Blue Rubber Bleb Nevus Syndrome - A Rare Cause of Anemia in Children

ABSTRACT
A 10 year old boy presented with generalized weakness and progressive pallor requiring multiple blood transfusions. The skin demonstrated multiple raised, bluish-black lesions over extremities, trunk and oral cavity. He also suffered from several episodes of maelena & upper GI bleeding, chronic anemia and growth retardation. The endoscopic examination of GIT revealed multiple bluish black, sessile, venous malformation of various sizes. A diagnosis of Blue rubber bleb nevus syndrome was made which is a rare disease characterized by distinctive cutaneous and gastrointestinal venous malformation causing massive or occult gastrointestinal hemorrhage and iron deficiency anemia.

INTRODUCTION
Blue Rubber Bleb Nevus Syndrome (BRBNS) is a rare entity consisting of distinctive venous malformation in the Skin, gastrointestinal tract & less often in other organs, leading to occult or profound GI bleeding & chronic anaemia. So far only 150 cases reported in the world literature [1]. Most cases are sporadic but autosomal dominant inheritance has been reported. BRBNS was probably first observed by Gascoyen in 1860 [2]. A century later, Bean coined the term blue rubber bleb nevus syndrome for this rare disorder. Diagnosis may not be readily apparent in the absence of sub cutaneous nodules [3]. BRBNS is an important syndrome because of its potential for serious or fatal bleeding. Here in we are reporting a case of Blue rubber bleb nevus syndrome in a 10 year old boy. The case is being reported for its rarity and for adding to the existing literature.

CASE REPORT
A 10 year old boy presented with history of recurrent gastrointestinal bleeding and progressive pallor requiring recurrent blood transfusions for the last 2 years. He had a history of a wall-nut sized cutaneous swelling located on the right side of his forehead since birth and the swelling was resected when he was 1 year old. Parents have noticed bluish black swelling over upper & lower extremities, trunk and oral mucosa two years back. There was no family history suggestive of bleeding disorder and parents were not anemic. There was no history of consanguinity.

On examination severe pallor and growth retardation (weight-22kg, height-123 cms) were noted. There was an incisional scar over the right side of forehead. The skin had multiple raised, bluish-black lesions over extremities trunk and oral cavity ranging in size from 1 to 2 cm in diameter [Table/Fig-1]. Oral cavity showed sub mucus blebs over soft palate and posterior pharynx (0.2-0.5cm). Systemic examination revealed hyperdynamic precordium and systolic murmur (grade 2/6) and there was no organomegaly.
are present at birth or may appear in childhood & increase in size & number with age [4, 5]. The skin & GI system are most frequently involved, with multiple vascular blebs or nodules. Skin nodules are usually multiple, protuberant, bluish, compressible blebs, a few millimeters to several centimeters in diameter and varied in shape. Most of these are symptomatic but some may be spontaneously painful or tender when palpated. Skin lesions rarely bleed unless traumatized [6].

Gastrointestinal system involvement usually becomes evident during early childhood. Venous malformations may occur anywhere from oral to anal mucosa but predominantly occur in the small bowel [4, 7], leading to massive hemorrhage and iron deficiency anaemia that may be severe and require blood transfusions & iron supplementation.

Blue rubber bleb nevus have also been reported in the nasopharynx, eyes, thyroid, parotids, central nervous system, skeletal muscles, pleura, peritoneum, pericardium, mesentry, lungs, kidneys, liver, spleen, penis, vulva and urinary bladder [8].

Symptoms and signs vary depending on the organ system involved. Patients may report fatigue from occult blood loss. Hematemesis, melena, or rectal bleeding may prompt emergency presentation and this was also the presentation of the disease in our patient.

Fecal occult blood test should be performed in order to screen for the occult blood loss from the GI lesions. Screening for the iron deficiency anaemia has to be performed [9]. Radiographic images may be useful in suspected bone & joint involvement & radiographic contrast techniques detect GI lesions but endoscopy is considered to be superior. Endoscopy also provides the opportunities to treat & diagnose the lesions. MRI detects extracutaneous lesions in asymptomatic family members.

The important differential diagnosis are Kaposi sarcoma, Klippel Trenaunay-Weber syndrome and Maffucci syndrome from which it needs to be differentiated.

The treatment of GI venous malformations depends on their number, location, size & symptoms. Bleeding from GI lesions is usually managed conservatively with iron supplement & blood transfusion when necessary. Endoscopic coagulation or removal is an effective modality in case of repeated bleeding. Endoscopic sclerotherapy & endoscopic laser (Nd: YAG) photocoagulation & plasma argon coagulation have been used successfully for the lesions in the gastrointestinal tract. If vascular lesions are confined to a segment of the GI tract, resection of the involved segment of gut may be indicated. Being a rare entity, awareness about this condition helps in appropriate diagnosis and management [1, 10].
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REFERENCES


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