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The Role of Ultrasound in Diagnosing Fetal Anterior Abdominal Wall Anomalies

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ABSTRACT

Introduction: Anomalies of the fetal anterior abdominal wall result from defective embryonic development or failure of embryonic fusion, potentially leading to life-threatening conditions. The incidence is approximately 6.3 per 10,000 pregnancies [1]. Prenatal ultrasound serves as a key diagnostic tool for early detection and management planning [2].

Methods: This study is a retrospective case series conducted over three years (January 2021 to December 2023) at the Kairouan Maternity Hospital. We analyzed medical records of seven cases diagnosed with fetal anterior abdominal wall anomalies via prenatal ultrasound.

Results: Among the seven cases, four were diagnosed with omphalocele, two with gastroschisis, and one with bladder exstrophy. Two cases were identified early (13 and 22 weeks of gestation), leading to medical termination of pregnancy. The remaining cases were followed to term, with postnatal surgical management.

Conclusion: Fetal anterior abdominal wall anomalies, though rare, significantly impact fetal prognosis due to potential associated malformations and chromosomal abnormalities. Prenatal ultrasound remains an essential tool for early diagnosis, prognosis assessment, and guiding clinical management.

KEYWORDS
Anterior abdominal wall, omphalocele, gastroschisis, bladder exstrophy, prenatal ultrasound

INTRODUCTION

Fetal anterior abdominal wall anomalies (FAAWA) result from defects in embryonic folding or closure, affecting structures from the thoracoabdominal junction to the pelvic region These defects, which include omphalocele, gastroschisis, and bladder exstrophy, occur in approximately 6.3 per 10,000 pregnancies. They are classified based on their location: upper (thoraco-abdominal junction), middle (peri-umbilical region), and lower (pelvic region) coelosomies[3]. Given their potential lethality, early detection via ultrasound is crucial for optimizing perinatal outcomes. This study evaluates the role of ultrasound in diagnosing FAAWA and its impact on prenatal and postnatal management.

MATERIALS AND METHODS

Study Design and Setting

This is a retrospective descriptive case series conducted over three years (January 2021 to December 2023) at the maternity ward of Kairouan, Tunisia.

Study Population

Eligible participants included all pregnant women who underwent ultrasound examination at any gestational age within the study period. Cases with diagnosed fetal anterior abdominal wall anomalies were included.

Data Collection

Data were extracted from medical records, including:

- Maternal age, obstetric history, and prenatal care details
- Fetal ultrasound findings: type of anomaly, gestational age at diagnosis
- Associated anomalies and karyotype results
- Pregnancy outcome and postnatal management

Ultrasound examinations were performed by a senior obstetrician and validated by a second specialist.

RESULTS

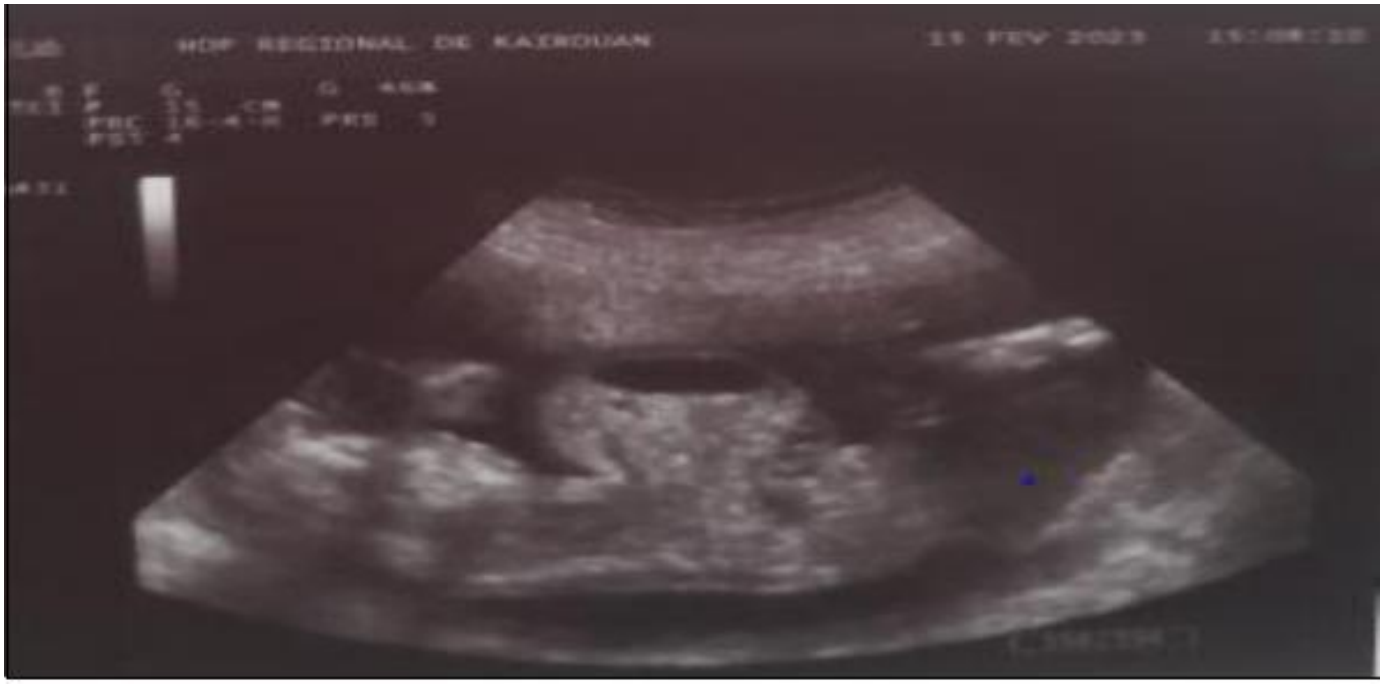
Seven cases of FAAWA were identified ; Among the seven cases, four were diagnosed with omphalocele, two with gastroschisis, and one with bladder exstrophy. The mean maternal age was 26 years, and the mean gestational age at diagnosis was 22 weeks.Two cases were identified early (13 and 22 weeks of gestation), leading to medical termination of pregnancy. The remaining cases were followed to term, with postnatal surgical management.

