

When ultrasonography reveals a fetal abdominal wall defect

Although these fetal defects are rare, be alert to their potential presence when early ultrasonography indicates structural abnormalities. Here, surveillance, planning, and appropriate patient counseling are reviewed.

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CASE Fetal anomalies detected on ultrasonography

A 34-year-old woman (G2P1) at 19 weeks' gestation presented for fetal anatomy ultrasonography evaluation. Ultrasonography demonstrated fetal demise with fetal size less than dates, oligohydramnios, and what appeared to be a full-thickness herniation of the thoracic and abdominal contents. Due to the positioning of the fetus and the oligohydramnios, the fetus appeared to have ectopia cordis and herniated liver and bowel; the bladder was not visualized. The patient was counseled regarding the findings and the suspected diagnosis of pentalogy of Cantrell. After counseling, the patient expressed desire to bury the fetus intact

according to her religious custom. She underwent a successful uterine evacuation with misoprostol administration and delivered a nonviable fetus that had a closed thoracic cage without ectopia cordis. Key findings were a very short 2-vessel umbilical cord without coiling that was tethered to the intra-abdominal organs, "pulling" the internal organs out of the abdomen, and lack of an anterior abdominal wall (**FIGURE 1**). Given these findings, a final diagnosis of body-stalk anomaly was made.

Fetal abdominal wall defects (AWDs) encompass a wide array of congenital defects, although they all involve herniation of 1 or more intra-abdominal content through a ventral abdominal defect.¹ Overall, the estimated incidence of AWDs is approximately 6 per 10,000 births.¹ Gastroschisis and omphalocele are the most common of these defect types.²

The majority of AWDs can be diagnosed during the first trimester of pregnancy via ultrasonography; however, during the first trimester the physiologic midgut herniation resolves by 12 weeks of gestation. It is therefore important to repeat imaging at a later gestational age to confirm the suspicion. Furthermore, the differential diagnosis should include the relatively benign condition of umbilical hernia.

While many AWDs share similarities, they differ significantly in prognosis and management. Early detection is therefore



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crucial for fetal surveillance, prenatal testing, perinatal planning, and patient counseling (TABLE, page 36). In this article, we outline antenatal surveillance and management of AWDs based on recommendations from the American College of Obstetricians and Gynecologists and the Society for Maternal-Fetal Medicine as well as on our experience and practice.

Gastroschisis is an increasingly prevalent AWD

Gastroschisis is a full-thickness, ventral wall defect that results in bowel evisceration; it typically occurs to the right of the umbilical cord insertion.³ It is one of the most common AWDs and its prevalence has increased in the past few decades, from 2 to 3 cases per 10,000 live births in 1995 to as high as 6 cases per 10,000 live births in 2011.^{2,4,5}

The cause of gastroschisis remains unclear. The main theory is that there is an ischemic disruption of the closure of the abdominal wall at or near the omphalomesenteric artery or the right umbilical vein.^{6,7} In addition, investigators have reported an increased incidence of gastroschisis in mothers exposed to cigarette smoking and certain medications, such as pseudoephedrine, salicylates, ibuprofen, and acetaminophen.^{8,9}

Making the diagnosis

Prenatal diagnosis using ultrasonography is possible at around 10 weeks of gestation. As previously mentioned, however, physiologic herniation of the midgut must be excluded by performing follow-up imaging at a later gestational age. In our practice, we typically do this at around 16 weeks of gestation.

Ultrasonographic features of gastroschisis include loops of bowel herniating through a small paraumbilical wall defect (usually 2–3 cm) floating in amniotic fluid without a covering membrane⁴ (FIGURE 2, page 38). Direct exposure to amniotic fluid causes small bowel inflammation and fibrin deposition, leading to a thickened, echogenic appearance. Polyhydramnios and intra-abdominal bowel dilation have been associated with the presence of intestinal atresia.¹⁰



FIGURE 1 Fetus with body-stalk anomaly



This fetus had a very short 2-vessel umbilical cord without coiling tethered to the intra-abdominal organs and lacked an anterior abdominal wall. The final diagnosis was body-stalk anomaly.

