

# Moving On with Mito

*A Guide for Teens and Young Adults Living with Mitochondrial Disorders*



**Boston Children's Hospital**  
Until every child is well™



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“I look at how Mito made me think differently about life and how much I enjoy it. If they strive to, someone with a medical condition can live a happy life.”



## Dear Reader,

We created this guide for emerging adults living with mitochondrial disorders to help you succeed with transitioning to adult health care and the next phase of life. Patients, families, and health care providers have expressed a need for resources like this and we've heard you loud and clear. We've researched medical resources, and have talked to young people, parents, doctors, nurses, and patient advocates to find out what you want and need to know about. This guide includes: a simple explanation of Mito for friends, teachers, and anyone else who doesn't understand; tools to help guide your transition to adult health care; tips for success in college, employment, and independent living; and stories of real people with real experiences living their lives to the fullest.

The best way to use this guide is to first look through it to see what interests you most. Make a mental note of the other sections and come back to them when those situations come up in your life.

- If you are trying to figure out how to work around your health condition to do the things you like, read the **Life Goals** section.
- If you want to know what other people have experienced, read the **Personal Stories** first.
- If you are wondering what exactly transition means, read the sections on **Adult Health Care** and **About Transition**.

We hope you find this guide informative and useful, and that it will be an encouraging and supportive resource as you move on with Mito.

Best Wishes,

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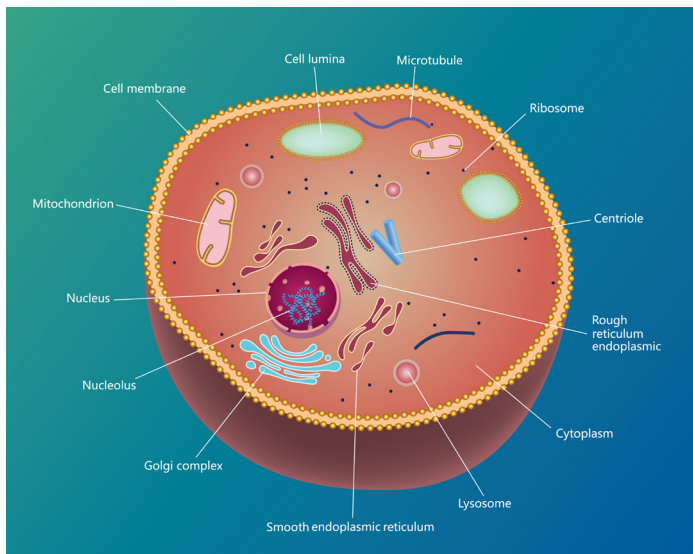
# Mitochondrial Disorders – The Basics

This overview provides an introduction to mitochondrial disorders, their symptoms, and treatment. You can show it to friends, teachers, school nurses, coaches, new doctors, family members, and anyone else who you feel needs to understand your condition.

## What are Mitochondrial Disorders?

In our bodies, mitochondria are the parts of cells that make energy from food and oxygen. With a mitochondrial disorder, something goes wrong with that energy-making process. When a cell is unable to produce enough energy, it may lose some of its ability to function. When enough cells in a certain part of the body are weakened that body part may not work correctly.

Mitochondria are found in almost all the cells in our bodies. In different people who have a mitochondrial disorder, different sets of cells, different body organs, or different body parts may be affected. Even people from the same family, with the same disorder, can have different health problems.



Parts of a typical human cell

Depending on which cells are affected, major symptoms might be:

- Muscle weakness or “heaviness”
- Vision or hearing problems
- Liver or kidney disease
- Diabetes
- Gastrointestinal problems
- Brain problems such as seizures

In people with a mitochondrial disorder, symptoms often show up in three or more different systems of the body; for example, in the eyes, muscles, and stomach. The body systems that are affected provide clues for doctors who can then test for a mitochondrial disorder. Many of the tests are typical, such as getting blood drawn. Other tests are more invasive, such as a muscle biopsy. Doctors will schedule the more extensive tests if they feel it’s necessary to learn more about their patient’s condition, and to confirm a diagnosis. Some people with mitochondrial disorders may have trouble with anesthesia, which may limit a doctor’s ability to perform certain tests. So a doctor may decide to give the patient a diagnosis based on his/her observations, on analysis of test results, and on his/her experience treating mitochondrial disease.

Since mitochondria are involved with making energy in the body, a lot of people living with a mitochondrial disorder become very tired, very suddenly. They may become so tired that they can’t pay attention or even stay awake. They may have to stop what they’re doing and find a place to rest and recharge. People sometimes call this “crashing”. It can come on quickly, because the heart, brain, lungs, muscles, and internal organs may be using a lot of energy during a certain activity or under certain circumstances. It’s like an electrical “brownout” – when the lights go dim because everybody in town is using so much electricity that the power-plant can’t keep up.

Two symptoms of mitochondrial disease are the body’s inability to properly regulate temperature,

and to stay hydrated. This may cause the person to faint during mild exercise or on warm days. Other symptoms can include: sensitivity to bright lights, muscle spasms, or trouble with digestion and nutrition.

People with mitochondrial disorders have good days and bad days. One day they may wake up feeling alright and have a productive day. Another day they may wake up feeling awful or not have any energy, even though they had a good night's sleep. Sometimes people may have good and bad *parts* of days (including “crashing”). Their symptoms may get better or worse over time, or new symptoms may develop. It can sometimes be difficult to predict how new situations and events will affect people with a mitochondrial disorder. They may need to work from home or miss classes, meetings or other events in order to save their energy for completing duties and assignments.

People with mitochondrial disorders may take longer than usual to recover from a common cold. During, and for about two weeks after a typical sickness such as a cold or flu, they may be in danger of getting a more serious infection. So, in addition to extra rest, they may need antibiotics to stop an infection, or intravenous fluids like saline or blood substitutes. It's important for each person to know his/her own body signs, and to know when to get help from health care providers.



**“I made myself sicker and ended up in the hospital...My body was trying to tell me to slow down. Listening to it has made a lot of difference.”**

## Types of Mitochondrial Disorders

Here are short descriptions of some of the more common mitochondrial disorder syndromes. There are others, and some people are only affected by one or a few symptoms. This list will give you an idea of the challenges that a person with a mitochondrial disorder may be dealing with. The person you know with a mitochondrial disorder may be affected with different combinations of symptoms in different amounts:

### **Chronic Progressive External Ophthalmoplegia (CPEO)**

CPEO is characterized by weakness of the eye muscles which may cause droopy eyelids and reduced ability to move the eyes. CPEO may sometimes be part of another syndrome, or may be one of many symptoms in an individual without a specific mitochondrial disorder syndrome.