Case 1: Omphalocele associated with Trisomy 18

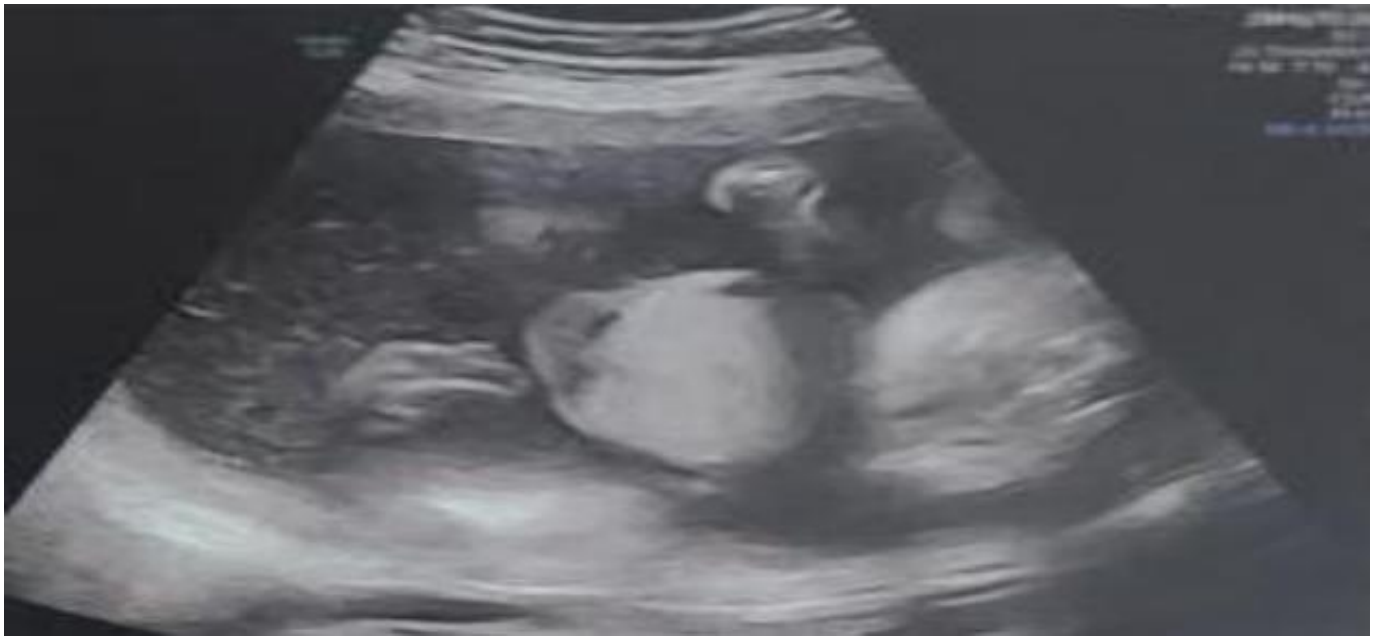
A 22-year-old woman, with no prior medical history, G2P1A0, with a scarred uterus and a healthy child, presented at 22 weeks + 1 day of gestation for pregnancy follow-up due to inadequate prenatal care.

A detailed morphological ultrasound was performed, revealing a singleton pregnancy in breech presentation, with biometric measurements consistent with gestational age, excessive amniotic fluid, and a polymalformative syndrome with the following abnormalities:

- **Cranial findings:** Anencephaly was observed.
- **Abdominal findings:** A hypoplastic thorax and a significant omphalocele containing intestinal loops surrounded by a membrane were identified.
- **Limb findings:** Arthrogryposis was noted, with no other detected anomalies.



Ultrasound image showing a giant omphalocele with its neck at 22 weeks of gestation.



Ultrasound image showing an associated anencephaly.

