Cystic Hygroma

Patient Information Series – What you should know, what you should ask.

What is Cystic Hygroma?

Cystic Hygroma (CH) is a developmental abnormality of the lymphatic system visible and recognized by ultrasound from 11 weeks onwards. It is characterized by fluid-filled multiseptated spaces, located in the fetal neck (occipito-cervical region) and often accompanied by fetal skin edema or fluid collection in other fetal parts of the baby's body (hydrops, also known as "space suit"). This finding, increases the risk of chromosomal abnormalities, cardiac and non-cardiac malformations, and the chance that the baby may die before birth.

How does Cystic Hygroma happen?

The anomaly results from failure of the vessels carrying the lymph to connect with the main bloodstream vessels in the fetal neck. The lymph accumulates in large cavities in the baby's neck and at a later stage of the disease fluid can also accumulate in other fetal compartments such as skin, chest and belly. This is called hydrops fetalis. The anomaly is more often seen in babies with chromosomal anomaly that are more common in older women.

How are chromosomes relevant to a Cystic Hygroma?

CH is associated with abnormal chromosomes and genetic syndromes, which in turn increase the risk of miscarriage, fetal death and neonatal death. Abnormal chromosomes are present in 60% of cases. The most common chromosomal abnormalities are Trisomy 21 (Down's syndrome), Turner syndrome (XO), and Trisomy 18. Turner syndrome is very common when the nuchal and extra nuchal fluid accumulation is massive.

What congenital anomalies are associated with Cystic Hygroma?

Major body anomalies are found in 44% of fetuses with abnormal chromosomes and 13-29% in fetuses with normal chromosomes. Anomalies affecting the baby's heart are the most common congenital anomaly found, followed by urinary, central nervous system, skeletal, abdominal wall defects and more.

Should I have more tests done?

Additional tests are required for babies with CH to look for underlying causes. These tests help to rule out chromosomal abnormalities and anatomical/structural anomalies.

Tests to ask about include:

- **Chorionic villous sampling** (sample taken from the placenta), or an **amniocentesis** (sample of amniotic fluid surrounding the baby). The choice depends on the gestational age when the CH is diagnosed.
- **Detailed ultrasound examination** of the baby anatomy (advanced scan)
- **Fetal echocardiography**: A detailed evaluation of the baby's heart by ultrasound to rule out cardiac abnormalities.

- **MRI scan:** This may be helpful when the baby has several other anomalies or when the ultrasound examinations are particularly difficult.

Can cystic hygroma disappear?

If the genetic tests are normal and no other anomalies are found later in pregnancy, the fluid accumulation can sometimes resolve, especially if this was not too severe. Your doctor will be able to inform you if you require any further follow-up appointments.

What are the things to watch for during the pregnancy if the condition persists?

For ongoing pregnancies, a regular ultrasound at least every four weeks is required to rule out any fluid accumulation in the baby's body (hydropic changes), assess the amount of amniotic fluid, the growth of the baby and its overall health. In some countries a test of fetal wellbeing called a Biophysical Profile (BPP) can be offered after 32 weeks of gestation.

Where should I deliver the baby?

Your baby should be born in a center that offers more advanced pediatric care, if needed. For this reason, you will need to deliver in a specialized tertiary care hospital. It may be recommended that you give birth a few weeks before your due date (38 weeks) or even earlier if any concerns about your baby arise.

Will it happen again?

The recurrence mostly depends on the underlying cause. In cases with abnormal chromosomes, there may be a slightly increased risk of recurrence of the specific chromosomal abnormality. In cases of previous history of CH in the setting of a genetic syndrome the recurrence risk will depend on the inheritance pattern and type of genetic syndrome. Genetic and maternal fetal medicine counselling are recommended to discuss future pregnancies.

What other questions should I ask?

- Are there any other abnormalities associated with the Cystic Hygroma?
- How often will I have ultrasound examinations done?
- Would my baby benefit from surgery inside the womb?
- Where will the baby receive the best care after it is born?
- What does it mean for my baby after it is born?
- Can I meet the team of doctors that will be assisting my baby in advance?

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