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Multiple Severe Fetal Anomalies with Suspected Trisomy 18 Versus Pena–Shokeir Syndrome: A Case Report

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Abstract

Congenital anomalies affect approximately 3–5% of all pregnancies worldwide and remain a leading cause of perinatal morbidity and mortality. The risk increases significantly when multiple structural abnormalities coexist, often suggesting an underlying chromosomal or genetic syndrome [1].

Trisomy 18 (Edwards syndrome) is the second most common autosomal trisomy after Trisomy 21 and is associated with severe multisystem malformations. Prenatal ultrasound findings typically include clenched hands with overlapping fingers, rocker-bottom feet, omphalocele, congenital heart disease, growth restriction, and central nervous system anomalies [6,8].

Fetal akinesia deformation sequence (FADS), also known as Pena–Shokeir syndrome, represents a heterogeneous group of disorders characterized by reduced or absent fetal movements. The lack of fetal motion leads to secondary deformities such as arthrogryposis, limb contractures, facial anomalies, pulmonary hypoplasia, and polyhydramnios [1,4]. Sonographically, FADS may closely mimic chromosomal aneuploidies, making prenatal differentiation challenging [4].

The presence of neural tube defects, such as encephalocele, and abdominal wall defects, such as omphalocele, further complicates diagnosis and significantly worsens prognosis [2,3,9].

1. Case Presentation

An occipital encephalocele was identified, characterized by a posterior skull defect with herniation of brain tissue, a finding commonly associated with syndromic and chromosomal disorders [2]. Additionally, a large omphalocele containing herniated liver was visualized, a feature known to carry poor prognosis when associated with chromosomal abnormalities [3,5,9].

Bilateral claw-like hand deformities with fixed flexion contractures were noted, suggestive of arthrogryposis and reduced fetal movement, which are hallmark features of fetal akinesia deformation sequence [1,4]. Lower limb evaluation revealed rocker-bottom feet, a classic prenatal marker of Trisomy 18 [6,8]. Polyhydramnios was also present, likely secondary to impaired fetal swallowing due to neurological or neuromuscular dysfunction, as described in both Trisomy 18 and FADS [7]. Given the strong association between Trisomy 18 and congenital heart disease, fetal echocardiography was recommended [6,10].

2. Discussion

This case illustrates the diagnostic complexity encountered when multiple severe fetal anomalies are detected antenatally. Trisomy 18 is classically associated with clenched hands, rocker-bottom feet, omphalocele, and central nervous system anomalies, all of which were present in this case [6,8]. Survival beyond the neonatal period is rare, with high rates of intrauterine and early neonatal mortality [6].

Pena–Shokeir syndrome results from reduced fetal movement due to neuromuscular or central

nervous system dysfunction. The resulting fetal akinesia leads to arthrogryposis, limb contractures, polyhydramnios, and pulmonary hypoplasia [1,4]. Phenotypic overlap with chromosomal aneuploidies necessitates comprehensive genetic evaluation [1].

The coexistence of encephalocele and large omphalocele containing liver strongly suggests a syndromic etiology and is associated with a particularly poor prognosis [2,3,9].

Polyhydramnios further increases obstetric risk, including preterm labor and malpresentation [7].

Definitive prenatal diagnosis relies on genetic testing, beginning with conventional karyotyping and chromosomal microarray analysis. When results are normal and clinical suspicion persists, advanced molecular testing such as targeted neuromuscular gene panels or whole-exome sequencing may be required [1].

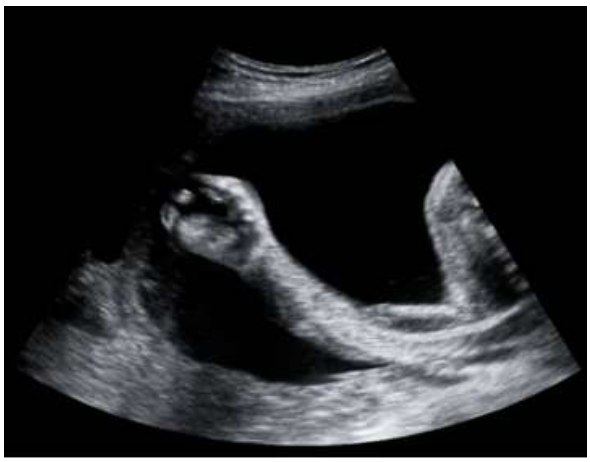

Figure 1. *Bilateral claw-like hand deformities suggestive of arthrogryposis.*

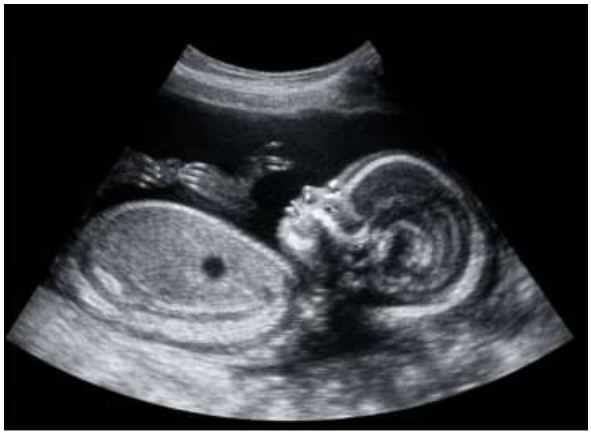

Figure 2. *Occipital encephalocele with associated omphalocele and liver herniation.*