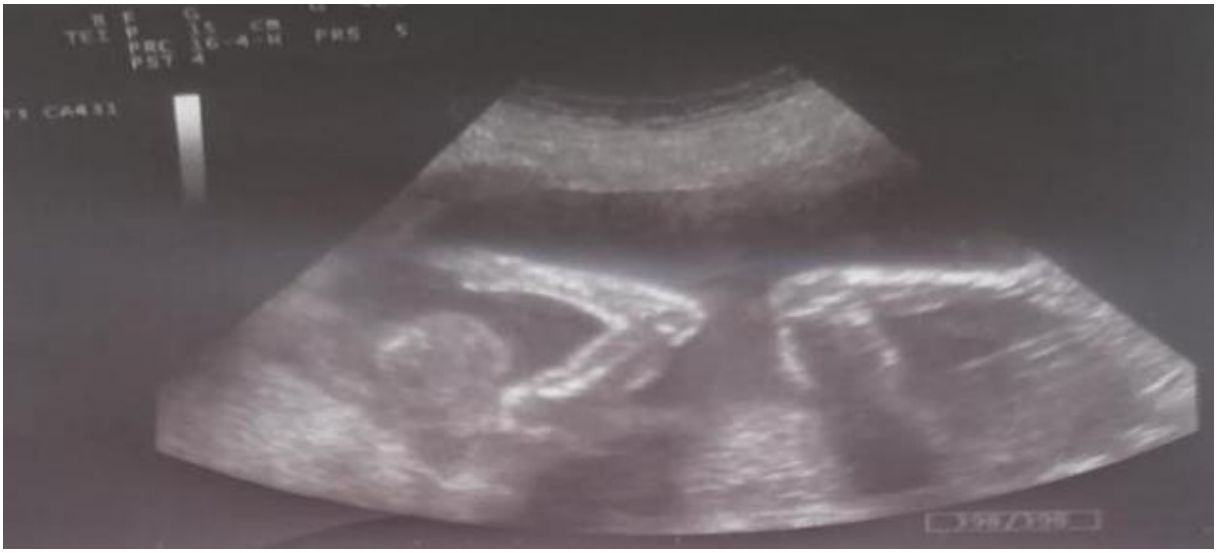


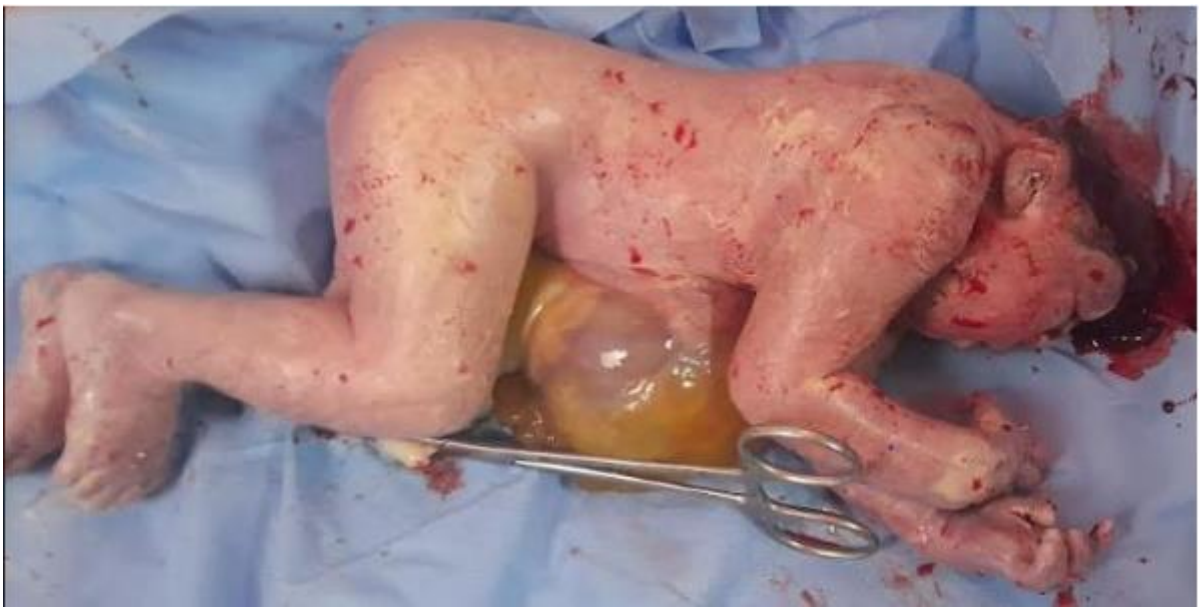
Image showing an associated arthrogryposis.

An amniocentesis was performed. Molecular cytogenetic analysis confirmed a fetal karyotype of 47, XY, +18.

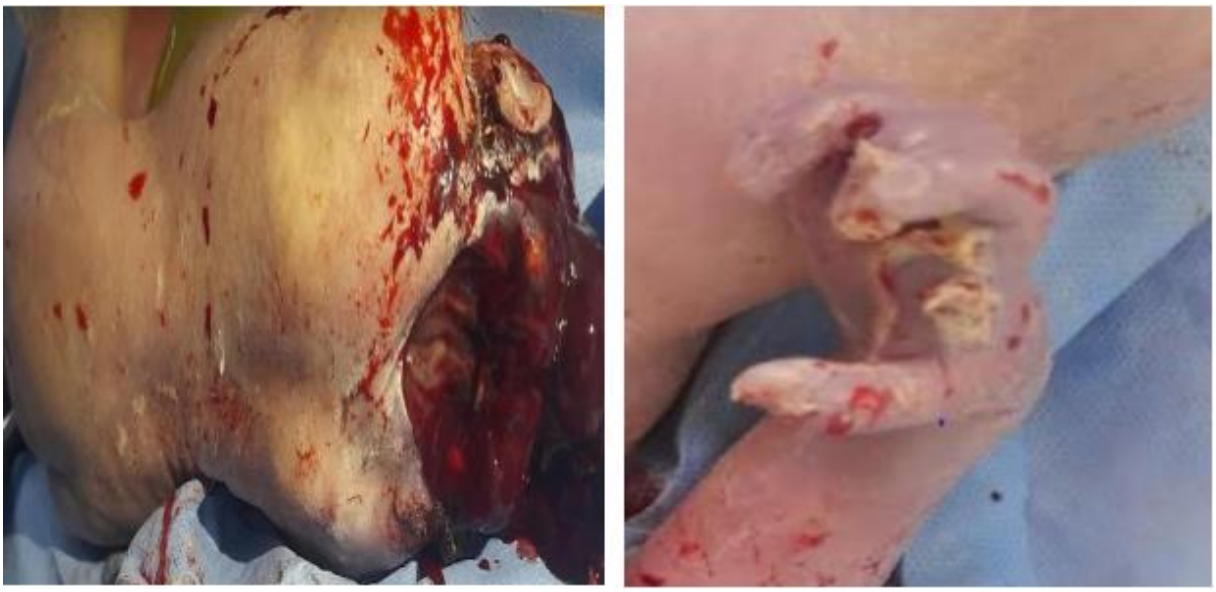
The patient was offered a medical termination of pregnancy, which she declined, and she was subsequently lost to follow-up.

At 33 weeks of gestation, she returned for consultation due to pelvic pain and significant fluid leakage. Upon examination, cervical dilation of 8 cm was noted, with breech presentation.

An emergency cesarean section was performed, delivering a male newborn with an Apgar score of 1 and an estimated fetal weight of 1400 g. The newborn presented with a complex polymalformative syndrome incompatible with life, including total anencephaly, a giant omphalocele with externalized intestinal loops, and arthrogryposis. Cardiorespiratory arrest occurred 15 minutes after birth.



Ventral wall hernia centered on the umbilical cord, where the externalized viscera are covered by the avascular amniotic membrane at 33 weeks of gestation.



Anencephaly Arthrogryposis
Actual images taken at 33 weeks of gestation showing the associated anomalies.

