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Recommended Citation

Rentea RM, Gupta V. Gastroschisis. In: StatPearls. Treasure Island (FL): StatPearls Publishing; 2020.

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StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2020 Jan-.

Gastroschisis

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Last Update: May 2, 2020.

Introduction

Gastroschisis is a paraumbilical, full-thickness abdominal wall defect associated with protrusion of the bowel through the defect. It is rarely associated with genetic conditions. A membrane does not cover the bowel exposed in utero and, as a result, may be matted, dilated, and covered with a fibrinous inflammatory rind. Infants have a high proportion of intrauterine growth restriction. Diagnosis is often made on the 20-week ultrasound with free-floating bowel loops in the uterine cavity. Maternal serum alpha-fetoprotein (AFP) is elevated in pregnancies with gastroschisis. Compared with other abdominal wall defects diagnosed prenatally such as omphalocele, only 10 percent of cases with gastroschisis are associated with malformations outside of the gastrointestinal tract. Additional gastrointestinal problems occur in up to a quarter of cases. Infants can be classified as simple or complex, which can help stratify outcomes and care for infants born with gastroschisis. Complexity is based on the absence or presence of intestinal atresia, stenosis, bowel perforation, necrosis, malrotation, or volvulus. Infants may benefit from delivery in a facility with resources such as high-risk obstetrics, neonatology, and neonatal intensive care unit and a pediatric surgeon. A trial of labor rather than scheduled cesarean birth for most patients. Spontaneous delivery usually occurs between 37 to 38 weeks gestation. A trial of spontaneous vaginal delivery is supported. The exposed infant bowel is protected following birth, an orogastric tube is placed, as are peripheral IVs. The airway is stabilized. Gastroschisis closure can be performed operatively or through slow bowel reductions utilizing a spring-loaded silo to contain the bowel. While a small percentage of infants have intestinal atresia, bowel loss, and prolonged hospitalizations, the overall survival is greater than 90 percent.

Etiology

The cause of gastroschisis is incompletely understood. Gastroschisis is caused by a failure of the formation and development of the ventral body wall during embryogenesis, resulting in herniation of the bowel. There are several implicated causative factors that may contribute to the development of gastroschisis, including tobacco, specific environmental exposures (nitrosamines, for example, atrazine), cyclooxygenase inhibitor use (aspirin and ibuprofen), and decongestants (pseudoephedrine and phenylpropanolamine).[1][2][3]

Epidemiology

Gastroschisis occurs in 1 in 4000 live births.[4] The incidence of gastroschisis is increasing worldwide.[5] The incidence of gastroschisis between male infants is similar to that of females. There is a higher incidence in Hispanic, singleton pregnancies, and younger women less than 20 years of age.

Pathophysiology

During the 4th week of gestation, the abdominal wall forms through the craniocaudal and mediolateral directions. The liver and midgut herniate through the umbilical cord during the sixth week of gestation. The midgut has completed rotation and returns to the abdominal cavity at the 10th week of gestation.

History and Physical

The characteristic clinical finding in newborns born with gastroschisis is a full-thickness paraumbilical abdominal wall defect (to the right of the umbilicus) with the associated evisceration of the bowel. The abdominal wall defect tends to be approximately 4 cm in size. Unlike omphalocele, there is no membrane covering the bowel. The bowel is often thick and matted. The exposure to the amniotic fluid, and the amount of time the bowel has been in contact with the amniotic fluid influence the amount of bowel wall thickening and rind. Within the matted bowel, potential bowel atresias or in-utero ruptures can be visualized. Additional abdominal contents can also herniate through this defect, including the stomach, liver, and bladder. Rarely, the bowel can be exposed and present on a small stalk with a tiny gastroschisis defect. This finding is often concerning, as this finding may be associated with a significant loss of bowel length (vanishing gastroschisis).

