Omphalocele (exomphalos)

Patient and family information, brought to you by the Education Committee of APSA

Overview - "What is it?"

An omphalocele is an opening in the middle of the belly where the muscle and skin is not present. Instead, the organs are covered by a thin transparent sac (Figure 1). The umbilical cord (belly button) is located in the center of the sac.

Figure 1: Courtesy of M. Arca 11/2016

Giant omphalocele: If the opening is greater than five centimeters in diameter and contains mostly liver, it is a giant omphalocele. Infants with giant omphaloceles have more problems with breathing, feeding and overall survival compared to those with smaller defects.

Omphaloceles are associated with older mothers and occur in between 1 in 3,000 – 10,000 live births. In some cases, omphalocele can be associated with chromosomal syndromes (abnormal genes) and 20-50% of babies with omphaloceles are born with cardiac abnormalities.

Signs and Symptoms - "What symptoms will my child have?"

Early signs: Most omphaloceles are diagnosed during prenatal ultrasound. If the omphalocele is small, it may be diagnosed after the baby is born**.**

Conditions that mimic this condition: Other defects of the belly wall such as gastroschisis, exstrophy may mimic omphalocele.

Diagnosis - "What tests are done to find out what my child has?"

Prenatal Studies: Once the baby inside the mom is suspected or found to have omphalocele, other studies may be needed. Not all of these may be needed:

- **Blood test:** Alpha fetoprotein is a substance that is elevated in the blood stream.
- **Ultrasound:** The mother will have frequent ultrasounds to evaluate the growth of the fetus and make sure that the sac covering the hole remains intact.
- **Magnetic Resonance Imaging (MRI):** Some centers obtain an MRI to evaluate the baby's heart, lungs, and other organs.
- **Amniocentesis:** Fluid from inside the uterus may be aspirated to look for chromosomal abnormalities.

Treatment - "What will be done to make my child better?"

A baby with omphalocele should deliver in a hospital that has ready access to surgeons and specialists that can take care of the baby right away (usually a dedicated children's hospital). Your obstetric doctor will discuss options about delivery. In a baby with large omphalocele, a Caesarian section (C-section) may be recommended, as passage through the birth canal can injure the liver, causing bleeding. Ideally, the parents can meet with surgeons and infant specialists before the baby is born to get an idea where and how your child will be cared for.

After initial stabilization, a thorough exam is done and investigations are arranged to evaluate for other associated abnormalities. These include an echocardiogram to look at the structure of the heart, a chest x-ray and further chromosome analysis**.**

Surgery: The size of the defect, the amount of liver and intestines that are out into the sac, the size of the baby and presence of other abnormalities determine the surgical approach.

Primary closure: In general, if the defect is small and the baby is healthy, the muscle and skin can be closed within a couple days after birth.

Delayed closure: When the hole is large, the size of the belly cavity is correspondingly small, replacing all the organs that have been out of the belly all at once creates a tight space that may slow down the blood flow to these organs.

In these cases, the sac covering the defect is encouraged to mature into regular skin. This process takes months. During skin formation, there is a chance that infection can occur. The sac is protected by medicines that fight infections. During the first few months or weeks, the sac is

fragile and can tear. This creates a situation where surgery is needed to place temporary coverage on the exposed organs.

When the infant has grown to the degree that the organs fit into the abdominal cavity, the muscles can be closed. Closure usually takes several surgeries spaced between months. During this time, infants with large omphaloceles may establish full feeds allowing them to grow.

Informed consent: A consent form is a legal document that states the tests, treatments, or procedures that your child may need and the doctor or practitioner that will perform them. Before surgery, your doctor should tell you what the operation is, the goal of the surgery and other possible treatment options that are available. Your doctor should explain the risks and benefits of the surgery. You give your permission when you sign the consent form. You can have someone sign this form for you if you are not able to sign it. You have the right to understand your child's medical care in words you know. Before you sign the consent form, make sure all of your questions are answered. It is important to know that during surgery, there are things that can happen that your doctor may have not predicted before going in. He or she will explain these to you after the surgery.

Preoperative preparation: For a small omphalocele, closure is done a few days after birth. The infant will receive antibiotics through the vein to decrease infection.

For a large omphalocele, closure is scheduled months after birth when there is usually mature skin covering the omphalocele sac. A bath or shower the night prior to or the morning of the operation is encouraged. The infant will receive antibiotics through the vein to decrease infection.

Postoperative care: After an omphalocele is closed, the infant is cared for in a monitored setting. Feedings will resume once intestinal function returns.

Pain medicine: Pain medicine can include acetaminophen (Tylenol®), ibuprofen (Motrin®) or narcotics. These medicines can be given by vein or by mouth.

Benefit: The benefit of surgery is having organs protected by the muscle and skin. The benefit of the delayed closure approach is that often the infant can be discharged from the hospital and return for closure as a planned, elective admission.

Risks: Returning organs to a tight abdominal space can decrease the blood flow to those organs. This should be monitored closely whether the repair is done days or months after birth. Wound infection, need for a post-op ventilator, bleeding, damage to internal organs that may need repair, as well as anesthesia risk are all risks of surgery.

Home Care - "What do I need to do once my child goes home?"

Care of infants with omphaloceles can be more complicated if they have problems with other organs such as the heart and lungs.

Diet: If the infant is ready to go home, in general, they will be eating normally. If they have heart or lung problems, they may need to have supplemental feedings by a tube that goes into the stomach or nutrition by vein. Supplementation is more the exception than the rule.

Activity: In general, normal activities according to the child's developmental stage should continue. If the child has a giant omphalocele that is still maturing, it is advisable to have a protective device over the omphalocele. This is usually fashioned in the hospital by occupational therapy.

Wound care: The surgeon will give instructions for wound care. If a complete closure is done, the wound would be ready for baths upon discharge.

Medicines: By the time of discharge, minimal oral pain medication such as Tylenol® can be administered. Postoperative pain should have almost resolved.

What to call the doctor for: Wound redness, drainage, fever. Vomiting, especially green vomiting, can indicate intestinal obstruction and requires immediate attention.

Follow-up care: If the post-operative course is uneventful, the infant can see the surgeon in one or two weeks after discharge in the office. You will also be directed to see other specialists if your baby has other conditions.

Long Term Outcomes - "Are there future conditions to worry about?"

Infants with omphaloceles can have a long recovery phase. Conditions that exist in addition to the omphalocele influence survival.

The size of the defect influences outcome and some report a 25% mortality rate for giant omphaloceles. Death is usually due to respiratory failure, infection, or prolonged intravenous feedings (TPN) that damage the liver. Prognosis is good for isolated small omphaloceles and no other comorbidities. Survival rates are between 75-95% for these infants.

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