

## A case of prenatal detection of trisomy 13

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### Objective

To describe the prenatal detection of trisomy 13.

### Methods

Case report.

### Results

A 39 years old, G4P3, was referred to our unit at 22 weeks with observed structural defects during the anomaly scan at her local hospital. The patient did not have any screening for chromosomal abnormalities during the 1st or 2nd trimester of pregnancy. Our ultrasound examination revealed hydrocephalus, occipital meningocele, interventricular communication and unilateral multicystic kidney. We performed an amniocentesis which indicated fetal trisomy 13. The parents opted for termination of pregnancy. A 24 -year-old multipara late registrant woman admitted to our hospital at 18 weeks of gestation for first routine examination. Her past medical and obstetric histories were unremarkable. There was no history of genetic disorders or structural anomalies noted in the family history of the both parents. Sagittal and axial sonography of the fetal face depicted a midline soft-tissue mass measuring 1.8 × 1.5 × 1.3 cm projecting anteriorly from the lower aspect of the fetal forehead above the nasal bridge and between the fetal orbits (Figure 1). The soft tissue mass appeared to contain both tissue and fluid. No other abnormalities were noted in detailed USG. Following genetic counseling, the patient declined amniocentesis and the parents were decided to continue the pregnancy. Structural defects were confirmed by autopsy postnatally.

### Conclusion

Trisomy 13 is a rare disease, however it is important to screen for this condition in the first trimester so an early and safer termination can be offered to the parents.