### **Kearns-Sayre Syndrome (KSS)**

KSS usually starts before age 20. Common symptoms are: a gradual inability to move the eyes (CPEO), difficulty seeing in the dark, cardiac problems, and uncoordinated muscle movements. Other symptoms may include: hearing problems, kidney disease, diabetes, or short stature.

### **Mitochondrial Encephalomyopathy, Lactic Acidosis, and Stroke-like Episodes (MELAS)**

MELAS usually starts in childhood to early adulthood. It can cause migraine-type headaches, vomiting, seizures or stroke-like episodes. Other common symptoms are: inability to move the eyes, hearing loss, diabetes, digestive problems, and muscle weakness.

### **Myoclonus Epilepsy with Ragged Red Fibers (MERRF)**

This disorder usually starts in late childhood or the early teen years. Muscle twitching, weakness, and lack of coordination are common. Other possible symptoms may include: seizures, hearing loss, eye and speech problems, as well as exercise intolerance (muscle cramping, spasms, fatigue), and short stature.

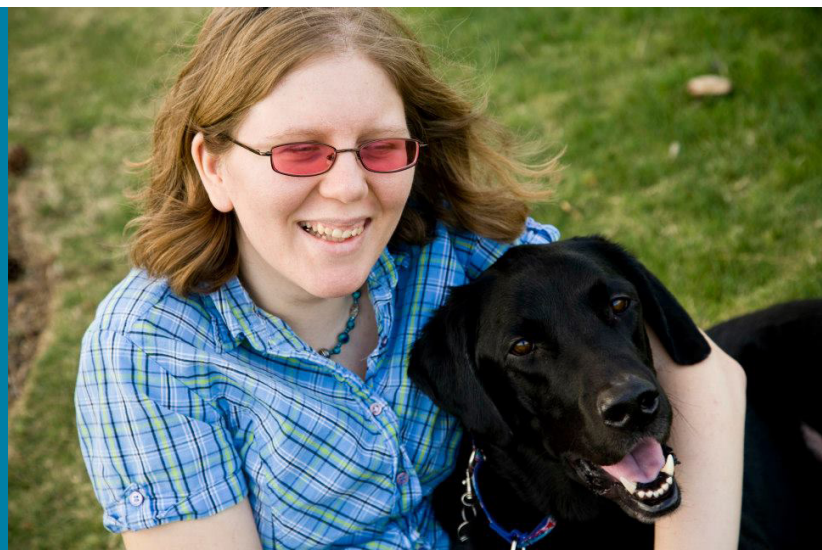
### **Neuropathy, Ataxia and Retinitis Pigmentosa (NARP)**

Primary symptoms of NARP may include: sensory impairment, uncoordinated movement, and gradual loss of vision. It can also cause epileptic seizures.

*Not everyone with a mitochondrial disorder will fit neatly into one of these syndromes. Many people are affected by different combinations of these or other symptoms to different degrees.*

For more in-depth information about mitochondrial disorders check out the **Resources** section of this guide. If you're a patient, you can fill out the **Medical Summary** section together with your metabolic specialist. Working on it with your doctor can help you learn more about your specific condition. Filling out the **Health Readiness Assessment** will show you where your knowledge is strong and what you might want to learn more about. The **Glossary** contains words and definitions you might find useful.

**“I (changed the practice of) finding identity in what I do to finding identity in myself as a person.”**





## Achieving Life Goals While Living With A Mitochondrial Disorder

**U**se this section to help you plan for and stay encouraged about achieving your goals. The people we talked to have done lots of things with their lives, and you can too. Here are some of the jobs they have or are training to do:

- Nurse
- Gymnastics Instructor
- Writer
- Patient Advocate
- Construction Worker
- Teacher's Aide
- Occupational Therapist
- Jewelry Maker
- Emergency Medical Technician
- Nutritionist
- Strength and Conditioning Coach
- Graphic Designer
- Child Psychology Specialist
- Film Maker

Here are some of their hobbies:

- Gardening
- Writing
- Crafts
- Reading
- Volunteering
- Science Club / Key Club
- Play Piano
- Crochet
- Running
- Social Media
- Horseback Riding
- Traveling

What are the things that you like to do? What do you think you'd like to do for work?



## Going to College

Seeking higher education is a popular goal for young people. If you are preparing to attend college after high school here are some things to think about to help you be successful.

### Accommodations

Many of the people we talked to discussed getting accommodations for special academic needs. You may currently receive help overcoming disabilities in public school, through an Individual Education Plan (IEP) and a 504 Plan (Section 504 of the Rehabilitation Act of 1973). Colleges and universities don't have to follow IEP's but their Disabilities Offices will take previous 504s into consideration. For students with Mito, accommodations might often include: extra time taking tests, access to a computer for written essays, getting someone's notes from the professor, or waiving the attendance policy. You may have to be polite but firm in advocating for what you need.

Everybody wants to fit in with people their own age, so sometimes in order to not "stick out" the people we talked to didn't ask for accommodations from the disabilities office. Sometimes they had their professors sign the paperwork at the beginning of the semester but never reminded them about what they needed, or never asked for the accommodations they were entitled to. One young man describes it like this:

***"I know what went through my head was - I'm sick, that's why I'm having problems - but I didn't understand that I was causing more problems and hurting myself by not asking for help."***

When he started using his accommodations his grades went from a C average back up to A's. He says his professors didn't judge him, and that the other students "are too worried about their own tests to notice" that he was taking his test in another room. Another young man told us: "I realized that the people I did tell [about my Mito] treated me the same... a lot of the other students respected me more."

You can learn more about IEP's and 504 plans on these websites:

[www.ncl.org/students-disabilities/iep-504-plan](http://www.ncl.org/students-disabilities/iep-504-plan)  
[www2.ed.gov/about/offices/list/ocr/504faq.html](http://www2.ed.gov/about/offices/list/ocr/504faq.html)

Remember to be persistent and to take a collaborative approach to working with the college staff to meet your needs.

### Plan for Health

Going to college and meeting new people is exciting, and you'll want new friends to think well of you. Sometimes you are going to want to prioritize your social life. You and your friends may not be thinking much about going back to your dorm rooms for a good night's sleep, and nobody expects you to. Other times you might be working on a paper and feel you're really making progress. You are going to push yourself – it's totally natural.

