Global Genes®
RARE Toolkits

AUGUST 2021

How a Genetic Counselor Can Help You

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

Since most rare diseases are caused by changes in your **genes**, **genetic testing** is often part of the journey. But understanding your options may be challenging. Partnering with a **genetic counselor** can help you make the best choice for you and your family.

What is a Genetic Counselor?

A genetic counselor is a healthcare professional with special training in counseling and medical genetics. They can help you understand the diagnostic process and can advocate for you to obtain emotional and financial support and get the best possible care.

WHAT GENETIC COUNSELORS ***DO***

Make complex genetic topics and tests easier to understand

Take time to explore your fears, questions, and concerns

Provide information about testing options

Identify the most cost-efficient approach in your situation

Help you navigate care, arrange referrals, and connect for support

WHAT GENETIC COUNSELORS ***DON'T DO***

Make decisions for you about testing or having children

Practice as licensed therapists or psychologists

Prescribe medications or diagnose your disease or condition

Replace other healthcare team members like physicians

Should I See a Genetic Counselor?

There are a variety of situations when genetic counseling may be useful, including the following:

Before or During Pregnancy

- You want to know if you are a carrier of a condition that could affect your children.
- You and/or your reproductive partner have a child or family member with a genetic condition and are considering pregnancy.
- Your age suggests a greater risk of having a child with a genetic condition.
- You have a history of miscarriages or stillbirths.
- Routine screening, such as an ultrasound, indicates an increased risk or concern.

At Birth or in the Neonatal Intensive Care Unit (NICU)

- Your baby has significant health concerns (i.e., low birth weight, failure to thrive) not related to preterm delivery.
- Your baby has abnormal external features (hands, feet, torso, etc.) that may suggest a genetic condition.
- Your baby has seizures or unusual eye, cardiac, or neurological findings.
- Genetic counselors may also be involved in communicating newborn screening results.

In Early Childhood and Beyond

- Symptoms continue or worsen, but doctors have been unable to provide a diagnosis.
- You see multiple specialists for various health issues (vascular, vision, hearing, etc.) and it's not clear why.

PRO TIP

"If things aren't going as expected, you should feel comfortable asking, 'Could we be missing something? Should we consider genetic testing?' The answer may be no. But I don't want you to be afraid to ask."

– ALLISON GOETSCH WEISMAN, GENETIC COUNSELOR, ANN & ROBERT H. LURIE CHILDREN'S HOSPITAL OF CHICAGO; ASSISTANT PROFESSOR OF PEDIATRICS, NORTHWESTERN UNIVERSITY FEINBERG SCHOOL OF MEDICINE

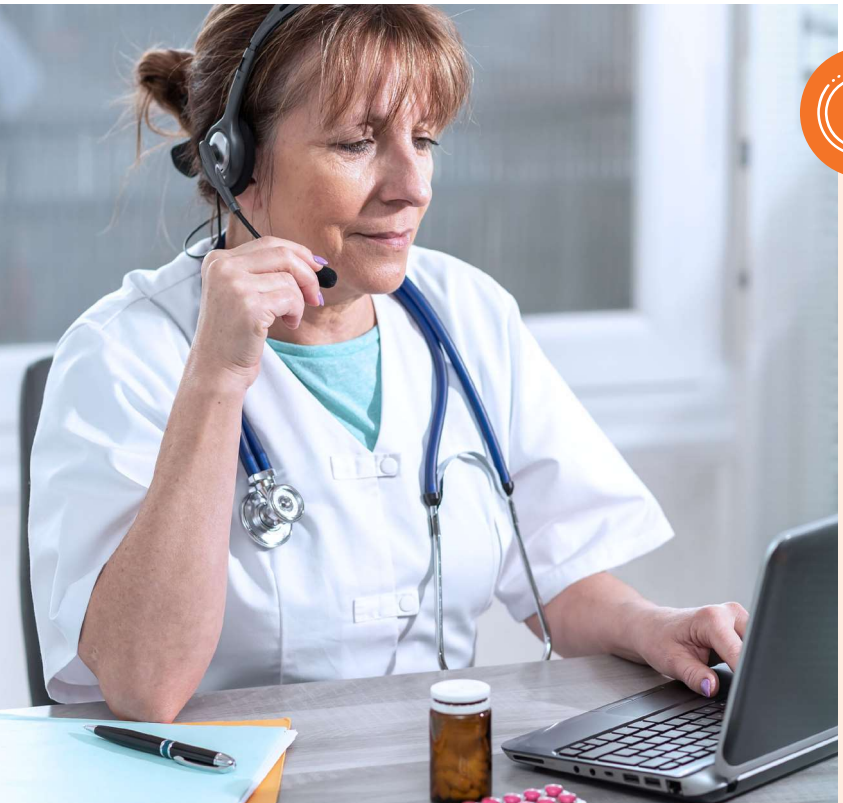
- Your child is not growing well or growing too much.
- Your child's development is delayed.
- Your child is regressing (losing physical or cognitive skills).
- You need a genetic test to confirm a clinical diagnosis so you can access treatment or enroll in a trial.
- You want to identify the exact genetic variant, so you'll know what symptoms to watch for and what treatment will be appropriate.

