Mitochondrial Dysfunction

Mitochondrial disease is a broad term that covers a wide range of mitochondrial disorders and syndromes. Mitochondrial disease is divided into two distinct categories – primary mitochondrial disease and secondary mitochondrial dysfunction. The distinction is important as each has a unique genetic basis, diagnosis and treatment strategies.

Primary mitochondrial disease (PMD): PMDs are long-term, progressive and caused by genetic mutations often inherited from one or both parents. In some rare cases, spontaneous or random mutations occur. This happens at the time of conception or just before conception. PMDs exist from birth, but symptoms can present at any age.

PMDs are usually diagnosed when mutations are identified in the genes responsible for creating ATP. PMDs can affect almost any part of the body, including the brain, kidneys, liver, nerves, muscles, heart, eyes and ears and can cause physical, developmental, and cognitive disabilities.

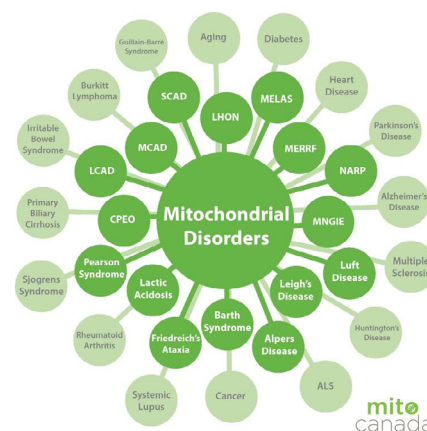
Secondary mitochondrial dysfunction (SMD): Occurs when the mitochondria do not function as well as they should. This dysfunction can be caused by gene mutations that are either inherited or develop at some point during a person's life. They can be caused by environmental factors such as exposure to a drug, toxic substance, or viral infection. These types of gene mutations do not directly affect how energy is generated but impact the function of mitochondria in some way.

Environmental exposure to cigarette smoke, asbestos and drug toxicity can also impair the mitochondria's ability to produce energy, creating dysfunction [\(1\)](#). SMD can also be acquired through illness, infection, inflammation, aging and strenuous medical procedures.

Other terms used for mitochondrial disease: Mitochondrial cytopathy and mitochondrial myopathy are other terms used in place of mitochondrial disease. These older terms are used less often. The medical community is trying to use more straightforward terms, like primary mitochondrial disease, to eliminate patient and caregiver confusion or misconceptions.

Historically, the term “possible mitochondrial disease” has been used when mito is suspected but cannot be confirmed with a genetic diagnosis. The challenge in using “possible mitochondrial disease” is that there are many conditions that look like or are similar to mito but are not mito. When genetic testing cannot confirm a mitochondrial disease, using the term “possible mitochondrial disease” can cause harm by creating anxiety or delay finding an accurate diagnosis. Not having an accurate diagnosis may result in providing the wrong care and management to a patient.

Currently, “diagnosis uncertain” with a detailed description of symptoms and test results is the preferred term when mito cannot be genetically confirmed.



* dark green are PMD, light green are SMD

Symptoms

Mito symptoms can be mild or severe, differ by individual, and depend on which cells of the body are affected. The two most common mito symptoms are muscle weakness and exercise intolerance which leads to feelings of exhaustion. Mito affects different parts of the body, and patients often have symptoms affecting multiple organ systems. Organs most commonly affected including the brain, nerves, heart, pancreas, liver, eyes, and kidneys. This is because each of these relies very heavily on energy production from the mitochondria to function properly.

Common mito symptoms:

1. muscle weakness



2. exercise intolerance



3. extreme exhaustion



Mito symptoms may include impaired hearing and vision, ataxia (challenges with balance, coordination and speech), seizures, learning disabilities and poor growth. Children with mito may have difficulty developing certain skills, such as sitting, crawling, walking, speaking and learning. Remember that no one symptom can give a diagnosis; it's a combination of symptoms and how they continue to present over time, and during certain activities, that need to be considered.

Because most people with mito experience symptoms that affect multiple systems simultaneously, common symptoms are grouped and referred to as syndromes. Symptoms of mito syndromes are outlined in the charts in the following pages.