What would be great, is if you collected **all the important information about your medical condition**, especially including things that only you and your parents and doctors would know about, and made an **appointment to explain it all to the head of the campus health office**. You can do this shortly after you first arrive at college.



This information should include your symptoms, your baseline health status, what brings symptoms on for you, and especially what **your particular urgent treatment needs** are, like antibiotics or IV fluids. That



way, at those times when you push yourself (you know you will - that's part of the college experience!) you will have a plan in place and a medical professional who knows how to treat you. This will help keep you from missing classes, team practice, or time with new friends.

The sections in this guide on the **Basics of Mitochondrial Disorders**, your **Medical Summary**, and the **Transition Plan** will help you gather this information with your current doctor. And it will become part of a packet you can take with you when you travel to college or elsewhere. If you're going to be living far from home it's a good idea to **find out about hospitals in the area** and to start finding a primary care physician (PCP) or team of specialists who you can work with when you get there. The idea is to **have a plan in place before you arrive at college**, so that if you get sick, you will already be in touch with health care providers who know about you and about Mito.

## Organization

If you're headed to college you're probably an organized student, but what we mean by "organization" is being able to anticipate problems, and plan ahead to ensure success. Here are some things to think about:

*Class Schedule* – Some of the people we talked to said that they made sure to schedule their classes with time in between to get a snack, to visit the nurse, or just walk slowly. Some people needed to take naps in the afternoon, so they scheduled classes in the morning. The disabilities office can help you get priority to register early for classes that might be full on the regular registration day.

*Planning Calendar* – Having a big calendar on your wall and a day planner to write appointments, phone numbers, assignments, and activities in will help you keep track of things. These are things that busy, successful people do!

*To-Do Lists* – Some people make lists every morning of what they want or need to get accomplished that day. This is a good way to feel good about getting things done. Even when you don't finish everything on your list, you can see that you've still made progress. It feels great to cross things off your list, and writing it

**“Sometimes (my professors) would forget and it was my responsibility to remind them and let them know what I needed. That communication was really important because if you didn't communicate it they didn't know...I found that to be key..”**

down makes it more “real”. You'll see that your tasks and accomplishments aren't just all in your head. Try this, it really works!

*Sleep Habits* – We mentioned staying up late in the health planning section, but it's important to talk a little bit more about that. Obviously there will be times when you want to push your body past its limits. There's nothing wrong with that, but *having a routine to follow is a good idea*. If you live in a dormitory a lot of your neighbors may not have the kind of discipline you have already developed. They may stay up late all the time and not take very good care of their bodies. Some will flunk out of school because of it. If you set a reasonable schedule for going to bed and getting up - and stick to it for the most part - then, on the few occasions when you have to break that routine, you can get right back on track without being too tired or getting sick.

Other problems may surprise you. Sometimes a new environment causes stress that can cause fatigue and lower your body's defenses. Maybe you will need a single room to make sure you get enough sleep and don't catch colds or the flu from roommates. Perhaps you will need to go home to your parents to rest and recharge if possible. Doctors who work with teens with mitochondrial disorders say that they are usually ahead of other kids when it comes to knowing what works for them and what doesn't. The biggest thing to remember from this section is to try to **plan ahead, trust your instincts, and do what you think is best for you.**

“Finding a great fit for work is important: finding the right kind of work that works for your body... start that search early.”



## Working Full Time

Whether or not you've received education or training beyond high school, you may want to find a full time job. Here are a few things to keep in mind:

- Job interviews are not just for your employer to learn about you, they are for you to learn about your employer. You will be interviewing them too! Make sure the job is right for you and that you will not be expected to do things you are not capable of (like standing all day if that's a problem for you).
  - Starting a new job might affect your status for receiving medical assistance. It's important that you ask about your prospective employer's health insurance policy, and ask if it will cover the kind of health care you need. Some employers have a month-long waiting period before your insurance starts up. You can decide if that works for you.
  - You do not have to disclose a disability on a job application. The only time you should disclose a disability on an application is if the employer is explicitly recruiting employees with disabilities. Look up the **Office of Vocational Rehabilitation** in your state if you have a disability and want help finding appropriate work.
- Employers can't discriminate against people with disabilities, it's against the law. Job interview questions that could elicit information about a disability are illegal. This includes questions about accommodations you might need, how many sick days you took in your last job, or what medications you are on.
  - Although employers must provide "reasonable accommodations" for a known disability, you are required to work together with an employer to find those accommodations, and you still must be able to perform the essential duties of the job.

“At 24 I’m trying to settle down. I have a girlfriend I’ve been with for about a year. I’m looking to start the major part of my life: the adult part of my life.”



## Living Independently

Moving out of your parents’ house is a big step, but one that you can accomplish if you are prepared and have your supports in place. Living independently means you’ll be taking care of your needs on your own or with reliable help.

Here’s a summary of how you can approach it:

**Learn what you need to know, be realistic about your expectations, do what you can to prepare yourself, and have a plan.**

If you feel confident getting your driver’s license, that can be a good place to start. Driving yourself to school, medical appointments, and fun events is a great way to gain independence. If you are going to college, think about living in the dorms or going to a community college for the first two years while you practice doing all the things you need to do for yourself, like buying groceries and washing laundry.

Some of the people we talked to had lived in apartments with classmates and friends, and eventually moved in with a girlfriend or boyfriend. Some got help from the state they lived in or received Social Security Disability Insurance if they couldn’t work, and found housing through related programs. Some went away to college and then moved back into their parents’ homes, but have a new relationship with their parents after being away. They are more independent and their parents worry less about them. That might work for you too.

When you were younger you may have had to rely on your parents for a lot of help. Then, when you started needing more independence as a teenager, it may have been difficult for you and your parents to agree on new rules, especially if they were concerned about your safety. This may have led to some tension or fighting. It can be frustrating dealing with parents and Mito! Your parents probably mean well, and are trying the best they can to help you and keep you safe. Be patient with them, with yourself, and with the situation. Keep talking with your parents and letting them know what you need. Sometimes a family counselor can help all of you learn how to communicate better, especially if tensions are running high.

Remember, it’s OK to ask for help. Just because you are working toward becoming independent from your parents doesn’t mean you have to do everything alone. As one young woman says:

***“When you are learning to advocate for yourself it’s OK to get help from others... to be an efficient adult means working with others, not that you have to do it by yourself.”***



## Personal Stories

Doctors can't always be sure if someone has a mitochondrial disorder. Before you were diagnosed, you may have been afraid or angry because you didn't understand what was happening. It may have been quite a relief to finally find out what the problem was. Or maybe you've known about your condition all your life and you felt "different". Understanding your Mito is very important. Sometimes though, people may look to their health condition to explain everything about them. You might also feel like Mito took away your identity and all you are is a sick kid.