Connecting with a Genetic Counselor

Genetic counselors usually work on a team at a major medical center. Some work at clinics specializing in cardiology, neurology, neuromuscular diseases, cancer, and metabolic diseases.

If you receive care at a hospital that doesn't provide genetic services, you can ask your physician for a referral. If you live in the United States, you can also reach out directly to a genetic counselor for an appointment, either live or virtual. Ask the clinic and your specific insurance plan about expected costs related to a genetic counseling appointment.

If you live outside the United States, you may need to be referred for genetic testing by a physician. In the UK, for example, *https://www.nhs.uk/conditions/genetic-and-genomic-testing/*



PRO TIP

Some practices, like Grey Genetics, specialize in telehealth visits with genetic counselors. Telehealth is a good option for those who live in an area where there are no genetic counselors, or in a country outside of the United States.

Get in Touch with a Genetic Counselor

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.

Canadian Association of Genetic Counsellors
Provides a search tool to locate a genetics clinic in Canada.

Board of Genetic Counseling, India
Professional organization dedicated to education, training and practice.

British Society for Genetic Medicine
Information for public, patients and families, including organisations offering information and support

Find a Genetic Counselor Directory
This tool from the National Society of Genetic Counselors offers access to over 3,300 genetic counselors in the U.S. and Canada. You can filter searches for “live” or “telehealth.”

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

What to Expect

Genetic counseling is based on **shared decision making**. You can expect your genetic counselor to listen to your thoughts and feelings, help you think through your values, answer any questions you have, arm you with information and support you in making the best choice for you, your child or your family.

Preparing for Your Appointment

Anything that you can gather and send to your genetic counselor ahead of time will make your session more valuable and efficient.

- You may be asked to fill out a questionnaire before the meeting.
- You may receive a call for a pre-session "intake" where family

RARE TIP

"A genetic counselor should be helpful and friendly and make you feel good when you leave, even if the conversation is heavy. They should make you feel like they have your best interests at heart, but you need to be receptive to what they're saying. You can always leave or switch. There is no harm in admitting someone isn't meshing with you."

- MELISSA HAYDEN, RARE DISEASE COMMUNITY MEMBER

history and brief medical history will be collected.

- Ask your physician to send any relevant medical records, test results and family history information to the genetic counselor – or bring them to your first session.
- If you haven't already done so, this is a good time to ask your extended family about any medical conditions, birth defects or disabilities that have occurred in your family, or any genetic testing or results they can share.
- Contact your insurance company to find out if your genetic consultation will be covered, or if you will have to pay for all or some of the appointment. You may want to ask the genetics clinic whether and how your consultation will be billed to your insurance.

