Abdominal Wall Defects: Omphalocele and Gastroschisis

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**Learning Objectives**

1. Define the difference between omphalocele and gastroschisis.
2. List the six most common chromosomal abnormalities associated with omphalocele.
3. Explain the formation of the embryonic abdominal wall.
4. Explain how the embryonic gut and abdominal wall forms.
5. Explain the significance of an elevate alphafetoprotein (AFP) level in the pregnant woman.

Omphalocele and gastroschisis are the two most common major congenital abdominal wall defects, occurring in 1 in 2,000 live births.\(^1\) Omphalocele is characterized by a midline defect of abdominal muscles, fascia, and skin that results in herniation of the intra-abdominal structures into the base of the umbilical cord. The herniated mass is covered by parietal peritoneum and amnion, with Wharton jelly intervening between the two membrane layers. Prevalence of omphalocele is reported to be similar to that of gastroschisis with a male-female ratio of 1:1. Unlike gastroschisis, there is a higher incidence of omphalocele and chromosomal abnormalities with increased maternal age. The incidence of chromosomal anomalies in omphalocele is reported as high as 40% to 60%. Of these abnormalities, the most common are trisomy 13, 18 and 21, together with Turner, Klinefelter and triploidy syndromes. In addition, the association with anomalies has also been reported to increase if the herniation contains small bowel only or when associated with oligohydramnios or polyhydramnios. The heart is affected in 50% of anomalies.\(^2\)
Gastroschisis is a periumbilical defect involving all the layers of the abdominal wall. The hallmark of gastroschisis is free-floating bowel. The herniation of the intestine occurs through a small abdominal wall defect located just lateral, and usually to the right of, an intact umbilical cord. Diagnosis on ultrasound is made by demonstrating a normally situated umbilical cord and herniated free-floating intestine in the amniotic fluid. Because the defect involves all layers of the abdominal wall, an open communication exists between the amniotic milieu and the intra-abdominal space, with no protective layer over the herniated bowel. Herniation of liver or stomach through this defect is uncommon. Since the bowel is in direct contact with the amniotic fluid, both serum and amniotic fluid alphafetoprotein (AFP) levels are elevated more than they are with omphalocele. Most gastroschisis defects occur spontaneously with no increased incidence of chromosomal abnormalities. In contrast, intrauterine growth restriction accompanies gastroschisis in up to 77% of cases, with intrauterine fetal death occurring in as many as 4.6% of cases.

The developing embryo is transformed from a flat disc (the fetal pole) to a cylindrical shape through a progressive sequence of events that start around the fifth to sixth menstrual week. This process is accomplished by the folding of the cranial, caudal, and lateral ends of the embryo simultaneously. This transformation, accompanied by the development of fetal organs, gradually establishes the body form. At the end of the 10th week, the embryo assumes a recognizable human appearance.
Abdominal wall defects have been detected by ultrasound as early as ten menstrual weeks, but the forces that lead to their formation occur between the fifth and tenth menstrual weeks. It is during this period that a physiological abdominal herniation develops. The primitive gut begins development within the fourth week of gestation. Embryonic folding toward the midline results in the fusion of the abdominal wall on the midline. Fusion is completed by the eighth week and physiologic umbilical herniation occurs. This phenomenon is prompted by the rapid growth of small bowel and the lack of intra-abdominal space which is filled primarily by the relatively large liver and kidneys. The midgut grows faster than the abdominal cavity and physiological bowel herniation occurs. Within the umbilical cord, the midgut loop rotates 90° counterclockwise around the axis of the superior mesenteric artery. During the 10th fetal week (12th menstrual week), the intestines return rapidly to the abdomen. As the large intestines return, they undergo a further 180° counterclockwise rotation. This phenomenon is referred to as reduction of the physiologic midgut hernia.1 This embryonic period is the time during which all major internal and external structures develop. Any aberration in the normal sequence of events can result in developmental anomalies that lead to a wide spectrum of abdominal wall defects.2 Fusion failure results in an abdominal defect that allows the abdominal organs to protrude into the amniotic fluid either with or without a covering membrane.

