BLUE RUBBER BLEB NEVUS SYNDROME- A RARE CASE

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INTRODUCTION

Blue Rubber Bleb Nevus Syndrome (BBRNS) is a rare angiomatosis characterized by distinctive cutaneous and gastrointestinal venous malformations that result in gastrointestinal hemorrhage and iron deficiency anemia secondary to bleeding episodes. The causes of this syndrome are unknown. Its most common presentation is in the form of sporadic cases but dominant autosomal inheritance has been also described specifically with a locus found on chromosome 9p.

Approximately 250 cases have been reported in the literature to date. The syndrome affects both males and females with equal frequency. Blue rubber bleb nevus syndrome (BRBNS or Bean's syndrome) was first recognized by Gascoyen in 1860. [1] Hundred years later Bean described BRBNS in detail and coined the term Blue rubber bleb nevus syndrome. [2] The most common symptoms are gastrointestinal bleeding and secondary iron deficiency anemia. The syndrome may also present with severe complications such as rupture, intestinal torsion, and intussusception, and can even cause death. Cutaneous malformations are usually asymptomatic and do not require treatment. The treatment of gastrointestinal lesions is determined by the extent of intestinal involvement and the severity of the disease.

CASE REPORT

A 22-year-old female came to the accident and emergency with complaints of increased fatigability and shortness of breath for 1 month. It was associated with episodes of nausea and vomiting for the last two days. The vomiting was sudden in onset with coffee ground color. She denied having melena, hematochezia, menorrhagia, dyspnea, and stomach ache.

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There was no history of NSAID intake, peptic ulcer, and chronic liver disease. On examination, the patient had an extremely pale face and pale conjunctiva with multiple bluish swellings on her hands and feet. After initial resuscitation in A and E, baseline investigations were done. Her blood reports showed Hb= 2.2g/dl (MCV: 60; MCH: 24; HCT: 26; TLC: 13,000/cmm; Platelets: 2,27,000/cmm. Detailed examination and history taking revealed that the patient had been suffering from the presenting symptoms since the early age of 4 years. She was born as a result of consanguineous marriage with the rest of her 5 siblings disease free. However, she had episodes of epistaxis and hematemesis since early childhood. She had been admitted multiple times for blood transfusions at an early age.

On examination, the patient had a pale face with pale conjunctiva and a bluish swelling on the lower lip. The swelling was about 1x1cm, tender with a rubbery consistency. The edges were well-demarcated with no ulceration.

The same kinds of lesions were present all over the body with a higher concentration on all four limbs and the lower abdomen (Fig:01, 02). Further workup revealed: S. ferritin: 4.2ng/ml; Peripheral smear: Hypochromic microcytic anemia; Hb electrophoresis was normal, Stool for occult blood was positive, and a Bone marrow biopsy revealed absent iron stores.

Invasive testing as of endoscopy revealed vascular hemangioma-like lesions seen in the Esophagus, the whole of the stomach involved with multiple vascular hemangioma lesions extending to the duodenum (FIG:03). Colonoscopy revealed several hemangioma lesions seen in the colon up to the splenic flexure. Histologic staining of skin biopsy showed Angiokeratoma. Colonic biopsy revealed acute self-limited colitis with Angiokeratoma circumscription. Hence the diagnosis of Blue Rubber Bleb Nevus Syndrome was made and the patient was treated by a multidisciplinary team involving a hematologist, gastroenterologist, endocrinologist, and dermatologist. As the patient had no dyspnea, dysphagia, or blurred vision, she was given iron supplementation and blood transfusions. Two weeks later, a routine blood test showed that the he-

moglobin was 7.7 g/dL. A fecal occult blood test was negative. She was discharged and asked to attend outpatient follow-up monthly.

DISCUSSION

BBRNS also known as Bean syndrome belongs to a group of disorders "BENGAL". Rare congenital vascular anomaly in which malformed veins or blebs appear on the skin and surfaces of internal organs. Patients present with multiple venous malformations in various organ systems including the liver, spleen, heart, eye, and central nervous system. It is usually a sporadic disorder; however, autosomal dominant modes of inheritance are also reported, specifically with a locus found on chromosome 9p. [3] The clinical manifestations vary according to the different organ involvement. The cutaneous lesions are asymptomatic but some of the patients complain of painful lesions (5%). [4] The condition presents at birth or in early childhood with multiple blue to violaceous soft compressible nodules on the skin or mucous membranes. They are often born with a "dominant" lesion and develop numerous venous malformations over their lifetime which are rubbery in consistency and may be painful or tender when compressed. Pain is especially prevalent at night time. They range in size from only a few millimeters in diameter to up to 4 to 5 cm in diameter. They can increase in size with time, and more lesions may develop in the skin or gastrointestinal tract. Large blue marks and disfiguring may appear as well. Uniquely, lesions tend to swell in gravity-dependent positions, and patients have focal areas of hyperhidrosis overlying these lesions. Venous malformations may be located in the heart, spleen, liver, central nervous system, and gastrointestinal tract. The small bowel is the most common site of gastrointestinal involvement. The patient may present with severe iron deficiency anemia from recurrent intestinal hemorrhages. Rare complications of BRBNS have been reported such as blood coagulation disturbance (four cases), thrombocytopenia (three cases), and disseminated intravascular coagulopathy (two cases) and the reasons for these complications are unclear.

