

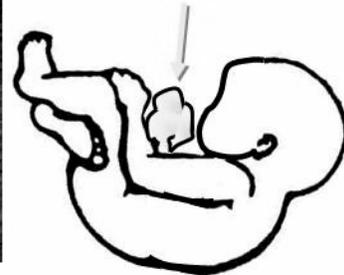


OMPHALOCELE*

*This information is intended to supplement your consultation with a pediatric surgeon, regarding your unborn child with omphalocele. The same information is also available on our web site: www.fetal-treatment.org. Underlined words are explained in a glossary at the end.

WHAT IS OMPHALOCELE?

Omphalocele (sometimes called "exomphalos") refers to a condition in the fetus whereby some abdominal contents (small and/or large intestine, stomach, and even liver) protrude through a hole in the abdominal wall. Unlike in gastroschisis, the hole is in the middle of the abdomen, right where the belly button would be. Instead, there is a variable size defect (hole) covered by a membrane (which somewhat protects the exteriorized organs). The umbilical cord of the fetus inserts at the top of this membrane, rather than on the abdomen itself. Although both omphalocele and gastroschisis appear the same (intestines protruding outside the abdomen), each condition has its own features. Abdominal wall defects can be detected by ultrasound from the third month of pregnancy on (14 to 15 weeks). As the pregnancy progresses, diagnosis becomes more accurate: loops of intestine can then be seen outside the abdomen, "floating" into the amniotic cavity (arrow).



HOW COMMON IS IT?

Omphalocele occurs somewhat less often than gastroschisis, and is estimated to be present in 1 of every 5,000 live births. It can be an isolated finding, but omphalocele is also seen in a number of chromosomal anomalies and other syndromes. The most common associated anomaly is a heart defect; others include the Pentalogy of Cantrell (which includes heart, diaphragm and other defects) and cloacal exstrophy (a severe anomaly involving the intestines, the bladder and pelvic organs). Omphaloceles are also seen in trisomy 13 and trisomy 18, two severe chromosomal anomalies. In all these cases, the omphalocele is only a small component of the fetal condition, and the outcome will be largely depend on the other anomalies, not on the omphalocele itself.

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Because of the relatively common association of omphalocele with other, vaster syndromes, many have, in the past, painted a grim picture for all omphaloceles. However, isolated omphaloceles have a prognosis similar to gastroschisis: once the extruded organs can be replaced in the abdomen and the defect closed, most of these children will have a normal life.

WHAT CAN BE DONE BEFORE BIRTH?

There is no reason to treat omphaloceles before birth (i.e., try to operate on the fetus). However, some measures can be taken once an omphalocele, or gastroschisis, has been diagnosed by ultrasound. Additional diagnostic tests may be necessary, particularly with omphalocele: an amniocentesis may be indicated, with chromosomal analysis; and efforts should be made to detect heart anomalies. The course of the pregnancy can be altered in three ways:

1. Mode of delivery

If intestines and other organs are outside the abdomen, it would seem logical that they would be at an increased risk of being damaged during normal delivery. Some have therefore advocated Cesarean section ("C-section") for all cases of gastroschisis and omphalocele. In fact, the risk of injury is only theoretical, and vaginal delivery does not put the baby at an increased risk of complications. For that reason, most (although not all) physicians now recommend normal delivery, even for gastroschisis, unless there are obstetrical reasons to proceed with a C-section. The main exception may be cases of "giant" omphalocele," where a large portion of the liver is exposed as well: here, there may be an increased risk of liver trauma with vaginal delivery.

2. Place of delivery

As long as he or she is inside the womb, the fetus with an omphalocele is relatively well shielded from trauma and complications. After birth, however, the exposed intestines and/or liver have to be protected from direct trauma and infection. The baby can be safely transported to a treatment center, as long as certain precautions are taken. However, if the diagnosis of omphalocele has been made beforehand, it would seem logical to have the baby be born directly in such a treatment center (i.e., a center with a neonatal intensive care unit and immediate access to a pediatric surgery service). Therefore, we generally recommend that, if you are pregnant with a fetus with gastroschisis or omphalocele, you plan to deliver in such a tertiary institution. Your care will likely be transferred to a Maternal-Fetal Medicine specialist at our institution, to facilitate the transition to peri- and postnatal care.

