

Mitochondrial Disease

A Guide for the Newly Diagnosed



A tool kit for patients, their families, &
caregivers

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Introduction

This guide is designed for those recently diagnosed with mitochondrial disease. It was developed to provide you with support and guidance during an often-challenging time for patients and their families when facing a new diagnosis. It is a step toward finding the help you need and planning the next steps you need to take.

This guide will help you learn more about:

- The basics of mitochondrial disease
- How mitochondrial disease may impact your family
- Finding strategies and resources for care
- Finding support so you do not feel alone or isolated

Learning that you or your loved one has been diagnosed with mitochondrial disease is a powerful moment in your life. Suddenly life may feel very different from what you expected it to be. You may feel relief at finally having found an answer. But, worries may also arise about your family and what this diagnosis will mean for their life experiences. You may worry about how you and your family will adjust to this in the years ahead. You may worry about the day-to-day challenges of caring for you or your loved one. You are also likely faced with a tidal wave of information. Getting emotional support and accurate information to help you cope and promote a positive future for you and your family will be critical during this period.

As you read this and other information sources, please note that there is not just one mitochondrial disease. In fact - there are hundreds. While there are similarities between the different mitochondrial diseases and the types of medical problems they cause, each one is quite unique in how it unfolds and the severity of medical problems that it causes. **Not all mitochondrial diseases act the same way or affect the person with the same severity.** Even within a family, two people with the same disease may have different symptoms and different severity of those symptoms. Please keep this in mind as you learn more about mitochondrial disease. Everything you read may not apply to you or your family - or may not apply for many years to come.

Please be aware, this information is up to date to the best of our abilities and changes will be made over time to try and keep this as up to date as possible, however mitochondrial medicine is always evolving as we learn more about this condition. As always, please discuss all medical concerns and potential changes to your medical care with your medical team.

Mitochondrial Disease

What is genetic mitochondrial disease?

Mitochondrial diseases are genetic, or DNA based conditions where the body's mitochondria do not work well.

Mitochondria use the food we eat and the oxygen we breathe to create energy (also known as ATP) for our cells. They are like power plants or batteries for our cells. They also have other important roles including controlling how long a cell lives and decreasing the number of unstable molecules (reactive oxygen species) in a cell, which can cause damage to your DNA.

When mitochondria do not work well, the cell cannot get enough energy to function effectively or may not live as long as it should. Cells in organs that require the most energy often struggle first, which can lead to those organ not performing their jobs – causing medical symptoms to arise.

Symptoms can begin at any age and are sometimes triggered during a time of medical illness or when undergoing a medical procedure that might be hard on the body. It is not just a pediatric disease, and some individuals may show no or very few symptoms until they are adults.

[More information is available in a booklet at this link](#)

How common is mitochondrial disease?

Recent estimates are that 1 in 5,000 people have mitochondrial disease. It affects males and females equally. To better understand how common mitochondrial disease is, further research is being done.

What are the symptoms of mitochondrial disease?

Mitochondrial disease can affect multiple parts of the body - especially those parts that use a lot of energy, such as the brain, muscle or nerves. With most mitochondrial diseases, no one symptom alone makes a diagnosis of mitochondrial disease, but it is how symptoms present in combination with each other or how they develop over time.

The nervous or neurologic system is often involved, and problems can occur with how the brain, muscle, and/or nerves work. These symptoms include:

- Developmental delays in an infant or child
- Loss of brain function at any age
- Strokes, where the brain gets injured due to a lack of energy, may occur at a young age
- Low body tone (hypotonia)
- Muscle weakness (myopathy)
- Imbalance, with or without falls (ataxia)
- Trouble with how the nerves send signals to the brain (neuropathy)

Other symptoms throughout the body may include:

- Problems with moving the eyes (ophthalmoplegia)
- Drooping of the eyelids (ptosis)
- Poor health of the retina of the eye (retinopathy)
- Sudden onset loss of central vision (optic neuropathy)
- Hearing loss due to poor health of the auditory nerves (sensorineural hearing loss)
- Hormonal deficiencies, including of thyroid or parathyroid hormone
- Diabetes mellitus
- Thickening of the heart muscle (cardiomyopathy)
- Heart rhythm disturbances (arrhythmias)
- Slower bowels and gut motility (dysmotility)
- Poor weight gain (failure to thrive)
- Short stature
- Physical and mental fatigue (though this symptom does not exist in isolation)

What causes mitochondrial disease?