It's OK to get discouraged when things don't go your way or if you get sick at just the wrong time. In fact, *whatever you feel is OK*. There's no such thing as a correct or incorrect feeling. But nobody wants to stay discouraged or feel isolated. Yes, your mitochondrial disorder has a big impact on the person you are, but *aren't you so much more?* People who are around mitochondrial patients a lot say that on average they are *ambitious, intelligent, creative, and sympathetic*.

What do you do, or what would you like to do, that helps you define yourself the way you want to think of yourself? Do you listen to or play music, get good grades, write stories? Paint? Share pictures or blog on social media? Are you an avid reader who happens to have a mitochondrial disorder? Are you a skateboarder, being treated for mitochondrial disease? Or maybe you're a gamer, with Mito?

Here are some real people who don't let Mito define them - whose identities are based more on what they like to do than on their medical conditions.

### Maryanne

I have had abdominal pain my whole life. At 16 it started to get really bad and I missed most of my junior and senior year of high school. That's when I started on IV feeding and medication and received homebound education. At first I didn't

want anything to do with the IVs but the nurses and my mother taught me how to take care of them and before long I was connecting the tubes, priming the bag, flushing my line, and everything.

Some of my doctors told me I'd never work full time or live independently. They were wrong. I learned that **I can still pursue my interests, but sometimes I need to refocus them:**

- I joined the Phi Beta Kappa Society at my local community college, and did a lot of volunteer day trips. **I had to make sure I had all of my medical supplies and IV bags**, and I always had someone trained to help me. Eventually I started doing overnight trips, and even went to Washington DC for two weeks, representing my state in a Women's Leadership program. **I found out where the hospitals were near the conference center. I had all my medical information written down** in case I needed to go to the emergency room. My stomach was bad for a few days and I had to take a break one day to make sure I could do the next, but **I had a plan** if it got worse.
- Being sick so much in high school was isolating, and the medical stuff seemed to take over my life, so when I got involved in volunteering I wanted to make lots of new friends and not talk about my medical conditions. What I did was **I started reading three articles in a newspaper every morning, and I would make a point to talk to at least three different people**. It helped me get out of my shell.



- I had been a competitive gymnast as a girl, and **I figured out a way to teach gymnastics** that didn't require me to lift the kids. I piled up mats and lowered the bars. They loved it! I loved it!
- I wanted to live independently so I rented a house with a cousin, then an apartment with a boyfriend, and eventually bought my own house. I think some of that was hard for my mom, but she let me do what I needed to do. **You have to gain your independence medically, but there's also gaining independence from parents.**
- I found work where they consider my Mito a benefit because it helps me to understand my clients. I am in the process of figuring out how I can spend more time with my family and still do my work.

## Alan

I'm a varsity athlete at a division II college and I have a mitochondrial disorder. In fact, I got an athletic scholarship to come here. Despite my Mito I was a three-sport captain in high school. My parents made sure I had an IEP (individual education plan). They used to take care of all of my medical appointments and stuff, but when I got into college they said they wanted me to take over.

I got hooked up with the disabilities office and was given the accommodations I'd need to be successful. One of my accommodations was to waive the attendance policy, so that as long as I kept up with the work I didn't have to attend every class. I made sure to talk to my professors early in the semester and have them sign the accommodations paperwork. They would all be like "no problem, we'll work with you."

But more than once it happened that I had like a 95 percent in the class and I got failed for the attendance policy. They didn't understand Mito; **if you're not in a wheelchair they don't understand.** They looked at me like I was normal because on the outside I appear to be normal. With my disorder it's almost like they didn't believe me. If it's not in writing and signed by a doctor, they won't take your

word for it. I went to the ER for three days in a row, and had all my discharge papers. They just thought I was some kid who wanted to skip classes.



It would have been good if someone taught me gradually how to handle my own stuff. It went from my parents doing 90% of it to them just stopping. If I had someone who had been through this stuff I still would have made my share of mistakes, but I probably would have been quicker at correcting them than I did on my own. That's why I'm telling my story: so that my mistake's aren't for no reason.

NOTE: Other people have pointed out that it's their responsibility to remind professors about their accommodations. Alan didn't know that. And he didn't have resources, like this guide, to help educate his professors. **Since Mito can be an "invisible disease", you can't assume that people will remember that you need accommodations. It's your responsibility to make sure they understand.**

*Finding a peer-mentor, i.e. someone who has Mito and who has recently gone through what you are experiencing, can be a good way to get guidance. Mito social media groups can be a useful resource for finding a mentor.*

## Laura

I was 14 when I was diagnosed with Mito. Being diagnosed as a teen can be tricky since you are right on the cusp between pediatric and adult care. Even at 27, one of my docs is in pediatrics. Four of my docs see both children and adults. Six of my docs only see adults. Not every doctor is the same so it really is a trial and error process. What works best for me is when doctors have an open mind and accept that my body doesn't function as one would expect, even in a typical Mito patient.

I was inpatient three times at the very end of senior year. As a result, I was short of graduating with my class. Fortunately, the school allowed me to walk with my class! I'll never forget walking across that stage! As I walked back to my seat the dean stepped into my path and gave me a huge hug. She was obviously aware of my hard work and determination. Walking with my peers was huge for me and I earned that diploma over the summer!



I chose to go to college in Boston because I knew families who had moved to Boston for the good medical care, specifically Mitochondrial Disease. Since others were successful, I knew I would be. When my school's health center was repeatedly ignoring my GI symptoms, I just went down the street to Beth Israel Deaconess Medical Center. That was the first time **I found my inner advocate. At that point, I realized that I can choose my own doctors.**

I double majored and double minored in college. I was inpatient a couple times and needed more time for exams as well as more leeway when it came to deadlines. I followed the advice I was given that **even if you don't think accommodations are necessary, get them anyway** right at the beginning of term. It is always best to have them and not need them. The work load was the same as for other students, but at the end of semester I could get that extension if need be so I didn't have to pull all-nighters like the rest of my classmates. Obviously, with Mito an all-nighter is a bad idea.

