



Why was my child referred to a medical genetic clinic?

(What is Medical Genetics?)

Why was my child referred to a medical genetic clinic?

Some children are born with differences in body structure, growth, brain development, or body chemistry that can lead to problems with health, development, school performance, and/or social interaction.

Your child has been referred to genetics clinic because you or your physician are concerned about why your child may have these medical problems or developmental (learning) problems.

What is medical Genetics?

- Medical genetics is a medical specialty, just like cardiology, or the ear, nose, and throat doctor.
- Medical geneticists and genetic counselors are the health care providers that work in medical genetics.
- Medical genetics deals with health problems that are caused or thought to be caused by changes in our genetic material
-

What is a gene?

Genes are the body's blueprints that determines the way our body works including our physical features, such as hair color, eye color but also how we are formed and developed. Genes can cause genetic conditions or disease that may be passed from parents to their children ("inherited").

There are thousands of documented genetic conditions, but the most common conditions include the following:

- Chromosomal changes (e.g. Down syndrome), which cause birth defects, intellectual/developmental disabilities and/or reproductive problems
- Single gene disorders such as cystic fibrosis, muscular dystrophy, Huntington's disease and sickle cell disease

- Birth defects such as spina bifida, cleft lip and cleft palate may have a genetic component

Why is it important to go to a genetics clinic if my child's medical problems are already being treated?

First, if your child is diagnosed with a specific genetic condition, medical providers may be able to give you more information about what can be done for your child including treatment or what can be expected in the future. Second, your child's condition may be inherited. It may have been passed down from family members who may or may not show signs of the medical condition. If your child's condition is inherited you can learn if this condition may also affect you, other family members or possible future pregnancies in your family.

Who can refer my child to the genetic clinic?

A doctor, physician assistant or nurse practitioner can refer your child to a genetics clinic. Sometimes parents can also call the genetics clinics directly to schedule an appointment.

How long will I have to wait for an appointment?

As there are few genetic service providers in most areas, you may have to wait from 3 to 6 months for an appointment at the genetics clinic.

What happens at a medical genetics clinic visit and after?

Who will my child see at a genetic clinic?

Genetic services are often provided through a team of medical providers.

- A medical doctor, a physician assistant or a nurse practitioner who specializes in medical genetics
- A genetic counselor
- Other medical professionals such as nurses, medical students or resident doctors.

What should I expect at my child's appointment?

Most genetic clinic appointments last 45 to 60 minutes. You may want to bring someone with you that can help you ask questions and take notes. At a genetics clinic you can expect the following:

- You will discuss your child's health and developmental concerns with the genetic provider.
- The genetic counselor will collect your child's family health history:
 - You will be asked about the health/developmental history of your child's siblings, parents, aunts, uncles, cousins, and grandparents. This information helps the genetics

- team figure out if your child's condition may be related to other health issues in the family.
- Please bring as much information about the health of family members as possible.
 - A physical examination of your child and possibly other family members will be performed by the medical geneticist.
 - The medical geneticist may take photographs to document certain physical features of your child.
 - The medical geneticist will explain any findings to you and discuss options for further evaluation if needed.
 - You will be encouraged to ask questions, and you, the geneticist and the genetic counselor will decide together about any further testing and evaluations.

What kind of tests are done in a genetic clinic?

- Tests on blood and/or urine samples may be ordered for your child. These samples may be collected on the day of the clinic visit. Others testing such as x-rays or MRIs may be ordered but generally cannot be done on the day of the clinic visit.
- Some genetic testing may require authorization from your insurance company due to cost, before it can be started. The insurance authorization process can take up to three months. If the insurance company approves the test and a sample is obtained and sent, it may take 4 to 6 weeks to receive results. Sometimes insurance companies may not approve genetic testing, and the testing may not be performed unless you can pay for the test yourself which can be thousands of dollars.

Will my child be diagnosed at the first genetic clinic visits?

Probably not. It can often take a while, maybe many months, before a child is diagnosed. Sometimes a child may not be diagnosed.

What happens next if my child is diagnosed with a genetic condition?

A genetic counseling appointment may be offered to discuss your child's condition, how this condition may be inherited and whether other family members may benefit from testing. A letter will be written to the referring health professional about your child's genetic condition and a copy may be sent to you. Your child may or may not need to return to genetics clinic or be referred to other medical specialists. Many times, the referring medical provider can provide care for the condition with guidance from the genetics team.

What happens next if my child is not diagnosed with a genetic condition?

The referring health professional will receive a letter from the genetics team summarizing your visit and the findings. The referring health provider will continue with your child's care. However, your child may still return to the genetics clinic once every one or two years in case new testing becomes available for your child.