

# Understanding and Coping with Mitochondrial Disease

A guide for patients and parents  
from the Neuromuscular and Metabolic Centre



**McMaster Children's  
Hospital**

## **From the parents' view ...**

When we first heard that our daughter, Alexis, was diagnosed with Mitochondrial disease, we really didn't know what it was. We also learned that not too many people had heard of it before.

Our daughter had many tests performed to reach the diagnosis of Mitochondrial disease. So once we received the news we were actually glad to finally have some answers. No parent wants their child to be ill, but we have learned to accept the disease and live with it on a day to day basis.

We also learned that each person with Mitochondrial disease is affected differently by it. Alexis is severely affected by the disease, so each day with her is special.

Meeting other children with Mitochondrial disease and their families have become a great support for us. We can relate to their daily struggles and if we need advice, they are there for us.

My advice that I would give to others affected by Mitochondrial disease is to not give up. We can all fight this disease together. The hardest part sometimes is people not knowing enough about the disease. We really need to start educating people about it.

*Chris and Stephanie*



# **Understanding and coping with Mitochondrial disease**

## **A guide for parents**

The health care team at the Neuromuscular and Neurometabolic Centre wrote this book to answer some common questions about Mitochondrial disease. We hope that you will find it helpful.

During your child's care, you will meet the members of our team. We will work closely with you and your child to meet your needs. Along the way, we will give you information and support, so that you can make informed decisions and take part in your child's care.

We strive to provide a comfortable and caring environment for you and your family. Please feel free to talk with us about your child's condition, care and any concerns that you may have. We welcome your questions at any time.

from the Neuromuscular and Neurometabolic Team  
McMaster Children's Hospital, Hamilton Health Sciences

**We wish to thank Mr. Warren Lammert and Ms. Kathy Corkins  
for their kind and generous donation.**

**We are also grateful to Giant Tiger and the McMaster  
Children's Hospital Celebration.**

**Their contributions, helped create the Neuromuscular and  
Neurometabolic Centre at Hamilton Health Sciences.**

### **Author:**

Laura Bainbridge, RN, BScN, MEd  
Registered Nurse

### **Medical Consultant and Editor:**

Mark Tarnopolsky, MD, PhD, FRCP(C)  
Medical and Research Director, Neuromuscular and Neurometabolic Centre



# Table of contents

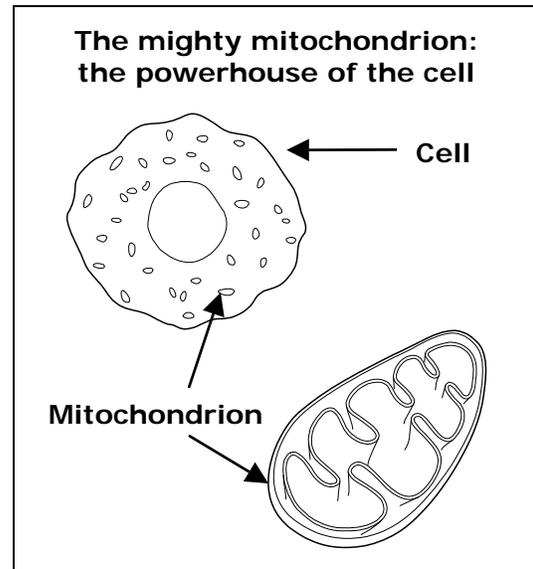
	<b>Page</b>
What is Mitochondrial disease?	1
How do mitochondria work?	2
How do you get Mitochondrial disease?	3
What are the signs and symptoms?	8
How is the diagnosis made?	10
What happens when Mitochondrial disease is diagnosed?	15
Will I know the type of disease?	16
How is Mitochondrial disease treated?	17
How can the Neuromuscular and Neurometabolic Centre help?	22
What can we expect in years to come?	23
How could this disease affect future generations?	25
Where can we get information and support?	27
Dictionary	28
Acknowledgements	30



## What is Mitochondrial disease?

The term “Mitochondrial disease” refers to a group of disorders. Each of these conditions involves a problem with mitochondria.

Mitochondria are tiny structures inside almost every cell in your body; all the way from your skin to the organs inside your body. Their main job is to use the food and oxygen that enter the cells to make energy. Almost all of the energy your body needs for daily life and growth comes from mitochondria.



There are so many types of Mitochondrial disease it would be impossible to name them all, and many have yet to be discovered. Each condition is the result of a genetic mutation – a specific change in the genetic material of the mitochondria. The mutation causes the mitochondria to fail in their task of making energy.

When mitochondria fail, less and less energy is made in the cells. The cells may stop working or die. Depending on where the affected cells are, parts of the body may not function properly and many health problems can result. The symptoms of Mitochondrial disease can range from mild to severe.

**At least 1 in 6000 people have Mitochondrial disease**

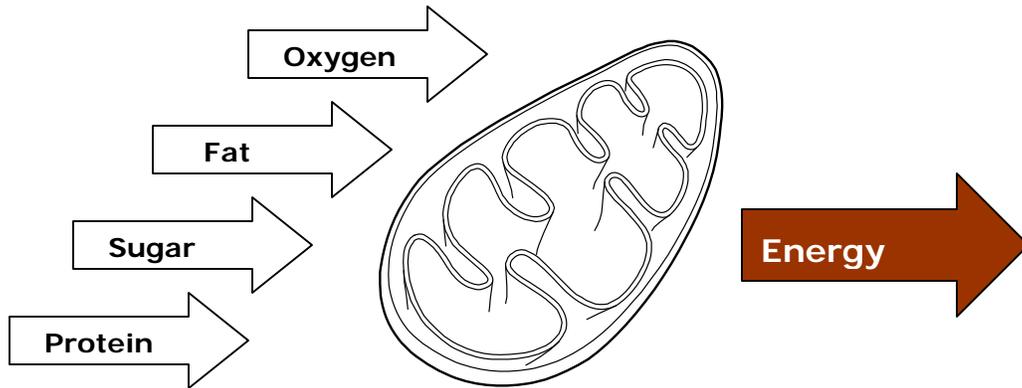
There is no cure for mitochondrial disease. However, many people have a normal life span with their disease well managed.