Primary mitochondrial diseases are genetic conditions. This means that the DNA instructions in a person's body that tells our body how to make the mitochondria and keep them healthy has a spelling change. These DNA spelling changes that cause our bodies not to work properly are called "mutations" or "pathogenic variants."

The mitochondria use two types of DNA instructions - nuclear DNA that comes from each of our parents and a special mitochondrial DNA that is primarily "maternally inherited," i.e. comes from the egg at conception. These two DNA instructions work in combination to make and maintain healthy mitochondria. Variants in either DNA instruction can cause mitochondrial disease, with approximately 20% being caused by changes to our mitochondrial DNA. Variants can be inherited from a parent or may be a new change that occurs in an individual for the first time.

There are hundreds of types of mitochondrial disease and so each type may look quite different from another one. It is important to note that while DNA testing is available, we are not yet always able to find a genetic cause for everyone's mitochondrial disease.

What else can look like mitochondrial disease?

Many other neurologic and genetic disorders can mimic mitochondrial disease. Some symptoms that raise the concern for a mitochondrial disease can also occur due to conditions such as infections, rheumatologic or autoimmune diseases. Some of these other causes can even lead to findings of the mitochondria looking unhealthy on testing. This is called "secondary mitochondrial dysfunction." A comprehensive and thorough evaluation is needed to ensure that these conditions that overlap with symptoms and/or mitochondrial dysfunction are not the underlying cause of you or your loved one's symptoms.

This is one reason why a diagnosis of mitochondrial disease is often not provided until an underlying genetic cause is found.

What is not mitochondrial disease?

Having findings in tests of blood, urine, skin, saliva, buccal swabs (cheek swabs), or muscle suggesting that the mitochondria are not healthy is not enough to diagnose a mitochondrial disease. There are hundreds of causes of abnormal mitochondrial function that are not primary genetic mitochondrial diseases.

A mitochondrial expert can help you and your family make sense of the results.

How is mitochondrial disease diagnosed?

A concern for mitochondrial disease is generally raised by neurologists or geneticists. On occasion, other types of specialists such as a cardiologist, endocrinologist or ophthalmologist may raise this concern. They may be able to perform testing that helps lead to a preliminary or confirmatory diagnosis.

Testing is typically obtained through blood and/or urine samples to see if there are signs that the mitochondria may not be healthy. This testing often includes blood levels of amino acids, acylcarnitines, lactate, pyruvate, and urine organic acids. These tests do not always show a problem. They can also sometimes show problems for reasons other than mitochondrial disease. If there is a high concern of primary or genetic mitochondrial disease, DNA testing (genetic testing) is needed. If DNA testing is not confirmatory, additional specialized tests may be obtained in a small piece of muscle, typically taken from the thigh (muscle biopsy).

There are also mitochondrial disease specialists that can help make or confirm the diagnosis. A mitochondrial disease expert to help with diagnosis and testing can be found through the [Mitochondrial Care Network](#). Formal diagnosis guidelines are available for your medical professional [online](#) at bit.ly/mitodiagnosis.

Why is a genetic diagnosis of mitochondrial disease needed?

A genetic diagnosis of mitochondrial disease is helpful for several reasons.

There are many different types of mitochondrial diseases - each one having a different set of symptoms and prognosis. Knowing exactly which mitochondrial disease a person has can help a family better understand what to expect over time and can help their doctors better plan the medical care needed.

Confirming a diagnosis of mitochondrial disease can also help ensure that another, different condition is not being overlooked and end the need for more diagnostic testing.

Finding the genetic diagnosis can be especially important to help you and your family learn if the condition might run in the family and whether other family members, including siblings or parents, may need to be concerned about the condition. A genetic diagnosis can also help with family planning, if future children may have a chance to also be affected and sometimes if testing the condition prior to or early in pregnancy is an option.

