Global Genes®
RARE Toolkits

SEPTEMBER 2021

Making Informed and Shared Decisions About Genetic Testing and Clinical Trial Participation

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

When healthcare providers work together with patients and caregivers to make decisions about tests, treatments, and procedures, the process they use is called **shared decision-making**. Making decisions together helps ensure that your healthcare choices are based on clinical evidence about risks and benefits, your own personal preferences, and your values.

PRO TIP

"Shared decision-making is another way of saying, 'thinking things through together.' If you start thinking about your values, goals, and priorities before major appointments, you will have a better sense of which questions are most important to ask and you will make it easier for your healthcare provider to focus your conversations around what matters to you. "

– ELEANOR GRIFFITH, FOUNDER, GREY GENETICS

Studies have shown that people who participate in healthcare decisions are more likely to report:

- better overall quality of life
- satisfaction with treatment
- fewer regrets about treatment choice

Knowing how to make shared decisions is valuable throughout your rare disease journey. But it's especially helpful when you're making choices related to **genetic testing**, **gene therapy**, and participating in **clinical trials** for therapies in development.

More than 70 percent of rare diseases are genetic in origin. Our knowledge about these diseases and how to treat them is growing rapidly. But there is still so much that is not known. There are rarely clear answers—and the answers we have are never "one size fits all."

In this toolkit, you'll learn how to consider what's right for you—and how to apply shared decision-making skills to the choices you make related to genetic testing and gene therapy clinical trials.

Understanding Shared Decision-Making

There was a time when physicians made most healthcare decisions with little input from patients. But over time, we learned that patients do better if they have more of a voice in the process.

Making healthcare decisions comes naturally to some people. “My body, my decision,” makes sense to them. Other people, especially those from varied cultural backgrounds, may not have considered the idea that physicians are experts in medical matters, but patients are experts too—in living with their disease. They have a right to share in healthcare decisions.

If you are new to shared decision-making, keep in mind that it’s a skill you learn by practicing it. Throughout your rare disease journey, you will be faced with choices. By asking questions, gathering facts, considering your beliefs, preferences, values and priorities, and sharing your thoughts with your healthcare provider, you’ll gain confidence in your ability to make choices that are right for you and your family.

Shared Decision-Making Tools

Making healthcare decisions can be challenging. The language of medicine is unfamiliar to many people and the choices often carry a great deal of emotional weight. Putting everything down on paper will help you identify information gaps and clarify your thoughts and feelings. Here are some tools to help you.

Getting Started

The "Ask Share Know" Approach

Ask Share Know, is a website sponsored by the Australian General Practice (GP) network. They recommend a three-step approach to improving shared decision-making.

Ask

When you are confronted with a choice of healthcare options, ask three questions:

1. What are my options?
2. What are the possible risks and benefits?
3. How likely are these risks and benefits?

Ask for an explanation if there is anything you don't understand. If your concerns aren't fully addressed, ask where you might be able to get more information.

Share

The more information you share about medical history, individual risk factors, lifestyle preferences, and what tests or treatments you would prefer, the more your healthcare professional will be able to give you the information you need.

Know

Knowing how likely the benefits and risks are is important. Medical tests and treatments are seldom 100 percent accurate and effective. Knowing what the procedure involves and what the impact will be on quality of life will also help you choose the best option for you.

You can find more details about how to use this approach, along with role play videos of conversations between patients and physicians on *Ask Share Know*.

PRO TIP

"Take your time. Talk to people you feel close to. But be selective. You can become overwhelmed with too many opinions. Once you make the decision, don't look back. You made the best decision you could, and you don't want to live in the 'what-ifs'."

– NICOLE GLENN, RARE CAREGIVER, PEDIATRICIAN, INTERNATIONAL DELEGATE, THE YELLOW BRICK ROAD PROJECT

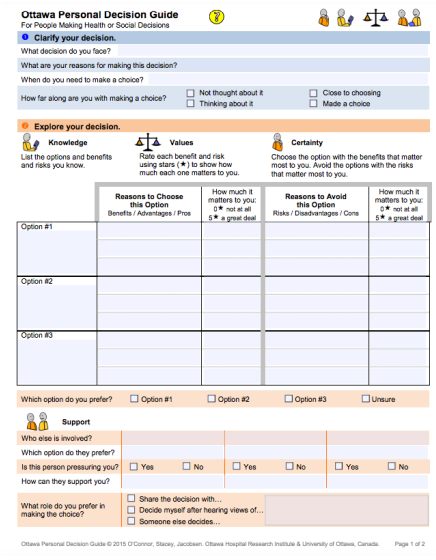
Comparing Your Options

The Ottawa Personal Decision Guide

When you are making a choice between two or more options (including not taking any action at all) how do you compare the pros and cons? The Ottawa Decision Guide provides a way of rating risks and benefits so you know what each choice means to you and feel comfortable deciding.

You can *access an interactive PDF* on the Ottawa Hospital website which will guide you step-by-step through the process of gathering information and identifying:

- What you know about each option's benefits and risks
- What matters most to you
- Where to turn for information, support, and advice
- What to do if you feel pressured by someone to make a choice
- Steps you can take if you are still uncertain about your decision


Ottawa Personal Decision Guide
For People Making Health or Social Decisions
Clarify your decision.
What decision do you face?
What are your reasons for making this decision?
When do you need to make a choice?
How far along are you with making a choice?
Not thought about it
Thinking about it
Close to choosing
Made a choice
Explore your decision.
Knowledge
List the options and benefits and risks you know.
Values
Rate each benefit and risk using stars (★) to show how much each one matters to you.
Certainty
Choose the option with the benefits that matter most to you. Avoid the options with the risks that matter most to you.
Reasons to Choose this Option
Benefits / Advantages / Pros
How much it matters to you: 0★ not at all 5★ a great deal
Reasons to Avoid this Option
Risks / Disadvantages / Cons
How much it matters to you: 0★ not at all 5★ a great deal
Option #1
Option #2
Option #3
Which option do you prefer?
Option #1
Option #2
Option #3
Unsure
Support
Who else is involved?
Which option do they prefer?
Is this person pressuring you?
Yes
No
How can they support you?
What role do you prefer in making the choice?
Share the decision with…
Decide myself after hearing views of…
Someone else decides…
Ottawa Personal Decision Guide © 2015 O'Connor, Stacey, Jacobsen. Ottawa Hospital Research Institute & University of Ottawa, Canada.
Page 1 of 2

The Ottawa Personal Decision Guide is available in multiple languages and in a format designed to be used by two people sharing their thoughts and working together to make a decision. You can also access videos on the *Ottawa Decision Guide website* that model the discussions you will have.