Research is underway at our centre and across the world that will help us learn more about these diseases and find new treatments and therapies.

In this book, we talk generally about mitochondrial disease, because the questions and needs of patients are similar, whether a specific mutation has been found or not.

## How do mitochondria work?

Mitochondria use oxygen, and the sugar, fat and protein from foods to make energy. The body uses that energy for daily function and growth.



To understand how they work, think of mitochondria like a furnace.

Furnace	Mitochondria
gas and oxygen enter the furnace ↓	food and oxygen enter the mitochondria ↓
gas and oxygen are used to make heat ↓	food and oxygen are used to make energy ↓
if the furnace has a problem ↓	if the mitochondria have problems ↓
gas and oxygen come in but are not used properly and smoke is produced ↓	food and oxygen enter but aren't used properly and lactic acid is produced ↓
little or no heat comes out ↓	little or no energy is made ↓
the house isn't kept warm	parts of the body can't work properly

## How do you get Mitochondrial disease?

### Primary disease

Mitochondrial disease is a genetic disease, meaning that most people are born with it. It is inherited or passed down from one or both parents.

In some rare cases, the disease is caused by a “spontaneous mutation”, meaning that it occurs randomly at or before conception, despite healthy (normal) genes in both parents.

Parents should never be “blamed” or feel that the disease is their “fault” as it is impossible to control how genes are naturally inherited.



**Mitochondrial disease is not infectious.  
You can't "catch" it from another person.  
It is not caused by something you did.**

When mitochondrial disease is the primary condition, it exists right from birth. The severity of the disease determines whether symptoms appear at birth or later in life. The severity of symptoms among family members can vary because the location of the mitochondria that have a specific genetic mutation are completely random at each birth.

<b>For example:</b>	A large number of mitochondria with a specific genetic mutation in the <b>brain</b> .	Can lead to	<ul style="list-style-type: none"><li>• Developmental delay</li><li>• Seizures</li><li>• Movement disorders</li><li>• Poor balance (ataxia)</li><li>• All of the above</li></ul>
	A large number of mitochondria with a specific genetic mutation in the <b>heart</b> .	Can lead to	<ul style="list-style-type: none"><li>• Thickening of the heart muscle walls</li><li>• Heart failure (cardiomyopathy).</li></ul>

## How is mitochondrial disease inherited?

Mitochondria contain 2 types of genetic material:

- mitochondrial DNA, which are passed on from the mother to all children
- nuclear DNA, which is passed on from both parents

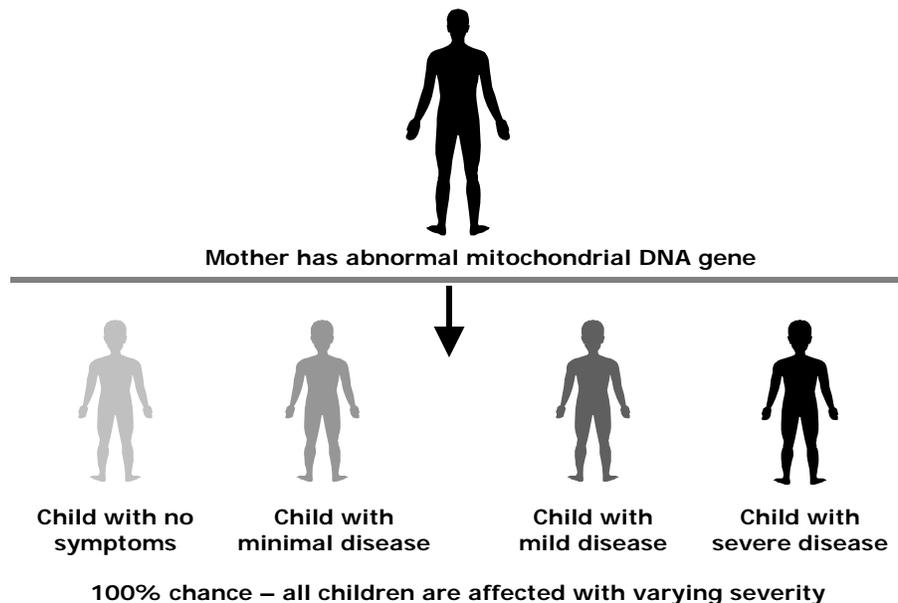
These are the ways that mitochondrial disease can be inherited.

### 1. Maternal inheritance



A mother with a mitochondrial DNA gene mutation will pass this abnormal gene to all of her children. The children will all be affected, with different degrees of severity. This is called maternal inheritance.

This does not mean that the children are going to be affected in the same way as their mother. It is rarely possible to predict how the children will be affected. This creates a lot of stress for those who are planning a family.