Evaluation

Gastroschisis is often diagnosed on a prenatal ultrasound performed at approximately 20 weeks' gestation.^{[6][7][8]} In the presence of gastroschisis, serum alpha-fetoprotein (AFP) level is elevated.^{[6][8]} On ultrasound, a defect to the right of the umbilical cord and free-floating bowel loops are visualized. Bowel exposed to amniotic fluid may become thickened and dilated. Late diagnosed gastroschisis, such as at the time of delivery, can result in changes in neonatal management. Late diagnosed gastroschisis often has fewer changes from the amniotic fluid. However, there may also be bowel loss if the defect that the bowel herniated through was small and compromised the intestine as the abdominal wall defect attempted to close.

Infants with gastroschisis may have intrauterine growth restriction (IUGR). Pregnancy may become complicated by fetal demise and spontaneous preterm birth.^{[9][10]} Approximately 10 percent of cases are associated with malformations outside of the gastrointestinal tract. In up to 25 percent of cases, additional gastrointestinal problems (e.g., intestinal atresia, stenosis, perforation, necrosis, malrotation, volvulus) are present. The karyotype is abnormal in 1 percent of cases, usually in the setting of associated abnormalities.

Treatment / Management

Pregnancy Management

Infants with gastroschisis should be monitored for fetal growth. Growth restriction can be seen in up to 60% of cases. If there is a concern or risk of intrauterine demise, then antenatal testing can be considered. The prevalence of chromosomal anomalies in infants with isolated gastroschisis is that of the baseline population risk. However, in the setting of extraintestinal structural abnormalities, then the risk of chromosomal anomalies increases, and amniocentesis for parental decision-making and management of the newborn may be warranted.^[11] Once gastroschisis is diagnosed, fetal growth and amniotic fluid volume are charted via ultrasound at 3 to 4-week intervals starting at 24 weeks gestation.^[12] Oligohydramnios may be related to fetal growth restriction and is a risk for cord compression, while polyhydramnios may be predictive of bowel atresia.^[13] Growth restriction in fetuses in the setting of abdominal wall defects may predict increased adverse neonatal outcomes.^{[14][15]}

Delivery

Infants with gastroschisis have no contraindication to vaginal delivery based on gastroschisis alone. The timing of delivery is based on gestation age (lung maturity), ultrasound findings (fetal growth profile, bowel appearance), and fetal testing results. Care for the infant is usually managed by a group of specialists, including a maternal-fetal medicine specialist, neonatologist, and pediatric surgery to discuss patient-specific factors before delivery. The mean gestational age for spontaneous delivery in women carrying infants with gastroschisis is the 36th week of gestation.^{[16][17]} A survey of practice patterns demonstrated forty percent of maternal-fetal medicine specialists deliver infants at 37 weeks, 30 percent deliver at 39 weeks if gastroschisis is stable. The remainder delivers infants before 37 weeks.^[12] Overall, many maternal-medicine specialists allow a trial of vaginal delivery. When there is marked liver herniation, a cesarean section should be considered. One factor that some specialists utilize to determine continued gestation is if the bowel does not dilate greater than 25 mm after 37 weeks gestation.

Management of the Neonate

Management of the neonate begins with delivery room care. Evaporative fluid losses through the exposed bowel are 2.5 times the amount of that of a healthy newborn.[18][19][20] However, over resuscitation has detrimental effects, including bowel and total body edema, an increase in time to abdominal wall closure, and an increased risk of abdominal compartment syndrome.[20][5] Initial management includes protecting the exposed bowel by placing the lower half of the infant into a bowel bag. This maneuver allows the protection of the bowel as well as visualization of the blood flow and perfusion of the bowel. An orogastric tube should be inserted to decompress the stomach. Placement of peripheral intravenous access to provide antibiotics and maintenance fluid is performed. Intravenous fluids are administered two to three-fold due to the exposed bowel fluid losses. The airway should be evaluated and maintained.