What is Informed Consent?

Before you go through some medical procedures, tests, or participate in a clinical trial or registry, you will talk to a healthcare provider or researcher who will explain:

- The procedure, protocol, plan, or test
- Its risks, benefits, costs, and limitations
- Other options, including no intervention
- Possible outcomes – both positive and negative

If you choose to proceed with the test or treatment, you will be asked to sign a document saying that you understand and give your permission.

This process is called **informed consent**. It is an ethical and legal requirement that's designed to protect your right to understand your options, and choose whether or not you proceed based on accurate, complete, and unbiased information.

PRO TIP

"Informed consent is a powerful tool. There is often a feeling that you have to do what your healthcare provider says, but the opposite is true. In most cases, they can't do anything if you don't say it is ok."

– MEGHAN HALLEY, RARE CAREGIVER, RESEARCH SCHOLAR, CENTER FOR BIOMEDICAL ETHICS, STANFORD UNIVERSITY

The informed consent process is a time to get answers to your questions. If anything you hear or read isn't clear, ask for more details. You have a right to receive information in understandable language. If the information is not provided in your native language, you can request a translation.

Going Deeper

Learn more about informed consent in this resource from Global Genes.
Informed Consent: Important for Treatment Decisions and Research

Your Healthcare Rights

1. The right to be treated with respect
2. The right to obtain your medical records
3. The right to privacy and confidentiality
4. The right to discuss risks, benefits, costs, and alternatives
5. The right to guidance based on your healthcare providers' professional judgment
6. The right to get information and answers to questions in understandable language
7. The right to refuse drugs, treatments, or procedures to the extent permitted by law
8. The right to get another doctor's opinion at your request and expense
9. The right to be advised of any conflict of interest your physician may have
10. The right to receive care without regard to race, color, religion, disability, sex, sexual orientation, national origin, or source of payment

Along with these rights, patients and caregivers have responsibilities. You are responsible for seeking care, being honest, providing complete health information, and sharing any needs or concerns you have.

Adapted from the AMA Code of Medical Ethics.

Some things to remember about informed consent are:

- It is an ongoing process of information-sharing, not a one-time event.
- Ask for a copy of the informed consent to take home and discuss with family, friends, and advisors. Review it as many times as you need before you feel confident about making your decision.
- It's not a contract. You may refuse to proceed with testing even after you have signed and samples have been drawn.
- You can also withdraw from a clinical trial at any time, for any reason.

Dealing with Uncertainty

Even when you ask all the right questions, making decisions on testing and treatment for rare disease is not simple because so little is known about these conditions. If you or your child has a disease that causes symptoms, but remains undiagnosed, you will have the added challenge of trying to decide what is best when you don't even have a name for the disease.

Under the circumstances, it's natural to feel a mix of emotions: sadness and frustration when you have limited options, confusion about what's best, and fear of making the wrong decision. When there is little evidence to guide you down one path vs. another, it may help to consider the possible impact of each choice on quality of life, alongside the potential medical risks and benefits. Regardless of which option you choose, you might have to accept living with a degree of uncertainty, knowing you've done your best to make an informed choice for yourself or your family member.

PRO TIP

"I would argue that physicians who are more willing to recognize and discuss the role of uncertainty in the recommendations they make regarding their patients' care, and who are willing to keep quality of life at the center of discussions with patients and families, will ultimately do what is best for their patients and their families."

– MEGHAN HALLEY, RARE CAREGIVER, RESEARCH SCHOLAR, CENTER FOR BIOMEDICAL ETHICS, STANFORD UNIVERSITY SCHOOL OF MEDICINE

In Their Words

Finding out how other rare disease patients and caregivers think through healthcare decisions may help you navigate the choices you face. Here are two good examples:

Deciding When Not to Treat Our Child with a Degenerative Disease
A first-person story by Daniel DeFabio, Global Genes' community manager and co-founder of the Disorder Channel, on making healthcare choices for a child with a rare disease.

When All You Have is Quality of Life: Making Decisions in the Face of Medical Uncertainty
Meghan Halley, a rare caregiver who is also a Stanford bioethicist, discusses shared-decision making for her undiagnosed son in this article published in the New England Journal of Medicine.

Deciding for a Child or Dependent Adult

Adult patients (unless they are incapacitated) have the moral and legal right to make decisions about their own medical care. Because young children and dependent adults do not have the ability to make complex medical decisions, parents/caregivers are typically the primary decision makers for them.

Children *do* play an important role in shared decision-making, however. Many patients under the age of 18 (under 16 in some countries) cannot legally provide informed consent, but they can and should provide their **assent** (agreement) for decisions that affect their health, life, and death.

Protecting and Empowering Your Child

Your pediatrician or other healthcare provider may include your child, as early as 7 years of age, in shared decision-making discussions about treatment by explaining procedures in language he or she can understand. Your child should be given plenty of time to ask questions and express any concerns before asking for his or her assent.

You can help nurture the ability to make good decisions by including your child in discussions at home and modeling good processes for thinking about risks, benefits, values, and preferences. As your child grows toward young adulthood, he or she will become an integral part of the shared-decision-making process in preparation for one day making these decisions independently.

The AMA's recommendations for *Pediatric Decision-Making* include:

- Evaluate minor (age range) patients to determine if they can understand risks and benefits and seek their **assent** to the extent that they are able to understand what the decision means.
- Negotiate a shared understanding of the pediatric patient's medical and psychosocial needs in the context of family relationships and resources
- Provide a supportive environment and encourage parents/guardians to discuss the child's health status with the patient.
- Offer education and support including putting the family in touch with others who have faced similar decisions and have offered their support as peers.

If you are considering clinical trials, pediatric assent is often a part of the discussions you will have. Although it's not a legal requirement like informed consent, many **Institutional Review Boards** (IRBs), the regulatory bodies that oversee clinical trials, require that minor children **assent** (agree to participate) beginning at about age 7. Children may also **dissent**, which means they do not agree.