RARE TIP

"Do your best to collect your family's medical history. But remember some family members may not want to share intimate details and that is ok. "

- PARVATHY KRISHNAN, RARE DISEASE CAREGIVER, GLOBAL GENES FOUNDATION ALLIANCE MANAGER

Pre-Test Counseling

Genetic counseling often has two parts. The goal of the first part is to help you understand potential benefits, risks and limitations of various genetic tests, and the possible consequences of test results. This **informed consent** process will help you decide whether to pursue, delay or decline genetic testing.

At the first session, you can expect to:

- Share your family history information and/or any relevant test results. Genetic counselors are expert listeners who want to know the details of your story, which can sometimes point to a possible diagnosis.
- Have an opportunity to share your feelings, emotions, thoughts, and concerns. Genetic counselors understand the emotional issues that surround undiagnosed disorders, chronic disease, and the uncertainty that may accompany genetic testing.
- Get answers to your questions related to genetic conditions, testing, possible results, and financial impact. Don't be afraid to ask! If your genetic counselor doesn't know the answer, he or she can likely find it for you.

Pre-test counseling

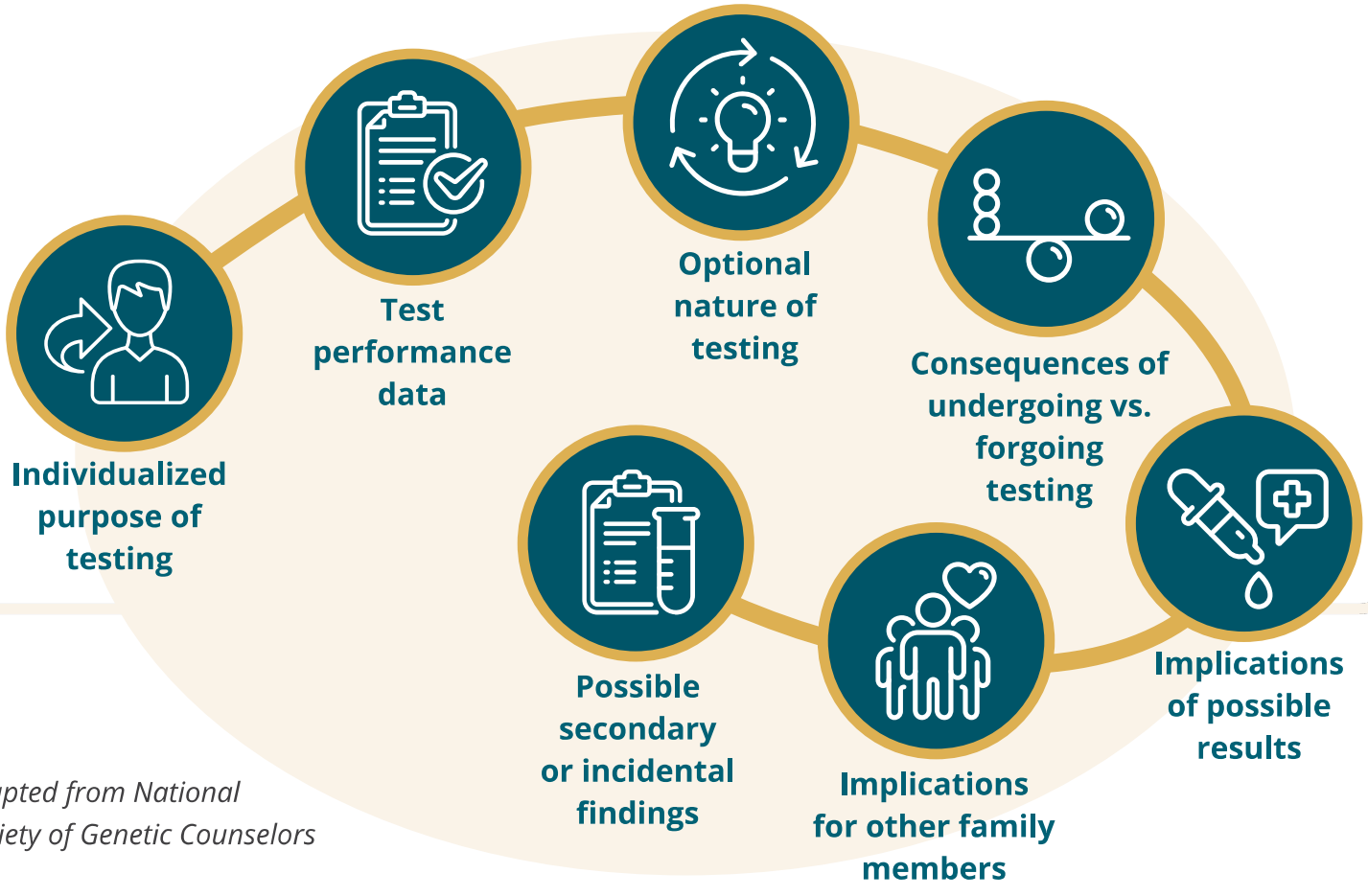
- Addressing questions
- Assessing personal and family history
- Culturally-sensitive
- Discussing benefits, risks and limitations of genetic testing
- Informed consent
- Sharing information
- Supporting informed choice regarding whether to have testing

