

Radiological findings of blue nevus syndrome. A case report

Manifestaciones radiológicas del síndrome del nevus azul. Presentación de caso

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Summary

The blue rubber bleb nevus syndrome is a rare vascular disorder entity that consists of multiple venous malformations that involve the skin, gastrointestinal tract and viscera. Patients may be asymptomatic or may present with various manifestations depending on the site of these vascular alterations and organs involved. The different diagnostic modalities allow for clear identification of vascular malformations; however, they are usually considered an incidental finding and are rarely the reason for the study to be performed. We describe the case of a 65-year-old patient with a diagnosis of blue nevus syndrome with evident venous malformations in computed tomography, ultrasound and magnetic resonance imaging.

Key words (MeSH)

Nevus, blue
Vascular malformations
Diagnostic imaging

Resumen

El síndrome del nevus azul es una entidad infrecuente que consiste en múltiples malformaciones venosas que comprometen la piel y el resto de los órganos. Los pacientes pueden ser asintomáticos o pueden cursar con diversas manifestaciones dependiendo del lugar donde se encuentren estas alteraciones vasculares. Las distintas modalidades diagnósticas permiten identificar claramente las malformaciones vasculares; sin embargo, usualmente son consideradas un hallazgo incidental y rara vez son el motivo de la realización del estudio. A continuación, se describe el caso de un paciente de 65 años de edad con diagnóstico de síndrome del nevus azul, con malformaciones venosas evidentes, en modalidades como tomografía computarizada, ultrasonido y resonancia magnética.

Palabras clave (DeCS)

Nevo azul
Malformaciones vasculares
Diagnóstico por imagen

Introduction

Blue nevus syndrome, also known as Bean syndrome, is a rare disease characterized by venous malformations of the skin and viscera, the most common location being the gastrointestinal tract (1). Macroscopic lesions include a spectrum of different morphologies; however, it is known that with the passage of time they increase in size and number; they can produce in some cases bleeding episodes (2). Nevi can be congenital or appear late and are usually not linked to a specific genetic pattern, although cases of autosomal dominant expression have been described (3). In general, this syndrome affects males and females equally; there is a slight predominance in Caucasians, although it is possible to find this syndrome in all races (4).

The following is the case of a 65-year-old patient with a history of blue nevus and multiple venous malformations in the body.

Clinical case

A 65-year-old male patient attended the outpatient diagnostic imaging service for a neck ultrasound due to the presence of a mass in the left supraclavicular region, which corresponded to a cyst of epidromic inclusion.

Additionally, there were findings consistent with low-flow vascular malformations with numerous venous sacs and phleboliths extending to the ipsilateral retropectoral region (Figure 1). On questioning, the patient referred to a history of blue nevus and previous studies also revealed vascular malformations in the left upper limb (Figure 2), in the chest wall (Figure 3), in the retroperitoneum (Figure 4) and in the neck (Figure 5). Some of these malformations were visible on physical examination as rounded violaceous lesions on the upper limbs and dorsum. Because of this, blue nevus syndrome was considered as a diagnostic impression.

Discussion

The first case of blue nevus syndrome was described by Gascoyen in 1860 and later characterized by Bean in 1958. For this reason it is also known as Bean's syndrome (1), which consists of a spectrum of vascular malformations in the skin and, simultaneously, in any other organ of the human body, frequently in the gastrointestinal tract, which means that the symptomatology varies depending on its location (1). Regarding skin lesions, they may present as a disfiguring structure that sometimes increases in size or compresses adjacent organs (type I); as a bluish-colored, rubber-like, compressible sac (type II); or as a blue or blackish mucous

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membrane or pulla (type III) (1). They are usually present from birth or infancy, although extracutaneous malformations may take a little longer to appear (2). In the patient in this case, his lesions are consistent with a type II, distributed on the upper limbs and dorsum.

Various diagnostic aids are used to characterize the syndrome. In the musculoskeletal and cutaneous manifestations, as in the case described here, the usefulness of ultrasound with Doppler and magnetic resonance imaging (MRI) has been demonstrated, and the latter is ideal for evaluating the involvement of bone and muscle tissues adjacent to the lesions (5). In MRI these malformations are observed as well-defined structures, of intermediate signal similar to that of the muscle in sequences with T1 information and of high signal in sequences with T2 information, with a prominent enhancement before the administration of contrast medium. Additionally, it is possible to find thrombi or phleboliths that are identified as focal se al voids (5). Phleboliths, in general, represent a diagnostic key for venous malformations (6). On ultrasound, they are identified as hypoechogenic, compressible lesions, with no flow or with low velocity monophasic flow on Doppler (6).

