Down syndrome—novel associations: antenatal strawberry-shaped skull, severe neonatal hepatitis and an unusual form of portosystemic hepatic shunt

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DESCRIPTION
A fetal anomaly scan at 13 weeks gestation showed cystic hygroma and amnion separation.

Further scans at 19 weeks revealed a ‘strawberry-shaped skull’ (figure 1). Additional findings were increased nuchal translucency, ventriculomegaly and polyhydramnios. These were suggestive of trisomy 18. An antenatal sonographic finding of a strawberry-shaped skull is a marker of serious fetal malformation, most commonly associated with trisomy 18 and thanatophoric dysplasia.1 The risk of miscarriage is increased with amnion separation.

The parents declined a termination.

At 22 days of age, the patient presented with prolonged conjugated jaundice. He was exclusively breastfed.

Figure 1  Strawberry-shaped skull.

Figure 2  Anomalous communication between left portal vein and left hepatic vein.

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References

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breast fed, his stools were never pale in colour and he remained well.

His highest bilirubin was 138 μmol/L with a direct component of 35 μmol/L.

Abdominal ultrasound Doppler scan showed ‘increased peri-portal echoes’ throughout the liver, and an anomalous vascular communication between the middle hepatic vein and right intra-hepatic portal vein (figure 2). Virology screen was negative. Metabolic screen was normal and disorders of bile acid synthesis were also excluded. A liver biopsy result was in keeping with severe neonatal hepatitis.

Congenital intrahepatic venous shunts in Down syndrome are rare.2 However, anomalous communication between left portal vein and left hepatic vein has not been previously described in Down syndrome. Portosystemic shunts are clinically insignificant and resolve without intervention.3

This is the first reported case of Down syndrome with an antenatal strawberry-shaped skull, neonatal hepatitis and specific type of portosystemic shunt.

These findings have not been previously described in this common genetic disorder.

The patient made a full recovery from his hepatitis.

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