Components of informed consent

- Consequences of undergoing vs. forgoing testing
- Implications for other family members
- Implications of possible results
- Individualized purpose of testing
- Optional nature of testing
- Possible secondary or incidental findings
- Test performance data

Post-test counseling

- Communication of the genetic test results
- Discussion of implications for the patient and family members
- Discussion regarding future health and family planning decisions
- Follow-up support and resource identification/ provision
- Medical management recommendations of individual/family



Adapted from National Society of Genetic Counselors

Post-Test Counseling

If you decide to have genetic testing, you will likely speak with your genetic counselor again when results are back. If the results of your test show a **genetic change**, the genetic counselor will explain what that means for you and your family. Most genetic changes are harmless and do not cause disease. However, sometimes a genetic change is found that indicates a clear genetic diagnosis.

At other times, a change is found that is not currently well understood. These genetic changes are often called **"variants of unknown significance."** This means that scientists at the testing lab cannot determine whether the change is harmless or disease causing. Genetic test results are not always clear-cut.

If a genetic test fails to make a diagnosis, keep in mind that does not rule out an underlying genetic diagnosis. There may be a genetic change present that could not be detected with current testing options.

Genetic Testing:
Getting to a Diagnosis
Learn more about testing in this *resource* from Global Genes.

Since genetic testing can be very complex, genetic counselors often provide written information for both families and their healthcare team, summarizing results and what they mean.

PRO TIP

"A genetic counselor gives you the time and space to digest what the results mean for you and your child. You can't advocate for your child or push forward on treatments and trials if you don't understand the result yourself."

– JERICA LENBERG,
RADY CHILDREN'S INSTITUTE
FOR GENOMIC MEDICINE

Diagnosis

Receiving a diagnosis for a genetic condition is beneficial for many different reasons.

- It may lead to effective treatment.
- It will help you avoid harmful or useless treatment.
- It provides you with a better idea of what to expect.
- It may open up possibilities for enrollment in clinical trials.
- It could answer questions of recurrence in families.

Ending the diagnostic journey provides you with facts. Some of them may be hard to bear. But having a name for the condition will also open opportunities to connect with researchers interested in the disease and link you to families who share your journey.

If genetic tests point to a diagnosis, your genetic counselor will likely:

- Explain the results and answer any questions you have
- Share information about the condition and what you can expect
- Provide you with a plan that outlines any recommended changes to care and/or other health issues that your child should be screened for now or in the future
- Let you know if the gene variant was **inherited** so you can tell family members who may want to seek testing
- Arrange any imaging or lab work for baseline assessments
- Provide referrals to specialists
- Recommend a timetable for follow-up visits
- Offer helpful resources, such as books, websites, patient-advocacy groups, clinical and research trials
- Support you through the emotional journey that accompanies a rare diagnosis

Some institutions may not have bandwidth to provide all of these services. If they don't, your pediatrician can help. Families with complex medical needs may also qualify for a care coordinator through their insurance company. Ask for what you need and follow-up if needed.