Syndrome Symptoms

Barth Syndrome <i>Infancy or early childhood (sometimes adulthood)</i> <i>Mainly impacts males</i>	<ul style="list-style-type: none"> • Enlarged heart • Increased rate of infections • Delays in growth before puberty • Muscle weakness and low muscle tone (hypotonia) • Specific facial appearance (round face, full cheeks, pointed chin, large ears, deep-set eyes) • Certain laboratory findings: high lactate, low white blood cell count, low cholesterol, increased 3-methylglutaconic acid and 2-ethylhydracrylic acid in urine or blood, increased monolysocardiolipin: cardiolipin ratio
CPEO Chronic Progressive External Ophthalmoplegia <i>Adolescence or early adulthood</i>	<ul style="list-style-type: none"> • Weakness of the eye muscles leading to decreased ability to move the eyes • Ptosis (weakness of the eyelid muscle leading to drooping of the eyelids)
KSS Kearns-Sayre Syndrome <i>Before age 20</i>	<ul style="list-style-type: none"> • Weakness of the eye muscles with decreased eye mobility • Ptosis (weakness of the eyelid muscle leading to drooping) • Abnormal pigment in the back of the eye (pigmentary retinopathy) which may affect vision • Failure to thrive • Abnormal heart rhythm • Ataxia (poor muscle control) • Certain laboratory findings (increased protein in cerebrospinal fluid)

Syndrome Symptoms

Leigh Syndrome When inherited through mtDNA, it may be called Maternally Inherited Leigh Syndrome (MILS) <i>Infancy or early childhood</i>	<ul style="list-style-type: none"> • Rapid loss of developmental skills, including head control, sitting, standing, or walking following earlier normal development • Decreased level of consciousness • Difficulty breathing • Weakness, seizures, low muscle tone (hypotonia), abnormal movements • Ataxia (poor muscle control) • Certain laboratory findings (increased protein in cerebrospinal fluid)
MDS Mitochondrial DNA Depletion Syndrome <i>Infancy to adulthood</i>	<ul style="list-style-type: none"> • Impaired energy production affecting muscle, liver, brain and kidneys • Intractable seizures • Liver problems • Ataxia (poor muscle control) • Weakness and numbness in the limbs • Hearing loss • Difficulty with eye movements and drooping eyelids • Difficulty gaining weight
MELAS Mitochondrial Encephalomyopathy Lactic Acidosis, and stroke-Like Episodes Syndrome <i>Childhood to early adulthood. Onset may be as late as 40 years</i>	<ul style="list-style-type: none"> • Dementia • Seizures • Migraine headaches • Stroke-like episodes • Recurrent vomiting • Weight loss • Exercise intolerance and weakness • Hearing loss
MNGIE Mitochondrial Neurogastrointestinal Encephalomyopathy <i>Infancy to adulthood, usually before age 20</i>	<ul style="list-style-type: none"> • Difficulty moving food through the digestive tract • Peripheral neuropathy (numbness/weakness in limbs, hands, feet) • Ptosis • Ophthalmoplegia • Hearing loss • Leukoencephalopathy (abnormal white matter in brain)
MERRF Myoclonus Epilepsy with Ragged Red Fibres <i>Childhood to adulthood</i>	<ul style="list-style-type: none"> • Muscle twitching (myoclonus) • Seizures • Ataxia (poor muscle control) • Muscle weakness and exercise intolerance • Hearing loss • Dementia • Enlarged heart muscles
NARP Neuropathy, Ataxia, and Retinitis Pigmentosa <i>Infancy to adulthood</i>	<ul style="list-style-type: none"> • Peripheral neuropathy • Muscle weakness • Ataxia (poor muscle control)
Pearson Syndrome <i>Infancy</i>	<ul style="list-style-type: none"> • Anemia that usually requires frequent transfusions • Signs of pancreas dysfunction

Causes

What causes mitochondrial disease? In many patients, mitochondrial disease is caused by an inherited gene mutation or multiple mutations. In others, external environmental factors may be responsible for mitochondrial dysfunction. Many different types of mito can develop in patients in contrasting ways. It often takes more work for healthcare teams to define direct causes.

