Congenital Diaphragmatic Hernia or CDH

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

What is a CDH?

A CDH happens when the diaphragm, which is the muscle between the abdomen and the chest, does not develop completely. This results in a hole forming, through which, organs from the abdomen, (like the bowel, the stomach and sometimes the liver), go up inside the chest, where the lungs and the heart are.

How does a CDH happen?

It is not clear why a CDH occurs. It is a rare condition and happens in about 1 in 5000 babies. In most cases, a CDH occurs simply due to bad luck and not due to other factors. Almost half of the babies with a CDH will have other problems, most often with their heart or gut. In 2 out of 10 cases, there is a change in the number of chromosomes, and in 1 in 10 cases there will be a change in the information within the chromosomes themselves.

How are chromosomes relevant to a CDH?

Chromosomes are where most of our genetic information is kept. We usually have 46 of them matched in pairs: 23 come from one parent and the other 23 come from the other parent. For example, people with Down syndrome have an extra chromosome number 21, and people with CHARGE syndrome have a change in the information in one of the chromosome number 8. Both of those conditions are seen in babies with a CDH: if a change in information within the chromosomes occurs, such genetic disorders can arise and the disorder would be the cause of the CDH.

Should I have more tests done?

Many women will choose to have more tests done to know more about the condition of the baby. The tests available depend on where you are. Tests you should ask about include:

- an **amniocentesis** to look for problems with the number of chromosomes and some of the problems within the chromosomes. This is done by removing small amount of amniotic fluid surrounding the fetus.
- a **fetal echocardiography** - a specialized ultrasound of the heart of the baby during the pregnancy, which uses sound waves that “echo” off of the structures of the fetus' heart.
- If available, an **MRI scan** can sometimes be done to provide information on the condition of the baby. This scan uses strong magnetic fields and radio waves to create detailed images of the inside of the body.

What are the things to watch for during the pregnancy?

Babies with CDH are at risk of some problems during the pregnancy. That is why most specialists will recommend regular ultrasound examination at least every 4 weeks. The ultrasound will help identify if the baby is going into heart failure due to the pressure on the heart because of the other organs in the chest. Many women will also accumulate extra amniotic fluid around the baby. This condition is called **polyhydramnios**. It can stretch the uterus too much and cause

early labour well before the due date. Knowing this in advance, your doctor or midwife can help with decreasing the risk of an early birth.

What does it mean for my baby after it is born?

The organs that are developing in the chest are affected by the upward movement of the stomach organs, and this is the biggest problem for babies with CDH. The organ that is most often compromised is the lungs. Most babies with a CDH will have lungs that are smaller. If it is the case that the lungs are too small to allow for enough oxygen to enter the baby's body, the baby may die. Ultrasound and MRI (a different way to obtain images of a fetus inside the womb), can be used to try to predict whether the baby has a higher risk of having very small lungs.

Babies who also have had the information within their chromosomes altered have even more problems after being born. This will depend on the genetic disorder.

When the baby is stable, a surgeon will perform one or more surgeries to attempt to fix the hole in the diaphragm. The baby often requires very complex care in a specialized hospital for weeks after birth.

When they grow up, babies who have a CDH and survive have more problems with their lungs, heart, gut, muscles and brain. Babies who are more sick after being born have a higher risk of dying or being left with long term problems like being deaf, having learning difficulties or having problems with exercising.

Will it happen again?

If no genetic reason is found to explain the CDH, the risk of this happening again is less than 1 in a 100. If there is a genetic reason, this will determine the risk, and a consultation with a specialist may be helpful to help sorting this out.

What other questions should I ask?

- Does this look like a severe CDH?
- Is the liver in the chest?
- How big do the lungs seem to be?
- How often will I have ultrasound examinations done?
- Is surgery during the pregnancy available?
- Is my baby one that could benefit from surgery inside the womb?
- Where should I deliver?
- Where will the baby receive the best care after it is born?
- Can I meet the team of doctors that will be assisting my baby when it is born in advance?

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