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Case report

Blue Rubber Bleb Nevus Syndrome: A case report

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INTRODUCTION

Blue Rubber Bleb Nevus Syndrome (BRBNS) is a rare disease associated with multiple venous malformations in the skin, gastrointestinal tract and other internal organs. Usually it presents with extensive haemorrhages (hematemesis, melena, rectal bleeding), or iron deficiency anemia (due to occult bleeding).

The cause of this syndrome is unknown. Common presentation is sporadic, although a dominant autosomal inheritance has been described.

It was first described by Gascoyen in 1860. In 1958 William Bennet Been described another similar condition with vascular nevi of the skin and gastrointestinal tract and hemorrhage. He named this syndrome due to the bluish color of the nevi and to their rubbery consistency at palpation. Since then aproximatinely 200 cases have been reported with no malignant transformation.

The skin and the GI tract are most frequently involved. However, case reports have demonstrated that central nervous system, thyroid, parotid, eyes, oral cavity, musculoskeletal system, lungs, kidney, liver, spleen and bladder may be affected.

Typical findings are bluish lesions ranging from few millimeters to several centimeters in diameter. The skin lesions are principally located in the trunk and upper extremities and are often apparent at birth or early childhood. Later onset of the syndrome has been reported. In the GI tract it may occur anywhere from mouth to anus, but more frequently it is found in the small bowel. In contrast to the skin lesions, the GI lesions often bleed.

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Symptoms vary depending on the system involved. Patients may report fatigue, hematemesis, melena or rectal bleeding. When bones are involved, patients may complain for joint pain. Pathologic fractures have also been reported. When kidneys are involved hematuria may be the main symptom.

CASE REPORT

We present a case of a 62 year old man admitted for melena and severe anemia. He had a previous episode of melaena 10 years ago, requiring blood transfusion. Upper and lower endoscopy were negative at that time.

On admission, physical examination showed paleness and tachycardia. Lab tests showed Ht 29 mg% with normal WBC, PLT. Peripheral blood smear showed microcytosis and hypochromia. Coagulation studies and routine biochemistry values were normal.

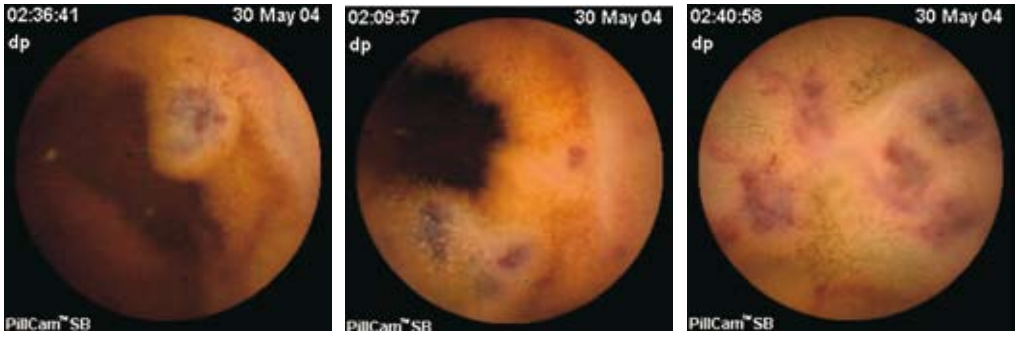
Gastroscopy showed multiple venous malformations in the oesophagus. Colonoscopy showed diverticulae as well as two small sigmoid polyps excised and hemorrhoids. Small bowel capsule endoscopy showed multiple venous malformations of different size from the Treitz ligament up to the terminal ileum. No signs of active bleeding were seen

Treatment was conservative with PPIs, blood transfusions and iron replacement.

DISCUSSION

BRBNS is a rare syndrome characterized by *multiple venous malformations* in the skin, gastroidestinal tract and other internal organs. These lesions usually appear at early childhood and increase in number and size with age. When the disease is suspected gastrointestinal endoscopy is recommended for detecting the extent of the lesions.

MRI, Computerized tomography and angiography



Images of small bowel venous malformations in a patient with BRBNS.

have been used as diagnostic tools.

The treatment of GI venous malformations depends on their number, location, size and symptoms. In our case the lesions were present in esophagous and small bowel. A conservative approach is usually recommended when the bleeding episodes are mild. When the bleeding is significant and lesions are localized in one segment of the GI tract, resection of the involved segment is recommended. However, surgical treatment does not exclude recurrence of the lesions in the remaining part of the GI tract.

GI lesions can also be treated endoscopically with sclerotherapy, band ligation for the pedunculated lesions and Argon plasma coagulation. In some patients a pharmacolocic treatment has been tried with corticosteroids, interferon and vincristine with good response, although lesions returned to their previous levels soon after the treatment was discontinued. Long term benefits of pharmacotherapy are still controversial.

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