Like informed consent, assent is not a one-time event. Your healthcare provider or research team may use videos, graphics, and other age-appropriate tools to explain what will be involved. Your child will be encouraged to ask questions. It may take several sessions before the child understands and assents or dissents.

Making decisions on behalf of your child may feel like a heavy responsibility, especially when the stakes are high. Remember that it is not something you have to do alone.

PRO TIP

"What I think about when I make decisions for my daughter is, 'How would it benefit her? ' I don't think, 'Will it make my life easier?'"

– NICOLE GLENN, RARE DISEASE CAREGIVER, PEDIATRICIAN, INTERNATIONAL DELEGATE, THE YELLOW BRICK ROAD PROJECT

ASSENT

Agree to participate

DISSENT

Do not agree to participate

Family Considerations

Sometimes healthcare decisions have an impact on not only the patient, but also immediate and extended family members.

For example, genetic tests may reveal information that will affect your relatives' decisions on whether or not they pursue genetic testing. Sharing genetic information with them could be viewed as helpful, alerting them to an unanticipated risk. But it could also create tension, confronting family members with fears and hardship they would prefer not to face.

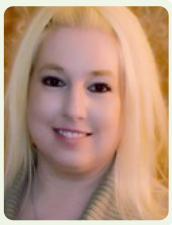
How to Discuss Genetic Disease with Your Loved Ones

This *toolkit from Global Genes* can help you plan the conversations you have with people who share your genes.

Families who receive a genetic diagnosis may also struggle with unwarranted guilt. Even the fear of guilt can impact the decisions people make. For example, expectant parents who learn their unborn child is sick may feel guilty about whether or not they choose abortion. Adult children who are diagnosed with an inherited condition may be reluctant to share the diagnosis with parents to spare them the pain.

RARE TIP

"Guilt happens in every culture, but it's more prevalent in some. In the Latino community, there are sometimes problems with genetic diagnoses because of the cultural respect for elders and ancestors. That is something we work through with education. It's not my fault. It's not your fault. It's science."



– KHRYSTAL DAVIS, RARE DISEASE CAREGIVER, FOUNDER TEXAS RARE ALLIANCE

Cultural and Religious Diversity

Your cultural and religious background may influence healthcare choices in a variety of ways, including by shaping:

- the values that guide you
- your long-term goals
- the priority you place on getting advice from family elders or other community members

In cultures with a tradition of marrying relatives such as cousins (consanguinity), for example, carrier screening may address the risk of having an inherited genetic condition. But the attitudes of the extended family and the risk of social stigma often impact decision-making.

PRO TIP

"When you are trying to engage elders in the family who have decision-making power, consider looking for a genetic counselor who can speak the same language and offer visuals on how recessive conditions come about. I would also recommend making connections to hospital chaplains of different faiths as we do in the UK."

– ASYA CHOUDRY, PROJECT MANAGER, VOCAL, MANCHESTER UNIVERSITY NHS FOUNDATION TRUST

Religious beliefs regarding pregnancy termination sometimes cause couples to rule out prenatal testing. A genetic counselor can help you consider all aspects of this decision, including the fact that termination is not the only option. If a problem is discovered, genetic testing can help you prepare for your child's medical needs and connect to a supportive community.

Ethical choices like these are both individual and cultural. Genetic counselors are trained to support you as you think through your values. They can provide facts you can discuss with a chaplain or religious leader. But the choice will ultimately be yours.

Overcoming Barriers to Shared Decision-Making

Shared decision-making is a conversation. Like any discussion, it requires active participation from all partners, each of whom has an important viewpoint to contribute. What should that discussion look like—and what should you do if you run into obstacles? Here are some ideas.

What's happening?	What might you say or do?
Your healthcare provider makes a recommendation without engaging in shared decision-making.	*Acknowledge the provider's expertise while asserting your need for more information. For example, "I appreciate your opinion. It would help me feel better about our next steps if I had a better understanding of all of the options first before moving forward."*
You're not sure whether you want to say "yes" or "no" and you feel pressured to make a decision.	*Give yourself the space and time to consider your options. Even if the situation is urgent, you can ask questions or request a break to talk to someone you trust. This will give you more time to formulate your response.For example, you might say, "Could you help me understand the thinking behind your recommendation?" Or "I'd like to take some time to think about it (or talk with my spouse, child, or other key person)?" Keep in mind that you have the right to say "no" and to request additional information to help you make the best decision possible.*
You are hesitant to proceed and are afraid of making a wrong decision.	*As rare disease patients and caregivers, we often have to make really tough decisions with little scientific evidence to guide us. Take your time to think through the range of possible outcomes of a given decision, how likely each outcome is to occur, and how you would feel if each of these outcomes came to be. Once you have made a decision, give yourself grace. You may regret decisions down the road, but try to remember that hindsight is always clearer, and you can only make the best decision you can with the information you have at the time.*
You consistently find communicating with your healthcare provider challenging.	*Unfortunately this is not uncommon. Many rare disease patients and caregivers will encounter at least one provider who is a poor communicator in their journey. Be polite but persistent in the pursuit of the information you need to make the decisions that are right for you or your family member. However, if the communication is chronically poor, you may need to consider finding a new provider.*

PRO TIP

"Don't feel pressured to do what you think your doctor wants you to do. He or she will provide you with excellent care no matter what choice you make."

– JENNIFER BRASWELL, EXECUTIVE DIRECTOR, ARM FOUNDATION FOR CELL AND GENE MEDICINE

What to Think About When Considering Genetic Testing

Genetic testing often leads to a life-changing diagnosis. Having a genetic diagnosis helps individuals receive appropriate care and helps communities grow their registries and natural history studies, advancing knowledge of the disease and accelerating the search for treatment.

Even if there is no treatment available, having a diagnosis may help you find better ways to manage the disease and advocate for services. The information you receive may be important for family planning or medical care for siblings.

Testing can also be useful even if you receive a genetic report with names of variants, but no official diagnosis. Having a name for your disease makes it easier to find your community – even if the name is “undiagnosed.”

Getting Connected

If your hospital or practice hasn't introduced you to a genetic counselor or geneticist, here are some resources to help you.

