Omphalocele

What is omphalocele?

Omphalocele is a condition in which loops of intestines (and sometimes parts of the stomach, liver and other organs) protrude from the fetus' body through a hole in the abdominal wall. The hole is located at the belly button and is covered by a membrane, which provides some protection for the exteriorized organs. The umbilical cord inserts at the top of this membrane rather than on the abdomen itself. Omphalocele is often confused with gastroschisis, a similar condition in which the hole in the abdominal wall is located to the side (usually the left) of the umbilical cord. Omphaloceles come in all sizes: they may only contain one or two small loops of intestine and resemble an umbilical hernia, or they may be much larger and contain most of the liver. These are called "giant" omphalocele and are more difficult to treat.

How common is it?

Omphalocele occurs in approximately one in 5,000 births and is associated with other conditions and chromosomal anomalies in 50 percent of cases.

How is it diagnosed?

Omphalocele can be detected through ultrasound from 14 weeks of gestation; however, it is easier to diagnose as the pregnancy progresses and organs can be seen outside the abdomen protruding into the amniotic cavity. Because of the high risk of associated conditions, a prenatal test called an amniocentesis may be performed to help detect chromosomal and heart anomalies.

- ***Amniocentesis:*** Under ultrasound guidance, a fine needle is inserted through the abdomen into the uterus. A small amount of amniotic fluid is removed from the sac surrounding the fetus. The fluid contains cells that have been shed by the fetus, which can be analyzed for chromosomal anomalies.

What can happen before birth?

Even large omphaloceles do not cause fetal distress. Associated anomalies; however, may affect the fetus' growth and well-being. Chromosomal anomalies, in particular, can increase the risk of complications for the fetus.

The prognosis of omphalocele depends in large part on the presence of associated anomalies, rather than on the size of the defect. Some of the more complex chromosomal and other syndromes are associated with a very small omphalocele, while many giant omphaloceles are isolated without associated diseases or conditions. As the pregnancy progresses, it may be easier to identify other anomalies by ultrasound, which helps the physician determine the likely course of events at birth and the long-term outcome.

What can be done before birth?

Omphalocele is best treated after birth because fetal intervention poses too many risks for the mother and fetus.

What are my delivery options?

Small omphaloceles do not require pre-term delivery or Cesarean section. Giant omphaloceles, however, pose a significant risk and a Cesarean section is usually recommended—particularly if a portion of the liver is exposed. In those patients, there is a high risk of liver trauma with vaginal delivery.

It is recommended that mothers deliver in a hospital that has immediate access to a specialized NICU, with a pediatric surgical specialist present. Although the abdominal organs are typically protected from the outside world by the membrane that covers the omphalocele, this membrane can rupture and lead to the

same complications associated with gastroschisis. The intestines may be exposed and become dehydrated, and they may even have thick rind, also known as a "peel."

What will happen at birth?

In the presence of neonatologists, the mother will deliver her baby and, if the omphalocele is very small, she will be able to see and hold her baby immediately. The baby will then go to the NICU for assessment and treatment. If the omphalocele is large, and especially if it contains the liver, the baby may be at risk of dehydration and direct trauma to the exposed organs. He or she will receive an intravenous line for fluids and be placed under a warmer. It is possible that he or she will be intubated and placed on a respirator to aid breathing.

Because omphaloceles—even the very small ones—can be associated with other anomalies, several tests may be performed shortly after birth. Since heart anomalies are the most common associated disorders with omphalocele, an echocardiograph may be performed. Once it is clear that there are no major problems, the baby will undergo surgery to place the intestines and organs back in the abdomen and close the abdominal wall. The type of procedure depends on the size of the baby and how much of the intestines and other organs is exposed.

With giant omphaloceles, so much of the liver is exposed through the hole in the abdominal wall that trying to "push it back in" during one procedure would be dangerous. It is often safer to temporarily protect the organs and continue the procedure when the baby is a little older. There are many techniques that can be used to close a giant omphalocele, and no single approach is perfect for all babies. If rapid repair is not possible at birth, the medical team may choose to proceed in stages. First, the skin is closed over the omphalocele (even if the liver cannot be pushed back completely) and then progressive compression dressings are applied to gradually force the organs back into place. Another option is to enlarge the abdominal cavity with special devices before trying to push back the liver. Because this is often a difficult and lengthy process, infants with giant omphaloceles typically remain in the hospital for several months.

What is the long-term outcome?

Babies with small omphaloceles have an excellent outcome and will require little care once the hole is closed. Babies with giant omphaloceles may require much lengthier hospital stays and sometimes multiple operations in infancy and early childhood. However, the overall long-term outcome is excellent.

If the omphalocele is associated with other conditions or anomalies, the baby may need additional procedures, and his or her prognosis may be affected. Most often, the prognosis and outcome depend on the associated anomalies, rather than on the omphalocele itself. Complex syndromes associated with omphalocele include cloacal exstrophy, which involves bladder, intestinal, anorectal and spinal anomalies; and pentalogy of Cantrell, which affects the heart, lung and diaphragm. These patients require many complicated operations involving a multitude of specialists. The overall outcome is variable.

Other associated conditions may determine the immediate outcome of the baby with omphalocele and have lasting effects. For example, Beckwith-Wiedemann syndrome is an "overgrowth" syndrome that can manifest at birth as an overactive pancreas. This can lead to increased insulin secretion and dangerously low blood sugar, and also places the toddler at risk of certain malignant tumors. Close surveillance of these children is necessary.