



## **DISEASES THAT OCCUR DURING THE FETAL PERIOD**

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### **Annotation:**

A birth defect is something visibly abnormal, internally abnormal, or chemically abnormal about your newborn baby's body. The defect might be caused by genetics, infection, radiation, or drug exposure, or there might be no known reason. Examples of birth defects include phenylketonuria, sickle cell anemia and Down syndrome.

**Keywords:** down syndrome, chemical imbalance, healthcare.

A birth defect is something abnormal about your newborn baby's body. Every four and a half minutes, a baby in the United States is born with a birth defect. A defect, which can affect almost any part of your baby's body, can be:

- Visibly obvious, like a missing arm or a birthmark.
- Internal (inside the body), like a kidney that hasn't formed right or a ventricular septal defect (a hole between the lower chambers of your baby's heart).
- A chemical imbalance, like phenylketonuria (a defect in a chemical reaction that results in developmental delay).

Your baby can be born with one birth defect such as a cleft lip (a gap in their upper lip) or multiple birth defects such as a cleft lip and cleft palate (a hole in the roof of their mouth) together, or even a cleft lip and cleft palate with defects of the brain, heart and kidneys.

Your healthcare provider won't be able to detect all birth defects right when your baby is born. Some defects, such as scoliosis, might not be apparent until your child is several months old. An abnormal kidney might take years to be discovered.

How common are birth defects?

Birth defects are common. Between 2% and 3% of infants have one or more defects at birth. That number increases to 5% by age one (not all defects are discovered directly after your child's birth). One out of every 33 babies born in the United States are affected by birth defects.

What are some other examples of birth defects?

- Duodenal atresia, an obstruction in the small intestine. It can cause polyhydramnios (extra fluid around the fetus in pregnancy), which can increase the risk of preterm birth. It is sometimes associated with other genetic syndromes.
- Dandy walker malformation, an abnormal development of the posterior fossa (a space in your baby's skull) and cerebellum (a section of the brain). This birth defect can cause a variety of problems.
- Limb defects, which happen when the fetal amnion (the inner lining of the amniotic sack) wraps around parts of the fetus (like a finger or foot).



Every human body cell contains 46 chromosomes, and each chromosome contains thousands of genes. Each gene contains a blueprint that controls development or function of a particular body part. People who have either too many or too few chromosomes will therefore receive a scrambled message regarding anatomic development and function.

Down syndrome is an example of a condition caused by too many chromosomes. Because of an accident during cell division, individuals with Down syndrome have an extra copy of a particular chromosome (chromosome 21). This extra chromosome can cause a typical constellation of birth defects. Characteristic features of Down syndrome can include developmental delay, muscle weakness, downward slant of the eyes, low-set and malformed ears, an abnormal crease in the palm of the hand and birth defects of the heart and intestines.

With Turner syndrome, a female lacks part or all of one X chromosome. In the affected women, this can cause short stature, learning disabilities and absence of ovaries.

Trisomy 13 (Patau Syndrome) and Trisomy 18 (Edwards Syndrome) are caused by inheriting extra copies of the 13th or 18th chromosome, respectively. These are rarer, more serious conditions which cause severe birth defects that are incompatible with survival after birth.

In addition to inheriting an extra or absent chromosome, deletions or duplications of single genes can also cause developmental disorders and birth defects. One example is cystic fibrosis (a disorder that causes progressive damage of the lungs and pancreas).

Defective genes can also be caused by accidental damage, a condition called “spontaneous mutation.” Most cases of achondroplasia (a condition that causes extreme short stature and malformed bones) are caused by new damage to the controlling gene. In addition, recombination errors can cause translocations of chromosomes which may lead to complex birth defects.

Environmental factors can increase the risk of miscarriage, birth defects, or they might have no effect on the fetus at all, depending on at what point during the pregnancy the exposure occurs.

The developing fetus goes through two major stages of development after conception. The first, or embryo stage, occurs during the first 10 weeks after conception. Most of the major body systems and organs form during this time. The second, or fetal stage, is the remainder of the pregnancy. This fetal period is a time of growth of the organs and of the fetus in general. Your developing fetus is most vulnerable to injury during the embryo stage when organs are developing. Indeed, infections and drugs can cause the greatest damage when exposure occurs two to 10 weeks after conception.

Diabetes and obesity can possibly increase your child’s risk of birth defects. Your healthcare provider may suggest that you do your best to manage these conditions before you get pregnant.

Some medicines and recreational drugs can cause birth defects, which are most severe when used during the first three months of pregnancy. Thalidomide, an anti-nausea medicine prescribed during the 1960s, caused birth defects called phocomelia (absence of most of the arm with the hands extending flipper-like from the shoulders).



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