The Abdominal Wall

Normal Anatomy of the Abdominal Wall

The superior wall of the abdominal cavity is formed by the diaphragm. This muscle can be seen as a hypoechoic line separating the lung and fetal liver (Fig. 6-1). In cases of severe hydrops, the structure appears as a hyperechoic sheet between the pleural effusion and the ascites. When a massive pleural effusion is present, the muscle can be inverted.

The floor of the abdominal cavity is formed by the pelvic diaphragm. The pelvic bones (iliac crests, ischial ossification centers) and pelvic muscles can be imaged with ultrasound but have limited diagnostic interest (Fig. 6-2).

The anterior abdominal wall is formed by the skin, subcutaneous tissue, and muscles. The muscles are visible as hypoechoic structures (Fig. 6-3). In the past, the image of the muscles has been confused with ascites and referred to as "pseudoascites."1,2 The entrance of the umbilical cord into the fetal abdomen should always be imaged to screen for the presence of an omphalocele (Fig. 6-4). Examination of the anterior abdominal wall should include visualization of its infraumbilical portion in order to rule out caudal fold defects (e.g., bladder exstrophy). Figure 6-5 shows the normal contours of the lower portion of the anterior abdominal wall.

The posterior wall of the abdomen can be easily imaged. Structures that can be identified include the spine and the paraspinal muscles.

REFERENCES

Figure 6-2. Coronal scan of a male fetal pelvis. A curved arrow points to the right iliopectoas muscle. IW, iliac wings; IT, ischial tuberosities.

Figure 6-3. Transverse section of the abdomen of a third trimester fetus. The abdominal muscles appear as hypoechogenic structures (arrowheads).

Figure 6-4. Longitudinal scan showing the insertion of the umbilical cord in the abdomen of a first trimester fetus.

Figure 6-5. Longitudinal scan of the fetal abdomen between the insertion of the umbilical cord (UC) and the gender. B, bladder; P, penis.
Diaphragmatic Hernia

Definition
Diaphragmatic hernia consists of protrusion of the abdominal organs into the thoracic cavity through a diaphragmatic defect. It includes posterolateral diaphragmatic hernia, Bochdalek hernia, retrosternal diaphragmatic hernia, Morgagni hernia, and eventration of the diaphragm.

Incidence
The incidence of diaphragmatic hernia varies depending on the type of study. The best estimates vary from 0.033 to 0.05 percent of births. Neonatal surgical series report a frequency of 0.012 percent. This difference is due to an underestimation of the incidence of the disease because of its association with stillbirths and early neonatal deaths. The male to female ratio in the two largest series to date varies between 0.67 and 0.77.

Etiology and Risk of Recurrence
Congenital diaphragmatic hernia can be both a sporadic and a familial disorder. The pattern of inheritance for familial cases is unknown, but a multifactorial type of inheritance has been suggested, indicating a recurrence risk for siblings of 2 percent. Familial cases have a higher male to female ratio (M:F=2.10 versus 0.67), a higher incidence of bilateral defects (20 percent versus 3 percent), and a lower incidence of life-threatening malformations than sporadic cases. However, no specific features allow identification of a familial case. Diaphragmatic hernia has been associated with Fryns syndrome, Beckwith-Wiedemann syndrome (see p. 221), Pierre Robin syndrome, and congenital choanal atresia. The defect has been associated with chromosomal defects (see section on associated anomalies).

The etiology of sporadic cases is largely unknown. Maternal ingestion of bendectin, thalidomide, quinine, and antiepileptic drugs has been reported in association with diaphragmatic hernia. In animal models, hypovitaminosis A was able to induce diaphragmatic hernia in rats. The etiologic role of diabetes is considered weak.

Embryology and Anatomy of the Diaphragm
The diaphragm is a dome-shaped septum dividing the thoracic and abdominal cavities. It consists of a central or aponeurotic segment and a peripheral or muscular one. It is formed by the fusion of four different structures: (1) the septum transversum, (2) the dorsal esophageal mesentery, (3) the pleuroperitoneal membrane, and (4) the body wall. The septum transversum is a mesodermal structure that migrates from the cranial portion of the embryo to the definitive location of the diaphragm. It gives origin to the central tendon of the diaphragm. The dorsal esophageal mesentery contributes to the median portion of the organ. The pleuroperitoneal membranes are structures that close the pleuroperitoneal cavity. Although they form a large segment of the embryonal diaphragm, their contribution to the final and fully developed diaphragm is relatively small. The participation of the body wall is limited to a narrow peripheral segment corresponding to the insertion of the muscle to the ribs and sternum (Fig. 6-6). The diaphragm is completely formed by the end of the 8th week of conceptional age. However, modeling continues throughout gestation. Expansion of the lungs results in the formation of the costodiaphragmatic recesses, which have a dome-shaped configuration.

The normal diaphragm allows the passage of organs, vessels, and nerves from the thoracic to the abdominal cavity. This is accomplished by three main orifices, allowing passage for the aorta, esophagus, and inferior vena cava (Fig. 6-7). The thoracic duct and azygos vein cross through the aortic foramen, and the vagal nerves use the esophageal foramen. The right phrenic nerves cross through the inferior vena cava orifice. Congenital diaphragmatic hernias occur when a diaphragmatic defect allows the protrusion of abdominal visceral content into the thoracic cavity. The spectrum of embryologic defects is wide, ranging from complete absence of the diaphragm through pathologic orifices (Bochdalek foramen) to congenital hiatal hernia, in which the viscera protrude through a physiologic orifice.

