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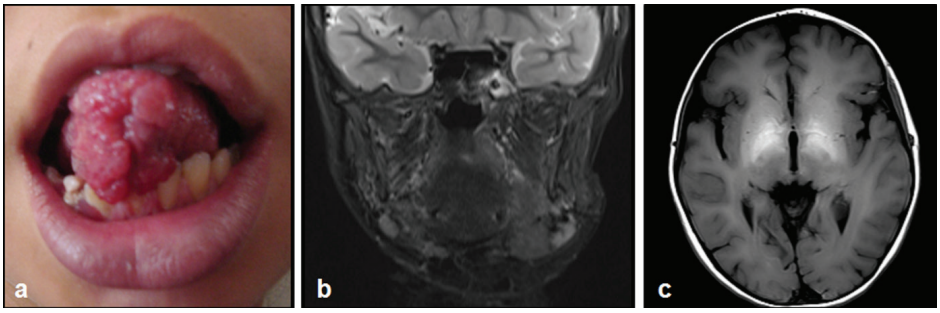

Figure 1. (a) The photograph of the mouth reveals noticeable enlargement of the tongue with poorly demarcated midline. The photograph also depicts advanced dental development on the left side. **(b)** Coronal T2 weighted MRI reveals marked thickening of soft tissues of the left hemiface. **(c)** Axial T1-weighted image demonstrates bilateral symmetric high signal intensity at the globus pallidus.

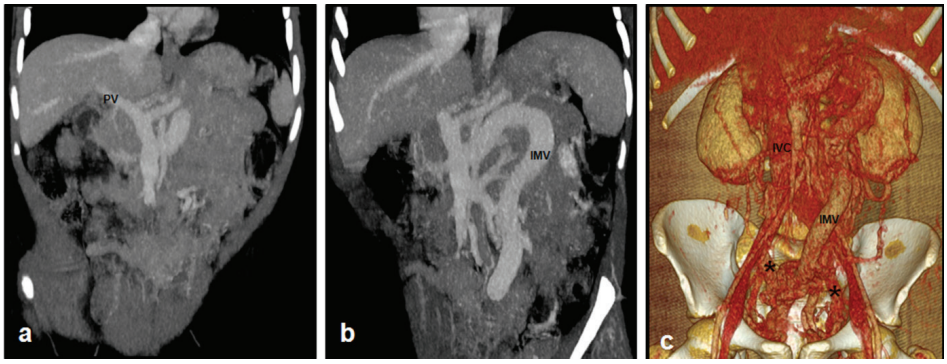

Figure 2. (a, b) Coronal maximum intensity projection images from contrast material–enhanced CT reveal an abnormally dilated inferior mesenteric vein (IMV). Decrease in calibration of the portal vein (PV) is clearly shown. **(c)** Volume rendered CT angiography image shows the splenic vein and the superior mesenteric vein joining and draining into the right and left iliac veins (*) through the dilated inferior mesenteric vein (IMV). Increase in calibration of the inferior vena cava (IVC) is also clearly visible.

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A ten-year-old girl was admitted to the emergency clinic with recurrent oral bleeding. Physical examination showed facial hemihypertrophy and a microcystic lymphatic malformation of the tongue as the source of bleeding (Figure 1a). There were also cutaneous hemangiomas localized in the lumbosacral and gluteal region. In neurologic assessment, mild mental retardation was detected. Laboratory results were consistent with mildly increased liver enzymes and hyperammonemia.

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Coronal T2 weighted craniofacial magnetic resonance imaging (MRI) showed left hemifacial hypertrophy (Figure 1b). Increased signal intensities were also detected in bilateral globus pallidus on T1 weighted MRI (Figure 1c). Multiplanar reconstruction, maximum intensity projection and three-dimensional volume rendered computed tomography (CT) images revealed hypoplastic portal vein with dilated tortuous inferior mesenteric vein bleeding (Figure 2a-2c). At the pelvic level, multiple porto-systemic shunts were delineated between the inferior mesenteric vein and iliac veins.

What is your diagnosis?
See the next page for your diagnosis.

Photoclinic Diagnosis:

Co-existence of Abernethy malformation and Klippel-Trenaunay-Weber syndrome

Complete absence of the portal vein and drainage of splenic and mesenteric veins into systemic veins occur as a rare condition. This spectrum of disease is also known as congenital extrahepatic portosystemic shunt and was first described by Abernethy. The Klippel-Trenaunay-Weber syndrome is, on the other hand, an uncommon but well recognized congenital disorder. It is characterized by cutaneous capillary malformations, soft tissue or bony hypertrophy and varicose veins or venous malformations.

We present a case of Klippel-Trenaunay-Weber syndrome with Abernethy malformation in a ten-year-old girl who was admitted to our clinic with oral bleeding as the initial symptom. To our knowledge this is the first case of coexistence of both Abernethy malformation and Klippel-Trenaunay-Weber syndrome in the literature.

Morgan and Superina[1] classified congenital extrahepatic portosystemic shunt into two types. Type 1 shunts are characterized by absence of the intrahepatic portal vein and complete end-to-side shunt, and have two subtypes, (i) separate drainage of the superior mesenteric and splenic veins into the IVC, iliac veins, or renal veins (subtype Ia) and (ii) superior mesenteric and splenic veins joining to form a short extrahepatic portal vein which drains into the inferior vena cava (subtype Ib). Type 2 shunts are marked by presence of a patent intrahepatic portal vein and a partial side-to-side shunt. Our patient had this type of Abernethy malformation.

Klippel-Trenaunay syndrome is characterized by the following triad of features: 1) cutaneous capillary malformations 2) soft tissue or bony hypertrophy and 3) varicose veins or venous malformations, often with persistent lateral embryologic veins.[2] The findings are usually limited to one extremity; however, involvement of multiple extremities, unilateral, or even whole body involvement have been reported. While the legs are the most commonly affected site, the arms, trunk, and rarely the head and neck may also be involved.

Currently, these abnormalities are usually diagnosed by noninvasive cross-sectional imaging techniques such as ultrasound, CT or MRI. CT angiography which shows the shunt and any intrahepatic portal vein branches can confirm the Abernethy malformation diagnosis.

References

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