
Newborn Screening for Critical Congenital Heart Disease

A Summary of the Evidence and Advisory Committee
Decision

Report Date: 03 September 2010



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EXECUTIVE SUMMARY

This summary reviews the information the federal advisory committee used when deciding whether to recommend adding Critical Congenital Heart Disease (CCHD) to the Recommended Uniform Screening Panel (RUSP) in 2010.

About the condition

CCHD is a group of the most serious heart disorders present at birth. About 200 out of every 100,000 babies have CCHD. Children with CCHD are born with a wide range of problems with the heart's structure. These problems prevent the heart from pumping blood normally to the lungs and rest of the body. Because blood carries oxygen, parts of the body may not get enough oxygen. There are many types of CCHD, and all can cause serious health problems. These problems can worsen quickly and cause death in early childhood. Babies with CCHD need treatment early in life.

Treatment for CCHD

Babies with CCHD need treatment early in life. Many babies need surgery right away. Other babies need medicines or other procedures to help with blood flow or heart problems. The type of treatment a baby needs depends on the baby's heart problem and symptoms.

Detecting CCHD in newborns

CCHD may be found before birth with a prenatal ultrasound. After birth, newborn CCHD screening usually happens when a baby is between 24 and 48 hours old. Newborn CCHD screening uses a device called a pulse oximeter to measure oxygen in the baby's blood. Newborns with low blood oxygen are at higher risk for CCHD. They need more tests to diagnose the condition.

Public health impact

Detailed data on how newborn CCHD screening would affect public health were not available at the time of the report. However, newborn screening for CCHD saves lives by allowing diagnosis early in life. Babies who get a diagnosis before leaving the hospital may avoid both life-threatening symptoms and treatment delays that can cause death.

Committee decision

The Committee voted in 2010 to recommend adding CCHD to the RUSP. As of 2011, the RUSP recommends that state newborn screening programs include CCHD.

ABOUT THIS SUMMARY

What is newborn screening?

Newborn screening is a public health service that can change a baby's life. Newborn screening involves checking all babies to identify those few who look healthy but who are at risk for one of several serious health conditions that benefit from early treatment.

Certain serious illnesses can be present even when a baby looks healthy. If the baby does not receive screening for these illnesses early in life, a diagnosis may be delayed. Later treatment might not work as well as earlier treatment. Newborn screening programs have saved the lives and improved the health of thousands of babies in the United States (US).

Who decides what screening newborns receive?

In the US, each state decides which conditions to include in its newborn screening program. To help states determine which conditions to include, the US Secretary of Health and Human Services provides a list of conditions recommended for screening. This list is called the Recommended Uniform Screening Panel (RUSP).

Progress in screening and medical treatments can lead to new opportunities for newborn screening. To learn how a condition is added to the RUSP, see **Box A**.

What will this summary tell me?

In 2010, the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) requested an evidence review of newborn screening for Critical Congenital Heart Disease (CCHD). This summary presents key review information that the Committee used to make its decision about whether to recommend adding CCHD to the RUSP. It will answer these questions:

- [What is CCHD?](#)
- [How is CCHD treated?](#)
- [How are newborns screened for CCHD?](#)
- [Does early diagnosis or treatment help patients with CCHD?](#)
- [What is the public health impact of newborn CCHD screening in the US?](#)
- [What did the Committee decide about adding CCHD to the RUSP?](#)

Box A: Adding a Condition to the RUSP

A committee, called the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC), makes a recommendation to the US Secretary of Health and Human Services about adding specific conditions to the RUSP. The Committee bases its decision on a review of the condition, the screen, the treatment, and the ability of newborn screening programs to check for the condition. To learn more about the ACHDNC, visit this [website](#).

UNDERSTANDING THE CONDITION

What is CCHD?

CCHD is a group of the most serious heart disorders present at birth. Children with CCHD are born with a wide range of problems with the heart's structure. Most of the time, the cause of these problems is not known. These problems prevent the heart from pumping blood normally to the lungs and rest of the body. Because blood carries oxygen, parts of the body may not get enough oxygen. Therefore, babies with CCHD need treatment early in life to help oxygen-rich blood reach the body. Treatment can lower the risk of serious illness or death.

