CASE REPORT

Multimodality Treatment for Blue Rubber Bleb Nevus Syndrome in a Child: A Case Report

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Abstract

Aim and background: Blue rubber bleb nevus syndrome (BRBNS) is a rare congenital venous malformation with predilection for the skin and gastrointestinal (GI) tract. We report a child who presented with symptomatic anemia and received multimodality treatment, including propranolol, endoscopic band ligation (EBL), and sirolimus.

Case description: An 8-year-old girl presented with symptomatic anemia requiring multiple blood transfusions since 4 years of age. She was diagnosed with BRBNS at 5 years of age and had a gastric nevus and multiple colonic nevi. Initial treatment was done with EBL and nonselective β-blockers, but sirolimus was added a year later due to partial response to β-blockade. The child responded well to the treatment.

Conclusion and clinical significance: A single form of treatment may not be enough to control the anemia in BRBNS, and this case adds to the scanty Indian literature on the entity.

Keywords: Blue rubber bleb nevus syndrome, Case report, Endoscopic band ligation, Propranolol, Sirolimus.

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Introduction

Blue rubber bleb nevus syndrome (BRBNS) is a rare congenital venous malformation with predilection for the skin and gastrointestinal (GI) tract.[1] Apart from visible skin lesions, patients often present with symptomatic anemia, along with occult or overt GI bleeding.[2] Apart from the treatment of anemia, treatments include endoscopic band ligation (EBL) of the GI lesions, nonselective β-blockers (propranolol), and sirolimus.[2] We report a child with BRBNS with occult GI bleeding who received multimodality treatment in the form of EBL and propranolol (with partial response), and subsequent addition of sirolimus with good response.

Case Description

A 5-year-old girl presented to us in 2021 with gradually progressive paleness of the body and fatigue for the last 1 year, requiring blood transfusions. There was no family history of recurrent skin lesions or GI bleeding. Laboratory investigations at admission showed a picture of iron deficiency anemia, with other system parameters being normal, including a normal hemoglobin variant analysis. Diagnostic upper GI endoscopy showed a bluish polypoidal lesion (10 mm diameter) at the midbody of the stomach. Colonoscopy showed similar lesions at multiple locations (sigmoid, transverse, ascending colon, and cecum), each of size 5 mm × 5 mm. EBL was done for the lesions, which were endoscopically accessible, and the child was started on propranolol. In spite of adequate iron supplementation and titrating the propranolol dose up to 3 mg/kg/day, the child continued to remain anemic and required further blood transfusions and EBL sessions over the next year. Subsequently, sirolimus was added at the age of 6 years at a dose of 1 mg/day (in two divided doses). After this, the requirement of blood transfusions started decreasing (twice over the last 2 years). Her last presentation was at the age of 8 years with symptomatic anemia requiring blood transfusion. Upper GI endoscopy was normal, and colonoscopy showed multiple nonbleeding colonic nevi. Her sirolimus dose was increased to 1.5 mg/day. At outpatient follow-up a month later, the patient was asymptomatic with a stable hemoglobin level.

Discussion

Blue rubber bleb nevus syndrome has been known since the mid-19th century,[3] though the entity was vividly illustrated a century later by Bean, after whom it is also called "Bean syndrome."[4] As mentioned, GI manifestations are generally overt or occult bleeding causing anemia.[4] GI lesions of BRBNS may be present anywhere in the GI tract, though most commonly they are found in the small intestine and colon. We could not perform wireless capsule enteroscopy due to logistic issues. Less

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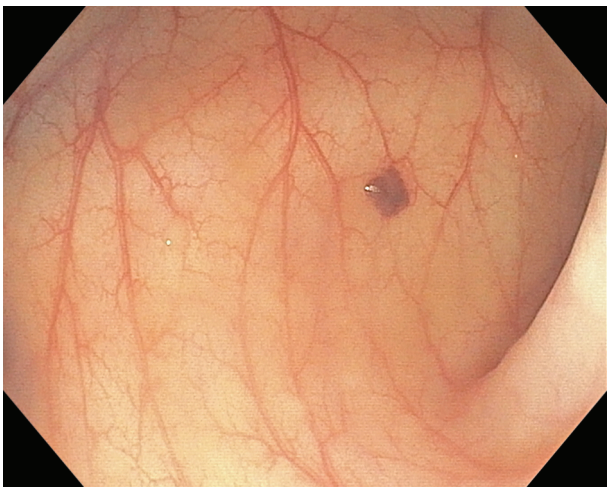

Fig. 1: Colonoscopic image of a nonbleeding nevus

commonly, patients may have severe abdominal pain due to intussusception or other related causes of luminal obstruction. In such cases, cross-sectional imaging is necessary to ascertain the depth of the lesions. Since our patient did not have abdominal pain, we did not perform the same. Therapeutic modalities utilized have included drug therapy with steroids, propranolol, interferon, and sirolimus.[5] The common approach is to try β-blockade first (which acts due to its antiangiogenic effects), and subsequently shift to sirolimus in case of poor response to propranolol. Sirolimus acts by inhibiting the mammalian target of rapamycin (mTOR) protein complex, which is involved in cellular proliferation.[6] For endoscopically accessible lesions, EBL has been found useful.[2] Our patient was treated on similar lines, using EBL and β-blockade initially. Due to poor response to β-blockade, we instituted sirolimus, to which the response was found to be satisfactory (Fig. 1).

Conclusion and Clinical Significance

Our patient reiterates the logical approach to the management of symptomatic BRBNS patients with EBL, propranolol, and the addition of sirolimus in cases of poor response. Sirolimus is a promising agent in the treatment of this entity.

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