Genetic Counseling for Adults with Congenital Heart Defects

When a child is born with a congenital heart defect, two of the most common questions are: “Why did this occur?” and “Will it happen again in our family?” These same questions come up when a person with congenital heart disease considers having children. Genetic counseling can help answer these questions and address your concerns about starting a family.

What happens in genetic counseling?

The goal of genetic counseling for adults with congenital heart disease is to determine the likelihood that their children will also have a heart defect. The chance for a heart defect to happen again in a family depends on its cause.

In your genetics evaluation the genetic counselor and/or geneticist will try to determine the cause of your heart defect by looking at your medical and family history and doing a physical exam. Genetic testing on a sample of your blood may also help determine the cause. Sometimes it’s helpful to obtain blood samples for genetic testing on other members of your family, especially if they also have a heart condition.

Causes of heart defects

Unknown cause: We don’t know the exact cause of most heart defects. They’re likely due to a combination of multiple genetic and environmental factors. There’s usually a 2 to 15 percent chance of a heart defect happening again in the family. The odds depend on what type of defect you have and whether anyone else in your family has a heart defect.

Genetic syndrome: Some people with congenital heart defects have a specific genetic condition that can include other health problems. They may or may not know that they have such a condition. The chance for their child to also have this condition can be as high as 50 percent. These conditions can vary widely in their severity, so children may have less serious or more serious health problems than their parents.

Single gene: Rarely, congenital heart defects are caused by changes in a single gene. Often when this is the case more than one person in the family has a heart defect. The chance for another family member to have a heart defect can be as high as 50 percent.
Environmental exposure: Heart defects can also be caused by something your mother was exposed to in her pregnancy with you, such as an infection or a drug. In this case, the chance that your children will have heart defects is no higher than that of the average person.

Who should have genetic counseling?

Genetic counseling can be helpful for anyone with a heart defect who wants to know the cause of it or the chance that their children will have a heart defect. Genetic counseling is relevant for both men and women with congenital heart defects. The chance for children to have a heart defect rises when either the mother or the father has a heart defect.

Genetic counseling is particularly important if others in your family have heart defects, or if you or someone in your family has other birth defects, deafness, psychiatric conditions, liver disease or learning problems.

When is the best time to have genetic counseling?

The best time to have genetic counseling is before you get pregnant. That way you'll know the risks before you become pregnant. You'll also know if any special testing is needed in the pregnancy. If you become pregnant and haven't had genetic counseling yet, it can still be helpful to get it during the pregnancy.

During the pregnancy

If a genetic cause is found for your heart defect, you may be able to do genetic testing during the pregnancy to see if the baby inherited that genetic condition. In most cases, it's a good idea to have a fetal echocardiogram done by a pediatric cardiologist who is an expert in imaging congenital heart disease.

Taking part in research

There's still a lot that we don't know about why heart defects happen. It's possible that future research will discover these causes. As an adult with congenital heart defects, you may be able to help improve our understanding by taking part in research. This could help your family and other families better understand their heart defect and the chance it will occur again in the family. Ask your genetic counselor about research studies you could take part in.