3. Time of delivery

One of the concerns with gastroschisis is that the exposed bowel becomes so damaged, that function is impaired and the baby may end up staying in the intensive care for a long time. It is known that many infants with gastroschisis have what appears to be damaged bowel, with very thick, rigid loops of intestines containing a "peel." One of the theories for this peel (and for the fact that some babies have little or no peel at all) is that prolonged exposure of the bowel to the amniotic fluid causes progressive damage. In other words, limiting the amount of time that the bowel is floating in this fluid (or even diluting that fluid by infusing sterile saline water inside the womb) could theoretically decrease the amount of peel and intestinal damage.

In omphaloceles, this is rarely a problem, because a membrane envelops the organs and shields them from exposure. However, that membrane can have ruptured (so-called ruptured omphalocele), exposing the intestines to the same potential trauma as with gastroschisis.

WHAT WILL HAPPEN AT BIRTH?

If everything goes as planned, you will deliver at a tertiary care center with direct access to a neonatal intensive care unit. The neonatologists will be present at delivery, so that they can immediately assess your baby and start treatment, if necessary. At the same time, the pediatric surgeons will be alerted, so that surgical correction can be performed as soon as possible. In most cases, however, you will be able to see (and hold) your baby after delivery.

Your baby will be "stabilized" in the intensive care unit. An intravenous line will be placed in an arm or a leg, so that fluids can be given. Because of the exposed intestines, your baby is likely to lose a lot of fluid by evaporation, and is likely to cool off more rapidly as well. Your baby will therefore be placed under a warmer, and if the omphalocele membrane is ruptured, the loops of bowel will be carefully wrapped to protect them from the outside. If your baby shows signs of distress, it is possible that he will be intubated, so that we can help him breathe better.

If it is clear that there are no other major problems, your baby will be ready to undergo surgical repair of the defect. How this is done will depend on how much intestines and other organs are exposed, and how big your baby is. In many cases, all the intestines can safely be placed back in the abdomen (so-called "primary repair"), and the abdominal wall can be closed. Of course, this is done in the operating room with your baby under anesthesia. Often, however, there is so much out that this cannot be safely replaced all at once. In that case, we try at least to protect the intestines until they are ready to be put back in the abdomen. For this, we place a "silo" (a clear plastic or silicone pouch) over the intestines, so that they are now shielded from trauma, infection and dehydration. This can be done at the bedside, in the intensive care unit, or in the operating room.

Once the swelling has gone down and the abdomen has become used to the presence of more bowel, the silo can be removed and the abdomen closed over the intestines. This typically takes a few days to a week.

If the membrane around the omphalocele is intact, it acts as a silo, and surgical intervention can be delayed somewhat. Small omphaloceles can be treated as described above. With giant omphaloceles, where a large portion of the liver is exposed, surgical intervention may be more difficult. If complete correction cannot rapidly be achieved, the first goal is to close the skin over the abdominal organs, so that they can be protected. Repair of the muscle defect may have to occur later.

Of course, associated anomalies may have to be addressed as well; as discussed, these anomalies (chromosomal or other) may be the determining factor for the baby's outcome.

WHAT HAPPENS NEXT?

On average, it may take 2 to 3 weeks before the intestinal tract functions properly again. During that time, your baby will be fed through the veins only, by "total parenteral nutrition," or TPN. He will get all the calories necessary to grow, until he can be fed by mouth again. Once gut function returns, it will likely take a while before your baby can tolerate full feeds, and that nutrition through the veins can be stopped. Your baby is likely to stay in the hospital for at least 1 month. Sometimes, this can be much longer, depending on the degree of prematurity and the associated anomalies.

COMPLICATIONS AND LONG-TERM OUTCOME

The overall outcome of isolated omphalocele is excellent: some infants may have minor intestinal problems in the first few months, but will recover from that and lead a completely normal life. Although the belly button may not look perfectly normal, there should be minimal scarring.

As mentioned, omphaloceles can be associated with other conditions. This happens in approximately 50% of patients with an omphalocele. If there are associated anomalies, these may have to be addressed as well. Trisomy 13 and trisomy 18 are severe chromosomal anomalies with a generally poor prognosis; cloacal exstrophy is not an immediately life-threatening condition, but is a complex anomaly that will require multiple surgical interventions and the input of many specialists. The prognosis of children with Pentalogy of Cantrell depends mostly on the degree of heart anomaly and whether the heart is exposed or not.