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TABLE Overview of antenatal surveillance and management of fetal abdominal wall defects

Abdominal wall defect	Imaging findings	Associated structural abnormalities	Associated chromosomal abnormalities	Fetal evaluation	Delivery timing/method	Survival rate
Gastroschisis	<p>Full thickness ventral wall defect</p> <p>Typically to the right of the umbilicus</p> <p>No membrane covering herniated contents</p> <p>May contain the liver</p>	<p>Increased incidence of IUGR (25%)</p> <p>Intestinal atresia (10%–15%)</p>	Usually none	<p>Genetic testing</p> <p>Fetal echocardiography</p> <p>Serial fetal growth ultrasonography</p> <p>NST/BPP weekly starting at 32 weeks</p>	<p>No consensus: vaginal versus cesarean delivery</p> <p>If significant liver involvement, some pediatric surgeons recommend cesarean delivery due to the risk of hepatic rupture</p>	91%–94%
Omphalocele	<p>Midline abdominal herniation through the base of the umbilical cord</p> <p>Covered by a membrane</p> <p>May contain liver (80% of cases)</p>	<p>Cardiac defects</p> <p>Gastrointestinal defects</p> <p>Genitourinary defects</p> <p>Neural tube defects</p> <p>Diaphragmatic defects</p> <p>Orofacial clefts</p> <p>Polyhydramnios is common</p> <p>Associated with IUGR</p>	<p>30% have chromosomal abnormalities (trisomy 13, 18, 21)</p> <p>Small (<5 cm) lesions without hepatic involvement are associated with a fetal aneuploidy</p> <p>Large (≥5 cm) liver containing lesions are associated with euploid fetuses</p> <p>Associated with Beckwith-Wiedemann syndrome</p>	<p>Genetic testing</p> <p>Fetal echocardiography</p> <p>Beckwith-Wiedemann syndrome testing</p> <p>Serial fetal growth ultrasonography</p> <p>NST/BPP weekly starting at 32 weeks</p>	<p>Timing: individualized expectant management until spontaneous labor, other indication for delivery, or at least 39 weeks</p> <p>Method: no evidence-based guidelines for term fetuses</p> <p>Large defects are at risk of rupture</p> <p>Preterm induction of labor not advised as approximately 50% neonatal mortality rate</p>	<p>Isolated defects 50%–90%</p> <p>Presence of aneuploidy and/or other anomalies is strongly associated with a poor prognosis</p>
Body-stalk anomaly	<p>2 of the 3:</p> <p>Exencephaly or encephalocele with facial clefts</p> <p>Large anterior abdominal wall defect</p> <p>Limb defects</p>	<p>A short or absent umbilical cord</p> <p>Severe kyphosis or scoliosis</p> <p>Oligohydramnios</p> <p>Increased nuchal translucency</p>	None	<p>Genetic testing</p> <p>Fetal complete anatomical evaluation</p>	<p>Elective termination of pregnancy often is advised</p> <p>Women who continue pregnancy are at increased risk of preterm labor and gestational hypertension</p>	<p>Nearly incompatible with life</p> <p>Only case reports of surviving neonates</p>

TABLE Overview of antenatal surveillance and management of fetal abdominal wall defects (continued)

Abdominal wall defect	Imaging findings	Associated structural abnormalities	Associated chromosomal abnormalities	Fetal evaluation	Delivery timing/method	Survival rate
Pentalogy of Cantrell	Anterior chest wall, cardiac, pericardial, and midline abdominal defects	Abnormalities to anterior diaphragm Ectopia cordis	Majority are idiopathic Possible X-linked inheritance on Xq25-q26.1	Genetic testing Fetal echocardiography Fetal MRI	Individualized based on severity of the case Elective termination of pregnancy often is advised	Depends on the type and severity of the cardiac and extracardiac manifestations Severe ectopia cordis cases have a 5%–10% survival rate
OEIS complex	Omphalocele Exstrophy of the cloaca Imperforate anus Spinal defects	Absent bladder	None Multifactorial	Genetic testing Fetal echocardiography Fetal MRI Beckwith-Wiedemann syndrome testing Serial fetal growth ultrasonography NST/BPP weekly starting at 32 weeks	Individualized based on severity of the case	Depends on the type and severity of the defects Neonates who are candidates for surgical repair have a near 100% survival rate

Abbreviations: BPP, biophysical profile; IUGR, intrauterine growth restriction; MRI, magnetic resonance imaging; NST, nonstress testing; OEIS, omphalocele-exstrophy-imperforate anus-spinal defects.

Management

There is no expert consensus regarding optimal prenatal management of gastroschisis.¹¹⁻¹⁷ Prenatal care, patient counseling, and delivery planning should be individualized based on the defect and should be determined in a multidisciplinary discussion with specialists in maternal-fetal medicine, neonatology, and pediatric surgery, as necessary. In our practice, if the gastroschisis is isolated and uncomplicated, our generalist obstetricians manage the patient with maternal-fetal medicine consultation, increased fetal surveillance as described below, and delivery at our tertiary care institution.