Lastly, having a genetic diagnosis may create the opportunity to be in research studies, including drug treatment studies, which require a DNA-test confirmed diagnosis in order to participate.

What to do if you or your loved one do not have a genetic diagnosis

Mitochondrial disease is often difficult to diagnose. If experienced physicians are involved, a diagnosis can sometimes be made through a combination of clinical features, laboratory evaluations, imaging studies, and/or tissue (often muscle) biopsies. Despite the advances in technology and knowledge, many people still do not receive a specific diagnosis.

If thorough genetic testing is negative, it is important to revisit genetic testing in the future as well as consider that you or your loved one's symptoms may be caused by another condition. Genetic testing technology is advancing quickly, and testing options continue to improve and so genetic testing may be negative/not conclusive now, but in 3-5 years' time may be able to find a diagnosis. When clinical testing does not confirm a diagnosis, research studies may also help to provide additional information.

Other words for mitochondrial disease

Over the years, many different labels have been used for mitochondrial disease. These labels have included: "mitochondrial cytopathy," "mitochondrial myopathy," and a listing of individual disorders by the changes found on muscle biopsy tests such as "Complex One Deficiency." These labels are used less and less. These other terms are no longer preferred, and the expert medical community is trying to not use them anymore, because they often mean different things to different people.

Historically, "possible" mitochondrial disease was used by clinicians when a mitochondrial disease was suspected, but the genetic testing did not confirm the diagnosis. However, there are many conditions that can look very similar to, and mimic, mitochondrial disease.

In instances when a genetic diagnosis cannot be confirmed, using a diagnosis of "possible" mitochondrial disease may feel like the right thing to do but may potentially harm patients and their families, creating anxiety, delaying finding the actual diagnosis, and leading to incorrect management and care. Presently, a label of "diagnosis uncertain," together with a specific description of the symptoms or testing results identified, is preferred when a mitochondrial

disease cannot be genetically confirmed. More information about this for your physician is available online at bit.ly/possiblemito.

What about other family members or future children?

Depending on the type of genetic mitochondrial disease, other siblings or future children may be at-risk for developing the condition. If a genetic diagnosis of mitochondrial disease has been made, your medical provider and a genetic counselor can help you better understand if other family members are at-risk of developing symptoms or passing on the condition and may consider testing. In regard to future children, a genetic counselor can better guide you as to options your family may have available for pre-conception or prenatal testing.

You, Your Family, and Mitochondrial Disease

Caregivers and Stress

A diagnosis of mitochondrial disease can provide relief, but also feel overwhelming and scary. Having a diagnosis will allow you or your loved one to end the need for more diagnostic testing and focus on treatment. However, a diagnosis of mitochondrial disease may raise the concern of ongoing or worsening health problems and the need for more medical care.

Like any major event in your life, this diagnosis may impact everyone in your family. Below are examples of new feelings you or your family may encounter:

- May feel overwhelmed with the everyday responsibilities
- May feel alone or isolated, as others may not understand the disease
- May feel pressure to become an expert on mitochondrial disease and learn everything overnight
- May worry about you or your family's future, especially with schooling, school friends, and the ability to work and live independently
- May have trouble maintaining friendships and keeping up with social activities
- May have difficulty finding balance and time to manage household tasks, other children, daily activities, etc.
- May feel less inclined to share feelings
- May become stressed about finances, health insurance, and the unknowns of the situation
- May find pre-existing mental health conditions worsening or may develop new ones due to this life change
- May feel grief at the loss of the ability to do previous activities or the loss of your imagined life plan
- May develop other problems such as stress and anxiety that can affect work life

If you or your family members are experiencing several of the following signs and symptoms over a period of time, you may want to seek professional counseling:

- Trouble sleeping
- Difficulty controlling or stopping negative thoughts
- Loss of appetite or enjoyment in activities that previously brought you joy
- Irritability, short-temperedness or more aggression than usual
- Consuming more alcohol than normal or engaging in other reckless behavior
- Thoughts that life is not worth living

If you or your loved one are having trouble working through these emotions or feel unable to function in usual ways, it may be a good idea to seek professional help. Professional help can prevent serious problems from developing in the future. It can help to empower us to face challenges more effectively.