Surgical management of gastroschisis

The primary goal of gastroschisis repair is to return the exposed bowel and new organs to the abdominal cavity while minimizing intestinal injury or increased intra-abdominal pressure. Two treatment options are present for gastroschisis. The first is primary repair, and the second is delayed closure (usually utilizing a temporary silo and performing serial reduction of bowel contents). Primary closure near birth is performed either operatively or following successful bowel reduction back into the abdominal cavity and performing sutureless gastroschisis repair. A randomized control trial evaluated primary closure versus delayed primary closure following silo reduction in which there were no differences seen between two groups in length of stay, time to enteral feeds, or ventilator times.[21]

The umbilicus is now preserved during the closure as it leads to excellent cosmetic results. A prospective randomized study comparing sutureless closure to sutured repair noted that the time to full feeds and length of stay was significantly longer in the sutureless group, despite no additional complications.[22] However, a multi-institutional review demonstrated that sutureless abdominal wall closure of neonates with gastroschisis was associated with less general anesthetics, antibiotic use, surgical site/deep space infections, and decreased ventilator time.[23]

The bowel is inspected at the time of reduction for obstructing bands, perforation, or atresia. However, bowel anastomosis in the setting of possible atresia is usually not commenced in the setting of edematous bowel. At the initial assessment of the gastroschisis, obvious bowel atresia can be turned into an end ostomy to allow an earlier resumption of feeding while waiting for the intestines to normalize. Care is exercised during the reduction of bowel contents. Perfusion of the bowel is monitored frequently as the mesentery can be compromised based on the pressure of the silo, the bowel at the fascia as it is being reduced, as well as once the bowel is reduced into the abdominal cavity. Abdominal compartment syndrome with pressures higher than 10 to 15 mmHg is often associated with decreased renal and intestinal perfusion, whereas above 20 mmHg, they correlate with organ dysfunction and complications.[24][25]

Differential Diagnosis

The differential diagnosis of gastroschisis in a newborn is those of newborn abdominal wall and umbilical defects. [26] Omphalocele is the first differential diagnosis. The membranous sac covering the bowel contents as well as the often intact umbilical cord assist in differentiating the two diagnoses. The membrane covering the omphalocele may have ruptured during in utero development or at birth. However, the location of the liver and cord vessel insertion site may assist in differentiating omphalocele (cord inserts into the apex of the omphalocele membrane) vs. gastroschisis (cord inserts adjacent to the abdominal wall defect with the defect usually to the right of the umbilicus). Additional diagnosis to consider: umbilical hernia (cord inserts into the hernia sac), pentalogy of Cantrell (lower sternal defect, anterior diaphragm defect, pericardial defect, omphalocele, congenital heart anomalies), bladder exstrophy (low umbilical cord insertion with non-visualization of the bladder), cloacal exstrophy (cord insertion is low with exposed bowel and bladder and omphalocele), amniotic band sequence (constriction of rings and limbs - also called limb body wall complex).[27][28]

Prognosis

Compared with infants born with other abdominal wall defects, infants born with gastroschisis have the most favorable prognosis, with excellent long term outcomes.[29] The overall survival rate for live-born infants is 98 percent in neonates born in North America.[30]

Categorizing infants born with gastroschisis into “simple” or “complex” based on the absence or presence of intestinal atresia, stenosis, bowel perforation, necrosis, malrotation, or volvulus aids in stratifying infant outcome further. [31] Up to 75 percent of gastroschisis cases are simple, and 25 percent are complex. Infants born with complex gastroschisis have more gastrointestinal, respiratory, and infectious complications in the neonatal period.[32] A systematic review and meta-analysis compared simple versus complex gastroschisis outcomes. Infants born with complex gastroschisis were associated with having a higher risk of in-hospital mortality, short bowel syndrome, bowel obstruction, necrotizing enterocolitis, parenteral nutrition, and tube feedings on discharge.[32] Infants with complex gastroschisis also were likely to have a 2-month longer hospitalization.[33]

Most infants with gastroschisis have an intestinal rotational anomaly, which is not repaired. The incidence of volvulus between infants born with omphalocele versus gastroschisis is higher for infants born with omphalocele (4.4% vs. 1.0%).[34] Intestinal adhesions and adhesive bowel obstruction are at increased risk after gastroschisis. [35] Cryptorchidism (undescended testicles) is associated with 15% to 30% of gastroschisis cases, with observational management for the first year of life, at which time an orchiopexy can be performed.[36]