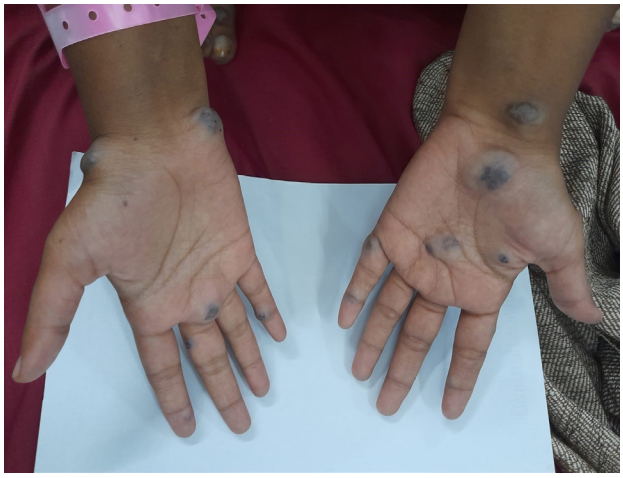

Fig 1: BBRNS lesions on hands

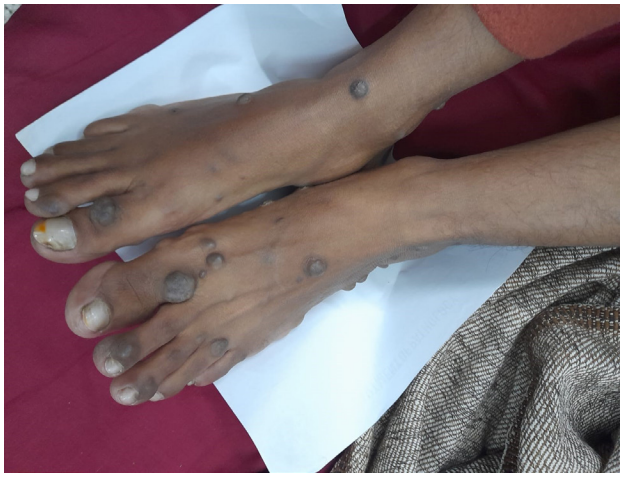

Fig 2: BBRNS lesions on feet

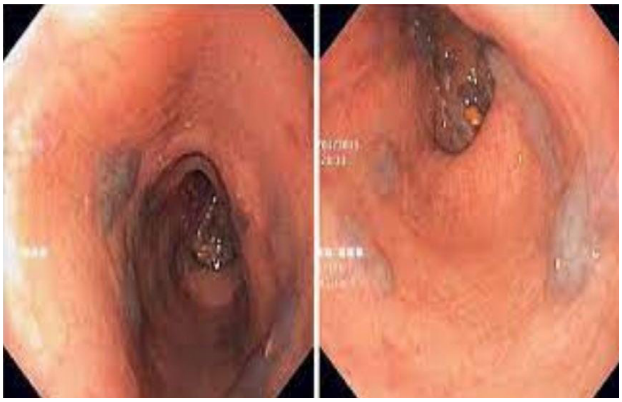

Fig 3: Vascular malformations in the Esophagus on endoscopy

The diagnosis of BRBNS is based on the presence of characteristic cutaneous lesions with or without GI bleeding and/or the involvement of other organs. [5]

For GI lesions, a push endoscopic examination is the most important diagnostic method and mucosal resection, Argon plasma coagulation, Laser photocoagulation, Sclerotherapy, or Ban d ligation are often necessary. [6] Initial diagnostic study of choice include: a) Endoscopic ultrasound: gastrointestinal venous malformation b) MRI with intravenous contrast, arterial and venography c) CT, barium studies, and skin biopsy. Blue rubber bleb nevi can be examined under Dermoscope with features of superficial, light red arborizing veins, maculae with undefined borders on the palms and soles, and blue-purple nodules with lacunae divided by white linear structures. The treatment is largely symptomatic. The most important step is the evaluation of gastrointestinal lesions and preventing severe bleeding and Iron replacement or blood transfusion for anemia. Vascular malformations require endoscopic sclerotherapy, band ligation, or laser photocoagulation. Somatostatin analog-subcutaneous Octreotide can be used to decrease splanchnic blood flow in patients

with gastrointestinal hemorrhages. Corticosteroids, Interferon-alpha, IVIG, and vincristine are also modalities of choice. The most concerning complications of blue rubber bleb nevus syndrome are severe gastrointestinal hemorrhages or intestinal intussusception. As a result, patients often require multiple blood transfusions throughout their lifetime.

CONCLUSION

It is important to counsel patients and parents about the risk of severe gastrointestinal bleeding and intussusception in blue rubber bleb nevus syndrome. There should be a low threshold for patients to present for medical evaluation if they have abnormal bowel function or abdominal pain. Patients with blue rubber bleb nevus syndrome will require a multispecialty approach to management. They require an interprofessional team approach with Hematology, dermatology, gastroenterology, surgery, nurse practitioners, and other specialties caring for the patient. There is no cure for the disorder and treatment is supportive. Patients do need to be monitored as they are prone to potential complications such as volvulus, intussusception, infarction, and gastrointestinal bleeding.

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