I've done some great things since graduating. I just said "yes" to every chance I've been given. After speaking to Congress representing the Mito community, I accepted invitations to write for the United Mitochondrial Disease Foundation's newsletter and to deliver a speech at Next Step's fund-raising gala. Last year I interned in the patient advocacy department of a large biotechnology company in DC. I really found my niche there, doing meaningful and rewarding work, so now I'm looking for a full time job in patient advocacy!



# The Adult Health Care System

You may have heard a lot of talk about the health care system, and you may be wondering what it all means. The best way to think about it is like a whole collection of businesses that work together.

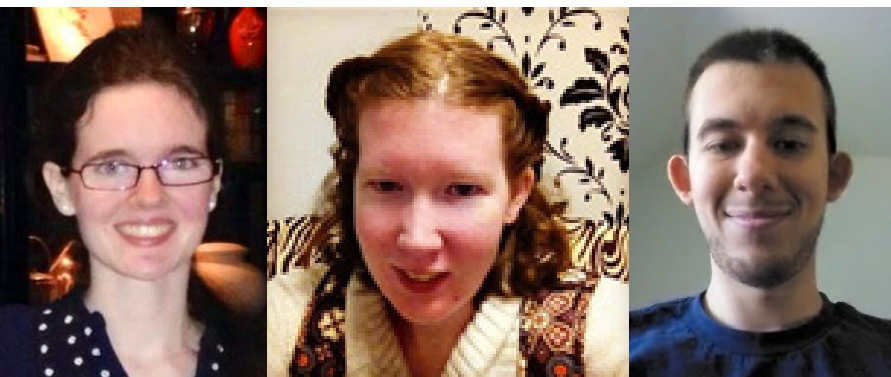
In this section we'll describe a typical adult health care arrangement. We'll also discuss the ways that your health care may be different.

## Who's Involved and What They Do

- **Patient** – You need quality medical care, health information, and health resources. Doctors aim to provide “patient-centered care”, which means you have a say in the treatment you receive and in how doctors share information with you.
- **Primary Care Physician (PCP)** – This is the doctor that provides your first level of medical care. A primary care physician is who you see for routine visits like yearly physical exams and flu shots. They are also usually the first doctor to go to when something isn't right but is not an emergency, like a bad rash, twisted ankle, or ear infection. Your primary care physician will prescribe medication, or send you for testing, like blood tests and X-rays. If they think you need it, they will refer you to a specialist.
- **Specialists** – These doctors specialize in a certain part or system of the body. Neurologists specialize in the brain and nerves. Cardiologists are experts in the heart and blood circulation. Certain surgeons specialize in joint surgery, and others specialize in eye surgery, for instance.
- **Internists** – These are doctors who have studied the adult body and know more about

adult diseases and conditions than about children's health. It's just another word for an adult physician.

- **Pediatricians** – These doctors have studied and treat children and teens. They know how to deal with issues like physical growth and brain development – things that stop or slow down once a person reaches adulthood.
- **Group Practices** – Sometimes a group of health care providers practice together, perhaps sharing a suite of offices. This allows them to share the cost of office and exam room space, and administrators. If one care provider is on vacation, the others can fill in, so patients don't have to wait.
- **Hospitals** – Often group practices and doctors who practice on their own will be associated with a particular hospital. In a big city there may be quite a few hospitals competing for the opportunity to care for you, and doctors want to make sure they can get their patients into a good hospital when needed, so they make business partnerships. Many specialists will only work in one hospital. They may even have their office and medical facilities in the hospital.
- **Other Facilities** – Testing facilities, physical therapists, and other providers specialize in other things, like Magnetic Resonance Imaging (MRI) or rehabilitation after a big surgery.
- **Pharmacies** – Pharmacies provide you with the correct amount of medicine that your doctor prescribes, and can help you get information on how to correctly take medicine, or on how multiple medicines you are taking may interact.



**“I'm choosing to be positive about my Mito and using it to help other people through their chronic illness journey, and I think it's made me more ambitious.”**

## Health Insurance

In the U.S. there are two basic ways health care bills get paid, either through a private health insurance company, or a public health care program.

Most people who have private health insurance get it through their workplace. The person's employer makes a business arrangement with a health insurance company, like Blue Cross/Blue Shield or Kaiser Permanente. The workplace pays for part of the health insurance plan, and the remaining cost is taken out of the worker's paycheck automatically every time he or she gets paid. Then when the person has medical bills and expenses, part of the cost is covered by the insurance company, and the rest is covered by the person.

When you have insurance through a health insurance company, the company has a contract with you, called a *policy*. The policy specifies the amount that the company will pay for doctor visits, hospitalizations, medicines, and other services. Your policy may cover the cost of some services or medicines, but not others.

Some people whose employers don't offer health insurance buy a policy directly from an insurance company. This is a very expensive option, so most people don't do this. Some people seek alternatives such as Affordable Care provided in their state.

Here are some aspects of health insurance that are helpful to know about:

- **Co-Pay** – A co-pay is the amount of money that you are required to pay your provider (doctor, pharmacy) at the time of your visit or purchase.

For instance, \$20 for an office visit, or \$15 for a prescription. The rest of the cost is covered by your health insurance company.

- **Deductible** – A deductible is a set amount of money that you have to pay out-of-pocket before your insurance company starts to chip in its share. For example, a \$500 annual deductible means you will have to pay \$500 towards your first medical bill, then the insurance company will cover the rest of the costs for that year.
- **Co-insurance** – Co-insurance describes an arrangement where you and the insurance company each pay a certain set fraction of your health care costs. For example, if you have 80/20 co-insurance, this means your insurance company will pay 80% of the cost of health care services, and you will pay 20%.
- **Affordable Care Act** – Nicknamed “Obamacare”, the Affordable Care Act is the US government's way for people to get health insurance if they don't have it through their employer. Through this act, which was launched in 2010, each state in the US has a State Health Insurance Marketplace which offers a variety of health plans, often with ways to help make them more affordable. To learn more about what's available in your state and whether you might be eligible for financial help, go to <http://healthcare.gov>
- **Public Programs** – Some people get their health care bills paid through public benefit programs like Medicare, Medicaid or the Children's Health Insurance Program (CHIP). Funding for these programs comes from

“She taught me to write down everything I wanted to go over. My doctors know now – I come in and they’re looking for my list.”



federal and/or state tax dollars. There are strict income, age and other limits on who is eligible for these programs. The cost to patients is much less, because these programs were designed for low income people who don't have another way to pay for health care. You can find out whether you're eligible for Medicaid in your state by filling out the same application for Marketplace coverage at <http://healthcare.gov>

## Getting The Health Care You Need With Mito

Not all *internists* and *pediatricians* have expert knowledge about mitochondrial disorders. You may have been going to the same pediatric *specialist* for many years, so you and that doctor are really the experts on your condition. If your doctor works at a children's hospital, eventually you are going to become too old for him/her to treat. This is called **aging out** of the pediatric system.

