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Antenatal diagnosis of lethal multisystem fetal anomalies: a case report

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Abstract

Background: Multiple congenital anomalies are an important cause of perinatal morbidity and mortality, particularly when abnormalities involve several organ systems. Antenatal ultrasonography is essential for early recognition of major structural abnormalities and for appropriate parental counseling.

Case presentation: A 33-year-old woman was referred at 34 weeks of gestation for detailed fetal ultrasonography because of a previous intrauterine fetal demise associated with an undiagnosed congenital anomaly. The current pregnancy was complicated by severe polyhydramnios. Ultrasound demonstrated discordant fetal biometry, a large umbilical vein varix, non-visualization of the stomach, collapsed bowel loops, hepatomegaly, a small thoracic cavity, leftward displacement of the heart, clubfoot, and generalized fetal skin edema. The fetal brain and spine appeared normal, whereas facial assessment was limited by fetal position. The combination of severe polyhydramnios and multiple structural abnormalities raised strong suspicion of a severe multisystem congenital anomaly with a poor prognosis. Following delivery, esophageal atresia and severe pulmonary hypoplasia were confirmed, and the neonate died within a few minutes of birth.

Conclusion: This case highlights the importance of systematic antenatal anomaly assessment, particularly in pregnancies complicated by severe polyhydramnios and a previous pregnancy affected by congenital anomalies. Early detailed ultrasound examination may facilitate recognition of lethal abnormalities, multidisciplinary counseling, and appropriate pregnancy and perinatal management.

Keywords: Congenital Anomalies, Antenatal Diagnosis, Fetal Ultrasonography, Polyhydramnios, Esophageal Atresia, Pulmonary Hypoplasia, Iraq

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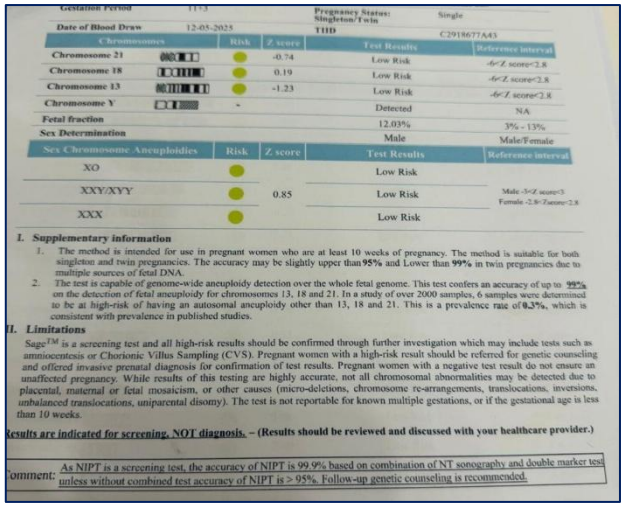
Background

Congenital anomalies are an important cause of perinatal morbidity and mortality and represent a continuing challenge for maternal and child health services, particularly in resource-limited settings.

The World Health Organization emphasizes that congenital disorders may be identified antenatally, at birth, or later in life, while adequate care before and during pregnancy and appropriate preventive interventions can reduce some preventable conditions [1]. In Iraq, available evidence indicates a substantial burden of fetal and congenital abnormalities, although national surveillance remains limited. A recent study from Tikrit reported that central nervous system, musculoskeletal, gastrointestinal, and cardiac anomalies constituted major proportions of congenital abnormalities presenting to hospital services [2]. An Iraqi study evaluating antenatal detection also emphasized the value of ultrasound examination, particularly among high-risk pregnancies, in identifying fetal structural abnormalities [3]. Furthermore, a recent study from Al-Qadissiyah documented a considerable burden of neural tube defects and found that antenatal diagnosis was achieved in approximately two-thirds of affected pregnancies, while inadequate antenatal supervision and folic acid use remained important concerns [4]. Recent Iraqi research has also highlighted the importance of maternal knowledge regarding pregnancy-related risk factors and adequate antenatal care [5]. This case describes the antenatal recognition of multiple severe fetal structural abnormalities associated with severe polyhydramnios and a subsequent lethal neonatal outcome, emphasizing the importance of timely and systematic fetal anomaly assessment in high-risk pregnancies.