Primary mitochondrial disease (PMD) exists from birth and is genetic, meaning the disease is inherited or passed down from one or both parents. In some rare cases, mito can be caused by a “spontaneous mutation”, meaning it occurs randomly at or before conception ⁽²⁾.

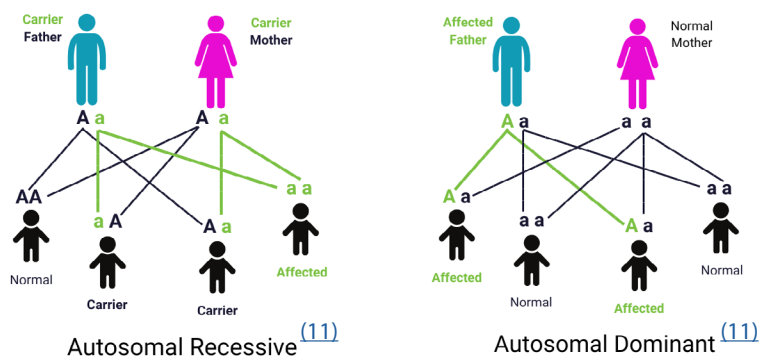
Within the mitochondria are two types of genetic material, **mitochondrial DNA (mtDNA)** and **nuclear DNA (nDNA)**. mtDNA is passed on from the mother to her children, and nDNA is passed on from both parents ⁽³⁾.

Maternal inheritance: A mother with a mitochondrial DNA (mtDNA) gene mutation can pass along the mutation to all of her children. Typically, all children will be affected but with varying degrees of severity, from no symptoms to severe disease. This does not mean that all of her children will be affected in the same way, and it is rarely possible to predict how children will be affected (causing stress for family planning) ⁽⁴⁾.

Autosomal recessive inheritance: The nuclear DNA (nDNA) that makes up part of the mitochondria is inherited by both parents, half from each. Autosomal recessive mitochondrial disease can only be passed on if BOTH parents are carriers. Meaning, each parent carries the mutated gene but not the disease, so they do not have any symptoms ⁽⁵⁾.

When both parents are carriers, there is a 25% chance of having a child that will not carry the mutation nor the disease, a 25% chance of having a child with mito and a 50% chance of having a child who will carry the mutation but will not have mito ⁽⁶⁾.

Autosomal dominant inheritance: If one parent has a dominant nDNA gene mutation and the other parent does not have any nDNA mutations, 50% of their children will inherit the mutation with the disease and symptoms, and 50% of their children will inherit normal genes; therefore, no disease or symptoms ⁽⁷⁾.



X-linked inheritance: X-linked conditions are caused by genetic variants on the X chromosome, one of the two sex chromosomes. Because females have two X chromosomes and males only one, the disorders can appear different in the two sexes. Males are nearly always more severely affected. The rules for inheritance are also different since men cannot transmit X-linked genes to their sons but will always transmit them to their daughters.

Diagnosis

How is mitochondrial disease diagnosed? As mito can affect each person differently, and many family physicians are unfamiliar with the disease, the process leading to a diagnosis can be long and frustrating. This process usually begins with a family physician sending a referral to a specialist.

Specialists who diagnose mito can include neurologists, metabolic specialists, geneticists, cardiologists, neuro-ophthalmologists or endocrinologists. The type of specialist you see may depend on which part of your body is affected. Specialists will take a medical history, conduct a thorough physical evaluation, measure your strength and endurance, and possibly order a series of specialized tests.

Certain blood tests may be ordered to look at amino acids, acylcarnitines, lactate, or pyruvate levels. Urine tests are often ordered as they can show indicators of reduced mitochondrial function.

A genetic test may be ordered to screen for known, rare and unknown genetic mutations and requires either a saliva, blood or muscle tissue sample. A muscle biopsy may be ordered, which involves removing a small piece of muscle tissue. The tissue is treated with a dye that stains the mitochondria, revealing certain mitochondrial enzymes, proteins or muscle cells with excessive mitochondria.