Congenital diaphragmatic hernias are classified according to the location of the diaphragmatic defect:

1. Posterolateral defect, or Bochdalek hernia, occurring through the primitive communication of the pleuroperitoneal canal or foramen of Bochdalek
2. Parasternal defect, or Morgagni hernia, located in the anterior portion of the diaphragm between the costal and the sternal origins of the muscle (foramen of Morgagni or sternocostal hiatus)
3. Septum transversum defects, occurring because of a defect of the central tendon
4. Hiatal hernias, occurring through a congenitally large esophageal orifice

Another pathologic entity, eventration of the diaphragm, is frequently considered with congenital diaphragmatic hernias because they have a similar pathophysiologic sequence. However, eventration of
the diaphragm consists of an upward displacement of the abdominal content into the thoracic cavity because of a congenitally weak diaphragm, which is virtually reduced to an aponeurotic sheet.

The mechanisms responsible for the origin of a diaphragmatic hernia are unknown. The two main hypotheses are (1) delayed fusion of the diaphragm and (2) a primary diaphragmatic defect. The bowel normally returns to the abdominal cavity between the 10th and 12th weeks of gestation, at which time formation of the diaphragm should be completed. Delayed closure of the communication between the abdomen and the thorax would allow part of the abdominal contents to pass into the thorax, and abdominal viscera would prevent complete closure of the diaphragm. An alternative hypothesis suggests that a primary defect occurs in the formation of the diaphragm, and this creates the condition for a subsequent migration of the abdominal organs into the thoracic cavity. The negative pressure created by fetal thoracic wall movements (fetal breathing) could be responsible for migration of the viscera into the thorax. Two observations support the first hypothesis. (1) In some cases, organs firmly attached to the abdomen in late fetal life, such as the pancreas, can be found in the thorax. This implies a precocious migration of abdominal organs into the thoracic cavity. (2) The discrepancy between the size of the defect and the dimensions of the herniated organs also suggests an early migration of the viscera and secondary partial closure of the defect.

The main cause of death of infants with congenital diaphragmatic hernia without associated anomalies is respiratory failure due to pulmonary hypoplasia. This can be easily understood if it is considered that development of the normal lung is an active process from the 5th week of gestation. The fetal lung goes through four different stages (Figs. 6-8, 6-9): (1) embryonic period, from conception to the 5th week, (2) pseudoglandular period, from the 5th to the 17th week, (3) canicular period, from the 17th to the 24th week, and (4) terminal sac period, from the 24th week until term. By the 16th week of gestation, the development of the constrictive part of the airways is completed (from trachea to terminal bronchioles). From this time until birth, respiratory airways are fully developed. This includes respiratory bronchioles and sacules. Most alveolar development is a postnatal event. Their number increases until the age of 7 years, and their size keeps up with changes in thoracic volume until maturity. The development of the pulmonary circulation follows that of the airways. The total adult complement of blood vessels irrigating the conducting portion of the airways (also known as preacinar) is completed by 20 weeks of gestation. The alveolar or intraacinar portion devel-
Knowledge of changes in the thickness and extension of the muscular component of the media is important in order to understand the development of postsurgical pulmonary hypertension. The muscle of these vessels is limited to the terminal bronchioles during fetal life. Postnatally, there is progressive extension peripherally to the respiratory bronchioles, alveolar ducts, and alveolar vessels. There is also a progressive decrease in the thickness of the muscle wall (Figs. 6-10, 6-11).

Infants with diaphragmatic hernia have profound changes in both the respiratory and the vascular components of the lungs. These are more pronounced in the ipsilateral than in the contralateral lung, depending on the timing and the degree of herniation. An early insult (before 16 weeks) could reduce the number of conducting airways. After this time, it also impairs the development of respiratory airways. After surgical correction, gains can be achieved in respiratory airways as their normal development continues postnatally. However, catchup growth cannot occur in conducting airways, and therefore, the total number of alveoli is diminished (pulmonary hypoplasia). The vascular changes in congenital diaphragmatic hernia include a reduction in the number of vessels, an increase in arterial medial wall thickness, and an extension of muscle peripherally into smaller preacinar arteries. These morphologic changes are the basis for the occurrence of pulmonary hypertension in the postoperative period, which can be triggered even by mild acidosis, hypoxia, or hypercapnia and can lead to right-to-left shunt and persistent fetal circulation. The presence of a transitory, postoperative, asymptomatic period shows that the cause of death is not lung hypoplasia but intense pulmonary vasospasm. Survival in these cases is related to surgical ligation of the ductus arteriosus and on a thorough pharmacologic treatment aimed at dilating the pulmonary vascular bed and at preventing acidosis and hypoxia.

**Pathology**

Among the previously described varieties of congenital diaphragmatic hernia, the three most common are posterolateral or Bochdalek type, parasternal or...
Figure 6-10. Bar graph showing progression of muscle in the walls of arteries within the acinus. In fetuses, there is no muscle within the acinus. With age, there is gradual extension into the acinar region, but, even at 11 years, muscular arteries have not reached the alveolar wall, where they are found in adults. (Reproduced with permission from Reid: AJR 129:777, 1977.)

Figure 6-11. Pulmonary artery structure at distal end. Complete muscle coat (muscular artery) gives way to spiral of muscle (partially muscular artery) before it completely disappears, leaving nonmuscular artery. In cross-section, vessels within the spiral region have a crescent of muscle. (Reproduced with permission from Reid: AJR 129:777, 1977.)

Morgagni type, and eventration. Hiatal hernia has little relevance for prenatal diagnosis.

Bochdalek type is the most common, accounting for 85 to 90 percent of all congenital diaphragmatic defects found in the neonatal period.\(^{11,19}\) It occurs in the left side in 80 percent of cases, in the right side in 15 percent of cases, and is bilateral in the rest.\(^{11,19,28}\) An explanation for this predominance is that the right pleuroperitoneal canal closes before the left. A true sac is present in only 5 to 10 percent of patients. The small bowel is involved in about 90 percent of cases, the stomach in 60 percent, the spleen in 54 percent, and the colon in 56 percent. Less frequently, the pancreas (24 percent), liver, adrenal glands, and even kidneys (12 percent) may be found.\(^5\) In the chest, the heart and mediastinum are shifted generally to the right. When the hernia is located in the right side, the main organs include the liver and gallbladder. Incomplete rotation of the intestine and anomalous mesenteric attachment are the rule.