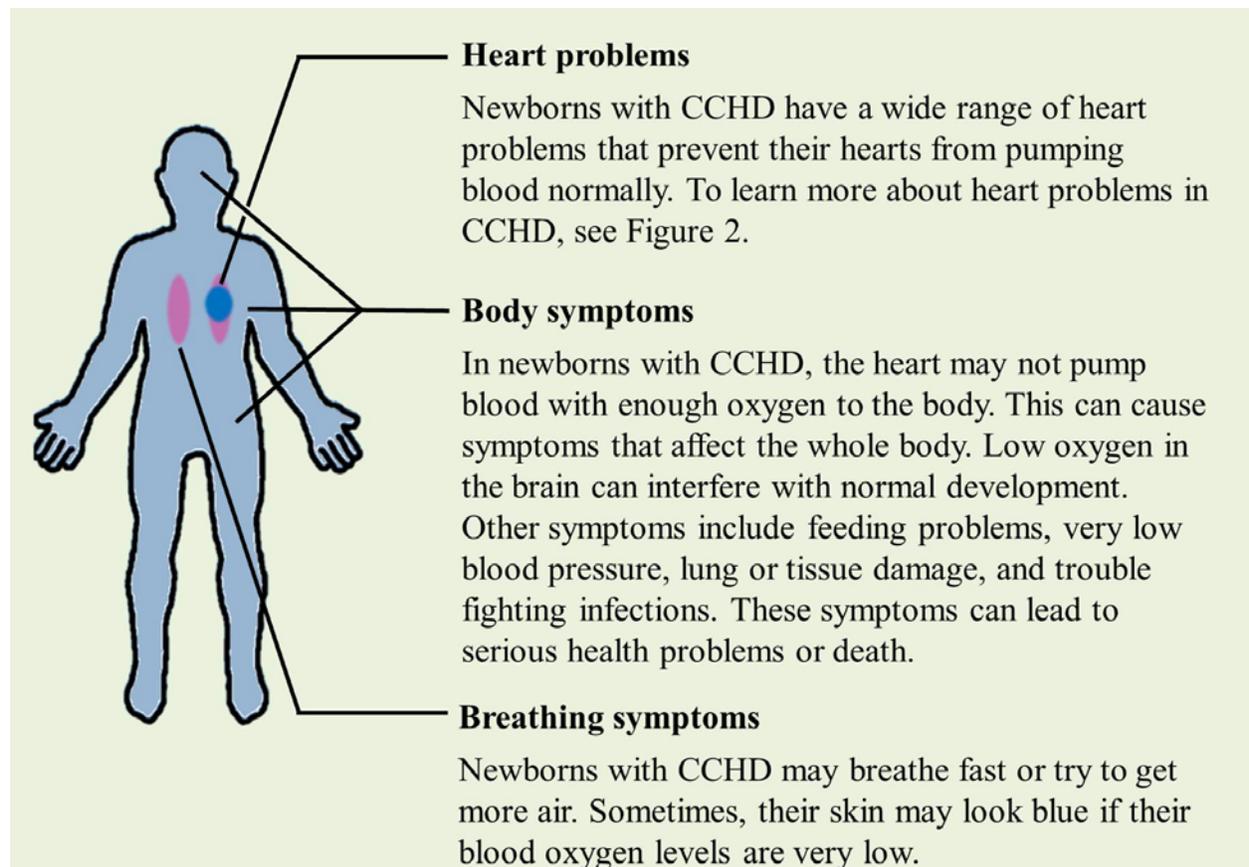
How common is CCHD?

- About 200 out of every 100,000 children have CCHD.

What kinds of health problems does CCHD cause?

Babies with CCHD have problems with the heart. CCHD can also cause other symptoms that affect other parts of the body (Figure 1).

Figure 1: CCHD Symptoms.



Are there different types of CCHD?

Yes. Doctors classify CCHD into many types based on the structure of the heart. To learn more about key parts of the heart that may have problems in CCHD, see Figure 2. In most types of CCHD, blood flowing to the body does not have enough oxygen.

All types of CCHD can cause serious health problems. All types require treatment early in life. Early detection is crucial for all types of CCHD.

Figure 2: Key Parts of the Heart.

