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Diagnosis or No Diagnosis: What's Next After Genetic Testing?



About **Global Genes**[®]

Global Genes is a 501(c)(3) non-profit organization dedicated to eliminating the burdens and challenges of rare diseases for patients and families globally. In pursuit of our mission we connect, empower, and inspire the rare disease community to stand up, stand out, and become more effective on their own behalf—helping to spur innovation, meet essential needs, build capacity and knowledge, and drive progress within and across rare diseases. We serve the more than 400 million people around the globe and nearly 1 in 10 Americans affected by rare diseases. If you or someone you love has a rare disease or are searching for a diagnosis, contact Global Genes at 949-248-RARE or visit our resource hub at www.globalgenes.org.

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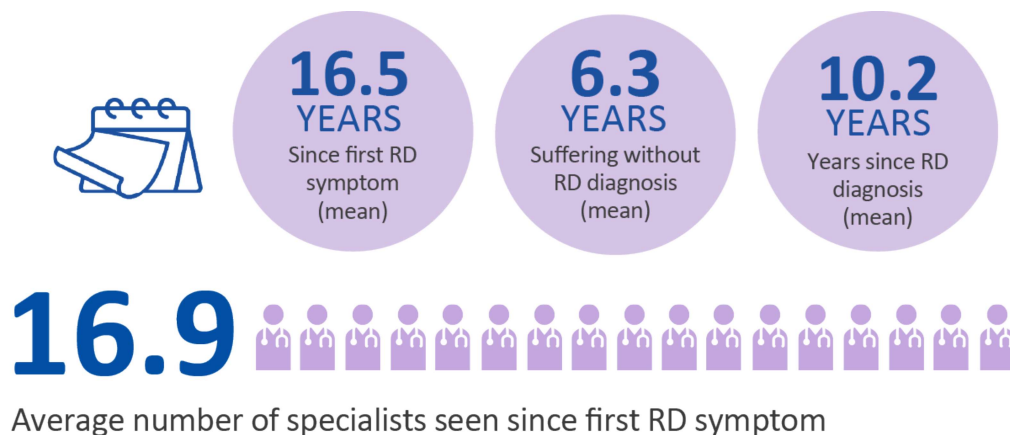
Introduction

If you are seeking a diagnosis, **genetic tests** along with a physical exam, medical history, and other lab tests can sometimes provide a clear answer that explains your symptoms and helps your doctors make decisions about treatment. But it's also possible that your test results won't produce a diagnosis—at least not right away.

There are many types of genetic tests and they are constantly improving, together with our knowledge of the variants that can lead to genetic disease. It sometimes takes multiple rounds of testing over a period of years to get the answer you are seeking. But if you are persistent, the diagnosis you ultimately receive can lead to better care and connect you to a supportive community.

Study Results:

RD Impact Survey Captures Medical Burden, Long Diagnostic Odyssey



Graphic from [The National Economic Burden of Rare Disease Study](#)

Whether you receive a diagnosis, or you are still searching, this toolkit will help you get your bearings. It will also help you explore your options and decide what you want to do next.

Need More Information?

Here are [*helpful resources*](#) on genetics and genetic testing from Global Genes

[Genetic Testing: Getting to a Diagnosis](#)

Learn Genetics 101 and review the types of testing and results that can be expected.

[Genetic Test Comparison Table](#)

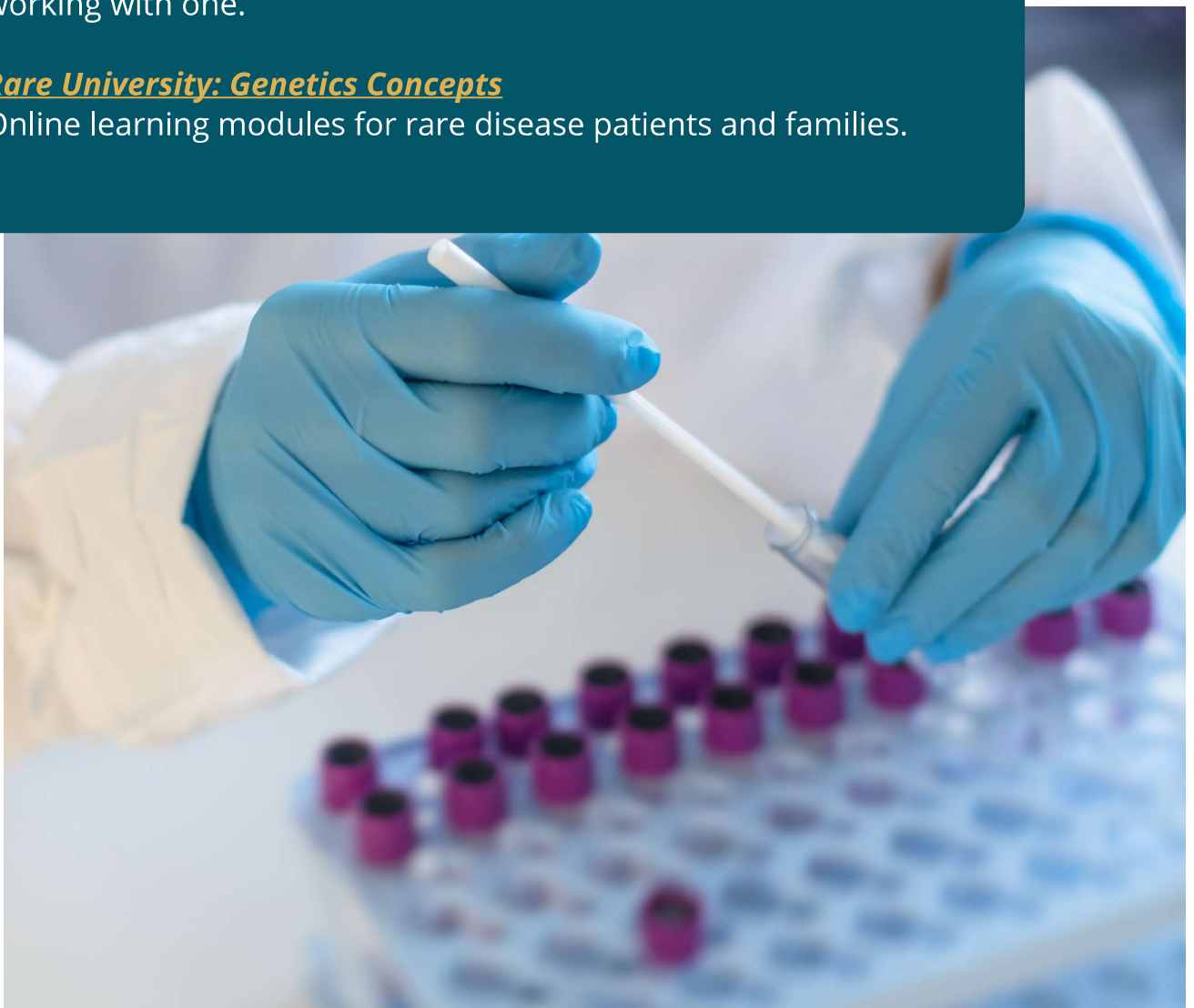
At-a-glance list of types of tests, uses, limitations and costs.

