

# Understanding Mitochondrial Disorders

Special thanks to Sheila Klywak and Jean Shepherd for their valuable input in the original making of this education pamphlet.

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## What are Mitochondria?

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Mitochondria are tiny, cucumber-shaped “factories” that are responsible for turning the food we eat into **ATP**, a form of energy that can be used by all body cells. There are many mitochondria in all cells of the body.

Although all tissues make and need mitochondrial energy, the areas of the body that rely on it most are the brain, spinal cord, heart, muscles, kidneys, stomach, intestines and endocrine glands (e.g. thyroid and pancreas). This is because these tissues have a large percentage of mitochondria. Some cells like red blood cells have very little or no mitochondria in them. Cells like these are therefore not affected by the mitochondrial disorder.

## What are mitochondrial disorders?

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Mitochondrial disorders are a group of genetic diseases that are caused by defects in the way that mitochondria make energy for the body. The disorders affect the organs and tissues that use the most mitochondrial energy (see above). By their effects on these areas of the body, mitochondrial disorders can cause blindness, deafness, dementia, movement problems, seizures, heart disease, muscle disease, diabetes and kidney problems and can even affect growth and body temperature. **It is important to note that all of these symptoms do not affect everyone; they vary widely among individuals.** Mitochondrial disorders can affect people of all ages, from newborns to adults.

Until fairly recently, scientists knew how mitochondria worked, but did not know how people were affected when mitochondria did not work. It is now believed that mitochondrial disorders may be playing a role in diseases whose cause was previously unknown.

## Helpful resources

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The Adult Metabolic Diseases Clinic  
Vancouver General Hospital  
Diamond Health Care Centre  
4<sup>th</sup> floor, 2775 Laurel Street  
Vancouver, BC V5Z 1C6  
Tel: 604-875-5965  
Fax: 604-875-5967

Mito 411 Support Line  
Tel: 1-888-MITO-411

Mitoaction: Mitochondrial Disease Action Committee  
Email: [info@mitoaction.org](mailto:info@mitoaction.org)  
Website: <http://www.mitoaction.org/>

MitoCanada  
Website: [www.mitocanada.org](http://www.mitocanada.org)

The National Organization for Rare Disorders Inc.  
Website: [www.rarediseases.org/](http://www.rarediseases.org/)

United Mitochondrial Diseases Foundation  
Website: [www.umdf.org](http://www.umdf.org)

*Examples are:*

“I’ll take care of the kids on Friday night so you and Bob can have a night on your own”

“I’m going to the grocery store tomorrow morning. What can I pick up for you?”

“Why don’t you go and lie down and I’ll vacuum the family room?”

“I’ll take Anne shopping so you can have a break”

Concrete offers like these are easier to accept than offers that the person may perceive as you just “trying to be nice”. Also, there is no decision-making involved, something the person you are trying to help may not have a lot of energy for.

- For people with severe physical limitations from their disorder, offer as much “normal” activity as possible. Include them in activities. Don’t treat them as different. Don’t make assumptions about their capabilities based on visible disabilities.
- For people with less severe limitations from their disorder, remember that their energy level will vary greatly during the day. Learn to look for the telltale signs of fatigue such as droopy eyelids, slow movement or memory problems. Expect less when fatigue is showing.
- Offer to give caregivers breaks.

Learn as much as you can about the disorder and help to explain it to others. This not only increases awareness of mitochondrial disease, it helps the person affected to have a break from explaining it over and over again.

### Examples of mitochondrial disorders are:

- Kearnes-Sayre syndrome
- Leigh’s disease
- LHON
- Mitochondrial myopathy or encephalomyopathy
- MELAS
- MERRF
- Congenital lactic acidosis
- NARP

## How do people get mitochondrial disorders?

Millions of years ago, mitochondria were independent creatures. Somewhere along the line, they were adopted into our cells. Mitochondria therefore come with their own set of genetic material, or recipe for new mitochondria. This is called DNA. Mitochondrial DNA differs from the regular (nuclear) DNA found in cells in one important way: mitochondrial DNA is only passed from the mother to baby, whereas, nuclear DNA is passed from the mother or father.

Damage to mitochondrial or nuclear DNA resulting in mitochondrial disorders can be **inherited** or can happen by **chance**. If the disorder is inherited, you should see a doctor who understands genetics to help you to understand just how the disorder can affect you and your family.



## What are the symptoms of mitochondrial disorders?

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Remember that every cell in the body has many mitochondria. Although all tissues make and need mitochondrial energy, the areas of the body that rely on it most are the brain, spinal cord, heart, muscles, kidneys, stomach, intestines and endocrine glands (e.g. thyroid and pancreas). This is because these tissues have a large percentage of mitochondria.

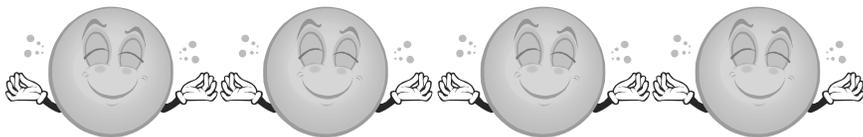


In mitochondrial disorders, there is a mixture of mitochondria in the cells. Some are sick



and some are healthy.

Sometimes the cell makes extra mitochondria to do the work of the sick mitochondria. These extra mitochondria make the energy to make the area of the body work properly.



Think back to the comparison of mitochondria being like factories. Imagine a city that depends on factories to make power. Some of these factories do not make enough power and so others are working overtime. The city has to build new factories to make up for the ones not making power. This keeps the city running smoothly.

