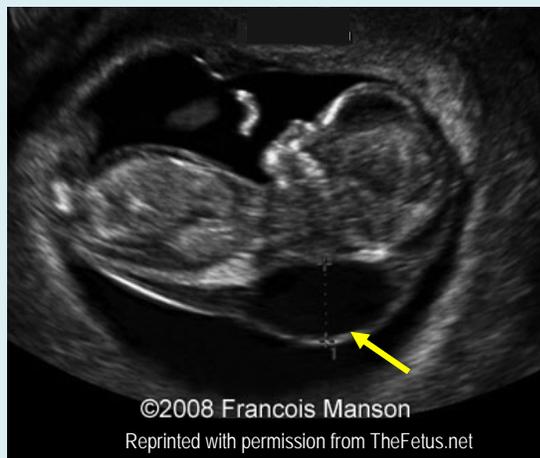


PRENATAL ULTRASOUND FINDINGS

What is a Cystic Hygroma?

A cystic hygroma is a large fluid-filled area found at the back of the neck in a developing baby. This ultrasound finding is seen in fewer than 1 out of 100 pregnancies. It is almost always detected before the 20th week of pregnancy.



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How does a cystic hygroma happen?

A cystic hygroma happens when the baby's lymph system has not developed in the usual way. This causes fluid to build-up in two lymph sacs located at the back of the neck and sometimes in other areas of the body.

What is the lymph system?

Lymph is a clear fluid that makes up part of the blood. This fluid naturally moves out of the blood into the cells of the body. The lymph system helps the fluid return to the blood.

How does the lymph system develop?

There are two lymph sacs at the back of the fetal neck that form around 7 to 8 weeks after your last menstrual period. These two sacs collect lymph fluid from the body of the developing baby. The sacs eventually connect with blood vessels, so lymph fluid can return to the blood. If this connection does

not happen at the usual time, fluid starts to build up in the lymph sacs and forms a cystic hygroma. Lymph fluid can also build-up in the body of the developing baby. This is called hydrops.

Can a cystic hygroma cause problems for the baby?

Cystic hygroma is often the sign of a problem in a developing baby. There are a number of different conditions that can be found in babies with this ultrasound finding, including:

Chromosome conditions: About half of babies with a cystic hygroma have a chromosome condition. The most common chromosome conditions seen with this finding are Turner syndrome, Down syndrome, and trisomy 18.

Chromosomes are the packages of genetic information found in the cells of the body. Normally, there is a set of 46 chromosomes in every cell. Extra or missing chromosome material almost always causes intellectual disabilities and physical differences. Chromosome conditions can range from relatively mild to very severe.

Birth defects: Babies with a cystic hygroma are also more likely to have birth defects in other parts of the body. Heart defects are the most common type of birth defect found, but many different types of birth defects have been reported.

Genetic syndromes: There are many different genetic syndromes that can cause a baby to have a cystic hygroma. Most of these conditions are rare and are often difficult to diagnose before a baby is born.

Fetal loss (miscarriage/fetal demise/stillbirth): If a connection between the lymph system and the blood vessels does not form at some point, extreme fluid build-up (hydrops) in the developing baby can lead to the loss of the pregnancy.

Are more tests needed?

When a cystic hygroma is seen, additional tests are offered to learn more about the pregnancy.

You may be offered one or more of these tests:

Chromosome testing

- Prenatal diagnostic testing - Either CVS or amniocentesis test chromosomes directly from the pregnancy. These tests give you the most complete information about chromosome conditions in the baby. There is a very small risk for miscarriage associated with these procedures (about 1 in 1000).
- Cell-free DNA screening - A blood test from the pregnant person can identify pregnancies that are more likely to have common chromosome conditions. This test is sometimes called non-invasive prenatal testing or NIPT. If the screening test shows a high chance for a chromosome condition, a diagnostic test is still needed to find out for sure. A screening test will miss some causes of cystic hygroma that could be found by diagnostic testing.

Ultrasound

- Level 2 ultrasound - A level 2 (detailed) ultrasound is done to carefully look at the baby. Some physical birth defects can be found by ultrasound; however, many birth defects are difficult to find during pregnancy.
- Fetal cardiac ultrasound - If there is concern about a possible heart defect, a special ultrasound of the baby's heart may be arranged.
- Follow-up ultrasounds - Your doctor may recommend follow-up ultrasounds to watch for changes in the amount of fluid build-up. Extreme fluid build-up in the pregnancy may lead to health concerns for both the developing baby and the pregnant person.

Will the cystic hygroma go away?

Sometimes the cystic hygroma goes away (resolves). It is more likely to go away when the cystic hygroma is small and seen before 14 weeks gestation.

When a cystic hygroma goes away, the developing baby's chance for survival improves. However, a cystic hygroma that disappears does not provide reassurance about chromosome conditions, birth defects, or genetic syndromes. A cystic hygroma can go away even when the developing baby has Down syndrome, Turner syndrome, or another medical condition.

What if all the follow-up tests are reassuring?

Babies have been born healthy after finding a cystic hygroma. In these cases, the chromosome studies were normal, there were no other birth defects seen on ultrasound, and the cystic hygroma went away. However, because there are many different reasons for a cystic hygroma to form, normal follow-up tests during pregnancy cannot guarantee your baby will be healthy at birth.

Where can I get more information?

You can speak with your doctor, nurse practitioner, nurse midwife, or a genetic counselor if you have additional questions about this ultrasound finding.

Kaiser Genetics Departments

Website: <http://genetics.kp.org/>

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| Fresno | (559) 324-5330 |
| Modesto | (916) 614-4075 |
| Oakland | (510) 752-6298 |
| Sacramento | (916) 614-4075 |
| San Francisco | (415) 833-2998 |
| San Jose | (408) 972-3300 |