

Questions and Answers About:

Omphalocele

What is An Omphalocele?

Omphalocele is a congenital birth defect in which the contents of the abdomen push out of the abdominal cavity into the umbilical cord. (Figure 1). A thin membrane of tissue covers the intestines. Omphaloceles vary from the size of a walnut to the size of a large grapefruit.

The cause of large vs. small omphaloceles differs. Normally, the intestines pass out of the abdominal cavity and into the umbilical cord at about 6 weeks after conception. In small omphaloceles, the intestines fail to return into the abdominal cavity, as they do normally at about 10 weeks after conception. These small omphaloceles contain only intestine and not other organs.

In large omphaloceles, the cause is a disturbance in the normal fusion of the two body folds that normally merge to form the abdominal wall muscles. This incomplete formation of the abdominal wall muscles causes a weakness at the base of the umbilical cord. This weakness allows the intestines, liver, and other organs to enter the umbilical cord. Large omphaloceles that contain intestines, liver, or other organs are often called “giant” omphaloceles.

Diagnosis

A fetal ultrasound often identifies the omphalocele in the second or early third trimester. Most omphaloceles are now diagnosed before delivery. This helps the pediatrician and surgeon prepare for neonatal care. The diagnosis by physical examination is easy after delivery. However, if the baby is not born in a perinatal/neonatal center, transfer to specialized NICU will be necessary.



Figure 1. Unrepaired omphalocele with intestines protruding through enlarged umbilical cord. Photo courtesy of Dr. David Rustad.

How Often Does Omphalocele Occur?

A small omphalocele (one in which only the intestine protrudes outside of the abdomen) occurs in one of 5,000 births. A large or “giant” omphalocele occurs in roughly one of 10,000 births.

Omphalocele in Subsequent Pregnancies

After a couple has had one baby with omphalocele, their risk of having another depends on the cause of the first. If the omphalocele is not associated with a syndrome, such as Beckwith-Wiedemann syndrome (see below), and is not due to abnormal chromosomes (too much or too little genetic material), the recurrence risk is low, 1% or less. However, in a small number of families a major genetic cause may exist. In such situations omphalocele may recur in as many as 50% of subsequent children. Therefore, couples should meet with a genetic counselor to receive an individualized assessment of their

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risk of having a subsequent baby with omphalocele.

Complications

About 30% of babies with omphalocele also have a major chromosome abnormality. In these cases, the chromosome abnormality causes the omphalocele and also causes abnormalities in many other body systems and organs. These babies rarely survive (Table) and if they do survive, they suffer severe handicaps.

About 50% of all babies born with an omphalocele have other birth defects in the heart, kidneys, or other organs, even if the chromosome tests are normal. Approximately 35% of babies with omphalocele will have heart defects. Therefore, when a baby has an omphalocele, we always examine the baby for other birth defects and perform a chromosome test.

Beckwith-Wiedemann Syndrome (BWS)

BWS consists of excessive growth of the fetus (large-for-gestational age); a large liver, a large, protruding tongue, grooves on the ear lobe, and a low blood sugar. BWS occurs in about 1 in every 15,000 births, although the incidence is probably higher in babies conceived by in vitro fertilization. BWS is also important because these infants have a higher risk (6-7%) of kidney cancer in childhood (Wilm's tumors).

What Are My Baby's Chances of Survival

The overall survival of infants with omphalocele is 77%. Three factors influence survival:

- **Prematurity.**
Infants who are born at <36 weeks gestation have a low survival rate, 57% (16/28). Survival increases with increasing gestational age and reaches 87% at a gestational age of \geq 36 weeks.
- **Size of the omphalocele.**
If the omphalocele contains liver, it is considered to be a giant omphalocele. Many of these patients have underdevelopment of the lungs (pulmonary hypoplasia) as well as the abdominal cavity. Such babies have great difficulty breathing and need ventilator support to survive. If a baby has small



Figure 2. Picture of silo placed on protruding bowel. the silo is rolled down over time like a tube of toothpaste. Photo courtesy of Dr. David Rustad.

lungs, survival drops to 50% regardless of the gestational age.

- **Presence of other birth defects.**
Babies born with birth defects in addition to omphalocele often have a long hospital stay and have a lower survival rate. For examples, see table below:

Table 1

Incidence and Survival with Additional Birth Defects and Omphalocele

<u>Birth Defect</u>	<u>Incidence</u>	<u>Survival</u>
Heart defect	34%	63%
Imperforate Anus (malformed anus)	15%	69%
Trisomy (One type of chromosome problem)	30%	1 %

