DERMATOLOGICA SINICA xxx (2018) 1–4

Contents lists available at ScienceDirect

Dermatologica Sinica

journal homepage: http://www.derm-sinica.com

CASE REPORT

Blue rubber bleb nevus syndrome associated with adult-onset cutaneous lesions and spontaneous intracranial hemorrhage: A case report and literature review

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ARTICLE INFO

Article history:
Received: Jun 17, 2017
Revised: Dec 21, 2017
Accepted: Jan 7, 2018

Keywords:
Bleeding
Central nervous system
Venous malformation
Blue rubber bleb syndrome
BRBNS

ABSTRACT

Blue rubber bleb nevus syndrome (BRBNS) is a rare disorder. Since 1860, only 200 case reports of BRBNS have been reported. BRBNS is characterized by numerous malformations of the vascular system that significantly involve the skin, gastrointestinal tract, and other internal organs. We describe a 72-year-old Taiwanese man with BRBNS. Besides typical adult-onset cutaneous lesions, he had venous malformation in the central nervous system, which caused massive intracerebral hemorrhage. He has been in a stupor ever since. Physicians should recognize cutaneous BRBNS lesions early and be aware of the possible complications that arise from internal organs involved.

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Introduction

Since described in 1860, there have been only about 200 case reports of blue rubber bleb nevus syndrome (BRBNS).[1] Typical cutaneous lesions appear as nipple-like, bluish nodules that easily compress and refill slowly.[1] Extracutaneous lesions most frequently affect the gastrointestinal (GI) system, causing bleeding and chronic anemia.[2] Besides the GI system, these lesions have been shown to affect multiple organs, including the central nervous system (CNS).[3] Although rare, BRBNS has the potential to cause fatal bleeding, especially in cases of CNS involvement. It is important to recognize its unique skin findings and multiple complications. Herein, we report a case of BRBNS in a patient with adult-onset cutaneous lesions and CNS involvement that led to spontaneous intracranial hemorrhage.

Case report

A 72-year-old Taiwanese man presented to the emergency room because of a change in consciousness. He had a generally healthy status until the morning of this visit when he was found to be drowsy. He had hypertension and attended regularly follow-up visits in clinics. The neurologic examination showed right hemiplegia and drowsiness. The physical examination revealed multiple bleb-like cutaneous nodules with variable diameters ranging from 1 to 2 cm on his chest, neck, and lips (Fig. 1A). The lesions were bluish, rubbery, and compressible. The results of other physical examination were normal. The family reported that the bleb-like skin lesions had developed in the patient's late 60s and were sometimes tender. He had previously visited another hospital for assessment of the lesions, but no specific diagnosis was made. His family history was negative for similar lesions. The brain computed tomography (CT) scan demonstrated a hyperdense area in the left basal ganglia and left frontal lobe (Fig. 1B). Blood tests were unremarkable except microcytic anemia, and the occult stool test result was positive. The patient underwent emergency left craniotomy, revealing a greyish nodule on the left basal ganglia, and the pathologic examination showed some vessels with thickened, hyaline walls in the brain tissue. Biopsy results of the cutaneous lesion on the chest wall showed dilated venous lumen lined with a thin layer of endothelial cells with packed red blood cells (Fig. 1C and D). Further investigations were refused by the family. Upon the follow-up visit after half a year, the patient has had a bedridden status, stupor consciousness, and tracheal intubation with ventilation.

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https://doi.org/10.1016/j.dsi.2018.01.002
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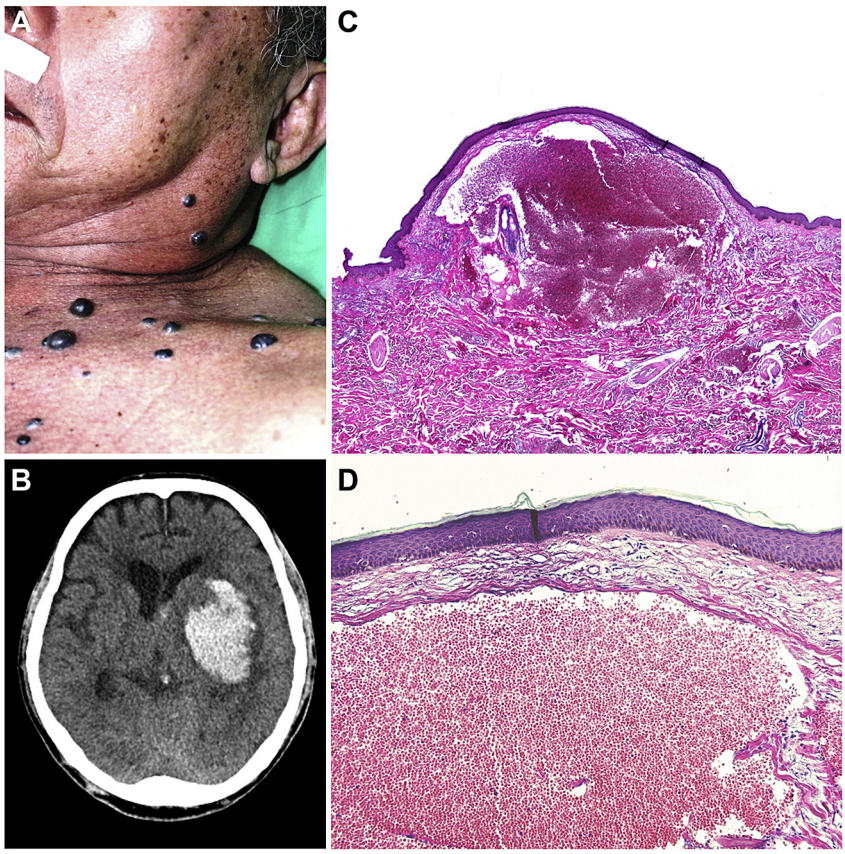