Finding support

There are many organizations that can help guide your way. Some are specifically dedicated to support rare disease patients and families. These organizations and their staff offer information, guidance, and resources. Most of the organizations offer newsletters to keep you up to date regarding advances and new knowledge in the field, as well as forums for discussions. Many hold regional and national symposiums with families and clinicians/scientists.

Mitochondrial Disease Support Networks

There are many patient advocacy organizations focusing specifically on mitochondrial disease. The **United Mitochondrial Disease Foundation** (umdf.org) and **MitoAction** (mitoaction.org) are two of the larger groups that focus on all types of mitochondrial disease and provide many support and educational services. There are others that focus on a particular disease type, for example **Cure Mito Foundation** (curemito.org) and **People Against Leigh Syndrome** (peopleagainstleighs.org) for Leigh Syndrome or **Champ Foundation** (thechampfoundation.org) for Pearson Syndrome.

Rare Disease Support Networks

There are many other support networks for families. The **Courageous Parents Network** (courageousparentsnetwork.org) provides resources to help parents and providers caring for seriously ill children. **Global Genes** (globalgenes.org) is an organization that provides information to those affected by rare diseases in general and **ANGEL AID** (angelaidcares.org) provides a variety of mental health and wellness services to rare disease families including rare mothers' wellness retreats and online support groups facilitated by professional psychologists or therapists.

Social Media and Online Support Groups

Many online platforms (such as Facebook) have groups that have been specifically created for mitochondrial disease. While these groups can be a way to connect to a larger community, it is important to note that in the vast majority of cases, such groups are not monitored by medical professionals, and it is important to exercise caution when asking for medical advice or sharing you or your family's medical reports (including genetics) in public forums. Even if a group or

page is “private” or “closed,” any information put in an online group should be viewed as publicly available.

When joining support groups, it is important to remember when comparing your experiences to others that there are hundreds of different types of mitochondrial disease and causes of mitochondrial dysfunction. Support groups can help you advocate for you or your loved one, find care, services, resources, learn how to develop a good care team of local medical and other resources, and much more. Some advice you read about may not be relevant for your family’s type of mitochondrial disease and in some cases may even cause harm. You may find some individuals who are extremely vocal and passionate about their personal medical experiences, but may be sharing advice that does not apply to experiences of others with mitochondrial disease.

As you navigate these forums, please be discerning, ask questions, learn, and always confirm medical advice with your healthcare provider. Be mindful to protect you and your family’s privacy on social media when sharing about your journey with mitochondrial disease. Your medical team should always remain as your first choice to ensure that you have the information that applies to you or your family member’s specific type of mitochondrial disease.

Palliative Care

Palliative care can be another helpful resource. There is a misconception that palliative care is only for individuals who are dying or at the end of their lives, while in reality palliative care exists to help those living with a serious illness. Palliative care is meant to enhance a person’s current care by focusing on quality of life for them and their family. UMDF, MitoAction, and the Courageous Parents Network offer helpful resources and/or videos on Palliative Care.

Getting Care

Finding a Provider

Depending on the expertise of the provider you or your family are currently seeing, you may find that they feel overwhelmed or underqualified to treat mitochondrial disease, and so you may need to explore finding a new physician to see.

Most hospital systems have a Genetics department. Physicians that specialize in Genetics often see both pediatric and adult patients. Geneticists learn about diagnosing and managing mitochondrial disease as part of their training. Pediatric and Adult Neurologists may also be knowledgeable about mitochondrial disease. They have all learned about mitochondrial disease in their training, though some specialists may be more knowledgeable than others.

Certain physicians specialize in caring for mitochondrial disease and may run a mitochondrial disease center. Some mitochondrial disease centers have joined a collaborative network known as the Mitochondrial Care Network (MCN). You can find an MCN site close to you by visiting the website: mitonetwork.org.

Assembling your Medical Team

The team of specialists you need depends on the type of mitochondrial disease you or your loved one have, and the specific symptoms being experienced. Your or your family member's geneticist or mitochondrial expert can help provide information as to which doctors need to be part of the care team. One of these physicians may act as a point person to help you and your loved one coordinate medical care with all of these specialists.