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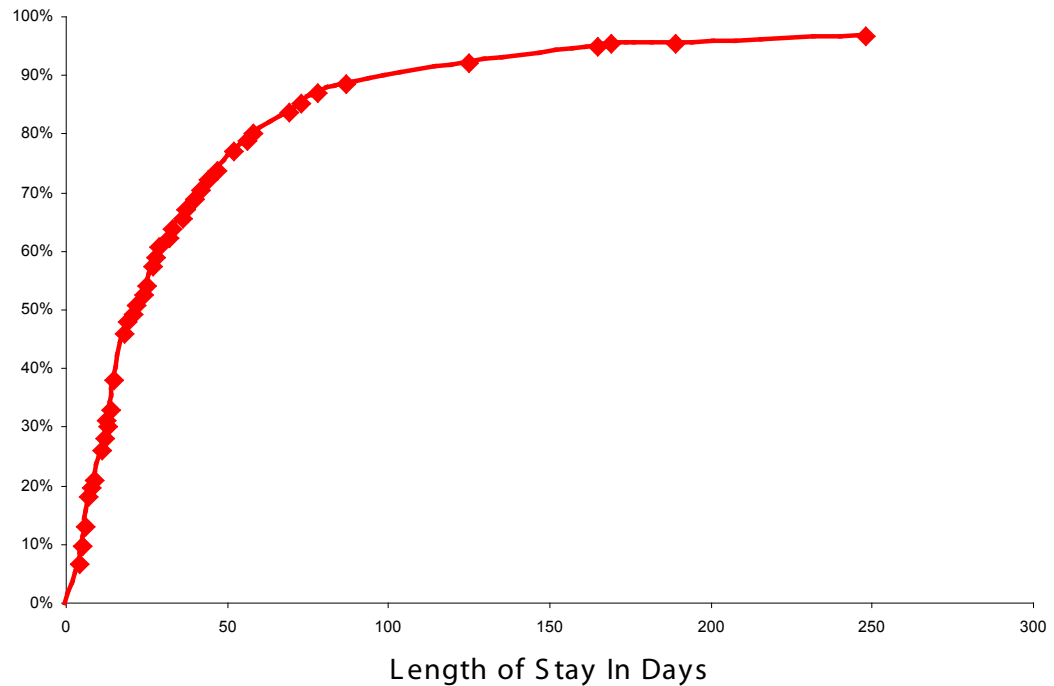


Figure 3. The X-axis is the length of stay in days. The Y-axis of the graph is the cumulative percentage of patients who have gone home at the corresponding day. Two babies who stayed in the hospital for significantly longer than 300 days were not included on this graph. Data is from Children's Hospitals and Clinics- Minneapolis.

When babies with omphalocele are born at >35 weeks gestation, with normal chromosomes, and no birth defects of the heart, lungs, kidneys and intestine, survival is 97% (36/37). With any of these complications, survival varies greatly depending on the nature of the chromosomal or other problem.

Do I Need a Cesarean Delivery?

For many years doctors thought that babies with omphalocele should be delivered by cesarean section to prevent rupture of the membrane covering the omphalocele. More recent research disputes this and shows no advantage to cesarean delivery

Treatment

The intestines and other organs must be returned to the abdominal cavity and the abdominal wall defect repaired. Some large omphaloceles require multiple operations to completely repair the abdomen. If multiple surgeries will be required, the first stage is often the application of a silo to protect the intestines and other organs while the abdominal cavity

is stretched (Figure 2). A silo is a plastic bag that is placed over the intestines, when the abdominal cavity is too small for a complete repair with just one surgery. The silo provides a sterile, protected environment for the intestines. The silo is gradually rolled down, like a tube of toothpaste, gradually forcing the intestines back into the abdomen and stretching the abdominal cavity. When the intestines have been completely returned to the abdominal cavity, the silo is removed and the abdomen is repaired. Most babies require a silo for only 7-10 days. Occasionally, patients will have such a large omphalocele, that repair requires multiple surgeries and the placement of a synthetic patch.

In addition to treating the omphalocele, other birth defects may also require treatment before discharge home. For example, some infants with omphalocele and heart defects will also need heart surgery.

Length of Hospital Stay (LOS)

The median (average) hospital stay is about 30 days (Figure 3). However, the actual LOS var-

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ies widely. Patients with very long LOS usually have had pulmonary problems, feeding difficulties, or other major birth defects.

Long-term Prognosis

Most babies with omphalocele do well after their abdominal wall is repaired. A few have intestinal problems, but these are uncommon. Birth defects other than omphalocele also influence the outcome. The most common, serious other defect involves the heart. These problems often require surgery and extended follow-up in the pediatric cardiology clinic.

Gastro-esophageal reflux (when food frequently sloshes up into the esophagus from the stomach, called acid reflux in adults) has also been a problem for some of our patients. This can usually be treated with medicines, but has occasionally required a special feeding tube through the abdominal wall into the stomach.

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Internet Resources

1. Mothers of Omphaloceles Organization (MOO) website offers many stories and pictures as well as a support network, chat room, and opportunities to contact other parents of babies born with an omphalocele <http://www.omphalocele.com/>
2. Beckwith-Wiedemann Support Network site is a peer support group for parents and professionals who deal with Beckwith Wiedemann Syndrome (<http://www.beckwith-wiedemann.org/>)