Omphalocele

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

What is an Omphalocele?

An omphalocele is a birth problem in which there is opening in the abdominal wall where the umbilical cord or belly button is normally located. Intestines and sometimes other abdominal organs, including the liver, gallbladder, stomach and/or bladder-can come through the opening, which produces a mass in the middle of the abdomen. The mass is covered by a membrane, not by skin, and the umbilical cord enters the mass. This problem is called an omphalocele. The word omphalocele comes from the Greek Omphalos (umbilicus) and Cele (cavity). A baby can have an isolated omphalocele, meaning that that is the only problem the baby has, or omphalocele can be one of many problems the baby has. When the omphalocele is not isolated, this increases the likelihood that the baby has a genetic problem which explains why there is more than one problem. Most babies with omphalocele have small or medium sized omphaloceles, but some have very large ones called giant omphaloceles. A giant omphalocele is an omphalocele where the defect after birth is larger than 2 inches or 5 cm and contains a portion of the baby's liver. An omphalocele is relatively rare, and happens in 3.4 of each 10,000 pregnancies and 1-2 of each 10,000 live births.

How does an Omphalocele happen?

It is not completely clear how an omphalocele happens. Approximately 80% of the babies with an omphalocele will have other problems, most often with their heart, intestines or kidneys. Often when a baby has multiple abnormalities, there is a problem with the number of chromosomes that the baby has, and this explains all of the baby's problems. Some babies with an omphalocele will have a change in the number of chromosomes or a change in the information within the chromosomes themselves. Chromosomes are where most of our genetic information is kept. We usually have 46 of them. 23 come from one parent and the other 23 come from the other parent. Sometimes, a baby has too many or too few chromosomes. As an example, people with Down Syndrome have an extra chromosome number 21. People with trisomy 13 or 18 have an extra chromosome number 13 or 18. Trisomy 18 and 13 are commonly seen in babies with an omphalocele and other abnormalities.

Some babies will have multiple problems as part of a syndrome. The most common syndrome in babies with an omphalocele is called Beckwith Wiedemann syndrome. Babies with Beckwith-Wiedemann syndrome are typically large and have extra amniotic fluid (a condition known as polyhydramnios), and may have large livers, kidneys and tongues at birth. Sometimes, these abnormalities only become obvious later in pregnancy. The care of patients with this syndrome after birth includes surgical correction of the omphalocele and monitoring of low blood sugar in the neonatal period and screening for cancer during childhood.

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 live. Tests to ask about include a CVS (chorionic villus sampling) which involve removing and testing a small sample of cells from the placenta, or an amniocentesis (where a thin needle is used to take some of the fluid from the womb) to look for problems with the number of chromosomes and some of the problems within the chromosomes, which can be found in Beckwith-Wiedemann syndrome. You should also ask for an advanced fetal ultrasound scan to look for other organ abnormalities, with special attention for the heart, intestines and kidneys of the fetus.

What are the things to watch for during the pregnancy?

Extra ultrasound examinations to check to see if the baby is growing well and to check the amount of amniotic fluid are typically recommended. Evaluation of the size of the liver kidneys and tongue are typically recommended to screen for Beckwith-Wiedemann syndrome. If a really good ultrasound of the fetal heart cannot be done when the omphalocele is found, sometimes a special ultrasound of the baby's heart, called a fetal echocardiogram is recommended. These scans can help you and your doctor ~~to~~ make decisions about where you should delivery so that the baby receives the best care possible after birth. Most babies with an omphalocele can safely deliver vaginally and do not need caesarean section just because they have an omphalocele.

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

After birth the baby will be admitted to a special nursery. The paediatric doctors will make sure that the baby does not have breathing problems and will determine the best way for the baby to feed. If a chromosome or other genetic problem is suspected, a Genetics doctor will see and evaluate the baby and talk with you about further management. Additional tests, including a special heart ultrasound may be done. Unfortunately, in up to 1 of every three babies with omphalocele, additional problems are found. The surgeon will decide how best to fix the omphalocele. Sometimes, the abdominal wall can be closed shortly after birth, but in other cases, the omphalocele may be need to be closed slowly over time. This is called a delayed closure.

Babies with a giant omphalocele have an increased risk of underdevelopment of the lungs which can cause short- and long-term breathing problems, feeding problems, long hospital stays and even death.

When children with an omphalocele grow up, most of them develop normally. Babies who have had a giant omphalocele with a delayed closure after birth may show some delays in motor (muscle) development at the age of 2 years compared to babies with a small omphalocele and a primary closure. Follow-up and care by a specialized team which can include a pediatrician, pediatric surgeon, and a physical therapist may be helpful.

Will it happen again?

When no other genetic reason is found to explain the omphalocele, the risk of this happening again is low and estimated to be approximately 1 in a 100. If there is a genetic abnormality, then the risk depends on the type of genetic abnormality. Consultation with a Genetics doctor may be helpful.

What other questions should I ask?

- Which organs are outside the abdomen of the baby?
- Does my baby have a small or a giant omphalocele?
- Do you suspect any other abnormalities?
- Do you suspect that the baby has a chromosome problem or a genetic syndrome?
- Do you recommend that I have a chorionic villus sampling or an amniocentesis to detect chromosome abnormalities?
- Are there abnormalities that are impossible or very difficult to detect before the baby is born?
- How often should I have ultrasound examinations done?
- Where should I deliver so that the baby gets the best care after it is born?
- Do you think that I will be able to deliver vaginally or do you recommend a caesarean delivery?
- Can I meet the team of doctors in advance who will be looking after my baby when it is born?
- Are there common problems that can happen after birth which are hard to predict?
- Can I see the nursery where my baby is admitted after birth?
- Do you think the omphalocele will be able to be closed in one operation or will it take some time to close it?
- How long do you think my baby will need to be in the hospital after surgery?

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