



BIRTH DEFECTS FACT SHEET

Background

About 150,000 babies are born in the United States each year with birth defects. The parents of one out of every 28 babies receive the news that their baby has a birth defect.

A birth defect is an abnormality of structure, function or metabolism (body chemistry) present at birth that results in physical or mental disability, or is fatal. Several thousand different birth defects have been identified. Birth defects are the leading cause of death in the first year of life.

What Causes Birth Defects?

Both genetic and environmental factors can cause birth defects. However, the causes of about 60 to 70 percent of birth defects currently are unknown.

A single abnormal gene can cause birth defects. Every human being has at least 30,000 to 35,000 genes that determine traits like eye and hair color, as well as direct the growth and development of every part of our physical and biochemical systems. Genes are packaged into each of the 46 chromosomes inside our cells.

Each child gets half its genes from each parent. A person can inherit a genetic disease when one parent (who may or may not have the disease) passes along a single faulty gene. This is called dominant inheritance. Examples include achondroplasia (a form of dwarfism) and Marfan syndrome (a connective tissue disease). Many other genetic diseases are inherited only when both parents (who do not have those diseases) happen to carry the same abnormal gene and pass it on to a child. This is called recessive inheritance. Examples include Tay-Sachs disease (a fatal disorder seen mainly in people of European Jewish heritage) and cystic fibrosis (a fatal disorder of lungs and other organs, affecting mainly Caucasians). There also is a form of inheritance (X-linked) where sons can inherit a genetic disease from a mother who carries the gene (usually with no effect on her own health). Examples include hemophilia (a blood-clotting disorder) and Duchenne muscular dystrophy (progressive muscle weakness).

Abnormalities in the number or structure of chromosomes can cause numerous birth defects. Due to an error that occurred when an egg or sperm cell was developing, a baby can be born with too many or too few chromosomes, or with one or more chromosomes that are broken or rearranged. Down syndrome, in which a baby is born with an extra chromosome 21, is one of the most common chromosomal abnormalities. Affected children have varying degrees of mental retardation, characteristic facial features and, often, heart defects and other problems. Babies born with extra copies of chromosome 18 or 13 have multiple birth defects and usually die in the first months of life.

Missing or extra sex chromosomes (X and Y) affect sexual development and may cause infertility, growth abnormalities, and behavioral and learning problems. However, most affected individuals have essentially normal lives.

Birth defects also may result from environmental factors such as drug or alcohol abuse, infections, or exposure to certain medications (such as the acne drug Accutane) or other chemicals. Many birth defects appear to be caused by a combination of one or more genes and environmental factors (called multifactorial inheritance). Some examples include cleft lip/palate, clubfoot and some heart defects.

What Are Some Common Types of Birth Defects?

Birth defects generally are grouped into three major categories: structural/metabolic, congenital infections, and other conditions.

- **Structural and Metabolic Abnormalities**

When a baby has a structural birth defect, some part of the body (internal or external) is missing or malformed. Heart defects are the most common type of structural birth defect, affecting one baby in 125. While advances in surgery have dramatically improved the outlook for affected babies, these remain the leading cause of birth defect-related infant deaths. Doctors usually do not know what causes a baby's heart



to form abnormally, although genetic and environmental factors are believed to play a role.

Spina bifida (open spine, in which the backbone never completely closes and the spinal cord is usually malformed) affects one in 2,000 babies. Affected babies suffer varying degrees of paralysis, and bladder and bowel problems. Both genetic and nutritional factors appear to play a role.

About one baby in 135 has a structural defect involving the genitals or urinary tract. These vary greatly in severity, ranging from abnormal placement of the urinary opening in males (hypospadias) to absence of both kidneys. The cause of hypospadias, which is surgically correctable, is unknown. Babies who lack both kidneys die in the first hours or days of life. This tragic defect is sometimes inherited.

Metabolic disorders affect one in 3,500 babies. These disorders are not visible, but can be harmful or even fatal. Most are recessive genetic diseases. These diseases result from the inability of cells to produce an enzyme (protein) needed to change certain chemicals into others, or to carry substances from one place to another. An example is Tay-Sachs disease. Affected babies lack an enzyme needed to break down certain fatty substances in brain cells. These substances build up and destroy brain cells, resulting in blindness, paralysis and death by age five. Phenylketonuria (PKU) is another metabolic disorder, in which affected babies cannot process a part of protein, which builds up in blood and causes brain damage. PKU is routinely detected with newborn screening tests, so affected babies can be placed on a special diet that prevents mental retardation.

- **Congenital Infections**

Rubella (German measles) probably is the best known congenital infection that can cause birth defects. If a pregnant woman is infected in the first trimester, her baby has a one-in-four chance of being born with one or more features of congenital rubella syndrome (deafness, mental retardation, heart defects, blindness). Fortunately, with widespread vaccination, this syndrome is now rare in this country.

The most common congenital viral infection is cytomegalovirus (CMV). About 1 percent (40,000 babies a year) of all newborns in this country are infected, although only about 10 percent of them (3,000-4,000) have serious consequences, including mental retardation, and loss of vision and hearing. Pregnant women often acquire CMV from young children, who usually have few or no symptoms.