Morgagni hernia accounts for only 1 to 2 percent of congenital diaphragmatic hernias.\(^{19}\) It is most frequently located in the right side and can also be bilateral.\(^{52}\) This type of hernia is considered to be the result of failure of muscle to develop from the transverse septum. The content of the hernia is usually liver; other organs, such as colon, small bowel, and stomach, may follow as well. The hernia is usually small because the liver may limit the degree of herniation. A peritoneal sac is always present, although on occasion rupture occurs, leaving little trace of the sac. Herniation into the pericardial cavity has been reported rarely. The heart may herniate through the foramen of Morgagni into the epigastric area.\(^4\)

Eventration of the diaphragm occurs in 5 percent of diaphragmatic defects.\(^{19}\) It is five times more common in the right side than in the left, and some bilateral instances have been reported. All or part of the hemidiaphragm is involved. The disorder can be considered a failure of muscularization of the diaphragm, which appears thin and lacks, either partially or totally, muscle fibers. Acquired eventration can be caused by paralysis of the phrenic nerve, and injury of this nerve during delivery may account for some cases of eventration in the neonatal period. In congenital eventration, the phrenic nerve is normal. The frequency of malrotation of the intestine in congenital eventration suggests that this disorder can occur during fetal life.

Associated Anomalies

Congenital diaphragmatic hernia has been associated consistently with a high incidence of other anomalies, excluding lung hypoplasia and gut malrotation, which are implied in the diaphragmatic hernia sequence. Three major studies have reported a remarkably similar incidence of major anomalies, ranging from 50 to 57 percent.\(^{11,19,47}\) These studies have included both stillbirths and neonatal deaths. The incidence of anomalies is much lower in survivors.\(^9,28\) Congenital diaphragmatic hernia has been considered as part of the schisis type of abnormalities, which include neural tube defects (anecephaly, cephaloceles, spina bifida), oral cleft (lip and palate),
and omphalocele. It is associated with these anomalies more frequently than is expected by chance. However, other anomalies have also been reported. Table 6-1 illustrates the associated anomalies in the largest reported series in the literature. The central nervous system is the most frequently involved. Cardiovascular abnormalities have been found in 23 percent of newborn babies with congenital diaphragmatic hernias excluding eventration. Ventricular septal defects and tetralogy of Fallot were the most common anomalies. Less frequent anomalies include coarctation of the aorta, ectopia cordis, atrial septal defects, absence of pericardium, and tricuspid atresia. Half of the infants with cardiac anomalies (5 of 11) had other extracardiac anomalies compared to those without cardiac disease (4 of 37, p < 0.05).

Congenital diaphragmatic hernia has been reported in association with chromosomal abnormalities. Hansen reported one trisomy 21, one trisomy 18, and one ring 4 in 75 newborns. Harrison reported 2 cases (trisomies 18 and 1 trisomy 21) in 143 patients. Boles and Anderson found 3 genetic anomalies (including 2 Down syndrome babies) in 58 infants. Other reports have suggested an association between trisomy 21 and Morgagni congenital diaphragmatic hernia. It is controversial whether eventration of the diaphragm is associated with a higher incidence of trisomy 18.

### Diagnosis

The diagnosis of congenital diaphragmatic hernia has been made in utero several times. A definitive diagnosis can be made if abdominal organs are seen in the thoracic cavity. Visualization of fluid-filled bowel at the level of the four chamber view of the heart is diagnostic (Figs. 6-12, 6-13, 6-14). The ribs and the inferior margin of the scapula can be used as landmarks when trying to establish the intrathoracic location of viscera. The visualization of these organs in the chest is difficult in early pregnancy. In late pregnancy, they can be seen as fluid-filled cystic structures that may peristalt in the thoracic cavity.

We have found that the most sensitive sign of the presence of a congenital diaphragmatic hernia is a shift in the position of the heart within the chest. Polyhydramnios is common and thought to be secondary to bowel obstruction. However, this mechanism does not explain the presence of polyhydr...
nios in right hernias or left hernias in which intestinal transit is not impaired.

The diagnosis of right diaphragmatic hernia is extremely difficult because of the similar echogenicity of liver and lung. A helpful hint is the identification of the gallbladder, which is frequently herniated into the chest. The presence of a pleural effusion and ascites may also aid in the differentiation between lung and liver. The mechanism for fluid accumulation in serous cavities is thought to be related to an obstruction of venous return. We made the diagnosis of right diaphragmatic hernia in one patient with a
duodenal atresia by realizing that there was malrotation of the stomach (see Fig. 7.5).

Small congenital diaphragmatic hernias may not be detected in utero. Presumably, these would have a better prognosis. In cases of uncertainty, other diagnostic maneuvers, such as amniography (Fig. 6-15) and computed tomography, can be used. This would be helpful in the differential diagnosis of cystic lesions of the chest, such as cystic adenomatoid malformation of the lung and mediastinal cystic processes (neuroenteric cysts, bronchogenic cysts) (see Table 5.1). The presence of a normally placed stomach can help to distinguish these two conditions.

A diagnosis of diaphragmatic hernia mandates a careful examination of the fetal anatomy.

Prognosis
The prognosis of congenital diaphragmatic hernia is poor. Infants with congenital diaphragmatic hernia are at risk for antepartum and neonatal death. In a retrospective review of such infants, 35 percent were stillbirths. Associated anomalies are thought to be responsible for most antenatal deaths, and they were present in 90 percent of stillbirths. An earlier study noted that abnormalities severe enough to account
for death were most frequently located in the CNS (anencephalus, iniencephalus, Dandy-Walker malformation, Arnold-Chiari malformation). The other frequent location of lethal anomalies is the cardiovascular system. A significant number of infants die within the first few hours of life. In Butler's series, 35 percent of all neonatal deaths occurred 1 hour after birth. The causes of immediate neonatal death are severe pulmonary insufficiency and associated congenital anomalies. Of infants who survive the immediate neonatal period and go to surgery, the mortality rate varies between 29 percent and 53 percent. The following are prognostic factors.

