A Rare Case of Severe Asymmetrical Limb Reduction Defects Diagnosed During Second-Trimester Scan

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Abstract:

Limb reduction defects (LRDs) are rare congenital malformations characterized by partial or complete absence of one or more limbs due to disruption of normal embryonic limb development during early gestation. With an estimated prevalence of 5–9 per 10,000 live births, these anomalies exhibit marked phenotypic and etiological heterogeneity, including chromosomal abnormalities, single-gene disorders, vascular disruption, amniotic band sequence, teratogenic exposure, maternal metabolic disorders, and sporadic developmental defects. Early prenatal diagnosis is essential for accurate phenotypic characterization, prognostic evaluation, parental counselling, and pregnancy management. We present an exceptionally rare case of severe asymmetrical limb reduction involving three limbs diagnosed during a routine second-trimester anomaly scan.

A 32-year-old woman Gravida 3 with two previous first-trimester spontaneous abortions underwent a detailed fetal scan at 16 weeks' gestation. The pregnancy was conceived spontaneously, and maternal history was significant for poorly controlled pregestational diabetes mellitus managed with insulin therapy. There was no history of teratogenic drug exposure, maternal infections, radiation exposure, alcohol or tobacco use, consanguinity, or family history of congenital anomalies. High-resolution ultrasonography demonstrated complete bilateral lower limb agenesis, complete absence of the right upper limb, and a hypoplastic left forearm with severe hand malformation. An isolated echogenic intracardiac focus was identified without additional major structural abnormalities. Following multidisciplinary fetal medicine consultation and comprehensive parental counselling, the pregnancy was medically terminated. Postnatal examination confirmed all prenatal imaging findings.

Conventional fetal karyotyping demonstrated a normal chromosomal complement, while whole-exome sequencing failed to identify any pathogenic or likely pathogenic variants explaining the phenotype. Incidental heterozygous carrier variants in HBB and SLC12A1 were considered unrelated to the congenital anomalies. The absence of a molecular diagnosis suggests the possible contribution of non-genetic mechanisms, including early vascular disruption, embryonic developmental arrest, amniotic band sequence, or diabetic embryopathy. This case highlights the pivotal role of meticulous

second-trimester ultrasonography combined with advanced genomic evaluation in the diagnosis of complex fetal limb anomalies. It further emphasizes the importance of multidisciplinary counselling, informed reproductive decision-making, and individualized recurrence-risk assessment. Recognition of genetically unexplained cases remains essential for improving our understanding of fetal limb morphogenesis and advancing prenatal diagnostic strategies.

Keywords:
Bilateral lower limb agenesis, congenital limb anomalies, diabetic embryopathy, limb reduction defects, prenatal diagnosis, whole-exome sequencing.