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Prenatal Detection of Isolated Fetal Amelia: A Case Report and Review of Literature

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Abstract

Background: Amelia, the complete absence of a limb, is a rare congenital anomaly with a complex and multifactorial etiology, including genetic, vascular, and environmental factors. Early diagnosis is crucial for optimal management and parental counseling. **Case:** This is a 27-year-old primigravida whose fetus was diagnosed antenatally with isolated right lower limb amelia at 29 + 5 weeks of gestation via ultrasound. The pregnancy was uneventful, with no identifiable teratogenic exposures or family history of congenital anomalies. The baby was delivered vaginally at term and postnatal examination confirmed complete absence of right lower limb along with a rudimentary thumb on the same side and bilateral undescended testes. Genetic testing was normal. **Conclusion:** This case highlights the importance of fetal anomaly scans in identifying rare congenital limb defects. Multidisciplinary care and early counseling play a key role in optimizing outcomes for both the infant and family.

Keywords

Amelia, Limb Deformities, Congenital, Prenatal Diagnosis, Ultrasonography, Prenatal, Congenital Abnormalities, Limb Buds, Pregnancy

1. Introduction

Amelia is a rare congenital anomaly characterized by the complete absence of one or more limbs. Clinically, amelia does not always imply complete absence of the limb, as patients may present with a distal limb bud lacking osseous structures [1]. The estimated incidence is between 0.05 and 0.095 per 10,000 live births, making it an uncommon but clinically significant diagnosis in prenatal care [2]. It results from disruptions in early limb development, typically occurring between days 24 to 36 post-fertilization, a critical period for limb bud formation [3].

The etiology of amelia is multifactorial. It may stem from genetic mutations in the WNT3 gene, where the gene itself undergoes homozygous or compound heterozygous point mutations known to cause tetra-amelia syndrome, which involves the absence of all four limbs, craniofacial, urogenital and cardiac anomalies [4]. Other causes include vascular disruptions—particularly from amniotic band syndrome (ABS), and teratogenic exposures, with thalidomide being the most well-known example [5] [6].

Advances in prenatal imaging, especially high-resolution and 3D ultrasound, have improved detection rates for fetal anomalies. Identifying limb defects in the second trimester with an anomaly scan necessitates comprehensive assessment for associated anomalies, family counseling, and delivery planning [7].

2. Case Presentation

A 27-year-old primigravida presented to the antenatal clinic at King Hamad University Hospital (KHUH) Bahrain at 29 weeks and 5 days of gestation with a previous private ultrasound report indicating isolated right lower limb amelia.

A follow-up scan confirmed the complete absence of the right lower limb (**Figure 1**). There were no other visible anomalies at the time. The patient had an unremarkable medical history, denied alcohol or tobacco use, and reported no known exposure to teratogens. There was no history of consanguinity or genetic disorders in the family.

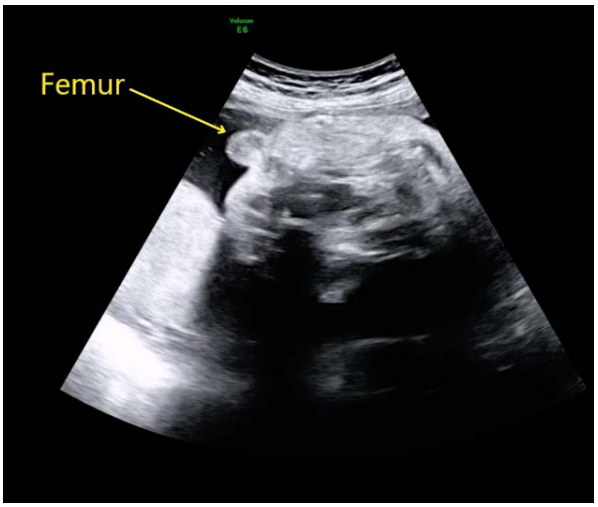

Figure 1. Absence of the femur of the right lower limb on ultrasound.

