



During pregnancy many women are concerned about the health and development of their baby. As you learn about genetic testing options, it can be overwhelming. It is important to remember that although you may be hearing about these genetic conditions and birth defects a lot during your pregnancy, they are usually quite rare, occurring in only 3-4% of all births. We like to remind people that 96-97% of babies are born without any birth defects or genetic conditions.

This number, 3-4%, is called the background risk or chance of a birth defect. What this means is that 3-4 out of every 100 babies born will have some type of birth defect; this also means that 96-97 of them will not.

There are many different causes of birth defects, and many birth defects have some genetic basis or influence. However, in many cases, doctors are not able to determine the cause or the reason for the birth defect.

Below is a list of some of the causes and/or types of genetic conditions and birth defects:

- [Chromosome Conditions](#) - Caused by an extra or missing chromosome. Examples are Down syndrome, Turner syndrome and Trisomy 18. Click [HERE](#) for more information on specific chromosome conditions.
- [Multifactorial Conditions](#) - Multifactorial means there are likely multiple genetic factors involved but also environmental factors. Examples of multifactorial birth defects include neural tube defects such as anencephaly and spina bifida, isolated cleft lip/palate, club foot and many others. Click [HERE](#) for more information on specific multifactorial conditions.
- [Single Gene Conditions](#) - This type of condition is caused by a harmful change (aka "mutation") in a single gene, not a whole chromosome. This may be a dominant, recessive or x-linked condition. Click [HERE](#) for more information on specific single gene conditions.
- [Microdeletion/Microduplication Syndromes](#)- These conditions are the result having an extra or missing piece of a small part of a chromosome or several genes vs. a whole chromosome. For more information about specific microdeletion conditions click [HERE](#).
- [Teratogen](#) - A teratogen is an exposure that can cause abnormalities in the form or function of a developing baby. Some examples of teratogens include street drugs, alcohol, prescription drugs, maternal illness or infection, heat and radiation.
- [Uterine factors](#) - Changes related to the uterus can be associated with birth defects. Some examples of uterine factors include differences in the amount of amniotic fluid (decreased), the shape uterus or tumors in the uterus.



- **Unknown** - The reason for many birth defects is often never determined.

Some of the most common genetic conditions and birth defects encountered or discussed during the pregnancy are those that can be detected through prenatal screening and/or diagnosis, are seen on prenatal ultrasound or are routinely discussed due to carrier screening recommendations. While it may be common to discuss these conditions, it is important to remember that most of these conditions are rare. There are many great resources available that provide information on these conditions and others; the Genetic Support Foundation has compiled up-to-date information and resource links to help you get started.