Case presentation

A 33-year-old woman was referred for a detailed antenatal ultrasound examination at 34 weeks of gestation because of a previous intrauterine fetal demise associated with an undiagnosed congenital anomaly. She had previously undergone chromosomal analysis, which was reported as negative (figure 1).



Gestation Period 11+3 Pregnancy Status: Singleton/Twin Single

Date of Blood Draw 12-05-2025 THD C29186774A43

Chromosomes	Risk	Z score	Test Results	Reference interval
Chromosome 21		-0.74	Low Risk	<6 Z score<2.8
Chromosome 18		0.19	Low Risk	<6 Z score<2.8
Chromosome 13		-1.23	Low Risk	<6 Z score<2.8
Chromosome Y		-	Detected	NA
Fetal fraction			12.03%	3% - 13%
Sex Determination			Male	Male/Female
Sex Chromosome Aneuploidies	Risk	Z score	Test Results	Reference interval
XO			Low Risk	
XXY/XYY		0.85	Low Risk	Male <5 Z score<3 Female <2.8 Z score<2.8
XXX			Low Risk	

I. Supplementary information

1. The method is intended for use in pregnant women who are at least 10 weeks of pregnancy. The method is suitable for both singleton and twin pregnancies. The accuracy may be slightly upper than **95%** and Lower than **99%** in twin pregnancies due to multiple sources of fetal DNA.
2. The test is capable of genome-wide aneuploidy detection over the whole fetal genome. This test confers an accuracy of up to 99% on the detection of fetal aneuploidy for chromosomes 13, 18 and 21. In a study of over 2000 samples, 6 samples were determined to be at high-risk of having an autosomal aneuploidy other than 13, 18 and 21. This is a prevalence rate of **0.3%**, which is consistent with prevalence in published studies.

II. Limitations

Sage^{TM} is a screening test and all high-risk results should be confirmed through further investigation which may include tests such as amniocentesis or Chorionic Villus Sampling (CVS). Pregnant women with a high-risk result should be referred for genetic counseling and offered invasive prenatal diagnosis for confirmation of test results. Pregnant women with a negative test result do not ensure an unaffected pregnancy. While results of this testing are highly accurate, not all chromosomal abnormalities may be detected due to placental, maternal or fetal mosaicism, or other causes (micro-deletions, chromosome re-arrangements, translocations, inversions, unbalanced translocations, uniparental disomy). The test is not reportable for known multiple gestations, or if the gestational age is less than 10 weeks.

Results are indicated for screening, NOT diagnosis. – (Results should be reviewed and discussed with your healthcare provider.)

Comment: As NIPT is a screening test, the accuracy of NIPT is 99.9% based on combination of NT sonography and double marker test unless without combined test accuracy of NIPT is > 95%. Follow-up genetic counseling is recommended.

Figure 1. Chromosomal analysis

The current pregnancy was complicated by severe polyhydramnios. Ultrasonographic examination demonstrated fetal biometric measurements that were discordant with the gestational age, with a biparietal diameter corresponding to approximately 35 weeks, femur length to 33 weeks, and abdominal circumference to approximately 31 weeks. Multiple structural abnormalities were identified (Figure 2:A,B,C,D,E), including a large umbilical vein varix, non-visualization of the stomach, collapsed bowel loops, hepatomegaly, a small thoracic cavity, leftward displacement of the heart, and clubfoot deformity. Generalized fetal skin edema was also observed. The fetal face could not be adequately assessed because the fetus remained in a dorsal position during the examination. The fetal brain and spine appeared structurally normal on ultrasound. Given the combination of severe polyhydramnios, absent stomach visualization, thoracic abnormality, hepatomegaly, umbilical vein varix, limb deformity, and generalized edema, a severe multisystem congenital anomaly was suspected, with a poor prognosis. The possibility of a lethal fetal outcome was discussed with the family. During the scan, the neonate was confirmed to have esophageal atresia and pulmonary hypoplasia, in addition to multiple congenital anomalies identified antenatally. The overall clinical and imaging findings were consistent with a severe multisystem congenital anomaly syndrome. Sever pulmonary hypoplasia incompatible with life after delivery. Despite delivery, the neonate survived for only a few minutes and subsequently died, confirming the multiple congenital anomalies and the lethal neonatal outcome. The case highlights the importance of detailed congenital anomaly scan at second-trimester in pregnancies complicated by severe polyhydramnios. It is particularly important in pregnancies with a previous pregnancy affected by congenital anomalies. This may allow early termination of pregnancy, particularly when multiple structural abnormalities are present.

