Case Report

Blue rubber bleb nevus syndrome: Prominent oral findings

G. N. Suma, S. M. Ravi Prakash, Dayashankara Rao, Sumit Goel

Department of Oral Medicine and Radiology, Oral and Maxillofacial Surgery, Kothiwal Dental College and Research Centre, Mora Mustaqueem, Moradabad (U.P.), India

Address for correspondence:
Dr. G.N. Suma,
C/O Dr R.P. Singh,
Dhanwantri Nursing Home,
Sarai Khalsa, Behind Head Post Office, Moradabad (U.P.) 244 001, India. E-mail: suma-gndaya@yahoo.com

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ABSTRACT

Blue rubber bleb nevus syndrome is a rare disorder characterized by multiple cutaneous venous malformations in association with visceral lesions, most commonly affecting the gastrointestinal tract. Oral cavity lesions occur in 59 to 64% of cases. We report a unique presentation of this syndrome in a 25-year-old male patient with prominent oral findings. This is a sporadic case, started during early childhood, progressively increasing in number and size. Oral vascular lesions were part of gastrointestinal involvement. Associated cardiac abnormalities were also observed. An early diagnosis of this syndrome is required as it gets complicated with bleeding, anemia and other systemic complications.

Key words: Blue rubber bleb nevus syndrome, venous malformations, oral cavity, hemorrhage

INTRODUCTION

Blue rubber bleb nevus syndrome (BRBNS) describes a clinical entity that presents with cutaneous and visceral venous malformations, especially in the gastrointestinal tract.[1] In 1860, Gascoyen first described an association between cavernous hemangiomas of the skin and similar lesions in the gastrointestinal (GI) tract. In 1958, Bean further described these lesions and coined the term blue rubber bleb nevus syndrome (also called as Bean syndrome). BRBNS is an important syndrome because of its potential for serious or fatal bleeding.[1,2] Cutaneous lesions are often apparent at birth or manifest in early childhood characterized by multiple, protruberant, dark blue, compressible blebs which progress in size and number with advancing age.[1,2] GI venous malformations may occur anywhere from oral to anal mucosa but predominantly occur in the small bowel.[1,3] This case report discusses a unique presentation of this syndrome in a 25-year-old male patient with prominent findings in the oral cavity.

CASE REPORT

A 25-year-old male patient presented with a complaint of difficulty in mastication due to multiple swellings on his tongue since childhood. He also reported a history of similar lesions on face, neck and upper limbs. These lesions first appeared on the right hand during his childhood and the lesions increased in number and size with age. History of spontaneous bleeding from the lesions was absent. He denied dyspnea, dysphagia or weight loss. His medical and family history was unremarkable except for the presence of gastrointestinal bleeding (melena).

At physical examination, patient was pale and tachycardiac with asymmetry of face due to a swelling on the left cheek [Figure 1]. General examination revealed multiple, dark, sessile, bluish, smooth surfaced blebs distributed on neck and upper limbs ranging in size from 2 to 5 cms. Overlying skin was smooth with hyperhidrosis at the lesion sites. Size of the lesion increased on aligning them with gravity. Lesions were non tender, soft, rubbery and compressible with a

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feeling of cutaneous herniation, refilled after removal of digital pressure [Figures 2 and 3].

The swelling on the left cheek was diffuse, 5*4 cm in size extending from lower border of the orbit to corner of the mouth. Skin surface was normal in color and texture. On palpation, swelling was non pulsatile and non tender. Intraorally, the swelling obliterated the right upper buccal vestibule. Increase in the size of the swelling was observed on bending the head.

Intraorally, multiple bluish vascular swellings involving left buccal mucosa, right labial mucosa [Figure 4], dorsum and ventrolateral aspect of tongue were present [Figure 5]. On palpation the lesions were soft, compressible and non tender. On the basis of history and clinical examination, patient was provisionally diagnosed as a case of blue rubber bleb nevus syndrome. Differential diagnosis included Maffuci syndrome, Osler Weber Rendu syndrome and Multiple Glomangiomatosis.

Ultrasonography of tongue lesions showed compressible cystic septate masses with low velocity color flow detected on both color Doppler and power Doppler suggestive of hemangiomatous lesions [Figure 6]. Masses on the the neck, cheek and upper phalanges showed similar findings on ultrasonography.

Laboratory investigations of the patient pointed to anemia. Radiographic examination revealed no abnormalities in the bones and joints. ECG of the patient showed right ventricular hypertrophy. Upper GI endoscopy showed no lesions in bowel. Stools showed occult blood. The patient refused to undergo further investigations and histopathological examination.

A diagnostic search for a clinical condition presenting with vascular lesions of skin and oral mucosa, iron deficiency anemia, history of melena, ventricular hypertrophy and low flow lesions on ultrasound confirmed the case to be BRBNS.

Patient is under long term oral iron supplements. He is advised sclerotherapy and carbon dioxide LASER treatment.