Mitochondrial disease centers usually have a core team of physicians knowledgeable about mitochondrial disease already organized. They can help coordinate medical care and help advise local physicians should questions in care arise.

A good primary care physician that is open to caring for patients with complex medical needs and working with multiple specialist providers is also strongly recommended.

If you live in a state where no expert mitochondrial disease center is available but you have a local physician who is willing to help you and your family learn more about mitochondrial disease, they can reach out to the [Mitochondrial Medicine Society \(MMS\)](#) or [Mitochondrial Care Network \(MCN\)](#) for more information.

Communicating with your medical team

You will likely find your decisions and approach to medical care is guided not only by medical information you receive, but also by your and your family's beliefs, values, preferences, and priorities. If there are considerations that are important to you for your medical team to know, it is best to communicate these from the start. Additionally, sometimes after receiving difficult news, it may take some time until you're ready to ask questions and process more information. If you feel overwhelmed or need some time to think the information over, it is important to say so. The following guide from Courageous Parents Network may help: [CPN-Family-Meeting2.pdf \(courageousparentsnetwork.org\)](#)

There are many tools to help you keep records and better prepare yourself for doctor appointments. The following are two resources to help you manage your medical journey with mitochondrial disease:

- **MitoAction Mobile** (https://www.mitoaction.org/mobile_new/) – an app that allows you to track your symptoms, medications, doctor's appointments, etc.
- **mitoSHARE** (<umdf.org/mitoSHARE>) – an online registry stewarded by the UMDf that includes tools to track symptoms, medications, doctor appointments, etc.

Symptom Management

Treatment

Mitochondrial disease care consists of providing preventative care and managing ongoing symptoms. Appropriate clinical care should be available to everyone - especially through the Mitochondrial Care Network. There are many things that can be done in terms of clinical care to

optimize a person's quality of life and ensure that certain medical problems that may develop with mitochondrial disease do not turn up unexpectedly.

Staying as active and mobile as possible is important for mitochondrial health. Your care team may involve physical, occupational, speech, or other types of therapists to help with this. Routine exercise is also important, and therapists can help develop a personalized exercise regimen.

Healthy nutrition is needed for good mitochondrial health and a nutritionist is often consulted for guidance. Currently there is not a special diet to follow, but many clinicians believe that eating a healthy and balanced diet is optimal.

Certain vitamins or supplements, such as folic acid and L-arginine or L-citrulline, might be used depending on the type of mitochondrial disease one has to combat some of the effects mitochondrial diseases has on the cells.

There are many other vitamins and supplements that have historically been used to attempt to help mitochondrial function. Commonly called the "mito cocktail", there is no scientific proof that any component of them is effective for everyone with mitochondrial disease. The managing mitochondrial physician can decide which vitamins and supplements to take based on the individual's specific underlying genetic disease or blood work. Due to the lack of strong scientific evidence, there is a lot of variability in which supplements are used in the mitochondrial disease community depending on the medical team you or your family see. In addition, insurance does not typically reimburse the expense for these supplements so this must be taken into consideration. More information about the vitamins and supplements historically used for mitochondrial disease treatment is available [online](https://bit.ly/mitosupplements) at bit.ly/mitosupplements.

New medications and therapies to slow down or stop the progression of mitochondrial disease are being developed and currently undergoing research trials, but none are available at this time.

Prevention

Mitochondrial disease is often progressive, involves other parts of the body, or presents with new symptoms over time. A key part of mitochondrial disease care is monitoring one's health status with a regular evaluation by a provider and periodic lab work and tests. Such monitoring will help ensure that any new symptoms or problems are picked up and treated by the medical team as early as possible.

For example, some individuals with mitochondrial disease may develop diabetes or thyroid disease over time. Routine monitoring for both conditions is often performed.

Guidelines for local physicians to assist in preventative care and medical management are available online at bit.ly/mitocare.

Diet & Nutrition

It is not yet known if one specific type of diet is optimal for mitochondrial health. A healthy diet - one rich in fruits, vegetables, grains, complex carbohydrates, proteins, and healthy fats (such as from nuts and olive or canola oil) may be optimal. Avoiding food with saturated fats, preservatives, and chemical additives may also be beneficial.