Find a Genetic Counselor
A search engine provided by the National Society of Genetic Counselors which provides both live and telehealth options.
https://findageneticcounselor.nsgc.org/?reload=timezone

How a Genetic Counselor Can Help
Find questions to ask and tips for locating a genetic professional in your area— as well as how to arrange an appointment via telehealth.
https://resource-hub.globalgenes.org/kb/article/298-gene-based-diagnosis-101/

National Coordinating Center for the Regional Genetics Networks
This organization works to improve access to quality genetic services for medically underserved populations.
https://nccrcg.org

Carrier Screening
A website and video from the Jewish Genetic Disease Consortium that explains who should consider screening, what screening is recommended, and how it's done.
https://www.jewishgeneticdiseases.org/genetics-and-carrier-screening/

Genetic Testing: Getting to a Diagnosis
Learn Genetics 101 and review the types of testing and results that can be expected.
https://resource-hub.globalgenes.org/kb/article/298-gene-based-diagnosis-101/

Global Genes RARE Concierge
https://globalgenes.org/rare-concierge/

Women with Rare Disease: The Reproductive Years
Includes a discussion of the challenge of family planning with rare disease.
https://resource-hub.globalgenes.org/kb/article/4-women-with-rare-disease-the-reproductive-years/

But as with all medical procedures, it's important to remember that there are both benefits and risks. For this reason, genetic testing requires a process of informed consent that will likely include information such as:

- General description of the test and purpose
- How the test will be done (for example, a blood sample)
- What the test results mean
- Physical or emotional risks associated with the test
- Whether the results might provide information for other family members
- What will happen to the sample after the test is complete
- Whether the results can be used for research purposes

RARE TIP

"Some families say 'no' to testing because they don't want to live with the fear each day. It's a personal decision. They don't see it as not wanting something—but rather wanting something different."

– PARVATHY KRISHNAN, RARE CAREGIVER, GLOBAL GENES FOUNDATION ALLIANCE MANAGER

Considering Genetic Testing

Here are some factors to consider as you think about the risks and benefits of genetic testing.

Health - The results you receive from genetic testing

- **May guide medical care.** You could be referred to a specialist or receive recommendations for a different diet, medications, or procedures such as surgery.
- **May reveal unexpected findings,** including the possibility that other family members or future children may be at risk for a genetic disorder.
- **May not be immediately useful,** although your doctors may suggest additional tests or recommend you have your data reanalyzed at some point in the future.

Emotions – You may feel

- **Disappointed** if your genetic tests did not reveal a diagnosis.
- **Frightened or sad** if the disease is chronic, progressive, or possibly fatal.
- **Relieved** to receive a diagnosis you have been seeking.
- **Overwhelmed** if you are facing complex medical care and decisions.

Quality of Life – The results of genetic tests

- **May connect you to a community** and people who will share your journey.
- **May change how you advocate** for supportive services and educational needs.
- **May enable early detection** of complications and early intervention to slow progress of the disease.

Finances – Genetic tests

- **May be costly,** but are often covered by insurance or Medicaid.
- **May not be used to determine coverage or rates** for your health insurance, because of the Genetic Information Non-Discrimination Act (GINA).
- **May prevent you** from obtaining long-term health insurance or life insurance.

Other things to consider before deciding on genetic tests:

- **Religious and cultural values** regarding genetic tests and their uses.
- **Impact on other family members** and how you will discuss the results.

What to Think About When Considering Gene Therapy Clinical Trials

Decision-making in rare disease is complex. There are often no clinical practice guidelines or standards of care that have been established. There are few recognized experts. And only 5 percent of the more than 7000 rare diseases have approved treatments.

As a result, the only interventions many rare disease patients have to choose from are those that are being studied by researchers in clinical trials. Participating in a clinical trial is an individual decision. It should be made by talking to your doctor about your medical options and considering your own needs and preferences.

Because participating in one clinical trial may disqualify you from others, it's also important to consider the risks and benefits of all options likely to be available to you. This includes your current treatment, other clinical trial options, and no intervention at all.

PRO TIP

"One of the best questions to ask is, 'What is this going to exclude me from in the future?'"

- DEBORAH COLLYAR, FOUNDER AND PRESIDENT, PATIENT ADVOCATES IN RESEARCH (PAIR)

If you are considering enrolling in a clinical trial, it's important to understand the difference between participating in clinical trial research and receiving an approved treatment.

What is a Clinical Trial?

Clinical trials are research studies performed in people. The goal of the research is to evaluate a medical, surgical, or behavioral intervention. Clinical trials are the primary way that researchers find out if a new treatment is safe and effective in people.

An intervention that is being studied in a clinical trial is new, untested, or different from the treatment that's currently used. It has not yet been proven to be safe or effective. It may not be better than, or even as good as, existing treatments.

What is an Approved Treatment?

Approved treatments have been studied in clinical trials and shown to be reasonably safe and effective for treating a particular disease or condition. These treatments are generally approved by a regulatory agency, such as the Food and Drug Administration (FDA) or the European Medicines Agency (EMA).

PRO TIP

"When you are among the first people to participate in trials, there are risks we can foresee and some that are unforeseeable. We are the research. It's not on paper."

–NICOLE GLENN, RARE CAREGIVER, PEDIATRICIAN, INTERNATIONAL DELEGATE, THE YELLOW BRICK ROAD PROJECT

Learn About Clinical Trials

These resources provide more in-depth information on rare disease clinical trials.