**Age in Hours at Time of Operation.** Infants who clinically manifest respiratory failure immediately after birth have a poor prognosis. In fact, the survival rate is almost 100 percent in infants operated on 24 hours after birth, but it drops to 38 to 64 percent in infants less than 24 hours old. Unfortunately, most of the babies treated surgically are in the latter group.

**Blood Gas Data.** Infants with acidosis, hypercapnia, and hypoxemia have a poor prognosis. The predictive value of blood gas determinations does not seem to be purely dependent on the volume of lung parenchyma, since no correlation has been found between lung weight and blood gas data in dead infants.

**Chest X-ray Data.** The presence of a pneumothorax, location of the stomach above the diaphragm, and volume of aerated ipsilateral and contralateral lungs have prognostic value. On the other hand, the degree of mediastinal shift and the amount of visceral distention have not been found to be statistically different between surviving and nonsurviving groups.

**Associated Anomalies.** The presence of additional major malformations, excluding gut malrotation and patent ductus arteriosus, which are often part of the disease process, portends a poorer outcome. In one series, the postsurgery mortality rate was significantly higher (p < 0.02) in infants with cardiac abnormalities (73 percent) than in those without cardiac disease (27 percent). In another series, the death rate among infants with associated serious congenital anomalies was higher (70.6 percent) than the overall mortality rate (36 percent). After excluding infants with associated anomalies, fetuses with congenital diaphragmatic hernia do not seem to be at higher risk for preterm delivery and intrauterine growth retardation.

Overall, the prognosis for infants with congenital diaphragmatic hernia diagnosed in utero is extremely poor. Harrison reported a mortality of 100 percent in nine consecutive patients. We have made 10 prenatal diagnoses of diaphragmatic hernia and have had only 1 survivor, in whom diagnosis was made at 23 weeks of gestation. The poor prognosis for infants diagnosed in utero may reflect a selection bias, as smaller hernias may not be detectable with ultrasound. Abnormally low lecithin:sphingomyelin (L:S) ratios have been reported in infants with congenital diaphragmatic hernia. Such observation has been attributed to decreased surfactant production by a hypoplastic lung. However, it has not been established whether a mature L:S ratio has prognostic value for infants diagnosed in utero.

One study provided information in long-term follow-up of 21 infants with congenital diaphragmatic hernia operated on within 24 hours of life. Infants were followed for an average of 8 1/2 years, and they were found to be in good health, vigorous, active, and without evidence of growth failure. Although some infants had respiratory problems in the first 3 to 4 years of life (2 had pneumonia, 2 had recurrent upper respiratory infections, I was said to have "chronic asthma"), all complaints subsequently disappeared. Other authors have performed pulmonary function tests in infants with congenital diaphragmatic hernia and have found residual defects in ventilatory functions and impaired blood flow to the lung in the side of the hernia, but normal airway resistance and distribution of ventilation. Interpretation of some long-term follow-up studies is difficult because they have included infants operated on at different ages. In one study involving 30 infants, 3 had evidence of mental retardation that was attributed to hypoxia after birth, although no evidence was presented to support this contention.

**Obstetrical Management.** Before viability the option of pregnancy termination should be offered to the parents. Karyotype determination and echocardiography should be performed in each infant in whom a diagnosis of diaphragmatic hernia is made. There are no data to justify delivery before fetal maturity or to alter the mode of delivery. Labor should occur in a tertiary center where a neonatologist and a pediatric surgeon are immediately available.

Diaphragmatic hernia is one of the conditions for which there is solid experimental basis for in utero surgery. An animal model for this condition has been developed in the fetal lamb by inflating an intrathoracic balloon or by producing a dia-
hemodynamic changes observed in the lamb are similar to those seen in affected human fetuses. The ventilatory and hemodynamic changes observed in the lamb are similar to those seen in affected human fetuses. Furthermore, intrauterine correction by deflation of the intrathoracic balloon or by closure of the defect has been associated with a favorable outcome. This procedure has been attempted in the human fetus at the time of this writing, with poor results.

REFERENCES

36. Harrison MR, Bjordal RJ, Langmark F, et al.: Congen-


Omphalocele

SYNONYM
Exomphalos.

DEFINITION
Omphalocele is a ventral wall defect characterized by herniation of the intraabdominal contents into the base of the umbilical cord, with a covering amnioperitoneal membrane (Fig. 6-16). Pentalogy of Cantrell is a term used to describe the association of five anomalies: (1) midline supraumbilical abdominal defect, (2) defect of the lower sternum, (3) deficiency of the diaphragmatic pericardium, (4) deficiency of the anterior diaphragm, and (5) intracardiac abnormality.

The Beckwith-Wiedemann syndrome is characterized by the association of macroglossia, visceromegaly, and omphalocele.

INCIDENCE
Omphalocele is estimated to occur in 1 in 5800 to 1 in 5130 live births. Beckwith-Wiedemann syndrome has an incidence of 1 in 13,700 live births. Pentalogy of Cantrell is a very rare disorder.

ETIOLOGY
Most cases of omphalocele are sporadic. Often the condition is associated with chromosomal aberrations, such as trisomies 13 and 18.

The familial occurrence of this anomaly with a sex-linked or autosomal pattern of inheritance has been reported. The recurrence risk for isolated omphalocele cases appears to be less than 1 percent. However, when an omphalocele is identified in association with trisomies, a careful evaluation of the karyotype should exclude the possibility of a balanced translocation, which increases the recurrence risk.