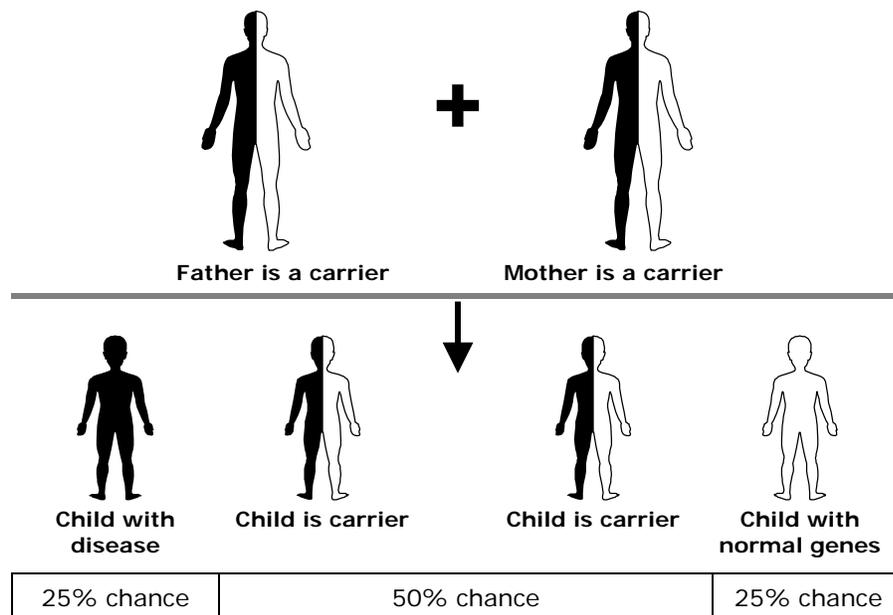


## 2. Autosomal recessive inheritance

The nuclear DNA that make part of the mitochondria is inherited from both parents (half from each parent). Autosomal recessive mitochondrial disease can be passed on only if BOTH the mother and father are “carriers”. This means that they carry the mutated gene, but not the disease – so they don’t have any symptoms. This is called autosomal recessive inheritance.

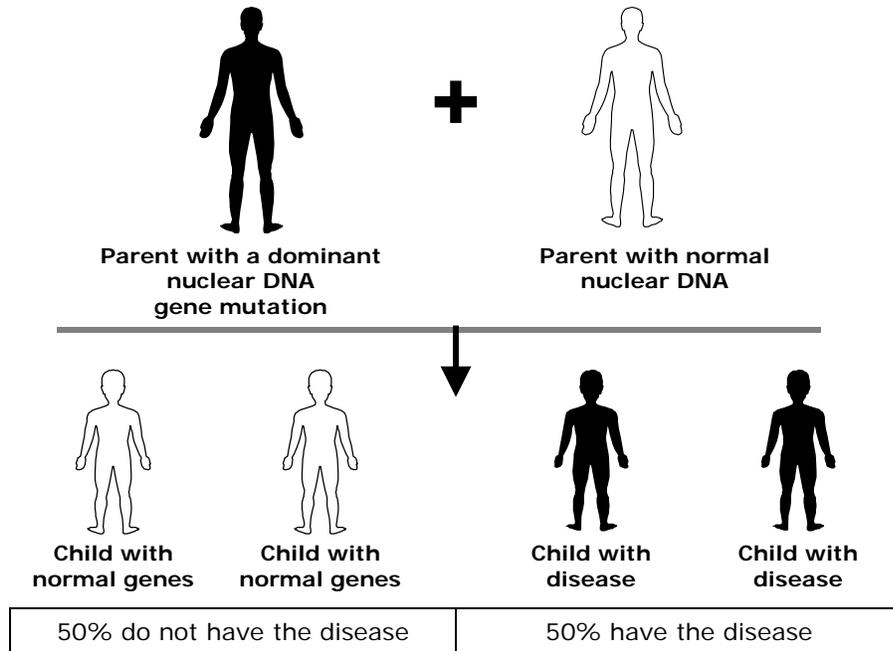
When both parents are carriers, there is:

- a 25% chance of having a child with the disease
- a 50% chance of having a child who is a carrier like the parents (has the mutation, but not the disease)
- a 25% chance of having a child that is not a carrier and does not have the disease



### 3. Autosomal dominant inheritance

If one parent has a dominant nuclear DNA gene mutation, this abnormal gene will be passed on to 50% of his or her children and lead to symptoms. If the other parent has normal nuclear DNA, the children will inherit a normal nuclear DNA gene from him or her, but the dominant gene (with the mutation) will prevail in causing symptoms.



## **Secondary disease**

It is also possible to develop mitochondrial disease later in life, without a primary DNA mutation. This is called secondary Mitochondrial disease.

Secondary Mitochondrial disease may develop due to:

- the natural process of aging - the mitochondria slow down and work less effectively as the body gets older
- taking certain prescription medications
- damage to mitochondria from stressors such as smoking or alcohol abuse

## What are the signs and symptoms?

Every cell in the body, except red blood cells, contains hundreds to thousands of mitochondria working to make energy. The mitochondria in some areas of the body may be working properly, but not in other areas. This can cause a wide variety of symptoms.

There is no one identifying sign or feature of Mitochondrial disease. Symptoms can vary and range from mild to severe, even among affected family members. In mild cases, young people may learn to cope and adapt to the amount of energy they have and don't realize they have symptoms, or adults may comment that they were very healthy as a child, but not really athletic.

People with Mitochondrial disease often have one of more of these symptoms:

