



## BASIC INFORMATION

### DESCRIPTION

A birth defect is described as any abnormality present at birth. Birth defects are sometimes referred to as congenital defects or a congenital anomaly. Defects may be inherited genetically, acquired during pregnancy, or caused by a birth injury. There are about 120,000 babies born each year with a birth defect. They can affect the baby's looks and health as well as the ability to function. Birth defects are the leading cause of infant deaths in the U.S. There are some birth defects that can be prevented, and some birth defects that can be treated with medication or surgery.

### FREQUENT SIGNS AND SYMPTOMS

Birth defects can be minor, such as a birthmark, or major, such as spina bifida (the spinal canal does not close completely). Congenital heart disease is the most common birth defect. Many birth defects can be seen right away. Others appear later in life, although they were present at birth.

### CAUSES

- The causes of about 50% of all birth defects are unknown.
- Chromosome defects - children are born with more or less of the normal 23 pairs of chromosomes, are missing a portion of a chromosome, or have a portion of a chromosome that is out of the normal sequence.
- Genetic or hereditary defects - these are defects inherited from one or both parents.
- Harmful agents (teratogens) - certain prescription medications or drugs of abuse taken by the mother, smoking, alcohol consumption, and other chemicals.
- Infections - some illnesses during pregnancy can cause birth defects, such as rubella (German measles) or toxoplasmosis.
- Factors involving the uterus - known causes such as too little fluid around the fetus or other causes that are unknown.
- Sometimes, a mix of inherited traits and exposure during pregnancy is the cause of a birth defect.

### RISK INCREASES WITH

- Maternal age 35 or older when the baby is due.
- Paternal age over 40 at time of conception.
- Family history of congenital heart defects, Down syndrome, neural tube defects, hemophilia, muscular dystrophy, cystic fibrosis, Huntington disease, or fragile X syndrome.
- Family members or siblings have any other genetic diseases, chromosomal disorders or birth defects.
- Parents are of Mediterranean or Asian descent.
- Parents are of eastern European Jewish or French Canadian descent, and there is a family history of Tay-Sachs disease.
- Parents are of eastern European Jewish descent, and there is a family history of Canavan disease.
- Parents are African-American, and there is a family history of sickle cell disease or trait.

- Exposure to harmful substances during pregnancy (e.g., chemicals, alcohol, drugs of abuse, certain medications, or higher than recommended doses of vitamin or mineral supplements).
- X-ray exposure of the fetus increases the risk of malformations.
- Maternal metabolic disorder such as diabetes or phenylketonuria. Women with these disorders should consult their doctor before becoming pregnant.
- In addition, there are many babies with birth defects that are born to couples with no known risk factors.

### PREVENTIVE MEASURES

- If possible, seek medical advice before becoming pregnant.
- During prenatal care, some tests are offered to all women (screening tests), and some are offered only to those with known risk factors (diagnostic tests). These tests, along with genetic counseling, will help tell patients about their risk of a problem.
- Get genetic counseling if either parent has any risk factors for hereditary or genetic abnormalities. Genetic counseling can help assess the risk for having a baby with a birth defect and recommend the appropriate tests. Choosing whether to have a test done is up to you. Some couples choose not to be tested for birth defects. Other couples find that testing and counseling can help them decide whether to become pregnant or continue a pregnancy. No test is considered perfect. Your baby may have a birth defect even if a test result doesn't indicate a problem.
- Take care of yourself during pregnancy.
- Avoid exposure to harmful substances and drugs of abuse.
- Protect yourself against infection during pregnancy.
- Start folic acid supplements one month prior to conception.
- Don't smoke or use alcohol during pregnancy.
- Ask your obstetric provider about the medications you currently take.
- Don't take any medications, vitamins, or minerals during pregnancy unless approved by your obstetric provider.
- Avoid X-rays, even dental X-rays, from the time you suspect or know you are pregnant.
- Currently, there are some birth defects that cannot be prevented. There are ongoing studies as to whether environmental pollutants cause birth defects, developmental disabilities, or other adverse reproductive outcomes. There are also many questions about whether various occupational hazards, dietary factors, medications, and personal behaviors cause or contribute to birth defects.
- For more information, visit your local library or do a web search.



## NOTIFY OUR OFFICE IF

- You or a family member is considering pregnancy.
- You are pregnant and are concerned about the possibility of birth defects.