It is not yet clear if a high amount of carbohydrates, fats, or proteins are preferable - though some individuals may respond better to an increased amount of one of these macronutrients in the diet. In general, a very restricted diet is not preferred and special diets such as a ketogenic or Atkins/low carbohydrate diet may not be tolerated and can sometimes make symptoms worse.

Some individuals, but not all, may respond better to small, frequent meals and avoiding fasting. A multivitamin, specific vitamin (such as vitamin D), or iron supplementation may be needed.

Working with a dietitian is recommended to optimize one's caloric intake and ensure that no macro- or micronutrient deficiencies develop. This is especially needed should there be any need for specialty feeding, such as via a gastrostomy tube, or if there is difficulty gaining or maintaining weight.

Exercise & Therapy

Fatigue and exercise intolerance are frequent symptoms of mitochondrial disease. Being involved in an exercise program can help decrease some of these symptoms or potentially keep them from getting worse. Exercise can also help the body and muscles generate more healthy mitochondria.

An evaluation by a physical therapist is generally recommended before starting an exercise program to ensure that it is safe, and the degree of difficulty increased at an appropriate rate. A short period of exercise may be all that is initially tolerated. More rest intervals between exercise days may be needed. Increases in exercise programs should be made more slowly than in other medical conditions.

Not all physical therapists are familiar with mitochondrial disease, but they can develop a program used for individuals with neuromuscular weakness. It is important to find a physical therapist that is receptive to your differing needs and can adapt accordingly.

Before starting any exercise, it is important to check with your doctor to see if any precautions or testing are needed. A baseline electrocardiogram (ECG) and/or echocardiogram are sometimes obtained to check on your heart function. Some mitochondrial patients are less efficient in how they use oxygen for energy during exercise, which can limit their activity level. If this is felt to be the case, a formal exercise test, called cardiopulmonary exercise test, or CPET, may be needed.

Starting slowly in intensity and length of exercise, with slow progression in difficulty is very important. A combination of aerobic exercise and resistance training (weightlifting) can be beneficial for the mitochondria. Listening to one's body is very important and extra rest should be taken for fatigue and soreness. Exercising on a reclining bicycle or in a therapy pool might be better tolerated and safer - especially if there is muscle soreness, pain, or balance problems. Exercise should occur in a neutral temperature (not too hot or cold) and never when sick (fever, cold symptoms, infection, etc.) or while fasting.

Children may benefit from modified/adaptive physical education class in school with permission to self-regulate how much they are exercising. They may have formal physical therapy instead of gym class based on their individualized plan at school.

More resources on exercise can be found at these links:

<https://www.umdf.org/ask-the-mito-doc/>

<https://www.mitoaction.org/resources/exercisenutrition/>

<https://www.mito.org.au/resource/exercise-and-mito/>

<https://vch.eduhealth.ca/PDFs/FA/FA.500.M697.pdf>

Medication Contraindication

It was previously believed that many medications needed to be avoided in individuals with mitochondrial disease. This is not the case. While many medications may impact mitochondrial function, especially in a research setting, they are generally well tolerated when used in routine care. Caution is still needed with certain medications when used for the first time. Unfortunately, there are many older lists on the internet that caution against a wide range of medications. As we have gained more knowledge about mitochondrial disease, these lists have become outdated. For new and updated guidelines, please ensure your medical team visits the [Mitochondrial Medicine Society website](http://mitosoc.org) (mitosoc.org).

Valproic acid (brand names Depakote, Depakene) is the one medicine that should not be used in most cases of mitochondrial disease, or only used when there are no other options. There are certain mitochondrial diseases such as *POLG*-related disease and mitochondrial diseases with liver involvement where it is absolutely contraindicated.

Some mitochondrial diseases may cause a sensitivity to general anesthetics and an anesthesiologist should be notified that the person has mitochondrial disease, as the person may need a lower or adjusted dose of the anesthetic medication. Certain anesthetics, such as propofol, should be used for short procedures or induction of anesthesia only. While use for a short period of time is often tolerated with no difficulties, a long period of use may lead to worsening of acidosis and so is generally avoided.