Prenatal testing includes determining the levels of maternal serum alphafetoprotein (MS-AFP) and acetylcholinesterase (AchE) with abnormal levels raising suspicion for congenital defects of the brain, spine, or abdomen. Sonography is a valuable tool in diagnosing abdominal wall defects as an isolated defect or part of a syndrome.6 Although not specific to one type of malformation, an elevated AFP raises suspicion for a neural tube defect (NTD), an abdominal wall defect, or
any malformation that results in exposure of internal fetal tissue to the amniotic fluid. Abdominal wall defects are the second most common cause of elevated AFP values which includes gastroschisis and omphalocele.³

The levels of AFP found in conjunction with an anterior wall defect will vary depending on the specific type of malformation present. The MS-AFP values are elevated with the gastroschisis defect due to direct contact of the fetal bowel and abdominal organs with the amniotic fluid. Omphalocele membranes reduce the contact of the fetal bowel with amniotic fluid, resulting in a lower MS-AFP. The difference in lab values results in the detection of approximately 85% of gastroschisis and 50% of omphalocles.³

Because abdominal wall defects occur early in embryologic development, it is theoretically possible to detect all major defects before the age of viability. In actual practice, small abdominal wall defects may be missed during routine obstetric scanning. This emphasizes the importance of routine views of the anterior abdominal wall and umbilical cord insertion site as recommended by the guidelines adopted by the American Institute of Ultrasound in Medicine.⁵

Prenatal detection of anterior abdominal wall defects has significantly improved over the past 10 years. Ultrasound has proven to be a very effective tool in determining the presence of anterior abdominal wall defects in utero. It is among the more definitive diagnoses that can be made in a routine obstetric ultrasound examination.⁶ The American Institute of Ultrasound in Medicine (AIUM) and the American College of Radiology (ACR) both publish guidelines suggesting that the routine prenatal sonogram should include images of the anterior abdominal wall. Familiarity
with the normal sonographic appearance of the anterior abdominal wall is essential in identifying
defects. Any variation of the typical abdominal wall relationships raises suspicion for an anterior
wall malformation.7

When detected prior to fetal viability (approximately 24 menstrual weeks), knowledge of an
abdominal wall defect provides the prospective parents the opportunity to make important
decisions regarding the pregnancy.1 The prognosis for fetuses identified with gastroschisis is
generally excellent; the mortality rate has steadily and dramatically improved during the last
three decades and is currently less than 10%. This remarkable achievement can be attributed to
improved perinatal management including use of total parenteral nutrition, improved surgical
skills, and the overall improvement of prenatal sonograms due to published guidelines by the
AIUM and the ACR.

The prognosis for fetuses with omphalocele depends primarily on the presence and severity of
concurrent anomalies. For this reason, the overall mortality rate for omphalocele during the last
two decades has not improved as much as for gastroschisis. The presence of one or more
concurrent malformations is associated with perinatal mortality of 80%, and the presence of a
chromosome abnormality or a major cardiovascular malformation increases the mortality rate to
nearly 100%. When no other anomalies are present, however, the mortality rate drops to nearly
10%, which is similar to that for gastroschisis.1

Because excellent results can be expected following surgical correction of an isolated abdominal
wall defect, parents may choose to continue the pregnancy when the defect represents
gastroschisis or an omphalocele that is not associated with concurrent anomalies or chromosome abnormalities. On the other hand, pregnancy termination may be elected when the defect represents a complex malformation or when additional anomalies are present. These diverse options emphasize the importance of establishing the correct diagnosis and identifying major concurrent anomalies when an abdominal wall defect is detected.¹

Prenatal sonography is a well-established and efficient method of improving pregnancy outcomes through early detection of life threatening malformations. The use of serum alphafetoprotein as a maternal screening tool has also increased the detection of neural tube and abdominal wall defects. Accurate diagnosis of an abdominal wall defect encompasses an array of fetal anatomic anomalies ranging from the repairable gastroschisis or omphalocele to the often fatal limb-body wall complex (LBWC), an anomaly of multiple organ systems. The use of increasingly sensitive and advanced maternal serum testing, modern sonographic technology, and standardization of prenatal imaging protocols has contributed to the development of a reliable group of tests available to clinicians to aid in prenatal diagnosis of congenital defects.⁶
References