Other tests may include:

CT or CAT Scan Computed (Axial) Tomography	Using computers and rotating X-ray machines, the CT creates images to provide detailed information about soft tissues, blood vessels, and bones in various parts of the body.
ECG or EKG Electrocardiogram	This fast and simple test records the electrical signals in your heart. Electrodes connect to the ECG machine through wires placed on certain spots on your chest, arms and legs. Specialists will be looking for signs of arrhythmia or cardiomyopathy.
Echo Echocardiogram	An echocardiogram looks for irregularities in the structure of your heart using ultrasound. This test is used to look for signs of cardiomyopathy.
Electromyography	This test assesses the health of muscles and nerves. An electrode is inserted through the skin into an affected muscle. The machine records electrical activity of the muscle and can determine whether muscle weakness is caused by the muscle itself or the nerves that control the muscle. An electromyography is often carried out at the same time as a nerve conduction study.
Genetic Testing	Genetic testing can determine whether someone has a genetic mutation in the nDNA or mtDNA that causes mitochondrial disease using blood, muscle or saliva samples, as described above. Note: there is no genetic test that completely rules out a genetic condition.

Other tests may include:

Laboratory tests	Blood or urine are standard tests used to detect problems with various organs, including the liver and kidney as described above. These tests also look for elevated lactic acid levels, which is common for those living with mitochondrial disease.
MRI Magnetic Resonance Imaging	MRI machines use a magnetic field and computer-generated radio waves to create detailed 3D images of organs and tissues that can be evaluated by a specialist.
MR Spectroscopy Magnetic Resonance Spectroscopy	Using the MRI machine, this test measures levels of certain chemicals within a tissue which may suggest mitochondrial dysfunction.
Muscle Biopsy	This procedure involves removing a small sample of muscle, usually from the thigh, as described above. The sample is then treated with a dye. Muscle fibres affected by mitochondrial disease show as a distinct red and have a ragged appearance.
Nerve Conduction Study	This test evaluates the ability and speed of nerve impulses and can be used to rule out conditions other than primary mitochondrial myopathies (PMM) .
Neurological Test	This test uses tuning forks, flashlights and/or reflex hammers to evaluate motor and sensory skills, hearing, vision, speech, coordination, and balance.
Spinal Tap or Lumbar Puncture	A needle is inserted between two vertebrae to remove a small amount of cerebrospinal fluid (CSF) to measure glucose, protein and/or lactic acid levels. CSF surrounds and protects the brain and spinal cord and abnormal lab values may indicate mitochondrial disease.

Why is a diagnosis helpful?

Although receiving a diagnosis can often be a long and uncertain process, knowing exactly which mitochondrial disease you have can help you and your loved ones better understand the disease and plan a path forward. It can also help you understand the long-term realities of the disease and help your health care team create a supportive treatment plan. Getting a diagnosis may ensure other diseases are not being overlooked, prevent the wrong treatments from being used, and can stop the need for additional or unnecessary diagnostic testing.

Receiving a genetic diagnosis can identify certain inherited mitochondrial diseases. If this is the case, your specialist may recommend that family members see a genetic counsellor and undergo genetic testing. A confirmed genetic diagnosis also aids family planning discussions to learn how the inherited disease may affect future children. There are mitochondrial reproductive options, such as pre-implantation genetic diagnosis (PGD) and tests, such as amniocentesis, to determine mitochondrial disease risk.

Receiving a genetic diagnosis is also helpful, should you ever be interested in enrolling in a clinical trial. Some clinical trial studies may require confirmation of diagnosis through genetic testing to participate.

Genetic Counselling

How does mitochondrial disease affect family members and future children? Depending on what type of mitochondrial disease you have, your siblings or future children may be at risk of developing mito. Passing on mito from parent to child depends on many factors, including whether the disease is caused by genetic mutations in the nuclear or mitochondrial DNA.



When a genetic diagnosis is made, healthcare teams and genetic counsellors will help determine if other family members are at risk and if that particular type of mito can be passed on to future children. You and your family may consider genetic testing and advise other family members to do the same. MitoCanada is here to provide you with all the resources you need. If you have questions about genetic counselling, reach out to us at info@mitocanada.org.