[How a Genetic Counselor Can Help You](#)

How to find a genetic professional and get the most from working with one.

[Rare University: Genetics Concepts](#)

Online learning modules for rare disease patients and families.



Getting Your Test Results

Preparing for Your Post-Test Appointment

First, find out how you will receive the results. Your genetic test results will be communicated in person or by having a phone call or video visit. Either way, it helps to have someone with you taking notes, so you won't have to trust your memory. Make arrangements for childcare, if needed, so you won't be distracted.



PRO TIP

"You may walk out of that room with nothing in your mind other than 'I'm walking out this door.' Anything beyond that is a score for you. Because what you are walking into requires a Herculean effort of your mind and nervous system. It's the Olympics of mental/emotional balance. So have your team around you."



– BECKY SANSBURY, DIRECTOR OF CAREGIVER SUPPORT, RARE DISEASE INNOVATIONS INSTITUTE

There will likely be a lot of information provided, much of which could be unfamiliar. To make things easier for you, most **geneticists** and **genetic counselors** provide summaries for families and their healthcare team. Before the appointment, ask if you will receive a summary. Also, find out the best way to follow up if you have questions later.



RARE TIP

"Understanding some genetic terms is a good first step in understanding the diagnosis."



– LAUREN KESSLER, RARE DISEASE FAMILY MEMBER

Ideally, you will receive:

- **genetic test** results
- an explanation of what your results mean
- supportive resources
- an overview of potential treatment

options or clinical trials if available

- referrals to specialists for a personalized medical-management plan

It's helpful to let your healthcare team know how you want to receive medical information. Some people like written information and some prefer videos. Some people want all of the details right away and some prefer to absorb what they've heard before thinking about what comes next.

You can ask for a referral if you aren't connected to a genetic counselor by your healthcare team and feel it would be helpful. You can also choose to make an appointment on your own. It's fine to call different healthcare networks or hospitals and ask if they have genetic counselors or nurse navigators who can help you with next steps. The National Society of Genetic Counselors also offers a directory (<https://findageneticcounselor.nsgc.org>) that allows you to search for a genetic counselor in your area or one who offers telehealth visits.

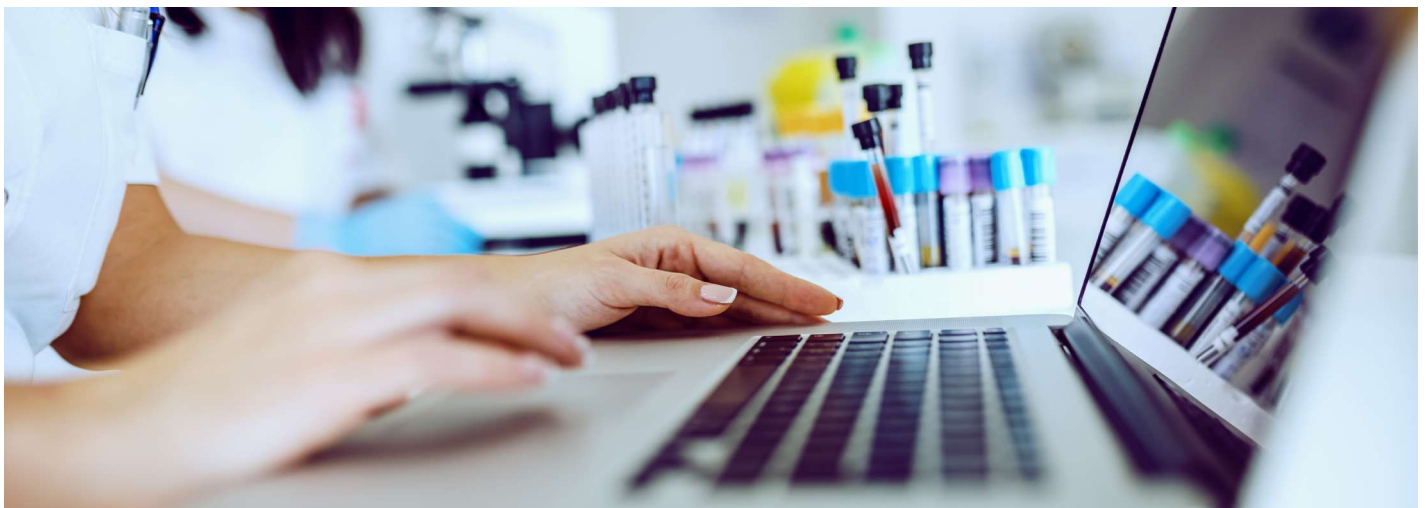


PRO TIP

There may be long waits or restrictions regarding the type of referrals a genetic counselor near you can see. But there are also many telehealth genetic counseling options. In many cases, you may be paying out-of-pocket, but the cost is not prohibitive for most people. If you don't think you have a good understanding of your test results or what to do next, it may be one of the best investments you make."