Case 2: Isolated Omphalocele

A 29-year-old woman, with no prior medical history, G2P1 (vaginal birth), was referred to our clinic for a detailed ultrasound examination. The parents were healthy, non-consanguineous, and had no family history of congenital anomalies.

The pregnancy was uneventful, with no signs of hyperemesis gravidarum, infections, or exposure to teratogens.

An ultrasound performed at 24 weeks of gestation revealed a 30 mm anterior abdominal wall defect containing digestive structures, with the umbilical cord centrally inserted within the defect. These findings suggested a ruptured omphalocele, with no associated anomalies detected.

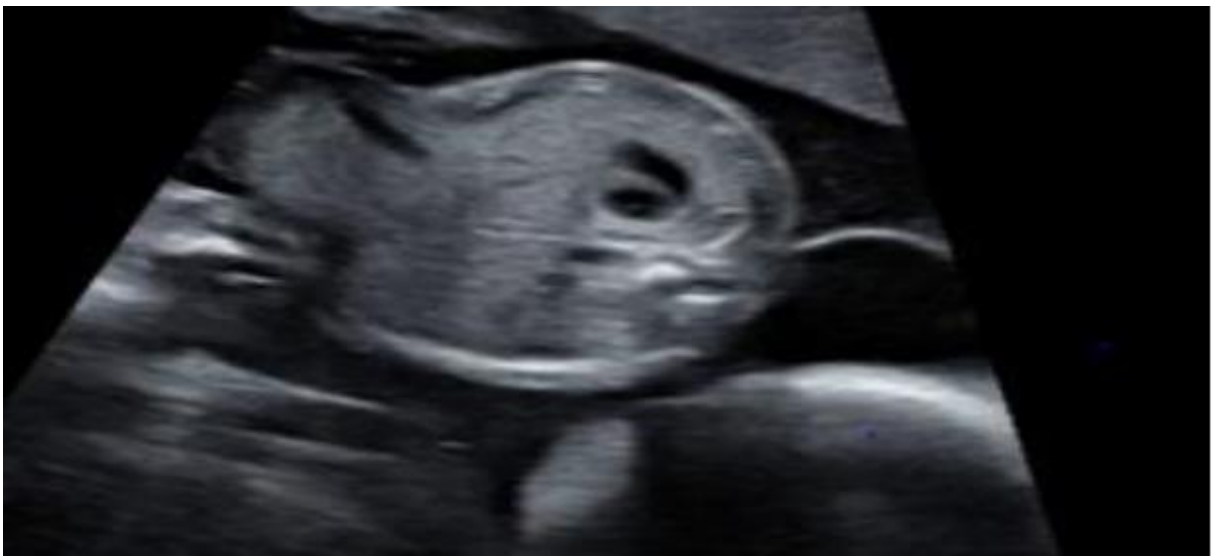


Image showing a 30 mm anterior abdominal wall defect with digestive content, with the umbilical cord centrally inserted within the defect.

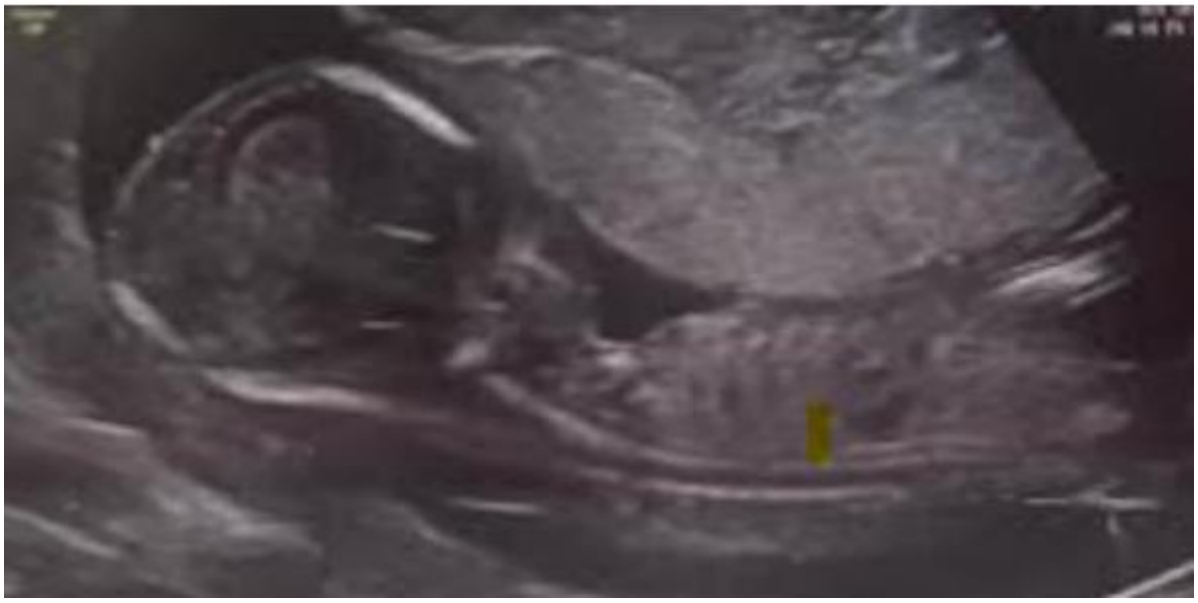
The patient went into spontaneous labor at 38 weeks and 1 day of amenorrhea, delivering a female newborn weighing 2860 g with an isolated omphalocele.

The baby was referred to a pediatric surgery unit for surgical management.

Case 3: Giant Omphalocele with Hygroma

A 32-year-old patient, G1P0 with no prior medical history, presented with first-trimester vaginal bleeding at 13 weeks of gestation.

Ultrasound examination revealed a **giant omphalocele**, with herniation of intestinal loops and liver, associated with a **multiloculated cystic posterior cervical mass**, suggestive of a **cystic hygroma**.