A multidisciplinary perinatal team, including obstetricians and neonatologists, reviewed the case. After counseling the patient and her family, spontaneous labor and vaginal delivery. Routine antenatal labs were normal, and the pregnancy was monitored closely.

At 38 weeks and 5 days of gestation, the patient presented with spontaneous rupture of fetal membranes, a high vaginal swab was taken and PCR examination for Group B Streptococcus infection was negative. She received epidural analgesia

and had a normal spontaneous vaginal delivery of a live male baby weighing 2.7 kg, with APGAR scores of 9 and 10 at 5 and 10 minutes, respectively.

Neonatal examination confirmed the absence of the right lower limb, as well as the presence of a rudimentary thumb on the same side (Figure 2 & Figure 3). Neonatal ultrasound confirmed bilateral undescended testes otherwise no other associated internal anomalies. Genetic testing, including chromosomal microarray, returned normal results. The neonate was referred to pediatric urology and orthopedics for follow-up care. At follow-up visits, the infant was thriving and showed age-appropriate developmental progress.

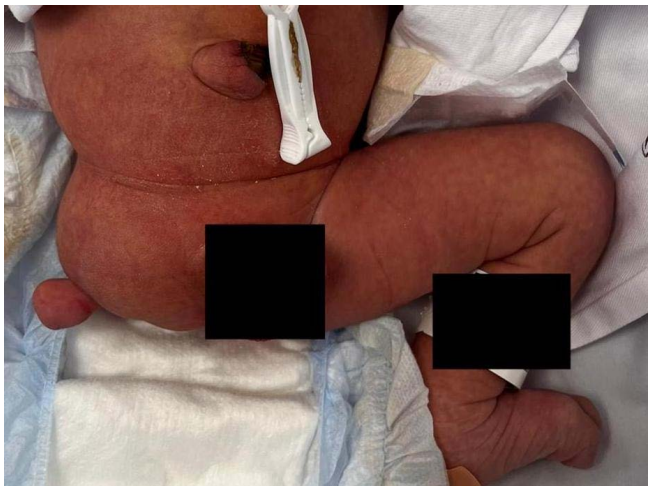

Figure 2. Showing the right lower limb amelia.

Figure 3. Showing the presence of rudimentary thumb on the ipsilateral side.

3. Discussion

This is a case of isolated amelia involving the right lower limb, accompanied by a rudimentary ipsilateral thumb and undescended testes. Isolated amelia without a syndromic or genetic basis is rare, particularly in the absence of teratogenic exposure or family history.

Limb development occurs between the 4th and 5th weeks of gestation, guided by interactions between the apical ectodermal ridge (AER) and the underlying mesenchymal core, which together regulate proximodistal outgrowth and limb patterning through FGF, WNT, and SHH signaling pathways [2] [6]. Disruptions in this critical window, whether from transient vascular insufficiency or localized signaling failures, can result in partial or complete limb agenesis.

In this case, the absence of the femur indicates an interruption in proximal limb development, while the rudimentary thumb reflects a disruption affecting the distal preaxial structures of the upper limb bud. Both align with a timing of insult during the 5th week of embryogenesis, when limb segment differentiation occurs in a proximodistal sequence.

The finding of bilateral undescended testes may also be related, as testicular descent involves hormonal and mechanical processes occurring later in gestation but requires normal development of the gubernaculum and abdominal wall structures, which begin forming during the 7th - 12th weeks of gestation. Subtle developmental disruptions affecting mesenchymal tissue or intra-abdominal pressure gradients can contribute to cryptorchidism, especially in association with limb anomalies.

The combination of limb deficiencies and undescended testes in the absence of genetic abnormalities or teratogenic exposures supports the possibility of a sporadic vascular or developmental disruption during early embryogenesis, which may have variably affected limb and urogenital development. This highlights the intricate timing and interdependence of embryologic processes and underscores the importance of early detection, multidisciplinary evaluation, and family-centered counseling in managing such rare presentations.

