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Unraveling Dual Anomalies: A Case Of Acrania With Gastroschisis

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ABSTRACT

Acrania and gastroschisis are rare congenital anomalies that are typically incompatible with life. Acrania is a severe neural tube defect characterized by the absence of the cranial vault, often progressing to anencephaly. Gastroschisis is a congenital anterior abdominal wall defect with herniation of bowel loops, usually isolated but occasionally found in association with other anomalies. The coexistence of acrania with gastroschisis is exceptionally rare, with very few cases reported in the literature. We report the case of a 28-year-old gravida 2, para 1 woman who presented at 16 weeks of gestation with abdominal pain. She had a previous cesarean section and a history of severe oligohydramnios in her first pregnancy. The current pregnancy was un booked, and she had not taken folic acid supplementation. Antenatal ultrasonography revealed a fetus with acrania and gastroschisis. Clinical examination confirmed a 16-week-sized uterus with a closed cervix. In view of the poor prognosis, a second-trimester medical termination of pregnancy was performed. The abortus was a male fetus weighing 88 grams, with external features confirming the ultrasound findings—absence of the cranial vault and evisceration of intestines through the anterior abdominal wall. This case emphasizes the importance of early antenatal care, prenatal screening, and periconceptional folic acid supplementation in the prevention and early diagnosis of major congenital anomalies. Increased awareness and timely interventions are essential to reduce the incidence and associated morbidity of neural tube defects and abdominal wall anomalies in low-resource settings.

Keywords: *Acrania, Gastroschisis, Neural Tube Defect, Folic Acid, Medical Termination, Antenatal Care*

1. INTRODUCTION

Congenital anomalies are a major cause of perinatal morbidity and mortality worldwide. Among these, **neural tube defects (NTDs)** such as acrania and structural abdominal wall defects like **gastroschisis** represent significant challenges in prenatal diagnosis and management. While each condition independently is well-documented, the co-occurrence of acrania with gastroschisis is exceedingly rare and sparsely reported in the literature [1]. **Acrania** is a fatal congenital malformation characterized by the complete or partial absence of the flat bones of the cranial vault. It occurs due to a failure of the cranial mesenchyme to migrate and ossify, resulting in an exposed brain that degenerates over time. It is often considered an early form of anencephaly and can be detected as early as 11 weeks of gestation through ultrasonography. The condition is universally fatal, with affected fetuses either being stillborn or dying shortly after birth [2].

Gastroschisis, on the other hand, is a paraumbilical full-thickness defect of the anterior abdominal wall through which abdominal contents, particularly bowel loops, herniate without a covering sac. Although often isolated and survivable with surgical correction after birth, its association with other anomalies—especially neural tube defects—is uncommon and portends a poor prognosis [3]. Folic acid supplementation before conception and during early pregnancy plays a critical role in preventing neural tube defects. Lack of such supplementation, especially in unbooked pregnancies with limited prenatal care, remains a significant risk factor in many low-resource settings [4].

In this case report, we present a rare and fatal case of acrania with gastroschisis identified in the second trimester of pregnancy in an unbooked patient who did not take folic acid. This report underscores the need for early antenatal booking, routine ultrasound screening, and nutritional counselling to reduce the burden of preventable congenital anomalies**.**

2.CASE PRESENTATION

A 28-year-old woman, gravida 2 para 1 living 1, presented to the labor ward at 16 weeks of gestation with complaints of lower abdominal pain persisting for one day. The pain was dull, non-radiating, and not associated with vaginal bleeding, discharge, or fever. She had been married for six years and had one previous live birth delivered via lower segment cesarean section (LSCS) four years ago due to severe oligohydramnios. Her previous child was a healthy female with no congenital anomalies. This pregnancy was unbooked, and the patient had not received any antenatal care before presentation. She had not taken folic acid or iron supplementation during the periconceptional period or during the early weeks of gestation. There was no history of chronic illness, exposure to teratogenic substances, radiation, or familial genetic disorders. She denied any history of smoking, alcohol use, or illicit drug intake. Her menstrual history was regular, and there was no consanguinity between her and her partner.

Physical Examination

On general physical examination, the patient was conscious, alert, and hemodynamically stable. Her vital signs were within normal limits: blood pressure was 110/70 mmHg, pulse rate 84 beats per minute, respiratory rate 16 breaths per minute, and temperature 36.8°C. There was no pallor, icterus, cyanosis, clubbing, or edema. Cardiovascular and respiratory examinations were unremarkable. Abdominal examination revealed a well-healed Pfannenstiel scar consistent with a previous cesarean section. The uterus was palpable just above the pubic symphysis, corresponding to 16 weeks of gestation. Mild lower abdominal tenderness was noted, but there were no signs of guarding or rigidity. Bowel sounds were audible and normal. There were no palpable masses, and no evidence of ascites or peritonism. Per speculum examination was normal with no cervical discharge or bleeding. On per vaginal examination, the cervix was firm and closed, and the uterus was anteverted and non-tender.

Investigations

A transabdominal obstetric ultrasound was performed, which revealed a single live intrauterine fetus with a crown-rump length consistent with 16 weeks of gestation. Two significant congenital anomalies were noted:

a. **Acrania:** The fetal cranium was absent above the level of the orbits. Brain tissue appeared exposed and was surrounded by amniotic fluid without a bony covering. The facial bones and orbits were visualized normally, but the cranial vault was completely absent. This finding was consistent with acrania.

b. **Gastroschisis:** An anterior abdominal wall defect was observed to the right of the umbilical cord insertion, with free-floating bowel loops visible in the amniotic fluid. There was no membranous sac covering the herniated viscera, distinguishing it from omphalocele. The stomach and urinary bladder were visible and appeared normal. The umbilical cord had a normal three-vessel structure and inserted at the normal position, separate from the abdominal wall defect.