Although most cases of Beckwith-Wiedemann syndrome are sporadic, familial occurrence has been described, suggesting autosomal dominant, recessive, sex-linked, and polygenic patterns. Cantrell’s pentalogy is also a sporadic condition.

EMBRYOLOGY
The development of the anterior abdominal wall depends on the fusion of four ectomesodermic folds (cephalic, caudal, and two laterals). Failure of the cephalic fold to fuse with the other folds usually results in the association of omphalocele with ectopia cordis and sternal and diaphragmatic defects. Failure of the lateral folds to meet in the midline (between the 3d and 4th weeks) leads to the formation of an isolated omphalocele. Defective fusion of the caudal fold results in cloacae exstrophy of the bladder.

PATHOLOGY
The defect is located in the midline, and the protrusion of the intraabdominal contents occurs through the base of the umbilical cord. Bowel loops, stomach, and liver are the most frequently herniated organs and are covered by a membrane made up of two layers: internally the peritoneum and externally the amnion. The umbilical cord inserts into the sac. The size of these defects is variable, ranging from a very small hernia containing a few bowel loops to a very large mass containing most of the visceral organs.

With pentalogy of Cantrell, the diaphragmatic defect is embryologically different from the hernia of Morgagni. The lesion is located in the anterior portion of the diaphragm and is rarely associated with

Figure 6-16. Typical omphalocele in a newborn. The lesion occurs in the midline and is covered by a membrane. (Courtesy of Dr. Robert Touloukian.)
Figure 6-17. Prenatal diagnosis of an omphalocele at 15 weeks of gestation. Transverse scan of the abdomen at the level of the umbilicus demonstrating the lesion (*). Sp, spine.

Diagnosis

The diagnosis of omphalocele relies on the demonstration of a mass adjacent to the anterior ventral wall representing the herniated visceral organs (Figs. 6-17 through 6-20). Differential diagnosis is mainly with gastrochisis. Omphaloceles are located in the midline. The umbilical cord enters into the hernia, and the herniated organs are covered by a membrane that is continuous with the umbilical cord. Gastrochisis is a lateral defect, devoid of a surrounding membrane and separated from the umbilical cord insertion. These signs permit an accurate differential diagnosis in almost all patients. A possible exception may be those cases of omphalocele in which rupture of the amnioperitoneal sac occurs in utero, an exceedingly rare complication. Pentalogy of Cantrell can be suspected in the presence of ectopia cordis. In many cases, the defect in the diaphragm at the level of the pericardium is small and cannot be demonstrated by prenatal sonography. Suspicion should arise when the apex of the heart deviates inferiorly (under nor-

Associated Anomalies

The frequency of trisomies in infants with omphalocele varies between 35 and 58 percent. Chromosomal aberrations include trisomy 13 and 18. Car-

* Our review of the literature indicates that this syndrome accounts for 4 percent of all cases of omphalocele.
Figure 6-19. Omphalocele in a third trimester fetus. The herniated organs are the liver (L) and bowel (B). The amniotic peritoneal membrane is seen (arrows). hv, hepatic vein.

Figure 6-20. Small omphalocele in a fetus with hydrops. The insertion of the umbilical cord (uc) in the abdomen is interrupted by a lesion that contains small bowel (b). The amniotic peritoneal membrane is outlined by the short arrows. Ascites is present in the peritoneal cavity. B, intraabdominal bowel loops.

nal circumstances, the heart is horizontal within the thoracic cavity) and bulges under the skin of the chest due to the sternal defect.

A specific prenatal diagnosis of Beckwith-Wiedemann syndrome has not been reported. The condition should be suspected when an omphalocele is associated with visceromegaly and macroglossia. Nomograms are available for assessing the size of kidneys, heart, and spleen. However, the value of these measurements in the diagnosis of Beckwith-Wiedemann syndrome has not been tested. This condition has been prenatally visualized twice, but a specific diagnosis of Beckwith-Wiedemann syndrome was not made before birth. Polyhydramnios is a frequent finding and is probably responsible for the increased incidence of premature labors.

Ventral wall defects may result in an elevation of the maternal serum alpha-fetoprotein (MSAFP). It has been reported that MSAFP screening has a sensitivity of 52 percent in the detection of anterior wall defects and 42 percent for omphalocoeles. Therefore, the evaluation of the anterior abdominal wall is an important part of the monographic assessment of pregnancies with elevated AFP. Since an increased incidence of neural tube defects has been found in infants with omphalocele, identification of the latter lesion should not result in overlooking the fetal spine.

Prognosis

A small defect can be repaired in a one-stage operation. Larger defects usually require a two-stage operation, generally using a Silastic or Teflon membrane to cover and reduce the herniation of the intraabdominal organs. Prognosis of omphalocele depends largely on the presence of associated anomalies. Losses are mainly due to cardiac abnormalities, chromosomal aberrations, and prematurity. Kirk and Wah have reported 38 cases of omphalocoeles, with a mortality rate of 29 percent. Of the 11 deaths, 5 occurred in infants with multiple congenital anomalies and 3 in infants with associated congenital heart disease. Carpenter et al. reported an overall mortality rate of 40 percent. Among the infants who died, two had trisomies, two had other severe anomalies (esophageal atresia, tetralogy of Fallot, pulmonary hypoplasia), and three had pentalogy of Cantrell.

Cephalic fold defects carry a worse prognosis than do the lateral and caudal fold defects. In the series by Carpenter et al., the mortality rate among cephalic fold defects was 78 percent versus 19 percent in the lateral fold defects. In a review of the literature by Toyama, the survival rate among individuals with pentalogy of Cantrell was only 20 percent. Since this report is based on data collected before 1970, it is quite likely that the outcome for infants with this condition has improved due to advances in cardiothoracic surgery.

