



## **BIRTH DEFECTS FREQUENTLY ASKED QUESTIONS (FAQs)**

### ***What is a birth defect?***

A birth defect is a problem that happens while the baby is developing in the mother's body. Most birth defects happen during the first 3 months of pregnancy.

A birth defect may affect how the body looks, works, or both. It can be found before birth, at birth, or anytime after birth. Most defects are found within the first year of life. Some birth defects (such as cleft lip or clubfoot) are easy to see, but others (such as heart defects or hearing loss) are found using special tests (such as x-rays, CAT scans, or hearing tests). Birth defects can vary from mild to severe.

Some birth defects can cause the baby to die. Babies with birth defects may need surgery or other medical treatments, but, if they receive the help they need, these babies often lead full lives.

### ***What are the most common birth defects?***

One of every 33 babies is born with a birth defect. A birth defect can affect almost any part of the body. The well being of the child depends mostly on which organ or body part is involved and how much it is affected.

Many birth defects affect the heart. About 1 in every 100 to 200 babies is born with a heart defect. Heart defects make up about one-third to one-fourth of all birth defects. Some of these heart defects can be serious, and a few are very severe. In some places of the world, heart defects cause half of all deaths from birth defects in children less than 1 year of age.

Other common birth defects are "neural tube defects," which are defects of the spine ([spina bifida](#)) and brain ([anencephaly](#)). They affect about 1 of 1,000 pregnancies. These defects can be serious and are often life threatening. They happen less often than heart defects, but they cause many fetal and infant deaths.

Birth defects of the lip and roof of the mouth are also common. These birth defects, known as "orofacial clefts," include cleft lip, cleft palate, and combined cleft lip and cleft palate. Cleft lip is more common than cleft palate. In many places of the world, orofacial clefts affect about 1 in 700 to 1,000 babies.

Some birth defects are common but rarely life threatening, though they often require medical and surgical attention. "Hypospadias," for example, is a fairly common defect found in male babies. In babies with hypospadias, the opening of the urethra (where urine comes out) is not at the tip of the penis but on the underside. Treatment depends on how far away from the tip the opening is and can involve complex surgery. This defect is rarely as serious as the others listed above, but it can cause great concern and sometimes has high medical costs. It rarely causes death.

These are only some of the most common birth defects. Two final points are worth noting. First, genetic conditions, though not mentioned so far, also occur often. Down syndrome, for example, is a genetic condition that affects about 1 in 800 babies, but it affects many more babies who are born to older women. Second, a woman who is pregnant may miscarry a baby (fetus) early, before it is time for the baby to be born. This often happens when the fetus has a severe birth defect. To know the true impact of birth defects and how often they occur, we not only need to look at babies born but also, if possible, look at all pregnancies.

### **What is my chance of having a baby with a birth defect?**

In the United States, about 3% of babies are born with birth defects. Some women have a higher chance of having a child with a birth defect. Women over the age of 35 years have a higher chance of having a child with Down syndrome than women who are younger. If taken when a woman is pregnant, certain drugs can increase the chance of birth defects. Also, women who smoke and use alcohol while pregnant have a higher risk of having a baby with certain birth defects. Other women have a higher chance of having a baby with a birth defect because someone in their family had a similar birth defect. To learn more about your risk of having a baby with a birth defect, you can



talk with a [genetic counselor](#). (To find a genetic counselor, see [Where can I find a geneticist or genetic counselor?](#)) Also, to reduce your chances of having a baby with a birth defect, talk with your health care provider about any medicines that you take, do not drink alcohol or smoke, and be sure to take 400 micrograms of the B vitamin folic acid every day. It is the amount of folic acid found in most multivitamins.

***Does my risk for having a baby with a birth defect increase as I get older?***

Women who are 35 years of age or older have a greater chance of having a baby with Down syndrome. Of the known causes of mental retardation, Down syndrome is the most common. It affects about 1 in 800 births. Down syndrome happens when there is an extra chromosome 21 (“trisomy 21”). Scientists have not proven that other birth defects, genetic or otherwise, are linked to the mother’s age.

***Do genetic factors play a role in causing birth defects?***

Yes, some birth defects “run in the family.” Babies with certain types of birth defects may have an extra or a missing chromosome. Birth defects can also happen when just a piece of a chromosome is missing or if just an extra piece is added. Also, certain genes may make a fetus more sensitive to things that cause birth defects.