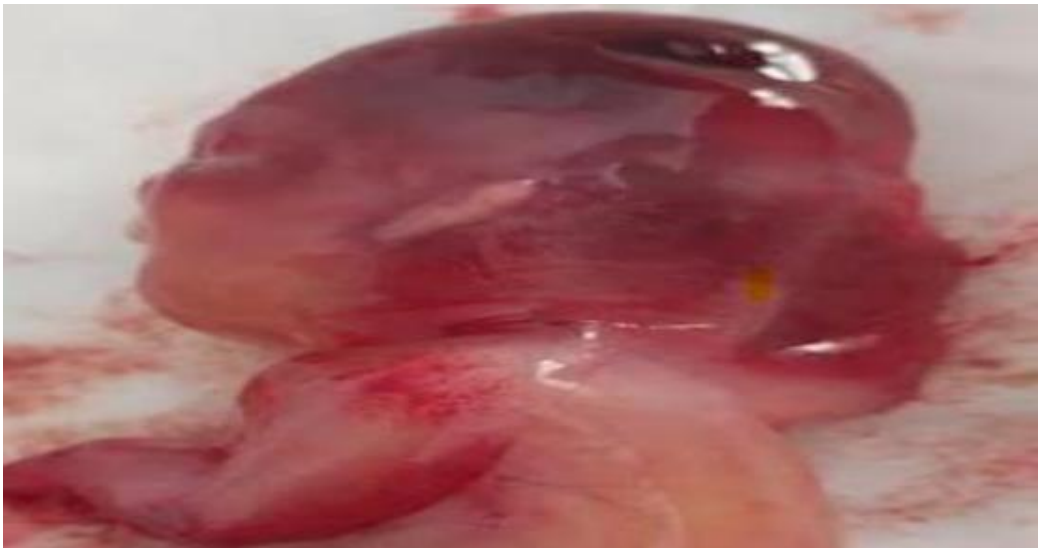


Ultrasound image showing a cystic hygroma at 13 weeks of gestation.

A trophoblastic biopsy was performed, and molecular cytogenetic analysis confirmed a fetal karyotype of 45, XX.

A medical termination of pregnancy (MTP) was proposed with the patient's consent.

The MTP was carried out using the Cytotec protoc



Real image showing a cystic hygroma at 13 weeks of gestation.

Case 4: Omphalocele in a Diabetic Mother

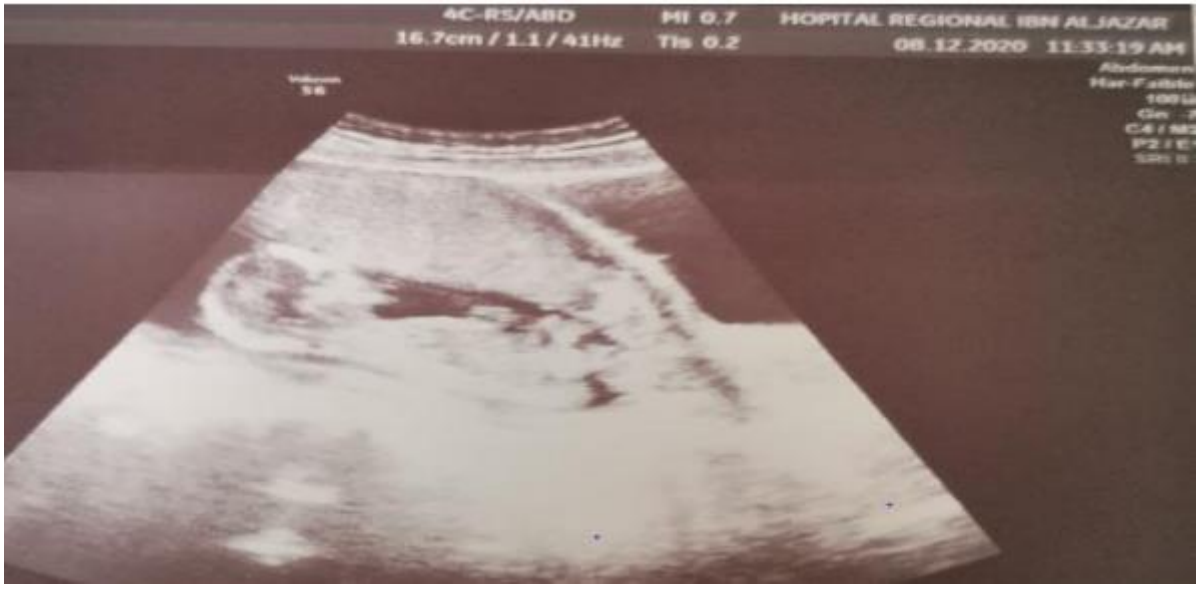
A 23-year-old patient, insulin-dependent diabetic, G3P2 (2 vaginal births), with two healthy children, presented for pregnancy follow-up.

Ultrasound at 29 weeks of gestation showed a singleton male fetus with an omphalocele, where the neck diameter measured less than 2 cm. The content of the omphalocele consisted exclusively of intestinal loops, with an intact membrane and an umbilical cord insertion located below the mass. The amniotic fluid volume was normal, and no additional malformations or syndromes were detected.

The patient was monitored by serial ultrasounds and delivered vaginally at 39 weeks of gestation. The newborn was immediately referred to the pediatric surgery department for further management.

Case 5: Gastroschisis with Fetal Demise

A 19-year-old primigravida at 13+4 weeks presented for routine follow-up. Ultrasound showed a paraumbilical abdominal wall defect with exteriorized loops of bowel, without a covering membrane, diagnostic of gastroschisis. The patient was lost to follow-up until 33 weeks, when she presented with decreased fetal movements. Ultrasound confirmed an intrauterine fetal demise with associated polyhydramnios.



Ultrasound image showing intestinal loops floating in the amniotic fluid without a hernial membrane.