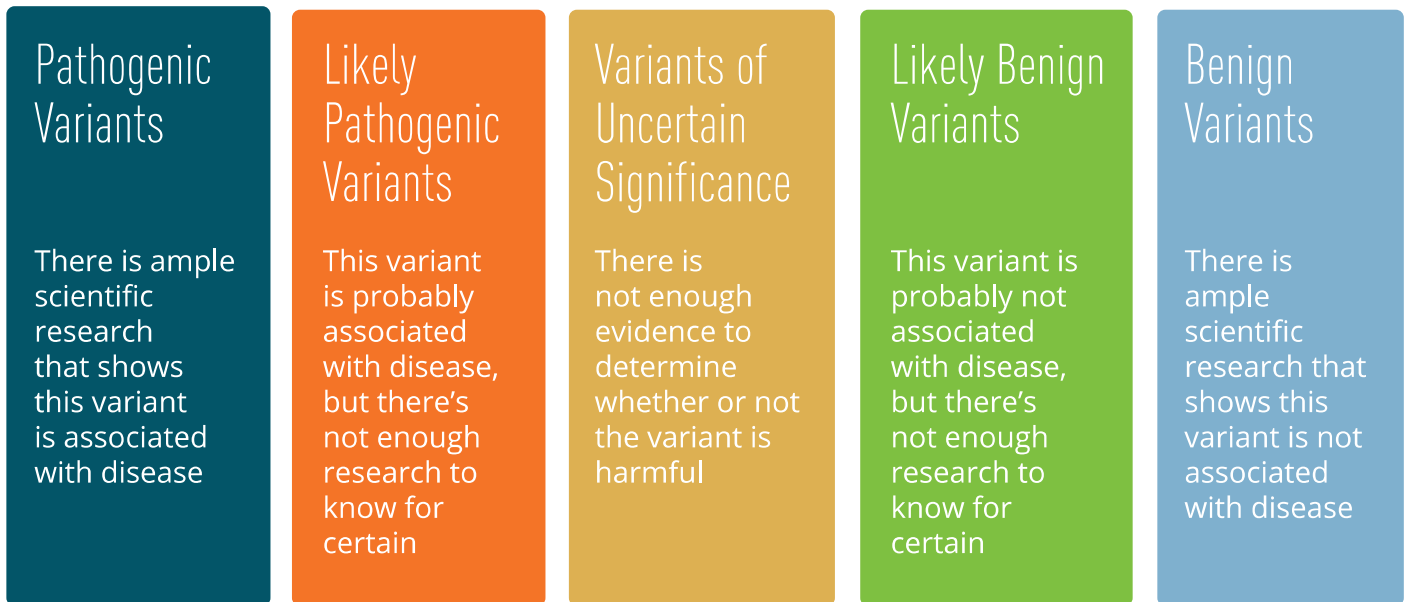
– ELEANOR GRIFFITH,
FOUNDER, GREY GENETICS



Understanding Genetic Test Results

Genetic testing identifies changes in genes, called variants. But only a small percentage of variants cause genetic disorders. Most variants have no impact on health or development.

To help determine if a variant is associated with a genetic disorder, scientists group them as follows:



If you receive a **positive result** from your genetic test, it means the lab found a change in a particular gene, chromosome, or protein of interest. Depending on the purpose of the test this could confirm a diagnosis, determine that a person is a carrier of a variant, or has a risk of developing a disease in the future. Note that healthcare professionals usually can't use a positive result to determine the exact risk or predict the severity of the condition.

If you receive a **negative result**, it means the lab did not find a change in a gene, chromosome, or protein that they considered. This could mean the

person isn't affected by a particular disease and does not have a risk of developing it. But it's also possible that the particular test used did not detect the change. Further testing may be needed to confirm a negative result.

An **uninformative result** means the test did not provide any useful information that would either confirm or rule out a specific diagnosis or indicate that the person has an increased risk of developing a disorder. In some cases, testing other affected and unaffected family members can help clarify the result.

If You Receive a Diagnosis

There are many benefits to getting a genetic diagnosis.

- It may lead to effective treatment
- It will help you avoid harmful or unnecessary treatment
- It will help you explain your condition to others
- It may open up possibilities for enrollment in clinical trials
- It could answer questions of recurrence in families

But it can also be overwhelming. You may feel relief that you finally have a name for the disease. At the same time, you could be fearful, grieving, angry, depressed, or even shut down. All of these reactions are normal and acceptable. Be patient with yourself. Take time to adapt. Ask for help when you need it. You don't have to do this alone.



Questions to Ask When You Are Diagnosed

- What is the result?
- What does this result mean for me (my child, my spouse, etc.)?
- Are there any treatment options?
- Would you recommend changes to my medical care?
- What specialists should I see?
- Is this disease inherited? Should other family members be tested?
- Where can I find reliable information on this topic in patient-friendly language?
- Can you connect me to any support groups or patient-advocacy groups?
- Do you know of any clinical or research trials I could consider?
- Are there registries I could participate in?
- Can I follow up with you if I find I need more information?

If you have been diagnosed with a rare disease, you may be referred to a specialist who has in-depth knowledge, such as a hematologist, neurologist, or endocrinologist. You may need to be seen at a major medical center that offers specialized care. If you don't receive a referral, consider asking your physician if this would be helpful.



RARE TIP

“A genetic diagnosis can help you understand what to look out for and what to expect.”



- JENN MCNARY, RARE DISEASE CAREGIVER, CONSULTANT AND FOUNDER OF ONE RARE

One advantage of working with a specialist is that he or she will likely be familiar with the latest research and clinical practice guidelines for your rare disease. Unfortunately, many rare genetic diseases have few or no treatment options. But your doctor may know of investigational treatments available through clinical trials.

If You Don't Receive a Diagnosis

What should you do when you go through genetic testing and don't receive a diagnosis? The short answer is, “Keep going.”



RARE TIP

“Don't lose hope. Keep it up. Stay in contact. Don't worry about being a pest to your doctor. We have come amazingly far since the beginning. Every year we are in a better place.”



- JANIS CREEDON, RARE CAREGIVER AND ADVOCATE

Our knowledge of rare diseases and genetic testing techniques is always improving. So, it may be helpful to have additional tests done or reanalyze your data from **whole genome sequencing** (WGS) or **whole exome sequencing** (WES).

Follow-up If You Are Undiagnosed

If your genetics team doesn't specify when you should come back, be sure to ask them if you should follow up in six months or a year. You should also alert your genetics team if:

- You have new symptoms
- You have a new health issue
- Someone else in your family was diagnosed with a genetic condition
- You have new contact information, so they can reach you if new tests are indicated

It can be frustrating to wait for scientific knowledge to catch up before you are able to obtain a genetic diagnosis. But keep in mind that you have taken a step forward. A “non-diagnosis” can help rule things out, redirect your path and determine next steps.



RARE TIP

“If I had to come up with a tagline for the undiagnosed, it would be one word: perseverance. There’s a lot of feeling and emotion in that word. The journey never ends. Your life has to continue. You have to keep fighting because you only have one body.”



– GINA SZAJNUK, UNDIAGNOSED PATIENT, CAREGIVER OF THREE UNDIAGNOSED CHILDREN AND FOUNDER OF THE RARE UNDIAGNOSED NETWORK (RUN)

Helpful Resources for the Undiagnosed

Becoming an Empowered Patient: A Toolkit for the Undiagnosed

Information designed to move you from feeling afraid and anxious to feeling prepared, confident and educated on how to manage the next steps in your healthcare journey.

NIH/NCATS Genetic and Rare Diseases Information Center (GARD)

Guide to research programs, clinical trials, and financial advocacy for the undiagnosed.

