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Case Report

Paediatrics Section

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

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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.

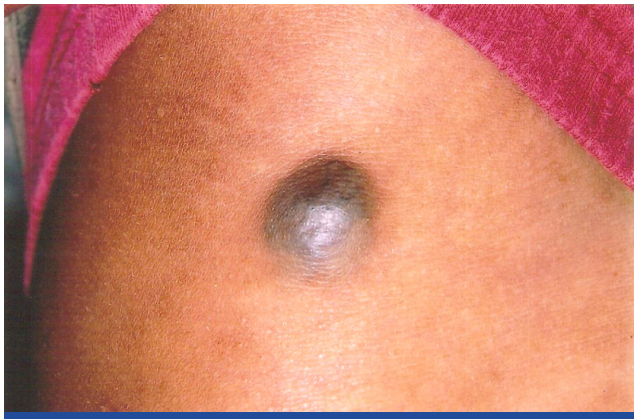
Key Words: Blue rubber bleb nevus syndrome, GI bleeding, Venous malformations

INTRODUCTION

Blue Rubber Bleb Nevus Syndrome (BRBNS) is a rare entity consisting of distinctive venus 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 cuteneous 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.

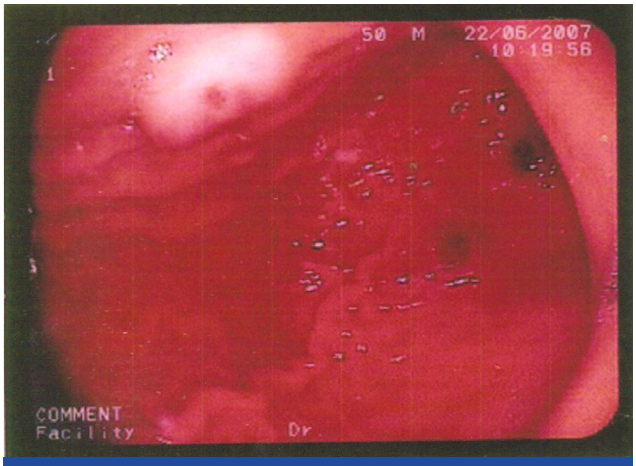
CASE REPORT

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.

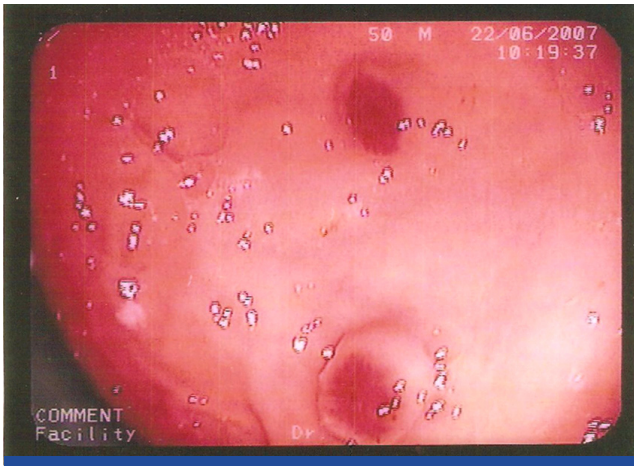


[Table/Fig-1]: -Black nodule over lower back (2.5cm- 3cm)

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.



[Table/Fig-2]: Upper GI Endoscopy showed multiple GI vascular malformations, Bluish-red submucosal hemangiomatous lesion measuring (1x2 cm), present in the stomach (fundus)



[Table/Fig-3]: There are 2-3 bluish-red submucosal hemangioma measuring 1x2 cms without stock, flat lesions, in the second part of duodenum

The laboratory investigations revealed a Hb of 2.2gm%, Total count of 4200/cumm, platelet count of 2.4 lac/cumm. Prothrombin time and partial thromboplastin time were normal. Peripheral smear showed microcytic hypochromic anaemia. Serum ferritin level was 4ng/ml (Normal: 9-120ng/ml). Serologic investigation for Hepatitis B and HIV were negative. Upper GI endoscopy showed multiple sub mucosal bluish blebs in the stomach, duodenum & jejunum [Table/Fig-2 and 3].

Histopathology was suggestive of hemangioma (cavernous venus malformation). Fundus examination did not reveal any vascular lesions. CT scan of brain for CNS lesions was negative. The child is on regular blood transfusions and iron supplementation (conservative management).

DISCUSSION

The typical symptom of BRBNS is venus malformations, which 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].

Gastro intestinal system involvement usually becomes evident during early childhood. Venus malformations may occur any where from oral to anal mucosa but predominantly occur in small bowel [4, 7], leading to massive hemorrange 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, thyroids, parotids, central nervus system, skeletal muscles, pleura, peritoneum, pericardium, mesentry, lungs, kidneys, liver spleen, penis, vulva and urinary bladder [8].

Symtoms and signs vary depending on the organ system involved. Patients may report fatigue from occult blood loss. Hematemesis, malena, 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 extracuteneous 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, recection 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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