Our standard practice is to use the initial ultrasonography imaging to evaluate the size and contents of the defect, measure the nuchal translucency, and evaluate for additional abnormalities. Serial ultrasonography

monitoring of the fetus is required to assess the size and quality of the herniated intestine, amount of amniotic fluid, and fetal growth.¹⁰

As gastroschisis is a full-thickness defect of the anterior abdominal wall, the abdominal contents are exposed to amniotic fluid. This exposure causes progressive intestinal damage, which can be identified on ultrasonography as bowel thickening and dilation.¹²⁻¹⁴ Currently, intestinal thickening and dilation is not considered an indication for delivery as it is assumed that the intestinal damage has already occurred. It is debatable whether delivery around 37 weeks compared with delayed delivery beyond 37 weeks improves outcomes and decreases the stillbirth rate.^{11,13} Studies show that neonates delivered prior to 37 weeks have worse outcomes compared with those delivered after 37 weeks.^{14,15}

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FIGURE 2 Ultrasound scan demonstrating gastroschisis



Ultrasonography reveals loops of bowel herniating through a paraumbilical wall defect and floating in amniotic fluid without a covering membrane, features of gastroschisis.

Fetal surveillance. As standard practice, we evaluate the fetus at around 16 weeks and then again at around 20 weeks. In the absence of fetal growth restriction, which is associated with 25% of cases,^{16,17} our standard practice includes performing serial growth ultrasonography every 3 to 4 weeks starting at 28 weeks and biophysical profiles and non-stress testing weekly starting at 32 weeks. Fetal echocardiography can be offered. However, unlike with omphalocele, which has a high incidence of associated cardiac structural anomalies, gastroschisis has a low incidence of congenital cardiac anomalies, estimated to be between 2.5% and 4%.^{18,19}

Delivery considerations. Little agreement exists regarding when and how to deliver pregnancies complicated by fetal gastroschisis. While some advocate for induction of labor at 36 to 38 weeks, most infants with gastroschisis can be delivered safely at term via either vaginal or cesarean delivery.^{14,15}

Delivery timing should consider the clinical picture and incorporate performance on antenatal testing, fetal growth, the size and

contents of the gastroschisis, and consultation with maternal-fetal medicine. Fetuses with gastroschisis often have non-reassuring antenatal testing. This can necessitate early delivery, although cesarean delivery should be reserved for obstetric indications, with the caveat that if there is large liver involvement, some pediatric surgeons recommend cesarean delivery due to the risk of hepatic rupture. **Neonate management.** The survival rate of gastroschisis is reported to be as high as 91% to 94%.² Morbidity is related to intestinal complications, such as strictures, adhesions, and volvulus.

In the case of simple gastroschisis, when the bowel is in good condition, the treatment method of choice is primary reduction.²⁰ If performed in the operating room, an immediate sutured closure of the defect can be done. The benefits of primary repair include decreased length of stay, fewer intensive care bed days, and less time to achieve full feeds.^{20,21} Primary reduction has a reported success rate of 50% to 83%.²² A reduction with a delayed spontaneous closure also can be performed at bedside in the neonatal intensive care unit.²²

For complex gastroschisis, characterized by bowel complications such as inflammation, perforation, ischemia, atresia, necrosis, or volvulus, primary closure may not be possible and reduction may need to be achieved through silo application.²²⁻²⁵ Additionally, further bowel surgery, such as stoma formation and bowel resection, may be required.²⁵

Omphalocele often is associated with abnormal karyotype

Also known as exomphalos, omphalocele is a relatively common defect, with an estimated prevalence of 2 to 3 cases per 10,000 live births.² In this condition, there is a midline defect in which intra-abdominal contents herniate through the base of the umbilical cord. Omphaloceles are covered by amniotic membranes, making them distinguishable from gastroschisis, which has no covering, and congenital umbilical hernias, which are covered by intact skin and subcutaneous tissue.²⁶⁻³³

Additionally, in omphalocele the umbilical cord insertion site varies, whereas in gastroschisis the umbilical cord insertion is usually to the right of midline. An omphalocele is often categorized based on whether or not it contains the liver (extracorporeal liver) or only the bowel (intracorporeal liver).

