

Case Reports

Congenital intrahepatic portosystemic shunt presenting with mucocutaneous bleeding

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Introduction

Congenital portosystemic shunts (CPSS) are rare vascular abnormalities characterized by complete or partial shunting of portal blood into systemic veins bypassing the liver¹. Anatomically, CPSS are classified into extra- and intra-hepatic shunts based on their location². Some of these shunts are asymptomatic and resolve spontaneously whereas others cause marked metabolic abnormalities affecting a variety of organ systems³. Common presentations of CPSS include neonatal cholestasis, encephalopathy, liver tumours and hepatopulmonary syndrome; however, severe coagulopathy is rare⁴. Here, we report a child with congenital intrahepatic portosystemic shunt presenting with a bleeding diathesis.

Case report

An 8-year old Sri Lankan boy presented with epistaxis and ecchymosis of 3 days duration. He started to have a few ecchymotic patches on the back which then progressed throughout the body during the past three days. There was no haematemesis, melaena, haematuria or evidence of bleeding into any other organs. He also complained of unsteady gait and excessive sleepiness which lasted 24 hours before resolving spontaneously.

At 1 year of age, he has had an episode of nephrotic syndrome which was complicated by haematuria and hypertension that was later diagnosed as mesangio-proliferative glomerulonephritis by renal biopsy. Nonetheless,

he did not have further relapses. At five years, he developed an afebrile generalized tonic clonic seizure without an identifiable provoking factor. He subsequently had three further seizures of similar semiology and was started on sodium valproate 20mg/kg/day for idiopathic generalized epilepsy. Electroencephalogram was normal. He was born to non-consanguineous parents with a birth weight of 3.3kg and required phototherapy for neonatal jaundice. Growth and childhood development were age appropriate except that he was a slow learner with poor school performance. He had no family history of liver diseases or sudden and unexplained deaths.

On examination, his growth parameters were within normal limits. He had multiple ecchymotic patches on the body but did not have any peripheral stigmata of chronic liver disease, hepatosplenomegaly or ascites. The remainder of the physical examination, including nervous system, was clinically normal.

The full blood count revealed: haemoglobin 11.1g/dL, haematocrit 32.1%, white blood cell count $9.8 \times 10^3/\mu\text{L}$ (neutrophils 20%, lymphocytes 73%) and platelet count $19 \times 10^3/\mu\text{L}$. Blood picture showed a mixed population of normochromic normocytic red cells, macrocytes and acanthocytes and a marked reduction in platelets. Liver function tests revealed: total bilirubin 39 $\mu\text{mol/L}$ (normal 1-24 $\mu\text{mol/L}$), direct bilirubin fraction 33%, aspartate transaminase 54U/L (normal 10-40U/L), alanine transaminase 13U/L (normal 10-40U/L), alkaline phosphatase-185U/L (normal 104-345U/L), gamma glutamyl transferase 17U/L (normal 9-48U/L) and serum total protein 5.3g/dl (albumin 2.7g/dL, globulin 2.6g/dL). The erythrocyte sedimentation rate, C-reactive protein and renal functions were normal.

His coagulation profile was deranged. His prothrombin time (PT) was 22s (normal 12-16s) with an INR of 1.7 while the activated partial thromboplastin time (APTT) was 39.3s (normal 24-35s). The thrombin time was normal (19.1s). PT and APTT following 50:50 correction were normal (15.7s and 29.9s respectively). His fasting plasma

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ammonia level was 74 μ mol/L (normal <40 μ mol/L).

Abdominal ultrasonography showed a portosystemic shunt between the right branch of portal vein and inferior vena cava (IVC) with associated chronic parenchymal changes of the right lobe of the liver. Contrast enhanced computed tomography of abdomen with portal venography confirmed the presence of directly communicating intrahepatic portosystemic shunt between the right portal vein and the IVC (Figure 1) with faintly visible focal lesions in segment VIII of the liver during arterial phase. Echocardiogram did not show evidence of pulmonary hypertension. The child is awaiting surgical closure of the shunt.



Figure 1: Contrast enhanced computed tomography image of upper abdomen demonstrating intrahepatic portosystemic shunt draining from right branch of the portal vein into the IVC (arrow).

Discussion

CPSS was first described by John Abernethy in 1793 and can be intrahepatic or extrahepatic⁵. Extrahepatic CPSS is further subdivided into types 1 and 2 based on the patency or apparent non patency of the portal trunk and intrahepatic portal system⁶. CPSS can cause a wide variety of clinical manifestations in older children including encephalopathy, liver tumours, hepato-pulmonary syndrome and pulmonary hypertension but coagulation abnormalities are sparsely reported⁴.

The most unusual feature in our patient is that he came to medical attention following mucocutaneous bleeding. Investigations confirmed coagulopathy derangement in both intrinsic and extrinsic pathways along with severe thrombocytopenia. Although inadequate production of clotting factors due to hepatic derangement could explain the coagulopathy in this child, it will not clarify the severe thrombocytopenia. Thrombocytopenia could be due to splenic sequestration of platelets but, our patient neither had splenomegaly nor portal hypertension.

Therefore, we believe that deranged PT, INR, APTT and severe thrombocytopenia of this child are probably due to a consumptive coagulopathy within the shunt.

Neurological manifestations of CPSS include irritability, dyslexia, lethargy, EEG abnormalities, extrapyramidal signs, and epilepsy^{7,8}. These manifestations are most likely to be due to bypass of hepatic detoxifying mechanisms of toxic compounds, including ammonia, leading to increased plasma levels⁴. Our patient did not have overt encephalopathy but had four seizures which may have been provoked by episodes of hyperammonaemia.

CPSS can also cause nodular lesions, focal nodular hyperplasia, hepatocellular adenoma, hepatocellular carcinoma and hepatic sarcoma in affected patients⁷. Focal nodular hyperplasia shows homogeneous strong enhancement in the arterial phase⁹. The faintly visible liver focal lesions during the arterial phase, which was seen in our patient, are most likely to be due to focal nodular hyperplasia. Other associated feature of CPSS includes intrauterine growth retardation, membrano-proliferative glomerulonephritis, congestive heart failure, pancreatitis, pulmonary hypertension, hepato-pulmonary syndrome and autoimmune disorders of which our patient had glomerulonephritis in the past^{4,7}. In summary, this case report highlights an unusual presentation with coagulopathy of a rare anatomical malformation involving the portal venous system.

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