Syndromes Without a Name UK (SWAN)

Help for the undiagnosed, part of the Genetic Alliance UK.

Rare Undiagnosed Network (RUN)

Dedicated to empowering rare and undiagnosed patients and their families with genomic information and community through advocacy, networking and support.

Undiagnosed Diseases Network (UDN)

A research study funded by the National Institutes of Health Common Fund. Its purpose is to bring together clinical and research experts from across the United States to solve the most challenging medical mysteries using advanced technologies. The application is patient-driven and has a portion that must be filled out by your medical team.

Undiagnosed Diseases Network International (UDNI)

When you submit your story to UDNI, it is shared with specialists who visit the UDNI website looking for cases similar to those they have seen. If yours is a fit, you or your doctor will be contacted.

Undiagnosed Rare Disease Patient Registry

A natural history study of undiagnosed rare disease patients conducted by the National Organization for Rare Disorders (NORD).

What Happens Next?

Adapting to life with a rare disease is challenging, whether you've received a genetic diagnosis or remain undiagnosed, at least for the time being. In this section, you'll find tips on where to find sources of support to better manage your care.

Finding People Who Understand

Connecting to a community is a valuable first step. Because genetic diseases are rare, many people have never heard of them until they're diagnosed. Even physicians, including specialists, generally don't know much about them.

What Is a Rare Disease?

In the United States, a rare disease is defined as a condition that affects fewer than 200,000 people. Other countries have their own definitions. In the European Union, a rare disease is one that affects fewer than 1 in 2000. An ultra-rare disease affects fewer than 1 in 50,000. There are more than 7000 rare diseases and 95 percent lack treatments. They affect 3.5 to 5.9 percent of the world's population, half of them children.

Perhaps that's why patients and caregivers living with rare diseases are so committed to helping each other. They understand how you feel and have faced the same challenges. They are some of the most generous and inspiring people on the planet and they will gladly share what they've learned.



RARE TIP

"For years, I had been all alone, spending endless hours on the internet pulling resources together. The beautiful piece about being part of a community is that they'll link you with others and help answer your questions. I found my sister's doctor through a Facebook contact on her rare disease page."



– NANCY KESSLER, BOARD MEMBER, SYNGAP RESEARCH FUND

Belonging to a community provides practical as well as emotional support. You can turn to them for tips on how to work with your healthcare team, how to meet your child's educational needs, recommendations for specialists, and options for financial assistance. They will also keep you informed as new knowledge and treatments emerge.



RARE TIP

Even without a diagnosis, it's really important to have a support group. We knew all along that my son's condition was ultra rare even before we had a name for it. Global Genes was my support group at that point when we were undiagnosed.



– CAROLINE LOEWY, CO-FOUNDER AND BOARD MEMBER, KCNQ2 CURE ALLIANCE, GLOBAL GENES BOARD OF DIRECTORS

In rare diseases, you will often find patient groups driving research, creating registries, and advocating for legislation. They work with each other, with academic institutions, and with government agencies to do everything in their power to accelerate treatment and improve lives. Altogether, there are 400 million people globally living with rare diseases and they are working together to support you. They understand that it is normal to feel overwhelmed and are waiting to help you when you are ready.

If you are undiagnosed, there are organizations you can turn to with that specific focus. There are also umbrella organizations, like Global Genes, that can serve as an anchor and source of information and encouragement while you continue your search.

Get in Touch

Here are some ways you can connect to a community, even if there are only a handful of other people living with the same condition across the globe.

Global Genes RAREConcierge

Provides a customized list of available resources, patient advocacy groups and disease-specific information, as well as personalized support from a network of healthcare providers, researchers and advocacy leaders.

Genetic and Rare Disease Information Center

Provides a database of disease information, organizations that provide support and services, research studies, clinical trials, and genetic testing, services, and phone support if you have additional questions.

Managing Your Care

Whether you are diagnosed, undiagnosed or have had a diagnosis corrected, genetic conditions are complicated to manage because they often require care from many different specialists. It's not unusual to have to balance appointments with a dozen different specialists—sometimes at different hospitals.



RARE TIP

“You get to the point where it doesn't matter why. The question is ‘What?’ What are we going to do? What is next and how can I advocate for my child? There is nothing you won't do to get your child help.”



– JANIS CREEDON, RARE CAREGIVER AND ADVOCATE

If you receive care at a major medical center, there may be a patient navigator on staff to help you make arrangements for these visits. You can also check with your insurer to see if they can provide a case manager to help you. Depending on your age and condition, a complex care team, pediatrician, neurologist, immunologist, or other specialist may coordinate care. But often the job of managing medical care will be yours.

Tips for Coordinating With Your Rare Disease Medical Team

Courtesy of Dr. Nicole Glenn, Rare Mom and Pediatrician, and The Disorder Channel

1. Have a one-page “cheat sheet” with the key “must know” information.
2. Be prepared for the appointment, write down questions, set the agenda and goals.
3. Learn who the “team members” are and what their role is.
4. Identify your quarterback, or prepare to be the lead yourself if necessary. Your primary care physician may not be the one to lead. Sometimes a specialist like a neurologist might take the lead.
5. Help your team members communicate, verify the plan, and close the loop across the whole care team.
6. Set clear follow up plans, establish communication channels for additional questions, know who to call for concerns.
7. Ask for help. It's ok to get more opinions!

As seen on *Once Upon a Gene TV* episode 4

Organizing Your Medical Information

Staying organized is important for patients and caregivers living with rare diseases. Having a system to help you maintain your schedule of appointments, track symptoms, order medications, manage a special diet, and access health and insurance data will make life easier to manage. It's even more helpful if your record keeping system is portable—either digital or a care notebook.

Easy access to information about your rare disease and how to best manage it is especially important when you receive care at different hospitals.

In case of emergency, it's best to keep information about your condition in several places to increase the chance that paramedics and emergency personnel will find it (on a medical alert bracelet, on a wallet card, on a form near your medicine bottles, etc.).



RARE TIP

“Have a business card that explains the basics of your disease to hand out to strangers who stare or comment.”

