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Case Report

Blue Rubber Bleb Nevus Syndrome, an Unusual Cause of Upper Gastro Intestinal Bleeding: A Case Report

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Abstract

Blue rubber bleb nevus syndrome or bean's syndrome, is a rare congenital disorder characterized by the presence of vascular malformations in the skin and other internal organs with a predilection of the gastro intestinal tract. The most common clinical manifestations are gastrointestinal bleeding causing severe iron deficiency anemia, and bluish-purple soft compressible nodules on skin or mucous membranes usually present at birth or during early childhood. We presented a clinical case of upper gastrointestinal bleeding revealing a blue rubber bleb nevus syndrome treated symptomatically with good evolution.

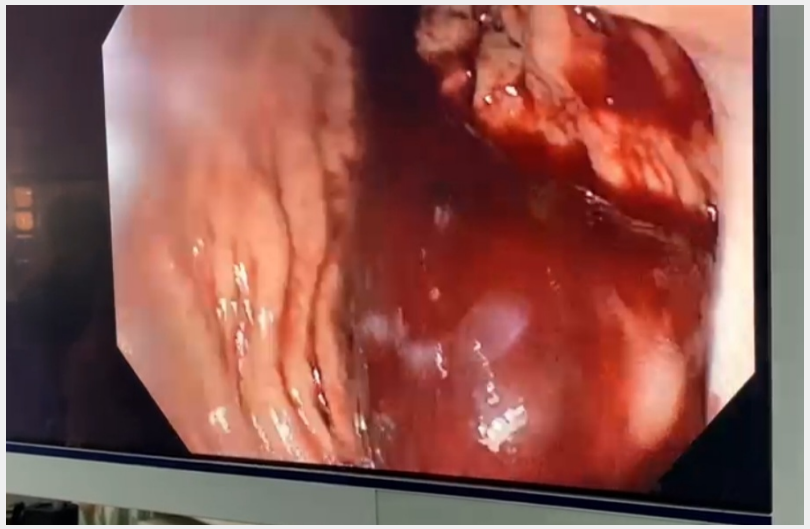
Introduction

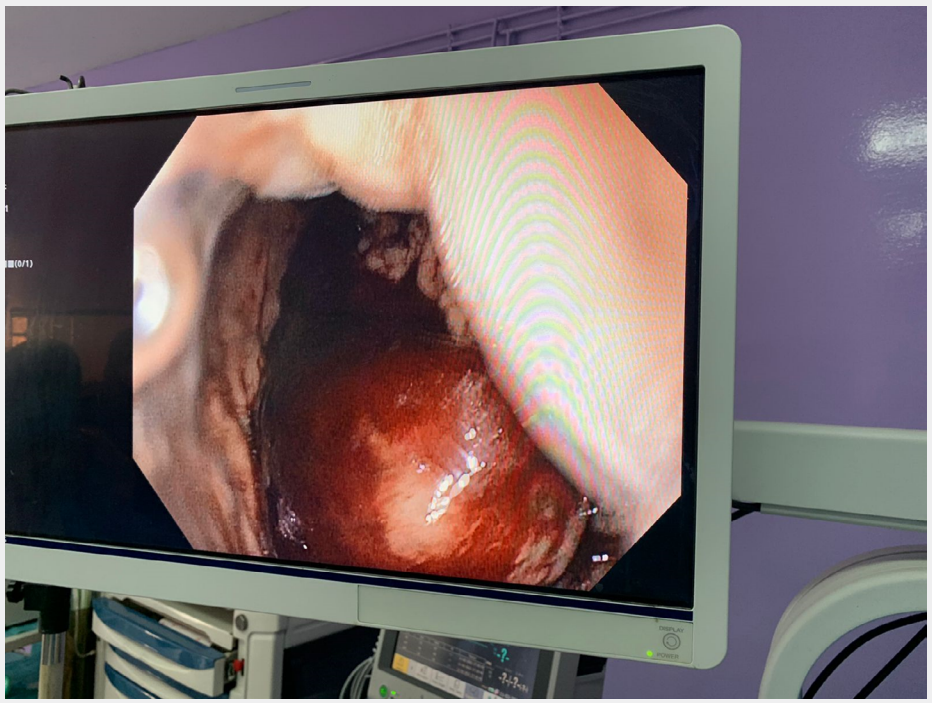
Blue rubber bleb nevus syndrome or bean's syndrome, is a rare congenital disorder characterized by the presence of vascular malformations in the skin and other internal organs with a predilection of the gastro intestinal tract [1]. It was first reported by Gascoyen and 100 years later William Bean described it in details and giving it a name according to the description of skin lesions: bluish color and rubber consistency at palpation [2]. Up to now, around 350 cases have been reported world-wide [3]. The most common clinical manifestations are gastrointestinal bleeding causing severe iron deficiency anemia, and bluish-purple soft