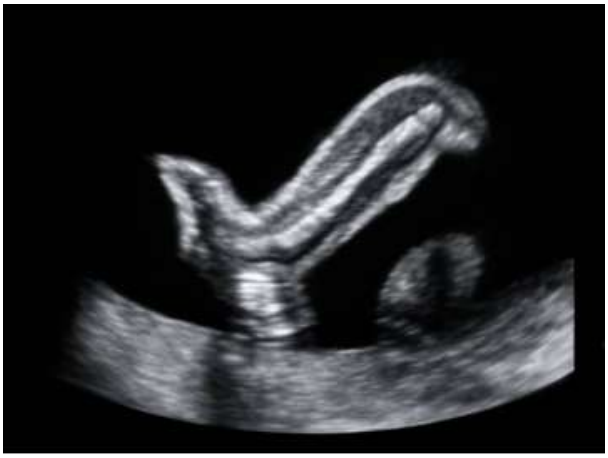

Figure 3. *Rocker-bottom feet deformity.*

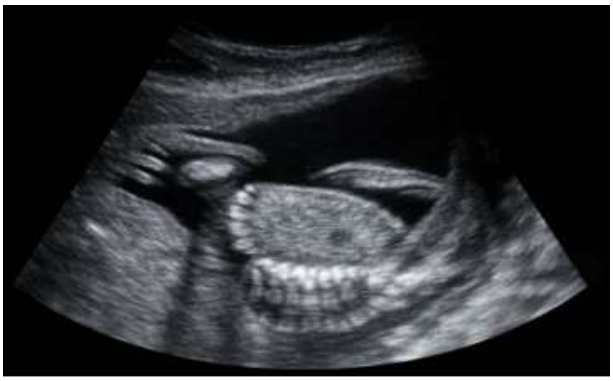

Figure 4. *Severe limb contractures consistent with fetal akinesia deformation sequence.*

3. Management and Counseling

The patient received comprehensive multidisciplinary counseling involving obstetrics, fetal medicine, neonatology, pediatric surgery, and clinical genetics. Counseling focused on explaining the suspected diagnoses, the severe and likely lethal prognosis, and available diagnostic and management options.

The role of amniocentesis for chromosomal analysis was discussed in detail, including potential benefits, risks, and limitations. Options for pregnancy continuation versus termination were explored in accordance with local legal and ethical frameworks. The importance of perinatal planning, including neonatal palliative care, was emphasized.

A plan was established for serial ultrasound monitoring, fetal echocardiography, and coordinated care with neonatal and surgical teams should the pregnancy continue.

4. Discussion

This case illustrates the diagnostic complexity encountered when multiple severe fetal anomalies are detected antenatally. Trisomy 18 is classically associated with clenched hands, rocker-bottom feet, omphalocele, and central nervous system anomalies, all of which were present in this case. Survival beyond the neonatal period is rare, and most affected fetuses experience intrauterine or early neonatal demise.

Pena–Shokeir syndrome results from reduced fetal movement due to neuromuscular or central nervous system dysfunction. The resulting fetal akinesia leads to arthrogryposis, limb contractures, polyhydramnios, and pulmonary hypoplasia. Phenotypic overlap with chromosomal aneuploidies necessitates comprehensive genetic evaluation.

The presence of encephalocele and large omphalocele containing liver further worsens prognosis and strongly suggests a syndromic etiology. Polyhydramnios in this context reflects impaired swallowing and is associated with increased obstetric complications, including preterm labor and malpresentation. Definitive prenatal diagnosis relies on genetic testing, beginning with conventional karyotyping and chromosomal microarray analysis. When results are normal and clinical suspicion persists, advanced molecular testing such as targeted neuromuscular gene panels or whole-exome sequencing may be required.

5. Conclusion

The antenatal detection of multiple severe fetal anomalies requires a structured diagnostic approach and early multidisciplinary involvement. Distinguishing between chromosomal aneuploidies such as Trisomy 18 and neuromuscular conditions like fetal akinesia deformation sequence is essential for accurate prognosis, parental counseling, and perinatal planning.

6. Ethics Approval and Consent to Participate

Ethical approval was obtained in accordance with institutional guidelines.

7. Consent for Publication

Written informed consent was obtained from the patient for publication of this case report and accompanying ultrasound images.

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