It makes a lot of sense to get as much information from your current health care providers written down to bring to your new doctor. A lot of doctors who haven't treated mitochondrial disorders before, say that information written for parents and patients is very useful to them. So if you can work together with your pediatric specialists to put your conditions into words that you understand, you can help bridge the gap between your old doctors and your new doctors, and in the process you learn even more. Use the section called **My Medical Summary** in this guide to help you do that.

## Handling Your Health Care

As you start managing your own health care, things that you will need to do are:

- **Know when your next appointments are** and make arrangements to **get there on time.**
- Get your **prescriptions filled** if you don't already.
- Call to **make appointments** when you think you need to.
- Be your own advocate and take it upon yourself to **make sure you get the right treatment.**
- **Find a new doctor** by talking to friends or asking questions on support forums online.

These are things that aren't too hard, but take some **organization**, so a lot of people get a **big calendar** and an **address book**, or use the **calendar app in their cellphones** to keep track of appointments.

The adult health care system requires a lot from you as a patient, but you will find that it is satisfying to advocate for yourself, and doing so will help you in other areas of your life. Many young people use it as a way to gain independence from parents and to have more control over their lives. It can lead to more freedom and opportunity in other areas, such as independent living and attending college. One of the people we talked to says it like this: **“You're sort of in charge of your own destiny... I mean they have the tools to help you but you can't get those tools if you don't advocate for yourself.”**





## About Transition

### Exactly what does “transition” mean, anyway?

As a teenager or young adult getting ready to live independently, you are beginning to take control of how you handle your Mito and your health in general. You are also starting to switch from pediatricians and child-focused specialists, to searching for and getting care from adult-focused health care providers.

This process of switching to adult-focused health care is called **medical care transition**. It also means changing the way you work with your doctors, so that you share some of the responsibility for your own care.

Medical care transition takes time and planning, but once it’s done, it frees you up to do the things you want to do with your life, like go to college, move into an apartment, and start living and working independently.

Just like planning and practicing plays to score baskets is part of the basketball team’s game plan, your medical care transition is part of your game plan. Planning and taking steps toward transition will help you be ready to react to whatever challenges show up as you work toward your goals.

Medical care transition is a journey, not a destination, so you don’t have to try to do it all at once! Our hope is that you will start early, and go at a pace that works for you. The tools in this **Transition Toolkit** will help you get a solid start and guide you along the way.

## Transition Toolkit

This section is designed to help you get organized and prepared. You can use the paper forms in this brochure, or the online forms here: <http://newenglandconsortium.org/for-families/transition-toolkit/> Read through the materials and fill out the forms to:

**Measure** your health independence by taking the **Health Readiness Assessment** and, over time, work toward answering yes to every question.

**Ask** a doctor or social worker for help filling out the **Medical Health Summary** and **Transition Plan** at your next appointment.

**Save** these forms in a folder, on your computer, or on a USB flash drive, and bring them to every medical appointment. Keep the flash drive in your wallet or purse and save any changes or updates. The online versions of the forms are here:  
<http://newenglandconsortium.org/for-families/transition-toolkit/>

As you work through the Transition Toolkit it will help you to: Decide when you’re ready to act as your own health spokesperson; Prepare for doctor’s appointments and fill out health forms; Talk to doctors and nurses about your medical condition and about what roles they should play in your medical care; Keep track of all of your important medical and health information.



## How's My Health Readiness?

Are you ready to be responsible for your own health care as an independent adult? Take this quiz to find out:

#		YES	NO
1	I know my height, weight, birth date, and social security number		
2	I know who to call in case of an emergency		
3	I know the name of my condition		
4	I can tell you how my condition affects me		
5	I can tell you what I need to do to care for myself		
6	I know the symptoms and medical problems related to my condition		
7	I know the basic medical treatment for my condition		
8	I have a support network for my condition		
9	I know what my diet should include and not include		
10	I know the names of my supplements and medicines		
11	I know how to schedule a doctor's appointment		
12	I know how to order my medications or get my prescriptions filled		
13	I can get myself to medical appointments (or can arrange transportation myself)		
14	I carry my health insurance card(s) with me		
15	I keep a schedule of medical appointments on my calendar		
16	I prepare questions for my health care provider before appointments		
17	I know what my co-pays are for office visits and prescriptions		
18	I know what kind of medical insurance I have		
19	I have documents explaining my condition to give to new providers		
20	I have a primary care physician		
21	I visit my metabolic specialist regularly		
22	I have a specialist for every system affected by my condition		
23	I know how to prepare my own food		
24	I know how to shop for my own food		
25	I have discussed issues about sex with my doctor		
26	I know how to get birth control and stay protected against sexually transmitted diseases (STDs)		
27	I have discussed the use of tobacco, alcohol and drugs with my doctor		

Now count the number of times you checked YES. If you checked YES for:

### 18 to 27 statements – You're there!

You are already accountable for your health care needs. If you haven't already, complete the Transition Plan with your doctor, school nurse, or other adult helper. Let's get your safety net set up.

### 9 to 17 statements – You're on your way!

You are actively taking on many important responsibilities for your health care. See what else you can learn from [Mitochondrial Disorders: The Basics, The Adult Health Care System](#) and [Resources](#) in this guide, and use the questions that got a NO to prepare questions for your doctors, school nurses, and parents or guardians.

### 0 to 8 statements – Let's get started!

Transitioning to college, career, and living independently requires you to become a better health advocate for yourself. That takes time and planning. Now's a perfect time to start, so when you're ready to take on new challenges, you'll have a plan for staying healthy if something unexpected happens. Pick a few of the responsibilities that got a NO answer and spend the next month learning and doing them. You can seek help from your doctors, school nurses, social workers, parents or guardians, or any other trusted adult helper.

Update and save this Health Readiness quiz every month until you've answered all of the tasks with a YES!



## My Medical Summary

Use this form to keep important medical information in one place, and to summarize the specifics of your mitochondrial disorder. This information can be helpful for your family members, friends, teachers or colleagues. It can help them understand how your condition affects you, and what can help you feel and function better at home, school, or work.