Infants with Beckwith-Wiedemann syndrome have respiratory and feeding difficulties because of the large tongue. The oral cavity may grow with time, and the tongue eventually fits inside the mouth. In some patients, glossectomy has been required. The excessive rate of growth often slows down after the first few years. Neonatal hypoglycemia is a serious complication, which has been implicated in the mild to moderate mental deficiency noted in some of these infants. Steroids have been used to control hypoglycemia. Ten percent of these infants develop neoplasms. In a review of 17 cases of Beckwith-
Wiedemann syndrome associated with tumors, a 47 percent mortality rate was reported.26

Obstetrical Management
Identification of a fetal omphalocele should prompt a careful search for associated anomalies. Fetal karyotyping18 and echocardiography are indicated. Diagnosis before viability may allow parents to opt for pregnancy termination. In continuing pregnancies and in those cases diagnosed after viability, serial ultrasound monitoring is indicated to look for signs of intestinal obstruction and intrauterine growth retardation. In recent years, the optimal mode of delivery of fetuses with omphalocele has been a subject of debate. Some authors have suggested that delivery of these infants should be performed by cesarean section to avoid birth injury with rupture of the amnioperitoneal sac. However, in two large retrospective and uncontrolled series, no benefits of cesarean section versus vaginal delivery could be documented.2,3,10

The major limitation of these studies is their design. The incidence of complications in infants delivered vaginally seems to be small. However, we believe that there is a sub group of patients who may benefit from a cesarean section. An example of this is the large omphalocele with external protrusion of a large part of the liver.1 A careful search for associated anomalies is indicated also in those cases diagnosed in the third trimester. Omphalocele may be accompanied by anomalies incompatible with life (e.g., trisomies 13 and 18), and the recognition of such disorders could alter obstetrical management.

REFERENCES
Gastroschisis

Definition
Gastroschisis is a paraumbilical defect of the anterior abdominal wall associated with evisceration of abdominal organs.

Incidence
The incidence of gastroschisis ranges from 1:10,000 to 1:15,000 live births.1,2,8

Etiology
Most cases are sporadic. Familial occurrence has been documented in five families.1,9,10,13,15 The findings in two families indicate the possibility of an autosomal dominant inheritance, with variable expressivity.

Embryology and Pathogenesis
Gastroschisis is a defect resulting from vascular compromise of either the umbilical vein or the omphalomesenteric artery.5,6

Human embryos initially bear both left and right umbilical veins. Involution of the right umbilical vein occurs between the 28th and the 32d day after conception. Premature involution may lead to ischemia and to the resultant mesodermal and ectodermal defects.

The omphalomesenteric arteries (OMAS) branch from the primitive dorsal aorta and extend to the right along the omphalomesenteric duct toward the yolk sac. The left OMA involutes, whereas the right one is transformed into the superior mesenteric artery. The terminal portion extends into the extraembryonic coelom, which is now located in the body stalk. Disruption of the distal segment could result in right-sided periumbilical ischemia and the paramedian defect characteristic of gastroschisis. Ischemic injury to the territory of the superior mesenteric artery may account for the high incidence of jejunal atresia found in association with gastroschisis.5,6,11

Pathology
Gastroschisis is characterized by a full-thickness defect of the abdominal wall, usually located to the right of the umbilical cord, which has a normal insertion. The defect in the abdominal wall is generally quite small (3 to 5 cm). The herniated organs include mainly bowel loops covered by an inflammatory exudate possibly resulting from chemical irritation by exposure to amniotic fluid. They appear edematous and are not protected by a membrane. Hepatic herniation is less frequent with gastroschisis than with omphaloceles. Meconium is frequently found in the amniotic fluid of these fetuses. Its presence probably reflects intestinal irritation.2,14 At birth, infants have low serum albumin and total protein levels, which probably indicate intestinal chronic sclerosing peritonitis.

Associated Anomalies
In contrast to omphalocele, gastroschisis is not associated with an increased incidence of other anomalies. However, in 25 percent of patients, gastrointestinal problems secondary to the vascular impairment and adhesions are found, including bowel malrotation, atresia, and stenosis.3 Intrauterine growth retardation has been reported in up to 77 percent of infants.2

Diagnosis
The diagnosis of gastroschisis relies on the demonstration of a mass adjacent to the anterior ventral wall representing the herniated visceral organs. Differential diagnosis from omphalocele can be made in almost all cases because, in gastroschisis, (1) the defect is usually located in the right paraumbilical area, (2) the umbilical cord is normally connected to the abdominal wall, and (3) the herniated organs are not covered by a membrane but float freely in the amniotic cavity (Figs. 6-21, 6-22). An omphalocele is a central defect surrounded by a membrane on which the umbilical cord is inserted.12 A possible exception may be those cases of omphalocele in which rupture of the amnioperitoneal sac occurs in utero, an exceed-

Figure 6-21. Newborn with gastroschisis. There is no covering membrane, and the defect is on the right paraumbilical area. (Courtesy of Dr. Robert Touloukian.)
GASTROSCHISIS

225

Figure 6-22. Sagittal scan of a 20-week fetus with a gastroschisis. Bowel is seen floating in the amniotic cavity (curved arrow). The normal insertion on the umbilical vessels is demonstrated. UC, umbilical cord inserting into the anterior abdominal wall; Sp, spine.

ingly rare complications. Polyhydramnios is a frequent finding, and it is probably related to impaired gastrointestinal transit.

Ventral wall defects may result in elevation of the MSAFP. It has been reported that MSAFP screening will identify 77 percent of these fetuses. Therefore, examination of the anterior abdominal wall is an important part of the monographic evaluation of pregnancies with elevated AFP.

Prognosis
In three different series, the mortality rate ranged from 7.6 percent to 28 percent. Death was caused by prematurity, sepsis, and intraoperative complications. In one series, herniation of the liver was associated with a higher mortality rate (50 percent versus 7 percent).

