

Case Report

Left-sided gastroschisis with placenta findings: case report and literature review

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Abstract: Gastroschisis is a congenital abdominal-wall defect that typically occurs to the right of the umbilicus. Only twenty-one cases of left-sided gastroschisis have been described in the literature. Here we report a large left-sided gastroschisis with pulmonary hypoplasia, scoliosis, ventricular septal defect and absence of gallbladder. Section of placental membranes revealed vacuolization of the amnion, without increased macrophage infiltration of the chorion. Postmortem comparative genomic hybridization micro array did not identify a specific genetic abnormality. Some of the previously reported cases were complicated by additional abnormalities and comparisons with these cases are discussed.

Keywords: Left gastroschisis, pulmonary hypoplasia, scoliosis, ventricular septal defect

Introduction

Gastroschisis is an uncommon abdominal wall defect, but its occurrence to the left of the umbilical cord is rare. We report a neonate with a large left-sided gastroschisis which incorporated the liver, stomach, small intestine, colon and spleen.

Case report

A 27 year old Hispanic gravida 2 para 1-0-0-1 at 35 weeks gestation presented in active preterm labor and was taken for an uncomplicated repeat cesarean. Her pregnancy was complicated by fetal gastroschisis first diagnosed on an 11 week prenatal ultrasound. Follow-up ultrasound at 29 and 4/7 weeks gestation confirmed a large left sided abdominal defect with liver, large bowel, and small bowel exteriorized (**Figure 1**) as well as a small chest, scoliosis and ventricular septal defect. Amniocentesis demonstrated a normal 46XY karyotype. The patient had been counseled extensively regarding the poor prognosis for this pregnancy in light of the size of the abdominal defect and possible pulmonary hypoplasia secondary to the small fetal chest size. The patient desired full intervention for the fetus.



Figure 1. Ultrasound at 29 and 4/7 weeks gestation: left sided abdominal defect with liver, large bowel, and small bowel exteriorized. Courtesy of the Center for Fetal Evaluation, University of California, Irvine.

The 1920 gram neonate had Apgar scores of 0, 2, 3, and 5 at 1, 5, 10, and 15 min, respectively. The neonate required several intubation attempts as well as multiple doses of epinephrine and chest compressions. Pulmonary hypoplasia was confirmed on chest x-ray. Pediatric surgery was consulted, but further surgery or silo placement was not considered feasible due to the inability to internalize the infant's organs given the lack of abdominal cavity, especially in

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the presence of pulmonary hypoplasia. Upon extensive discussion with the infant's family, the decision was made to discontinue life support, and he passed away three hours after delivery due to respiratory failure. An autopsy limited to the chest, abdominal cavity and pelvis was performed. The placenta was sent to pathology for examination.

Pathologic findings

Postmortem examination revealed the following measurements: crown-rump length 28 cm (expected: 30.9 cm), crown-heel length 41 cm (expected: 42.3 cm), foot length 6.8 cm (expected: 7.2 cm), head circumference 33 cm (expected: 31cm), chest circumference 20 cm (expected: 28.5 cm), and abdominal circumference 18.5 cm (expected: 24cm). The body weighed 1920 grams (expected: 2093g). The neonate had a depressed nasal bridge, slightly low-set ears and hypognathia. The left hand showed a transverse palmar crease. Otherwise the upper and lower extremities were normal. Gastroschisis was identified involving the left abdomen, 0.5 cm to the left of the umbilicus. The defect measured 7.5 X 4.5 cm and the extra-abdominal organs included stomach, left liver, small intestine, colon and spleen. No membrane covering the organs was identified. The surface of the liver was dry, dark red, with green patches on the right lateral surface (**Figure 2**). The gallbladder was absent. Scoliosis with left thoracic convexity was associated with anterior displacement of the right ribs and inferior displacement of the left hemidiaphragm. The lungs weighed 13 g combined, with the expected weight being 35.2 g. The left pleural cavity contained 4 ml of light brown clear fluid. A

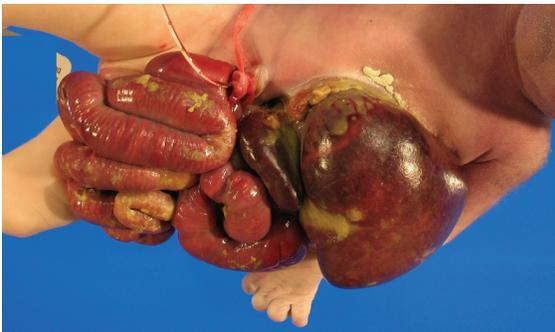


Figure 2. The large, left-sided gastroschisis included liver, small intestine, colon and spleen. No membrane covering the organs was identified.

ventricular septal defect measuring 0.4 cm was identified in the heart. There was no evidence of situs inversus of the heart or lungs.

Microscopically, squamous debris was adherent to the liver capsule, spleen, serosa of small intestine and colon. The spleen was congested. An adrenal rest was identified near the left testicle. Petechial hemorrhage was found in the thymus. Post mortem comparative genomic hybridization micro array did not identify a specific genetic abnormality.

