

There are many different kinds of genetic conditions. Some are caused by whole extra or missing [chromosomes](#) (such as [Down syndrome](#)), while others are caused by changes in a specific gene (such as sickle cell anemia). There is a third group of genetic conditions called microdeletion syndromes. A microdeletion is when a small area of a chromosome (which is made up of our [DNA](#)) is deleted, or missing.

Each section of our DNA has genes that perform different functions in our body. For example, if part of our DNA is deleted that contains genes that help in brain development, that can cause difficulties in brain function. Identifying what section of DNA is missing can sometimes help determine what the related health concerns may be. Other times, the section of DNA that is missing may contain genes that we are not yet familiar with and do not know what they do, meaning that we don't necessarily know what sort of medical concerns, if any, to expect.

There are many microdeletion syndromes that have been fairly well defined. Click below to get more information:

- [DiGeorge syndrome \(22q11.2 deletion\)](#)
- [Jacobsen syndrome \(11q deletion\)](#)
- [Wolf-Hirschhorn syndrome \(4p deletion\)](#)
- [Cri-du-chat syndrome \(5p deletion\)](#)
- [Langer-Giedion syndrome \(8q deletion\)](#)

Click [here](#) to learn more about scheduling a genetic counseling appointment for pregnancy-related questions.

Click [here](#) to learn more about scheduling a genetic counseling appointment for infertility or preconception questions.

Click [here](#) to learn more about scheduling a genetic counseling appointment for questions about pediatric or adult genetic conditions.

Related Articles

- [Down Syndrome](#)
Down syndrome, also known as trisomy 21, is a genetic condition caused by an extra copy of the 21st chromosome. Each individual with Down syndrome is unique.

Individuals with Down syndrome have variable health issues, including developmental delays and some degree of intellectual disability. What is life like for people...

- [Trisomy 18 \(Edwards Syndrome\)](#)

Trisomy 18, also known as Edwards syndrome, is a genetic condition caused by an extra chromosome 18. Babies with trisomy 18 generally have many complex medical complications, including heart defects, growth restriction, a small abnormally shaped head, and clenched fingers with overlapping fingers. Because of the severity of medical complications...

- [Trisomy 13 \(Patau Syndrome\)](#)

Trisomy 13, also known as Patau syndrome, is a genetic condition caused by an extra chromosome 13. Babies with trisomy 13 generally have many complex medical complications, including heart defects, brain and spinal cord abnormalities, very small or poorly developed eyes, cleft lip and/or cleft palate, and low muscle tone...

- [Open Neural Tube Defects](#)

Open neural tube defects (ONTDs), which includes spina bifida and anencephaly, is a range of birth defects that can include the brain and/or spinal cord. This is generally something that happens very early in pregnancy, and can vary widely in severity and expected outcome. Although the cause of most cases...

- [Sex Chromosome Variations](#)

X and Y chromosome variations are conditions that result when a baby have a different number of sex chromosomes, also known as X & Y chromosomes, than we would expect to see in some or all of the cells in their body. Typically, girls have two X chromosomes while boys...