- ✓ **Know your rights**
- ✓ **Speak up for your family**

Parent Toolkit

How to ask your doctor about genetic testing

Start Genetic

What is genetic testing?

→ **Each baby born has a unique genetic code.**

This code is called our DNA, and it is like a personal instruction manual guiding a person's growth, development, and body functions.

→ **Harmful changes can happen to the DNA.**

Each person's DNA code is made up of 3 billion letters. Sometimes, changes in a person's code—like missing words, or misspellings—cause problems.

→ **GENETIC TESTING is a spell-checker for DNA.**

Genetic testing checks a person's DNA for changes that cause problems. Several types of genetic tests check in various ways for different kinds of changes.

PARENT HACKS

✓ It's important to know:

A child can have a genetic condition *even* if it does NOT "run in the family".

- Some genetic conditions are "inherited" from parents who have the same condition, or whose relatives have it.
- But many genetic conditions are caused by ***new*** changes that happen ***only*** in an individual person's DNA.
- So, if your child has certain kinds of symptoms, **it is always worth doing genetic testing, *even if no one else in the family has the same symptoms.***

Start Genetic

How to ask about genetic testing

*You have the right to find out about accessing **reliable genetic testing**.*

→ **Have an open conversation with your doctor.**

They can assess your child's specific situation and determine if genetic testing could be helpful.

→ **Consider talking to a genetic counselor.**

These experts in the field of genetics can also discuss testing options and help you make decisions to meet your family's needs.

→ **Speak up for your child and your family.**

There are programs that can help you find genetic counselors and testing options if you need additional guidance.

PARENT HACKS

✓ If a doctor says a genetic diagnosis won't make a difference for treatment, point out these potential benefits:

- ➢ **monitoring for future concerns**
- ➢ **genetically-tailored treatments***
- ➢ **recommendations for other therapies**
- ➢ **medications to try or to avoid**

✓ Some doctors are unaware of genetic testing guidelines. Direct them to the "for healthcare providers" section of StartGenetic.org.

***in some cases**

Starting the conversation

→ **Consider asking:**

“My child has [autism, congenital anomalies, global developmental delay, epilepsy, CP, or other condition], and I understand there are professional guidelines for genetic testing for people with my child’s condition. How can we get genetic testing done?”

→ **Alternatively, try asking:**

“I don’t know if there are professional guidelines for genetic testing for people with my child’s condition. Can you tell me more about genetic testing?”

PARENT HACKS

✓ If the doctor is reluctant to order genetic testing, ask them to review these professional guidelines and explain them to you:

- **Autism/ID/Congenital Anomalies**
- **Epilepsy**
- **Cerebral Palsy**
- **Other conditions**

✓ Your child’s pediatrician or specialists usually CAN order the test, so you generally do NOT need a referral to a genetics specialist to receive genetic testing.

Ask about ALL genetic tests

→ **Even if you already did genetic testing, *ask again*.**

There are many different types of genetic tests.

*If previous tests did not find a genetic condition, **your child still might have a discoverable genetic condition**.*

Newer tests can often detect genetic changes that older or less precise tests cannot, but sometimes insurance companies or Medicaid require trying the older tests first.

→ **Consider asking:**

“Have we done ALL the recommended or available genetic tests for a person with my child’s symptoms? Is a sequencing test an option?”

PARENT HACKS

✓ If the *name* of your past genetic testing does NOT include the word **“sequencing”**, ask your doctor about **“exome” and “genome” sequencing tests.**

✓ Doctors can find detailed information on available genetic tests in the “for healthcare providers” section of StartGenetic.org.

Paying for genetic testing

→ **Genetic testing is often covered by insurance or Medicaid.**

- "Prior authorization" is usually required because they want to understand how the testing will help.
- Your doctor's office will work with the genetic testing lab to determine if testing will be covered.

→ **If insurance denies the genetic testing, your doctor can help.**

- Your doctor can request a "peer-to-peer" review to discuss medical necessity.
- Your doctor can also *appeal* an insurance denial.

PARENT HACKS

✓ ***Do not do any testing*** *until insurance has confirmed it will cover it*: make sure that prior authorization is obtained before you do any genetic testing.

✓ If the doctor is unsure about the insurance process, tell them *the genetic testing lab may provide guidance*, and direct them to the "for healthcare providers" section of StartGenetic.org.

✓ Tests offered online by companies that do not work with your doctor or with your insurance are not recommended.

Start Genetic

Know your rights

→ It is your **RIGHT** to understand your child's health information, including **genetic conditions**.

→ In the United States, the **Genetic Information Nondiscrimination Act (GINA)** protects you against discrimination based on your genetic information when it comes to **health insurance and employment**.*

PARENT HACKS

✓You can ***always*** ask for a **second opinion**.

✓You have the right to **speak up for your child & your family**

*** GINA *might* not apply to:**

- health insurance plans with fewer than 15 employees
- life insurance
- disability insurance
- long-term care insurance

You've got this!

Start Genetic and make an appointment to talk to your doctor about genetic testing today!

If you have more questions about genetic testing, email **takeaction@startgenetic.org**

Selected Resources

to Share with Your Doctor

Please direct your doctors to StartGenetic.org for more comprehensive resources for healthcare providers.

Does my patient need genetic testing?

Indications for genetic testing according to the ACMG:

- Developmental delay[1,2]
- Developmental regression[5]
- Intellectual disability[1,2]
- Autism[3]
- Growth differences, such as poor growth, or overgrowth
- Birth defects[1,2]
- Congenital hearing loss
- Cerebral palsy[6]

There are many reasons that genetic testing should be considered.

For pediatric patients having neurodevelopmental features, there are American College of Medical Genetics and Genomics (ACMG) genetic testing practice guidelines.

While ACMG does not have guidelines for patients with epilepsy alone, professional recommendations curated by the National Society of Genetic Counselors (NSGC) for **unexplained epilepsy** were published in 2022.[4]

It is important to note to patients and their families that a genetic diagnosis explains the genetic etiology of a patient's medical features but does not establish clinical diagnoses such as autism, which relies on a set of behavioral characteristics to have a diagnosis.

Does my patient need genetic testing?

References

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Start Genetic

Resources for Healthcare Professionals
more comprehensive list available at
StartGenetic.org

Who can provide additional genetic testing guidance?

Below is a list of companies you might consider. Genetic testing labs may also provide genetic counseling services. Start Genetic does not endorse any companies that provide telemedicine genetic counseling.

Location of Licenses to Practice	Company Name / Website / Services
Rhode Island, Maryland, Pennsylvania, California	**CeGaT**
All 50 US states	**InformedDNA** (Can do pre- and post-test counseling but testing needs to be completed through their company)
All 50 US states	**GreyGenetics** (Pre- and post-test counseling offered for any genetic testing, and offer genetics training for healthcare providers)
All 50 US states	**Ambry Genetics** (Post-test counseling for testing completed through Ambry)
All 50 US states	**Genetic Support Foundation** (Post-test counseling)
All 50 US states and Canada	**Genome Medical** (Pre- and post-test counseling, help with ordering)