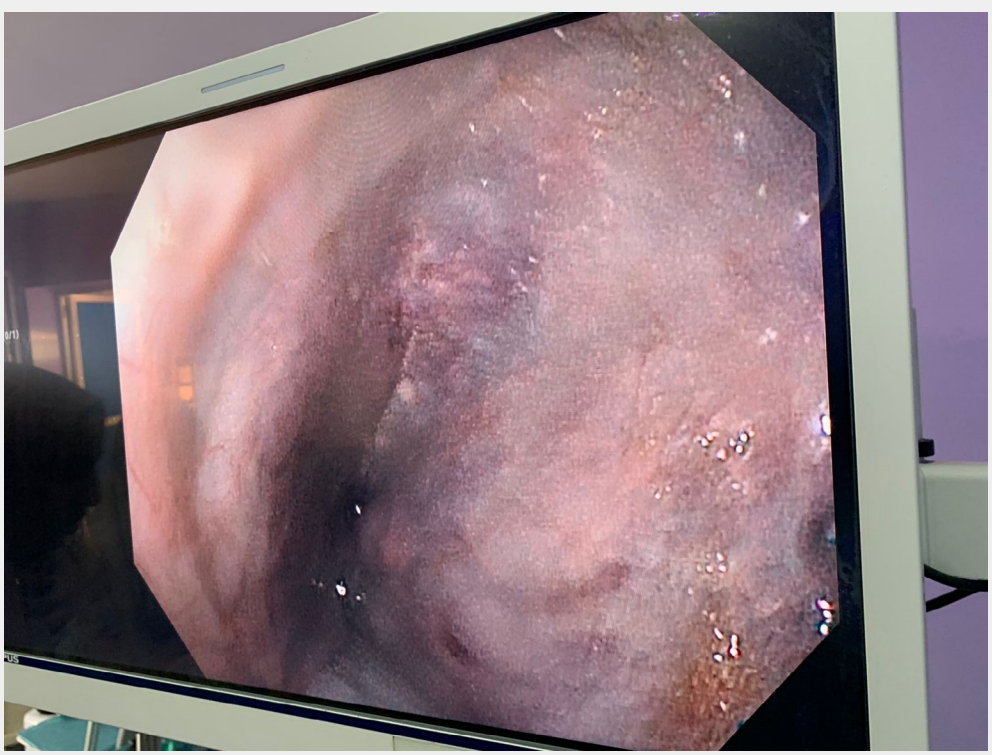
compressible nodules on skin or mucous membranes usually present at birth or during early childhood. Complications as intussusception of the bowel, bowel infarction are extremely rare [4]. We presented a clinical case of upper gastrointestinal bleeding revealing a bleu rubber bleb nevus syndrome.