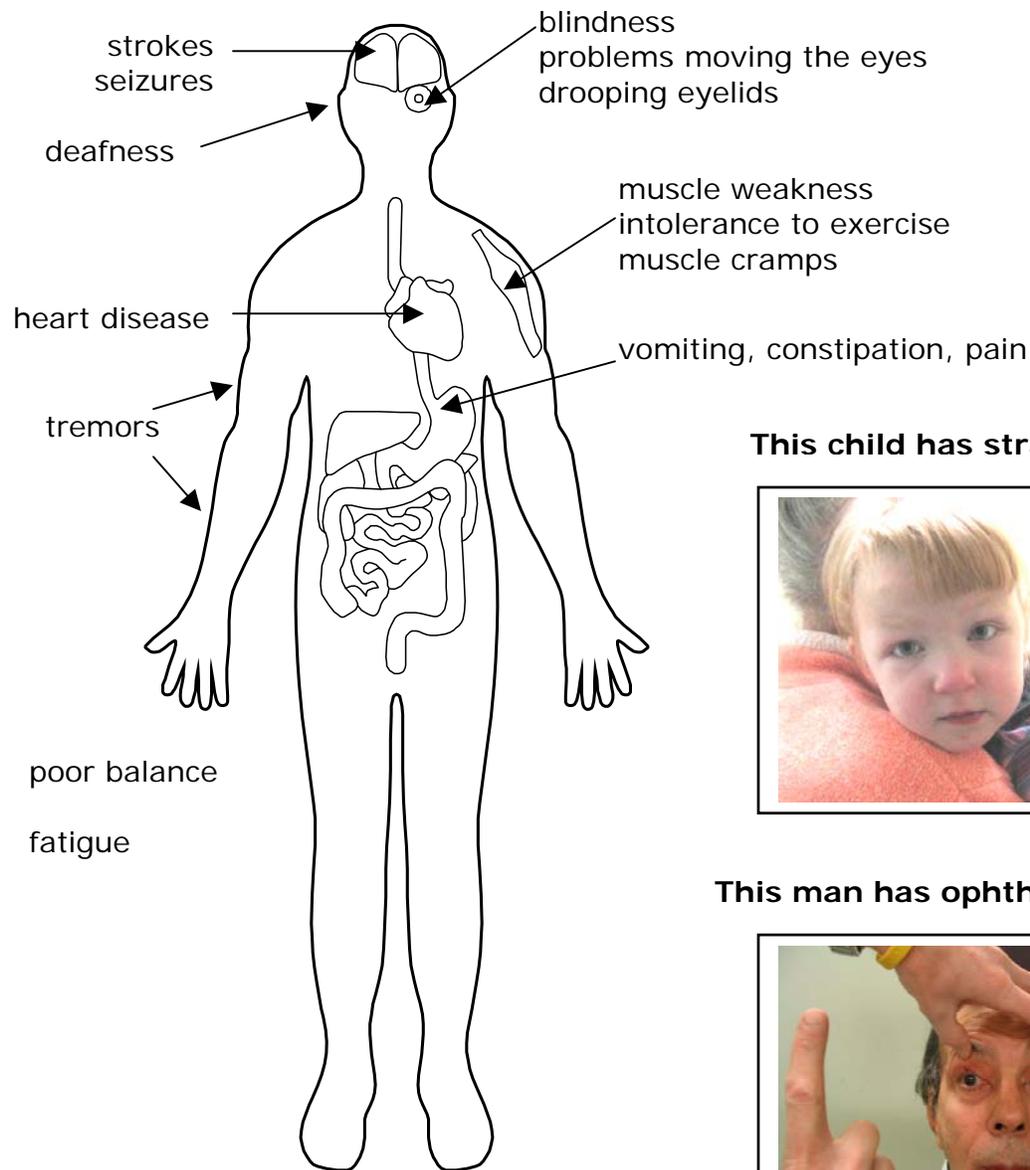
- developmental delay or regression in development
- seizures
- migraine headaches or strokes
- muscle weakness (may be on and off)
- poor muscle tone (hypotonia)
- poor balance (ataxia)
- painful muscle cramps
- unable to keep up with peers (low endurance)
- chronic fatigue
- stomach problems (vomiting, constipation, pain)
- temperature problems from too little or too much sweating
- breathing problems
- eyes are not straight (strabismus)
- decreased eye movement (ophthalmoplegia)
- loss of vision or blindness
- droopy eye lids (ptosis)
- loss of hearing or deafness
- heart, liver or kidney disease at a young age
- parts of the body are shaky (tremors)

Although some of these symptoms are common in the general population, people with Mitochondrial disease are usually affected with multiple symptoms at a young age.

If your child has any signs of mitochondrial disease, see your doctor. The doctor may send your child to a specialist for further examination and testing.

This can be a difficult step as it is the first time you realize that there is a medical problem. But getting help early can improve the chances of your child living a long and healthy life, with as few disabilities or complications as possible. It will also help you to learn as much as you can.

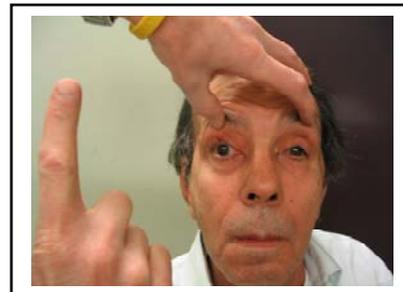
**These pictures show some of the symptoms of mitochondrial disease**



**This child has strabismus**



**This man has ophthalmoplegia**



## How is the diagnosis made?

Mitochondrial disease is difficult to recognize, because there are so many possible symptoms and they can range from mild to severe.

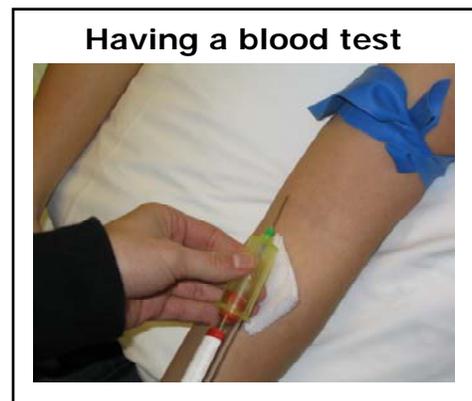
There are some physical signs and symptoms (such as developmental delay, seizures, poor muscle tone, vomiting, vision and hearing loss), which could lead someone to suspect mitochondrial disease.

You may notice one or more symptoms before you recognize that something may be wrong with your child. If you have a feeling that something isn't right, have your child seen by a pediatrician.

