Newborn with an Omphalocele and Ventricular Septal Defect

Robert Listernick, MD

This baby boy was the 4,465-gram product of a 37-week gestation to a 39-year-old G2P2 female. The pregnancy was complicated by ultrasoundography that revealed an omphalocele as well as ventricular septal defect and possible tetralogy of Fallot. Prenatal surgical consultation was obtained.

Robert Listernick, MD, moderator: Just to be clear, what’s the difference between an omphalocele and gastroschisis?

Marleta Reynolds, MD, pediatric surgeon: Omphalocele is a defect of the abdominal wall due to failure of fusion of the umbilical ring. Intestinal contents herniate through the defect covered by a sac, unless it has ruptured. There’s a high incidence of associated extraintestinal defects. In gastroschisis, the defect is always to the right of an intact umbilicus. It results from rupture of a dysplastic abdominal wall or a vascular defect. There is neither a sac nor any associated defects.

Dr. Listernick: What issues should be addressed at this prenatal consultation?

Kathy Barsness, MD, pediatric surgeon: We need to discuss both treatment of the omphalocele as well as the potential for associated anomalies.

Dr. Listernick: Let’s start with the omphalocele itself.

Dr. Barsness: The outcome is highly dependent on the size of the omphalocele and the state of the omphalocele sac. For a very small omphalocele, often called a hernia of the cord, in which the contents come up into a sac which has the insertion of the umbilical vessels within it, associated intestinal complications are very low. Think of it as a large umbilical hernia. We close these defects at birth. We also close at birth larger omphaloceles that simply have intestinal contents in the sac, assuming the sac has not ruptured.

Dr. Listernick: When does it get more complicated?

Dr. Barsness: As more abdominal contents enter the sac, the more difficult it is to close and the incidence of complications rises dramatically. Once the liver enters the sac, it becomes extremely difficult to replace all the sac’s contents back into a small peritoneal cavity. The retroperitoneal structures — the duodenum, kidney and cecum — may be the only things that remain in the abdominal cavity which was never forced to grow in utero because all the normal structures were in the omphalocele. In addition, if the liver is in the sac, it’s at an angle which kinks the inferior vena cava potentially causing ischemia to the liver or impeding venous return to the heart.

Dr. Listernick: And the treatment for these large omphaloceles?

Dr. Barsness: As long as the sac is intact, we typically paint the sac with dilute Betadine and use the sac as a temporary membrane to protect the abdominal visceras. We delay closure until the peritoneal cavity has enlarged enabling us to replace the visceras safely.

Dr. Reynolds: Another important issue of which to make parents aware is the potential for associated pulmonary hypoplasia because the chest has not had a chance to expand in utero. An important reason to delay closure of giant omphaloceles is these infants’ lack of lung reserve. If they are taken to the operating room in the neonatal period, they may never be able to be safely removed from the ventilator.

Dr. Listernick: What should be said to the parents about potential associated defects?

Barbara Burton, MD, pediatric geneticist: Knowing that there are two
anomalies and a 39-year-old mother, I’d be very concerned about a chromosomal abnormality. Omphaloceles are particularly seen in infants who have trisomy 13 but may also be seen in those with trisomy 18. Beckwith-Wiedemann syndrome (BWS) is certainly another syndrome to consider. Even in babies who don’t have a recognizable genetic syndrome, there’s a significantly increased risk of cardiac defects in babies who have an omphalocele.

Dr. Listernick: What about counseling about the delivery itself? Can these infants be delivered vaginally?

Daniel Robinson, MD, neonatologist: Children with some abdominal wall defects can be delivered vaginally. However, infants with a giant omphalocele are different. The obstetrician has to decide whether the baby can be safely delivered without rupturing the sac which could have fatal complications from liver hemorrhage. There are even some data suggesting that infants with small to moderate myelomeningoceles can be delivered safely vaginally as long as there aren’t significant in utero hydrocephalus and macrocephaly.

Dr. Listernick: This baby was born by Cesarean section due to failure to progress. Apgar scores were 2 at 1 minute, 8 at 5 minutes. An intact omphalocele was noted at birth and was wrapped in sterile gauze in a plastic bag. The family history was significant for a male sibling who was delivered at 24-weeks gestation and passed away; we have no further information about that infant.

