https://doi.org/10.3329/cbmj.v15i2.91583

Exploring the Relationship between Fetal Malformations and Perinatal Outcomes: A Case Report of Stillbirth

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Abstract

Stillbirth remains a devastating perinatal outcome. This case report details the fetal and postmortem findings of a stillborn male fetus with severe lower limb reduction defects. It highlights the critical relationship between antenatally detectable anomalies, uteroplacental insufficiency, and fetal demise. A 28-year-old woman with an unremarkable medical history presented for routine prenatal care. First-trimester combined screening was low-risk for common aneuploidies. At 18 weeks, ultrasound revealed a short fetal nasal bone and moderate polyhydramnios; parents declined non-invasive prenatal testing. At 22.5 weeks, detailed ultrasonography demonstrated absent right tibia and fibula, absent left fibula, short and bowed left tibia, bilateral talipes equinovarus, and partial left-hand absence. At 26 weeks, uterine artery Doppler showed notching and an elevated resistance index, indicating uteroplacental insufficiency. Despite bi-weekly biophysical profiles, fetal demise was confirmed at 32 weeks. Examination of the stillborn fetus showed symmetrical limb development, intact cord, and normal external genitalia (male); however, internal autopsy was refused. This case illustrates the complexity of managing pregnancies complicated by fetal limb malformations and concurrent uteroplacental insufficiency. The absence of internal autopsy and genetic testing limited definitive syndromic classification, but does not diminish the importance of reporting the association between structural anomalies and stillbirth.

CBMJ 2026 July: Vol. 15 No. 02 P:404-408

Keywords: Lower limb deformities, uteroplacental insufficiency, stillbirth, perinatal mortality

Introduction

Congenital anomalies affect approximately 2-3% of all pregnancies and represent a leading cause of perinatal mortality worldwide. Among these, limb reduction defects are particularly challenging to detect prenatally, with population-based registry studies reporting detection rates as low as 53.6% even for severe isolated anomalies.[1,2] This diagnostic gap has significant clinical implications, as timely identification of fetal malformations enables informed parental counseling, appropriate peripartum planning, and in select cases, in utero intervention.[1] The presence of associated ultrasound findings often complicates the prenatal evaluation of limb anomalies. Short nasal bone, a well-established soft marker for aneuploidy, has been extensively studied in the second trimester; however, its significance in the context of structural malformations without chromosomal abnormalities remains incompletely understood.[3] When nasal bone hypoplasia coexists with major structural defects, the risk of chromosomal aberrations increases substantially, warranting invasive diagnostic testing.[3] Polyhydramnios represents another important sonographic finding that may accompany fetal anomalies, particularly those affecting the central nervous system or gastrointestinal tract. Idiopathic polyhydramnios, in which no specific cause is identified prenatally, has been associated with adverse perinatal outcomes, including preterm delivery, low Apgar scores, and increased rates of neonatal intensive care unit

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admission.[4] In cases with documented fetal anomalies, polyhydramnios may further compound the risk of poor pregnancy outcomes. Uteroplacental insufficiency, characterized by abnormal uterine artery Doppler parameters such as notching and elevated resistance index, represents a distinct pathologic process that can occur independently or coincidentally with fetal structural anomalies. Mid-gestation uterine artery Doppler assessment has demonstrated predictive value for placental dysfunction-related stillbirth, particularly for events occurring before 37 weeks of gestation.[5] The co-occurrence of fetal malformations and uteroplacental insufficiency presents a complex clinical scenario with limited published guidance. This case report describes a pregnancy complicated by severe lower limb reduction defects, polyhydramnios, and progressive uteroplacental insufficiency culminating in fetal demise at 32 weeks. The objective is to explore the relationship between fetal malformations and perinatal outcomes, highlighting the diagnostic and management challenges inherent in such cases when parents decline definitive testing.

Case Presentation

A 28-year-old woman, gravida 2 para 1, presented for routine prenatal care at a tertiary care center. Her medical and surgical history was unremarkable, with no chronic conditions such as diabetes mellitus or hypertension, and no prior history of preterm labor. Her first pregnancy resulted in a full-term, uncomplicated vaginal delivery of a healthy baby girl who remains in good health. The current pregnancy was initially uneventful. First-trimester combined screening was reported as low-risk for common aneuploidies (trisomies 21, 18, and 13). However, at 18-week of gestation, a routine second-trimester anomaly scan revealed a short fetal nasal bone and moderate polyhydramnios (Fig. 1). The parents were offered non-invasive prenatal testing (NIPT), which they declined. At 22.5-week of gestation, a follow-up ultrasound confirmed the short nasal bone but documented normal cranial and thoracic anatomy with no significant structural defects. However, a detailed fetal ultrasonogram performed at this time revealed severe lower limb reduction defects (Table-I).

