Introduction

Agenesis of the Corpus Callosum (ACC) involves a partial or complete absence of the main commissural pathway that connects the two cerebral hemispheres and can be an isolated abnormality or complex, coexisting with other abnormalities. It is rare although a precise incidence and prevalence is difficult to ascertain mostly due to selection bias. It is usually identified at 20-weeks ultrasound scan, however it remains a diagnostic challenge and may be missed leading to late detection in utero or postnatally.

Case Summary

A 29-year-old G2 P1 lady with a previous SGA baby delivered by SVD, smoker with BMI 23, got referred in by her midwife at 34+4 weeks gestation with suspected IUGR when ultrasound scan identified severe fetal ventriculomegaly with head circumference (HC) measuring above the 95th centile. Follow-up at FMU initially suspected a major vascular event. Fetal MRI confirmed ACC with interhemispheric cyst and severe bilateral ventriculomegaly. Her virology screen (CMV, Toxoplasma & Rubella) remained negative with no evidence of autoimmune thrombocytopenia. Aminoacetic acid confirmed a normal karyotype. Due to guarded prognosis, with high likelihood of significant neurological problems, the couple decided to proceed with termination of pregnancy. Mem was induced to labour at 35+4 weeks gestation following a feticide with KCI injection to arrest fetal heart delivering a male baby weighing 2840gm (77% birth centile) by SVD. Post-mortem examination was declined by parents and placental histology and swab culture were normal.

Avoiding Diagnostic Pitfalls in Future

Complete agenesis of the corpus callosum (cACC), the most common commissural anomaly diagnosed prenatally, is often suspected on fetal ultrasound when the cavum septi pellucidi (CSP) is absent and additional signs, such as teardrop-shaped lateral ventricles, are present. In cases of partial agenesis of the corpus callosum (pACC) the CSP is often present, which makes it difficult to suspect anomalies of the corpus callosum prenatally. Kari et al. propose a reproducible simple method with the potential to identify fetuses at high risk for pACC. By calculating CSP length-to-width ratio between 20 and 34 weeks’ gestation and by constructing reference ranges in relation to biparietal diameter (BPD), they show that fetuses with a normal-sized corpus callosum have a rectangular-shaped CSP, with a CSP ratio > 1.5 in the second half of gestation (a). Most fetuses with pACC have an abnormally shaped, wide and short CSP, with a decreased CSP ratio (b, c, d).

Images

[Images showing ultrasound scans and MRI images]

Discussion

Chromosomal abnormality is associated in 18% cases of ACC with conditions like Aicardi and Andermann syndrome and non-syndromic cases like fetal alcohol syndrome. This case is a reminder that imaging in pregnancy as in ultrasound for fetal survey at 20 weeks is a screening test and will not detect all structural anomalies. Fetal MRI is an useful adjunct to ultrasound to confirm diagnosis alongside karyotyping. Prognosis of the condition varies and hence antenatal counselling and management poses ethical dilemma and controversies amongst patients and clinicians. Due to 3-4% risk of recurrence, this patient is offered scan in FMU at 20 weeks gestation in future pregnancies.

References


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