Case 6: Gastroschisis with Postnatal Complications

A 21-year-old primigravida presented at 29 weeks with preterm contractions. Ultrasound revealed free-floating bowel loops within the amniotic fluid, characteristic of gastroschisis. Serial ultrasounds were performed every 7–15 days to monitor intestinal health. At 35 weeks, bowel dilation and caliber disparity suggested bowel compromise. Emergency cesarean section was performed, but the neonate died on day 2 due to severe intestinal dysfunction.

Case 7: Bladder Exstrophy

A 38-year-old multiparous woman was referred at 22 weeks for suspected bladder exstrophy. Ultrasound confirmed persistent non-visualization of the bladder and a subumbilical mass corresponding to the

exstrophied bladder plate. The couple opted for medical termination of pregnancy, which was performed successfully.

DISCUSSION

Ultrasound in Diagnosis and Prognostic Evaluation

Ultrasound is the primary diagnostic tool for FAAWA, with a sensitivity approaching 90% in experienced centers[4]. Features such as defect location, sac presence, and associated malformations aid in differentiation[5]. Doppler ultrasound enhances accuracy, especially for assessing vascular supply and bowel viability in gastroschisis.

Obstetric ultrasound is the gold standard for FAAWA detection, enabling early identification of syndromic cases and guiding prognosis. The addition of first-trimester nuchal translucency screening aids in detecting aneuploidies.

Procedures such as chorionic villus sampling and amniocentesis confirm chromosomal abnormalities[6].

This study underscores the value of obstetric ultrasound in improving outcomes for fetuses with FAAWA. By enabling early detection and comprehensive evaluation, ultrasound provides critical information for parental counseling and clinical decision-making. Future studies should focus on optimizing ultrasound protocols and integrating advanced imaging techniques, such as fetal MRI, to further enhance diagnostic accuracy and patient care.

Classification and Pathophysiology

Fetal anterior abdominal wall anomalies (FAAWA) are categorized based on the location of the defect relative to the umbilicus. Middle coelosomies, including omphalocele and gastroschisis, are the most common[7]. Omphalocele arises due to a failure of midgut retraction into the abdominal cavity, whereas gastroschisis results from a right-sided paraumbilical defect exposing the intestines to amniotic fluid[8].

Embryological Basis

These anomalies originate from disruptions in embryonic folding between the 3rd and 6th weeks of gestation. Omphalocele occurs when the midgut remains in an extra-abdominal position beyond the 10th week, while gastroschisis is associated with vascular compromise of the omphalomesenteric artery[9].

Anomalies Detected by Ultrasound

Upper Coelosomies

Upper coelosomies are rare, with an incidence of approximately 1 in 50,000 pregnancies[10]. They result from a defect in the embryonic development of the thoraco-abdominal junction. Pentalogy of Cantrell, a severe form of upper coelosomy, includes omphalocele, diaphragmatic hernia, sternal cleft, ectopia cordis, and cardiac malformations. No cases of upper coelosomies were diagnosed in our study.

Midline Coelosomies

Midline coelosomies are the most common and primarily include omphalocele and gastroschisis.

Omphalocele: With an incidence of 1 in 3,000 live births, omphalocele results from the failure of the physiological herniation of the intestines to return to the abdominal cavity between the 8th and 12th weeks of gestation and should be re-evaluated at **15 weeks**. [11]. It is characterized by a midline abdominal wall defect covered by a membrane, with variable contents ranging from intestinal loops to the liver. The **size and contents** of the omphalocele vary:

- **Small omphaloceles** (≤ 4 cm), usually containing **only the small intestine**.
- **Giant omphaloceles** (> 5 cm), which may include **intestines, stomach, and liver**

Associated malformations are present in 70% of cases. In our study, four cases of omphalocele were diagnosed, with two associated with chromosomal abnormalities mainly **trisomy 18, 21, and 13**.

Gastroschisis: This anomaly presents as a paraumbilical right-sided defect with herniated intestinal loops not covered by a membrane, affecting all layers of the anterior abdominal wall. [12]. It is usually isolated,**not associated with chromosomal abnormalities** and more common in younger mothers. In our study, two cases of isolated gastroschisis were diagnosed. Prenatal

ultrasound findings include herniated intestinal loops without a membrane and a normally inserted umbilical cord to the left of the defect.

Lower Coelosomies

Lower coelosomies include bladder and cloacal exstrophy, which are rare and often associated with complex genitourinary and musculoskeletal anomalies [13].

- **Bladder Exstrophy:** Characterized by a defect in the lower abdominal wall and an open bladder. Prenatal ultrasound findings include the absence of a visible bladder, a normal upper urinary tract, and a protruding bladder plate in the infraumbilical region.

- **Cloacal Exstrophy**: A rare defect involving herniation of the bladder and intestines. No cases were diagnosed in our study.

Ultrasound in Diagnosis and Prognostic Evaluation

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The prenatal ultrasound diagnosis of omphalocele typically shows:

- A well-defined midline abdominal mass, surrounded by a membrane, containing hyper-echogenic intestines and liver.

