

First trimester diagnosis of fetal gastroschisis, case report and literature revisión

Diagnóstico ecográfico de gastrosquisis fetal en el primer trimestre: presentación de un caso y revisión de la literatura

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Summary

The spectrum of fetal abdominal wall defects includes omphalocele, gastroschisis, pentalogy of Cantrell, bladder and cloacal exstrophy, among others. These pathologies are usually diagnosed during second-trimester ultrasounds, generally during the anatomical detail sonogram performed between 20 and 24 weeks. However, careful assessment by the sonographer, starting in the embryonic phase, allows for the detection of these anomalies in the first trimester. It is important to differentiate the main morphological characteristics of these wall defects, since the suggestion of any of them as a diagnosis entails a chain of decision-making, involving the medical team and the expectant parents. Below, a case of gastroschisis in a primiparous woman who came for a first-trimester screening ultrasound is presented. The imaging findings were unequivocal and typical of the condition, which allowed for an early diagnosis and the decision-making considered pertinent by the parents.

Key words (MeSH)

Gastroschisis
Ultrasonography
Abdominal wall
Prenatal diagnosis

Palabras clave (DeCS)

Gastrosquisis
Ultrasonido
Pared abdominal
Diagnóstico prenatal

Resumen

El espectro de defectos de pared abdominal fetal incluye el onfalocele, la gastrosquisis, la pentalogía de Cantrell, la extrofia vesical y cloacal, entre otros. Estas patologías suelen ser diagnosticadas durante las ecografías del segundo trimestre, generalmente en el detalle anatómico entre las 20-24 semanas; sin embargo, la acuciosa valoración del ecografista, desde la fase embrionaria, permite la detección de estas anomalías en el primer trimestre. Es importante diferenciar las principales características morfológicas de dichos defectos de pared, ya que la sugerencia de alguna de ellas como diagnóstico lleva a una cadena de toma de decisiones, las cuales involucran al equipo médico y a los padres expectantes. A continuación, se presenta un caso de gastrosquisis en una primigestante que acude a realizarse la ecografía de tamizaje de primer trimestre, en la que se encontraron hallazgos imagenológicos inequívocos y típicos de la entidad, lo cual permitió un diagnóstico temprano y la toma de decisión considerada pertinente por los padres.

Introduction

Ventral defects of the fetal abdominal wall include a spectrum of abnormalities, the main examples of which are gastroschisis and omphalocele. Antenatal detection of both defects is important because their prognosis differs significantly (1).

Gastroschisis is a congenital defect of the fetal abdominal wall, which is typically located to the right of the navel. The intestines and, in some cases, the liver and stomach protrude into the amniotic cavity. Unlike in omphalocele, these organs are not covered by a membrane but instead float freely in the amniotic cavity (2).

This condition is usually diagnosed during a detailed anatomical ultrasound at 20-24 weeks; however, adequate knowledge of fetal and embryonic anatomy and a thorough examination of the fetus can lead to early detection, even in the first trimester, as illustrated in this case.

Case presentation and radiological findings

A 20-year-old primigravida woman attends an appointment for a first-trimester screening ultrasound. At the time of the examination, intrauterine pregnancy is confirmed, with a single live and active fetus, measuring 66 mm in cephalocaudal length, at 12 weeks and 5 days. The presence of the nasal bone (Figure 1), nuchal translucency within the expected range for gestational age (Figure 2), and a venous duct with antegrade flow throughout the cycle, without inversion or other alteration in any of the spectral wave components (Figure 3), were confirmed. The anatomy of the skull, viscerocranium, central nervous system, thorax and its components, limbs, and cord vessels are normal in number and morphology for gestational age, taking into account their early stage of development.

When exploring the fetal abdomen in sagittal and axial sections, a solid, microlobulated, irregular, echo-

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genic structure without vascular enhancement is noticeable to the right of the umbilical cord insertion site in the fetal abdomen (Figures 4 and 5). This image shows a serpentine structure, consistent with intestinal loops, which are still collapsed given the early stage of gestation. The described structure persists in all sections, is fixed in the fetal abdomen, and floats freely in the amniotic cavity, without membranes or connection to the base of the cord. The liver is not visible in the image. The diagnosis is a live fetus at 12 weeks and 6 days of gestation, with ultrasound markers for aneuploidy without alteration and gastroschisis. Pregnancy monitoring is not continued, as the pregnant woman decides to undergo voluntary termination, which is performed at another center seven days after the ultrasound diagnosis. This procedure resulted in a fetus of approximately 14 weeks of gestation, with a 4 × 4 mm wall defect to the right of the umbilical cord, through which intestinal loops, still collapsed, protrude.



Figure 1. Fetal profile with confirmation of the presence of the nasal bone and its measurement (markers).



Figure 2. Parasagittal profile with measurement of normal nuchal translucency.

