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Face Predicts the Brain: Prenatal Ultrasound Diagnosis of Alobar Holoprosencephaly with Multisystem Anomalies - A Rare Case Report

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Abstract: ***Holoprosencephaly is a rare congenital malformation resulting from incomplete cleavage of the forebrain and is frequently associated with severe craniofacial and systemic anomalies[1–3]. We present a case of a 16-week intrauterine fetus diagnosed on antenatal ultrasonography with alobar holoprosencephaly and multiple associated anomalies. Ultrasound revealed a monoventricle with fused thalami, absent midline structures, and severe facial dysmorphism including absent orbits and a midline proboscis. Associated findings included a cystic lesion suggestive of a foregut duplication cyst, complex congenital heart disease, and bilateral echogenic kidneys. These findings indicate a poor prognosis and are highly suggestive of a syndromic association, most commonly trisomy 13[4].***

Keywords: holoprosencephaly, fetal anomalies, prenatal ultrasound, trisomy thirteen, congenital malformations

1. Introduction

Holoprosencephaly (HPE) is a spectrum of forebrain malformations caused by failure of normal division of the prosencephalon during early embryogenesis[2, 6]. Based on severity, it is classified into alobar, semilobar, and lobar types, with alobar being the most severe form[6]. The condition is frequently associated with craniofacial anomalies and systemic malformations, particularly involving the cardiovascular and genitourinary systems[4, 7]. Prenatal ultrasonography plays a crucial role in early diagnosis and prognostication[5, 9].

2. Case Report

A routine antenatal ultrasound examination was performed in a pregnant woman at 16 weeks of gestation.

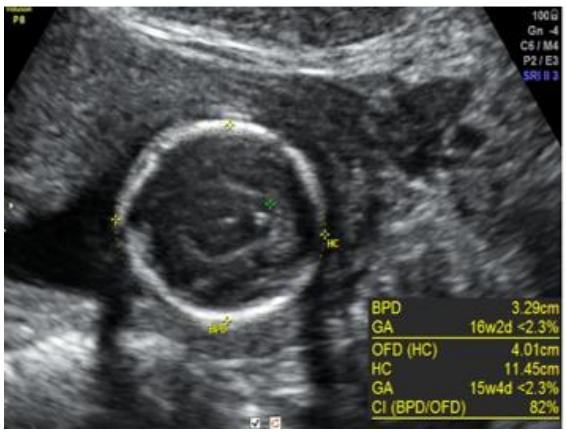
Obstetric Details

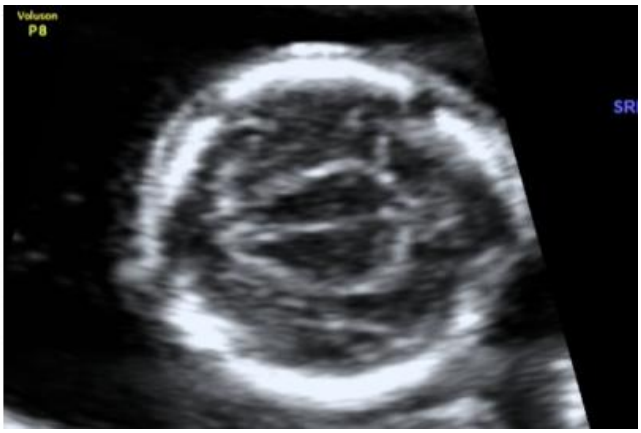
- Single live intrauterine fetus
- Gestational age: 16 weeks
- Placenta: Anterior, Grade I
- Estimated due date: 11/04/2026

Ultrasound Findings

Central Nervous System:

- Large single ventricular cavity (monoventricle)
- Fused thalami
- Absence of midline structures including falx cerebri→ Findings consistent with **alobar holoprosencephaly[1, 2]**