The gross placenta weighted 500 g (expected: 434g). The umbilical cord coiling was increased. The membranes were pink-tan, translucent and complete. Meconium was grossly absent. The fetal surface was purple-blue, smooth and glistening, with a normal vascular pattern. The amnion was intact over the fetal surface, and no amniotic bands were seen. Sections of placental membranes revealed vacuolization of the amnion, without increased macrophage infiltration of the chorion (**Figure 3**).

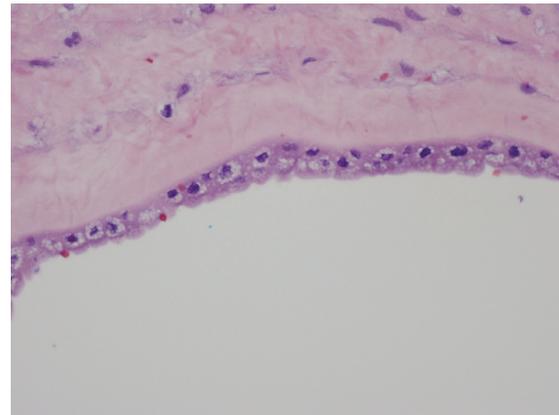


Figure 3. Enlarged distinctive vacuolated cells are identified at amnion. (400x)

Discussion and review

Gastroschisis is an uncommon abdominal wall defect, with the incidence rate of 0.3-1 in 10,000 births [1]. It is rare when occurring to the left of the umbilical cord. Only a handful of similar cases have been reported [2-16]. Most of the patients recovered very well after surgery. One patient developed respiratory difficulty and died 5 hrs later [8]. One fetus underwent autopsy after termination of pregnancy at 24

weeks of gestation following the prenatal diagnosis of bilateral multicystic dysplastic kidneys with severe oligohydramnios [6]. However, none of these studies evaluated the placenta or reported on the findings of an autopsy after live birth.

In the current case, the patient required intubation immediately after birth due to respiratory distress. Prenatal ultrasound examinations raised the suspicion of pulmonary hypoplasia, which was confirmed on autopsy. The findings of this case and previously reported cases suggest a correlation between the size of gastroschisis and the degree of pulmonary insufficiency. Ameh et al. reported that their patient developed respiratory difficulty after the 8 X 6 cm gastroschisis was repaired [8]. Further, other studies concluded that larger abdominal wall defects increased the incidence of respiratory insufficiency [17]. Low intra-abdominal pressure in utero may result in an excessively narrow lower thoracic cage, which limits the development of fetal lungs [1].

Among the reported cases, evisceration of the stomach, small and large intestine is common, and evisceration of the liver, kidney, urinary bladder and ovary have all been reported [6, 8, 10, 12].

Some of the previously reported cases of left sided gastroschisis were not complicated by other abnormalities [2-4, 7, 8, 11-13, 16]. However, one case was complicated with situs inversus [9]. Another case showed pubic diastasis, bifid clitoris, double vagina, anteriorly placed anus, atrial septal defect, and patent ductus arteriosus [10]. Jejunal atresia and microcolon, cerebral arterio-venous malformation, macrocephaly and stenosis of superior vena cava have also been reported [5]. In our case, the patient had pulmonary hypoplasia, scoliosis, ventricular septal defect and absence of the gall bladder. These diverse findings suggest that an unidentified syndrome is unlikely; rather, de novo mutations or in utero deformations possibly occurred in order to create these abnormal phenotypes. We suspect that the scoliosis and pulmonary hypoplasia were deformations of the fetus due to the abnormal displacement of organs outside the body cavity.

The pathogenesis of gastroschisis remains controversial, although multiple mechanisms have

been proposed [1]. It has been hypothesized that involution of the right umbilical vein, which normally occurs during embryogenesis, results in decreased viability of the surrounding mesenchyme. Because this mesenchyme is part of the body wall, death of its cells could result in a weakness in the region, allowing the internal organs to herniate. Regression of the left umbilical vein or left-sided omphalomesenteric artery might lead to a left sided gastroschisis [18]. However, the actual molecular mechanism underlying gastroschisis remains unclear.

It is important to differentiate gastroschisis from omphalocele. Gastroschisis is an abdominal wall defect which usually occurs to the right of the umbilicus, and, rarely, to the left of the umbilicus as in our case. A portion of the medial wall of the defect usually includes the umbilical cord. Omphalocele is a defect in the ventral abdominal wall wherein there is absence of abdominal muscles, fascia, and skin at or surrounding the junction of the umbilical cord with the abdomen. The area of omphalocele is covered by a membrane composed of amnion externally and parietal peritoneum internally, with mesenchymal connective tissue between. Microscopically, vacuolated amnion is specific for gastroschisis, and does not occur in the presence of omphalocele [19]. Gastroschisis results most likely from a vascular defect. Omphalocele is due to the persistence of the body stalk in the region normally occupied by the somatopleure and is often associated with a chromosomal anomaly. In our case, the position of the defect, the lack of covering membrane, and the placental pathology confirm that this is, indeed, a case of left sided gastroschisis. The absence of situs inversus rules out abnormal situs as the cause of the defect on the left rather than right abdomen. Moreover, the intact amnion on the placental surface exclude the possibility of early amnion rupture.

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