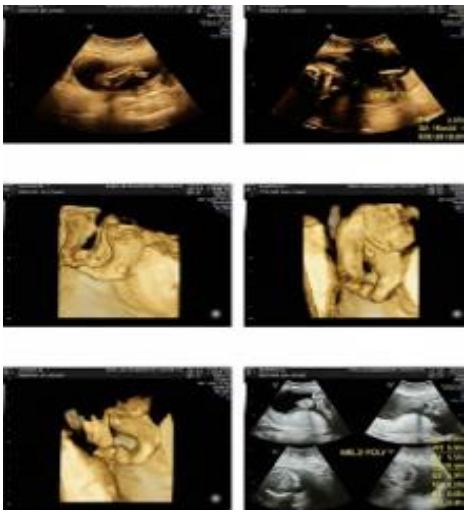

Fig. 1: 2D and 3D ultrasound at 18-week of gestation showing a short nasal bone and mild polyhydramnios – indicating possible fetal anomalies.

Table-I: Detailed ultrasonogram findings at 22½±2 weeks of gestation

Parameters	Findings
Gestational age by BPD	22½ ± 2 weeks
Fetal BPD	53 mm
Fetal FL	34 mm (corresponds to 21½ ± 2 weeks)
Fetal AC	164 mm
Estimated fetal weight	423 g (±10%)
Amniotic fluid index (AFI)	15.4 cm (adequate)
Fetal heart rate	137 bpm (regular)
Presentation	Transverse
Placental position	Posterior, away from the internal os
Right lower limb	Tibia and fibula absent
Left lower limb	Fibula absent; tibia short and bowed
Both feet	Deformed and adducted (bilateral talipes equinovarus)
Left hand	Partially absent
Other structures	Normal cranial and thoracic anatomy

At 26-week of gestation, color and pulsed Doppler ultrasound demonstrated uterine artery notching and an elevated resistance index, findings highly indicative of uteroplacental insufficiency. Normal cranial and thoracic structures were again documented (Fig. 2 & 3).

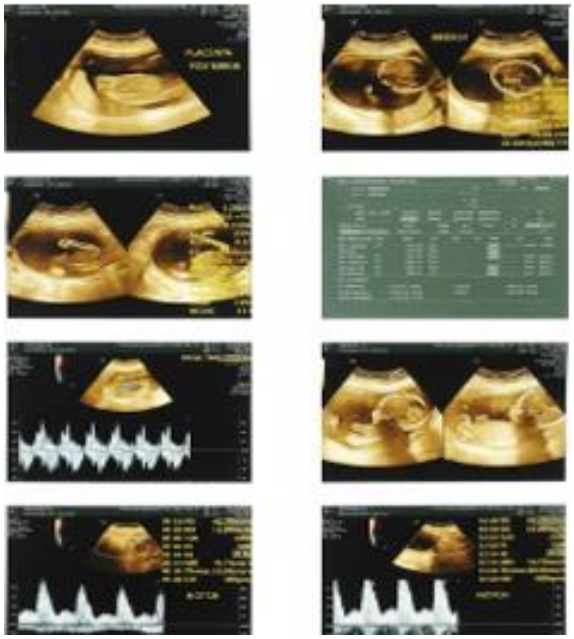

Fig. 2: Colour and pulsed Doppler ultrasound at 26-week of gestation showing uterine artery notching and elevated resistance index – suggestive of uteroplacental insufficiency.

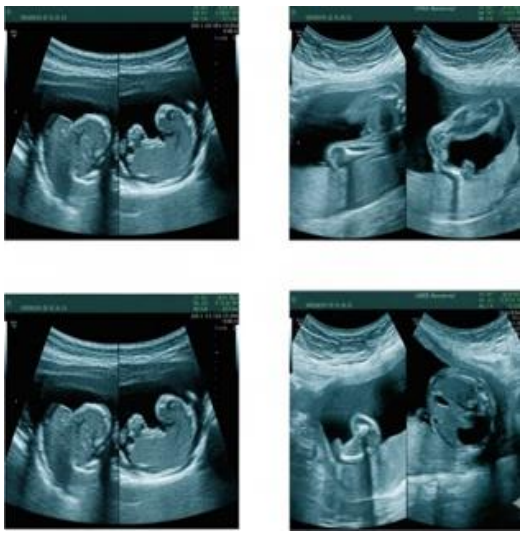

Fig. 3: Ultrasound at 26-week of gestation showing normal cranial and thoracic structures with preserved anatomic outlines.

