



General Population Risk for Birth Defects

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A “birth defect” is a health problem or physical change, which is present in a baby at the time he/she is born. Birth defects may be very mild in which the baby looks and acts like any other baby, or birth defects may be very severe, and you can immediately tell there is a problem. Some birth defects can be so severe that a baby may live only a few months or die at a young age.

Most babies are born healthy. In fact, 96 to 97 out of every 100 babies born are born healthy. About three or four out of every 100 babies born, however, have some type of birth defect. Birth defects occur in babies all over the world, in families of all nationalities and backgrounds. Anytime a woman becomes pregnant, there is a chance that the baby will have a birth defect. This chance is three to four out of 100, or 3 to 4 percent. This means that there is a 96 to 97 percent chance with each pregnancy for a baby to be born normal and healthy. The “3 to 4 percent” number is sometimes called the “background rate for birth defects” or the “population risk for birth defects.”

There are many reasons why birth defects happen. Most are due to a combination of environmental and genetic factors. About 30 percent of all birth defects have a known cause, such as: chromosome abnormalities, single gene defects, or a teratogenic exposure. The remaining 70 percent of birth defects do not have a known cause. You may find it surprising that scientists and physicians have not determined the cause for all birth defects. There is ongoing research into the causes and prevention of birth defects. Testing is available for some birth defects which have a known cause, depending on your medical background and family history.

In a family in which birth defects are already present, the chance for a couple to have a child with a birth defect may be higher than the background rate of 3 to 4 percent.

Here are some terms used to explain the different causes of birth defects:

Inheritance

Inheritance is a word used to describe a trait given to you or “passed on” to you from one of your parents. Examples of inherited traits would be your eye color or blood type.

Chromosome abnormalities

Chromosomes are structures in the nucleus of each cell that contain your genes. A normal chromosome study on a pregnancy does not rule out the possibility that a child will be born with a birth defect.

Single gene defects

Genes are what determine your traits. A child can inherit not only those genes responsible for their normal traits, but also disease causing genes that result in a birth defect.

Multifactorial inheritance

Multifactorial inheritance means that “many factors” (multifactorial) are involved in causing a birth defect. The factors are usually both genetic and environmental.

Teratogens

A teratogen is an agent which can cause a birth defect. It is usually something in the environment that the mother may be exposed to during her pregnancy. It could be a prescribed medication, a street drug, alcohol, or a disease that the mother has, which could increase the chance for the baby to be born with a birth defect.