**Answer the questions below together with your metabolic specialist and have him or her sign this form:**

Date updated \_\_\_\_\_

Last Name \_\_\_\_\_ First Name \_\_\_\_\_

Birthday \_\_\_\_\_ Gender \_\_\_\_\_ Weight \_\_\_\_\_ Height \_\_\_\_\_

Address \_\_\_\_\_

City \_\_\_\_\_ State \_\_\_\_\_ Zip Code \_\_\_\_\_

Insurance Name \_\_\_\_\_

Insurance ID & Group # \_\_\_\_\_

Insurance Address & Phone Number \_\_\_\_\_

Metabolic or Genetic Condition \_\_\_\_\_

Medications/Supplements and Dosages \_\_\_\_\_

\_\_\_\_\_

\_\_\_\_\_

Allergies to Food, Drugs and Other Substances \_\_\_\_\_

\_\_\_\_\_

\_\_\_\_\_

Emergency Contact Name & Relationship \_\_\_\_\_

Emergency Contact Phone Number \_\_\_\_\_

Pharmacy Name \_\_\_\_\_ Pharmacy Phone Number \_\_\_\_\_

Immunizations Type and Date: \_\_\_\_\_

\_\_\_\_\_

\_\_\_\_\_

\_\_\_\_\_

(continued)

**Information about your symptoms and care:**

What parts of your body are affected by mitochondrial disorder? \_\_\_\_\_  
\_\_\_\_\_  
\_\_\_\_\_

What happens to you when you have a bad spell, or a bad day? \_\_\_\_\_  
\_\_\_\_\_  
\_\_\_\_\_

What causes you to lose energy or have other symptoms? \_\_\_\_\_  
\_\_\_\_\_

What do you have to do to stay healthy? \_\_\_\_\_  
\_\_\_\_\_  
\_\_\_\_\_

If your mitochondrial disorder is a known syndrome, what is the medical name for it? \_\_\_\_\_  
\_\_\_\_\_

If you have multiple symptoms, what are their medical names? \_\_\_\_\_  
\_\_\_\_\_  
\_\_\_\_\_

What other things do you or your metabolic specialist think are important for teachers, school nurses, managers, roommates and friends to understand about your condition?  
\_\_\_\_\_  
\_\_\_\_\_  
\_\_\_\_\_  
\_\_\_\_\_  
\_\_\_\_\_  
\_\_\_\_\_  
\_\_\_\_\_

Patient Name: \_\_\_\_\_

Health Specialist Name and Title: \_\_\_\_\_

Health Specialist Signature: \_\_\_\_\_ Date: \_\_\_\_\_

## My Transition Plan

Use this plan to start collaborating with your doctors. There's room to fill in specifics and things that each person should agree to do. It's a good way to know what to expect from each of your doctors and what they should expect from you. Bring it to your appointments to fill in each section with the appropriate doctor.

Patient Name \_\_\_\_\_

Medical Record # \_\_\_\_\_

Primary Care Physician \_\_\_\_\_

PCP Phone Number \_\_\_\_\_ Email \_\_\_\_\_

Pediatrician \_\_\_\_\_

Pediatrician Phone Number \_\_\_\_\_ Email \_\_\_\_\_

Metabolic Specialist \_\_\_\_\_

Metabolic Specialist Phone Number \_\_\_\_\_ Email \_\_\_\_\_

Other Health Care Provider \_\_\_\_\_

Other Care Provider Phone Number \_\_\_\_\_ Email \_\_\_\_\_

Emergency Contact Name & Relationship \_\_\_\_\_

Emergency Contact Phone Number \_\_\_\_\_

### Medical Findings

Genotype/Laboratory Findings \_\_\_\_\_

Treatment Status \_\_\_\_\_

Current Medications/Treatments \_\_\_\_\_

Allergies/Adverse Reactions \_\_\_\_\_

(continued)



Past Medical Concerns or Hospitalizations \_\_\_\_\_

Living Situation/Employment \_\_\_\_\_

Mental Health Status/Cognitive Status \_\_\_\_\_

Other Health Risks \_\_\_\_\_

### **Contract**

To enhance collaboration, each person should agree on the following responsibilities:

Metabolic Specialist will:

- Obtain labs and tests. For example: \_\_\_\_\_
- Prescribe condition-related medications: \_\_\_\_\_
- Work with PCP to develop an emergency protocol: \_\_\_\_\_

Patient will:

- Remember to take medications. For example: \_\_\_\_\_
- Order and pick up supplies/supplements: \_\_\_\_\_
- Show-up to appointments: \_\_\_\_\_

Primary Care Physician will:

- Monitor patient's general health: \_\_\_\_\_
- Update metabolic specialist with important patient information: \_\_\_\_\_
- Contact metabolic specialist with medical concerns: For example: \_\_\_\_\_
- Other: \_\_\_\_\_

When you're done filling out the **How's My Health Readiness**, **My Medical Summary**, and **My Transition Plan** save them in a safe place. You can find the online versions of the forms here:

<http://newenglandconsortium.org/for-families/transition-toolkit/>

Congratulations! You are well on your way to being in charge of your own health.

## Resources

The online version of this **Moving On with Mito** guide is available on the web site of the New England Consortium of Metabolic Programs: <http://newenglandconsortium.org/>

Here's a list of web sites for people living with a mitochondrial disorder that the people we talked to found helpful. The United Mitochondrial Disease Foundation and Mito Action are excellent sources for information and support. The additional resources listed will fill in the gaps, including a link for additional information on transition from pediatric to adult healthcare.

### **Mito Action**

Mito Action works through support, education and advocacy initiatives to improve life quality for all who are affected by mitochondrial disorders.

<http://www.mitoaction.org/>

### **United Mitochondrial Disease Foundation**

UMDF seeks to promote research and education for the diagnosis, treatment, and cure of mitochondrial disorders, as well as to provide support to individuals and families.