What to do if there is no genetic diagnosis

Genetic testing technology is advancing quickly, and testing options continue to improve. If you or your family members do not have a genetic diagnosis, talk to your healthcare team or a physician experienced in mito care. They can assist in the diagnosis process using clinical tests, laboratory evaluations, image studies and/or tissue biopsies. As is often a requirement, the tests referenced earlier in this guide must be completed first before a request for genetic testing is made. Genetic testing is costly. In some cases, tests are shipped out of province or country for processing. In certain provinces, genetic testing first requires provincial funding approval to receive coverage.

If genetic test results come back negative, you may still have a mitochondrial disease. Consider requesting another panel of genetic testing in the future. Genetic testing may be negative or inconclusive now, but in 3-5 year's time may be able to determine a diagnosis.

Managing Mito

Currently, there is no cure for mitochondrial disease. Managing mito is an ongoing process; however, certain medications, supplements, exercise plans and nutrition management can help control and even ease some of the symptoms you may be experiencing.

Treatment Options

Medications: Treatment for mitochondrial disease is currently symptom-based management. Medications are quite variable and typically are used to treat the mito-associated symptoms (i.e. seizures, strokes).

Supplements: When considering the use of supplements, it is advised that you do so under the guidance of a healthcare provider, as specific supplements are chosen based on the type of mito you have been diagnosed with.

Many individuals with mito are prescribed the “mito cocktail.” This combination of supplements varies, depending on the condition being treated. Mito cocktail supplements may include creatine, L-carnitine, alpha-lipoic acid, riboflavin, and CoQ10.

Exercise as Medicine

The best exercise for you is the exercise you enjoy doing! Mindful exercises can be highly beneficial for those with mito. Exercise can improve mitochondrial quantity and function, improve muscle strength, and help reduce fatigue. The benefits of exercise extend to every part of the body and improve mitochondrial health.

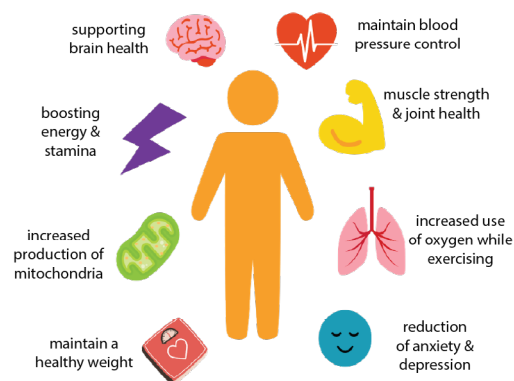
To understand what types of exercise are safe for you and the frequency at which you should exercise, speak with your doctor. There are two main types of exercises to consider: endurance and resistance/strength.

Endurance exercise, also known as aerobic exercise, involves many low-intensity repetitions that make your heart and lungs work harder. Some examples include walking, running, biking, jumping rope, swimming, treadmill, ski machine, Stairmaster, recumbent or upright bike (spinning), and pedal ergometer.

Resistance/strength exercise involves contracting muscles against resistance. Resistance can be your body weight or tools like dumbbells, rubber tubing, or items from around the house. It involves doing a low number of high-intensity repetitions. In the beginning, you may only be able to do one set. Over time, your goal is to do three sets of 12-15 repetitions of each exercise. Having an exercise buddy is a great way to stay motivated if you are new to exercise.

Visit our [Exercise as Medicine guide](#) for more information and examples of endurance, resistance and balance exercises.

Exercise advantages to consider are:



It is important to listen to your body to exercise safely. Creating an exercise schedule can help you stay on track and ensure you have adequate time to rest. It is essential not to exhaust yourself. It is ok to take a break and recover, especially when you are new to exercise. Using a heart monitor is helpful to track your heart rate while exercising and can help ensure you stay within any recommended target zones. Pushing yourself too hard can lead to fatigue, which can be dangerous for people with mito.

For children, find endurance and resistance exercises that are fun but appropriate for the type of mito they have. Modified and self-regulated physical education classes are beneficial, as exercises can be done at the child's own pace. Pets such as dogs can be a great motivator for children to exercise.

Be sure to watch for signs of fatigue or exhaustion.