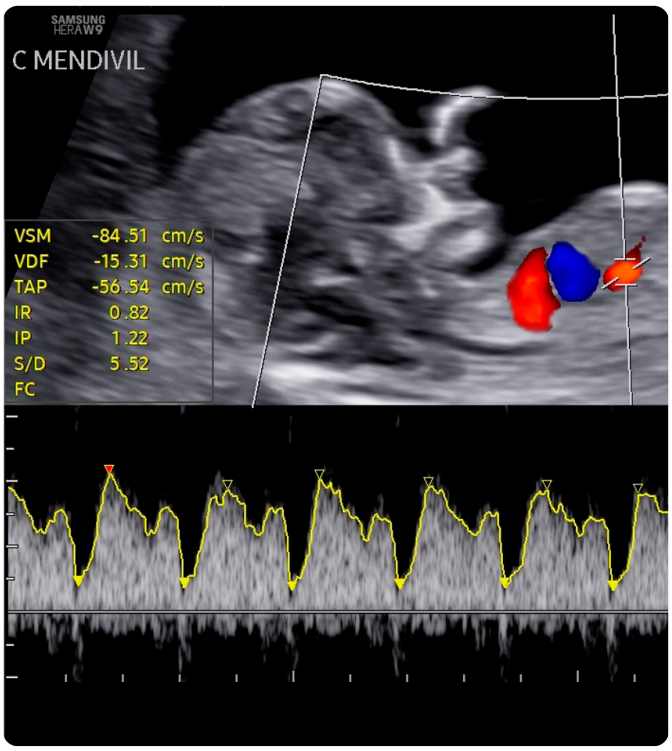


Figure 3. Measurement of the fetal venous duct, with spectral representation and peak systolic velocity measurement. No alterations in the wave components are observed.



Figure 4. Complete sagittal section of the fetal profile, marked with the word "intestines," showing them floating freely in the fluid, without membrane or vascular enhancement. The lack of intestinal content is due to early gestation.

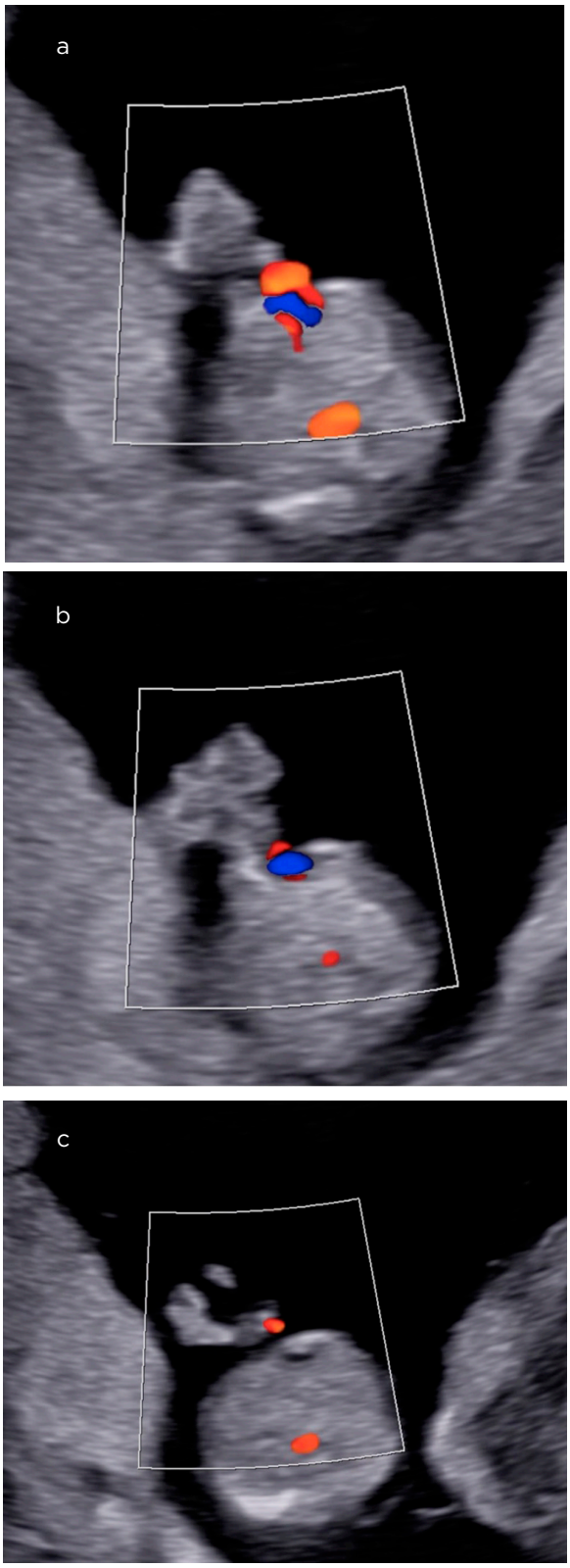


Figure 5. a) Axial section of the fetal abdomen at the level of the cord. Right-sidedness of the defect is confirmed with respect to the cord, which shows enhancement and normal morphology. The rounded image with flow on color Doppler examination corresponds to the abdominal aorta. b) Axial examination of the image in the fetal abdomen confirms a microlobulated morphology without enhancement, located in all lateral sections of the cord. c) A more superior section reveals the serpentine morphology of the intestinal loops.