CISCRP Webinar Series: Navigating Rare Disease and Clinical Research
Patients and organization leaders describe clinical trials protocol, what it's like to participate and how to prepare for when the clinical trial ends.
https://www.ciscrp.org/events/webinars/webinar-rare-disease-clinical-trials-being-informed/

Clinical Research Participation Basics Roadmap
Interactive map that takes you through the basics of how to find, participate in and leave a clinical trial.
https://www.ciscrp.org/wp-content/uploads/2021/03/WOW-ClinResRoadMap-3.22.21.pdf

Courageous Parents Network (CPN): Evaluating the Clinical Trial Option
A digital resource offering the perspective of a principal investigator, study coordinator, patient advocate and families who have considered and/or participated in a trial. The unit includes videos, podcasts and blogs. Three downloadable guides and a Guided Pathway, a curated learning experience, are available in English and Spanish. CPN also offers a list of questions to ask about clinical trials.
https://courageousparentsnetwork.org/esp

Informed Consent for Clinical Trials
This FDA document outlines information that researchers are obligated to provide to participants before enrolling in a clinical trial.
https://www.fda.gov/patients/clinical-trials-what-patients-need-know/informed-consent-clinical-trials

Participating in a Clinical Trial: A Step-by-Step Guide for Rare Disease Patients and Families
A Global Genes resource that walks you through the process of joining and participating in clinical trials.
https://globalgenes.org/2021/05/07/putting-the-patient-at-the-center-of-rare-disease-clinical-trials/

What to Ask When You're Interested in a Clinical Trial
This resource, co-developed with rare disease advocates, helps you think about how easy or difficult participation in a trial would be for you and the people close to you. It also lists questions to ask regarding what can be done to make the hard things easier.
https://prahs.com/insights/trial-development-toolkit-what-to-ask

What Makes Gene Therapy Clinical Trials Different Than Other Clinical Trials?

Gene therapy involves changing the genes inside your body's cells to treat or stop disease. Because it targets the cause of genetic disorders rather than just lessening the symptoms, gene therapy holds promise for rare diseases.

The science of gene therapy is still in development. There are a limited number of gene therapies that have been approved by the Food and Drug Administration (FDA) for rare diseases.

Understanding Gene Therapy

Get up-to-speed with these reliable and easy-to-understand sources of information on cell and gene therapy.

American Society of Gene and Cell Therapy

The patient education section of this site provides basic information on gene therapy, a clinical trial finder and information on treatments for specific rare diseases.
https://patienteducation.asgct.org/

FAQ for Gene Therapy Clinical Trials

Answers to your questions about risks, benefits, and other considerations.
https://resource-hub.globalgenes.org/kb/article/271-cell-and-gene-therapy-for-rare-disease-clinical-trials-faq/

Healing Genes

An introduction to cell and gene therapy for the public and advocacy groups from the ARM Foundation for Cell and Gene Medicine.
https://healinggenes.org/news/

All clinical trials have potential risks and benefits. But the risks and benefits of gene therapy are unique. The potential benefit of gene therapy is that:

- It targets the cause of genetic disease and may provide a way to slow or stop the progression of serious rare diseases that can result in disability or death.
- By altering the genes that cause the disease, gene therapy also has the potential to maintain the benefit over a long period time without the need for frequent treatments.

The potential risks of gene therapy differ, based on both the type of gene therapy and the type of **vector** used to deliver it. Some risks can be serious or fatal.

What is a Vector?

Vectors are carriers used to deliver genetic material into a cell. They are typically derived from a virus. All of the viral genes are removed, and the vector is modified to deliver only therapeutic genes. Choosing a vector that is safe and effective at targeting the right cell type is a priority for researchers. Learn more on the American Society of Gene and Cell Therapy's Patient Education website.

Potential risks are:

- **Unwanted immune response**
  Your body's immune system could see the vector (virus) as an intruder and attack it. If the immune response is uncontrolled, it could cause inflammation and, in severe cases, organ failure. The immune response could also make the treatment ineffective.
- **Inadequate immune response**
  High doses of some viral vectors could be toxic to some people, especially if they are immune compromised.
- **Innate immunity**
  If you have previously been exposed to a virus used as a vector your immune system may attack and destroy it before it can be effective. Often trials require you to be tested for innate immunity to be eligible for the trial.
- **Targeting the wrong cells**
  The virus (vector) could potentially infect cells that were not targeted. This could damage healthy cells and cause additional disease.

Medical researchers and regulatory agencies are working to make sure that gene therapies are as safe as possible, and researchers are continually honing and improving their processes over time.

PRO TIP

"If there is safety data, have your clinicians evaluate the relative risk you are taking. Ask them if they think the immune response mitigation strategies are sufficient. Ask, 'Will this improve or maintain my quality of life?' so you can better weigh benefit and risk."

– ERIC CAMINO, VICE-PRESIDENT, RESEARCH AND CLINICAL INNOVATION, PARENT PROJECT MUSCULAR DYSTROPHY

Other Things to Consider

Logistics of the trial

Along with the risks and benefits of the therapy itself, there may be additional impacts on quality of life when participating in trials of specialized treatments, such as gene therapy. Often the trials are offered at a limited number of sites. If travel and time commitments are significant, it could affect your job, other family members' needs, and potentially have financial impacts. For example, it is important to understand if study-related travel will

be paid for in advance by the clinical trial or if your family will be reimbursed for it after the travel occurs.

RARE TIP

"Sometimes the most "local" option may not be the best fit for your family. Learn all of the details involved:

- ***What are the travel requirements?***
- ***What are the time commitments now and in the future?***
- ***Can you be away from home for an extended period of time?***
- ***What does this mean for the entire family?***

The pandemic has taught us that things can be overturned at any moment. If you can't begin or continue treatment as planned, what will happen?"

– LAURIE TURNER, FAMILY SERVICES MANAGER, NATIONAL NIEMANN-PICK DISEASE FOUNDATION

Because researchers want to understand how gene therapy affects your body long-term, clinical studies can last many years. You may be asked to undergo regular monitoring to see if the treatment continues to be safe and effective.

Potential for redosing

Although the hope is that gene therapy can be delivered in a single dose and remain effective for life, long-term effectiveness and durability is not known for individuals in clinical trials. If the effect is not durable, redosing may not be an option using current techniques. It's possible that you could be eliminated from the trial and you may not be eligible for any other trial, even with a washout period.

RARE TIP

"Make sure you understand all the things this treatment will entail long term. Is the more invasive treatment with greater benefits a better option? If it doesn't work, would you be eligible to try something different? Is it better to have a one-time treatment or one that will be repeated the rest of your life, knowing that repeat means risk?"

– VERONICA HOOD, RARE DISEASE CAREGIVER, RESEARCH COORDINATOR, DRAVET SYNDROME FOUNDATION

What happens if you wait?