PRO TIP

"Just because you are told to come back only if you have a new concern doesn't prevent you from calling back and asking a question. It's ok to say, 'I know we talked about this in detail a month ago. But I was so overwhelmed I honestly don't remember a thing.' We are always available because even when the results are conclusive, they are not simple."

– ALLISON GOETSCH WEISMAN, GENETIC COUNSELOR, ANN & ROBERT H. LURIE CHILDREN'S HOSPITAL OF CHICAGO; ASSISTANT PROFESSOR OF PEDIATRICS, NORTHWESTERN UNIVERSITY FEINBERG SCHOOL OF MEDICINE

No Diagnosis

If you don't receive a diagnosis, it's not the end of the road. You may be referred for additional testing. But if you're not, it's still important to stay connected. Most genetic counselors recommend that you check in once a year.

Genetic testing technologies continually evolve and improve and so does our

knowledge of how to identify and treat rare conditions. One year from now, there may be new tests or there may be an opportunity to reanalyze previous tests.

If you have had **whole genome sequencing (WGS)** or **whole exome sequencing (WES)** your data can be reanalyzed. Most new diagnoses from reanalysis happen because of new conditions being described in the scientific literature. Therefore, most laboratories recommend waiting at least 6 months before requesting re-analysis.

Reanalysis may also be indicated if you develop a significant new symptom. For example, if someone was sequenced due to a heart problem but later develops seizures their doctor can request reanalysis of the genes associated with seizure conditions. Some laboratories perform their own periodic reanalyses. Laboratories will try to contact your medical provider for new diagnoses. It is recommended to keep your clinic updated on your contact information in case they receive a new result from reanalysis.

RARE TIP

"Our first and last meeting with our genetic counselor was to get my son's diagnosis. I had no idea we could have begun a recurring relationship with her. It might have been made perfectly clear at the time and I just couldn't hear that through the fog of a terrible diagnosis. I've since learned this can vary by hospital, state, or insurance plan. Some families get just 3 visits with a GC others can get many more."

- DANIEL DEFABIO, RARE CAREGIVER, CO-FOUNDER OF DISORDER: THE RARE DISEASE FILM FESTIVAL, GLOBAL GENES ASSOCIATE DIRECTOR, COMMUNITY ENGAGEMENT

You should also alert your genetics team if you have a new health issue, someone else in the family was diagnosed with a genetic condition, or there has been any other change from baseline.

PRO TIP

"Families don't always realize that we are only interpreting genes that could potentially be associated with the health issues reported. If you provide new information, it can change the way we analyze the data."

– ALLISON GOETSCH WEISMAN, GENETIC COUNSELOR, ANN & ROBERT H. LURIE CHILDREN'S HOSPITAL OF CHICAGO; ASSISTANT PROFESSOR OF PEDIATRICS, NORTHWESTERN UNIVERSITY FEINBERG SCHOOL OF MEDICINE

Insurance Eligibility

It's important to get the facts on what your insurance company will or won't cover related to genetic counseling and testing. Coverage often varies from one company to the next. Also, some insurance plans require referrals, while others do not.

Genetic counselors can play a significant role in helping your insurance company understand what is or is not being performed. It's not uncommon for genetic counselors to step in when genetic testing costs are being questioned or there is a need for a skilled expert to help the patient explain what he or she needs. Health systems in some countries do not provide coverage for genetic testing or genetic counseling, so patients and caregivers pay out of pocket. Crowdfunding is sometimes used to cover complex medical needs.

PRO TIP

"Ask the genetics clinic about expected costs. How they bill for genetic counseling can make a difference in whether or not it will be covered by insurance."