Another widely considered cause is the vascular disruption theory, which suggests that a temporary interruption in blood supply during early embryogenesis can lead to tissue necrosis and subsequent resorption of developing limb structures, resulting in defects such as transverse limb reductions [8]. Another cause is identified as amniotic band syndrome (ABS), where fibrous strands from a ruptured amnion can constrict or entangle fetal limbs. While ABS typically causes asymmetrical or irregular limb abnormalities, in more severe cases it can lead to complete limb loss [9]. However, in this case, there was no sonographic evidence of amniotic bands, making ABS an unlikely cause.

Several genetic conditions are known to result in limb defects. Tetra-amelia syndrome, caused by mutations in the WNT3 gene, is a genetic disorder characterized by the absence of all four limbs and often associated craniofacial and urogenital anomalies [3]. However, this syndrome is unlikely due to the absence of such features, normal karyotype, and unremarkable genetic testing.

Environmental teratogens are well-documented causes of congenital limb anomalies. Thalidomide, which inhibits angiogenesis, infamously led to thousands of limb defect cases in the 1950s-60s [4]. Other teratogens include alcohol, retinoic acid, and some anticonvulsants, all of which have been implicated in limb abnor-

malities [10] [11]. The patient denied exposure to any such agents.

This case highlights a patient diagnosed with fetal anomaly at 29 + 5 weeks gestation via ultrasound, typically anomaly scans are done at 20 weeks of gestation which will give both the patient and doctors time to evaluate and understand conditions that may present early on in pregnancies. Factors such as late clinic booking or patient noncompliance may lead to late diagnosis of such conditions, hence why this case highlights the importance of early diagnosis.

Prenatal ultrasound detection of the abnormality gave appropriate time for family counseling, delivery planning, and multidisciplinary coordination. Postnatal care focused on addressing the associated anomalies—namely the rudimentary thumb and undescended testes—while providing support for the family. Involvement of the orthopedic surgeon and pediatric urologist aided in the treatment of the rudimentary thumb and undescended testes.

Early fitting of upper-limb prostheses in children has long been recognized as important, but there is ongoing debate about when exactly "early" should be [12]. Sypniewski (1972) reviewed the literature and highlighted several reasons for early fitting, emphasizing its role in promoting functional use and integration of the prosthesis into daily activities [12]. However, clinicians continue to differ on which developmental milestones should guide this timing.

Prosthesis in early childhood is said to facilitate motor development, improve symmetry in posture and gait, and support psychosocial adaptation [13] [14]. Research indicates that early prosthetic fitting, ideally around 6 - 12 months of age, coinciding with the period when children begin to sit and develop bimanual activities, may enhance prosthesis acceptance and use [15] [16]. Delayed fitting may lead to prosthesis rejection due to the child's adaptation to functioning without the limb [17] [18].

Fisher (1976) explored this debate further, discussing how the development of visually guided reaching may depend on the opportunity to see the limb moving in space [13]. She questioned whether fitting a prosthesis before 3 to 4 months of age would truly support the development of visually guided reaching and positively affect future prosthetic use patterns [13]. This highlights the need for individualized timing, balancing the potential benefits of early fitting with each child's readiness and family context.

4. Conclusions

Amelia is a rare congenital anomaly that may occur as part of a syndrome or in isolation. This case describes a fetus with right lower limb amelia, detected via routine antenatal ultrasound, confirmed at birth, and managed successfully through a multidisciplinary approach. In the present case, the absence of teratogenic exposures, normal genetic testing, and lack of family history makes a sporadic event more likely. The presence of a rudimentary thumb suggests that initial limb bud formation may have occurred, followed by a disruption halting further development. Such sporadic disruptions are, unfortunately, challenging to predict or pre-

vent and emphasize the complexity of early fetal development.

Early detection of congenital anomalies plays a critical role in perinatal planning. Ultrasound remains a key tool in identifying such conditions, prompting comprehensive evaluation for associated defects. Effective communication and family-centered counseling are essential for preparing families for potential outcomes.

Conflicts of Interest

The authors declare no conflicts of interest regarding the publication of this paper.

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