Genetic studies

Approximately 67% to 88% of all pregnancies with omphalocele have an abnormal karyotype and/or associated malformations, including Beckwith-Wiedemann syndrome.³¹ Of the aneuploidies, trisomy 18 is the one most commonly associated with omphalocele, accounting for approximately 62% to 75%, while trisomy 13 accounts for approximately 11% to 24%.^{32,33} The presence of other anomalies is strongly associated with poor prognosis, and increased defect size is an independent predictor of neonatal morbidity and mortality, as neonates with large omphaloceles with extracorporeal livers can develop respiratory insufficiency and require more complex surgical repairs. It is interesting, however, that the absence of an extracorporeal liver is associated with a higher risk of aneuploidy than are cases with an intracorporeal liver.³³

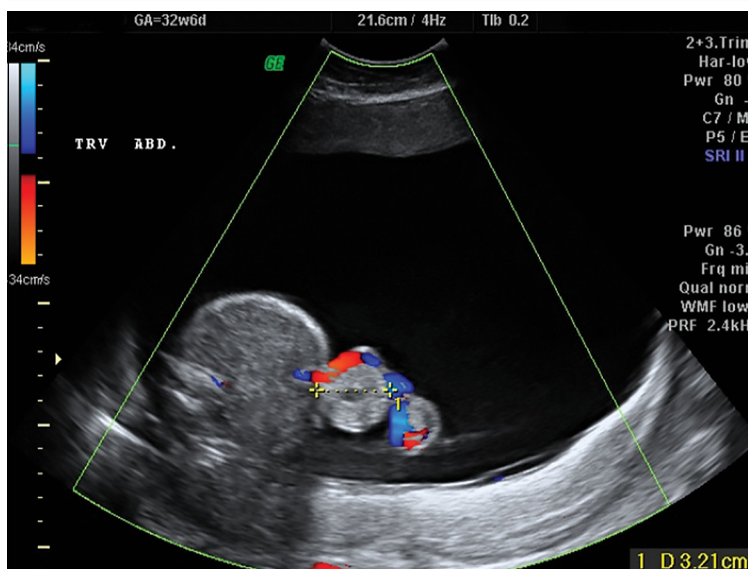
We offer chorionic villus sampling or amniocentesis to all patients with omphalocele. If the patient undergoes invasive diagnostic testing, the sample then undergoes karyotyping, chromosomal microarray, and testing for Beckwith-Wiedemann syndrome. If the patient declines diagnostic sampling, we perform a cell-free DNA screening to rule out aneuploidy.

Making the diagnosis

Omphaloceles can be diagnosed via prenatal ultrasonography as early as 11 to 14 weeks' gestation.²⁶ They are classified based on size, location, and contents of the sac.^{26,27} A small omphalocele is defined as a defect less than 5 cm with a sac that may contain a few loops of intestines (FIGURE 3).²⁷ A giant omphalocele is a defect with more than 75% of the liver contained in the sac.²⁹

Location can be epigastric, umbilical, or hypogastric, and both small and giant omphaloceles may have ruptured membranes that

FIGURE 3 Ultrasound scan showing omphalocele



Ultranography shows a small omphalocele (<5 cm), with intra-abdominal contents covered by amniotic membranes herniating through the base of the umbilical cord.

will result in exposure of the contained viscera.²⁷ Omphaloceles are associated with such structural anomalies as cardiac, gastrointestinal, genitourinary, diaphragmatic, and neural tube defects. We do not routinely perform magnetic resonance imaging (MRI) for evaluation of omphaloceles, but MRI may be used to help predict postnatal outcomes in the case of giant omphaloceles.²⁶

Management

Our standard practice is to use the initial ultrasonography imaging to evaluate the size and contents of defect, measure the nuchal translucency, and evaluate for additional abnormalities. As in cases of gastroschisis, serial ultrasonography monitoring of the fetus is required to assess the size and quality of the herniated intestine, amount of amniotic fluid, and fetal growth. We typically evaluate the fetus at around 16 weeks and then again at around 20 weeks. In the absence of fetal growth restriction, we recommend serial growth ultrasonography every 3 to 4 weeks starting at 28 weeks and biophysical profiles and nonstress testing weekly starting