Obstetrical Management
The critical issue is the differential diagnosis from an omphalocele, since this defect is associated with a higher incidence of associated anomalies and carries a worse prognosis. In the past, the consensus was that fetal karyotyping was not indicated in gastroschisis. We have recently seen two cases with karyotype abnormalities. When the diagnosis is made before viability, the parents may opt for termination of pregnancy. In continuing pregnancies and in those cases diagnosed in the third trimester, serial ultrasound examinations are recommended to detect intrauterine growth retardation and early signs of bowel obstruction. Polyhydramnios is a frequent finding and may contribute to the onset of premature labor. In this situation, tocolytic agents and amniotic fluid drainage are indicated to prolong the gestation.

In recent years, the optimal mode of delivery of fetuses with ventral wall defects has been a subject of debate. Some authors have suggested that delivery of these infants should be performed by cesarean section to avoid birth injury to the herniated visceral organs. However, in two large series, no benefits of cesarean section versus vaginal delivery could be documented. The major limitation with such studies is their retrospective and uncontrolled design. The incidence of complications in infants delivered vaginally seems to be small. Delivery in a tertiary care center is recommended.

REFERENCES
Body Stalk Anomaly

Definition
This is a severe abdominal wall defect due to failure of formation of the body stalk and characterized by absence of the umbilicus and umbilical cord.

Incidence
The incidence of body stalk anomaly is 1 in 14,273 births.²

Embryology
During the third week of embryonic life the flat trilaminar embryo is transformed into a cylindrical fetus by a parallel set of contiguous body folds: cephalic, lateral, and caudal. Folding separates the intraembryonic coelom (peritoneal cavity) from the extraembryonic coelom (chorionic cavity). The amnion then fuses with the chorion peripherally and forms the covering of the umbilical cord centrally. Body stalk anomaly results from severe maldevelopment of cephalic, caudal, and lateral embryonic body folds. The failure of complete extraembryonic coelom obliteration accounts for the absence of umbilical cord formation and the wide-based insertion of the amnioperitoneal membrane onto the placental chorionic plate. The intraabdominal contents persist in the extraembryonic coelom. Fusion of the amnion and chorion takes place only at the margin of the placenta.

Pathology
The abdominal organs lie in a sac outside the abdominal cavity (Fig. 6-23). This sac is covered by amnion and placenta and is attached directly to the placenta.

Figure 6-23. Body stalk anomaly. The placenta is attached to the herniated viscera without an intervening umbilical cord.

Figure 6-24. The umbilical vessels run along the wall of the sac.
The umbilical vessels are short and run along the sac walls (Fig. 6-24). The absence of the umbilical cord results in the fetus lying directly against the placenta and the uterine wall. This may lead to malposition and skeletal deformities, including kyphosis and scoliosis (Fig. 6-25).

**Etiology**

The disorder is sporadic in most cases, but concordance for this anomaly has been noted in twins.²

**Diagnosis**

This diagnosis should be suspected when the fetus is attached to the placenta and uterine wall and a large defect of the anterior abdominal wall allows protrusion of the viscera (Fig. 6-26). Prenatal diagnosis of this condition has been reported using MSAFP and ultrasound.¹²

**Associated Anomalies**

Anomalies, including neural tube defects, intestinal atresias, genitourinary and skeletal defects, anomalies of the chest wall, pericardium, heart, liver, and lungs, are nearly always present. One umbilical artery is usually absent.³

**Prognosis**

Potter and Craig³ and Mann et al.² have stated that this condition is uniformly fatal.²³

**Obstetrical Management**

The option of pregnancy termination for this uniformly lethal condition could be offered to the parents anytime the diagnosis is made.

**REFERENCES**

Bladder Exstrophy and Cloacal Exstrophy

Definition
Exstrophic anomalies are a group of disorders derived by a maldevelopment of the caudal fold of the anterior abdominal wall. In bladder exstrophy, the anterior wall of the bladder is absent, and the posterior wall of this organ is exposed. Cloacal exstrophy is a more complex anomaly in which there is involvement of both the urinary and intestinal tracts caused by a defect in the formation of the urorectal septum.

Incidence
Exstrophy of the bladder occurs in 1:30,000 deliveries, with a male to female ratio of 2.3:1. Exstrophy of the cloaca has an incidence of 1:200,000 live births without a sex preponderance.

Etiology
Most cases of bladder exstrophy are sporadic. Familial cases have been reported, and the risk of recurrence in a given family is 1 percent. The risk of having an affected offspring if one parent has bladder exstrophy is 1 in 13 live births, which is 500 times greater than the risk of the general populations. Presumably, cloacal exstrophy is also a sporadic disorder, although affected individuals have not reproduced.

Embryology
The cloaca is a blind pouch that receives the midgut and the allantoic duct. The anterior wall of the cloaca is formed by the cloacal membrane, which extends from the two lateral mesodermal ridges to the body stalk (the primordium of the umbilical cord). By the 6th week of conception, the cloaca is divided by a proliferating mesodermal ridge (the urorectal septum) into a urogenital sinus anteriorly and a hindgut posteriorly. The urorectal septum divides the cloacal membrane into an anterior portion, or urogenital membrane, and a posterior one, or anal membrane (Fig. 6-27).

Normally, the two mesodermal ridges fuse in the midline to form the genital tubercle, and the cloacal membrane retracts downward toward the perineum. The lower portion of the anterior abdominal wall is reinforced by the tissues derived from the mesodermal ridges (Fig. 6-28A). If the cloacal membrane does not retract normally, the two mesodermal ridges fuse inferiorly, and the cloacal membrane becomes the anterior wall of the bladder. By the 9th week, the cloacal membrane disappears and the posterior wall of the bladder is exposed, giving rise to bladder exstrophy (Figs. 6-28B, 6-29). If the membranes disappear before the urorectal septum divides the primitive cloaca, both bladder and rectum will be exposed, leading to cloacal exstrophy (Fig. 6-30).