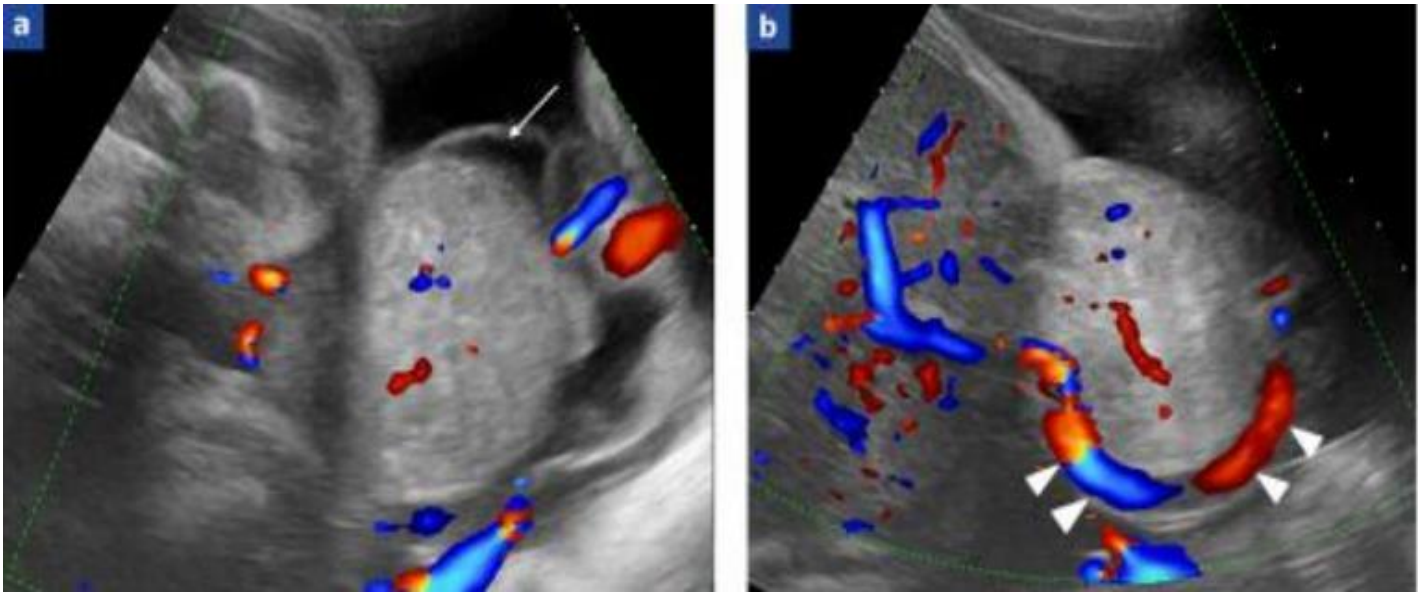
- Umbilical vein remains centered, an important diagnostic feature .

- Lower cord insertion relative to the mass.

- Doppler imaging helps visualize vascular elements.

- Possible ascites within the omphalocele sac and mild polyhydramnios .

When omphalocele is diagnosed, screening for associated malformations and chromosomal abnormalities is crucial for prognosis.



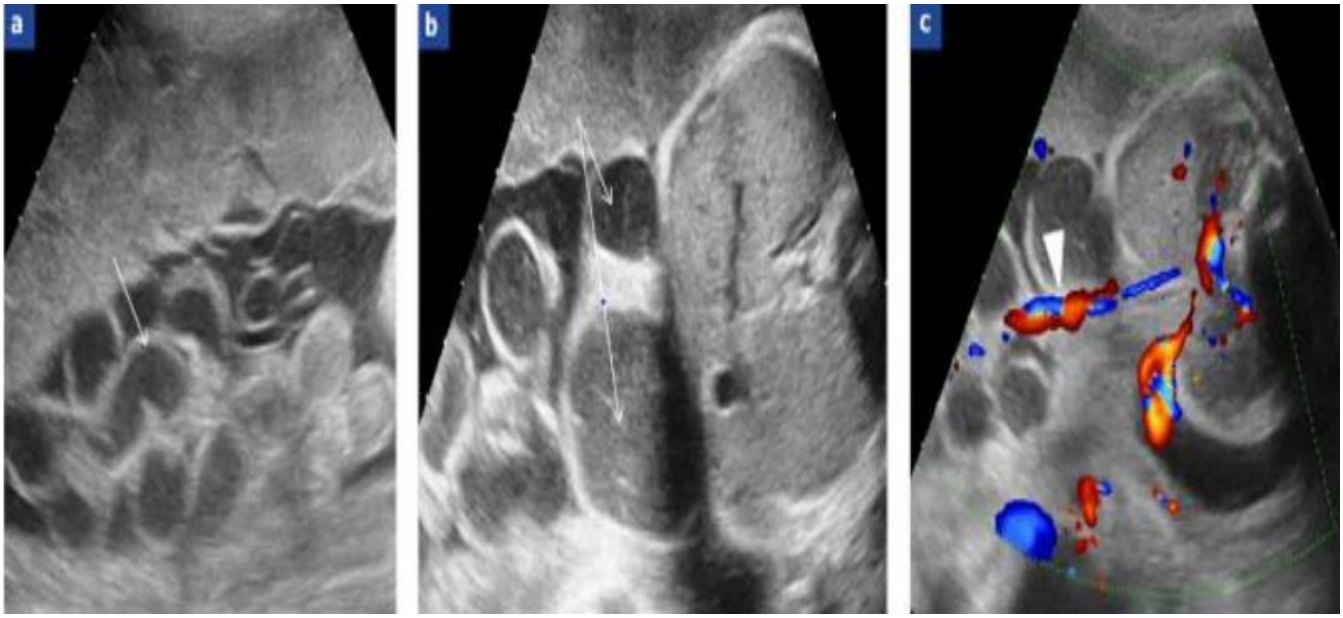
Axial sections (a, b) with color Doppler. Omphalocele with hyperechoic content, corresponding to the digestive tract. The umbilical cord runs along the wall of the hernia sac (arrowheads). The sac contains ascites (arrow) and is associated with mild polyhydramnios. [1].

Prenatal Ultrasound Features of Gastroschisis:

- **Absence of a hernial membrane** surrounding the intestinal loops.
- **Exteriorized intestinal loops** floating freely in **amniotic fluid** through a **right paraumbilical defect**.
- **Normal umbilical cord insertion** on the **left border of the defect** .

