



Genetic Fact Sheets for Parents Other Disorders

Screening, Technology, and Research in Genetics is a multi-state project to improve information about the financial, ethical, legal, and social issues surrounding expanded newborn screening and genetic testing – [http:// www.newbornscreening.info](http://www.newbornscreening.info)

Disorder name: Critical Congenital Heart Disease
Acronym: CCHD

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This fact sheet has general information about Critical Congenital Heart Disease (CCHD). Every child is different and some of this information may not apply to your child specifically. Certain treatments may be recommended for some children but not others. If you have specific questions about CCHD and available treatments, you should contact your doctor.

What is newborn screening for Critical Congenital Heart Disease (CCHD)?

Newborn Screening

Newborn screening for CCHD by pulse oximetry is now done in all states in the United States.

What is Pulse Oximetry?

Pulse oximetry is a simple, quick, and painless test used to help find babies who may have a CCHD. A pulse oximeter is a small device that measures a baby's pulse and how much oxygen a baby has in his or her blood (called oxygen saturation). Babies who have low oxygen levels in their blood may have a CCHD, but they may also have a different cause for low oxygen saturation that is not a CCHD.

Why is pulse oximetry used to screen for CCHD?

Pulse oximetry is used to screen for CCHD because it can be easily done soon after a baby is born, and it is the best way to detect a CCHD along with the pediatrician's physical exam. This should be completed after a baby is 24 hours (1 day) old.

A physical exam alone does not always detect CCHD since some CCHDs do not show outward symptoms. Ultrasounds done during pregnancy can be used to check the fetal heart. However, ultrasounds during pregnancy do not always detect all CCHDs.

Confirmatory testing

If a baby does not pass their pulse oximetry screening or is suspected to have a CCHD, multiple pulse oximetry screenings may be completed before the baby is examined by the primary care provider/specialist at the hospital. A baby may also need additional testing such as an X-ray, an electrocardiogram (a test that records the heart's electrical activity), or an echocardiogram (an ultrasound of the heart). The doctors will discuss the follow up care for the baby and any additional testing the baby may need. The baby may be referred to a pediatric cardiologist.

What is CCHD?

Critical congenital heart disease (CCHD) describes a group of heart defects that can cause life-threatening problems which need to be treated within the first days or first year of life. CCHD can usually be treated if found early. CCHD is a birth defect and this may be the easiest way to describe it to family and friends as you learn the details of your own child's CCHD.

CCHD can be caused by different reasons including problems with the heart's structure or an abnormal heartbeat. The heart is a very complicated organ with many structural components. If specific structures of the heart do not form properly during pregnancy, the result is a heart defect. Critical Congenital Heart Defects have varied severity. Some are severe and need many heart surgeries, others are less severe and need one heart surgery or catheter intervention, and some are somewhere in between.

CCHD can involve problems with the chambers of the heart, holes in the heart, abnormal connections in the heart, and abnormalities in how the heart functions. Most congenital (from birth) heart conditions affect people from childhood through adulthood. Here are a few examples of CCHDs:

- Hypoplastic left heart syndrome
- Pulmonary atresia (with intact septum)
- Tetralogy of Fallot
- Total anomalous pulmonary venous return
- Transposition of the great arteries
- Tricuspid atresia
- Truncus arteriosus

Some babies may have a combination of these heart defects. In this case, ask your doctor to explain the unique nature of your child's heart defects. Remember that CCHD is as complex as the heart itself, and it may take time to fully understand your child's unique heart defects.

What causes CCHD?

In most cases, the cause of CCHD is unknown. It is thought that both genetic and environmental factors likely play roles in the cause of CCHD. Approximately 10% of these heart defects are due to changes in a baby's genes or chromosomes. A pregnant woman's exposure to environmental factors, such as drugs or chemicals, can also affect how the baby's heart develops. Finally, maternal conditions, such as diabetes during pregnancy, can affect the developing baby's heart. However, your baby's CCHD is not your fault, and as of today, there is no known way to prevent CCHD. You or your partner did not do anything wrong.

What are the symptoms of CCHD?

The symptoms of CCHD range from moderate to severe. They can cause minor problems in some newborns and more serious health problems in others. Symptoms can begin to appear in the newborn period. However, some babies born with a CCHD may be healthy for the first days or weeks of life and only later begin to show severe symptoms that require emergency care. This is because the infant heart works differently before birth compared to after birth when the child becomes dependent on the lungs. Normal changes to the way the baby's heart and lungs work after birth may lead to the symptoms and discovery of CCHD.

Newborn CCHD symptoms may include:

- blue color of the skin, lips, and nailbeds (cyanosis)
- rapid heartbeat
- poor feeding/sucking or feeding difficulties
- low birth weight or delayed weight gain
- excessive sweating (especially on the forehead)
- rapid breathing or shortness of breath
- fatigue, abnormal drowsiness, lethargy
- pale, cool, or clammy skin
- repeated respiratory infections

Even if treated correctly, some CCHDs can restrict the person's ability to participate in sports or other high stress activities. Some individuals may have developmental delays, but most should be able to live a close to typical life. Many people with an isolated CCHD have finished college and have full-time jobs.

If left untreated, CCHD can cause serious chronic health problems that could lead to death at a young age. Many of the symptoms of CCHD can be controlled with medication and surgery. It is important that you see your doctor and follow a treatment plan tailored for your child's needs.