Clinical trials have **inclusion and exclusion criteria**, such as age, specific gene mutation, and health history, that determine whether or not you're eligible to participate. If the disease is chronic and degenerative,

you may have to weigh the risks and benefits of enrolling in a clinical trial now vs. waiting for a better option, only to find you are ineligible due to age or deteriorating physical condition.

RARE TIP

"Talk to every single site. Consider every possibility. Put your name on the list for all of them because you don't know your chances of getting in. Spread your bets. It's about creating options for yourself. Educate yourself. Talk to your physician. Watch the videos. View the presentations. If you can stomach it, read the research papers, and make your own decision. The world is big with opinions and everyone's choice will be different."

– CHRISTINE WAGGONER, RARE DISEASE CAREGIVER, PRESIDENT AND CO-FOUNDER, CURE GM1

As you collect information, you will be able to better judge which of the options available to you best matches your personal needs and preferences. Some patients and caregivers prioritize quality of life and some prioritize quantity of life. Neither is right or wrong. What matters is how you feel about these important choices.

RARE TIP

When you make a decision based on collecting all the resources you can gather and giving yourself time to reflect, you will feel that you have done the best you can in that moment; and this will help you feel you were the best parent you could be for your child, which helps protect against decisional regret."

– BLYTH LORD, COURAGEOUS PARENTS NETWORK

Help From Advocacy Groups

One of the best ways to gain perspective on gene therapy—or any type of clinical trial—is to contact a patient organization. Patient leaders stay up-to-date with treatment developments and keep their community informed of research progress and opportunities to participate.

RARE TIP

"Receiving clarity and honest answers is essential when navigating clinical trials and gene therapy. We encourage individuals and families to ask questions and share their concerns, so they are secure in knowing the decisions they make are right for them. Their questions ultimately inform the programs we create and offer families, as well as the conversations we have with industry."

– DIANA PANGONIS, NATIONAL TAY SACHS & ALLIED DISEASES ASSOCIATION (NTSAD)

Some provide webinars featuring physicians or researchers who can explain the process of gene therapy, the risks and benefits and the regulatory side of approval. Patient organizations can also be a good resource for translating this information and making it understandable.

Talking to others who have been through the process can be helpful, too. You may decide to seek a second opinion from a specialist, gather information from advocacy groups, or talk to others who have faced similar decisions. But in the end, your choice should be based on what matters most to you.

RARE TIP

"You have to be ready. Do your research. And don't think it's going to be easy. It's not. It's a time commitment, going to labs, going to radiologists. But in the end, I look back and think it was all worth it."

– STEVE TEJIRAM, HEMOPHILIA PATIENT

Considering Gene Therapy Clinical Trials

Understanding as much as possible about the process of gene therapy and the trial protocol will help you be confident in your decision.

Health Impact

- Is there evidence about safety or efficacy of the treatment and the vector from this— or other similar trials?
- Will there be a placebo arm or different treatment arms with different dosages?
- What risks are involved in the treatment or administration?
- What procedures are involved in preparing participants for the trial?
- What procedures are involved in administration?
- Will I be prevented from participating in other trials?
- If the treatment doesn't work, is redosing possible?

Quality of Life impact

- What is the time and travel commitment?
- How will being in the trial affect my regular activities?
- How long will I be expected to participate?

Emotional Impact

- How will I feel if the treatment doesn't work or I'm eliminated from the trial?
- How will I feel if I wait for better options and my child doesn't meet the criteria?
- Will there be an impact on other family members?

Financial Impact

- What are the direct and indirect costs and what costs will be covered by insurance?
- Will I be required to pay for routine care?
- Will I be required to pay for travel and lodging?
- Is there financial assistance available?

Other Considerations

- Are there religious or cultural values that might impact my choice?
- Do I have any ethical concerns?
- Are there scientific advances or regulatory changes I should consider?

Glossary

Assent: Assent means a child agrees to take part in a trial, treatment or procedure. They can also dissent, which means they do not agree. *https://www.cancer.gov/about-cancer/treatment/clinical-trials/patient-safety/childrens-assent*

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*

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

Clinical Trial: A research study in which one or more human subjects are assigned to one or more interventions (which could include a placebo or control) to evaluate the effectiveness of the intervention. *https://grants.nih.gov/policy/clinical-trials/definition.htm*

Consanguinity: Genetic relatedness of individuals who are descended from at least one common ancestor. *https://www.cancer.gov/publications/dictionaries/genetics-dictionary/def/consanguinity*
aucherdisease.org/gaucher-diagnosis-treatment/treatment/enzyme-replacement-therapy/

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*

Gene Therapy: An experimental technique that uses genes to treat or prevent disease. Researchers are testing several approaches to gene therapy, including replacing a mutated gene that causes disease with a healthy copy of the gene, inactivating or "knocking out" a mutated gene that is functioning improperly and introducing a new gene into the body to help fight a disease.
https://medlineplus.gov/genetics/understanding/therapy/genetherapy/

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 change in a person's genetic makeup, which 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/mutationsanddisorders/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/genetictesting/

Gene Therapy: A technique that uses genes to prevent or cure disease by replacing a copy of a mutated gene with a healthy one, "knocking out" a mutated gene that is functioning improperly, or introducing a new gene into the body to help fight disease.
https://medlineplus.gov/genetics/understanding/therapy/genetherapy/

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.
https://cser-consortium.org/system/files/attachments/cser_provider_toolkit.pdf.pdf

Genomics: Genomics is the study of all a person's genes including interactions of those genes with each other and with the environment.
https://www.genome.gov/about-genomics/fact-sheets/A-Brief-Guide-to-Genomics

Inclusion and exclusion criteria: Attributes of participants that are essential for their selection to participate.
http://www.unm.edu/~rrobergs/604Lect2.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

Institutional Review Board: A group that has been formally designated to review and monitor biomedical research involving human subjects. In accordance with FDA regulations, an IRB has the authority to approve, require modifications in (to secure approval), or disapprove research. This group review serves an important role in the protection of the rights and welfare of human research subjects.
https://www.fda.gov/regulatory-information/search-fda-guidance-documents/institutional-review-boards-frequently-asked-questions

Shared Decision-Making: Shared decision making (SDM) is a communication process by which patients and clinicians work together to make optimal health care decisions that align with what matters most to patients.
https://mghdecisionsciences.org/about-us-home/shared-decision-making/

Standard of Care: Treatment that is accepted by medical experts as the proper treatment for a certain disease.
https://www.cancer.gov/publications/dictionaries/cancer-terms/def/standard-of-care

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

Vector: Vectors are the vehicles used to deliver genetic material into a cell. Vectors are typically derived from viruses, because viruses have proven to be very efficient at finding their way into cells. In order to make vectors safe to use, all of the viral genes are removed, and the vector is modified to deliver only therapeutic genes
https://patienteducation.asgct.org/gene-therapy-101/vectors-101

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

Clinical Trials

CISCRP Webinar Series: Navigating Rare Disease and Clinical Research
Patients and organization leaders describe clinical trials protocol, what it's like to participate and how to prepare for when the clinical trial ends.