Other fetal structures, including the spine, heart, kidneys, and limbs, appeared normal. There was no evidence of hydrops or additional malformations. The amniotic fluid volume was within normal limits. A fetal echocardiogram was deferred due to the early gestational age and poor prognosis. Routine laboratory investigations including complete blood count, liver and renal function tests, fasting blood sugar, and serological screening for infections (TORCH panel, HIV, hepatitis B and C, VDRL) were all within normal limits. Blood group typing revealed A positive with negative antibody screening.

Diagnosis and Counseling

Based on clinical and ultrasonographic findings, a diagnosis of **acrania with gastroschisis** was made. The patient and her partner were counseled extensively regarding the poor fetal prognosis, the lethality of acrania, and the potential complications. After discussion and consent, a second-trimester medical termination of pregnancy (MTP) was planned and carried out as per hospital protocol. The procedure was uneventful. The abortus was a male fetus weighing 88 grams. On external examination, the fetus had a completely absent cranial vault with exposed brain tissue and a right-sided abdominal wall defect through which intestines were herniating, consistent with acrania and gastroschisis.

3. MANAGEMENT AND OUTCOME

Following confirmation of the diagnosis of acrania with gastroschisis through detailed ultrasonography, the patient was counselled extensively regarding the prognosis. It was explained that acrania is a lethal neural tube defect incompatible with life, and the coexistence of gastroschisis further complicated the fetal condition. The couple was informed about the nature of the anomalies, the absence of any curative treatment, and the likelihood of intrauterine or early neonatal demise should the pregnancy be continued [5]. Given the poor prognosis and associated maternal risks of continuing a pregnancy with major congenital anomalies, the patient opted for a **medical termination of pregnancy (MTP)**. Informed written consent was obtained after detailed counselling, including the potential complications and emotional implications of the procedure. The case was managed in accordance with the Medical Termination of Pregnancy Act guidelines and institutional protocols.

Figure 1: Acrania with gastrochisis

Figure 2 - Abortus with cranial vault absent

Termination was performed using a **second-trimester medical abortion regimen**. Mifepristone 200 mg was administered orally, followed 24 hours later by repeated doses of misoprostol 400 mcg administered vaginally every 3 hours until fetal expulsion was achieved. The patient was monitored closely throughout the procedure for signs of excessive bleeding, infection, or incomplete abortion. The MTP was completed without complications. The abortus was a male fetus weighing 88 grams. External examination confirmed the prenatal findings of absent cranial vault (acrania) and an anterior abdominal wall defect with exposed bowel loops (gastroschisis) [6]. No other anomalies were noted on gross inspection

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Parameter	Findings
Presenting Complaint	Abdominal pain for 1 day
General Examination	Stable vital signs; no systemic abnormalities
Abdominal Examination	Uterus corresponding to 16 weeks size; Pfannenstiel scar present
Per Vaginal Examination	Cervix closed; uterus anteverted
Ultrasound Findings	Acrania (absence of cranial vault); Gastroschisis (herniated bowel loops)
Fetal Sex and Weight	Male fetus; 88 grams
Other Anomalies	None observed
Laboratory Investigations	Within normal limits
Diagnosis	Acrania with Gastroschisis
Management	Second-trimester medical termination of pregnancy (MTP)
Method of Termination	Mifepristone 200 mg + Misoprostol 400 mcg regimen
Outcome	Successful MTP; no complications
Post-MTP Counselling	Contraception, periconceptional folic acid, early antenatal care in future

Table 1: Clinical Summary of the Case

The patient had an uneventful post-procedural recovery. She was given psychological support and counselled on the importance of **preconception care**, including **periconceptional folic acid supplementation**, and the need for early booking and regular antenatal care in future pregnancies [7]. She was discharged in stable condition with advice for follow-up and contraception counselling.

4. DISCUSSION

Acrania is a rare and fatal neural tube defect characterized by the absence of the cranial bones, often progressing to anencephaly. Gastroschisis is a congenital anterior abdominal wall defect, typically occurring to the right of the umbilical cord insertion, with herniation of bowel loops without a protective sac [8]. The coexistence of these two anomalies is exceedingly rare and not well documented in current literature. The etiology of such combined anomalies remains unclear but may involve multifactorial causes including genetic predisposition, teratogenic exposure, nutritional deficiencies, and environmental influences [9]. The lack of periconceptional folic acid supplementation is a known risk factor for neural tube defects, as seen in this case. While gastroschisis is often isolated, its presence alongside acrania suggests a more complex embryological disruption during early fetal development. Early antenatal diagnosis through ultrasonography is critical for detecting lethal congenital anomalies. In this case, the diagnosis at 16 weeks allowed for appropriate counselling and timely medical termination [10]. This highlights the importance of early booking and regular antenatal check-ups, especially in resource-limited settings.

Increased awareness, public health education, and promotion of folic acid use in women of reproductive age can significantly reduce the incidence of such preventable birth defects.

5. CONCLUSION

This rare case of acrania with gastroschisis highlights the critical importance of early prenatal care, timely ultrasonographic screening, and periconceptional folic acid supplementation in preventing major congenital anomalies. The absence of antenatal registration and folic acid intake in this patient contributed to a poor fetal outcome. Early diagnosis enabled appropriate counselling and management through second-trimester medical termination. This case underscores the need for increased public health awareness and strengthened antenatal services to ensure early detection and prevention of lethal fetal anomalies, ultimately improving maternal and neonatal outcomes in future pregnancies.

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