Fig. 1 (A) Multiple bleb-like nodules with variable diameters ranging from 1 to 2 cm on his chest and neck; (B) brain computed tomography (CT) scan demonstrating a hyperdense area in the left basal ganglia and left frontal lobe; (C) in the lower-powered field, a dilated venous lumen lined with a thin layer of endothelial cells (hematoxylin and eosin stain, ×40); (D) in the lower-powered field, the dilated venous lumen is filled with packed red blood cells (hematoxylin and eosin stain, ×100).

Discussion

BRBNS is a rare disorder characterized by discrete vascular malformations on the skin and visceral organs.[1,4] The condition was first mentioned by Gascoyen in 1860, but it was fully described by William Bean who called it the blue bleb nevus syndrome in 1958. It has since been called blue rubber bleb syndrome, Bean syndrome, blue bleb syndrome of Bean, and cellular blue nevus of Bean, which all refer to the same disease.[5] Lesions usually appear at or shortly after birth on any cutaneous surface, including the soles and palms, but the face, trunk, and limbs are the most frequently involved sites.[1] They often increase in size and number with age.[1] Only a few report have described adult-onset type BRBNS. Jin[6] et al. discovered that among 82 patients with BRBNS, only 4% of them had BRBNS that started in adulthood. Cases are found in all races, but BRBNS is rarely observed in African Americans.[6]

Most cases of BRBNS appear to be sporadic.[7] In 2017, Soblet et al. identified double (cis) mutations (two somatic mutations on the same allele) in *TEK*, the gene encoding TIE2, as the principal cause of BRBN.[8] They also found that BRBNS is one of disease of the spectrum of TEK-mediated venous phenotypes caused by somatic mutation in T1105N-T1106P.[8]

Typical cutaneous lesions are purplish, nipple-like lesions, which easily compress and refill slowly, but the manifestation may vary from small, blue-black punctate papules to large disfiguring vascular tumors.[1]

Extracutaneous lesions most frequently affect the GI system, predominantly the small intestines.[1] GI tract malformations may lead to frequent bleeding, causing occult blood loss and chronic anemia. Aside from the GI tract, BRBNS may also affect multiple organs, leading to different clinical findings, such as hematuria, epistaxis, focal seizures, and weakness, which can easily lead to a missed diagnosis or underdiagnosis. BRBNS has the potential to cause fatal bleeding, especially in cases of CNS involvement. Cases of BRBNS with CNS involvement with onset in adulthood are extremely rare. Symptoms depend on the location of the lesion.[1] Visual loss, exophthalmos, unilateral trigeminal neuralgia, and fatal brain hemorrhage have been reported (Table 1).[3,9–17]

The diagnosis of BRBNS is usually based on cutaneous characteristic lesions.[1] Numerous cutaneous, internal venous malformations, and GI lesions are pathognomonic.[8] Histologically, BRBNS lesions are characterized by blood-filled, tortuous, and ectatic vessels lined by thin endothelium, although gene analysis may identify a double mutation in *TEK* in a patient with BRBNS.[8]

In cases of possible CNS involvement, further imaging studies, such as brain CT or magnetic resonance imaging (MRI), are desirable, given the fragile nature of BRBNS venous malformation (thin endothelium) and the possible subsequent rupture and hemorrhage. It may also be fragile to minor trauma. Once rupture occurs, it may cause bleeding.

The differential diagnoses of BRBNS include Maffucci syndrome,[6] angiokeratoma corporis diffusum (Fabry disease),[10] and hereditary hemorrhagic telangiectasia (Osler–Weber–Rendu disease).[18] Maffucci syndrome manifests as deformation of the hands and feet, with enchondromas formations associated with a higher risk of cancer.[7] Fabry disease is an X-linked inherited disorder caused by a deficiency of the lysosomal enzyme α-galactosidase that leads to multiple angiokeratoma formations.

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