***What causes birth defects?***

We do not know what causes most birth defects. Sometimes they just happen and are not caused by anything that the parents did or didn't do. Many parents feel guilty if they have a child with a birth defect even if they did everything they could to have a healthy child. If you have a child with a birth defect, it might be helpful to talk with other parents who have had a child with the same condition (See [How can I get in touch with parents of a child with the same birth defect as my child?](#)) Sometimes the causes of birth defects are figured out after the baby is born. Whenever possible, it is important to know what you can do for a better chance of having a healthy child in the future. Some actions might increase the chances of having a baby with a birth defect. The questions and answers that follow talk about some of these known risks.

***Do prescription drugs cause birth defects?***

Some prescription drugs cause birth defects and should never be taken if there is any chance that a woman is pregnant or could become pregnant while taking the drug. Drugs that are used to treat a serious or life threatening illness should be avoided if possible. Prescription drugs known to cause birth defects include thalidomide (Thalomid<sup>®</sup>) and isotretinoin (Accutane<sup>®</sup>). A pregnant woman should always talk with her doctor about the risks and benefits of any drugs before taking them. Even a woman who is not pregnant now but who might get pregnant while she is taking these drugs should talk with her doctor. It is very important to use two reliable forms of birth control if you are taking these drugs.

There are a few drugs that do not have any link with birth defects when taken by a pregnant woman. We say those drugs have little risk. Most drugs are between the two extremes of having great risk and little risk. Also, there are no studies of how most drugs affect pregnant women. As a result, we do not know if it is safe for a pregnant woman to take these drugs. Women who could get pregnant should talk with their doctor about any drugs they are taking. Together, they can decide if the benefit of the drug is worth the possible risk. If a woman finds out that she is pregnant while she is taking a drug, she should talk to her doctor soon. Some drugs are needed to keep the mother healthy. Not taking some drugs while she is pregnant may put both the woman’s and her baby’s health in danger. It is important for the woman and her doctor to talk about any drug she might need to take. The doctor can consider the woman’s full medical history.

***Does alcohol cause birth defects?***



The U.S. Public Health Service has indicated that there is no safe level of alcohol use during pregnancy. If a woman drinks while pregnant, she puts her developing fetus at risk for a wide spectrum of adverse effects including spontaneous abortion; growth retardation; physical, mental, and behavioral abnormalities; facial abnormalities; and CNS impairment, such as developmental delay, speech or language delay, lower IQ, and decreased head circumference. In the worst cases, prenatal exposure to alcohol may result in fetal death.

<http://www.cdc.gov/ncbddd/fas/fasask.htm>

***Does smoking cause birth defects?***

A woman who smokes while she is pregnant has a greater chance of having a premature (early) birth, a small baby, or a stillborn baby. If the mother smokes while pregnant, there is also an increased risk of the baby dying during the first year of life. Some types of birth defects have been linked to the mother's smoking. Birth defects that may be increased when the mother smokes include: cleft lip, cleft palate, clubfoot, limb defects, some types of heart defects, gastroschisis (an opening in the muscles of the abdomen that allows the intestines to appear outside the body), and imperforate anus (there is no opening from the intestines to the outside of the body to allow stool or gas to be passed). Talk with your health care provider about ways to help you quit smoking if you are pregnant or can get pregnant.

***Do illegal drugs cause birth defects?***

Women who use illegal drugs, or "street drugs," can have babies who are small, premature, or have other health problems, such as birth defects.

Women who use cocaine while pregnant are more likely to have babies with birth defects of the limbs, gut, kidneys, urinary system, and heart. Other drugs, such as marijuana and ecstasy, may also cause birth defects in babies.

Women should not use street drugs while they are pregnant. It is also important that women not use street drugs after they give birth because drugs can be passed through breast milk and can affect a baby's growth and development. Talk with your health care provider about ways to help you quit using street drugs **before** you get pregnant.

***Does exposure to the MMR vaccine cause birth defects? Other vaccines?***

The MMR is a vaccine for measles, mumps, and rubella. If a woman gets one of these viruses while pregnant, it may cause her to have a miscarriage or to have a baby with birth defects. The MMR, like some other vaccines, is made with viruses that are alive but very weak. Because these viruses are alive, there is a very slight chance that they may cause harm to the baby. For this reason, a woman who may be pregnant should not get an MMR or other vaccine unless she is at high risk of getting a serious illness without it. She should talk about the risks and benefits of getting the vaccine with her doctor. A woman who has just gotten the vaccine and then learns she is pregnant should also talk with her doctor. Vaccines such as those for tetanus and hepatitis are made from dead viruses or parts of dead viruses and do not cause infection in the mother and should not harm the fetus.