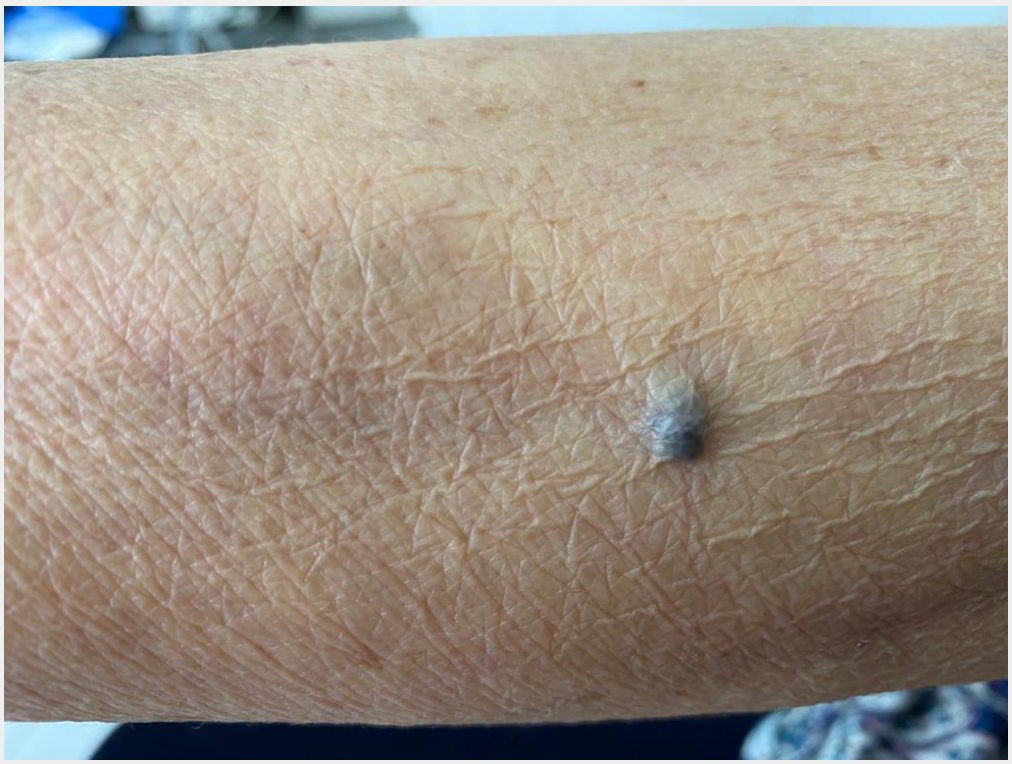
Case Report

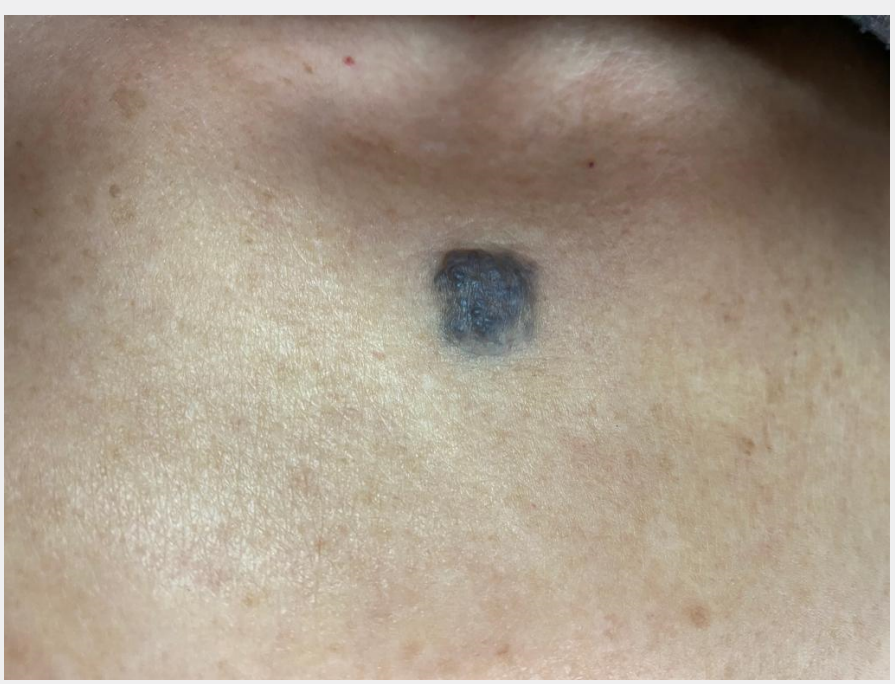
A 73 years old woman was admitted in 2012, because of hematochezia and melena in the Gastroenterology department where she was hospitalized for two months. The oesogastroduodenoscopy revealed a large vascular lesion in the distal part of the esophagus and the fundus. Colonoscopy showed no remarkable signs of venous malformations or bleeding signs. CT scan of the body revealed the presence of an esophageal large mass extended to the lesser curvature of the stomach with multiple micro calcifications. The patient underwent abdominal laparoscopy, that revealed the presence of multiple venous lesions in the greater omentum and the liver. Histology results confirm the diagnosis of hemangiomas. The patient was discharged after transfusion with a hemoglobin level of 9.2g/dl. She suffered from chronic anemia treated by iron supplementation. In 2021, the patient consulted in the emergency department because of a 3 months' history of melena complicated by 2 episodes of hematochezia, with fatigue, pallor and severe anemia with a hemoglobin level of 5g/dl. Physical examination revealed small bluish nodules on the trunk, the lower limb and the nose that have been present from birth. The abdomen was soft and no tender. Complete blood count revealed a hypochromic microcytic severe anemia with a hemoglobin level of 5.7g/dl. Oesogastroduodenoscopy revealed a large vascular mass in the lower third of the esophagus, with other vascular malformations in the fundus with active bleeding. The results of CT scan of the body were similar to the previous one done in 2012. Cerebral MRI showed no remarkable lesion. The diagnosis of blue rubber bleb nevus syndrome was established based on medical history, cutaneous lesions and the pathognomonic endoscopic lesion. The patient was treated by proton pump inhibitor and blood transfusion, with a regular follow up. The patient presents no other episodes of GI bleeding during the hospital stay and was discharged with a hemoglobin level of 10g/dl.











