Omphalocele

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

What is an Omphalocele?

An omphalocele, also known as exomphalos, is a birth defect characterized by an opening in the abdominal wall where the umbilical cord or belly button is normally located. This results in the protrusion of the intestines and sometimes other abdominal organs, such as the liver, gallbladder, stomach, and/or bladder, through the opening. The mass is covered by a membrane, not by skin, and the umbilical cord enters the mass. The term "omphalocele" is derived from Greek, with "Omphalos" meaning umbilicus and "Cele" meaning cavity. Babies with omphaloceles can have isolated cases or be part of a syndrome with multiple problems, which may suggest an underlying genetic issue. Most cases are small or medium-sized, but some are classified as giant omphaloceles, which are larger than 2 inches or 5 cm and contain a portion of the liver. Omphalocele is relatively rare with a reported prevalence of 3.38 per 10,000 pregnancies, approximately 1 in 4,200 live births in the United States。

How does an Omphalocele happen?

The exact cause of omphalocele is not fully understood. It occurs when the normal closure of the abdominal wall fails. It is often associated with other health problems, particularly in the heart, intestines, or kidneys, and chromosomal abnormalities. Approximately 80% of babies with an omphalocele have additional abnormalities, which may be related to the number or structure of their chromosomes. In the context of other structural abnormalities, 30%-70% of cases are associated with a chromosomal abnormality or multiple malformations syndromes. Common chromosomal conditions associated with omphalocele include trisomy 13, trisomy 18, and trisomy 21. Beckwith-Wiedemann syndrome is another condition frequently associated with omphalocele, characterized by large size at birth, extra amniotic fluid, and enlarged organs.

Should I have more tests done?

Prenatal testing can provide more information about the baby's condition. Tests may include chorionic villus sampling (CVS), amniocentesis, and advanced fetal ultrasound scans to detect chromosomal abnormalities and other organ malformation.

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 need to be closed slowly over time. This is called a delayed closure.

Babies with a giant omphalocele, which included more than 75% of the liver in sac and/or more than 5cm, 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?

The risk of recurrence without an identified genetic cause is low, approximately 1 in 100. If a genetic abnormality is present, the risk depends on the specific abnormality, and genetic counseling is recommended.

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?

Last updated December 2024