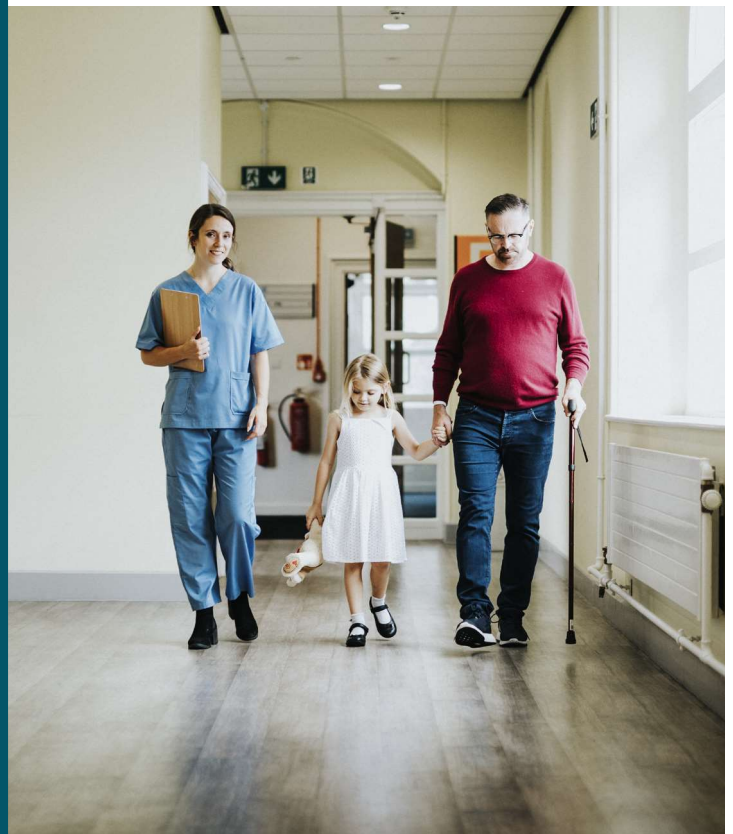


– BECKY BENSON, MOM TO MISS ELLIOTT, TAY-SACHS

What to Bring to Medical Appointments

From Parvathy Krishnan, Rare Caregiver and Global Genes Foundation Alliance Manager

1. Electronic medical record app containing updated lab results, medications, contact info for doctors and allergists
2. Your insurance card and cards related to your medical care (port, G-tube, pacemaker, etc.)
3. Home health agency related papers
4. Medical Order for Scope of Treatment (MOST) form, if you have one
5. List of concerns or information you'd like to ask your doctor about



Here are some resources to help you keep the information you need at your fingertips.

American Academy of Pediatrics National Center for Medical Home Implementation

Step-by step instructions on how to build a care notebook, plus an exhaustive list of information you will need to track, as well as forms you can download and fill out.

Exceptional Children's Assistance Center

Describes records and information to track and discusses the value of paper vs. digital and app record keeping systems.

NIH/NCATS Genetic and Rare Diseases Information Center (GARD)

Searchable list of disease information and orphan drugs.

Office of the National Coordinator for Health Information Technology

Provides the basics on how to access your health records and how to select apps to make your health information portable and easy to share.

Rare List

Information about 7000 rare diseases, support organizations, news, and clinical trials.

What to Put in a Hospital Go-Bag

A list of essentials for unexpected hospital visits.

Accessing Treatments

Only 5 percent of rare diseases have approved treatments. For the other 95 percent, physicians focus on managing symptoms and minimizing the health impact of the disease while waiting for treatments to be developed.

A small number of rare disease communities have developed clinical practice guidelines that contain

recommendations for diagnosis and treatment. Some also have designated centers of excellence staffed by teams of healthcare professionals who specialize in the treatment of a particular rare disease.

Finding a specialist who has experience with your disease, or managing your symptoms if you are undiagnosed, may be challenging. But it's definitely worth

the search. There are several ways to find specialists including contacting a patient advocacy group, looking for researchers who have conducted or are conducting clinical trials, and contacting authors of articles published in the medical literature.

Looking for a Specialist?

The National Institutes of Health Genetic and Rare Diseases Information Center (GARD) provides tips on [*how to find rare disease specialists*](#):

Even if there are no approved treatments for your rare disease, there may be clinical trials available. Your physician is the best source of information on which trial may be appropriate for your specific condition. But the decision on whether or not to enroll is a shared one. Educating yourself on what is required to be in a trial and what risks and benefits are involved is important to help you make this decision.

If you live outside the United States and there are no clinical trials or approved treatments available in your country, ask your physician if you might qualify for a pharmaceutical company's compassionate use program. Other options include crowdsourcing to pay for medications and contacting local or national healthcare officials.

How to Find a Clinical Trial

Begin with your medical team. They can connect you to a study site or principal investigator (PI) if you qualify.

The NIH/NCATS [*Genetic and Rare Diseases Information Center*](#) (GARD) provides in-depth information on clinical trials, types of studies, the process of informed consent, and what to consider in choosing whether or not to enroll.

For both U.S. and international clinical trials, you can find information on clinicaltrials.gov or by contacting disease-specific organizations.



Financial Literacy

Living with a rare disease is a financial challenge, as well as a medical one. There are many sources you can access to help with the financial burden. But it will likely require some study and self-advocacy to get the best care.

Here is a list of Global Genes' comprehensive financial advocacy resources, designed to serve as a reference for many of the questions you will have.

- Financial Literacy and Navigation
- Partnering with State and Federal Representatives
- Understanding and Navigating Medicaid
- Accessing and Maintaining Social Security Benefits
- Access to Newborn Screening
- *Financial Advocacy in RARE: Navigating the U.S. Health System for Young Adults*

Also from the NIH:

- *Tips for Finding Financial Aid*
- *Help with Travel Costs*

Coping and Hoping

Living with a chronic disease is always challenging, whether you have a diagnosis or not. But for rare disease patients and caregivers, there is the added stress of living with a condition that few people understand.



PRO TIP

“A whole range of emotions is possible and valuable. You may be scared, confused, angry, and may switch back and forth. It’s all normal and requires checking in with yourself to see how you are doing.”



– KATHLEEN BOGART
MOEBIUS SYNDROME
ADVOCATE, ASSOCIATE
PROFESSOR, SCHOOL OF
PSYCHOLOGICAL SCIENCE,
OREGON STATE UNIVERSITY

Adults with rare diseases have a 70 percent greater risk for depression and anxiety, according to Kathleen Bogart, associate professor in the school of Psychological Science at Oregon State University. Bogart was diagnosed with Moebius syndrome at the age of two and has devoted her career to studying the psychosocial impact of living with rare disorders and disability.

Problem Focused vs. Emotion Focused Coping

Psychologists talk about two main ways of coping. Both are valuable and can be useful in different circumstances, according to Kathleen Bogart.

- **Problem focused coping** involves handling stress by facing it head-on and taking action to resolve the underlying cause.
- **Emotion focused coping** works when you are in a situation that is not solvable at the moment. You regulate your feelings and emotions about the problem by meditating, doing art, spending time with people you care about, or writing in a journal.

