BLUE RUBBER BLEB NEVUS SYNDROME: A CASE REPORT

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ABSTRACT

Blue Rubber Bleb Nevus Syndrome (BRBNS) is a syndrome that is characterized with cutaneous venous malformations besides visceral involvement. Visceral involvement affects mostly the gastrointestinal (GI) system. As these venous malformations can lead to life-threatening serious GI bleeding, anemia is an important clue to diagnosis. A 18-year-old female presented to the department of internal medicine with complaints of weakness and paleness. Hypochromic microcytic anemia was diagnosed and upper and lower GI screenings were done. On endoscopy, reddish-purple polypoid lesions were detected throughout the GI tract and nodular lesions compatible with venous malformations were observed on both lower extremities. A diagnosis of BRBNS was made. Symptoms of iron deficiency anemia guide patients with BRBNS to general practitioners and internal medicine specialists. If cutaneous findings are present, collaboration with dermatologists at this stage will improve diagnostic accuracy.

Key words: Anemia, cutaneous hemangioma, gastrointestinal tract.

Introduction

Blue Rubber Blebs Nevus Syndrome (BRBNS) is a syndrome characterized by vascular tumors of the gastrointestinal (GI) system and cutaneous hemangiomas. Visceral involvement mostly affects the GI system and particularly the small intestine. The main significance of this syndrome is that these venous malformations can lead to life-threatening serious GI bleeding and anemia. Although it can affect multiple visceral organs, cutaneous findings and GI symptoms as it emerges particularly with cutaneous involvement and GI bleeding symptoms, internal medicine specialists and general practitioners must be aware of patients with symptoms of anemia and work in collaboration with gastroenterologists and dermatologists.

Case

A 18-year-old female patient was seen at the internal medicine clinic with complaints of fatigue and weakness. Upon examination of the patient; conjunctival pallor, purple lesions on the oral mucosa and the anterior right leg and at plantar surface of the left foot were detected. Dermatological consultation was sought from our department. Upon dermatologic examination, nodular lesion of 0.5 cm diameter, compatible with venous malformation, were observed on the upper lip mucosa (Figure 1).

Figure 1: Nodular lesion in the upper lip mucosa.
Similar lesions of 0.8 cm diameter were present on the right anterior leg and 0.5 cm diameter on the sole of the left foot. On dermatoscopy of the lesions, homogenous steel blue appearance and lacunar patterns were present. Iron deficiency anemia was diagnosed by routine laboratory tests (haemoglobin: 6.74 g/dL, ferritin: 8 ug/dL) and peripheral blood smear (hypochromic microcytic anemia). The platelet count, the reticulocytes, liver function tests, coagulation factors and albumin levels were normal. Serologic investigation for hepatitis B and C viruses and human immunodeficiency virus were all negative. On upper GI endoscopy that was performed to investigate the etiology of anemia; purple vascular lesions that are raised from the mucosa - two at the corpus of stomach measuring 0.5 cm and one in the duodenum (Figure 2) measuring 0.6 cm in diameter were detected. On colonoscopy, similar lesions - four in the transverse colon and one in the rectum - were observed. A diagnosis of BRBNS was made. No family history was present. Permission was obtained from the patient for publication of the case. The patient was informed of possible complications and was referred for endoscopic band ligation.

**Figure 2:** Purplish nodules in the duodenum.

**Discussion**

BRBNS, which is also known as Bean Syndrome, is characterized by multiple venous malformations that can affect the skin, GI tract and other viseral organs. BRBNS is a quite rare syndrome and number of reported cases in the literature are limited. These venous malformations are generally present at birth but can also commence during childhood. Their count and size can increase with increasing age. BRBNS is generally diagnosed with symptoms of abdominal colic, melena, fatigue and abundant GI bleeding in children or by iron deficiency anemia in adults. The clinical appearance of the cutaneous lesions range from small blue-black punctate papules to large disfiguring vascular tumors.

The nipple-like lesion, which is the most classic cutaneous lesion seen in BRBNS, rapidly compresses and refills slowly.

Gastrointestinal complications may lead to significant complications such as hemorrhage, intussusception, volvulus and infarction.

The most common clinical presentation of BRBNS is GI hemorrhage and is one of the rarer reasons underlying anemia. GI venous malformations are present commonly in the small intestine but may be found anywhere between the oral mucosa and anal mucosa. Oral lesions can be seen in 59-64% of cases. On oral mucosal examination of our patient, a nodular lesion was detected. Venous malformations of the small intestines are commonly the reason for chronic anemia; however they rarely can be related to acute and massive hemorrhage. Our patient presented with chronic iron deficiency anemia, the most common form. Until then, she had never seen a physician for her cutaneous lesions.

The central nervous system, liver, kidney, bladder, heart, thyroid, and spleen may be affected. Unlike cutaneous lesions, intestinal angiomas can bleed easily even with light touch. Therefore we did not perform a biopsy. For the diagnosis of the syndrome, most common diagnostic tools are upper GI endoscopy, colonoscopy and magnetic resonance imaging.

Rendu Osler syndrome may also be included in the differential diagnosis of the disease. Also known as hereditary hemorrhagic telengiectasia, Rendu Osler syndrome is a vascular autosomal dominant disease, leading to dysfunction in development of arteriovenous capillaries. This results in epistaxis, gastrointestinal bleeding and iron deficiency anemia. Our patient had no history of epistaxis, mucocutaneous and visceral telengiectasias and thus was differentiated from Rendu Osler syndrome.

The treatment of the disease is related with the severity of the clinical presentation and the amount of GI involvement. Supportive long term iron replacement treatment with accompanying blood transfusion if needed is the treatment of choice. Ferrous sulphate was administered to our patient and endoscopic band ligation was planned for her GI lesions.

In conclusion, although BRBNS is a very rare cause of iron deficiency anemia, we would like to emphasize the importance of a systemic approach and detailed dermatological examination during the evaluation of patients with symptoms of anemia.
References