If there are physical signs and symptoms or your family history suggests a problem with metabolism, the pediatrician may refer your child to a specialist in neurology or metabolic diseases. At your first visit, the specialist will ask you questions about your child's health and your family's medical history. As well as gathering these facts, the specialist will give your child a complete physical and neurological examination.

A diagnosis of Mitochondrial disease may be confirmed with diagnostic tests, which include:

- Genetic testing – a blood test to check for mutations in the genetic material of the mitochondria.
- Muscle biopsy – taking a tiny sample of muscle to examine under a microscope.
- Magnetic Resonance Imaging and Spectroscopy (MRI and MRS)
- Blood tests for lactate and pyruvate – these enzymes in the blood can help diagnose and track many diseases.
- Urine tests for organic acids, which may indicate problems with metabolism present since birth (called inborn errors of metabolism).
- Forearm exercise testing – to check for the build-up of lactate in the forearm muscles, during exercise with a cuff that restricts blood flow.
- Exercise and muscle strength testing – to check strength and tolerance for physical activity.



Other tests may include:

- a lumbar puncture (spinal tap) to test and evaluate fluid around the brain and spinal cord (cerebrospinal fluid or CSF)
- echocardiogram (ECHO) to check if the heart muscle is affected
- electroencephalogram (EEG) to check brain activity for seizures
- pulmonary function tests (PFT or spirometry) to check how well the lungs are working

## Genetic testing

Genetic testing examines a person's DNA from a sample of his or her blood.

- DNA makes up the genes, which are found on chromosomes. Chromosomes are tiny, thread-like structures inside the nucleus of every cell.
- DNA has the code for a “master plan” of the body. DNA provides instructions for how the body will develop, grow and function.
- There are about 1500 genes making up the mitochondria. Most are “coded” by the nuclear DNA. Only 37 are “coded” by the mitochondrial DNA.

Genetic testing also refers to a careful study of a person's family history to find out how certain characteristics and conditions are inherited (passed on from parent to child).

Genetic testing may be able to identify a genetic mutation for Mitochondrial disease and where that abnormal gene came from. Testing can confirm or exclude a suspected diagnosis of Mitochondrial disease or rule out certain other disorders. When a genetic mutation is identified, we may be able to tell you the probability for passing the disease to future generations. This can help you make decisions about family planning.

**Once a genetic mutation is discovered in a family, other family members may be tested.**

Genetic testing is a complex process and must be done in a specialty clinic such as ours.

The doctor will discuss genetic testing with you. You will need to carefully consider:

- the potential benefits and harms of genetic testing
- whether your child is capable of making his or her own decisions
- what is in the best interests of your child

**The main goal of genetic testing is to promote the well-being of your child.**

If you have a family history of Mitochondrial disease, you may question whether or not to test your child for the disorder. It is important to weigh the pros and cons.

Children under the age of 16 are not usually tested unless they are having problems with their health or growth and development. At this young age, they may not fully understand or be able to make decisions for themselves. It is likely that nothing can be done to prevent someone from getting Mitochondrial disease, and it is unclear whether giving treatment before symptoms appear will be of benefit. Therefore, genetic testing is usually put on hold for children without symptoms, until they are old enough to make their own informed decision.

**As many mitochondrial mutations are not yet known, genetic testing can sometimes leave you without answers and with more questions.**

## **Muscle biopsy**

A muscle biopsy is a procedure to take a small sample of muscle for testing. The sample is usually taken from the muscle on the top of the leg (quadriceps muscle), but may be taken from the upper arm (deltoid muscle).

### **Before:**

- Tell the doctor if your child is taking medication to “thin” the blood (anti-coagulants, aspirin).
- Children under 9 years of age and people with severe developmental delay will be given a sedative to help them stay still during the procedure.
- If sedation is required, the child or adult should have only fluids (nothing to eat) after midnight (the night before) and nothing to eat or drink after 4 am on the day of the procedure.

### **During:**

- The area is “frozen” with an injection of local anesthetic medication.
- The doctor makes a tiny incision (about ½ cm) and uses a needle to take a small piece of muscle, about the size of an eraser at the end of a pencil.
- The incision is closed with a small stitch, which will need to be removed at home in 5 days.
- The total procedure takes about 5 to 10 minutes. It is usually not painful, but your child may feel some pressure.
- The sample is sent to the laboratory for testing. Any extra muscle tissue is kept frozen in case more tests are needed in the future.

### **After:**

- Your child’s leg or arm will be wrapped in a tensor bandage, which can be taken off 1 hour after the procedure.
- The freezing will wear off after about 3 hours. It is usually not painful, but you can give your child acetaminophen (Tylenol) or ibuprofen (Advil or Motrin) for discomfort. Putting an ice pack on the area can also help.
- To prevent a skin infection, clean the area with alcohol each day and keep it covered with a Band-Aid. Your child should not have a bath or swim for 5 days, until the stitch is taken out.

- Your child may resume his or her regular activities when you get home. Regular stretching and movement can help prevent the leg or arm from getting stiff.
- There is a small chance (1 in 3000) of developing a minor skin infection after a biopsy. Signs of infection are: redness, swelling, discharge from the incision and fever - a temperature above 38.5° C (101.3° F). If you notice these signs, take your child to the doctor. An infection may be treated with a medicated cream or antibiotics.

## **Magnetic resonance imaging and spectroscopy (MRI and MRS)**

Magnetic resonance scanning uses a powerful magnet and radio waves to detect signs of Mitochondrial disease in the brain.

### **Before:**

- Children under 9 years of age and those with severe developmental delay may be given a sedative to help them stay still during the procedure.
- If sedation is required, your child should have only fluids (no food) after midnight (the night before) and nothing to eat or drink for 6 hours before the procedure.
- Tell the doctor if your child is afraid of closed spaces (called claustrophobia).

