



A Parent's Guide to Pediatric Genetic Testing



Children's Hospital Colorado
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INTRODUCTION

Navigating Pediatric Genetic Testing

If your child has been referred for genetic testing, you probably have a lot of questions. Our goal is to guide and support you through the entire process.

When you meet with our genetics team, they will ask you a lot of questions about your child's health and family history. You will also have to make decisions about the next steps in their care. This e-book will help you understand how to prepare for this visit. We want you to feel informed, assured and empowered to make the right choices for your child and family.

Understanding Genetic Testing



As you start this journey, it's important to understand what genetic testing is and what it is not. Our genetics team will walk you through the information we can get from genetic testing and its limitations.



What genetic testing is

Genetic testing is a medical tool that looks for changes in your child's DNA. DNA is a molecule in our cells. It contains instructions for how to build the proteins that make our bodies work. It's like a guide for how a child's body grows and functions.

When your child gets genetic testing, we take a sample of their cells and separate out the DNA using special machines. We usually take a swab of the inside of their cheek, but sometimes we need other samples, such as blood. We can then look closely at the DNA to check for anything that is:

- Added
- Changed
- Missing
- In a different order than expected








What genetic testing is not

Genetic testing is a valuable tool that helps your care team make important decisions about diagnoses and treatments. But it does not always give concrete answers. Genetic testing is:

- **Not a genealogy test:** Many of the genetic tests you can buy and use at home are designed to track your family's ancestry. These tests are different from clinical genetic tests because they are not designed for medical decision-making. They can also give incomplete or misleading results.
- **Not a paternity test:** Sometimes we recommend that parents and family members get genetic testing so we can gather more information. These tests give us biological information, but they are not the same as a paternity test.
- **Not a publicly shared record:** Any medical information we get through genetic testing is protected by the Health Insurance Portability and Accountability Act (HIPAA). That means we do not give your child's test results to anyone outside our health system without your consent.

Genetic testing myths vs. facts

	MYTH	FACT
	My child has to have genetic testing because their doctor suggested it.	Genetic testing is always optional. Our genetics team helps you decide how to move forward based on what's best for your family and will let you know if they think the test is necessary.
	Genetic testing labels my child.	Genetic testing is a tool that helps us understand your child's health and development. It does not define your child or limit their potential. Who you share your genetic information with is a personal decision.
	If there's no family history of a condition, there's no point to genetic testing.	Genetic conditions develop because of a change in your child's DNA. Many of these DNA changes are not passed down and can happen in children without a family history.
	Genetic testing is the same as an at-home DNA test.	Our genetic testing happens in certified medical labs. Our specialists read the results and help you make medical decisions based on the information. This is not true for home tests, which are designed primarily for entertainment.
	Genetic testing always leads to a treatment or cure.	In some cases, genetic testing can help guide us to the right treatment for your child. But it does not always change your child's day-to-day care, treatment plan or resources.

What's the difference between genetic and inherited conditions?

Genetic conditions occur because of a change in your child's DNA. The difference between inherited conditions and other types of genetic conditions is in how that change came to be:

- Inherited conditions are caused by a genetic change that was passed down from parent to child.
- Other genetic conditions are caused by a new genetic change in a child's DNA that does not run in the family.

Genetic testing can help us find the cause of both inherited and noninherited genetic conditions. Family history is an important part of genetic testing, but it does not tell us the whole story.

What to Know About Referrals



It's helpful to understand why and when doctors may suggest genetic testing.



Why do doctors refer children for genetic testing?

When your care team refers your child for genetic testing, they are looking for more information about a possible or confirmed diagnosis. A referral does not always mean that your child has a specific condition. And it does not always mean that something is “wrong.” Often, genetic testing referrals are just a part of a longer diagnostic and treatment journey.



Common reasons for referrals to genetic testing

Common reasons doctors refer children for genetic testing include signs of:

- **Birth differences**, such as congenital heart problems or cleft lip/palate
- **Developmental delays** or autism spectrum disorders
- **Family history** of a known or suspected genetic condition
- **Growth differences**, such as short stature, poor weight gain or overgrowth
- **Seizures**, epilepsy, movement disorders and other neurological concerns
- **Vision or hearing** differences

The role of genetic testing

Genetic testing can provide helpful information even if it doesn't change your child's day-to-day care. Your child may already be on the right treatment plan for their diagnosis, or there may not be any new treatments available. Additionally, the results may point your care team toward new treatments, such as molecular therapies, developing a new drug, repurposing an existing drug and other treatments that fall under the precision medicine umbrella. Genetic testing can help you and your child's care team answer questions such as:

- Are there specific specialists, screenings or other resources that could be helpful now or in the future?
- Could this condition cause future health effects?
- Does the condition affect other family members?
- Is it possible for this condition to affect your future children or your child's future family?
- Is there a known underlying reason for your child's symptoms?



Genetic testing for developmental delays

Signs of developmental delays are one of the most common reasons doctors refer children for genetic testing because genetic changes can play a role in developmental delays.