Sexually transmitted infections in the mother also can endanger the fetus and newborn. For example, untreated syphilis can result in stillbirth, newborn death, or bone defects. About one baby in 2,000 is affected.

- **Other Causes**

Other causes of birth defects include fetal alcohol syndrome, which affects one baby in 1,000. This pattern of mental and physical birth defects is common in babies of mothers who drink heavily during pregnancy. Even moderate or light drinking during pregnancy can pose a risk to the baby.

Rh disease of the newborn, which is caused by an incompatibility between the blood of a mother and her fetus, affects about 4,000 infants a year. It can result in jaundice (yellowing of the skin), anemia, brain damage and death. Rh disease usually can be prevented by giving an Rh-negative woman an injection of a blood product called immunoglobulin at 28 weeks of pregnancy and after the delivery of an Rh-positive baby.

Babies of mothers who use cocaine early in pregnancy may be at increased risk of birth defects. A large study has suggested that these babies are five times more likely to be born with urinary tract defects than babies of women who don't use cocaine.



Can Birth Defects Be Prevented?

While the causes of most birth defects are not known, there are a number of steps a woman can take to reduce her risk of having a baby with a birth defect. One important step is a pre-pregnancy visit with her health care provider. During this visit, the provider can obtain valuable information about a woman or couple's family history, which may help identify risk factors for birth defects or inherited genetic conditions. This information allows for appropriate testing and screening to be offered prior to or during pregnancy. During a pre-pregnancy visit, providers also can take a good look at a woman's health and lifestyle, and guide her in any changes that could improve her chances of having a healthy baby.

A pre-pregnancy visit is especially crucial for women with medical problems like diabetes, high blood pressure, and epilepsy, which can affect pregnancy. For example, women with poorly controlled diabetes are several times more likely than women without diabetes to have a baby with a serious birth defect. However, if their blood sugar levels are well controlled starting before pregnancy, they are almost as likely to have a healthy baby as women without diabetes.

If a woman has never had chickenpox (and has not been vaccinated), a pre-pregnancy visit is a good time to check whether she should be vaccinated prior to pregnancy. Like rubella, chickenpox can cause birth defects when contracted by the pregnant woman, although the risk is low. If she has not been vaccinated against rubella since childhood, she should ask her doctor about the rubella vaccine or a combination vaccine such as measles-mumps-rubella (MMR). She should avoid pregnancy for one month after chickenpox, rubella or MMR vaccination.

All women who could become pregnant should take a daily multivitamin containing 400 micrograms of the B-vitamin folic acid. Studies show that taking this vitamin prior to and in the early weeks of pregnancy reduces the risk of having a baby with certain birth defects of the brain and spine, including spina bifida. If a woman already has had a baby with one of these birth defects, she should consult her doctor prior to pregnancy about how much folic acid to take. Generally, a higher dose, 4 milligrams, is recommended.

A woman who is pregnant or planning pregnancy should avoid alcohol, smoking, and street drugs — these can cause birth defects and other pregnancy complications. She should not take any medication — prescription, over-the-counter, or herbal — without first checking with her health care provider.

Can Some Birth Defects Be Diagnosed Before Birth?

Some birth defects can be diagnosed before birth, using one or more prenatal tests including ultrasound, amniocentesis and chorionic villus sampling (CVS). Ultrasound can help diagnose structural birth defects, such as spina bifida, heart and urinary tract defects. Amniocentesis and CVS are used to diagnose chromosomal abnormalities, such as Down syndrome. They also can detect, or rule out, numerous genetic birth defects that may be suspected because of family history or ethnic background. Can birth defects be treated before birth?

A small percentage of couples will learn through prenatal diagnosis that their baby has a birth defect. While this news can be devastating, prenatal diagnosis sometimes can improve the outlook for the baby. Advances in prenatal therapy now make it possible to treat some birth defects before birth. For example, biotin dependence and methylmalonic acidemia — two life-threatening inherited disorders of body chemistry — have been diagnosed by amniocentesis and treated in the womb, resulting in the births of healthy babies.

Prenatal surgery has saved babies with urinary-tract blockages, rare tumors of the lung, and congenital diaphragmatic hernia (a hole in the muscle that separates the chest from the abdomen). More than 100 babies have undergone experimental prenatal surgery to repair spina bifida before birth. Preliminary results appear promising: fewer babies who have had surgery for spina bifida require shunts to drain fluid from their brain. However, it is too soon to know how well most of these babies will walk, and the procedure leads to preterm birth. Prenatal blood transfusions have saved numerous babies with severe Rh disease, and heart medications given to the pregnant woman have saved babies with serious heart rhythm disturbances. However, even when a fetus has a condition for which prenatal treatment is not yet possible, prenatal diagnosis permits parents to prepare themselves emotionally, and to plan with their provider the safest timing, location and method of delivery.



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Couples who have had a baby with a birth defect, or who have a family history of birth defects, should consider consulting a genetic counselor. These health professionals help families understand what is known about the causes of a birth defect, and the chances of the birth defect recurring in another pregnancy. Genetic counselors also can provide referrals to medical experts as well as to appropriate support groups.

Reference:

March of Dimes Perinatal Data Center. Maternal, Infant, and Child Health in the United States, 2001.