### **During:**

- Your child needs to lie still, on his or her back, inside a tube-shaped chamber for 30 to 60 minutes. If your child is not sedated, you may bring a music CD or DVD video for him or her to listen to or watch, as the machine is quite noisy.
- Your child will be able to hear and speak to the technician during the procedure through an intercom.

### **After:**

- Your child can resume his or her regular activities right away.
- If your child is sedated, you can expect to go home 1 or 2 hours after the procedure.

## **What happens when Mitochondrial disease is diagnosed?**

The doctor will review the result of all the tests with you to confirm the diagnosis of Mitochondrial disease. It may take a few months for these tests and procedures to be scheduled and the results to be ready. You will be informed as soon as possible once the results are available. All test results will be discussed in person during a follow-up visit with your doctor.

Hearing that your child has Mitochondrial disease can be a shock. You may feel scared, sad or angry. All these feelings are normal. You may have mixed emotions and your feelings may change from day to day, even minute to minute. It will help to have support from your family, friends and health care providers at this time. You may have trouble remembering the information that is shared by your nurse and doctor. Always feel free to ask questions during your care or call your doctor if you have concerns or questions afterwards.

After a diagnosis is made the doctor will likely recommend starting a cocktail of vitamins. You will have time to discuss and think about treatment options before making decisions. The health care team will continue to explain the disease and answer your questions as best as they can.

If you are feeling overwhelmed or alone please contact your specialist's office. There are supports and services available to you that will help. There are also other families affected by Mitochondrial disease who are available to listen and talk with.

Clinic staff will always try to be available to help you work through worries or concerns, but we do not provide emergency services. If you have immediate concerns about your child's health (such as breathing problems, heart problems or seizures), you should take your child to the nearest hospital emergency room or call 911.

Your child may see many health professionals and it is difficult to keep track of all that is said and done. We encourage you to keep a file about your child's health. Once the doctor has gone over test results with you in person, ask for a copy for your own records.

## Will I know the type of disease?

There are over 200 known types of this disease. Each is caused by a specific change (mutation) in the genetic material of the mitochondria. When a mutation is discovered, it is given a name and a number. The number describes the position of the mutation on the mitochondrial DNA.

Here are some types of Mitochondrial diseases and the mutations associated with them.

Name of Mitochondrial disease	Mutations
Chronic Progressive External Ophthalmoplegia (CPEO)	mt3243, polymerase gamma, twinkle, ANT
Leiber's Hereditary Optic Neuropathy (LHON)	mt11778, mt14459, mt3460
Mitochondrial Encephalopathy Lactic Acidosis and Stroke-like Episodes (MELAS)	mt3243, mt3271, mt3260
Myoclonic Epilepsy Raged Red Fibers (MERRF)	mt8844
Neurogenic Ataxia and Retinitis Pigmentosa (NARP)	mt8993
Myoneurogastrointestinal Encephalopathy (MNGIE)	thymidine phosphorylase
Leigh's syndrome	mt8993, SURF1, NDUFS1

Hundreds more mutations are possible, but have not yet been discovered.

Most people with Mitochondrial disease never know their specific mutation. This can be upsetting if you are planning a family, as we may not know exactly how the disease will affect future generations.

**The specialist will tell you the name of your child's disease and the specific mutation, if known.**

## How is Mitochondrial disease treated?

In the last decade, advances in medicine and science have helped us better understand, diagnose and treat mitochondrial diseases. Unfortunately, there is still no cure or even a treatment that can guarantee better health or function. The goals of treatment are to ease symptoms, improve function and slow down the progression of the disease.

The effectiveness of treatment varies from person to person, depending on the exact disorder and severity. Generally, those with mild disorders tend to respond to treatment better than those with severe disorders. Treatment will not reverse the damage that has already occurred, such as brain malformations or damage from a stroke.

The doctor will decide what types of treatment and therapy will best benefit your child's specific disease, based on a complete assessment of his or her health.

Many options are available, including:

- physical therapy
- occupational therapy
- respiratory therapy
- speech therapy
- massage therapy
- stretching and relaxation exercises
- exercise training
- counseling
- medications and supplements

### Medications and supplements

Medications are used to treat certain symptoms. For example:

- seizures can be controlled with medications called anti-convulsants
- muscle cramping and stiffness may be relieved with medications called muscle relaxants
- spasticity (tight or rigid muscles that constantly contract) can be eased with medications or injections of Botox<sup>®</sup>

Recent research has shown that several vitamin supplements can help relieve symptoms and improve function. They are often called the “Mitochondrial cocktail”. Your doctor may recommend some or all of these supplements:

- creatine monohydrate
- vitamin C
- vitamin E
- alpha lipoic acid
- co-enzyme Q10
- riboflavin
- thiamine
- L-carnitine
- L-arginine



A program called Inherited Metabolic Diseases (IMD) will cover the cost of these medications for people in Ontario with confirmed Mitochondrial disease. A metabolic specialist can apply to the program for you and arrange to have the mitochondrial cocktail prepared.

The IMD program only works with these hospitals:

- McMaster Children’s Hospital
- The Hospital for Sick Children (“Sickkids”)
- Children’s Hospital of Eastern Ontario (CHEO)
- Kingston General Hospital
- London Health Sciences Centre

Medications are only available from the pharmacies at these hospitals.

If you live more than 1 hour away, you can arrange for the pharmacy to deliver the supplements to your home for a small fee. Ask the specialist to provide a 3-month supply to reduce the number of trips you have to make to the hospital. When calling to get a refill of your prescription, please allow 3 or 4 days for the pharmacist to prepare the medications.

## Exercise and activity

People with Mitochondrial disease benefit from regular exercise. As well as the usual benefits for the body, mind and spirit, physical activity helps their mitochondria work and improves their ability to tolerate exercise. This can improve their quality of life.



