

EP06.92 Holoprosencephaly: prenatal diagnosis and associated anomalies in a case series of five fetuses

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Objective :

To evaluate prenatal imaging findings, associated anomalies, and chromosomal abnormalities in fetuses diagnosed with holoprosencephaly (HPE) .

Methods :

Retrospective longitudinal study was conducted over a 5-year period at Gynaecology and Obstetrics department B-Charles Nicolle Hospital, Tunis, Tunisia.

Fetuses diagnosed with HPE during prenatal ultrasound examination were included. Detailed fetal neurosonography was performed in all cases. Postnatal assessment and chromosomal analysis were reviewed when available. Clinical characteristics, associated malformations, and genetic findings were analysed.

Results:

- ✓ Five fetuses with HPE were identified.
- ✓ Mean gestational age at diagnosis: 15 weeks + 4 days.
- ✓ The alobar subtype was the most frequent, accounting for three cases, followed by one semilobar and one lobar form,
- ✓ Severe craniofacial abnormalities were mainly observed in the alobar cases and included features such as cyclopia, proboscis, and arrhinia.
- ✓ Milder facial anomalies such as hypotelorism and median facial cleft were observed in the semilobar and lobar forms.
- ✓ Chromosomal abnormalities were identified in several cases, with trisomy 13 being the most frequently detected abnormality.
- ✓ Additional structural anomalies, including neural tube defects were also observed in some fetuses.

CONCLUSIONS

This case series highlights the marked clinical and genetic heterogeneity of holoprosencephaly. Early prenatal diagnosis using detailed neurosonography is essential for identifying associated anomalies and guiding genetic evaluation and parental counseling.