What are the treatments for CCHD?

Babies born with a CCHD should be seen by a pediatric cardiologist and receive special care and treatment. Treatments can prevent death or disability early in life. The type of treatment recommended will depend on the type of CCHD that the baby has. For some children, special care may be needed throughout life. Sometimes, babies who have more than one medical problem may need additional treatments for their other medical problems.

What happens when CCHD is treated?

Most children with CCHD will survive into adulthood following successful treatment and can lead normal or nearly normal lives. Exercise and other activities may be limited. Some children with CCHD may have developmental delays or learning difficulties.

Is CCHD inherited?

Whether the CCHD is inherited or can happen again in your family depends on the cause of the CCHD:

Unknown cause: The exact cause of most CCHDs is not known. It is thought that genetics plays a role. Only a few genes have been linked to CCHDs. CCHDs are most likely due to both genetic and environmental factors.

If CCHD is present in your family, there is an increased chance of having a child with a CCHD. However, this chance depends on the type of CCHD in your family and the number of family members who have a CCHD.

Genetic syndromes: Sometimes a baby's CCHD is caused by a genetic syndrome that may include other health problems. These other health problems may or may not be obvious at birth or in infancy. Genetic syndromes can vary in their severity. As a result, people may have less serious or more serious health problems than other family members with the same genetic syndrome. In these families, the chance for another family member to have a CCHD can be as high as 50%.

Single gene: CCHD can be caused by changes in a single gene. Usually in these families, more than one person has a CCHD. In these families, the chance for another family member to have a heart defect can be as high as 50%.

Environmental exposure: CCHD can sometimes be caused by something the mother was exposed to in her pregnancy, such as an infection or certain drugs. If this is the case, the chance for another family member to have a heart defect may be low.

Is genetic testing available?

Genetic counseling is available to families or individuals with a CCHD. Genetic counselors and geneticists review medical information about the family and pregnancy of the person with the CCHD. They also do a physical exam of the person to try to determine the cause of the CCHD. Genetic testing may be offered to help figure out the cause. Sometimes more than one family member may be tested.

Can you screen for CCHD during a future pregnancy?

CCHD may be diagnosed before a baby is born. Fetal ultrasound and fetal echocardiograms are tools that can be used to screen for CCHD during pregnancy. An ultrasound done at 18-22 weeks of pregnancy can detect some CCHDs. A fetal echocardiogram can be done at 20-22 weeks of pregnancy to give a picture of the chambers of the heart. Unfortunately, it is not possible to detect 100% of CCHDs using routine ultrasound and fetal echocardiograms.

If a genetic cause has been identified in a child with a CCHD, DNA testing can be done before or during future pregnancies. Parents may also choose to use assisted reproductive techniques to decrease the chance that their future children would have CCHD.

During pregnancy, DNA testing can be done on a sample obtained by either CVS or amniocentesis. Parents may choose to have DNA testing before or during

pregnancy or wait until birth to have the baby tested. A genetic counselor can talk to you about your choices and answer questions about prenatal testing or testing your baby after birth.

How many infants are born with CCHD?

About 2 in every 1,000 babies in the United States are born with CCHD. CCHD occurs in all ethnic backgrounds.

Can I prevent CCHD while pregnant?

Since little is known about what causes CCHD, there is no certain way to avoid having a baby with CCHD. However, if you are female and are pregnant or planning to become pregnant, you can reduce the risk by:

- Getting vaccinated against rubella and the flu.
- Taking at least 400 micrograms of a folic acid supplement every day during your pregnancy. Research shows that this will lower the risk of giving birth to a child with CCHD among other types of birth defects.
- Talking with a health care provider about medications during pregnancy, including herbal remedies and over the counter medication.
- Trying to avoid contact with people who are known to have an infection.
- Trying to avoid exposure to chemicals, such as those used in dry cleaning, paint thinners, and nail polish remover.
- Avoiding alcohol, smoking, and street drugs.
- Maintaining a healthy diet with good blood sugar control

Does CCHD go by any other names?

CCHD is also sometimes called:

- Critical Cyanotic Congenital Heart Defect
- Cyanotic Heart Defect
- Congenital Heart Defect

Where can I find more information?

While in the hospital you can:

- Ask the hospital social worker for emotional support.
- Ask your primary care provider, nurses, or pediatric cardiologist for additional information and ways to talk with your family about your baby's CCHD.
- Ask the social worker if your hospital has a support group.
- Ask the social worker about other families in your area who have a child or children with a CCHD.

- Talk to the hospital's lactation consultant and social worker about your options for breastfeeding and storing breast milk while your baby is staying in the hospital or having a procedure.

For additional information and support, you can follow the links below:

Baby's First Test

<http://www.babysfirsttest.org/newborn-screening/conditions/critical-congenital-heart-disease-cchd>

Center for Disease Control and Prevention

<http://www.cdc.gov/ncbddd/heartdefects/index.html>

March of Dimes

<https://www.marchofdimes.org/find-support/topics/planning-baby/congenital-heart-defects-and-critical-chds>

National Heart, Lung, and Blood Institute

<http://www.nhlbi.nih.gov/health/health-topics/topics/chd/>

Children's Heart Association

<https://heartchild.info/web/>

The Children's Heart Foundation

<http://www.childrensheartfoundation.org/>

MedlinePlus

<https://medlineplus.gov/genetics/condition/critical-congenital-heart-disease/>

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