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A case of holoprosencephaly and dandy walker malformation associated with triploidy

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Objective

We report on a rare case of a triploidy fetus, co-existing with dandy walker malformation and holoprosencephaly.

Methods

Case report.

Results

A 31-year-old G1P0 with spontaneous pregnancy was referred to our centre at 16 weeks and 6 days gestation for further assessment of a choroid plexus cyst and low serum PAPP-A. Obstetric and medical histories were all unremarkable. Sonographic findings were dandy walker malformation, holoprosencephaly, oligohydramnios, asymmetric ventriculomegaly and right atrial isomerism. Amniocentesis revealed triploidy (69 XXX).

Conclusion

Holoprosencephaly and dandy walker malformation can be the result of triploidy.