Signs may include:

- lightheadedness
- severe muscle soreness
- hearing loss
- blurred vision
- heart palpitations or chest pain
- being unable to talk

It is important *not* to exercise:



- when you are sick or have the flu
- In extreme outdoor temperatures
- during long periods of fasting

Diet and Nutrition

There are no specific mito diets or general dietary guidelines for mito patients. Instead, your dietary and nutritional needs will depend on the type of mito you have been diagnosed with and your current state of health. Maintaining a well-balanced diet, one rich in fruits, vegetables, grains, complex carbohydrates, proteins and healthy fats, is typically recommended. However, speak to your doctor to ensure you are including the foods best suited for your nutritional and mito needs.

Your doctor may recommend vitamins, multi-vitamins or iron. Many patients respond well to small, frequent meals throughout the day, and avoiding fasting. Restrictive diets like ketogenic, Atkins or low-carb diets are not advised and could worsen symptoms. If you do or plan to incorporate regular exercise into your weekly routine, it is important to pay attention to nutrition. Staying well-nourished will help with pre- and post-exercise recovery.

Here are some nutritional tips to consider when exercising:

- Do not exercise after fasting for six or more hours
- Always stay hydrated before, during and after your workouts
- Have a light meal with carbohydrates before exercising
- Keep fats, protein and fibre-dense foods low before exercising
- After eating, wait at least 30 minutes before starting exercising
- Try to have at least 20 grams of protein and carbohydrates within 30 minutes after exercising

Medications to Avoid or Use with Caution

Historically, it was thought that mito patients should avoid many medications as they could impact mitochondrial function. However, we now know that many medications are well-tolerated when taken for routine care. It is advised that caution be taken when using a new medication, and should any side effects be observed, reach out to your doctor as soon as possible. They will advise how to proceed with the treatment. Any time a patient is starting to use a new medication, they should take extra care to monitor any adverse symptoms that may develop.

Want to learn more about medication use?



View our curated [list of medications to avoid or use with caution.](#)

Valproic acid (brand name Depakote, Depakene)

- This medication should be used with caution by patients with mitochondrial disease.
- For POLG-related (mutation in POLG gene) disorders and mito disorders with liver involvement, it is advised not to take this medication.

Anesthesia

- It is essential to tell the anesthesiologist if you or your loved one has a mitochondrial disease.
- Some mito patients may have anesthesia-related sensitivities. If this is the case, the patient may require a lower or adjusted dose when general anesthesia is required.
- Muscle relaxants in anesthesia (also called neuromuscular blocking agents) should be monitored in patients with pre-existing myopathy or decreased respiratory drive ⁽⁸⁾. Generally, local anesthetics are well-tolerated.

Antibiotics (aminoglycosides, linezolid, tetracycline, azithromycin and erythromycin)

- Antibiotics such as aminoglycosides, linezolid, tetracycline, azithromycin and erythromycin should be avoided for patients with specific mitochondrial conditions. Ensure your physician knows your diagnosis.
- If needed, these antibiotics should be taken with caution.
- Most mito patients are fine with other antibiotics.

Topiramate or Metformin

- When using either of these two medications, it is recommended to monitor use.
- Statins can sometimes worsen muscle weakness, monitor or avoid them.

Emergency Plans

An emergency care plan should be created to ensure you receive the best medical care for your mito needs, when your medical team cannot be present. It may include plans for emergency room visits or hospital admissions. The emergency plan provides hospital staff with your medical history and emergency protocols to follow, such as a list of treatments you should receive while in hospital, lab work to complete, and medications to avoid. As every mito patient presents their disease differently, your patient emergency protocols will be different and personalized to meet your unique needs.



It is recommended to have your emergency care plan in your local hospital's records.

Always bring a copy of your emergency care plan with you when visiting the hospital.

Surgical Care

When it comes to surgical procedures, extra precautions often need to be taken for mito patients. Pre-surgical preparation should be reviewed and possibly adjusted. Some medical procedures require no eating or drinking 12-24 hours beforehand, also referred to as NPOs (latin abbreviation for nothing by mouth). Fasting before a medical procedure can be dangerous for those with certain types of mito. Furthermore, some surgical plans may advise mito patients to be hospitalized the day before a surgery is scheduled to administer IV fluids. Be sure to speak to your doctor to learn more.

