

EP06.74 Isolated versus non-isolated fetal choroid plexus cysts: association with trisomy 18 and clinical implications

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Objective :

To evaluate the association between fetal choroid plexus cysts (CPC) and chromosomal abnormalities according to the presence or absence of additional ultrasound findings.

Methods :

Retrospective study was conducted over a 5-year period at Charles Nicolle Hospital, Tunis, Tunisia.

Fetuses diagnosed with CPC on first-trimester ultrasound were included. Cases were classified as isolated or associated with other anomalies. Chromosomal abnormalities and perinatal outcomes were analysed.

Results:

- Among 2286 first-trimester ultrasound examinations, 62 CPCs were identified (2.7%). Mean gestational age at diagnosis was 13 weeks + 2 days.
- CPC was isolated in 52 cases (83.8%), with normal karyotype in 96.2% (50/52). Two cases of trisomy 18 were identified (3.8%).
- In 10 cases (16.1%), CPC was associated with additional abnormalities, most commonly ventriculomegaly. Trisomy 18 was confirmed in 90% of these cases (9/10).
- Pregnancy termination was performed in all cases with confirmed aneuploidy. Postnatal outcomes were normal in all fetuses with normal karyotype.

CONCLUSIONS

Isolated CPC is associated with a low risk of chromosomal abnormalities, whereas the presence of additional ultrasound findings markedly increases the risk of trisomy 18. These results support a targeted approach to invasive testing based on ultrasound findings.