DISCUSSION

BRBNS is a rare disorder with only approximately 200 cases reported in the world's literature.[1] Although autosomal dominant inheritance has been reported, most cases do not show any clear pattern of inheritance as in the present case.[4] The pathogenesis of the disease is not clear.[1] Recent analysis has identified a locus on chromosome nine responsible for venous malformations.[2]

Cutaneous lesions are often apparent at birth and increase in number from few to hundreds with age, present predominantly on the upper limbs and trunk [1,3,5] In our case, lesions started in early childhood and increased in number to 21. Three types of cutaneous lesions have been described: (1) blue, rubbery, blood-filled sacs with a smooth or wrinkled surface that are easily compressible and promptly refill when pressure is released; (2) large, disfiguring, cavernous lesions that may compress vital structures; and (3) blue irregular macules.

The present case is associated with type 1 lesions intraorally, on neck and phalanges, however, the lesion of left middle third of the face presented similar to a cavernous lesion described in the literature.[1] An increase in size of the extraoral and skin lesions under gravity, as seen in our case, may explain the venous nature of the lesions.

Vascular malformations when affecting the gastrointestinal tract, may occur anywhere from oral to anal mucosa but predominantly occur in the small bowel.[1,2]

Although the syndrome is rare, it is usually associated with oral and maxillofacial lesions. Oral lesions occur in 59 to 64% of cases, most common sites being the buccal mucosa, retromolar trigone and ventrolateral tongue.[6,7] Most of these sites were involved in our case.

Complications include acute hemorrhage, iron deficiency anemia following chronic bleeding, thrombocytopenia, disseminated intravascular coagulation, intussusception, volvulus, bowel infarction, skeletal bowing, pathologic fracture, bony overgrowth, articular derangement and pain due to phleboliths.[1,2,4,5,8] Ventricular hypertrophy associated with BRBNS has been reported as seen in our case.[1]