Certain anesthetics may need to be avoided, as some mito patients are often vulnerable to “metabolic stress” when certain anesthetics are used. To avoid any sensitivity to anesthesia, preoperative patients must minimize fasting and have glucose added to preoperative IV fluids, unless the patient has adverse reactions to high glucose intake [\(9\)](#). Anesthetic options must be discussed with the patient by the medical team, including the anesthesiologist, to ensure a safe and well-tolerated anesthetic is selected.

Be your best advocate

Over time, mitochondrial disease may progress. This may lead to the development of new symptoms and may affect new parts of the body. Monitoring your health and reporting any changes to your doctor is essential to help address disease progression early on. Ensure you attend annual evaluations, keep up with requested lab tests and scans, keep a symptom journal to record any symptom changes you may be experiencing and attend routine check-ups.

As part of your surgical care, your healthcare team will create a plan to ensure the surgical team, including the anesthesiologist, is aware of extra precaution that need to be taken.



Healthcare Specialists

Finding Healthcare Specialists

Mito is interdisciplinary, meaning multiple body systems are affected. Depending on what organs are impacted by mito, you may need to see more than one specialist to manage the disease effectively; therefore, the need for multiple healthcare specialties may be necessary.

Your Healthcare Team

Specialists involved in managing mito care include, but are not limited to: neurologists, geneticists, cardiologists, rheumatologists, epileptologists, physiatrists, dietitians, physiotherapists, orthopedic surgeons, audiologists, endocrinologists, immunologists, ophthalmologists and sleep medicine specialists [\(10\)](#).

Mito Lingo

Below are a lists of specialists involved in managing mito:

Neurologist - a medical doctor who specializes in disorders of the brain and nervous system

Geneticist - a medical doctor who focuses on how genetic information is passed from parents to their children

Metabolic Specialist - a medical doctor who specializes in the processes that change food into energy to keep our cells healthy and working properly

Even though you will have new specialists helping you manage your mito, it is strongly recommended that you keep your family doctor. Your family doctor should be able to care for your more complex medical needs and be willing to work alongside your specialists to continue providing care.



MitoCanada has curated a provincial list of clinics that oversee the care of mito patients.

View our [metabolic clinic listing](#) for province-specific information.

Living with Mito

Managing Stress

After receiving a mito diagnosis, many individuals may experience similar challenges and uncertainties as other newly diagnosed patients. With tests to rule out other conditions and tests to gather evidence that mito may be at play, the investigative process can heighten anxiety.

For newly diagnosed patients, the diagnosis can be a relief because testing is no longer needed and now there is a focus on treatment options to manage symptoms. However, patients and their families often also feel overwhelmed and concerned about ongoing care, disease progression, impacts on family life, and financial hardships.

While it is important to learn about your new diagnosis, you cannot learn everything overnight. Your specialists and clinic nurses are there to build your knowledge and confidence in living with mito, as well as guide you in management strategies. MitoCanada is here to provide you with resources, tools and guidance to help introduce you to a supportive Canadian mito community.

Many individuals and families reach out to professional counselling services pre- and post-diagnosis to help cope with the challenges they have faced and the stress, anger and sadness they experience. It may be helpful to talk to someone who can support you during this period of uncertainty. If you or your family feel overwhelmed, consider seeking support from a professional counsellor.

View MitoCanada's list of [provincial and national resources](#) for emotional support.

Finding Support

MitoCanada is dedicated to empowering and enhancing the lives of Canadians living with mitochondrial diseases. From advances in research to educational, and support programs to ways you can get involved in MitoCanada events, you can find a multitude of resources on our website:

- [Energizing Lives newsletters](#)
- [Education guides](#)
- [Community and fundraising events](#)

Visit our Support Resources where you will find a [Regional Resource page](#) for financial, emotional, physical and social help and explore a health management app called [Zamplo](#), a tool to track mito health history, appointments, test results, medications and more.

Explore [MitoCanada's website](#) where can find a multitude of resources.

