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Case Report
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Unilateral Absence of Fetal Foot: A Case Report of Rare Congenital Anomaly

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Abstract

Objectives: Fetal ultrasonography is the key tool for offering prenatal detection of fetal anomalies. The objective of this case report was to explore a unique case of congenital unilateral absent fetal foot.

Methods: We report a rare case of unilateral absent fetal foot by early pregnancy ultrasound in IVF patient, previous 2 miscarriages then 4-year-old child, come with lower abdominal pain and vaginal spotting, with no other past medical or surgical history, no allergies.

Conclusion: An early-stage ultrasound scan can detect congenital limb reduction defects. It is important to give enough time for an ultrasound scan to clearly visualize all fetal parts.

Introduction

limb abnormalities occur in six out of 10,000 live births, with a higher incidence observed in the upper limbs compared to the lower limbs [1]. Limb abnormalities are more commonly unilateral than bilateral, and they are more frequently found on the right side than on the left [2]. The origin of limb abnormalities is notably complex, involving factors like single gene disorders [3,4], maternal diseases [5], intrauterine factors [6], vascular events [7,8], and chromosomal abnormalities [9]. Yet, in many instances, the specific cause remains unknown.

Detecting limb abnormalities can become a complex challenge without the implementation of an accurate diagnostic approach. The prenatal diagnosis and management of limb abnormalities require a multidisciplinary team, including radiologists, obstetricians, neonatologist, clinical geneticists, and orthopedic surgeons. This collaboration aims to provide parents with information about the disorder's origin, prognosis, pregnancy-related choices, and the likelihood of recurrence in future pregnancies [10,11].

Case Presentation

A 33 years old gravida 4 para 1, previous 2 miscarriages (IVF pregnancies), followed by term pregnancy with a healthy child (currently 4-year-old). She came with lower abdominal pain and vaginal spotting, with no other past medical or surgical history, no allergies.

By ultrasound, viable early pregnancy 12+ weeks, NT 2.2 mm (normal regarding CRL measurement), posterior placenta developing, left fetal foot was not seen, then patient was sent to Fetal Maternal Unit for further evaluation (Figures 1-3).

After a detailed counseling of the condition including genetic counseling, invasive testing was done (amniocentesis), eventually she underwent medical termination of pregnancy at 18 weeks. Follow up genetic testing showed Normal Karyotype and Microarray; however Whole Exome Sequencing (WES) showed Mosaic Denovo Likely Pathogenic variant in PORCN gene consistent with the diagnosis of Goltz Syndrome in the terminated fetus.

Discussion

Congenital limb reduction defects arise when a section or the complete upper or lower limbs fail to form normally during pregnancy [12]. By the eighth week, the upper and lower limbs have fully developed. Limb defects may coexist with other abnormalities, such as other limb anomalies, heart defects, and disorders affecting the gastrointestinal and nervous systems [13]. Any factors

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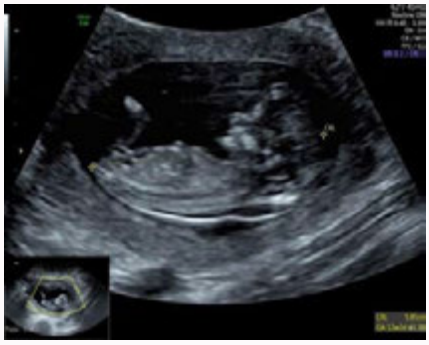

Figure 1: Fetal CRL showing normal nasal bone.

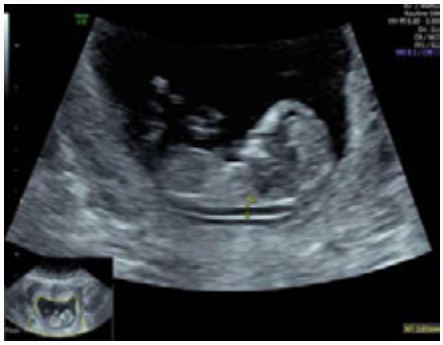

Figure 2: NT which is normal according to fetal gestational age (CRL).

influencing the development and differentiation of limbs during this period can lead to various limb abnormalities. Some of these factors include environmental exposure, chromosomal abnormalities, genetic disorders, and prenatal diagnostic procedures, can contribute to limb abnormalities [14].

Despite its rarity, the absence of a limb is highly noticeable and can be devastating for parents. It has the potential to significantly impact the daily functioning and quality of life of the individuals affected [15].

The initial opportunity to detect fetal limb anomalies occurs in the first trimester of pregnancy, with the optimal time within the first trimester is after 12 weeks of gestation.

When examining the fetus, it is essential to assess all three planes-coronal, longitudinal, and axial. Additionally, it is important to examine both left and right limbs, as the anomaly may affect only one limb. Ultrasound examinations are generally performed abdominally, and transvaginal assessments are utilized when deemed necessary. Evidence suggests that optimal results are achieved through a combination of both methods [16,17]. The best time to detect limb reduction defects is during the second trimester of pregnancy using an anomaly ultrasound scan. This is because, during this period, the uterus is fully elevated out of the pelvis, and the fetus has developed sufficiently and is likely in an optimal position for examination.

In the latter part of the second trimester and during the third trimester, the growing size of the fetus makes it increasingly challenging to conduct detailed morphological examinations [18].

Sonologists are advised to allocate a satisfactory amount of time for fetal ultrasound examinations. Andrikopoulou et al. demonstrated that an increase of less than five minutes in the duration of anomaly

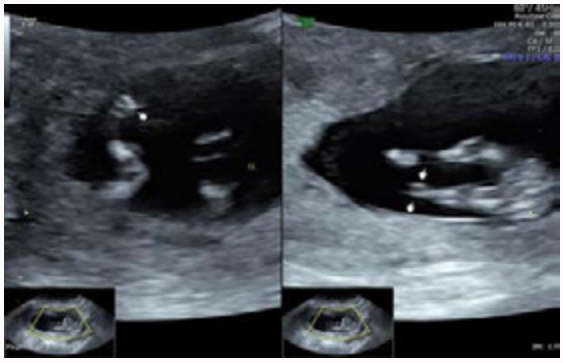

Figure 3: Unilateral absence of fetal foot.

ultrasound scans could more than double the detection rate of limb malformations [19]. The primary advantage of early prenatal diagnosis of limb defects is to offer parents the chance to undergo prenatal counseling [20]. This enables parents to discuss their child's abnormalities with various specialists. In cases of treatable malformations, it might be essential to gather a team of specialists for postnatal care. Conversely, families may consider pregnancy termination for untreatable malformations. Numerous studies have demonstrated an increased rate of pregnancy termination following the early diagnosis of major untreatable malformations [21,22].

Conclusion

Unilateral absent limb represents a rare congenital fetal anomaly, potentially posing significant challenges for parents due to its adverse effects on daily functioning and quality of life. The optimal time for detection during the first trimester is recommended after 12 weeks of gestation. A comprehensive fetal examination should encompass all three planes: Coronal, longitudinal, and axial. Examination of both left and right limbs is crucial, considering that only one limb may be affected. Abdominal ultrasound is typically employed, with transvaginal assessments used when necessary. Studies suggest that a slight extension of ultrasound time can improve the diagnostic rate. Early prenatal diagnosis not only allows for counseling but also provides an opportunity for decision-making.

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