In people with certain types of Mitochondrial disease, exercise intolerance is the only symptom. Regular activity can help free them from symptoms, as it builds up their tolerance for exercise.

People with Mitochondrial disease that affects their heart and other organs, also benefit from activity, but need an exercise program designed to meet their needs and abilities.

<p><b>The amount and type of activity recommended for each person varies.</b></p> <p><b>Your doctor can help you decide what activities are best for your child.</b></p>	
--	--

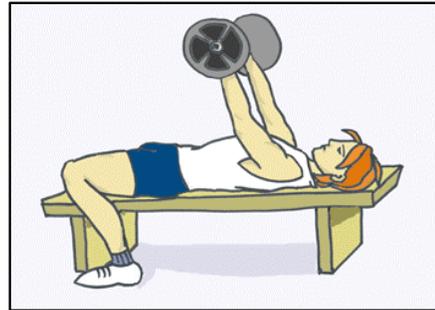
It is best for your child to start slowly. If a particular activity makes your child feel totally exhausted and it takes hours or days to recover, it means the activity is causing more harm than good to your child's muscles. Use how your child is feeling as a guide to balance exercise and rest. Help your child learn to do this by "listening" to his or her body.

Your doctor and physiotherapist can work with your child to design an exercise program, which may include:

- activities such as cycling, walking or swimming
- exercises to strengthen muscles

Older children may learn to strengthen muscles with resistance training. This usually involves lifting weights or using weight machines.

Your child should start slowly and gradually increase activity by “listening “ to his or her body. He or she should take care not to do too much. Becoming exhausted is not helpful and may be stressful.



### **Monitoring the effects of exercise**

As part of your child’s ongoing care, exercise and strength testing may be repeated to keep track of how well his or her body is responding to exercise. If exercise is stimulating the mitochondria to work, the tests will improve. If there isn’t improvement, it doesn’t mean that the treatment isn’t working. It may mean the treatment is working to slow down the progression of the disease.

Strength testing is usually done every year, or more often if your child is losing muscle strength. Muscle strength is tested with equipment such as:

- a Biodex machine, which measures the amount of force put out by a muscle
- a JAMAR tool, which measures the strength of the hand grip

## Helping your child with activity

**Exercise has important benefits for your child.  
Teach your child to make physical activity  
a part of his or her daily life.**

Keep it fun and start off with a wide variety of activities. See what your child likes and what he or she is able to do.

Your child may become more active if you do activities together.

## How can the Neuromuscular and Neurometabolic Centre help?

The Neuromuscular and Neurometabolic Centre is a new program at McMaster Children's Hospital, dedicated to improving the lives of people affected by Mitochondrial disease.

A team of health professionals and support staff, under the direction of Dr. Mark Tarnopolsky, provides care that focuses on the needs of patients and their families.

You and your family will work closely with the members of the health care team. Depending on your needs, you may meet:

- specialized doctors
- developmental pediatrician
- registered nurse
- neuropsychiatrist
- kinesiologists
- EMG technicians
- genetic counsellor
- occupational therapist
- speech and language pathologist
- physiotherapist



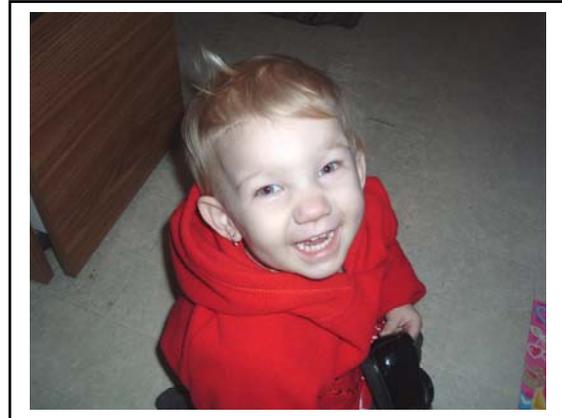
The Neuromuscular and Neurometabolic Centre was established with a generous donation from Warren Lammert and his family. This gift helped us create the first clinic in Canada that diagnoses, treats and cares for people with Mitochondrial disease throughout their life span.

Patients and families benefit from our extensive and ongoing research, cutting edge diagnostic and treatment facilitates and commitment to the highest quality of care.

## What can we expect in years to come?

This is an important question, but one that is not easy to answer. So much depends on the type of Mitochondrial disease and what parts of the body are affected.

We do know that the majority of people with Mitochondrial disease live for years with their disease well managed and their life expectancy unaltered.



However, there is no doubt that your child's diagnosis of a Mitochondrial disease has a significant impact on you and your family. The effects can be:

- physical
- emotional
- social
- spiritual
- financial

Many parents change their jobs because their child needs more time, attention and visits to the doctor and various specialists. As they grow and mature, children with Mitochondrial disease may need to change their life goals, if the disease limits what they can do.

Please talk with your health care team and discuss the options that are available to you. It is not always necessary to change your plans for education, employment and family. We can give you information, support and guidance so that you can make decisions and choices that are right for you and your family's future.

We are sad to say that the prognosis for children who are severely affected by Mitochondrial disease is poor. Mutations such as Leigh's disease and Lethal Infantile Mitochondrial Disease (LIMD) are very severe. Children whose hearts are affected at an early age may develop a thickening of the walls of the heart (called cardiomyopathy) which shortens their life. These children often do not live past the age of 16.

## Hope for the future

Our team and others around the world are continually working to improve the quality and duration of life for people with Mitochondrial disease, by:

- developing better diagnostic tools
- finding better treatments
- discovering more gene mutations
- searching for a cure

With each passing day, more people are being diagnosed, more people are getting the proper treatment and more lives are being restored.



## **How could this disease affect future generations?**

If you or your partner has Mitochondrial disease or a family history of the disorder, the thought of passing it on to your children is one of the most troubling aspects of this disease to deal with. Genetic counseling can help.