The antibiotics that belong in the aminoglycoside family (such as gentamicin, tobramycin, neomycin, and paromomycin) may need to be avoided in those with certain types of mitochondrial DNA (mtDNA) changes, but not most individuals with mitochondrial disease.

Certain medications, while not contraindicated, have the risk of worsening lactic acid levels. Some additional monitoring may be needed in these situations if medications such as topiramate or metformin are used. Others, such as statin medications, can sometimes worsen muscle weakness, even in those without mitochondrial disease. A person's symptoms should be tracked when taking this medicine to see if a problem develops.

If a medication, once tried, creates a specific problem - it should only be used again with caution, whether it is in this list of medications or not.

Emergency Care

Individuals with mitochondrial disease are vulnerable to a decline in health during times of "metabolic stress." Metabolic stress happens when the body needs more energy than normal during an illness, infection, fever, dehydration, starvation, surgery, or during other stress. Your mitochondrial team will provide you or your loved one with an individualized plan of care for these times to try and help prevent problems from developing. There are certain precautions that one might have to take especially when undergoing a medical procedure or fasting for a prolonged period.

Individualized care plans may include protocols for an emergency room visit or hospital admission with a high fever or one is not able to eat and drink well. There may be a list of treatments to receive at that time and a list of medications that should be avoided. The plan may include lab work that needs to be obtained when ill.

Some patients carry an "Emergency Letter", a protocol that the Emergency Department can use to help guide them in obtaining lab work and tests and providing care. There is no one-size-fits-all Emergency Protocol, and this letter should be customized to you or your family by your medical team and signed by your treating providers. Such letters can be placed into your local hospital's electronic medical record for easier access to the medical team during emergencies.

Surgical Care

Individuals with mitochondrial disease are sometimes vulnerable to a decline in health during the added stress of surgery and when receiving anesthesia. Your mitochondrial team will provide an individualized plan of care for these times to try and help prevent problems from developing.

Such plans may include a hospital admission beginning the day before and the day of surgery to receive intravenous fluids containing dextrose, a type of sugar. Your mitochondrial team might want to discuss the procedures and the precautions to be taken with the surgeon and the

anesthesiologist. The plan may include lab work that needs to be monitored during the admission.

Getting Services

After you or your family member has been diagnosed with a mitochondrial disease, you likely will wonder how to meet you or your family's needs. You might face feelings of guilt, isolation, confusion, and anger. These are common feelings; the key is to deal with them constructively by getting help if needed, addressing your concerns, and learning more about mitochondrial diseases and the challenges to come. On top of a myriad of doctors' visits, technological jargon, labels, and a mountain of information to process, you need to learn quickly to navigate a complicated system to figure out what services and treatments you or your loved one needs and how to get them. Some individuals may have at home support helpful, such as a personal care attendant (PCA), and some may end up deciding to apply for disability funding or housing. There are many available resources, both medical and financial, to help meet some of the needs for you and your family.

This section is still under development and will be updated over time. For now, a great resource on services and insurance is available on the Global Genes website:

[NAC RareCaregivers Guidebook FINAL.pdf \(caregiving.org\)](#).

Clinical Research

To better understand mitochondrial diseases and help improve how we diagnose, treat, and manage children and adults affected by them, clinicians and scientists may perform clinical research.

Understanding and Participating in a Mito Clinical Trial

These studies may involve simple observation, may involve testing, or may involve trialing various treatment options on individuals affected by mitochondrial disease. If you are interested in learning more about this type of research or are interested in participating in a clinical trial, you may visit these websites to learn more:

- The government funded website listing all ongoing clinical trials: <https://clinicaltrials.gov/>
- The Mitochondrial Care Network Research (MCN): <https://www.mitonetwork.org>
- The North American Mitochondrial Disease Consortium (NAMDC): <https://www.rarediseasesnetwork.org/cms/namdc>
- The United Mitochondrial Disease Foundation (UMDF): <https://www.umdf.org/clinical-trials/>
- MitoAction: <http://www.mitoaction.org>
- National Organization for Rare Disorders (NORD): <https://rarediseases.org>
- GlobalGenes.org: <https://resource-hub.globalgenes.org/kb/article/306-making-informed-and-shared-decisions-about-genetic-testing-and-clinical-trial-participation>

Patient Registries

Patient and caregiver participation in research is crucial for helping advance our understanding and treatment of mitochondrial disease.