Rare disease also takes a physical and mental toll on caregivers. In a 2018 study of Rare Disease Caregiving in America conducted by Global Genes and the National Caregivers Alliance, seven out of 10 caregivers of children under the age of 18 reported high levels of emotional stress.



RARE TIP

“You’ve likely heard of PTSD. In rare disease we talk about CTSD: chronic traumatic stress disorder.”



– DANIEL DEFABIO, ASSOCIATE DIRECTOR, COMMUNITY ENGAGEMENT, GLOBAL GENES AND CO-FOUNDER OF THE DISORDER CHANNEL

One emotion caregivers often experience is grief. “You may find yourself asking, ‘Did I not do my due diligence? Should I have done more?’” Bogart says. “Remember that parents have the best interests of their kids at heart and have done the best they could in the situation they found themselves in. It’s valid to have these feelings. But looking back is not productive or healthy.”



PRO TIP

“Grief is anything that causes a situation that I did not expect, I did not ask for, and I don’t like the result it is bringing.”



– BECKY SANSBURY, DIRECTOR OF CAREGIVER SUPPORT, RARE DISEASE INNOVATIONS INSTITUTE

Bogart recommends seeking psychological support, especially from someone who has experience with chronic illness or rare disease. She also

suggests connecting to a rare disease organization like Global Genes, or a disease-specific group if there is one.

“Community is so important for the rare disease community, most people feel they don’t get enough support through the health and mental health system,” Bogart says.



RARE TIP

“Take care of yourself and you’ll be better able to care for others.”



- NANCY KESSLER, BOARD MEMBER, SYNGAP RESEARCH FUND

“You have to be intentional about meeting others,” Bogart says.

“Organizations like Global Genes put on conferences that enable people to come together and meet each other and learn about advocacy and support. It’s wonderful to be surrounded by people who understand without an explanation and may look like you as well. Organizations can connect you to vital mentors and peers for social support and information as you move forward. That’s so important.”



Getting Back on the Road



Becky Sansbury, director of Caregiver Support for the Rare Disease Innovations Institute and author of the book, *After the Shock: Getting You Back on the Road to Resilience When Crisis Hits You Head On*, says the things you need to stay balanced are like the four wheels on a car.

- **Comfort** What brings you comfort each day? A piece of music? Coffee in the morning? A soft sweater? The things that comfort you in good times may be the things that will help you through really tough times.
- **Control** When your life feels out of control, try to get control in small things. Ask others to help you if needed. Your life will be full of big decisions and little ones. Each one helps get you where you need to go.
- **Community** Look for people who will support you and provide good distractions. If they can't be found close by, look online.
- **Connection** Stay in touch, whether it is with nature, a group you are a part of, a philanthropy you support, music, art, sports, or politics.

"Put on the frame of experience: your own and that of your medical team and counselors," Sansbury says. "Fuel yourself with resources and let the steering committee of your beliefs guide you.

"Remember that when you are walking into the unknown, it's important to know you can prepare. But also prepare not to be prepared.

"Your GPS for life before may not fit now and that's ok. Pack up our assumptions about life and put them in the trunk awhile. Somehow you will get down this road. You may have detours, hit potholes, bump into other cars, or find that a bridge is closed. But bit by bit, you'll get there."

Glossary

Carrier A carrier is an individual who carries and is capable of passing on a genetic change associated with a disease and may or may not display disease symptoms. Carriers are associated with diseases inherited as recessive traits. <https://www.genome.gov/genetics-glossary/Carrier>

Chromosomes Chromosomes are thread-like structures located inside the nucleus of animal and plant cells. Each chromosome is made of protein and a single molecule of deoxyribonucleic acid (DNA). Passed from parents to offspring, DNA contains the specific instructions that make each type of living creature unique. <https://www.genome.gov/about-genomics/fact-sheets/Chromosomes-Fact-Sheet>

Clinical Practice Guidelines Recommendations on how to diagnose and treat a health condition. <https://www.ncbi.nlm.nih.gov/books/NBK390308/>

Gene The gene is the basic physical unit of inheritance. Genes are passed from parents to offspring and contain the information needed to specify traits. Genes are arranged, one after another, on structures called chromosomes. A chromosome contains a single, long DNA molecule, only a portion of which corresponds to a single gene. Humans have approximately 20,000 genes arranged on their chromosomes. <https://www.genome.gov/genetics-glossary/Gene>

Geneticist A geneticist is a doctor who studies genes and heredity. <http://www.genesinlife.org/testing-services/working-healthcare-professionals/geneticist>

Genetics Human genetics is a branch of biology that studies how human traits are determined and passed down among generations. <https://www.ashg.org/discover-genetics/genetics-basics/>

Genetic counselor Genetic counselors are professionals who have specialized education in genetics and counseling to provide personalized help to patients who need to make decisions about their genetic health. <https://www.nsgc.org/page/whoaregeneticcounselors-473>

Genetic disease A disease or condition that is caused by a change in a person's genetic makeup that may or may not have been inherited from a parent. <https://www.aboutgeneticcounselors.org/Rare-Disease-Genetics>

Genetic mutation (see variant)
A gene mutation is a permanent alteration in the DNA sequence that makes up a gene, such that the sequence differs from what is found in most people. <https://medlineplus.gov/genetics/understanding/mutationsand-disorders/genemutation/>

Genetic test Genetic testing is a type of medical test that identifies changes in chromosomes, genes, or proteins. The results of a genetic test can confirm or rule out a suspected genetic condition or help determine a person's chance of developing or passing on a genetic disorder. <https://medlineplus.gov/genetics/understanding/testing/geneticstesting/>

Genetic variant Historically, disease causing variants were called mutations. To reduce confusion, all genetic changes—whether they cause a medical condition or have no impact at all—are now called variants. Genetic variants are classified on a 5-point scale: Pathogenic, Likely Pathogenic, Variant of Uncertain Significance, Likely Benign, Benign.

Pathogenic variants are proven to be disease causing. Likely pathogenic variants are suspected to be disease causing with ~90% certainty. Variants of Uncertain Significance do not have enough scientific evidence to know for certain if the variant is disease causing or not. Likely Benign variants are suspected to be Benign with ~90% certainty. Benign variants have been proven not to be disease causing, often because they are seen in many other individuals in the general population who do not have the health condition associated with the gene. https://cser-consortium.org/system/files/attachments/cser_provider_toolkit.pdf.pdf

Inheritance The process by which genetic material is handed on from parent to child. It's why members of the same family tend to have similar characteristics. <https://www.yourgenome.org/facts/what-is-inheritance>

Inherited disease A disease or condition caused by a change in a person's genetic makeup that was inherited from one or both parents.