Blood vessels

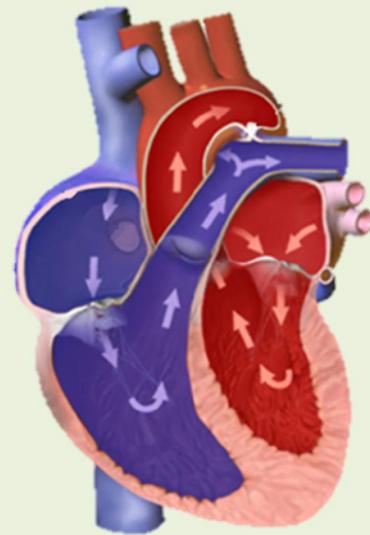
Some babies with CCHD have problems with the blood vessels leading to or away from the heart. These blood vessels may be missing, formed incorrectly, or connected to the wrong places.

Heart valves

Some babies with CCHD have problems with the valves that control blood flow through the heart. These valves may be missing or formed incorrectly. This means that blood cannot flow properly through the heart and lungs.

Areas of the heart

Some babies with CCHD have problems with specific areas of the heart. Parts of the heart may develop incorrectly or incompletely. This prevents the heart from pumping properly.



Note: This picture shows a normal infant heart. In CCHD, the heart may look different in many ways.

When do CCHD symptoms develop?

Babies are born with CCHD. However, many babies with CCHD have no symptoms at birth. These babies may look healthy at first, but many develop symptoms within the first days or weeks of life. Therefore, parents and doctors should monitor newborns for the CCHD symptoms shown in Figure 1.

Symptoms of CCHD can worsen quickly. Babies who have symptoms at home may not get care in time to prevent death. Some babies do not receive a diagnosis until after death.

Even with treatment, CCHD can cause health problems throughout life.

TREATMENT FOR CCHD

How is CCHD treated?

Babies with CCHD need surgery or other health care early in life. The type of care a baby needs depends on the baby's heart problem and symptoms.

Many babies need surgery right away, in the first few days or weeks of life. Some need multiple surgeries over several years for their CCHD problems.

Other babies do not need surgery right away. They may receive medicine or other procedures. Some need a medicine called 'prostaglandin.' Others may need a procedure in which a doctor threads a long, thin tube (catheter) through a blood vessel into the heart. These treatments can help with blood flow and heart problems early in life.

What are the risks of treatment for CCHD?

Surgeries for CCHD are serious procedures that require special surgeons, staff, and equipment. The risks of surgery depend on the baby's heart problem, the type of surgery, and other things. Surgeries may cause other problems or require lifelong treatment. The survival rate for babies having CCHD surgery is over 95%.

Medicines and other procedures for CCHD also have risks, like bleeding, infections, or other side effects. Because of these risks, doctors monitor babies who need medicines or other procedures for CCHD carefully. Doctors must understand these risks, along with the special features of CCHD, and work closely with families to best manage a child's care.

Box B: Where Can I Learn More?

Follow the links below to learn more about topics from this summary.

- To learn more about CCHD, visit these websites:
 - [National Institutes of Health CCHD](#) website.
 - [Centers for Disease Control and Prevention Hearing Loss and CCHD](#) website.
- Visit the Committee's website to learn more about:
 - [Nominating conditions to the RUSP.](#)
 - [Full CCHD evidence report.](#)
 - [The ACHDNC recommendation to the Secretary to add CCHD to the RUSP.](#)

FINDING NEWBORNS WHO HAVE CCHD

How are newborns screened for CCHD?

Screening for CCHD is done separately from blood spot-based screening for other disorders. Sometimes, CCHD is found before birth with a prenatal ultrasound. After birth, newborn screening for CCHD usually happens when a baby is between 24 and 48 hours old.

To screen for CCHD, a doctor or nurse uses a device called a pulse oximeter. The device has a sensor that attaches to a baby's right hand and either foot. It shines light through the skin to measure oxygen in the blood. The screening process is painless and happens at the bedside or in the nursery.

When a newborn has blood oxygen levels that differ from established guidelines (eg, levels that are too low or that differ in the hand and foot), the baby may need more tests. The baby's doctor will determine if the baby needs more tests or to see a specialist to tell for sure if the baby has CCHD.

How well does screening for CCHD work?

Screening detects babies with low blood oxygen. After more tests, some of these babies receive a diagnosis of CCHD. Many others do not have CCHD but may have other serious health problems, like lung infections or other problems, that also lower blood oxygen. Screening cannot diagnose CCHD, but it finds babies who need more tests or to see a specialist.