### **Genetic counseling**

Genetic counseling involves discussions with an expert in genetic disorders (a genetic counsellor), your doctor and other members of the health care team. They will give you information and support as you consider what might happen if you were to have more children, and make decisions that are right for you.

With autosomal recessive and autosomal dominant inheritance, the probability of a child inheriting the mutation is fairly clear. It is much more difficult to predict what may happen with maternal inheritance. Essentially every child will inherit the mitochondrial DNA from their mother, but the severity of their symptoms is random and variable. Children may not be affected in the same way as their mother and each child may be affected differently.

You may also be offered prenatal genetic testing. This is a good option but can only be done when a specific gene mutation is known within the family.

Prenatal testing checks for genetic mutations in a sample of cells taken from the placenta. These cells are similar to those of the developing baby (fetus). This procedure, called Chorionic Villus Sampling or CVS, is done about 12 weeks into the pregnancy.

Having this test available has helped couples decide to go ahead with planning a pregnancy, even when there is a history of severe mitochondrial disease. It improves their chances of having a baby without the disorder.

Before proceeding with counselling for prenatal testing, you and your partner may want to discuss what would happen if there were an abnormal result.

- Would you be willing to end the pregnancy? If you would not choose abortion, do you really want to proceed with testing, as the results will not change the outcome of the pregnancy.
- How you would prepare and cope with the emotional distress?
- What if the results are not definitive? Would this cause you more anxiety?

We recognize that this is a difficult process for you and your family. We are here to give you support and the best information that is available. Genetic counseling will let you explore the possibilities and make decisions that are best for you.

As a parent of a child with Mitochondrial disease, you may be worried or concerned about the future and whether it would be possible for your child to have healthy children. Individuals with Mitochondrial disease are able to have healthy children, but each person's condition and situation is unique. It is important to discuss your specific situation with your specialist and genetic counselor.

## **Where can we get information and support?**

Anytime you have questions or concerns, please feel free to talk with a member of your health care team. We want you to have as much information and support as you need to make decisions and take part in your child's care.

There will be times when you feel overwhelmed. Remember that you are not alone. We are here to help. There is also a Parent Support Group that includes families affected by Mitochondrial disease living across Southern Ontario. Call the number below for more information.

**The Neuromuscular and Neurometabolic Centre  
at McMaster Children's Hospital**

**905-521-7933**

You can also find information on the internet. If you do not have a computer at home, you can use one at your local Public Library.

Here are some websites that we recommend:

The United Mitochondrial Disease Foundation [www.umdf.org](http://www.umdf.org)

Neuromuscular Disease Center (USA) [www.neuro.wustl.edu/neuromuscular](http://www.neuro.wustl.edu/neuromuscular)

Please remember that information on other websites may not be accurate, complete or relevant to your situation. Talk with your health care providers about the information you find on the web, before taking any action.

## Dictionary

Word	Meaning
Ataxia	Poor balance and lack of muscle co-ordination due to problems with the nervous system.
Chorionic villus sampling (CVS)	A procedure done early in pregnancy to obtain a small sample of cells from the placenta. These cells are similar to those of the developing baby (fetus). The genetic make up of the cells is analyzed to identify errors (called mutations).
Echocardiogram (ECHO)	An ultrasound scan to examine the heart.
Electroencephalogram (EEG)	A test to check the electrical activity of the brain and detect seizures.
Forearm exercise test	An exercise test that involves blood tests during exercise and rest to measure ammonia and lactate levels. Also called forearm ischemic test.
Hypotonia	Decreased muscle tone. Babies with hypotonia seem floppy, like a rag doll, when held.
Lumbar puncture	A diagnostic test to measure the pressure in the spinal column and take a sample of the fluid that surrounds the brain and spinal cord (cerebrospinal fluid or CSF). Also called a spinal tap.
Magnetic resonance imaging and scanning (MRI and MRS)	A diagnostic test using a powerful magnet and radio waves to see inside the body. This test can detect signs of Mitochondrial disease in the brain.

<b>Word</b>	<b>Meaning</b>
Metabolism	The chemical reactions in the body that make energy. As mitochondria make 90% of the energy needed by the body, they are an important part of metabolism. As Mitochondrial disease affects metabolism, it is called a metabolic disease.
Mitochondria	Tiny structures inside almost every cell of the body (except red blood cells) where energy is produced. Almost all of the energy the body needs for life and growth comes from mitochondria. A single one is called a mitochondrion.
Muscle biopsy	A procedure to obtain a tiny sample of muscle for testing.
Mutation	A specific change or error in a gene.
Pulmonary function test (PFT)	A group of tests that measure how well the lungs are working. Tests measure how well the lungs take in and exhale air and how efficiently they transfer oxygen from air into the blood.
Urine organic acids	A urine test to check for metabolic disorders present at birth (called inborn errors of metabolism).
VO <sub>2</sub> MAX	The amount (volume) of oxygen (O <sub>2</sub> ) used when a person is exercising as much as possible (maximum capacity). The VO <sub>2</sub> MAX indicates a person's fitness level. People with Mitochondrial disease have a low VO <sub>2</sub> MAX, even lower than that of people with inactive (sedentary) lives.

## **Acknowledgements**

### **Educational Consultant:**

Lindsay Wizowski, BScN, MEd  
Patient Education Specialist, Hamilton Health Sciences

### **Document production:**

Dianne Tennant  
Document Production Specialist, Patient Education, Hamilton Health Sciences

### **Cover design and illustrations:**

Steve Janzen  
Senior Graphic Designer, Media Productions Services, McMaster University

### **Cover picture:**

Peter Foulds  
Photographer, Hamilton Health Sciences





## Patient Education

© Hamilton Health Sciences, 2010.