***When in pregnancy do birth defects happen?***

Birth defects happen before a baby is born. Inherited or genetic factors; things in the environment, such as smoking or drinking alcohol or not getting enough folic acid; and a woman's illness during pregnancy can cause birth defects. Most birth defects happen in the first 3 months of pregnancy, when the organs of the baby are forming. This is the most important stage of development. However, some birth defects happen later in pregnancy. During the last six months of pregnancy, the tissues and organs continue to grow and develop.



Some birth defects can be found before birth. If you want to know more about your risk of having a baby with a birth defect, contact a genetic counselor. See [Where can I find a clinical geneticist or genetic counselor?](#)

***What kind of health care provider can find out what caused my baby's birth defect?***

Birth defects are common in our country. Some birth defects are found before birth, some at the time of birth, and some are found during the first year of life. A few don't show up until the child is older. It is common for parents to want to know what caused their baby's birth defect. However, the causes for about 70% birth defects are unknown.

A primary care provider (PCP) usually looks at a child who may have a birth defect. The PCP is most often the child's pediatrician or the family's general physician. PCPs look for important clues in the child's first exam for a birth defect. The first exam includes a lot of questions about history, a physical exam, and sometimes testing. The PCP is trying to find a "diagnosis" (name or cause) for the child's type of birth defect. If a diagnosis cannot be made after the first exam, the PCP may refer the child to a specialist in birth defects and genetics. A clinical geneticist is a doctor with special training to evaluate patients who may have genetic conditions or birth defects. Even if a child sees a specialist, an exact diagnosis may not be reached.

Counseling the family of an infant with a birth defect is a large part of the PCP's job. PCPs may refer parents to a genetic counselor to help parents learn more about their infant's condition. A genetic counselor explains the diagnosis, the possible role of genes, and medical aspects of the birth defect. A genetic counselor can talk with parents about their risk of having future children with a birth defect. He or she also talks with parents about how to lessen their chances of having another baby with birth defects. Counseling can help a family adjust to and plan for their newborn.

***Who coordinates the health care of my child who has a birth defect?***

The PCP usually provides the basic care of a child with a birth defect. The PCP should know about sources of help for the child and the family. Help may include support groups, public health services, and current medical literature. The PCP also coordinates the child's care. For example, children with birth defects involving their bones may need to see an orthopedist, a doctor trained in problems with the bones. A child with a birth defect involving the brain may need to see a neurologist, who is trained to deal with problems in the brain and nervous system. The PCP may also send the child for special services that will help the child function better. For example, a child with a cleft palate may be sent to a speech therapist, someone with special training who works with people to improve their ability to talk. Another common referral is for physical therapy to improve the child's strength and movement.

Many children with birth defects have more than one problem and may need one or more specialists. The PCP coordinates the care of a child with a birth defect so that he gets all the special care he needs.

In summary, birth defects are common, but the causes for many birth defects are not known at this time. The PCP is generally the best person to coordinate the special care needed for a child with a birth defect.

***What does a genetic counselor do?***

A genetic counselor talks with you about birth defects and genetic conditions. Genetic counselors are part of the health care team and have special training to help families learn about birth defects and conditions passed down through a family. They can guide families to other resources for help. They also help families deal with feelings about how these conditions affect their family. People talk about both medical and personal questions during genetic counseling. Genetic counselors ask questions about family history and pregnancy history. They talk with families about tests used to find a condition and, if known, about how to prevent a condition.



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There are many reasons to see a genetic counselor. Some people go because of a family history of a genetic condition. Others see a genetic counselor because they have trouble getting pregnant or because they have had several miscarriages or infant deaths. Some women may see a genetic counselor after learning the results of a blood test or because of their age. Still others seek genetic counseling to learn about the effects of being exposed to things like x-rays, chemicals, illness, or drugs while pregnant.

***Where can I find a clinical geneticist or genetic counselor?***

Your best source is your health care provider. He or she will know about the resources in your area. Also, you can call the nearest university medical school or large medical center. To reach them, call the main telephone number and ask for "genetics."

Reference: <http://www.cdc.gov/ncbddd/bd/faq1.htm>