Figure 6-27. A-D. Developmental changes of the cloaca and cloacal membrane in the 4 to 16 mm embryo. Arrows show the direction of growth of the urorectal septum. (Reproduced with permission from Muecke: In Campbell’s Urology. Philadelphia, Saunders, 1986, pp 1856-1880.)
Figure 6-28. Schematic view of regression of cloacal membrane and formation of the primitive phallus. A. Normal sequential events. B. Genesis of the exstrophy group of anomalies by a persistent cloacal membrane impeding mesodermal flow. The paired genital folds fuse inferiorly, carrying the thin cloacal membrane along the anterior surface of the enlarging phallus. A weak, membranous anterior body wall persists, leading to the eventual catastrophic event of exstrophy. (Reproduced with permission from Muecke: In Campbell's Urology. Philadelphia, Saunders, 1986, pp 1856-1880.)

Pathology
In bladder exstrophy, there are protrusion of the posterior vesical wall, separation of pubic bones, low set umbilicus, incomplete descent of the testes, short penis pointing upward and epispadias in males, and cleft clitoris in females (Fig. 6-31). The size of the everted bladder is quite variable, ranging from a small area of the trigone to complete eversion of the posterior wall of the organ. The perineum is short and broad. Divergent elevator ani- and puborectal muscles may result in rectal incontinence and anal prolapse.

In cloacal exstrophy, there are two hemibladders each with its own ureteral orifice, separated by an area of intestine. This bowel mucosa probably corresponds to the cecum, since it receives the ileum superiorly. Other structures that can be observed include an umbilical hernia and diphallus (separation of the two corpora) (Figs. 6-32, 6-33).

Associated Anomalies
Associated anomalies are rare in bladder exstrophy. In contrast, associated anomalies are very frequent in cloacal exstrophy. Renal anomalies (renal agenesis, hydronephrosis, multicystic kidney, hydrourater, ureteric atresia) are present in 60 percent of patients. Skeletal defects other than separation of pubic bones were noted in 72 percent of patients. Spina bifida is by far the most common. Anomalies of the cardiovascular and gastrointestinal tract occur in 16 percent and 10.5 percent, respectively. Omphaloceles are present in 87 percent of patients. A double vena cava is also frequent.

Diagnosis
The prenatal diagnosis of bladder exstrophy has been reported. This condition was suspected because of the presence of a solid mass (47 mm in diameter) in the lower part of the fetal abdomen (Fig. 6-34). The mass did not contain cystic areas, and the bladder could not be identified. The penis was extremely short, and a normal amount of amniotic fluid was present. The diagnosis should be suspected any time a bladder cannot be visualized in a fetus with a

Figure 6-29. Diagram of events leading to classic exstrophy. (Reproduced with permission from Muecke: In Campbell's Urology. Philadelphia, Saunders, 1986, pp 1856-1880.)
normal amount of amniotic fluid. The differential diagnosis includes omphalocele, gastroschisis, and cloacal exstrophy. Visualization of a normal bladder and the relationship of the mass to the fetal abdomen are helpful hints in the differentiation of bladder exstrophy from the first two defects. A prenatal diagnosis of cloacal exstrophy with ultrasound has not been reported. Elevated amniotic fluid AFP has been reported in one case of exstrophy cloaca.1

Prognosis

The main problems of bladder exstrophy are urinary incontinence, presence of an abdominal wall defect, and the cosmetic consequences of the lesion in the male genitalia. These problems can be treated surgically by the performance of a primary bladder closure, reconstruction of the bladder neck, and epispadias repair. Alternatively, the approach may consist of a urinary diversion, cystectomy, and epispadias repair. An optimal surgical treatment between these two alternatives has not been established, and there are merits to both approaches. The genital defects in the male are quite serious, and sex reassignment may be required in 1 in 50 to 1 in 100 patients in whom an adequately functional penis cannot be created.5 Genital defects in the female are less complex. Approximation of the hemiclitoris can generally be accomplished. Vaginal dilatation and perineoplasty may be required for satisfactory sexual intercourse.

Patients with bladder exstrophy grow into adulthood and have an acceptable social adjustment.7,13 Fertility is decreased in both males and females.10 Pregnancy is possible, although there is an increased likelihood of uterine prolapse postpartum.6 This complication is due to hypoplasia of the cardinal ligaments.

Cloacal exstrophy is a very serious anomaly associated with a 55 percent mortality rate.3 The most common associated anomaly is a neural tube defect. Untreated infants frequently die from sepsis, short bowel syndrome, or renal or central nervous system defects. The correct surgical approach consists of a series of operations including repair of the omphalocele, functional bladder closure (neonatal period), antiincontinence and antireflux surgery at age 2 to 3 years, and vaginal reconstruction at age 14 to 18 years. Tank and Lindenauer2 have recommended the conversion of males to females because of the
uniformly disappointing results when trying to create a functionally acceptable penis. Gonadectomy is performed when sexual reassignment has been selected.12

Obstetrical Management
If a diagnosis of bladder exstrophy is suspected before viability, the option of pregnancy termination should be offered. After this point, no change in standard obstetrical management is required. There is no evidence that altering the mode of delivery changes the prognosis for these infants. After birth, the exposed bladder mucosa is very friable and easily denuded. Jeffs and Lepor3 have recommended that the umbilical cord be tied closely to its area of insertion so that a long cord does not add trauma to the bladder mucosa. It is preferable to tie the cord with a suture as opposed to the standard clamp, which may also traumatize the defect. The bladder mucosa may be covered with a nonadherent film of plastic wrap to prevent the mucosa from sticking to clothing. The traditional petroleum jelly gauze should be avoided, since it may lift the bladder epithelium when removed.

REFERENCES