Long term outcomes are not well defined but are overall favorable. The cosmetic appearance of the abdomen and umbilicus in infants where the umbilicus is sacrificed report psychosocial stress at the absence of an umbilicus. [36] An umbilicoplasty or umbilical reconstruction surgery can be considered in this patient group. Neurodevelopmental delay, learning issues, and overall health-related quality of life have not been well defined, but are reported in preliminary studies to be within a normal range.[35][37]

Complications

Complications

Preterm delivery is more common in infants with gastroschisis at 28% versus infants without 6%. Complications can occur in infants with gastroschisis based on the need for total parenteral nutrition (TPN) and resultant line sepsis, bowel configuration leading to necrotizing enterocolitis (NEC), and abdominal wound infections from the gastroschisis closure.

Risk Stratification

Patients can be defined as “simple” versus “complex” based on the presence of intestinal complications (atresia, ischemia, perforation, or development of necrotizing enterocolitis). Patients with complex defects have a higher mortality rate, require multiple operative interventions, and have a prolonged hospitalization, increased rates of sepsis, and higher rates of prolonged cholestasis and need for intestinal transplantation due to intestinal failure.[31][38][39][40]

Management of intestinal atresia in the setting of gastroschisis

Up to 10% of neonates with gastroschisis have associated atresia, most commonly jejunal or ileal.[41] Patients born with atresia have significantly worse outcomes.[32] The timing of surgical management of the atresia depends on the state of the bowel as a bowel anastomosis in the setting of an extensive inflammatory peel will not hold sutures well. A retrospective database review examined outcomes of early versus late operations for intestinal atresia associated with gastroschisis (defined as before or after 21 days of life) found there was no significant difference in outcomes and potential for early feeding between the two groups.[42] If the atresia is known, it may be reduced and re-explored at a 4 to 6-week interval to allow for decreased adhesions and inflammation. Atresia of the bowel can also be diagnosed following several weeks of no bowel function with a confirmatory contrast study. At the initial assessment of the gastroschisis, obvious bowel atresia can be turned into an end ostomy to allow the earlier resumption of feeding while waiting for the intestines to normalize.

Closing Gastroschisis

A “closing gastroschisis” is when the gastroschisis defect size decreases before delivery.[43] As the hole gets smaller, the blood supply to the bowel progressively diminishes, resulting in atresia and a variable loss of bowel. When a large amount of intestine is lost in utero, it usually results in short bowel syndrome.

Consultations

It is essential to consult with an interprofessional team of specialists that include an obstetrician, neonatologist, and pediatric surgeon. Often a prenatal consultation is performed with the family prior to delivery. The obstetrician assists in safe modes and timing of delivery as well as management of the mother and patient. The neonatologist assists in the critical care of the infant after birth. The pediatric surgeon assists in forming an operative strategy for managing the exposed bowel and slowly reducing it into the abdominal cavity to close the abdominal wall. Closure of the abdominal wall defect may take place with the aid of a silo (that the bowel is contained within it temporarily externally) as it is slowly reduced into the abdominal cavity.