<http://www.umdf.org/>

These sites also have good information on mitochondrial diseases:

### **North American Mitochondrial Disease Consortium**

<http://www.umdf.org/site/pp.aspx?c=8qKOJOMvF7LUG&b=7934687>

### **Myths and Facts about Mito from the Cleveland Clinic**

[http://my.clevelandclinic.org/disorders/mitochondrial\\_disease/hic\\_myths\\_and\\_facts\\_about\\_mitochondrial\\_diseases.aspx](http://my.clevelandclinic.org/disorders/mitochondrial_disease/hic_myths_and_facts_about_mitochondrial_diseases.aspx)

### **Muscular Dystrophy Association**

<http://mda.org/disease/mitochondrial-myopathies>

### **A Resource for your Primary Care Physician - From Exceptional Parent Magazine**

<http://biochemgen.ucsd.edu/mmdc/ep-toc.htm>

Mitochondrial disorders can often look like other diseases. Research is ongoing to find how Mito is linked to many other diseases. Learn more here:

### **Foundation for Mitochondrial Medicine**

<http://mitochondrialdiseases.org/related-diseases/>

### **UMDF links to other diseases**

<http://www.umdf.org/site/pp.aspx?c=8qKOJOMvF7LUG&b=7934637>

These two organizations were instrumental for the research that led to this toolkit. They have additional information for a broad spectrum of genetic conditions.

### **New England Genetics Collaborative**

<http://negenetics.org/>

### **Genetic Education Material for School Success**

<http://www.gemssforschools.org/>

**Got Transition** is an organization that aims to help the transition from pediatric to adult healthcare. There you'll find lots of resources for Patients & Families, Health Care Providers, and Researchers & Policymakers.

<http://www.gottransition.org/>

# Glossary

**Accountable** (ah-KOWNT-ah-bul) – able to take responsibility for.

**Adolescent** (add-oh-LESS-ent) – a person who is growing from childhood to adulthood.

**Adverse** (add- VERSS) – negative, bad.

**Advocate** (AD-voh-kayt) – to support or argue for.

**Allergy** (AL-er-jee) – an adverse reaction to a food or substance.

**Antibiotic** (an-tee-by-OT-ick) – medicine that kills bacteria to stop an infection.

**Birth control** (BERTH kun-TROHL) – medicine or device used to try to keep from getting pregnant. “The pill”, condoms, and IUDs are all forms of birth control.

**Cognitive** (KOG-nih-tiv) – having to do with thinking and brain activity.

**Collaboration** (kohI-LAB-uh-RAY-shun) – working together.

**Co-pay** (Koh-PAY) – the amount of money you must pay when you have a medical appointment. The health insurance company pays the rest.

**Crashing** – when a person with Mito runs out of energy suddenly. It’s like an electrical “brownout”, when the lights suddenly dim because everyone is using a lot of electricity all at once.

**Diabetes** (die-ah-BEET-eez) – when your body doesn’t produce enough insulin to offset the amount of sugar in your blood.

**Diagnosis** (die-agg-NO-sis) – the condition your doctor decides you have after examination.

**Dominate** (DOM-in-ayt) – to control.

**Empathetic** (em-pah-THET-ick) – able to understand what it would feel like to experience what another person is experiencing. This is more than sympathy.

**Experiment** (ex-PEHR-uh-ment) – a test.

**Gastrointestinal** (GAS-troh-inTEST-in-ul) – stomach, intestines, and other organs that digest food.

**Genetic** (jen-ETT-ick) – having to do with genes.

**Genes** (JEENZ)– the building blocks inside the cells of our body that determine how we inherit features, traits, or diseases from our parents.

**Genotype** (JEEN-oh-tiyp) – the total set of genes transmitted from a parent to child.

**Guardian** (GARD-ee-in) – a person who watches over and protects. In the instance of child-rearing, it is a person who takes the place of a parent.

**Identity** (eye-DENT-ih-tee) – how a person feels or thinks about him/her self.

**Independent** (in-deh-PEN-dint) – able to make one’s own choices; free from the control or influence of another.

**Insightful** (in-SIYT-ful) – ability to understand one’s own or others’ problems.

**Intravenous** (in-trah-VEEN-uss) – special liquids going directly into a person’s veins to address health issues.

**Metabolism** (meh-TAH-bul-iz-m) – chemical processes for making food and oxygen into energy.

**Mitochondria** (my-toe-KON-dree-ah) – the parts of a cell that make energy from food and oxygen.

**Mobility** (Mow-BILL-ih-tee) – ability to move without help.

**Mutation** (myoo-TAY-shun) – a sudden change. In this instance a change in genes.

**Responsibilities** (reh-SPON-sih-bill-ih-teez) – things you are expected to do.

**Savvy** (SAH-vee) – well informed. Knowledgeable and quick to understand.

**Seizures** (SEEZH-yurz) – sudden shaking, inability to control one’s body, sometimes fainting.

**Sexually transmitted diseases (STDs)** (SEX-yoo-ah-lee trans-MITT-idd diz-EEZ-ez) – diseases that are often spread from one person to another through contact with infected sex organs, skin, or body fluids. AIDS, herpes, and chlamydia are STDs.

**Specialist** (SPESH-ah-list) – a doctor who specializes in a specific part of the body, like metabolism or neurology (brain & nerves).

**Summarize** (SUMM-ah-riyz) – to gather and present just the important parts of a large amount of information.

**Supplements** (SUPP-leh-ments) – something added to make up for something that is missing.

**Sympathetic** (simp-ah-THET-ick) – able to understand how another person feels.

**Symptoms** (SIMP-tumz) – changes to your body’s normal function due to disease or disorder.

**Syndrome** (SIN-droh-m) – a number of symptoms occurring together that make up a disease or disorder.

**Transition** (tranz-IH-shun) – passing from one form or stage to the next.



## Acknowledgments

**W**e would like to acknowledge [Mito Action](#), [Next Step](#), and the [United Mitochondrial Disease Foundation](#) for their guidance and assistance in the development of this guide.

We also thank the many [doctors](#), [nurses](#), [advocates](#), [researchers](#), and especially [young adults living with mitochondrial disorders](#) who agreed to be interviewed. It is largely due to their perspectives, insights, and suggestions that we can bring you this information.

Thanks also to [Tim Edgar, PhD, Graduate Program Director for Health Communication at Emerson College](#) and [Karen Smith, Project Coordinator at the New England Genetics Collaborative](#) for their expertise and guidance.

Funding and programmatic support for this project has been provided by the [New England Genetics Collaborative](#), which is funded by [Grant No. H46MC24093](#) from the [Maternal and Child Health Bureau /Health Resources and Services Administration, Genetic Services Branch](#).

Lastly, we would like to thank the [Metabolism Program team at Boston Children's Hospital](#), [Genetics Education Materials for School Success](#), and the [New England Genetics Collaborative](#) for sharing their knowledge and enthusiasm, and for helping to create this guide.



## **Moving On with Mito: A Guide for Teens and Young Adults Living with Mitochondrial Disorders**

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This guide is available online at [newenglandconsortium.org](http://newenglandconsortium.org)

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