The differential diagnosis includes Maffucci syndrome,

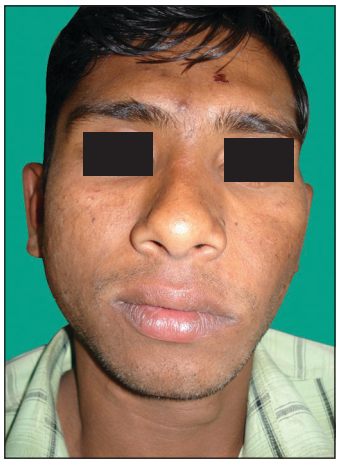

Figure 1: Diffuse swelling on the left cheek

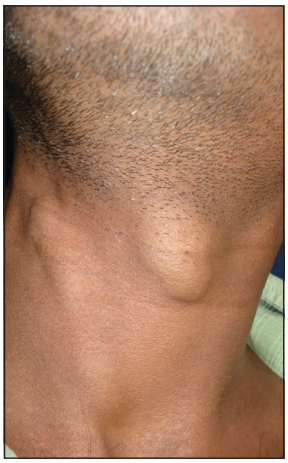

Figure 2: Multiple protruberant, blue colored, smooth surfaced blebs on the neck

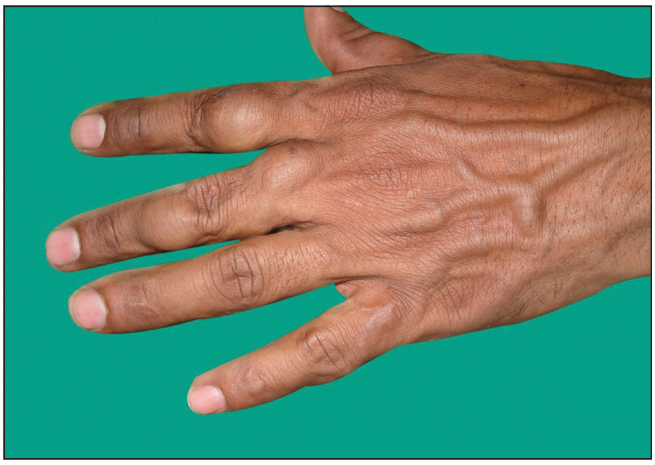

Figure 3: Multiple blue colored, blebs on the fingers

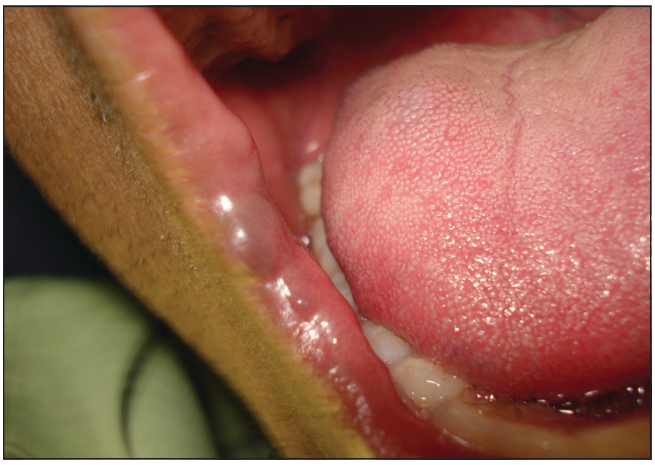

Figure 4: Similar swellings on the right labial mucosa

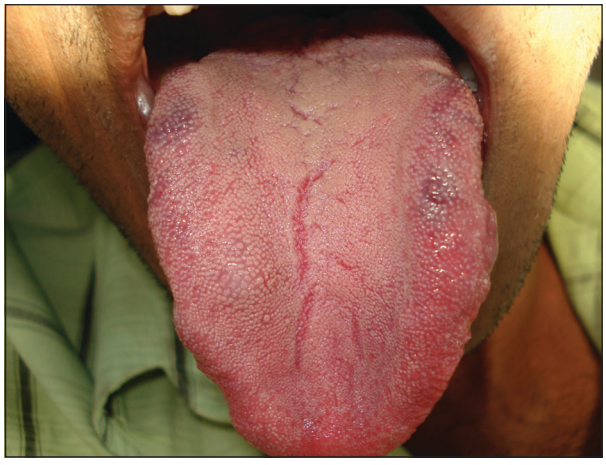

Figure 5: Blue colored, blebs on the dorsum and lateral border of tongue

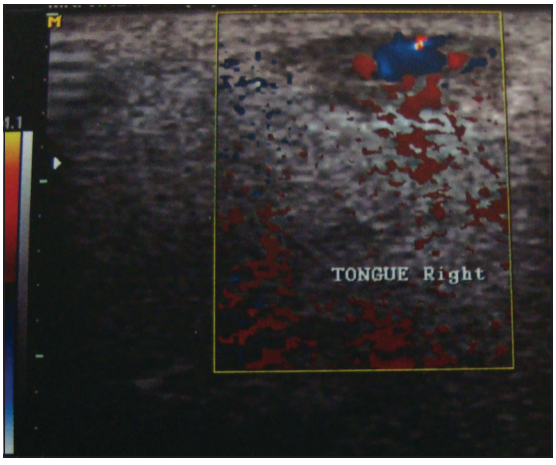

Figure 6: Ultrasonography of tongue lesions with compressible cystic septate masses and low velocity color flow detected on both color Doppler and power Doppler

Venous lakes, Osler Weber Rendu syndrome or hereditary hemorrhagic telengiectasias, disseminated hemangiomatosis and Kaposis sarcoma.[5,9,10]

Both sonography and MRI can confirm the nature of individual cutaneous venous malformations. Doppler interrogation can confirm the low-flow nature of the

lesions.[11] In the present case the low flow lesions on Doppler ultrasound is suggestive of venous malformations. MRI is helpful to depict the extent of deep lesions including the degree of involvement of underlying bones and joints.[11] MRI was not possible in our case due to claustrophobia.

Histopathologic examination of skin lesions revealed vascular tissue with tortuous, blood-filled ectatic vessels, lined by a single layer of endothelium, with surrounding thin connective tissue.[1-3] Due to fear of bleeding, patient did not agree for biopsy.

Though endoscopy failed to demonstrate gastrointestinal lesions in our case, the presence of skin lesions, vascular malformations of oral cavity as a part of GIT and along with symptoms of melena and occult blood in stools guided the diagnosis to BRBNS.

Treatment of cutaneous lesions on cosmetic or functional grounds includes electrodessication and curettage, liquid nitrogen, carbon dioxide laser or surgery. Bleeding from GI lesions is managed with iron supplementation, blood transfusions, endoscopic coagulation, endoscopic sclerotherapy and endoscopic laser (Nd:YAG) photocoagulation.[1,4,5] Octreotide has also shown varying degrees of success in treatment of gastrointestinal bleeding.[13]

Treatment proposed for the oral lesions include surgical therapy, intralesional injections of sclerosing agents, lasers, cryotherapy and radiation therapy. The sclerosant agents used are 5% sodium morrhuate, quinine urethrone, 5% ethanolamine oleate [EO], 1% pilidocanol and hypertonic saline.[3,12] The prognosis depends on the extent of visceral organ involvement. Most patients have a normal life span.[1]

CONCLUSION

BRBNS is a rare syndrome. The rarity is reflected in the rare reports in the dental literature. A patient with BRBNS is most likely to first present to an oral physician as it has been estimated that oral lesions occur in 59-64% of cases. Oromaxillofacial lesions may present as either blue rubber blebs or cavernous hemangiomas as in our case. A complete history and physical evaluation is a must to make a successful diagnosis to help in treatment planning and to prevent fatal potential complications. Treatment is symptomatic and conservative.

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