Deterrence and Patient Education

Families are given an option for a prenatal consultation to meet an interprofessional team when the diagnosis of gastroschisis becomes known. This allows the family to understand the diagnosis and prepare for the needs of the infant. The family is consulted on what gastroschisis is (full-thickness abdominal wall defect to the right of the umbilical cord). The most crucial issue is that as the organs are developing, the bowel which has not returned to the infant's abdominal cavity is in contact with the amniotic fluid. Early signs are bowel floating outside of the infant on ultrasound. Later signs include new organs being visualized through the defect as well, which include liver, stomach, and bladder. What is the treatment: the infant is delivered in a hospital that has access to surgeons and specialists to care for the infant immediately after birth. The best time for delivery is unknown, and most commonly, infants are delivered near term. When the infant is born, a nasogastric tube is inserted to decompress and drain the stomach, a breathing tube may be required to assist in breathing, peripheral intravenous access, and fluids will be administered. The bowel that is exposed will be protected with moist dressings and/or a lower body bag to avoid evaporative fluid losses as well as to monitor the bowel perfusion (blood flow). The surgical team will then assist in deciding if the bowel needs to be placed into a silo (a temporary plastic bag) that allows gravity to get the intestine back into the abdomen over a few days, or if the abdominal wall defect can be closed at birth. There are many ways to close the abdominal wall opening, which include primary closure or sutureless closure, and the surgeon will discuss which option is best for your child. Because the bowels have been exposed to the amniotic fluid, they may not be able to tolerate food for several weeks. Therefore, nutrition will be given through the infant's veins until they can start feeding. Risks/Benefits: some in cases reducing the organs and bowel back through the abdominal wall opening may injure the organs. The intestines may also have a chance of having blood supply issues such as when the space is too tight in the tummy or outside the body, and these issues can happen both in-utero and after birth. Up to one-third of infants experience an infection called necrotizing enterocolitis, and 10 to 15% of infants have intestinal atresia where the intestines are not in continuity and require additional operation(s) a few weeks after birth. Once the child goes home, there are usually no restrictions on a diet. There are no activity restrictions. The surgeon reviews wound care. If the infant develops feeding intolerance (vomiting) or the incision becomes red, then the surgical team needs to be called.

Pearls and Other Issues

Gastroschisis is a full-thickness right-sided paraumbilical abdominal wall defect, without a covering membrane. The pathogenesis is unknown but occurs early in embryologic development around the 4th week of gestation. Maternal serum alpha-fetoprotein (AFP) is elevated in pregnancies with gastroschisis. Prenatal ultrasound visualizes free-floating bowel loops and a paraumbilical abdominal wall defect. Compared with other abdominal wall defects diagnosed prenatally such as omphalocele, only 10 percent of cases with gastroschisis are associated with

malformations outside of the gastrointestinal tract. Additional gastrointestinal problems (e.g., intestinal atresia, stenosis, perforation, necrosis, malrotation, volvulus) are present in up to 25 percent of cases.

Pregnancy complications include increased risk of intrauterine growth restriction, fetal demise, spontaneous preterm birth, and bowel thickening and dilation. A fetal microarray molecular testing should be offered when gastroschisis is associated with additional nongastrointestinal structural abnormalities. Infants may benefit from delivery in a facility with resources such as high-risk obstetrics, neonatology, and neonatal intensive care unit and a pediatric surgeon. A trial of labor rather than scheduled cesarean birth for most patients (grade 2C), except if the liver is significantly herniated. There is no consensus on optimum timing of delivery of these pregnancies, and it is between 37 to 38 weeks gestation. In the delivery room, an orogastric tube is placed, as are peripheral IVs. The airway is stabilized. The exposed bowel is wrapped with sterile saline dressings covered with plastic wrap to preserve heat and minimize insensible fluid loss, or the lower half of the infant is placed into a plastic bag to minimize evaporative losses and aid in bowel visualization. Overall survival is greater than 90 percent.

Enhancing Healthcare Team Outcomes

Gastroschisis poses several challenges in the prenatal and early neonatal management of the infant with exposed bowel from an in-utero full-thickness abdominal wall defect with exposed bowel. These infants rarely have additional congenital issues besides intrauterine growth restriction (IUGR). The cause of gastroschisis is unknown and presumed to be multifactorial. While the physical exam at birth reveals an infant with exposed bowel, which may be matted depending on the duration of exposure to amniotic fluid, liver, stomach, and bladder may also have herniated through the abdominal wall defect depending on the size.

It is essential to consult with an interprofessional team of specialists that include an obstetrician, neonatologist, and pediatric surgeon. The nurses are also a vital member of the interprofessional group as they will monitor the patient's vital signs and assist with the education of the patient and family. The obstetrician assists in safe modes and timing of delivery as well as management of the mother and patient. The neonatologist assists in the critical care of the infant after birth. The pediatric surgeon assists in forming an operative strategy for managing the exposed bowel and slowly reducing it into the abdominal cavity to close the abdominal wall. Closure of the abdominal wall defect may take place with the aid of a silo (that the bowel is contained within it temporarily externally) as it is slowly reduced into the abdominal cavity.

