

FAMILY CENTER WORKGROUP **ON INCLUSION**

TIPS AND HIGHLIGHTS FOR FAMILIES



ENTERING THE WORLD OF PEDIATRIC GENETICS

INFORMATION TO CONSIDER FOR YOUR FIRST APPOINTMENT

Families may be asked to see a genetic professional for a variety of reasons. Genetics professionals are healthcare professionals with specialized degrees and experience in medical genetics and counseling. Genetics professionals include geneticists, genetic counselors, and genetics nurses. Trained in genetics. If you are a family in this situation, it is understandable that you may have many questions. We hope this information is helpful for you and your family to understand why an appointment to see a genetic professional may be needed and what you may expect.

WHY DO WE NEED TO SEE A GENETIC PROFESSIONAL

It is not uncommon for a pediatrician or other family doctor to suggest a family make an appointment with a genetics clinic. Referrals to a genetics professional can happen throughout your lifetime, including during pregnancy, after the birth of a child, during childhood, or as an adult. There are many reasons you might see a genetics professional, such as:

- ✓ You are pregnant or considering becoming pregnant and are concerned about the health of your baby.
- ✓ Your baby had an out-of-range result from newborn screening.
- ✓ You or your partner have a genetic condition that runs in your family.
- ✓ You are concerned about your child's health, growth, or development. Find a list of Red Flags for Genetics.



Here

- ✓ You, your child, or a family member has been diagnosed with a genetic condition.
- ✓ You are concerned that you, your child, or a family member has a genetic or inherited condition and would like more information.

WHO WILL BE **THERE?**

Usually there is a team working together to support families in caring for their child. A genetics clinic appointment might include meeting with several people including the geneticist, a genetic counselor, a genetics nurse, and perhaps a social worker. A clinical geneticist is a specialist who has received specialized advanced training and experience to give children the unique medical care they need. Genetic counselors share education, support, and resources to help families understand how your child's condition might affect them or your family. A genetics nurse is a licensed professional nurse with special education and training in genetics. A social worker can provide similar resources along with other community and government agency information.

WHAT WILL HAPPEN AT THE APPOINTMENT?

A genetic appointment is a conversation. You will be able to ask questions during the visit. It can last an hour or longer, especially if it's your first visit and depending on the information covered. This is a time for the genetics team to get to know your child and your family and talk with you about your family's medical and health history. Depending on the reason for the visit, some key things may happen during your visit:

- ✓ If the patient, couple, or family was referred to the genetics clinic, the genetic professional will review the reason for referral.
- ✓ The team will take a family health history of at least three generations, documenting all genetic conditions or health problems in each family member.
- ✓ A physician may provide a full physical examination and decide on laboratory tests to rule out or diagnose a genetic condition or refer you to other medical specialists. They may ask permission to take photographs to assist in diagnosis.
- ✓ The genetic counselor can provide supportive counseling and information about resources and support networks to connect with other families.
- ✓ The genetic professional will explain the diagnosis and any issues about the condition, including how the condition is expected to progress, the management of the condition, treatment options, whether genetic testing is available, and the chances of the condition being present in future pregnancies.
- ✓ The genetics professional will also discuss how the condition may or may not affect other family members and will tell you if testing is suggested for other family members.

This may be done all in one visit or over the course of numerous sessions. The healthcare team may ask you to come back for follow-up appointments.

TELEHEALTH

A growing number of medical providers are using technology to increase access to health care services. One of the ways to reach patients is with telehealth.

The Midwest Genetics Network created videos specifically for patients and their families to increase their awareness of the benefits of seeing a health care provider using telemedicine. These videos will introduce you to telemedicine, describe what to expect, and help you get the most out of your telemedicine appointment.

 <https://midwestgenetics.org/resources/telemedicine-resources/>


WHAT DO WE DO BEFORE **WE GO?**

You will likely get instructions from the genetics clinic on how to prepare for the appointment. Ask how long this first appointment might take so you can be prepared. Typical instructions before the visit might include:

- ✔ **Contact your insurance company** to find out if the genetic appointment and genetic testing will be covered or if you may need to pay for some or all the appointment. You can find resources regarding insurance coverage.

 [Here](#)

- ✔ **Find out** as much as you can about your family's health history. Talk to your family members and try to find medical information about your siblings, parents, aunts, uncles, cousins, grandparents, children, and grandchildren. To find out if a condition runs in the family, the genetic counselor will ask questions that relate to the problems for which the person was referred. You can find Family Health History resources.

 [Here](#)

- ✔ **Gather all of your medical records** and, if possible, for any family members who may also be affected by the particular genetic condition.
- ✔ **Think about bringing someone**, either a friend or family member, with you for support and to help you remember all of the information that will be provided to you.
- ✔ **Write down your questions** and concerns in advance. You may want to bring a notepad and pen to take notes during the meeting.

WHAT QUESTIONS MIGHT WE ASK DURING THIS **FIRST** **APPOINTMENT?**

You will likely have many questions. The following is an example of questions you might want to ask:



1. Does my child need genetic testing?
2. Why does my child need genetic testing?
3. What kinds of genetic testing will be done?
4. Where and how will it be done?
5. How long will it take to get the results?
6. Will my insurance pay for genetic testing? If not, what other options can I explore?
7. What will genetic testing tell me about my child?
8. If testing provides a diagnosis, what will this mean for my child?
9. Are there other family members that will need genetic testing?
10. Will you be able to tell me about any treatments that will help my child?
11. What happens if the genetic test does not give a diagnosis?
12. Will there be other tests that will need to be done?
13. What comes next?
14. Do we need to return for another appointment?
15. How long does it usually take between appointments?
16. Where can I find more information about genetics and genetic testing?
17. How can I meet other families who are dealing with similar issues?
18. Where can I find a support group or connect with other families?
19. What is the best way to contact you if I have more questions later?

WHAT HAPPENS **NEXT?**

The genetics team might ask for tests for your child such as a blood test or an x-ray. In some cases, blood tests may also be recommended for other family members like parents or siblings. The team will also write a report about your child's condition and the suggestions they have for next steps. The report will become part of your child's medical record for you and other doctors to read.

You may not receive a diagnosis for your child at the first appointment. It may be that your child doesn't have a genetic condition or the doctor may be waiting for test results to make a diagnosis.

If you have questions after your appointment, please get back in touch with the genetics office. There may be need for future genetics appointments, so stay in touch to see when to return. If you feel like you want to learn more or need to talk with someone who understands what you are going through, ask for resources that will be helpful for you and your family. You might also want to consider these national resources as a place to learn more:

The Family Center

Expecting Health

National Coordinating Center for the Regional Genetics Networks

Regional Genetics Networks

Global Genes

NORD (National Organization for Rare Disorders)

Genetic Alliance

P2P is a program that provides information and one-to-one emotional support to parents of children who have special needs. Trained and experienced parents are carefully matched in one-to-one relationships with parents who are new to the program.

Connect with your Parent Center for Parent-to-Parent (P2P) support.

Even if there is not a clear diagnosis at first, the genetics team can still provide information on next steps and resources.

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Molly Martzke

Bailey Perez

Cindy Weber

Mita Bhattacharya

Marie Torres Perez

Deepa Srinivasavaradan

Jeannette Mejias

Linda Hampton Starnes

Sarita Edwards

Michael Allen

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