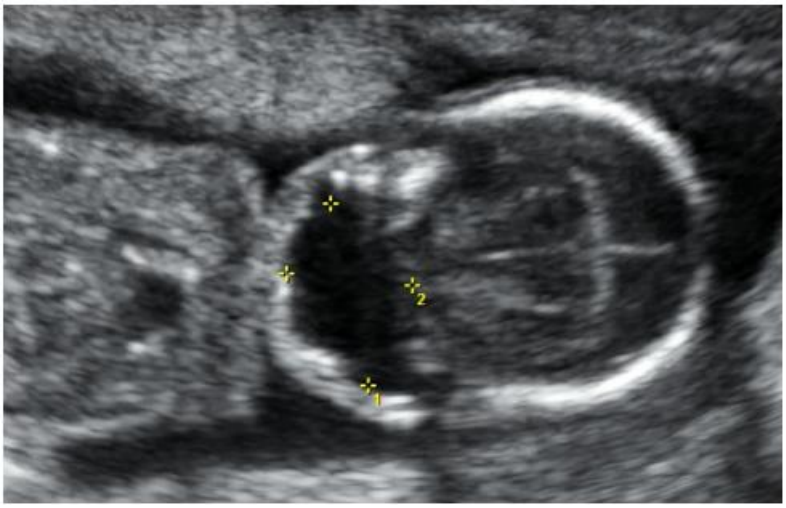


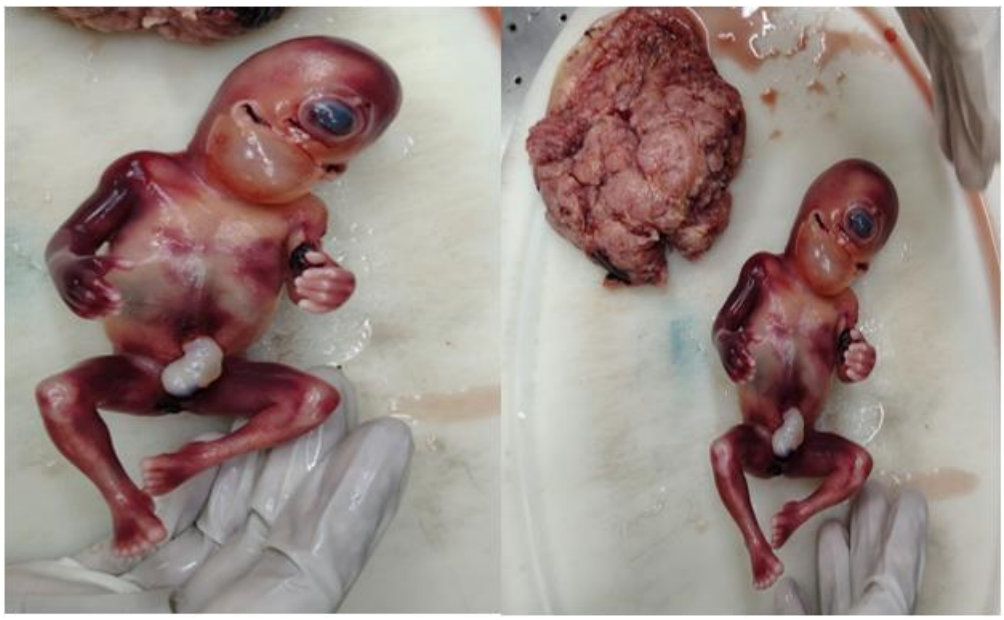
Facial Findings:

- Severe facial dysmorphism
- Absence of identifiable orbits
- Midline proboscis→ Suggestive of cyclopia spectrum anomaly[4]

Additional Findings:

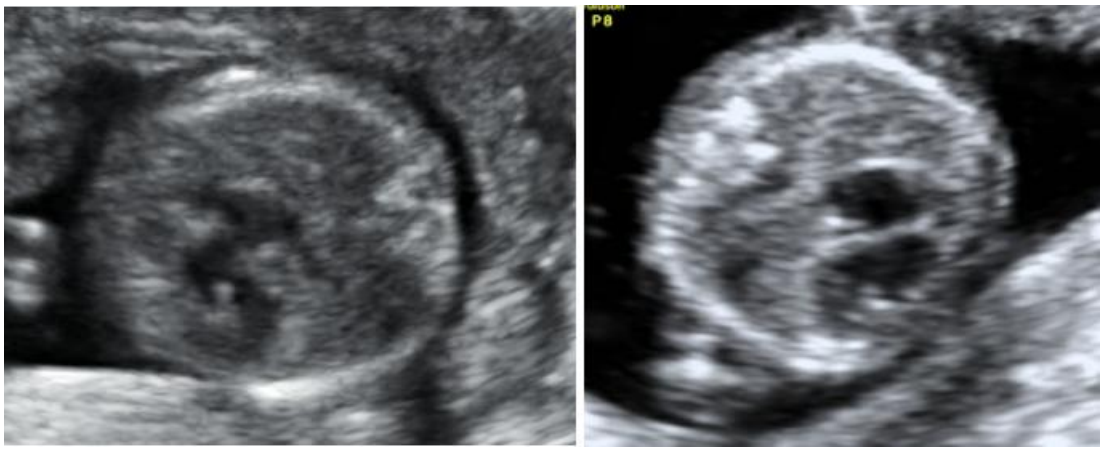
- Cystic lesion in lower facial/foregut region, likely foregut duplication cyst



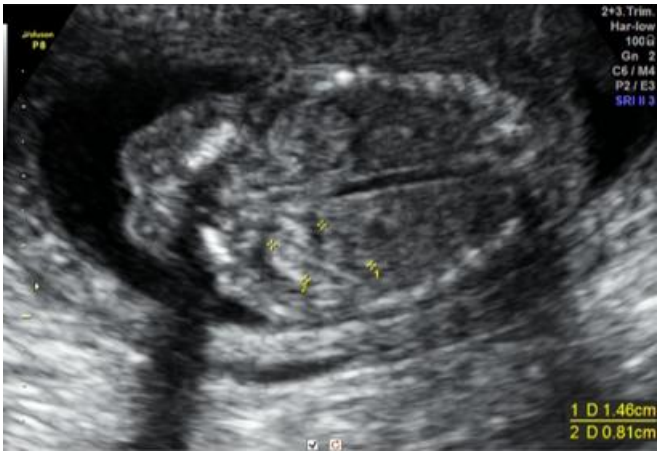


Cardiac Findings:

- Endocardial cushion defect
- Complex atrioventricular septal defect→ Frequently associated with HPE[4]



Renal Findings:
Bilateral echogenic kidneys → Suggestive of syndromic association



3. Discussion

Alobar holoprosencephaly represents the most severe end of the HPE spectrum and is characterized by complete failure of hemispheric division, resulting in a single ventricular cavity and fused thalami[2, 6]. The absence of midline structures such as the falx cerebri is a key diagnostic feature[1].

Craniofacial anomalies strongly correlate with the severity of brain malformation, supporting the concept that "the face predicts the brain"[8]. Findings such as proboscis and absent orbits indicate severe midline developmental defects[4].

Holoprosencephaly is frequently associated with:
- Chromosomal abnormalities (most commonly trisomy 13)[4, 7]
- Congenital heart disease (especially AVSD)[4]
- Renal anomalies
- Gastrointestinal malformations

Prenatal ultrasound remains the primary modality for diagnosis, with MRI serving as a complementary tool in selected cases[5].

4. Conclusion

Second trimester ultrasonography plays a crucial role in the early detection of severe congenital anomalies such as alobar holoprosencephaly[9]. The presence of associated multisystem anomalies indicates a poor prognosis. Early diagnosis enables appropriate genetic counseling and informed decision-making regarding pregnancy management.

5. Learning Points

- Alobar holoprosencephaly is characterized by monoventricle and fused thalami[1, 2]
- Facial anomalies such as proboscis and absent orbits indicate severe diseas[e]
- Frequently associated with cardiac and renal anomalies[4, 7]
- Strong association with chromosomal abnormalities, especially trisomy 13[4]
- Early ultrasound diagnosis is essential for counseling and management[5, 9]

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