The outcomes of gastroschisis depend on the infant's prematurity, additional comorbidities, as well as the state of the bowel from cumulative effects pre and antenatally (atresia, vanishing gastroschisis, vascular compromise of the mesentery supplying bowel before and after birth). However, to improve outcomes, prompt consultation with an interprofessional group of specialists is recommended.

Questions

To access free multiple choice questions on this topic, [click here](#).

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Figures

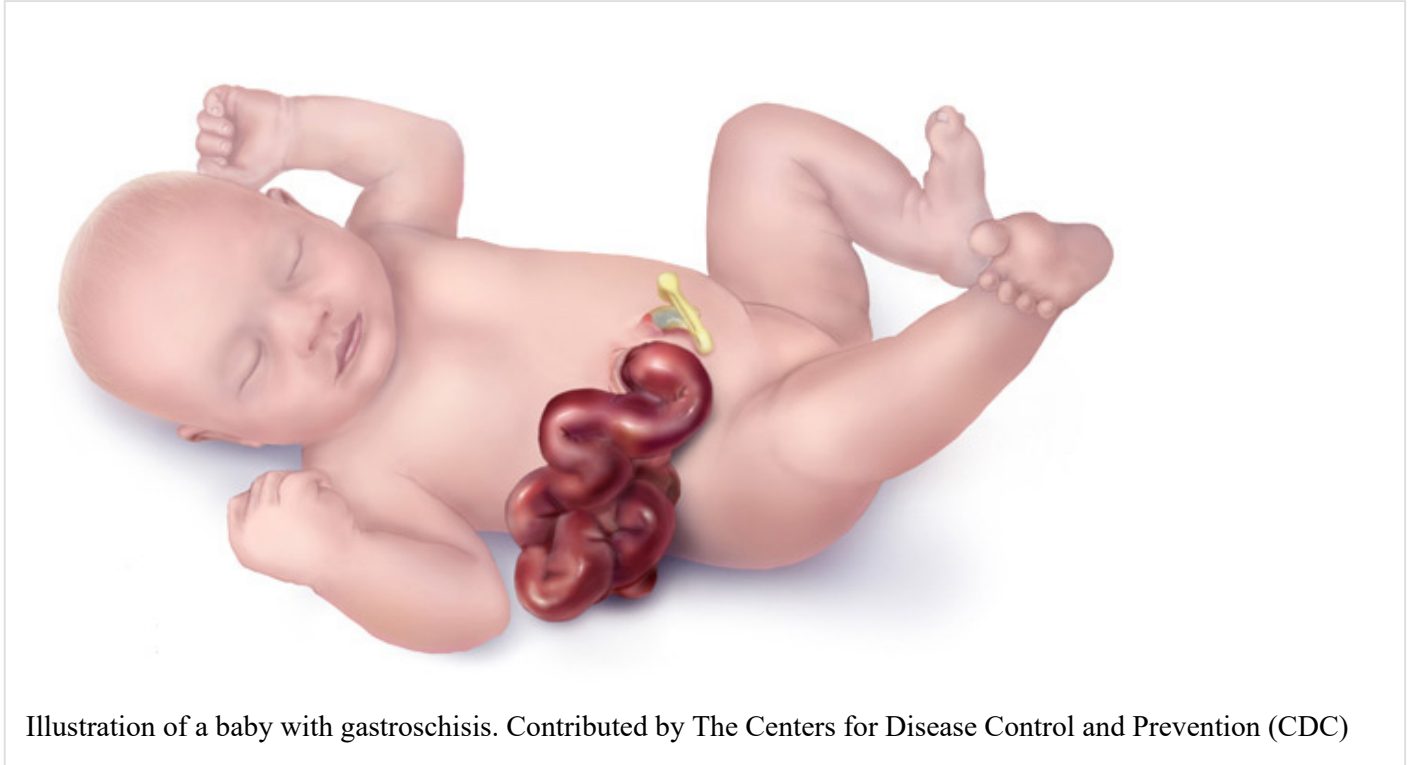


Illustration of a baby with gastroschisis. Contributed by The Centers for Disease Control and Prevention (CDC)

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