An intensified surveillance plan was initiated, including bi-weekly biophysical profiles and daily fetal movement counts performed by the mother. The possibility of antenatal corticosteroids for fetal lung maturation was discussed with the parents, as were the risks and benefits of early delivery. Given the gestational age of 26 weeks and the high risk of extreme prematurity, the decision was made to continue close monitoring to prolong gestation. The mother reported normal fetal movements until approximately 31 weeks of gestation. Unfortunately, at 32 weeks, an ultrasound confirmed the absence of cardiac activity, indicating fetal demise. Examination of the stillborn fetus showed symmetrical limb development, normal external genitalia (male), an intact umbilical cord, and mild skin discoloration consistent with postmortem changes. No obvious external malformations were noted on gross inspection. The parents refused an internal autopsy.

Discussion

This case report describes a pregnancy complicated by severe lower limb reduction defects, progressive uteroplacental insufficiency, and eventual fetal demise at 32 weeks of gestation. The findings highlight several important clinical considerations regarding the antenatal detection of structural anomalies, the relationship between fetal malformations and placental dysfunction, and the diagnostic limitations encountered when parents decline confirmatory testing. Lower limb reduction defects (LLRDs) represent a heterogeneous group of congenital anomalies with an estimated prevalence of approximately 5.2 per 10,000 live births.[6] The spectrum ranges from isolated fibular hemimelia to complex limb-body wall associations. In the present case, the combination of absent tibia and fibula on the right, absent fibula with a short and bowed tibia on the left, bilateral talipes equinovarus, and partial

left-hand absence represents a severe phenotype extending beyond isolated limb involvement. This pattern of deficiencies has been described in the literature under the acronym FATCO (femur, fibula, tibia deficiency with complex anomalies of the lower limbs), a rare sporadic condition first delineated in the 1990s.[7] FATCO syndrome typically presents with variable lower limb deficiencies that may be unilateral or bilateral, often accompanied by upper limb anomalies such as hand deformities, as observed in this case.[7] However, the diagnosis remains one of exclusion, as no specific genetic or molecular marker exists.[8] The antenatal sonographic detection of such limb anomalies presents considerable challenges. A large registry-based study reported that only 53.6% of severe isolated limb reduction defects were identified prenatally, with even lower detection rates for non-isolated cases.[1] The present case was detected at 22.5 weeks, which falls within the typical window for detailed anomaly scanning. Several factors may have contributed to earlier detection, including the presence of associated soft markers (short nasal bone) and polyhydramnios, both of which prompted closer sonographic scrutiny.[3,4] The coexistence of uteroplacental insufficiency with fetal structural anomalies raises important questions about potential mechanistic links or coincidental occurrence. Abnormal uterine artery Doppler parameters in the mid-trimester are established predictors of placental dysfunction-related stillbirth, particularly for events occurring before 37 weeks.[5] In this case, the emergence of uterine artery notching and an elevated resistance index at 26 weeks preceded fetal demise by six weeks, suggesting a progressive ischemic placental process. Whether the limb reduction defects and placental dysfunction share a common etiology remains speculative. Animal models have suggested that vascular disruption events during early embryogenesis can produce both limb deficiencies and placental abnormalities,[8] human data is limited though. Polyhydramnios observed at 18 weeks in this case warrants specific comment. While frequently associated with fetal anomalies affecting the central nervous system or gastrointestinal tract, polyhydramnios may also occur in the setting of limb reduction defects due to impaired fetal swallowing mechanics or reduced fetal movement.[4] The moderate degree of polyhydramnios documented here did not progress to severe levels, but its presence likely contributed to the decision to perform serial detailed sonographic examinations. Several limitations must be acknowledged. First, the parents declined both non-invasive prenatal testing and internal fetal autopsy, precluding any genetic or chromosomal analysis. Consequently, aneuploidy (including trisomy 18, which can present with limb anomalies and growth restriction) cannot be definitively excluded despite the low-risk first-trimester screening result. Second, radiographic imaging of the stillborn fetus was not performed, limiting the precision of skeletal anomaly characterization. Third, placental histopathological examination was not available, which might have identified specific lesions such as maternal vascular malperfusion or fetal thrombotic vasculopathy to explain the Doppler findings. Fourth, the absence of an internal autopsy means that visceral anomalies potentially associated with FATCO syndrome or other conditions could not be assessed. Despite these limitations, this case contributes to the limited literature on the co-occurrence of severe limb reduction defects and uteroplacental insufficiency. Clinicians managing similar pregnancies should maintain a high index of suspicion for placental dysfunction when fetal anomalies are identified, as serial Doppler assessment may provide prognostic

information.[5] When parents decline definitive diagnostic procedures, meticulous sonographic documentation and clear communication regarding limitations become paramount.

Conclusion

This case illustrates the complex relationship between fetal lower limb reduction defects, polyhydramnios, and progressive uteroplacental insufficiency culminating in stillbirth. When parents decline confirmatory genetic testing and internal autopsy, definitive syndromic classification remains unattainable. Nonetheless, serial Doppler surveillance and transparent counseling about prognostic uncertainty remain essential in managing such high-risk pregnancies.

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