Extrahepatic portosystemic venous shunt presenting as galactosaemia

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A newborn screening program identified a 23-day-old asymptomatic girl with elevated serum galactose (>18.3 mg/dl) after breast feeding. The enzymes that metabolise galactose showed normal activity, thereby excluding hereditary galactosaemias. Three-dimensional CT venography (Fig. 1) showed direct portocaval communication between the inferior mesenteric vein and the left internal iliac vein. Inferior mesenteric venous portography via the shunt confirmed that the flow was hepatofugal through the communication (Fig. 2). Intrahepatic

Fig. 1 3-D CT venography

Fig. 2 Inferior mesenteric portography

portal venous supply was preserved. The girl was treated with catheter embolisation.

Congenital extrahepatic portosystemic (PS) venous shunt is a rare form of PS shunt that can cause hypergalactosaemia in infants [1, 2]. These malformations may lead to severe complications, such as heart failure, pulmonary hypertension, encephalopathy and hepatic tumours, and necessitate life-long follow-up if closure is not performed [1, 2].

References

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