

Venous malformations as a rare cause of lower gastrointestinal bleeding in a young patient with Blue Rubber Bleb Nevus Syndrome

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Question

We report a case of 19-year-old male who presented with complaints of recurrent episodes of passage of black coloured stools since last 2 weeks. The patient reported that he had multiple blue coloured lesions in hands, legs and face since childhood. At physical examination, the lesions were of variable sizes ranging from 3-10 mm and resembled small bubbles of blue colouration. Biopsy taken from one of the lesions in childhood was suggestive of cavernous haemangioma. Laboratory investigations revealed microcytic anaemia (haemoglobin 9 g/dL; serum ferritin 22ng/ml; transferrin saturation 10%) with normal rest of the laboratory parameters. His stool sample tested positive for occult blood. Further colonoscopy done to look for the origin of bleed revealed multiple, variable sized vascular malformations involving the colon and visualized distal 10 cm of terminal ileum without any active bleed. Gastroscopy revealed multiple vascular malformations in the stomach and second part of duodenum.

Computed tomography of the abdomen done after distension with oral water demonstrated multiple well defined homogeneously enhancing polypoidal lesions along the intraluminal surface of ileum, few of which in the terminal ileum showed foci of calcifications within likely phleboliths. Homogeneously enhancing polypoidal lesions were also seen involving the ascending, transverse, descending colon and the rectum. The lesions were suggestive of multiple slow flow venous malformations. In addition, there was also presence of a large irregular lesion in the pelvis involving left iliacus muscle with no erosion/destruction of underlying ileum. The lesion showed homogeneous enhancement with few foci of calcifications suggesting venous malformation (Figure 1).

What is your diagnosis?

Answer

Based on the history, clinical examination and computed tomography findings, the diagnosis of Blue Rubber Bleb Nevus Syndrome (BRBNS) was made. The patient was conservatively managed and was given oral iron supplementation. Currently, the patient is

healthy and has not complained of any new episode of gastrointestinal bleeding. In case of rebleed, primary endoscopic intervention like argon plasma coagulation or endoscopic mucosal resection of the bleeding vascular malformations is intended.

Blue Rubber Bleb Nevus Syndrome also called as Bean syndrome is a rare disorder characterised by multiple cavernous venous malformations involving several organ systems particularly the skin and the gastrointestinal tract (1). The cause of this syndrome is unknown and an autosomal dominant transmission has been suggested. The cutaneous lesions are invariably present at birth and have a propensity to increase in size and frequency with age. Whereas, the skin lesions are predominantly seen in the upper extremities and trunk, small bowel and the colon are most frequently involved in the gastrointestinal tract (2). As opposed to skin lesions, the gastrointestinal lesions frequently bleed. Computed tomography owing to its high spatial and temporal resolution is an excellent modality to depict gastrointestinal lesions and to assess the complications like intussusception. In addition, involvement of other systems like left iliacus in the present case can also be depicted. The mainstay of the treatment lies in iron supplementation, oral or intravenous with blood transfusions as indicated. Newer treatment modalities include endoscopic techniques like laser, argon plasma or band ligation (3). The present case highlights that although BRBNS is a rarely reported disease, it can be responsible for lower gastrointestinal bleeding and chronic anaemia in young patients.

References

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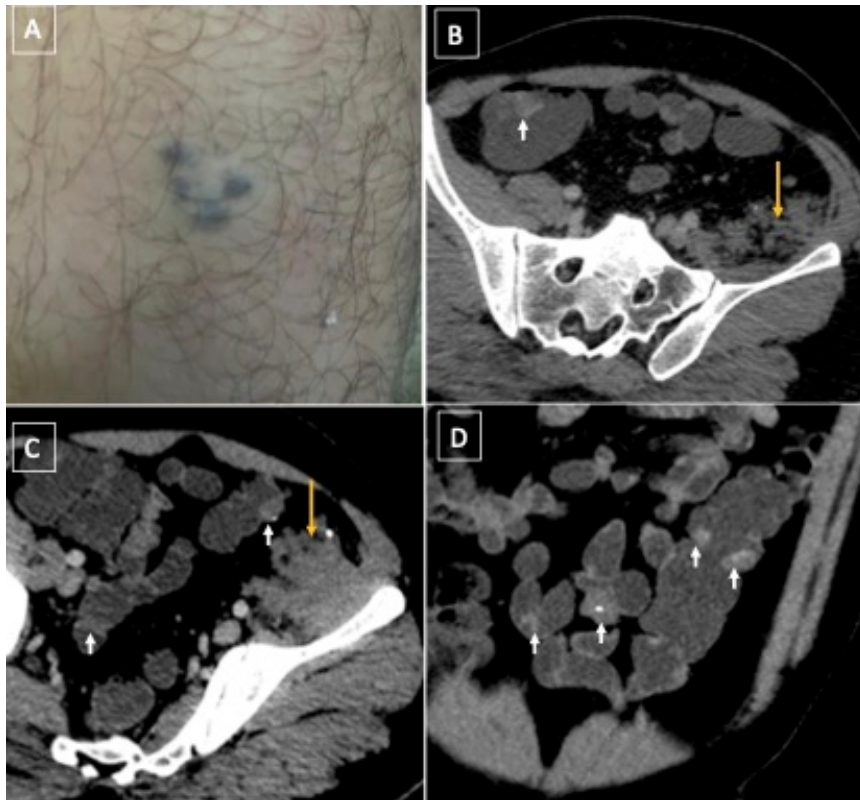


Figure 1. — Photograph of left leg (A) revealed multiple deep blue rubbery blebs of variable sizes present on the skin. Axial (B and C) and coronal image (D) demonstrated multiple homogeneously enhancing polypoidal venous malformations involving ileum and colon (demonstrated by white arrows). Irregular venous malformation with foci of calcification was seen involving the left iliacus muscle (yellow arrow).