CASE REPORT

Blue Rubber Bleb Nevus Syndrome: Case Report and Review of the Literature

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ABSTRACT

Blue rubber bleb nevus syndrome is a rare disorder characterized by multiple cutaneous venous malformations in the skin and gastrointestinal tract associated with intestinal hemorrhage and iron deficiency anemia. Almost all of the reported cases from India have been from the north of the country. We report the case of blue rubber bleb nevus syndrome on account of the rarity of the disease from our part of the country and review the current available literature on this topic.

Key words: Blue rubber bleb nevus syndrome – Gastrointestinal hemorrhage – vascular malformation - Iron deficiency anemia

Introduction

Blue rubber bleb nevus syndrome (BRBNS), is a rare disorder characterized by multiple venous malformations in the skin, gastrointestinal tract (GIT) and other visceral organs. It was first recognized by Gascoyen in 1860. One hundred years later, Bean further described the characteristic lesions and coined the term BRBNS.

The most common presentation is anemia secondary to GIT bleeding but it may rarely present with intussusception, hemotherax, hemopericardium, pulmonary hypertension, dementia, paraparesis, ataxia, cortical blindness and chronic consumption coagulopathy depending on the organ involved. We report the case of a female child with multiple lesions of skin and GIT who presented with severe iron deficiency anemia.

Case Report

A 14-year-old female patient was referred to us for evaluation of gastrointestinal (GI) blood loss in view of anemia and positive test for fecal occult blood. She had history of exertional dyspnoea, palpitations and syncope since childhood and had been on regular iron supplementation. She also gave a history of multiple bluish, bleb-like nodules in the skin all over the body since birth. She felt that these lesions were gradually increasing in size and number over time. She denied the occurrence of hematemesis, melena, hematochezia and overt bleeding elsewhere. She did not give history of peptic ulcer, chronic liver disease or NSAID intake. She was born of non-consanguineous marriage and did not have a family history of any major illnesses. She was pale and had a blood pressure of 100/60 mmHg and a pulse rate of 90/min. Skin examination revealed the presence of multiple, soft, compressible, tender, bluish-black, bleb like nevi all over the body including the scalp, tongue, palms and soles (Figure 1). Rest of the physical examination was normal. Laboratory investigations revealed hemoglobin of 3.1 g/dl, with normal total and differential WBC count, platelet count and liver function tests.

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Blue rubber bleb nevus syndrome is a rare disorder characterized by multiple vascular malformations involving the skin and GI tract. Rarely, the brain, liver, lung and skeletal muscles may also be involved. BRBNS has the potential for serious or fatal bleeding. Although frequently sporadic, autosomal dominant inheritance caused by mutation of chromosome 9p has also been described.

About 200 cases of BRBNS have been reported in the English literature. Indian case reports are few in number and most of them have been from North India. There has been only one published report from South India.

The skin lesions of BRBNS mainly appear in the trunk and upper extremities and these are characterized by their small size, bluish color, softness, absence of pain and the tendency to refill with blood after compression. The gastrointestinal lesions of BRBNS are usually distributed throughout the gastrointestinal tract, mostly in the small bowel and distal colon. Capsule endoscopy or double balloon enteroscopy is ideal for small bowel evaluation. The vascular malformations in BRBNS include telangiectasia, capillary hemangioma, cavernous hemangioma, venous angiomia and on rare occasion arteriovenous fistula.

The cases reported from India are all sporadic and mostly in males. The common presentation was anemia secondary to GI bleed. Haemopericardium has been reported in BRBNS. The single case report from South India described associated skeletal deformities in addition to cutaneous and GI vascular malformations.

Other diseases associated with vascular malformations involving the skin and GI tract include Mafucci’s syndrome and Rendu-Osler-Weber syndrome. Patients with Mafucci’s syndrome also have dyschondria, while Rendu-Osler-Weber syndrome often present with recurrent episodes of epistaxis, pathologic findings of telangiectasia and is inherited in an autosomal dominant pattern.

Several therapeutic modalities have been attempted to date for treating BRBNS. Medical treatment consists of iron supplementation for anemia, steroids, interferon α-2a and octreotide to reduce the frequency and severity of bleeding episodes. However, in most cases disease recur after discontinuation of these treatments. New therapeutic modalities for treating the GI lesions have recently been attempted such as endoscopic laser photocoagulation, sclerosis, band ligation and polypectomy. In the event of life-threatening hemorrhage, excision of the gastrointestinal lesions or segmental resection of the involved GI tract can be performed. Recurrence resulting in rebleeding often occurs. In a recent study performed by Fishman on ten patients with BRBNS, complete GI endoscopy was conducted followed by removal of all the gastrointestinal lesions by means of wedge resection, endoscopic polypectomy, suture-ligation, segmental bowel resection and band ligation (surgical eradication). On function tests. The patient underwent upper GI endoscopy and total colonoscopy to ascertain the cause of anemia. The upper GI endoscopy revealed multiple bluish-black nevi of varying sizes seen in esophageal and gastric mucosa (Figure 2). Colonoscopy showed similar lesions throughout the colon. However, no active bleeding was observed from any of these lesions. Capsule endoscopy was planned for small bowel evaluation, but patient refused. Skeletal system examination was normal. The chest X-ray and abdominal computed tomography were unremarkable. She refused to undergo a biopsy from the skin lesions. She has been receiving oral iron supplementation and the haemoglobin after 3 months follow up was 10.8 g%.
follow up for a mean of 5 years, GI bleeding recurred in only one patient who had received less extensive procedures.

Conclusion

BRBNS is a rare but important syndrome, because of its propensity for serious and fatal bleeding. The morbidity and mortality depends on the organ involved, particularly GI bleed. A complete history and physical evaluation is a must for a successful diagnosis to help in treatment planning and to prevent fatal complications. Treatment is symptomatic and conservative, except in the situation of life-threatening hemorrhage, where surgery is the option. The patient should be counselled about the chronicity of this condition and the need for regular follow-up.

References

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