Clinical pearls: Management of fetal abdominal wall defects

- Patients with fetuses with anterior wall defects should be referred to a maternal-fetal medicine specialist for co-management and advanced fetal imaging.
- The American College of Obstetricians and Gynecologists recommends microarray for all major fetal structural abnormalities, with the qualifier that karyotype can be offered if a specific aneuploidy is suspected based on the abnormality or prior genetic screening tests.
- If confirmatory testing is performed (amniocentesis or chorionic villus sampling), the sample should undergo karyotyping, chromosomal microarray, and if indicated, testing for Beckwith-Wiedemann syndrome. If the patient declines confirmatory sampling, performing cell-free DNA screening to rule out aneuploidy is recommended.
- Fetal echocardiography is recommended.
- Fetal magnetic resonance imaging should be considered in complex cases.
- Management should be individualized based on the type and severity of defect(s).
- Delivery timing and method should be individualized based on the defect(s) and determined in a multidisciplinary discussion with maternal-fetal medicine, neonatology, pediatric surgery, and pediatric cardiology, as necessary.
- The most common fetal abdominal wall defect is omphalocele, followed by gastroschisis.
- Maternal serum α -fetoprotein is usually elevated in all of the disorders.

at 32 weeks. Additionally, we routinely obtain a fetal echocardiogram to rule out cardiac structural abnormalities.

Delivery considerations. Fetuses that do not undergo spontaneous abortion or medical termination of pregnancy often are born at term.²⁶ We recommend expectant management until spontaneous labor, another indication for delivery arises, or at least 39 weeks' estimated gestational age. There are no evidence-based guidelines for the optimal mode of delivery in fetuses with omphalocele, although we recommend cesarean delivery for fetuses with large defects to avoid postnatal sac rupture and liver damage. Preterm induction of labor is not indicated as infants born preterm have about a 50% mortality rate.^{26,27}

Children born with isolated omphalocele typically have a good prognosis, with an estimated survival rate of 50% to 90%.^{32,33} However, compared to gastroschisis, omphaloceles are often associated with other anomalies.^{32,33}

Management of omphaloceles depends on the size of the defect. In our institution, our generalist obstetricians manage the standard

prenatal care with the addition of increased fetal surveillance and testing, interdisciplinary patient counseling with maternal-fetal medicine, pediatric surgeons, and neonatologists for delivery planning, and delivery is performed at our tertiary care center.

Neonate management. Small omphaloceles are amenable to primary early fascial closure.²⁶⁻³⁰ However, attempted primary closure of giant omphaloceles carries significant risks, including abdominal compartment syndrome and post-operative herniation.^{29,30} Instead, several options exist for staged surgical closure, in which there are multiple operations prior to final fascial closure, as well as nonoperative delayed closure for management of giant omphaloceles.^{29,30}

Conservative management of giant omphaloceles has certain benefits, such as earlier first feeds, decreased risk of abdominal compartment syndrome, and lower risk of infection.³⁰ Ruptured omphaloceles can be repaired through primary repair, employment of a synthetic or biologic mesh fascial bridge, or silo placement with delayed closure.²⁸

Body-stalk anomaly: Multiple defects and poor prognosis

Also known as limb body wall complex, body-stalk anomaly is a rare malformation that has a reported prevalence of approximately 0.12 cases per 10,000 births (both live and stillbirths).³⁴ Body-stalk anomaly is characterized by multiple defects, including severe kyphosis or scoliosis, a short or absent umbilical cord, and a large anterior abdominal wall defect.³⁴⁻³⁶ This malformation is almost entirely incompatible with life, resulting in abortion or stillbirth.³⁵ Survival is extremely rare and limited to case reports.

While the exact etiology of body-stalk anomaly is unknown, 3 possible causes have been hypothesized: early amnion rupture, vascular compromise, and embryonic dysgenesis.³⁷⁻⁴⁰

Making the diagnosis

Body-stalk anomaly typically can be diagnosed by 10 to 14 weeks' gestation via ultrasonography.³⁴⁻⁴¹ We currently follow the

diagnostic criteria proposed by Van Allen and colleagues, which requires 2 of the following 3 anomalies³⁴:

- exencephaly/encephalocele with facial clefts
- thoraco- and/or abdominoschisis (midline defect)
- limb defect.

Additional ultrasonographic findings can include the identification of evisceration of the abdominal contents, a short umbilical cord, and increased nuchal thickness.^{36,42} During the second and third trimesters, oligohydramnios may be seen.²

Management

Body-stalk anomaly is considered a fatal condition without specific therapeutic interventions. Maternal risks include an increased risk of preterm labor and gestational hypertension.³⁵ Research on body-stalk anomaly has not shown any correlation with patients' age, fetal sex, or abnormal karyotype, and the reported risk of recurrence for this anomaly is very low.^{42,43} Early diagnosis therefore is essential to provide families with information and counseling. Given the poor fetal prognosis, increased maternal risk, and low

recurrence rates, mothers can be advised toward elective termination of pregnancy.