Genetic testing does not always find a reason for your child's symptoms. But when it does, it may lead to:

- Access to new treatments through clinical trials
- Help with insurance approval for certain treatments
- More effective care planning for your child

CHAPTER 3

Benefits of Genetic Testing



Each child is unique, and you know your family's situation best. One of the roles of a genetic counselor is to help you understand the possible benefits of genetic testing. Knowing these benefits helps you decide whether testing is right for your child.



Potential benefits of getting a diagnosis through genetic testing

Sometimes, genetic testing finds or confirms a specific diagnosis. In these cases, genetic testing may help:

- Avoid tests that are unnecessary or repetitive
- Clarify your child's prognosis (long-term outcomes) or help you plan for possible future health effects
- Guide your care team's plan for managing and monitoring a condition, along with referrals to other specialists
- Identify opportunities to participate in clinical trials or research studies (if your family wants to)
- Inform family planning and clarify any risks to your child's relatives or future family.
- Make your child eligible for various medical and community resources
- Support access to certain therapies, insurance coverage or other services



Potential benefits when testing does not lead to a diagnosis

Even when genetic testing does not confirm a specific diagnosis, it can still offer benefits. Testing may help:

- Guide healthcare decisions in the future
- Inform further testing as science advances
- Provide reassurance
- Rule out certain genetic conditions



Connection and community: A major benefit of genetic testing

Many families find comfort in confirming a diagnosis even if their child's care plan doesn't change. Often, getting a diagnosis allows you and your child to find helpful community support.

Connecting with other families who have children with the same condition can be powerful. We often hear families say there's a deep sense of relief in "finding your people." Building this community may look like:

- Attending conferences, events or webinars
- Getting involved with advocacy groups or national foundations
- Joining online forums or discussion boards
- Participating in support groups
- Sharing your story on social media to raise awareness



How do I know if genetic testing is right for my child?

Genetic testing is always optional. When you meet with a member of our genetics team, they can help you decide whether or not to move forward with testing.

You may make this decision based on several factors, including:

- **Effects of test results:** Even if testing doesn't change a diagnosis, it may still inform your child's care plan or help you understand family risks. Testing can also help you qualify for insurance coverage for certain treatments. Our counselors will explain the possible impacts of test results so you can decide what's right for your child.
- **Family comfort and goals:** Some families want as much information as possible. Others may feel overwhelmed by a lot of information. Your preferences and comfort level can help you decide whether genetic testing is the right choice. You are always free to change your mind at any time.
- **Your child's diagnosis:** Some genetic conditions are easily diagnosed and managed. Your genetic medical provider/counselor can help you understand whether genetic testing is likely to offer useful insights.

CHAPTER 4

Preparing for Your Visit



Most families don't need to do a lot to prepare for their child's visit. But knowing what to expect and the types of conversations you might have can be helpful. You're always welcome to reach out to our team if you have questions before your child's appointment.



What you can do before your child's genetic testing visit

There are a few things you can do to make your child's appointment go smoothly.

Write down your child's medical history

Before your visit, it may be helpful to note information about:

- **Your child's current diagnosis:** Your genetics team usually starts the visit by asking what brought you in for care. They may ask about any testing or treatments your child has had in the past.
- **Your child's health:** Your genetics team will ask what symptoms your child has. They may also want to know about developmental milestones, past medical conditions, surgeries or hospitalizations.
- **Pregnancy and birth:** The genetics team may ask about your child's development before birth. They will also want to know about their infancy, including their gestational age at birth, their birth weight and whether they spent time in the neonatal intensive care unit (NICU).



If your child or an immediate family member has already had genetic testing, it's helpful to bring a copy of the report. Our genetics team members are experts in reading these reports.

Gather family history information

Our genetics team members typically ask about family history across 3 generations:

- Grandparents
- Parents
- Siblings

Knowing whether grandparents, parents or siblings share symptoms with your child can help us narrow down which genetic changes to look for. Gathering family history from 3 generations also helps us:

- Choose the correct test
- Interpret test results accurately
- Understand risks for other family members

In some cases, the genetics team may ask about first cousins or other extended family members. It's OK if you don't know the answers to all the questions. Whatever you are able to share is helpful.

Fill out questionnaires and provide medical records

Our clinic typically sends a questionnaire to complete before the appointment. It will have questions about your child's health and history. We provide this document in both English and Spanish.

If your child is a current patient at Children's Hospital Colorado, we will have access to their medical records. If your child is a patient at another health system, we may ask you to send us their medical records. Our team will let you know what records we need, including genetic testing reports, and how to send them.

Common questions genetic counselors may ask

MEDICAL HISTORY	FAMILY HISTORY
Were there any complications before or during your child's birth?	Do you have other children? Are there any known concerns about their health or development?
Did your child pass their newborn screenings?	Do the child's parents have known health or developmental concerns?
Did your child spend time in the neonatal intensive care unit (NICU)?	Do any of the child's grandparents have known health or developmental concerns?
What signs or symptoms have led to your child's current or suspected diagnosis?	Do any of the child's first cousins have known health or developmental concerns?
Has your child had any major surgeries or hospitalizations?	Do any relatives have similar symptoms to your child's?