Beckwith-Wiedemann syndrome, which may present at birth with pancreas anomalies (too much insulin secretion, resulting in a very low blood sugar), is important because of its associated risk of childhood tumors. While most of these tumors can be treated effectively today, early detection is important. Therefore, babies with Beckwith-Wiedemann syndrome need to be screened (usually by ultrasound) on a regular basis for the first few years of life.

GLOSSARY

Amniotic cavity: The space within the uterus in which the fetus resides, and bound by the amniotic membrane

Atresia: An absent portion of an organ. Intestinal atresia (ileal or jejunal atresia) refers to a missing portion of small bowel, a known complication of gastroschisis. If present, this will require an operation, to reconnect the two ends of bowel

Beckwith-Wiedemann syndrome: An anomaly that includes omphalocele, enlargement of some of the organs (often the pancreas), a large tongue and various degrees of gigantism (large baby). These children have a risk of developing some childhood tumors, such as Wilms tumor and hepatoblastoma.

Cloacal exstrophy: A complex anomaly of the abdominal wall, the intestinal tract and the pelvic organs. Infants with cloacal exstrophy have a short intestine, an absent anus, a large defect of the abdominal wall (omphalocele) and the bladder (which is exposed and lacks a front wall), and anomalies of the pelvic bone and genitalia. This is not the same as a bladder exstrophy (defect of the bladder, with or without anomaly of the penis in boys), which is a less severe condition.

Diaphragm: A large muscular sheath that separates the chest from the abdomen. Anomalies of the diaphragm include diaphragmatic hernia, a congenital defect in that muscle. The most common form is a posterolateral hernia, or Bochdalek type (whereby the hole is mostly on the side and in the back of the diaphragm, causing intestines to move into the chest). The anterior form, or Morgagni, is a defect behind the sternum (breast bone); this is the one most commonly associated with omphaloceles, as part of the Pentalogy of Cantrell.

Gastroschisis: refers to a defect (hole) in the abdominal wall of the fetus or newborn, through which intestines or other abdominal organs can protrude. In gastroschisis, the hole is to the (patient's) left of the belly button.

Omphalocele: Abdominal wall defect in the fetus, located in the umbilicus. Often, a membrane covers the exteriorized intestines. "Giant" omphaloceles contain not only intestines, but liver as well. If only a very small defect is present, this is often referred to as "hernia of the umbilical cord." Omphaloceles are to be differentiated from gastroschisis, where the defect is to the side (usually left) of the belly button. Omphaloceles are often associated with other anomalies, including congenital heart defects. Omphalocele can also be part of a syndrome, such as Beckwith-Wiedemann syndrome, pentalogy of Cantrell and cloacal exstrophy.

Peel: The thick layer of scar often seen in exposed bowel loops in gastroschisis.

Pentalogy of Cantrell: A rare syndrome that includes five anomalies: an omphalocele, a

diaphragmatic hernia (hole in the diaphragm), a heart defect, a defect of the pericardium (the membrane that envelops the heart), and a defect the sternum (breast bone), which in very severe cases can cause the heart to protrude out of the chest.

Silo: A silicone plastic ("silastic") sterile bag or membrane that can be placed over the exposed bowel loops in gastroschisis, to prevent dehydration and further damage to them. It is used for temporary cover if the intestines can not immediately be replaced in the amniotic cavity.

Total Parenteral Nutrition (TPN): Basic nutrients (carbohydrates, essential fats, proteins, vitamins and trace elements) given intravenously (through a vein), usually via a central venous catheter, that provide enough calories for an patient to survive *and* grow without any feeding by mouth. This life-saving technique allows a baby with gastroschisis to receive enough nutrition until he can feed normally (often 3-4 weeks later).

Trisomy: The presence of an extra chromosome (normally, humans have 2 sets of 23 chromosomes, or 46 chromosomes, in each cell; individuals with a trisomy have an extra copy of one chromosome per cell). The most common trisomy syndromes are trisomy 21 (Down syndrome), trisomy 13 and trisomy 18. The latter two are sometimes associated with an omphalocele.