



Image from: [Infant with spina bifida. \(Biophoto... in Gale Encyclopedia of Public Health\)](#)

Summary Article: **Birth Defects**

From *Health Reference Series: Genetic Disorders Sourcebook*

Facts about Birth Defects

¹Birth defects are common, costly, and critical conditions that affect 1 in every 33 babies born in the United States each year.

Birth Defects Are Common

Every 4 ½ minutes, a baby is born with a birth defect in the United States.

That means nearly 120,000 babies are affected by birth defects each year.

Birth defects are structural changes present at birth that can affect almost any part or parts of the body (e.g., heart, brain, foot). They may affect how the body looks, works, or both. Birth defects can vary from mild to severe. The well-being of each child affected with a birth defect depends mostly on which organ or body part is involved and how much it is affected. Depending on the severity of the defect and what body part is affected, the expected lifespan of a person with a birth defect may or may not be affected.

Identifying Birth Defects

A birth defect can be found before birth, at birth, or any time after birth. Most birth defects are found within the first year of life. Some birth defects (such as cleft lip) are easy to see, but others (such as heart defects or hearing loss) are found using special tests, such as echocardiograms (an ultrasound picture of the heart), X-rays or hearing tests.

Causes

Birth defects can occur during any stage of pregnancy. Most birth defects occur in the first 3 months of pregnancy, when the organs of the baby are forming. This is a very important stage of development. However, some birth defects occur later in pregnancy. During the last six months of pregnancy, the tissues and organs continue to grow and develop.

For some birth defects, like fetal alcohol syndrome, we know the cause. But for most birth defects, we don't know what causes them. For most birth defects, we think they are caused by a complex mix of factors. These factors include our genes (information inherited from our parents), our behaviors, and things in the environment. But, we don't fully understand how these factors might work together to cause birth defects.

While we still have more work to do, we have learned a lot about birth defects through past research. For example, some things might increase the chances of having a baby with a birth defect, such as:

- Smoking, drinking alcohol, or taking certain “street” drugs during pregnancy.
- Having certain medical conditions, such as being obese or having uncontrolled diabetes before and during pregnancy.
- Taking certain medications, such as isotretinoin (a drug used to treat severe acne).

- Having someone in your family with a birth defect. To learn more about your risk of having a baby with a birth defect, you can talk with a clinical geneticist or a genetic counselor.
- Being an older mother, typically over the age of 34 years.

Having one or more of these risks doesn't mean you'll have a pregnancy affected by a birth defect. Also, women can have a baby born with a birth defect even when they don't have any of these risks. It is important to talk to your doctor about what you can do to lower your risk.

Prevention

Not all birth defects can be prevented. But, there are things that a woman can do before and during pregnancy to increase her chance of having a healthy baby:

- Be sure to see your healthcare provider regularly and start prenatal care as soon as you think you might be pregnant.
- Get 400 micrograms (mcg) of folic acid every day, starting at least one month before getting pregnant.
- Don't drink alcohol, smoke, or use "street" drugs.
- Talk to a healthcare provider about any medications you are taking or thinking about taking. This includes prescription and over-the-counter medications and dietary or herbal supplements. Don't stop or start taking any type of medication without first talking with a doctor.
- Learn how to prevent infections during pregnancy.
- If possible, be sure any medical conditions are under control, before becoming pregnant. Some conditions that increase the risk for birth defects include diabetes and obesity.

What Are the Types of Genetic Tests?

Genetic testing can provide information about a person's genes and chromosomes. Available types of testing include:

Newborn Screening

Newborn screening is used just after birth to identify genetic disorders that can be treated early in life. Millions of babies are tested each year in the United States. All states currently test infants for phenylketonuria (a genetic disorder that causes intellectual disability if left untreated) and congenital hypothyroidism (a disorder of the thyroid gland). Most states also test for other genetic disorders.

Diagnostic Testing

Diagnostic testing is used to identify or rule out a specific genetic or chromosomal condition. In many cases, genetic testing is used to confirm a diagnosis when a particular condition is suspected based on physical signs and symptoms. Diagnostic testing can be performed before birth or at any time during a person's life, but is not available for all genes or all genetic conditions. The results of a diagnostic test can influence a person's choices about health care and the management of the disorder.

Carrier Testing

Carrier testing is used to identify people who carry one copy of a gene mutation that, when present in two copies, causes a genetic disorder. This type of testing is offered to individuals who have a family

history of a genetic disorder and to people in certain ethnic groups with an increased risk of specific genetic conditions. If both parents are tested, the test can provide information about a couple's risk of having a child with a genetic condition.

Prenatal Testing

Prenatal testing is used to detect changes in a fetus's genes or chromosomes before birth. This type of testing is offered during pregnancy if there is an increased risk that the baby will have a genetic or chromosomal disorder. In some cases, prenatal testing can lessen a couple's uncertainty or help them make decisions about a pregnancy. It cannot identify all possible inherited disorders and birth defects, however.

Preimplantation Testing

Preimplantation testing, also called preimplantation genetic diagnosis (PGD), is a specialized technique that can reduce the risk of having a child with a particular genetic or chromosomal disorder. It is used to detect genetic changes in embryos that were created using assisted reproductive techniques such as in-vitro fertilization. In-vitro fertilization involves removing egg cells from a woman's ovaries and fertilizing them with sperm cells outside the body. To perform preimplantation testing, a small number of cells are taken from these embryos and tested for certain genetic changes. Only embryos without these changes are implanted in the uterus to initiate a pregnancy.

Predictive and Presymptomatic Testing

Predictive and presymptomatic types of testing are used to detect gene mutations associated with disorders that appear after birth, often later in life. These tests can be helpful to people who have a family member with a genetic disorder, but who have no features of the disorder themselves at the time of testing. Predictive testing can identify mutations that increase a person's risk of developing disorders with a genetic basis, such as certain types of cancer. Presymptomatic testing can determine whether a person will develop a genetic disorder, such as hereditary hemochromatosis (an iron overload disorder), before any signs or symptoms appear. The results of predictive and presymptomatic testing can provide information about a person's risk of developing a specific disorder and help with making decisions about medical care.

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abnormalities in specific genes, such as hemophilia, sickle cell disease, and cystic fibrosis, chromosomal disorders, such as down syndrome, fragile x syndrome, and klinefelter syndrome, and complex disorders with environmental and genetic components, such as alzheimer disease, cancer, heart disease, and obesity; along with information about the human genome project, genetic testing and newborn screening, gene therapy and other current research initiatives, the special needs of children with genetic disorders, a glossary of terms, and a directory of resources for further help and information (6th ed.). Detroit, MI: Omnigraphics, Inc. Retrieved from https://search.credoreference.com/content/topic/birth_defects



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