What to expect at a pediatric genetic testing visit

Your child's first visit with our genetics team is mostly a discussion about your child's history, family history and genetic testing options. Usually, you meet with a genetic counselor, a medical geneticist or a nurse practitioner specializing in genetics. You discuss:

- Any questions you have
- Benefits and limitations of genetic testing
- Genetic testing options and possible results
- Previous test results if your child has had prior testing
- Your child's medical and family history
- Your goals for the appointment

Your child may also get a physical exam, depending on the reason for their visit. The first visit usually lasts between 45 and 60 minutes. You should plan to be at the clinic for about 90 minutes to account for check-in time.



Your child's genetics team will make space for you to share how you feel about the possible test results. If you are not ready to receive uncertain or unexpected information, you may decide testing is not the right choice at this time. That's OK — we want you to feel empowered to make the best decisions for your family.



What to expect after your child's genetic testing visit

At the end of the visit, the genetics team confirms whether you want to move forward with genetic testing. If you do, they will explain the next steps. These steps typically include:

- 1. Prior authorization:** Before we can continue, we must have prior authorization from your insurance provider. This process often takes 4 to 6 weeks. Our financial counselors will help you understand the costs involved and what insurance may or may not cover.
- 2. Sample collection:** Most likely, we will mail you a DNA sample collection kit. We provide instructions for collecting this simple cheek swab. Some patients may need to schedule an appointment to give a blood sample, but this is rare. We may also request samples from both parents, which can help us better understand your child's test results.
- 3. Sample processing:** You will mail the samples back to us. We will then process them in a clinical laboratory. Processing the results can take about 6 to 8 weeks.
- 4. Follow-up:** A genetic provider will reach out to review the results of your child's genetic test with you. This follow-up conversation may be over the phone or at another visit. We will discuss what the results mean and recommend next steps for your child.

The genetic testing process at a glance



STEP 1 Referral

Your child's primary or specialty care doctor refers them to our genetic testing team.



STEP 2 Clinic visit

You meet with the genetics team to discuss testing options, benefits and potential concerns. You decide whether to move forward with testing.



STEP 3: Insurance authorization

Our testing requires prior authorization from insurance. This process can take a few weeks or a few months, depending on the insurance provider and your plan.



STEP 4: Financial counseling

Financial counselors help you understand insurance coverage and out-of-pocket costs. We help you access financial support when needed.



STEP 5: Sample collection

Most samples are collected with an at-home cheek swab kit. We mail the swab kit to you with instructions to complete the test. You mail it back when the test is complete.



STEP 6: Follow-up

The genetics team reviews the test results with you over the phone or in a follow-up visit. We also share a report with your child's primary care or referring doctor so they can make any additional referrals.

CHAPTER 5

What to Expect From Test Results



After your child completes genetic testing, we upload the test report to their medical record. There are many types of genetic tests, and each can lead to different information and outcomes.

Genetic test reports can be very hard to interpret without help. Our genetics team will guide you through the results and what they mean.

Types of genetic testing results

During your first appointment, the genetic counselor explains what a genetic test may or may not find. Genetic test results can be:

- **Positive:** The test may find an explanation for your child's symptoms, leading to a clear diagnosis. This information can help guide your child's care, condition monitoring and access to resources.
- **Negative:** The test may not find a genetic cause for your child's symptoms. In this case, we may recommend follow-up testing.
- **Uncertain:** In some cases, the test may find rare or unique changes in your child's genes that may or may not be responsible for causing their condition. These results are known as "variants of unknown significance."
- **Unexpected:** Genetic testing can sometimes reveal information that is unrelated to the reason for testing. That might include genetic changes that are likely to cause a different disease in your child's lifetime or information about biological relationships.

It's important to understand that genetic test results are not always clear. Right now, our ability to test DNA is more advanced than our ability to interpret every result. But testing can offer benefits even if the results are uncertain.

Follow-up care after genetic testing

Your child's follow-up care depends on the test results. In general, we may recommend:

- Connecting with your child's doctor about care options such as therapies, treatments or school services, such as individualized education plans (IEPs)
- Getting follow-up testing to help us learn more
- Meeting with specialists for additional support
- Planning for additional tests

CONCLUSION

Ongoing Support for Your Child



After testing, we want to make sure you have access to all the resources you need. Our genetics team works alongside social workers, family navigators, financial counselors and other specialists dedicated to supporting your family.

These team members help you navigate every aspect of your child's journey, including:

- Access to therapies or medical equipment
- Costs and payments
- Insurance claims
- School accommodations

Our genetics team members are always available for further questions and ongoing support. Call **720-777-0020** to learn more about genetic testing and counseling at Children's Hospital Colorado.



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