Along with ultrasounds before birth and physical exams, screening detects most babies with CCHD. However, screening may miss a small number of newborns with the disease. That can happen for a few reasons, like if screening was done very early or the baby did not have very low blood oxygen at the time of screening. Parents and doctors should watch for CCHD symptoms in newborns, even if screening results are normal.

What happens if newborn screening indicates a high risk for CCHD?

If a baby has low blood oxygen, the baby needs a doctor's exam to see why the oxygen levels are low. If the baby's doctor thinks the baby might have CCHD, the baby may need an echocardiogram. This test is an ultrasound of the heart. It takes movies of the heart and shows the heart's blood flow. It is noninvasive and uses no radiation. Babies with low blood oxygen might also need other tests, like a chest X-ray or blood tests that measure oxygen or check for infections.

Low blood oxygen is a sign that a baby may have a serious health problem, so babies need these extra tests before they can go home. Sometimes, babies may need to go to another hospital to get the tests they need. Babies should not be discharged until the reason for their low blood oxygen levels is known.

What are some of the benefits and risks of newborn CCHD screening?

Table 1 describes the benefits and risks of newborn CCHD screening as of 2010.

Table 1: Benefits and Risks of Screening.

Benefits	Risks
<ul style="list-style-type: none">● Earlier diagnosis.	<ul style="list-style-type: none">● Some babies detected from screening do not have CCHD. All babies with low blood oxygen need more tests.● Follow-up tests may disrupt parent/child bonding.
<ul style="list-style-type: none">● Earlier monitoring and treatment, which lowers the risk of early death.	<ul style="list-style-type: none">● More anxiety about the future.
<ul style="list-style-type: none">● Detection of other serious health problems that lower blood oxygen.	<ul style="list-style-type: none">● A small number of babies with normal blood oxygen during screening may have CCHD. Screening offers false reassurance in these cases.

Do early diagnosis and treatment help patients with CCHD?

Yes. Early diagnosis and treatment of CCHD greatly improve a baby's chance of survival.

PUBLIC HEALTH IMPACT

How would newborn CCHD screening affect the health of the country?

Detailed data on how newborn CCHD screening would affect public health in the US were not available at the time of the report.

However, newborn screening for CCHD saves lives by allowing diagnosis early in life. Babies who get a diagnosis before leaving the hospital may avoid both life-threatening symptoms and treatment delays that can cause death.

What is the status of newborn CCHD screening in the US?

- In 2011, 3 states (Indiana, Maryland, and New Jersey) had laws mandating newborn screening for CCHD.
- To learn when and how states began CCHD screening, read this [report](#).

ADVISORY COMMITTEE DECISION

What did the Committee recommend?

The ACHDNC voted in 2010 to recommend adding CCHD to the RUSP. The Committee based its decision on the ability of screening to find babies with CCHD and early treatment being better than later treatment. In 2011, the US Secretary of Health and Human Services recommended that all newborns receive CCHD screening.

HELPFUL INFORMATION

Glossary

Term	Definition
ACHDNC	<u>A</u> dvisory <u>C</u> ommittee on <u>H</u> eritable <u>D</u> isorders in <u>N</u> ewborns and <u>C</u> hildren. The committee that oversees the RUSP.
Catheter	A long, thin tube used in some CCHD procedures.
CCHD	<u>C</u> ritical <u>C</u> ongenital <u>H</u> ear <u>D</u> isease. A group of the most serious heart disorders present at birth.
Echocardiogram	An ultrasound of the heart. It takes movies of the heart and shows the heart's blood flow.
Prostaglandin	A medicine that can help with blood flow early in life.
Pulse oximeter	A device that uses a special sensor to measure oxygen in the blood.
RUSP	<u>R</u> ecommended <u>U</u> niform <u>S</u> creening <u>P</u> anel. The list of conditions recommended for newborn screening.
Secretary of Health and Human Services	The head of the US Department of Health and Human Services. This person decides whether to add conditions to the RUSP.
Specialist	A doctor with expertise in a specific area of medicine.

Source

The information in this summary comes from the report *Evidence Review: Critical Congenital Cyanotic Heart Disease* (03 September 2010), commissioned by the ACHDNC. The report reviewed data on CCHD screening and treatments in children through June 2010. It included both published and unpublished research. To see a copy of the report, visit this [page](#).