<https://www.aboutgeneticcounselors.org/Rare-Disease-Genetics>

Proteins Proteins are an important class of molecules found in all living cells. A protein is composed of one or more long chains of amino acids, the sequence of which corresponds to the DNA sequence of the gene that encodes it. Proteins play a variety of roles in the cell, including structural (cytoskeleton), mechanical (muscle), biochemical (enzymes), and cell signaling (hormones).

<https://www.genome.gov/genetics-glossary/Protein>

Variant of unknown significance A variation in a genetic sequence for which the association with disease risk is unclear. Also called variant of uncertain significance, unclassified variant, and VUS. <https://www.cancer.gov/publications/dictionaries/genetics-dictionary/def/variant-of-uncertain-significance>

Whole exome sequencing (WES)

Exons are pieces of an individual's DNA that provide instructions for making proteins. All the exons in a genome are known as the exome, and the method of sequencing them is known as whole exome sequencing. <https://medlineplus.gov/genetics/understanding/testing/sequencing/>

Whole genome sequencing (WGS)

Whole genome sequencing determines the order of all the nucleotides in an individual's DNA and can determine variations in any part of the genome. <https://medlineplus.gov/genetics/understanding/testing/sequencing/>

Resources

General Information

[American Board of Genetic Counseling](#)

Questions to ask about diagnosed inherited conditions, genetic testing and future care.

[National Society of Genetic Counselors](#)

In-depth, family friendly information on what genetic counselors do, what genetic counseling involves, the genetic testing process, and how you'll receive results.

[American Society for Cell and Gene Therapy Patient Education](#)

Information on identifying a genetic disease and next steps with a diagnosis.

[NIH/NCATS Genetic and Rare Diseases Information Center](#)

Offers a comprehensive online database of information on rare diseases, how to find a specialist, financial information, and phone support.

[Rare Disease: What Role Do Genetics Play](#)

Webinar hosted by the National Society of Genetic Counselors, presented by Kelly East, MS, CGC.

[The National Economic Burden of Rare Disease Study Summary](#)

Comprehensive assessment of the total economic burden of rare diseases in a single year, from the EveryLife Foundation.

How to Locate a Genetics Professional

[American Board of Genetic Counselors](#)

A directory of genetic counselors by practice area.

[Australasian Society for Genetic Counsellors](#)

Represents over 280 members across Australia and New Zealand.

[British Society for Genetic Medicine](#)

Information for the public, patients and families, including organizations offering information and support.

[Canadian Association of Genetic Counsellors](#)

Provides a search tool to locate a genetics clinic in Canada.

[Find a Genetic Counselor Directory](#)

This tool from the National Society of Genetic Counselors offers access to more than 3,300 genetic counselors in the United States and Canada. You can filter searches for “live” or “telehealth.”

[Board of Genetic Counseling, India](#)

Professional organization dedicated to education, training, and practice.

[Organization for Rare Diseases India](#)

A national umbrella organization representing the collective voice of all patients with rare diseases in India.

[International Genetic Counseling Organizations](#)

A table of relevant websites for genetic counseling professional groups globally.

Understanding the Language of Genetics

[*NIH/NCATS Genetic and Rare Disease Information Center Glossary*](#)

Genetic and medical terms used on the GARD website.

[*National Society of Genetic Counselors*](#)

Genetic testing glossary of terms.

[*National Human Genome Research Institute Glossary*](#)

A talking dictionary of genetics terms.

Interpreting Results

Medline

[*Do all variants affect health and development?*](#)

Understanding mutations and disorders.

[*Understanding Testing*](#)

How to interpret the results of your genetic tests.

Help for the Undiagnosed

[*Becoming an Empowered Patient: A Toolkit for the Undiagnosed*](#)

Information designed to move you from feeling afraid and anxious to feeling prepared, confident and educated on how to manage the next steps in your healthcare journey.

[*Looking for Answers in Uncertainty - Managing When There is No Diagnosis or Prognosis*](#)

Managing psychosocial and emotional journey of having a child without a diagnosis.

National Human Genome Research Institute

Help for undiagnosed children

[Undiagnosed conditions in a child FAQ](#)

Help for undiagnosed adults

[Undiagnosed condition in an adult FAQ](#)

[NIH/NCATS Genetic and Rare Diseases Information Center \(GARD\)](#)

Guide to research programs, clinical trials, and financial advocacy for the undiagnosed.

[Rare Undiagnosed Network \(RUN\)](#)

Dedicated to empowering rare and undiagnosed patients and their families with genomic information and community through advocacy, networking, and support.

[Syndromes Without a Name UK \(SWAN\)](#)

Help for the undiagnosed, part of the Genetic Alliance UK.

[Systematic reanalysis of genomic data improves quality of variant interpretation](#)

A study that supports the benefits of reanalyzing whole exome sequencing and whole genome sequencing data.

[Undiagnosed Diseases Network](#)

A research study funded by the [National Institutes of Health Common Fund](#). Its purpose is to bring together clinical and research experts from across the United States to solve the most challenging medical mysteries using advanced technologies. The application is patient-driven and available online.

[Undiagnosed Rare Disease Patient Registry](#)

A natural history study of undiagnosed rare disease patients conducted by the National Organization for Rare Disorders (NORD).

[When Undiagnosed is your Diagnosis](#)

Podcast interview with Gina Szajnik, executive director of the Rare and Undiagnosed Network and Matt Might, director of the Hugh Kaul Personalized Medicine Institute at the University of Alabama at Birmingham School of Medicine.

Disease Information

[Global Genes' RARE List – powered by Genetic Alliance](#)

Information about specific diseases, support organizations and clinical trials.

[National Organization for Rare Disorders](#)

Rare disease database.

[NIH/NCATS Genetic and Rare Diseases Information Center \(GARD\)](#)

Searchable list of disease information and orphan drugs.

[Orphanet](#)

Alphabetical list of diseases, clinical signs and symptoms, genes, and emergency guidelines.

Managing Care

[American Academy of Pediatrics National Center for Medical Home Implementation](#)

Step-by-step instructions on how to build a care notebook, plus an exhaustive list of what you will need to collect with forms you can download and fill out.

[Disorder: The Rare Disease Film Festival](#)

How to coordinate a medical team that includes a lot of specialists.

[Exceptional Children's Assistance Center](#)

Describes what records and information to track along with a discussion of the value of paper vs. digital and app record keeping systems.

[Office of the National Coordinator for Health Information Technology](#)

How to access your health records, including a discussion of your right to health information and how to choose apps.