Should a patient desire expectant management, care can be provided by generalist obstetricians or care can be transferred to maternal-fetal medicine, with the addition of increased fetal surveillance and testing, interdisciplinary patient counseling with maternal-fetal medicine, pediatric surgeons, and neonatologists for delivery planning; delivery should be performed at a tertiary care center.

Multidisciplinary team strategy is essential

Based on our experience, when faced with an anterior AWD in utero, prenatal imaging, genetic testing, increased fetal surveillance, and a multidisciplinary team approach improves outcomes. We must emphasize that careful patient counseling is paramount in our practice. ●

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To read about more types of fetal abdominal wall defects, see the online version of this article at mdedge.com/obgyn.

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Pentalogy of Cantrell: Very rare, with variable prognosis

Pentalogy of Cantrell is characterized by a collection of defects in the midline abdominal wall, lower sternum, anterior diaphragm, diaphragmatic pericardium, and some manifestation of intra-cardiac defect.⁴⁴ It is thought to arise early in gestation due to abnormal differentiation, migration, and fusion of the embryonic mesoderm.⁴⁴ The condition is rare, with an incidence of about 1 in 5.5 million live births.⁴⁵

Making the diagnosis

The diagnosis of pentalogy of Cantrell can be made via prenatal ultrasonography as early as the first trimester, although it is diagnosed more commonly in the second trimester.⁴⁶ Three-dimensional ultrasonography and fetal MRI have been used to confirm the diagnosis.⁴⁷

Management

Typically, corrective operations are performed during the neonatal period, and cases of successful staged and one-stage operations have been reported.⁴⁸ Surgical treatment is determined based on the complexity of the condition and the presence of coexistent heart defects.^{49,50} However, very few patients survive surgical repair; mortality rates are estimated at around 50% to 60%, with high postsurgical morbidity risks for those who do survive.⁴⁵

Prognosis varies depending on the type and severity of the associated malformations and intracardiac anomalies.⁴⁶ Patients with partial ectopia cordis and incomplete presentation may have more favorable outcomes, but for patients with severe ectopia cordis, the survival rate is only 5% to 10%.⁴⁷

Depending on the severity of the defects, mothers can be advised toward elective termination of pregnancy. In our institution, prenatal care usually is transferred to the maternal-fetal medicine service, and delivery is planned at our tertiary care institution.

OEIS complex comprises abdominal, pelvic, and spinal defects

Omphalocele-exstrophy-imperforate anus-spinal defects (OEIS) complex is a congenital malformation syndrome characterized by the combination of midline abdominal and pelvic defects (including omphalocele, exstrophy of the cloaca, and imperforate anus) and spinal defects.⁵¹ The condition's etiology is unknown but is thought to be multifactorial.⁵¹⁻⁵³ It is a rare condition, with an incidence of around 1 in 200,000 to 400,000 pregnancies.⁵²

Making the diagnosis

Prenatal diagnosis of OEIS complex can be made as early as the first trimester via ultrasonographic identification of an infraumbilical abdominal wall defect with protruding mass, absent bladder, and spinal defects.⁵² When OEIS complex is suspected, fetal MRI can play a critical role in the diagnosis.

Management

As OEIS complex is rare, there are no evidence-based guidelines for optimal mode and timing of delivery. Cases are individualized based on their specific pathology, and we recommend cesarean delivery for fetuses with large defects to avoid postnatal sac rupture and liver damage.

The prognosis for infants with OEIS complex depends on the spectrum and severity of the structural defects.^{52,53} The many surgeries involved in the repair of OEIS have potential complications, such as urogenital and gastrointestinal dysfunction.^{52,53} Advances in medical and surgical treatment have resulted in improved survival and quality of life, and survival rates for OEIS complex are now close to 100%.⁵³ While many OEIS patients live with a permanent colostomy, improvements in management mean that more patients are now candidates for gastrointestinal pull-through procedures, which allow for natural bowel control and a higher degree of bowel cleanliness.⁵³

Prenatal care, patient counseling, and delivery planning should be individualized based on the defects present and determined in a multidisciplinary discussion with maternal-fetal medicine, neonatology, and pediatric surgery as necessary. In our institution, prenatal care usually is transferred to the maternal-fetal medicine service, and delivery is planned at our tertiary care institution.

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