Clinical Research Participation Basics Roadmap
Interactive map that takes you through the basics of how to find, participate in and leave a clinical trial.

Courageous Parents Network: Evaluating the Clinical Trial Option
A digital resource offering the perspective of a principal investigator, study coordinator, patient advocate and families who have considered and/or participated in a trial. The unit includes videos, podcasts and blogs. Three downloadable guides and a Guided Pathway, a curated learning experience, are available in English and Spanish. CPN also offers a list of *questions to ask* about clinical trials.

Data DIY
Educational program with training modules, workbooks, and live presentations that cover: the Why's and How's of Data Collection; Data Trusts and Governance; Developing Collaborative Research Networks; and Becoming a Data-Centric Community.

From Molecules to Medicine: Clinical Research
Global Genes' toolkit on how to find and begin participating in a clinical trial.

Gene Therapy Clinical Trials FAQ
A list of answers to your questions about what makes gene therapy trials unique.

Participating in a Clinical Trial: A Step-by-Step Guide for Rare Disease Patients and Families
How to learn about treatment options, what to consider, and what to expect

What to Ask When You're Interested in a Clinical Trial
This resource, co-developed with rare disease advocates, helps you think about how easy or difficult each trial would be for you and the people close to you who matter - and ask what can be done to make the hard things easier.

Genetics

Genes in Life
The Genetic Alliance's guide covers Genes 101, family history, testing, and research.

Help Me Understand Genetics
Medline's overview covers how genes work, how genetic conditions are inherited, genetic consults and testing, gene therapy and research.

Rare Disease: What Role Do Genetics Play?
Webinar hosted by the National Society of Genetic Counselors, presented by Kelly East, MS, CGC, Genetic Counselor, HudsonAlpha Institute for Biotechnology.

RARE University: Genetics Concepts
Online learning modules for rare disease patients and families.

Genetic Testing

Carrier Screening
A website and video from the Jewish Genetic Disease Consortium that explains who should consider screening, what screening is recommended and how it's done.

Child Neurology Foundation: Guide to Genetic Testing
Highlights include benefits of genetic testing, types of genetic tests, cost of genetic tests and the role of genetic counseling.

Family Planning
Summit session on family planning, technologies such as in-vitro fertilization with preimplantation genetic diagnosis, and other options for patients and carriers ready to start a family.

Find a Genetic Counselor
A search engine provided by the National Society of Genetic Counselors which provides both live and telehealth options.

Genetic Testing: Getting to a Diagnosis
Learn Genetics 101 and review the types of testing and results that can be expected.

How a Genetic Counselor Can Help You
Find questions to ask and tips for locating a genetic professional in your area— as well as how to arrange an appointment via telehealth.

How to Discuss Genetic Disease with Your Loved Ones
Factors to consider when deciding whether to tell others in your family about your genetic testing results and diagnosis.

National Coordinating Center for the Regional Genetics Networks
This organization works to improve access to quality genetic services for medically underserved populations.

Gene Therapy and Clinical Trials

A Guide to Gene Therapy
Gene Therapy 101 from Global Genes, including how it works and potential benefits.

American Society for Gene and Cell Therapy (ASGCT) Patient Education
Information on participating in gene therapy clinical trials.

Healing Genes
In-depth information on many different types of cell and gene medicine

Is Gene Therapy Right for Your Disease?
Global Genes' RARE Patient Advocacy Summit presentation on new developments in gene therapy technologies.

Mayo Clinic
Explains why gene therapy is done, as well as risks and potential benefits.

National Human Genome Research Institute Gene Therapy
Animated video showing how gene therapy works.

Platforms of Hope
A multimedia approach to ensuring this information is communicated in a digestible and shareable way.

Rare Webinar: Understanding Gene Therapy
Covers the basics of gene therapy and includes collaborations between companies and patient advocacy groups.

The Progress of Gene Therapy and Gene Editing
Global Genes summit presentation from September 2020.

Understanding Gene Therapy
Medline's guide to what gene therapy is, how it works, safety and ethical issues.

What is Gene Therapy?
FDA overview of the types of gene therapy.

Informed Consent

Courageous Parents Network
Questions to ask about a clinical trial as you navigate the informed consent process.

What is Informed Consent?
Elements of informed consent and information commonly included on forms.

Informed Consent FAQs
Basic elements of informed consent, what is and what is not allowed, and parental permission.

Informed Consent Important for Treatment Decisions in Advancing Reesarch
Global Genes' toolkit for patients and caregivers considering a clinical trial and advocacy leaders creating informed consent processes and documents.

Pediatric Decision-Making

AMA Pediatric Decision Making
Code of medical ethics on decisions for minors.

Health and Human Services Office for Human Research Protection
Regulations and permissions for involving children in research.

Informed Consent in Decision-Making in Pediatric Practice
American Academy of Pediatrics (AAP) policy on involving children and adolescents in medical decision making for research and clinical care.

Pediatric decision-making: Help parents protect, empower kids
Advice for physicians on facilitating decisions.

Shared Decision-Making

American Medical Association Code of Medical Ethics 1.13
A list of patients' rights that guide collaborations between physicians and patients on healthcare decision making.

Empowered Patient Coalition Decision Support Tool
An app that helps you identify areas where you may need help and support in making decisions.

National Institute for Health and Care Excellence (NICE) UK
Guidance on shared decision making and decision aids.

Ottawa Personal Decision Guide
A downloadable worksheet that helps you explore your knowledge, values, sources of support and your level of certainty about your decision.