[What to Put in a Hospital Go-Bag](#)

A list of essentials for unexpected hospital visits.

[Global Genes Financial Advocacy in Rare Disease Toolkit Series](#)

Coping and Hoping

[After the Shock: Getting You Back on the Road to Resilience When Crisis Hits You Head On](#)

Becky Sansbury's recommendations for coping.

[Disability is Diversity: What Everyone Should Know About Ableism](#)

Kathleen Bogart's Psychology Today Blog.

[Disorder: The Rare Disease Film Festival](#)

A rare disease Mom's tips for those newly diagnosed.

[Healthy Coping Skills for Uncomfortable Emotions](#)

Describes problem-focused and emotion-focused coping skills.

[Once Upon a Gene TV, Episode 4 , Dr. Nicole Glenn](#)

Conversation with rare mom and pediatrician Nicole Glenn and Rob Long of Uplifting Athletes to talk about quarterbacking your medical team.

[Rare Disease Caregiving in America](#)

A study of the unmet needs of 1406 rare disease caregivers conducted by Global Genes and the National Caregiver Alliance.

[Words of Wisdom from Jessica Fein](#)

An experienced rare caregiver's perspective.

Patient Stories

[Caren's Story](#)

A rare disease diagnosis that was 64 years in the making.

[Tess is Not Alone: A USP7 Story](#)

How a family who thought their daughter was one-of-a-kind found their community.

References

- Bogart K. Disability is diversity: What everyone should know about ableism. Psychology Today. <https://www.psychologytoday.com/us/blog/disability-is-diversity>. Accessed April 2021
- Do all gene variants affect health and development? Medline. <https://medlineplus.gov/genetics/understanding/mutationsanddisorders/neutralmutations/> Accessed April 2021.
- Morin A. Healthy coping skills for uncomfortable emotions: Emotion-focused and problem-focused strategies. <https://www.verywellmind.com/forty-healthy-coping-skills-4586742>. Very Well Mind. Accessed April 2021
- National Economic Burden of Rare Disease Study. Every Life Foundation. https://everylifefoundation.org/wp-content/uploads/2021/02/The_National_Economic_Burden_of_Rare_Disease_Study_Summary_Report_February_2021.pdf. February 2021.
- Rare disease caregiving in America. National Alliance for Caregiving and Global Genes. <https://globalgenes.org/resources/rare-disease-caregiving-in-america/> February 2018
- Sansbury B. After the Shock: Getting You Back on the Road to Resilience When Crisis Hits You Head On. Real Life Communication; 2015.
- What do the results of genetic tests mean? Medline. <https://medlineplus.gov/genetics/understanding/testing/interpretingresults/>. Accessed April 2021
- Rare Disease Caregiving in America
A study of the unmet needs of 1406 rare disease caregivers conducted by Global Genes and the National Caregiver Alliance.
- Healthy Coping Skills for Uncomfortable Emotions
Describes problem-focused and emotion-focused coping skills.
- <https://pubmed.ncbi.nlm.nih.gov/33108040/>

Rare Story



RARE TIP



“If I could go back in time, there’s so much I would want to say. First of all, I would say, ‘You’ve got this.’ But I’d also say, ‘This is a marathon, not a sprint.’”

– JESSICA FEIN, RARE DISEASE CAREGIVER
AND MERRF ADVOCATE

My daughter Dalia, 16, has MERRF Syndrome, a rare mitochondrial disease that affects about two in a million.

We adopted Dalia from Guatemala when she was six months old. Early on I had a hunch that something wasn’t quite right with her development—her speech was garbled and her gait was wobbly. But the doctors told us she just needed time to catch up and early intervention said she didn’t qualify for services. And so I questioned myself: Am I overreacting? Am I being a helicopter Mom? But that parental instinct is strong, and when you have the feeling that something is wrong, you need to honor it.

I convinced the doctor to do a hearing test when my daughter was four and discovered she had hearing loss. Because the hearing loss was unexplained, doctors recommended genetic testing. From there, things moved quickly. She was diagnosed with MERRF Syndrome. We were “lucky” in a way, because the diagnosis came easily. But the diagnosis was crushing. Honestly, I didn’t even know what mitochondria were, so it was really hard to understand the gravity

of the doctor's decree. He started pulling out charts and telling us about proteins and energy. We were trying to get a handle on how serious it was. We did what everybody does: we became addicted to Googling. And that was a double-edged sword because we learned a lot of important information and also a lot of devastating information.

What I would tell someone who is newly diagnosed is to give yourself some time. Let the news sink in and settle in a bit. The disease is going to define a lot of your life, but it does not define your child and it does not define you. It's going to be a long and complicated road. It's traumatic. It's tragic. But you're going to get through it. You will rise to the challenge if for no other reason than you don't have a choice.

The immediate feeling is total isolation. Not only might you be the only person you know with a sick child, but you are most likely the only one dealing with this specific diagnosis. We were able to connect to other families living with mitochondrial disease. But connecting with other rare parents is equally useful, because it doesn't matter exactly what the disease is, the feelings and the practicalities are often very similar.

Another way in which I am fortunate is that I have a strong partnership with my husband. My daughter is 16 now and has completely lost her ability to move. She is attached to a ventilator and fed through a g-tube. Suffice to say, her care involves a lot of tubing. I was so intimidated by it at first, so my husband said, "I'm the tubing guy." It was the most romantic thing he'd ever said to me.

The most valuable thing my husband and I were able to give each other was the gift of time. We each have one night off on our own and we also take one night off together. In order for us to go out, we need a nurse and a babysitter, but it's worth it. Every single week we get in the car and say the same thing: "We have never needed a date this much."

Grief can show up unexpectedly. Years after the diagnosis, we were out on a date and in between appetizers and cocktails, I started sobbing. My husband said, "Are you feeling okay? What's wrong? Should we leave?" Suddenly, out of nowhere, I was thinking about

the fact that I'd never go wedding dress shopping with our daughter. She was seven at the time. I realized later that I was grieving the loss of what I thought her life would be like, and, by extension, what mine would be like.

The question of grief is a really complicated one. On the one hand, I'm not grieving my daughter. As long as she's with me, I won't be grieving her. But there is also anticipatory grief and grief for all that she—and we—have lost, and that's okay.

One day after my father died, I found a letter he had written to me years earlier. He talked about the world being so loud and chaotic, and that it's too much for us to create a world of beauty. But you can create corners of beauty. There is no light at the end of the tunnel for my daughter. It's going to get worse. But we can still try to make each day as good as it can be. ***We can make it a beautiful tunnel.***

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