



Apert Syndrome: A Rare Case Detected Following Polyhydramnios

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INTRODUCTION/BACKGROUND

Apert syndrome, a rare autosomal dominant disorder, poses challenges for prenatal diagnosis. Early diagnosis allows for informed decision-making and appropriate management, including genetic counseling and potential termination of pregnancy if desired by the parents.

OBJECTIVES/AIMS

To emphasize the importance of detailed morphological assessment using three-dimensional (3D) ultrasound after the detection of polyhydramnios.

METHODS

We present a case of suspected Apert syndrome diagnosed at 31 weeks of gestation. The diagnosis was facilitated by 3D ultrasound imaging, which was instrumental in guiding parental counseling following the discovery of severe polyhydramnios.

CONCLUSIONS

In cases of severe polyhydramnios or abnormal ultrasound findings in the second trimester, it is imperative to perform a comprehensive morphological assessment to investigate possible underlying genetic syndromes, such as Apert syndrome. Key features suggestive of Apert syndrome include midface hypoplasia, a prominent forehead, low-set ears, hypertelorism, bilateral syndactyly (mitten hands), and foot deformities. When these findings are identified, genetic testing should be promptly pursued to confirm the diagnosis and guide prenatal counseling.

RESULTS

We report the case of a 37-year-old, Gravida 1, Para1, with a history of primary infertility for 7 years, who achieved spontaneous pregnancy. The second-trimester triple test showed a low risk for Trisomy 21, 18, 13, and neural tube defects. The second-trimester ultrasound was missed. At 31 weeks of gestation, a morphological study was performed following the discovery of severe polyhydramnios. It revealed the presence of midface hypoplasia with a prominent forehead (figure1), low-set ears (figure 2), hypertelorism , bilateral syndactyly of both hands (mitten hands appearance) (figure 3), and deformed feet (figure 4).

Genetic testing on an amniotic fluid sample concluded the presence of a heterozygous mutation in the FGFR2 gene (Fibroblast Growth Factor Receptor 2) specific to Apert syndrome. After informed consent, the parents decided to terminate the pregnancy. The morphological appearance of the fetus was characteristic of Apert syndrome and very similar to 3D ultrasound images (Figure 5).

Figure 1
midface hypoplasia with a prominent forehead in 3D (A) and 2D (B)

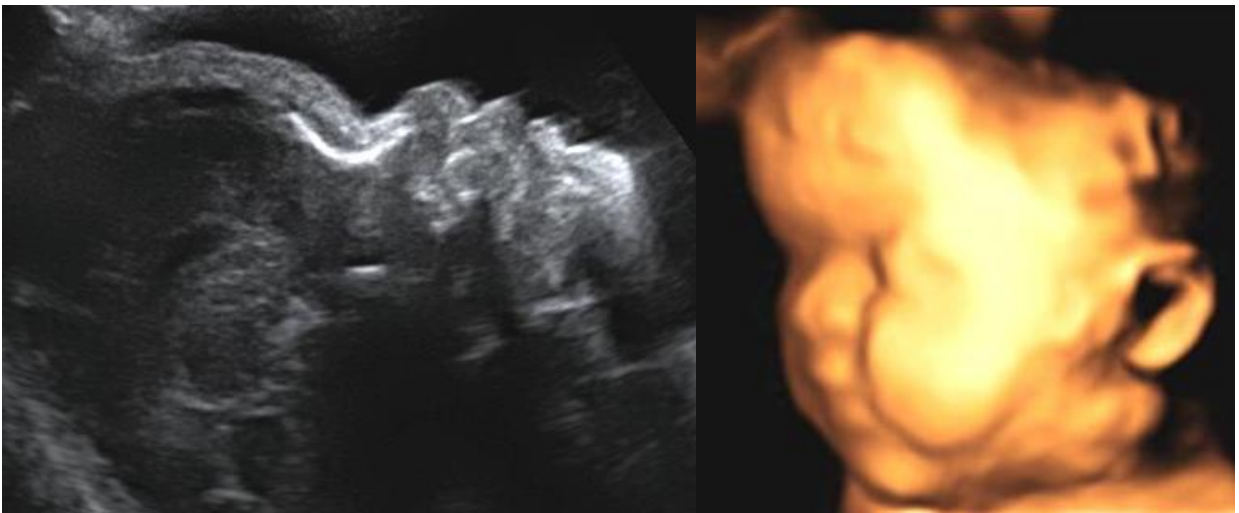


Figure 2
low-set ears



Figure 3
bilateral syndactyly of both hands (mitten hands appearance)



Figure 4
deformed feet



Figure 3
Comparison between 3D ultrasound findings and macroscopic fetal examination showing the same facial dysmorphism (A) and limb abnormalities (B,C).

REFERENCES

El-Bassyouni HT, El-Kamah GY, Afifi HH, Taher MB, Soliman DR, Hamed K, et al. Insight into Apert Syndrome: Reporting on Six Patients and Increasing Awareness. Mol Neurobiol 2025. <https://doi.org/10.1007/s12035-025-04902-9>.
Wu F-T, Chen C-P. Too Much of a Good Thing: Updated Current Management and Perinatal Outcomes of Polyhydramnios. Journal of Medical Ultrasound 2024;32:285. https://doi.org/10.4103/jmu.jmu_83_24.
Varlas VN, Epistatu D, Varlas RG. Emphasis on Early Prenatal Diagnosis and Perinatal Outcomes Analysis of Apert Syndrome. Diagnostics 2024;14:1480. <https://doi.org/10.3390/diagnostics14141480>.