Discussion

Blue rubber bleb nevus syndrome can occur in any age with no sex predilection, the estimated incidence is very low and is usually a sporadic disorder caused by a double mutation in TEK gene which encodes the TIE2(endothelial cell tyrosine kinase receptor for angiopoietin in humans) [5]. In the present case, the gastro intestinal bleeding was the main symptom revealing the disease. According to a single center study in China [3], the GI lesions were a major cause of morbidity and were found in 77.2% of patients, affecting any part of the GI tract from the esophagus to the anus, they are identified as vascular lesions that ranges from 1 to 10cm complicated in most of cases with GI bleeding and iron deficiency anemia. Sometimes, they became larger and can lead to intussusception, volvulus or infarction [6]. Soblet *et al* establish the diagnostic criteria for BRBNS:1) The presence of GI lesions, 2) the presence at birth of one 'dominant' visible lesion in combination with 10 or more small cutaneous lesions, 3) hyperkeratotic, palmoplantar lesions [5]. The cutaneous lesions in BRBNS mainly appear in the trunk and upper extremities and are characterized by their small size, softness, absence of pain and bleeding and tendency of refill with blood after compression [7]. The skin lesions in our patient had the same characteristic. The medical literature on treatment is limited to case reports. Since cutaneous lesions rarely bleed, the different therapeutic strategies were proposed for the GI bleeding. In most patients, the treatment was symptomatic and consisted of lifelong iron replacement and blood transfusion [8]. Several pharmacologic agents have been tried like antiangiogenic agents such as interferon alpha and corticosteroids. Octreotide has also been used in the hemorrhage phase to control the bleeding [9]. The last treatment that has been proposed is low dose Sirolimus which has antiangiogenic properties [10]. Endoscopic treatment as band ligation and sclerotherapy has been used but are associated with a high risk of perforation [11]. Surgery can be an option especially in complicated cases [12]. In our case, the therapeutic option was symptomatic treatment by iron supplementation, blood transfusion and close follow-up.

Conclusion

Rubber bleb nevus syndrome is a rare syndrome where the diagnosis is based on clinical history, characteristic physical appearance of the cutaneous lesions, and the pathognomonic endoscopic appearance of the gastrointestinal lesions. The therapeutic management remains not codified. Low dose of sirolimus was used in many cases. For widespread lesions, conservative treatment such as blood transfusion and iron supplementation is usually adopted which was the case in our patient.

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