Discussion

This case illustrates the diagnostic value of detailed antenatal ultrasonography in recognizing a combination of abnormalities suggestive of severe multisystem fetal disease. The coexistence of severe polyhydramnios, absent visualization of the stomach, hepatomegaly, a small thoracic cavity, umbilical vein varix, limb deformity, and generalized fetal edema provided important antenatal indicators of a poor fetal prognosis. Postnatal confirmation of esophageal atresia and severe pulmonary hypoplasia further supported the severity of the condition. The importance of antenatal detection is particularly relevant in Iraq, where available studies indicate a considerable burden of congenital anomalies and emphasize the role of ultrasonography in their identification. An Iraqi study reported a prevalence of approximately 14 congenital anomalies per 1,000 births and concluded that ultrasound has an important role in screening, diagnosis, and follow-up, particularly in high-risk pregnancies [3]. Similarly, recent data from Tikrit demonstrated a broad spectrum of congenital abnormalities involving multiple organ systems, highlighting the clinical burden imposed by these conditions [2]. The present case is also relevant because of the previous pregnancy affected by an undiagnosed congenital anomaly. Such a history warrants careful assessment in subsequent pregnancies, although the negative previous chromosomal analysis does not exclude structural abnormalities caused by non-chromosomal, genetic, or multifactorial mechanisms. The high frequency of neural tube defects and incomplete antenatal supervision reported in an Iraqi series further underscores the importance of strengthening antenatal screening and preventive maternal care [4]. Severe polyhydramnios should therefore be regarded as an important indication for detailed fetal evaluation, particularly when accompanied by abnormal fetal biometry or additional sonographic findings. The principal lesson from this case is that systematic anomaly scanning earlier in pregnancy, ideally during the second trimester, may permit earlier recognition of major structural abnormalities, appropriate parental counseling, multidisciplinary planning, and informed pregnancy management in accordance with applicable clinical and legal frameworks.

Conclusion

This case highlights the value of detailed antenatal ultrasonography in detecting severe multisystem fetal anomalies and supporting early prognostic counseling and appropriate perinatal management.