## Exercise

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Research has demonstrated that regular exercise is good for people with mitochondrial disease. Resistance or light weight training with some aerobic activity is recommended (daily if possible). Only you will be able to judge how much is too much. Take frequent breaks and start out slow is best.



## Check ups

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Regular visits to the metabolic clinic are needed to consult with the team. Referrals can be made to other specialists to closely monitor the effects of the mitochondrial disease on the body.

## How can friends and family help?

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Understanding that the person with mitochondrial disease is not “crazy” or “lazy” is the best help that you can offer. Recognize and accept that they have physical limitations and may tire easily. Become familiar with how their specific disorder affects them. Don’t make assumptions about their level of physical or mental functioning. Remember that the person is feeling frustrated by how quickly their energy level can change. The person may get angry or very depressed and sad. Sometimes the person may be forgetful or have difficulty with planning activities.

Some ideas...

- Make your offers of help concrete. Instead of saying, “What can I do to help?” or “If there’s anything I can do, let me know”, give clear offers.

## Diet

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Although there is no specific diet that has been shown to help improve the symptoms of mitochondrial disorders, maintaining a healthy diet according to Canada's food guide will help the person with a mitochondrial disorder to stay well.



## Energy conservation

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People with mitochondrial disorders tend to tire easily. It is important for them to pace themselves and to alternate between rest and periods of activity to conserve energy. Sleep is very important. Symptoms may increase when the person is fatigued. Exercise, stress, illness and time of day can cause fatigue.



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Regular visits to the metabolic clinic are needed to consult with the team. Referrals can be made to other specialists to closely monitor the effects of the mitochondrial disease on the body.

The body depends on mitochondria in much the same way. Think of the muscles in the legs. When the mitochondria in muscle cells are not making enough energy for the muscle to work, the muscle becomes tired and weak because the healthy mitochondria are working overtime. This same problem can happen with cells in the brain and other areas of the body that have lots of mitochondria.

Some people with mitochondrial disorders may have a mixture of “sick and healthy” mitochondria in their hearts, others in their eyelids, and others in their leg and arm muscles. Therefore, mitochondrial disorders have a wide variety of symptoms; they depend on the area of the body where the mitochondria “factories” are not working. Even two people with the exact same defect in their mitochondrial or nuclear DNA may not show the same symptoms. This is why family members who have all inherited a mitochondrial defect will not all show symptoms, or not show them to the same degree. Some disorders are not evident until adulthood and some never cause symptoms. Some are so severe that a child does not survive infancy or becomes increasingly behind in development.

## How are mitochondrial disorders treated?

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The symptoms of the specific mitochondrial disorder determine treatment. There is no miracle cure for the diseases. There are, however, different things that may help. The vitamin cocktail for mitochondrial disease is a combination of vitamins, cofactors and antioxidants that may help to improve energy production in the mitochondria. It may take up to three months to see any improvement in symptoms from any one of these supplements.

These supplements help symptoms in some people but not in others. They will not eliminate symptoms nor do they alter the progression of the symptoms over time.

# Vitamin cocktail

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## 1. Coenzyme Q 10, Vitamin C, Vitamin D, Multi-vitamin without iron

Coenzyme Q 10 helps to make energy in the mitochondria of the body. It also acts as an “antioxidant”. Oxygen free radicals are charged oxygen molecules that can destroy the walls of the cells. Coenzyme Q 10 acts as a “buffer” to protect the cell from these free radicals.



**Vitamin C** is important in the growth and repair of bones, teeth, skin and other tissues. It has many other roles in the body: increases your body’s absorption of iron from plant-based foods; helps to prevent cell damage by acting as an anti-oxidant and may reduce your risk for certain cancers and other chronic illnesses; and protects you from infections by keeping your immune system healthy. In mitochondrial disease, vitamin C has been shown to make Coenzyme Q10 work better in the body, helping to increase its benefit in enhancing mitochondrial function.

**Vitamin D** is best known for its role in keeping bones healthy. It is also involved in fighting infections, keeping blood pressure within a healthy range and helping control insulin production. Recent research suggests that vitamin D may also have benefits in preventing some types of cancer, especially colorectal cancer. It may help prevent diabetes and multiple sclerosis as well.

Some groups of patients, especially those with chronic diseases who tend not to eat well are at higher risk for vitamin deficiency and sub-optimal vitamin status. Taking a **multivitamin** daily may reduce the risk of developing other chronic diseases including coronary artery disease, cancer, and osteoporosis and help to meet your general nutritional requirements. We do not recommend iron supplements unless you have had a blood test that shows you are anemic.

## 2. Creatine

Creatine is a substance that is naturally made by our bodies. It is an amino acid (amino acids are the building blocks of protein) made by the liver and kidneys. Creatine is also derived from the diet through meat and animal products. Creatine is also sold as a nutritional supplement. In the body, creatine is changed into a molecule called “phosphocreatine” which serves as a storage reservoir for quick energy. Phosphocreatine is especially important in the muscles and nervous system, which periodically need large amounts of energy.

## 3. Riboflavin (vitamin B2) and Thiamine (vitamin B1)

Thiamine and riboflavin are essential in energy production in the body. Both vitamins act as antioxidants, protecting the body cells from free radical damage. One possible cause of migraine headaches is a reduction in energy production within the mitochondria of cerebral blood vessels. Riboflavin has been shown to decrease the incidence and severity of migraine headaches by increasing energy production in the mitochondria.

## 4. Alpha-Lipoic acid

Alpha lipoic acid helps to make energy in the mitochondria of the body and also acts as a powerful antioxidant. Oxygen free radicals are charged oxygen molecules that can destroy cell walls. Alpha lipoic acid acts as a ‘buffer’ to protect the cell walls from these free radicals.