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4. Health IT.gov National Learning Consortium. Shared Decision-Making Fact Sheet — December 2013. *https://www.healthit.gov/sites/default/files/nlc_shared_decision_making_fact_sheet.pdf*
5. International Society for Stem Cell Research. Patient Handbook on Stem Cell Therapies. Accessed 05/26/2021 *https://www.isscr.org/docs/default-source/patient-handbook/isscrpatienthandbook.pdf*
6. Joseph-Williams N, et al. Shared decision-making in rare disease in the United Kingdom. Prime Centre Wales Working Group White Paper *http://www.primecentre.wales/resources/210125_SDM%20in%20Rare%20Disease%20White%20Paper_V3_Final.pdf*
7. Perez-Ramos J, et al. Shared decision-making in rare diseases: An overview. Rare Dis Orphan Drugs 2015, 2 (3): 39-44. *https://rarejournal.org/index.php/rarejournal/article/view/83*
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9. Roth S. What is genomic medicine? J Med Libr Assoc. 2019 Jul: 107(3):442-448. *https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6579593/*

Global Genes resources to help you learn more about genetic topics

How a Genetic Counselor Can Help You
Connect to a genetic professional and get the most from working with one.

Genetic Testing: Getting to a Diagnosis
Learn Genetics 101 and review the types of testing and results that can be expected.

Diagnosis or No Diagnosis: What's Next After Genetic Testing
Finding people who understand, managing your care, and coping with emotions, whether you receive a diagnosis or are still undiagnosed and searching.

Data DIY
Educational program with training modules, workbooks, and live presentations that cover: the Why's and How's of Data Collection; Data Trusts and Governance; Developing Collaborative Research Networks; and Becoming a Data-Centric Community

A Guide to Gene Therapy
Gene Therapy 101 from Global Genes, including how it works and potential benefits.

Gene Therapy Clinical Trials FAQ
A list of answers to your questions about what makes gene therapy clinical trials unique.

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This online learning module from Global Genes is designed to give rare disease patients and families the background they need to understand how genes work.

Platforms of Hope
A multimedia approach to ensuring this information is communicated in a digestible and shareable way.

Wesley's Story

by Jamie Eckman

We were blessed with a beautiful 7 pound 20 inch boy on November 30, 2005 in Jacksonville, Florida. We named him Wesley Quinn Eckman. The only complaint this little newborn offered was reflux symptoms after meals and he would never take to the breast for feedings. Wesley would take the pumped breast milk from a bottle, but he would take longer than what we thought was normal to finish a feeding. We repeatedly brought this concern, along with reports of developmental delays, to our pediatrician's attention. The pediatrician's only response was that all babies develop at their own pace and he offered strengthening exercises to help meet infancy milestones.

Symptoms Worsen

Ironically, Wesley's symptoms worsened the week of his second set of immunizations. He now suffered from increased irritability, decreased interest in feedings, fever, and mouth ulcers. We were led to believe the immunizations held responsibility for these symptoms. Then, at 4 1/2 months of age, Wesley experienced a grand-mal seizure and was hospitalized for 21 days in All Children's Hospital in St. Petersburg, Florida. During his hospitalization, Wesley was treated for encephalitis with the probable cause being the herpes simplex virus (HSV). Although all the tests for infectious diseases came back negative, we were encouraged to treat him for HSV because of the stated symptoms. During our hospital stay, his symptoms appeared to decrease and no seizure activity was noted during multiple encephalograph (ECG) studies; however, repeated magnetic resonance imaging (MRI) tests revealed increased brain damage. We were given the explanation that the virus had attacked the neurovascular system, but we were told that with rigorous therapies, Wesley should be able to reach developmental milestones with only mild delays.

Searching for Answers

After a couple of months of treating Wesley with anti-seizure medication and physical therapy, we had hoped that he would return to the healthy, happy baby that he was before this nightmare began. But sadly, the seizure activity began to increase along with the feeding intolerance, so we sought a second opinion at Johns Hopkins Medical Center in Baltimore, Maryland. Without a definitive diagnosis from Johns Hopkins and his symptoms worsening daily, we took Wesley to Children's National Hospital in Washington, D.C. where we hoped to find the answers to our many questions as to why our baby's health continued to decline instead of improving.

A Devastating Diagnosis

Many more diagnostic tests were completed to reveal the dreadful diagnosis of Menkes disease when Wesley was 8 months old. We were told that Menkes is a terminal illness and palliative care was all that could be offered at this stage of the disease. We were instructed to go home with hospice care to manage his symptoms. We did go home and prayed that he would be the one baby to beat all the odds and be cured miraculously. His love for life was evident in his toothless grin that he offered every morning upon awakening. I truly believe he would have been happy all through the day had the side effects of the anti-seizure medications not been so severe. As the disease progressed, Wesley suffered to the bitter end. He had a story to tell and since he is no longer with us, we are gladly taking on the role of sharing his life story with all who are willing to listen.

Wesley fought this battle with such bravery that he will forever be our hero. The disease finally took over and Wesley earned his angel wings on November 15, 2006, two weeks shy of his one-year birthday.

Finding Support

We had a brief encounter with a genetic counselor by pure accident. My sister-in-law happened to share our story over a work meeting and one person in attendance was a genetic counselor from Johns Hopkins University Medical Center who had been in contact with another family whose son also shared the same Menkes diagnosis for 7 years.

She asked if it would be all right to share our email with this family so that a connection could be made. A few days later the emotional support we were desperately seeking came in a short and blunt email and 16 years later we have remained close with this family despite losing our sons to the disease.

Making a Difference for Others

We started the Menkes Foundation 6 months after our son's passing to serve as a liaison between families and the medical community. I feel our 11.5 month journey would have been a little less painful had we had a genetic counselor guiding us as we advocated for our son's short life.

Once the dust settled and we had time to comprehend the diagnosis he received only three months prior to his demise, we sought out genetic testing. Results came back that I was not a carrier of the disease and that this was a spontaneous mutation that happens very rarely in an already extremely rare disease. Fast forward eight years later I was asked to tour the exact lab that performed our genome sequencing while attending a fundraiser for Menkes disease by a family who also lost their son in Chicago, Illinois. This was a full circle moment in our lives.

Hope. It's in our genes.

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