Patient Information Sheet
Second Trimester Cystic Hygroma

You have recently learned that your baby has a cystic hygroma. We expect that you may have questions about what this could mean for your developing baby. Hopefully, this information will address some of your concerns; however, we encourage you to speak to your genetic counselor or healthcare provider for more information. It is important to remember that anyone can have a baby with a birth defect. Birth defects occur in 2-5% of all births and are rarely caused by something that the parents did or did not do before or during pregnancy. Currently, there are no tests available to detect all health problems.

What is a cystic hygroma?
A cystic hygroma is a fluid filled sac (called a cyst) that can develop in the baby. Most often these cysts develop in the neck but they can also occur in other areas of the body. The fluid inside the cyst is called lymph fluid because it comes from the lymphatic system, which is involved in defending our bodies against infection.

What causes a cystic hygroma?
A cystic hygroma is caused by problems in the formation of the lymphatic system. Early during the development of the baby, vessels form to carry the lymph fluid. These vessels connect with the blood vessels in the neck. A cystic hygroma can develop when the lymph vessels do not connect properly or when the connection is delayed. This prevents drainage of the lymph fluid. Without proper drainage, the fluid builds up and causes a cystic hygroma. Cystic hygromas can be many different sizes and may appear to be a collection of many fluid-filled cysts.

How many babies are found with a cystic hygroma?
Cystic hygromas are believed to occur in 0.2% to 1% of all pregnancies.

What problems could be associated with a cystic hygroma for my baby?
Sometimes, a cystic hygroma is seen on a prenatal ultrasound in a baby that will be healthy at birth. More often, though, a cystic hygroma is a sign of an underlying problem with the baby's development. A common cause of cystic hygroma is a chromosome problem. Other possible causes include heart defects and other rare genetic syndromes.

In order to understand a possible chromosomal cause or genetic syndrome cause, it is helpful to know a little about chromosomes. Chromosomes are packages of genetic information that carry the instructions (genes) necessary for our growth and development. Typically, there are 23 pairs of chromosomes in each cell of our bodies. One copy of each pair is from our mother and one copy of each pair is from our father. The first 22 pairs of chromosomes are the same in males and females. The last pair is known as the sex chromosomes and they determine our gender. Females typically have two X chromosomes and males usually have one X and one Y. It is important to have the correct amount of genetic material to have typical development. Sometimes there is an imbalance of genetic material that may cause birth defects. The imbalance may be caused by an extra or missing whole chromosome or sometimes only by extra or missing pieces of chromosomes. Some genetic conditions are caused by changes in one or more genes that result in a particular set of characteristics (also called a syndrome). A chromosome abnormality or genetic syndrome can produce birth defects and physical changes such as a cystic hygroma.

The most common chromosome problem associated with a cystic hygroma is Turner syndrome, though other chromosome abnormalities are possible as well. Individuals with Turner syndrome are missing a sex chromosome and, thus, they have a total of 45 chromosomes rather than the usual 46 chromosomes in the cells of their body. As a consequence, these individuals are females, typically with normal intelligence, who may be short in stature, infertile, and have other possible medical complications. The majority of babies with Turner syndrome are spontaneously miscarried during pregnancy or stillborn.

Another chromosomal issue associated with a cystic hygroma is Down syndrome. Down syndrome is caused by having an extra copy of chromosome #21. Individuals with Down syndrome often have characteristics in common including: a flattened facial appearance, up-slanting eyes, low muscle tone, and short stature. Individuals with Down syndrome typically have mild to moderate intellectual disability and are at an increased risk for medical problems, such as heart and bowel defects. Despite the challenges they may face, children with Turner syndrome and Down syndrome are more like other children than different. Your genetic counselor or health care provider can give you more information about these conditions if you wish.

When a baby has a cystic hygroma, there is a higher than usual chance for there to be abnormalities in other parts of the body. The most common birth defects in babies with cystic hygromas are heart defects. Some babies with a cystic hygroma will have extra

fluid in other parts of the body (called hydrops). This is usually a sign that the baby is not doing well. For many babies with cystic hygroma, the condition gradually worsens, and the baby may be stillborn.

What further testing may be offered and what will it tell me?

If the cystic hygroma is found during a routine ultrasound, then a more detailed ultrasound will usually be performed to look carefully at the baby's anatomy. A fetal echocardiogram may also be offered, which is a specialized ultrasound of the baby's heart. Screens and ultrasounds will only modify your chance of having a baby with a chromosome problem or genetic syndrome. They cannot diagnose or rule out the presence of a chromosomal problem or genetic syndrome.

Depending on your gestational age, two diagnostic testing options may be available: chorionic villus sampling (CVS) or amniocentesis. These are tests in which a small sample of placental tissue or amniotic fluid is obtained to examine the baby's chromosomes. Because these procedures are invasive, there is a risk, likely less than 1%, for complications that can lead to miscarriage.

If you choose to pursue diagnostic testing you may be offered a test called a chromosome microarray (CMA). CMA is used to identify small missing or extra pieces of the baby's chromosomes that may be associated with genetic conditions. CMA cannot detect all genetic conditions and may detect variations in the chromosomes that have uncertain clinical significance (VUS). Parental blood samples may help to clarify the meaning of a variation, but effects of these changes may not be known until after the baby is born. CMA may also detect information such as non-paternity and close relationships between parents.

If diagnostic testing is declined, there are several screening tests available in the first and second trimester. Screening tests can modify your risk for a chromosome abnormality, such as Down syndrome, and involve no risk of complications to your pregnancy. A screening test will not determine if your baby has Down syndrome, but it may help you decide whether or not you would like to pursue a diagnostic test.

Non-invasive prenatal testing, or NIPT, is a screening test for certain chromosome conditions. During pregnancy, some of your baby's chromosome material is in your blood, along with your own chromosomes. NIPT is a blood test that measures the amount of chromosome material in your blood to determine if your baby could have an extra chromosome, such as in Down syndrome where there is an extra 21st chromosome. This test also screens for two other more severe chromosome conditions, trisomy 13 and trisomy 18, and may screen for some less severe conditions that are caused by different numbers of the X or Y chromosome. NIPT is a highly accurate screen; however, it is not a diagnostic test. The detection rate for these chromosome conditions is typically between 90-99%.

Other screening blood tests (often called a triple, quad, penta, or first trimester screen) may also be offered. Your genetic counselor or health care provider can discuss these various screening options in more detail.

The risks, benefits and limitations of screening and diagnostic testing options should be discussed with you by your genetic counselor or other health care provider. Follow-up counseling and referrals for support can be made if a birth defect, chromosome condition, or other genetic condition is detected prenatally. As with all situations in which prenatal testing is discussed, it is your decision whether or not this test is done.

What do I do now?

This information is only intended as an introduction to some of the terms and tests that you have already heard or will be hearing about from your genetic counselor and healthcare provider. We hope that this information sheet will be helpful as you begin to understand more about cystic hygromas. We understand that any time something of concern is found through prenatal screening and testing, parents are going to be worried. Please don't hesitate to contact your genetic counselor with any questions or concerns you have. We are here to help you and your baby.

The decision to have one or more of these tests or to do no additional testing is a difficult and personal one. There is no one right decision. Some people will decide to have no further testing because they do not feel they need to know if their baby will have any of these conditions before delivery. Other people feel that they want more information as soon as possible. After gathering all of the information you need about cystic hygromas, you should make the decision that is right for you and your family. Please keep in mind that not all babies with a cystic hygroma will have the complications described in this information sheet. Though it is never possible to predict the long-term prognosis for a child with even an isolated cystic hygroma, some babies can be born without significant medical complications.