- KELLY EAST, LEAD GENETIC COUNSELOR, HUDSONALPHA INSTITUTE FOR BIOTECHNOLOGY

Insurance and Genetic Counseling

Learn more in a podcast entitled, "Demystifying Insurance," from the National Society of Genetic Counselors (NSGC)

RARE TIPS

"Even if you have been denied, don't give up. You can appeal with the insurance company or ask for advice from the genetic counseling or the testing company."

– NANCY KESSLER, BOARD MEMBER, SYNGAP RESEARCH FUND

"If you are denied coverage, you can also consider checking with your genetic counselor to see if there are research studies you can enroll in which include genetic testing."

- PARVATHY KRISHNAN, RARE DISEASE CAREGIVER, GLOBAL GENES

Questions to Ask Your Genetic Counselor

Sometimes it takes a conversation ahead of time to figure out whom you should see. If your child has a metabolic condition, for example, it's important that you go to a clinic with a metabolic care team (dieticians, metabolic geneticists, etc.) Consider asking:

Before you make an appointment

- Who provides the care here?
- Do you have geneticists?
- Do you have genetic counselors?
- Do you have advanced practice providers?
- What should I expect when I meet with the genetic counselor?
- How many appointments will I need?
- What should I send over before the appointment, or bring with me?
- Will my insurance pay for the counseling? Are there assistance programs?
- Is genetic testing part of the first session? If so, what will you sample?
- How will my privacy and the privacy of my family members be protected?

At Your First Session

- What type of genetic testing is available?
- What is involved in the process?
- How will I know if I need to have genetic testing done?
- What are the benefits and limitations of these tests?
- What information will the tests provide and how will I receive the results?
- How will this information help us?
- We have had genetic tests in the past that didn't provide answers. What will be different now?
- What is the chance that testing will provide a diagnosis?
- Could I be exposing myself of my family to discrimination based on genetic information?
- How will I pay for testing?

After testing

- What does this result mean for my family?
- Does this result mean I'm definitely going to get sick?
- Is there a treatment for this?
- You found this genetic change. Is there a way to correct it?
- Could I pass this on?
- What are the chances that my children, my siblings, my parents are affected by this?
- What is happening in the world of clinical trials around this?
- Do you know of other families or organizations you could connect me with?
- Who should I share my results with?
- I didn't receive a diagnosis. What's next?

Glossary

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

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 counseling Genetic counseling gives you information about how a genetic condition might affect you or your family. *https://www.cdc.gov/genomics/gtesting/genetic_counseling.htm*

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 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/*

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*

Informed consent The process of educating a person about genetic tests and obtaining permission to carry out testing. "Informed" means that the person has enough information to make an educated decision about testing; "consent" refers to a person's voluntary agreement to have the test done. *https://medlineplus.gov/genetics/understanding/testing/informedconsent/*

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*

Neonatal intensive care unit (NICU) Newborn babies who need intensive medical care are often put in a special area of the hospital called the neonatal intensive care unit (NICU). The NICU has advanced technology and trained healthcare professionals to give special care for the tiniest patients. *https://www.urmc.rochester.edu/encyclopedia/content.aspx?contenttypeid=90&contentid=P02389*

Pediatric intensive care unit (PICU)
The pediatric intensive care unit (PICU) is a specialized unit of the hospital where the sickest pediatric patients are admitted. *https://medicine.iu.edu/blogs/pediatrics/what-is-the-pediatric-intensive-care-unit-an-introduction*

Shared decision making Shared decision making is a model of patient-centered care that enables and encourages people to play a role in the medical decisions that affect their health. *https://www.ahrq.gov/cahps/quality-improvement/improvement-guide/6-strategies-for-improving/communication/strategy6i-shared-decisionmaking.html*

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.

Centers for Disease Control and Prevention
Explains common reasons that people seek genetic counseling

Genetic Alliance
Guide to prenatal, general, pediatric, cancer, and psychiatric genetic counseling.

HealthyChildren.org
Explains the role of a pediatric geneticist

Medline
Explains what a genetic consultation is, why someone might seek genetic counseling and what to expect during the consult.
Consultation
Reasons
Expectations

National Human Genome Research Institute
FAQs on genetic counseling

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.