Discussion

Gastroschisis is an anterior abdominal wall defect with an estimated incidence of 0.4-2.3 cases per 10,000 live births. Maternal cigarette smoking, abuse of vasoactive substances, and young maternal age are considered risk factors for the disease (3).

Although genetic studies in fetuses with this condition are usually normal, in some cases intestinal abnormalities may be found, ranging from motility disorders, obstruction, perforation, intestinal atresia, and stenosis to peritonitis or necrotizing enterocolitis. With the recent increase in comprehensive evaluation during the first trimester, in search of markers of morphological and/or genetic alterations, there has been an increase in the number of anterior wall alterations diagnosed in this early stage of pregnancy, which allows for appropriate follow-up, accurate planning of the delivery route, and the implementation of an interdisciplinary team prepared for eventualities during and after birth (4).

Embryological theories on the origin of omphalocele and gastroschisis have been studied and proposed, but none of them can explain the appearance of the latter on their own. Marquat et al., in a systematic review of 2024, sought to compile literature on the genetic etiopathogenesis of gastroschisis. They used data from fourteen published articles on human genetics, all with cohorts of varying sizes (from 40 to 1,966 individuals); In the end, they were unable to reliably identify the genetic component of this pathology or recognize chromosomal aberrations in the karyotype or microarray, concluding that, apparently, single variations in specific genes rarely cause gastroschisis. Therefore, studies combining genetic and epigenetic analysis are needed to determine the role of this component in the onset of the defect (5).

Once the prenatal diagnosis has been made, ultrasound scans are scheduled every three weeks, paying special attention to findings suggestive of fetal growth restriction (oligohydramnios) or intestinal atresia (polyhydramnios). The associated finding of fetal growth restriction is predictive of an adverse fetal prognosis. As for the timing of termination of pregnancy, it has been established that doing so before 35 weeks adds the morbidity of prematurity to that of gastroschisis, therefore, it is usually recommended to wait until 36-37 weeks (6, 7). Although the route of termination is usually cesarean section, there is conflicting evidence, depending on the population and institution, so this decision is left to the treating specialist team (8).

The purpose of surgical management is to reduce the herniated viscera into the peritoneal cavity and close the wall defect, while minimizing the risk of abdominal compartment syndrome. The condition of the exposed loops (amniotic fluid is harmful to them) and the degree of disproportion between the abdomen and herniated viscera dictate the type and timing of surgery, while taking into account factors such as neonatal weight and other associated comorbidities. There are two surgical options: primary surgery and two-stage surgery.

Primary reduction consists of bringing the herniated loops into the abdominal cavity without causing excessive intracavitary pressure, after which the defect in the wall is closed. This approach is sometimes associated with intra-abdominal compartment syndrome, a potentially fatal complication in which extensive intestinal ischemia, lower limb ischemia, and kidney ischemia develop (9).

When the pediatric surgery team suspects the possibility of progression to abdominal compartment syndrome, they may choose to place the intestinal loops inside a bag to protect them from the external

environment. The distal ring of this bag is inserted through the wall defect into the abdominal cavity. Subsequently, without the need for orotracheal intubation, the intestinal contents are gradually reduced into the abdomen on a daily basis. This approach has a survival rate of over 90% (10).

Newborns with gastroschisis tend to require prolonged parenteral nutrition, usually due to intestinal ileus and other disorders of intestinal peristalsis, which lead to varying degrees of intolerance to enteral feeding. The prognosis is favorable, with high survival rates; however, it is a condition associated with multiple morbidities that influence the final fetal prognosis. Complex gastroschisis—associated with complications arising from the pathology—is associated with up to a sevenfold increase in mortality compared to simple gastroschisis—without complications (11).

The relevance of the case presented, despite the voluntary termination by the parents—which is not indicated—is illustrated by the importance of considering first-trimester ultrasound as a study in which fetal malformations can be identified without the need to wait for a detailed anatomical sonogram and beyond altered nuchal translucency, ductus arteriosus, or nasal bone. At all stages of pregnancy, the embryo and then the fetus must be thoroughly evaluated in order to ensure timely and early diagnoses and provide adequate counseling to parents, culminating in a birth attended by a competent interdisciplinary team contextualized to the case.

Radiologically, the sensitivity and specificity of ultrasound for detecting gastroschisis in the first trimester are estimated to be variable, depending on the population studied. Sensitivity of up to 75% and specificity of up to 95% have been reported, making the use of other imaging modalities unnecessary. The key ultrasound sign for diagnosis is intestinal loops protruding through a defect in the anterior wall, often located to the right of the umbilical cord; this does not show enhancement on Doppler and, depending on gestational age, may be seen as collapsed loops (first trimester) or with intestinal contents (end of second trimester). Fetal monitoring is also performed by ultrasound, as this allows adequate assessment of the evolution towards probable complications in the loops compromised in the defect (ischemia). Magnetic resonance imaging plays an academic role in the study and monitoring of gastroschisis, but it is not the routinely proposed diagnostic method (12).

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