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

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Accepted 8 October 2015

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

Figure 1 Strawberry-shaped skull.

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

To cite: Losa I, Owoeye A.
BMJ Case Rep Published
online: [please include Day
Month Year] doi:10.1136/
bcr-2015-212852

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

His highest bilirubin was 138 $\mu\text{mol/L}$ with a direct component of 35 $\mu\text{mol/L}$.

Abdominal ultrasound Doppler scan showed 'increased periportal echoes' throughout the liver, and an anomalous vascular communication between the middle hepatic vein and right intrahepatic 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.

Acknowledgements The authors thank to Dr David Wright for technical editing of the manuscript.

Contributors AO revised the manuscript critically for important intellectual content.

Competing interests None declared.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

Learning points

- ▶ An antenatal scan finding of a strawberry-shaped skull could be a feature of Down syndrome.
- ▶ Severe neonatal hepatitis can present as prolonged conjugated jaundice in breast fed neonates.
- ▶ Anomalous communication between the left portal vein and left hepatic vein has not been previously described in Down syndrome.

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