The **main fetal complications** are intestinal, due to prolonged exposure to amniotic fluid and vascular compression at the defect, which may lead to:

- **Intestinal atresia**.
- **Stenosis, obstruction, or perforation**.
- **Necrosis and peritonitis**



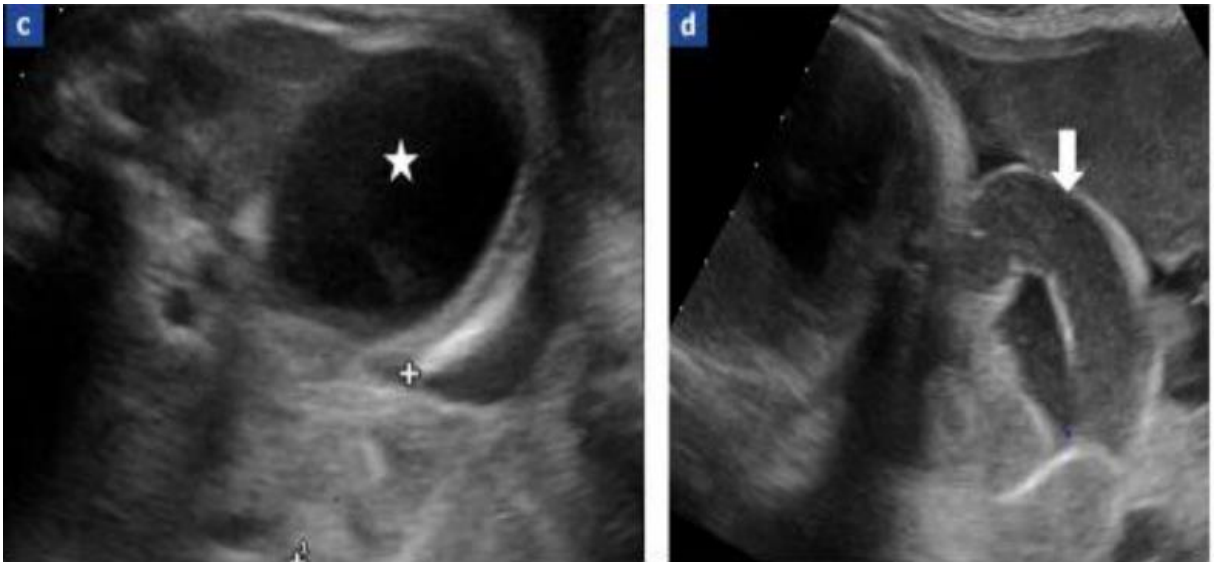
Axial sections without (a, b) and with color Doppler (c) showing gastroschisis with a narrowed defect and exteriorization of the stomach and small bowel loops with echogenic content floating in the amniotic fluid (arrows), accompanied by mesenteric vessels (arrowhead). [1].

Prenatal Ultrasound Diagnosis of Inferior Coelosomies:

- **Absence of fetal bladder visualization**, despite repeated ultrasound examinations.
- **Normal amniotic fluid volume**, ruling out upper urinary tract abnormalities.
- **Normal umbilical cord insertion**.
- **Infraumbilical mass**, corresponding to **exposed bladder mucosa**.

Bladder exstrophy is **usually isolated**, but it may be associated with other congenital anomalies, including:

- **Rectovesical fistula**.
- **Terminal colon agenesis**.
- **Anorectal malformations**.
- **External genital anomalies**.
- **Lower limb malformations**.



Sections c/d: showing dilated digestive loops (thick arrow) with a thickened wall and echogenic content, associated with a disparity in the caliber of intraperitoneal loops and gastric distension (star), suggesting intestinal distress. [1].

Comparison with Existing Literature

In this study, we identified four cases of omphalocele, two cases of gastroschisis, and one case of bladder exstrophy.

The prevalence of omphalocele in our cohort was four cases over three years, which is lower than what has been reported in previous studies.

Research conducted by Hidaka N et al. in Japan documented two cases per year [5].

Similarly, Floortje C et al. in the Netherlands observed an annual incidence of 3.3 cases [6].

Higher rates were noted in Germany, where Dingemann C et al. reported nine cases per year [7], and in Australia, where Kong JY et al. identified 9.7 cases annually [8].

The mean maternal age in our study was 26 years, which aligns with findings in the literature. Parity ranged from 0 to 3.

Regarding gestational age at diagnosis:

One case was identified in the first trimester.

One case in the second trimester.

One case in the third trimester. The mean gestational age at diagnosis ranged from 22 to 29 weeks, similar to findings from a 2017 Tunisian study, where 17% of cases were detected in the first trimester, while 71.4% and 77.8% were identified in the second and third trimesters, respectively [1]. In contrast, Mitanchez et al. (France, 2019) reported an earlier average detection time of 13 weeks [9]. Among the four omphalocele cases in our study, one involved both intestinal loops and the liver, a proportion similar to that found in a 2019 French study [10]. However, this rate was higher than that reported in a Tunisian study, which documented ten cases of intestinal loop herniation but only one case involving the liver [1]. Additionally, two out of four omphalocele cases were linked to chromosomal abnormalities, specifically trisomy 18 and monosomy X, consistent with findings in the literature [1,5].

A medical termination of pregnancy (MTP) was proposed in two cases due to non-isolated omphalocele.

Our study identified two cases of isolated gastroschisis.

The affected patients were generally younger than those diagnosed with omphalocele [1], with a mean maternal age of 20 years, a trend that aligns with existing literature.

Clinical Management and Prognosis

Management varies by anomaly type. Omphalocele cases associated with genetic syndromes have a high mortality rate, while isolated cases typically have good surgical outcomes[14]. Gastroschisis management involves monitoring bowel viability and timely delivery in cases of suspected ischemia.

Bladder exstrophy necessitates a multidisciplinary approach involving neonatologists and pediatric urologists. Surgical advancements have improved continence and reproductive outcomes.

Postnatal Outcomes

Postnatal management focuses on prompt surgical intervention. Gastroschisis requires immediate bowel protection and staged closure, while omphaloceles may need delayed repair depending on defect size[15]. Bladder exstrophy requires multiple reconstructive surgeries, emphasizing the need for long-term follow-up.

CONCLUSION

Fetal anterior abdominal wall defects, though rare, pose significant challenges for prenatal diagnosis and management. Obstetric ultrasound is a vital tool for early detection, prognosis assessment, and guiding clinical decisions. This study highlights the importance of ultrasound in improving outcomes for affected fetuses and underscores the need for multidisciplinary care in managing these complex anomalies. Early diagnosis and timely intervention can significantly enhance the quality of life for neonates with FAAWA and provide essential support for their families.

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