

Introduction: Understanding Congenital Heart Defects

The word “congenital” means existing at birth. The terms “congenital heart defect” and “congenital heart disease” are often used to mean the same thing, but “defect” is more accurate. The heart ailment is a defect or abnormality, not a disease. A defect results when the heart or blood vessels near the heart don’t develop normally before birth.

If your child is born with a heart defect today, the chances are better than ever that the problem can be overcome and that a normal adult life will follow. Recent progress in diagnosis and treatment (surgery and heart catheterization) makes it possible to fix most defects, even those once thought to be hopeless. As diagnosis and treatment continue to advance, scientists will develop treatments for other defects.

After your child is diagnosed with a congenital heart defect, more tests may be needed. They’ll help the children’s heart specialist (pediatric cardiologist) and the surgeon to decide whether a treatment (surgery or heart catheterization) is a good idea. But before that point is reached, you can learn a lot to make the best use of the help available to you.

Because no two children are exactly alike, children with the same heart defect may have different courses of treatment. Your doctor can advise you about your child’s specific heart defect and suggest treatment options. This section was designed to help you better understand the information your child’s doctor has given you about his or her defect. It will also tell you what to expect about tests your child may need, stays in the hospital, surgery and recovery at home.

Since having a child with a congenital heart defect is different than having a child with a healthy heart, your child will be best served at a center where people are skilled in caring for children. These people include board-certified pediatric cardiologists, cardiothoracic surgeons, intensive-care experts and a skilled team of nurses and therapists.

Heart centers, hospitals and clinics that specialize in cardiac diagnosis and treatment for children exist across the country. Discuss the experience of the heart surgery team and the hospital’s facilities and services with your doctor. Many tests and treatments can be done in your home state or one nearby; travel to distant hospitals is seldom needed. If possible, take location into consideration when deciding which hospital will treat your child because you may have to make several trips there or you might end up there for longer than you had anticipated.

Incidence of congenital heart defects

Most people are unaware that up to 1.3 million American alive today have some form of congenital heart defect and about one-half of these individuals are less than 25 years of age.

At least nine of every 1,000 infants born a year have a heart defect. That's almost one percent of live-born infants. Sometimes the defect is so mild that there are no outward symptoms. In other cases, it's so severe that the newborn becomes ill soon after birth. In still other cases, signs and symptoms occur only in later childhood.

In the United States, about 36,000 children are born with a heart defect each year. Most of these children can be helped by surgery even if the defect is severe.

How congenital heart defects develop

You may wonder why your child has a congenital heart defect. Congenital heart defects do not occur in a majority of children, and when they do occur, often there is no known cause. Because of this, in most cases there is nothing the parents could have done differently to prevent the heart defect. Congenital heart defects strike without regard to socio-economic status, race or cultural background. Don't feel it's your fault that your child has this problem.

Heart defects, especially those occurring as isolated problems in otherwise healthy infants, generally don't have readily identifiable genetic or environmental causes. Some error occurs early in the formation and growth of the heart when the fetus is developing that causes the defect.

Several genetic syndromes (such as Down, DiGeorge, Williams, Alagille, and Noonan syndromes) have cardiac malformations as well as defects in other organ systems. In some cases, a gene defect has been identified that causes the syndrome. For instance, in Down syndrome there is an extra chromosome 21 in each cell of the body (trisomy 21). In other syndromes, a genetic cause has not yet been established. These facts all point to a genetic component to the occurrence of congenital heart defects.

Numerous maternal environmental exposures early in pregnancy are also associated with congenital heart defects. These include the use of some medications for seizures, acne or bipolar psychiatric disorder, or having rubella (German measles), other viral diseases, or maternal conditions such as phenylketonuria and diabetes. Alcohol abuse and street drugs, as well as exposure to certain chemicals, have also been associated with heart defects, too.

Depending on the defect, the chances of a sibling also being born with the same or another heart defect are about 2 to 3 percent. While this is higher than the general population rate of about 1 percent, it's still a relatively small chance. Although exact causes for most congenital defect aren't known, remaining healthy and avoiding certain exposures may be helpful. If you or your partner does become pregnant again, tell your obstetrician about your other child. Fetal echocardiograms (ultrasounds) may help detect problems as early as mid-pregnancy.