On exam, he was a large infant whose weight was in the 90th percentile, length in the 97th percentile, and head circumference in the 10th percentile. He had a megal ear lobe creases in men are associated with a higher risk of coronary artery disease? File that fact away for use on a rainy day. So, what is the cause of BWS?

Dr. Burton: It’s complicated. In most patients it’s an epigenetic phenomenon in which there is no change in the genetic code but gene expression is altered due to genetic imprinting. The critical region is on chromosome 11, specifically 11p15.5. The critical BWS region is normally silenced by methylation of the maternal chromosome and active on the paternal chromosome. BWS occurs in approximately 50% of cases due to loss of methylation (activation) of this region on the
maternal chromosome. Twenty percent of cases are due to paternal uniparental disomy, inheritance of two copies of the paternal 11p15.5 region. Rarely, BWS may result from duplications or microdeletions that are detectable on whole genome microarray or from mutations of a specific gene which, if mutated, can impair methylation. These latter infants may have a family history of BWS.

**Dr. Listerick:** How do you confirm the diagnosis of BWS?

**Dr. Burton:** Once again, it’s complicated. We take a step-wise approach. Generally we start with whole genome microarray and methylation testing which detect approximately 60% of cases. Next, we may do testing for uniparental disomy which detects an additional 20% of cases.

**Dr. Listerick:** Which babies in the nursery should we suspect have BWS and perform testing?

**Dr. Burton:** That’s not clear. It’s easy when you have the full spectrum of anomalies such as in this baby. For instance, some geneticists recommend BWS testing in all infants with seemingly isolated hemihyperplasia, macroglossia or omphalocoeles.

**Dr. Listerick:** Why do babies with BWS develop hypoglycemia?

**Don Zimmerman, MD, pediatric endocrinologist:** Hypoglycemia occurs in approximately half of these children, usually lasting only days or several weeks. In about one tenth of those who are hypoglycemic, the hypoglycemia may persist for months or years. The mechanism is hyperinsulinism. It turns out that the 11p15.5 region contains a gene responsible for a potassium channel which regulates the release of insulin from the beta cells of the pancreas.

**Dr. Listerick:** Why does the hypoglycemia improve over time?

**Dr. Zimmerman:** That’s unknown.

**Dr. Listerick:** Does anything need to be done about this child’s macroglossia?

**Dr. Reynolds:** This tongue is not particularly large in the grand scheme of macroglossia. We’ve seen tongues so large that they are obstructing airways, necessitating tracheostomy and surgical reduction of tongue mass.

**Dr. Robinson:** Although the tongue does not look enormous, it is causing the child some difficulty eating and it produces significant upper airway obstruction on sleep study. We’re not out of the woods yet in so far as needing to deal with the macroglossia. The family is having a difficult time accepting the degree of macroglossia and are interested in a surgical solution.

**Dr. Reynolds:** Over time, the tongue will fit into the oral cavity. It might be beneficial to offer the family services through the Beckwith-Wiedemann Children’s Foundation which has its own website. They could contact other families with similar BWS children.

**Dr. Listerick:** We’d be remiss if we didn’t mention the increased risk of abdominal malignancies in BWS children.

**Dr. Burton:** There’s an increased risk of Wilms’ tumor, hepatoblastoma, and adrenocortical carcinoma. Recommendations include serial abdominal ultrasonography every 3 months up to the age of 8 years and serum alpha-fetoprotein measurement on the same timetable up to the age of 4 years.

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### Key Learning Points

1. Omphalocele is a defect of the abdominal wall due to failure of fusion of the umbilical ring. Intestinal contents herniate through the defect covered by a sac, unless it has ruptured. There’s a high incidence of associated birth defects.
2. Gastrocsisis results from rupture of a dysplastic abdominal wall or a vascular defect. There is neither a sac nor any associated extraintestinal birth defects. However, there is a high incidence of associated intestinal atresia and subsequent episodes of enterocolitis.
3. Omphalocoeles are associated with Trisomy 13 and Trisomy 18, as well as with Beckwith-Wiedemann syndrome (BWS).
4. Children with BWS have an increased risk of Wilms’ tumor, hepatoblastoma, and adrenocortical carcinoma. Recommendations include serial abdominal ultrasonography every 3 months up to the age of 8 years and serum alpha-fetoprotein measurement on the same timetable up to the age of 4 years.

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