Figure 2A

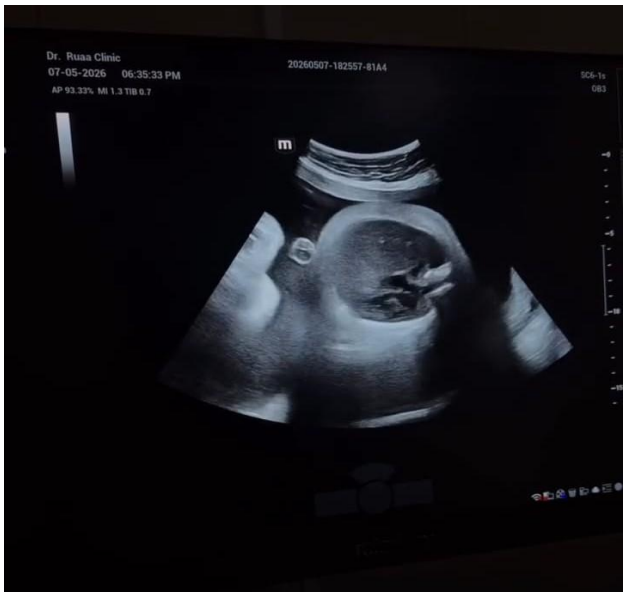


Figure 2B

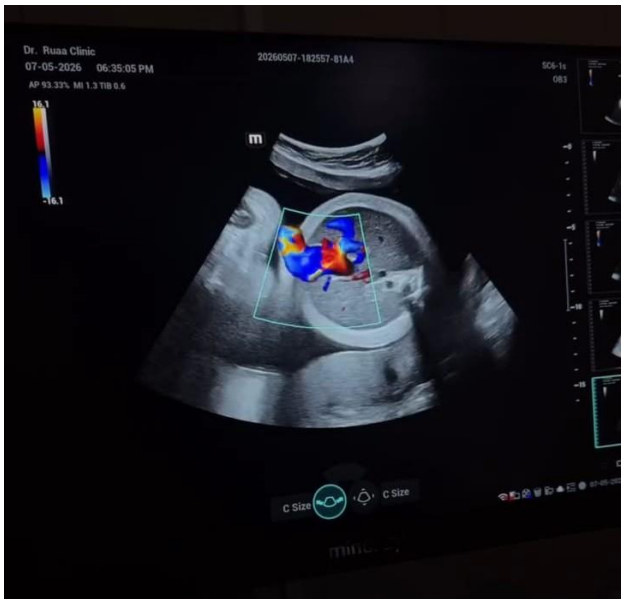


Figure 2C

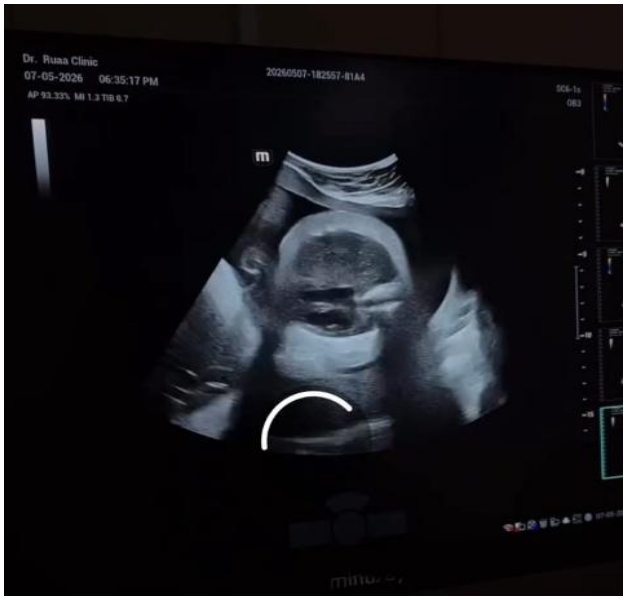


Figure 2D

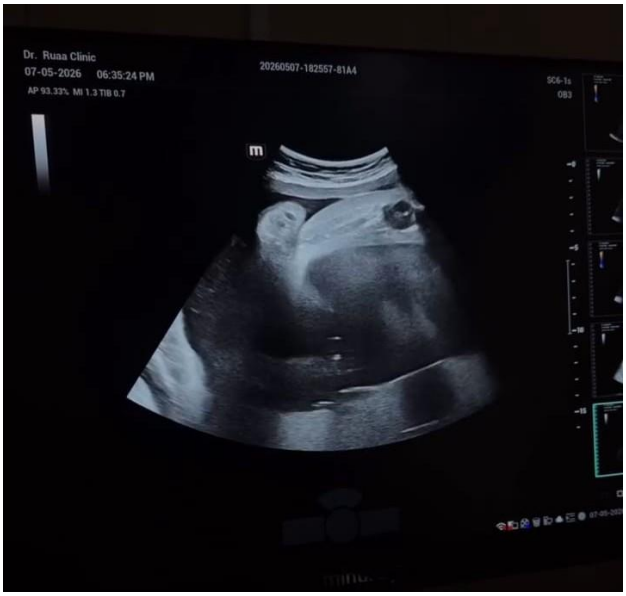


Figure 2E

Abbreviation
None.

Declaration

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Availability of data and materials
Data will be available by emailing dr.ruaa.salman@gmail.com

Authors' contributions
Ruaa Abdulkareem Salman (RAS): Conceptualization, data collection, manuscript drafting, and final manuscript preparation.

Ethics approval and consent to participate
This case report was conducted in accordance with the Declaration of Helsinki. Written informed consent was obtained from the patient for the publication of this report and any accompanying clinical images. The patient also consented to the use of this case to provide new insights within the field of radiology.

Consent for publication
Not applicable

Competing interest
The authors declare that they have no competing interests.

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