Prenatal Genetic Testing

Kids Health: Prenatal Genetic Counseling
Describes situations when doctors may recommend testing during pregnancy and what the process will be like.

March of Dimes
Key facts on prenatal visits to a genetic counselor.

How to Locate a Genetics Professional

American Board of Genetic Counselors
A directory of genetic counselors by practice area.

American College of Medical Genetics
Database for individuals who wish to locate a U. S. genetics clinic.

American Society of Human Genetics (ASHG)
Database of members (researchers and clinical geneticists), some of whom live outside of the US.

Australasian Society for Genetic Counsellors
Represents over 280 members across Australia and New Zealand.

Canadian Association of Genetic Counsellors
Provides a search tool to locate a genetics clinic in Canada.

Find a Genetic Counselor
This tool from the National Society of Genetic Counselors offers access to more than 3,300 genetic counselors in the U.S. and Canada and has a way to filter searches by "live" and "telehealth."

RARE Concierge
Global Genes offers a free service for people seeking a rare disease diagnosis and those who have questions about a diagnosis they have already received. Reach out through our Rare Concierge portal to be connected to resources, support and advice on accessing care.

Webinars

Rare Disease: What Role Do Genetics Play
Webinar presented by Kelly East, MS, CGC, on the role that genetic testing and genetic counselors play in rare disease. Resources for individuals impacted by rare disease living with or without a diagnosis are included throughout the presentation.

Video

Beyond Diagnosis: Working with a Genetic Counselor During Your Rare Journey
Genetic counselor Allison Goetsch Weisman, winner of the Code Talker Award, and Melissa Hayden tell how their partnership led to a long-awaited diagnosis.

A Patient's Story

by Melissa Hayden, Rare Patient

"Twenty-four years, hundreds of doctors, and countless genetic tests later and I finally, with the help of my dedicated genetic counselor Allison, found an answer. No longer am I a patient with an "undiagnosed genetic condition," and Allison was a driving force in this becoming a reality.

I was born prematurely with many other obvious signs that something wasn't right. Since then, I have had more than 10 brain surgeries, three spinal fusions, hundreds of days in the hospital, and more doctor appointments than I can count. I have a sister who passed away at age 5 from an aneurysm. I knew I had a genetic condition, but I never knew what type.

After years of testing and always hearing results of "inconclusive," I became weary of the process. I resigned myself to never knowing my condition. Luckily, I was blessed to be assigned to Allison, my genetic counselor. She was always so positive and encouraging, but honest too. She brought new suggestions, new test options, and more information. All of this is key to any genetic counselor's role.

RARE TIP

"I have come to find there is a special bond between genetic counselors and their patients — one unlike any other that I've had in the hospital."

- MELISSA HAYDEN, RARE PATIENT

Allison cared about my well-being as a whole person — not just the person she knew from my medical records and test results. She took the time to ask about my life outside of the hospital. She constantly reminded me that eventually we would find an answer and that she was working tirelessly to help me do that.

As a genetic counselor, she had to wear so many hats: intelligent genetics department staff with facts to deliver, translator to repeat those facts in

English, caring psychologist to help me cope with the lack of a diagnosis, and supportive friend. I knew how much she cared about me and how hard she was working to help find a diagnosis. I will never forget the day when she finally got to tell me something different.

RARE TIP

"I will never forget the day when Allison finally got to tell me something different: "I almost dropped the files when I got them back from the lab. We have a diagnosis!" she said. She was as excited as I was, and she shared in my joy."

- MELISSA HAYDEN, RARE PATIENT

She also brought every piece of medical journal literature about the syndrome to show us. She gave us every detail on the condition and information about the other nine patients in the world that have this condition. She answered every question we had to the very best of her ability.

Without Allison, I wouldn't have a diagnosis. Without Allison, I wouldn't be able to say that my medical history will be published for others to learn about the syndrome, which will hopefully save many lives in the future.

Hope. It's in our genes.

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