A relatively easy way to participate is by enrolling in a mitochondrial disease patient registry. A patient registry is a collection of information about patients who have a specific disease or condition. At this time, there are a number of different registries for mitochondrial disease patients. Some are run by medical organizations (NAMDC) and others are run by patient advocacy organizations. (*Note – the following is not inclusive of all registries*)

Mitochondrial Disease Registries:

- North American Mitochondrial Disease Consortium (NAMDC) – clinician-populated patient registry: <https://www.namdc.org>
- mitoSHARE – worldwide patient-populated registry stewarded by the United Mitochondrial Disease Foundation: umdf.org/mitoSHARE

Disease Specific Mitochondrial Disease Patient-Populated Registries:

- Leigh Syndrome-Specific Patient Registries:
 - CoRDS: curemito.org/cords – by Cure Mito Foundation
 - TriaLS: peopleagainstileighs.org/registry – by People Against Leigh Syndrome (PALS)
- Pearson Syndrome Patient Registry:
 - Champ Foundation Registry: cfr.thechampfoundation.org – by the Champ Foundation
- Leber Hereditary Optic Neuropathy (LHON) Patient Registry:
 - lhon.rare-x.org – participating organizations LHON and LHON Canada
- ARS Patient Registry:
 - CoRDS:
<https://cordsconnect.sanfordresearch.org/BayaPES/sf/screeningForm?id=SFSFL>
– by [CureARS](#)

Before joining any patient registry, you have the right to ask as many questions as necessary in order to make an informed decision. Some topics you may want to explore prior to joining are what is the purpose of the registry, how does the patient foundation plan to use the data, what is the purpose of any medical records you're asked to upload, can you expect to see the results and when, and how do I remove myself from the registry.

Addendum – Quick Reference of Links

- Mitochondrial Care Network (<https://www.mitonetwork.org/>)
- Formal Diagnosis Guidelines Medical Professional (bit.ly/mitodiagnosis)
- MitoLand: A Guide to Mitochondrial Disease for Patients and Parents (<http://www.mitosoc.org/s/Mitoland-2017-version.pdf>)
- Other Words for Mitochondrial Disease (bit.ly/possiblemito)
- Mitochondrial Medicine Society (MMS) (<https://www.mitosoc.org/>)
- Support Organizations
 - United Mitochondrial Disease Foundation (umdf.org)
 - MitoAction (mitoaction.org)
 - Cure Mito Foundation (curemito.org)
 - People Against Leigh Syndrome (peopleagainstleighs.org)
 - Champ Foundation (thechampfoundation.org)
 - Courageous Parents Network (courageousparentsnetwork.org)
 - Global Genes (globalgenes.org)
 - ANGEL AID (angelaidcares.org)
- Exercise Links:
 - <https://www.umdf.org/ask-the-mito-doc/>
 - <https://www.mitoaction.org/resources/exercisenutrition/>
 - <https://www.mito.org.au/resource/exercise-and-mito/>
 - <https://vch.eduhealth.ca/PDFs/FA/FA.500.M697.pdf>
- Tools to Manage Your Journey:
 - MitoAction Mobile (https://www.mitoaction.org/mobile_new/) – an app that allows you to track your symptoms, medications, doctor’s appointments, etc.
 - mitoSHARE (umdf.org/mitoSHARE) – an online registry stewarded by the UMDF that includes tools to track symptoms, medications, doctor appointments, etc.
- Vitamins and Supplements (bit.ly/mitosupplements)
- Preventative Care and Medical Management Guidelines for Local Physicians bit.ly/mitocare
- Rare Caregivers Guidebook - https://www.caregiving.org/wp-content/uploads/2021/07/NAC_RareCaregivers_Guidebook_FINAL.pdf
- Finding Clinical Trials:
 - <https://clinicaltrials.gov/>
 - <https://www.umdf.org/clinical-trials/>