MitoCanada's Patient Contact Registry

MitoCanda has created a Patient Contact Registry. This is a secure patient-populated database where individuals living with mito and their caregivers can share information about their lived experiences with mito and opt-in to be alerted about clinical trial opportunities in Canada.

All information entered into the registry is de-identified, meaning the information you share is private. Furthermore, entrusting MitoCanada with your lived-experience information helps ensure future patients and families will receive the guidance, education and support they need to live an optimal quality of life.

By participating in the registry, you will:

- Contribute to a growing understanding of mitochondrial disease in Canada.
- Receive information and opportunities about clinical trials, which includes new drugs and therapies.
- Help MitoCanada develop valuable education tools and resources.
- Help strengthen our efforts in raising awareness and advocating for the needs of the Canadian mitochondrial community to provincial and federal governments.

Read more about the [MitoCanada's Patient Contact Registry](#).

Clinical Trials

Clinical trials can be a fantastic opportunity to advance research and medicine. By volunteering in a clinical trial, you may have the opportunity to access medications that may positively impact your disease. Your involvement, first-hand experiences, and the perspectives you share in a trial all help bring a new treatment to Canada that improves quality of life. These elements can impact the health and lives of future mito patients. The truth is, patient participation is essential to clinical trials. Your time and involvement matter!

Clinical trials are controlled research studies that investigate the safety and efficacy of either a new drug treatment, device or procedure. A clinical trial study goes through an extensive research process and must gain approval from Health Canada before moving to patient trials. Once approved by Health Canada, a trial is then done in phases, in which a clinical trial team tests the safety and effectiveness of a new drug or device in different populations. Throughout each trial phase, reviews are completed and approvals are required to ensure a trial is conducted properly and are in the best interests of participants.



Watch [“Clinical Trials Explained for Kids”](#), an animation which shares an excellent overview of clinical trials and walks you through the consent (assent) process in a friendly, upbeat and entertaining way.

MitoCanada is here to support you throughout the clinical trial process and to answer any questions you may have. MitoCanada provides updated lists of clinical trial opportunities for Canadians. Visit our dedicated page to [find clinical trials](#).



Learn more about clinical trials by viewing our web presentation:

[Understanding Clinical Trials, the Patient Perspective](#)

Connecting Through Social Media

MitoCanada has social media pages on [Facebook](#), [Instagram](#), [LinkedIn](#) and [Twitter](#), where we share relevant, up-to-date information about mito management, research, investigational therapies, community events and much more. We offer a closed [Peer-to-Peer Facebook support group](#) to connect to a larger community and find the support you need.

Connecting with mito families and individuals, who live close to you, can be a great resource and source of comfort and connection. Be mindful that those participating in support groups come with a range of mito experiences that may not be quite similar to yours.

Support Organizations

Explore patient associations around the world supporting the mitochondrial disease community:

- [Lily Foundation](#) – UK
- [MitoFoundation](#) – Australia
- [The United Mitochondrial Disease Foundation](#) – USA
- [MitoAction](#) – USA
- [CureMito](#) – USA

About Us

Since 2010, MitoCanada has been supporting those living with mitochondrial disease with information, initiatives and opportunities to inspire, empower and connect while enhancing quality of life. We develop education and awareness programs. We advocate for those living with, or at risk of developing, mitochondrial disease and we fund research that is patient focused and transformational.

MitoCanada is the only national charity dedication to mitochondrial disease. We are the voice of Canadians living with or at risk of developing mitochondrial disease.

Request for support

MitoCanada is a national charity governed by a volunteer board of directors. We rely on donations from the public and the generosity of our partners to develop and deliver support and education programs and resources. Please consider including MitoCanada as one of your charities of choice to support.

[Contact Us](#)

MitoCanada Foundation
30022-478 Dundas Street West
Oakville, ON, L6H 7L8
info@mitocanada.org

[Make a Donation](#)

Make a donation at:
www.mitocanada.org/donate

Through research and public education, we aim to:



Disclaimer: The content found in this education resource guide is for informational purposes only.

The content found in this guide is not to be substituted for professional medical advice. Please seek the advice of your physician or other qualified health providers should you have any questions regarding a medical condition.

End Notes

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