Among the patient's studies there was a computed axial tomography (CAT) of the chest, which was performed due to pain in the left hemithorax. There multiple calcifications were evidenced in the soft tissues in the supraclavicular, pectoralis major, right axillary regions and in relation to the latissimus dorsi muscle on both sides. The above, in relation to the phleboliths. Similarly, ultrasound of the neck, adjacent to the epidromic inclusion cyst, showed hypoechogenic lacunar images that on Doppler examination showed slow flow and phleboliths. In the MRI of the abdomen, a mass with a chemical appearance was visualized adjacent to the left renal hilum, with a slight contrast medium relaxation. Also, both in the left upper limb MRI angiography and neck MRI showed saccular dilatations infiltrating the muscle and subcutaneous cellular tissue.

Treatment depends on the size, number and location of the lesions (7), so it must be an individualized approach. One option is surgical resection, which is performed for aesthetic purposes or in cases where it causes pain. This alternative is preferred in small lesions (2-4 cm) that do not invade other structures, but are superficial; although, in some cases in which these conditions are not met, prior embolization may be chosen.

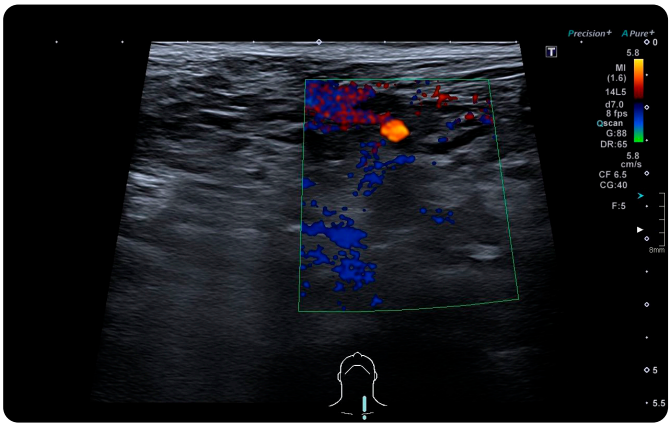


Figure 1. Doppler ultrasound of the neck. Multiple saccular images, hypoechoic, with diameters between 1 and 3 mm with slow flow and phleboliths.

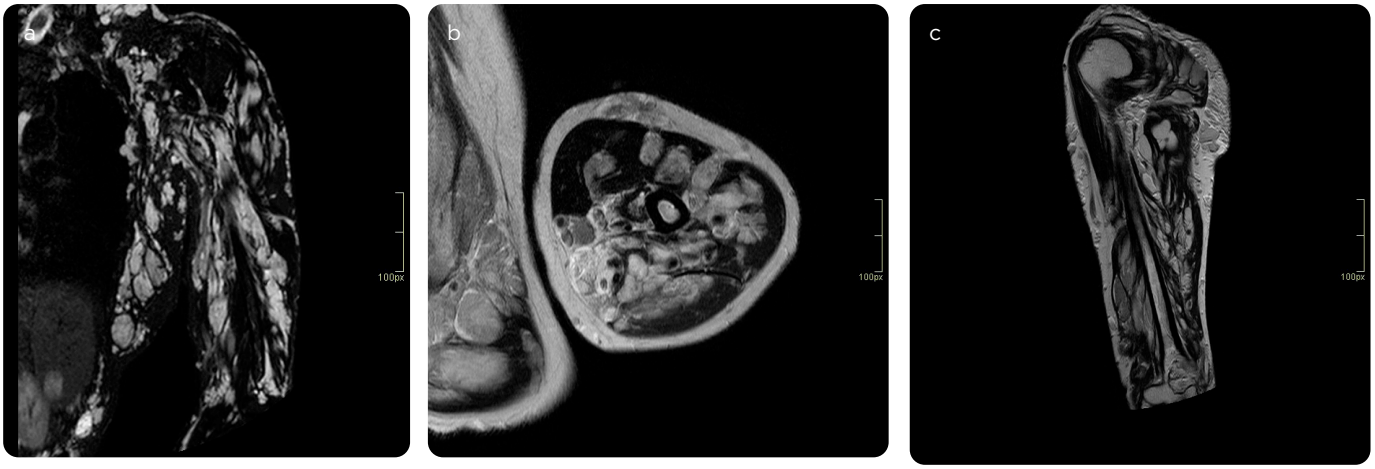


Figure 2. MRI of left upper limb. a) Coronal view in STIR sequence: multiple areas of high se al, of saccular form, involving the subcutaneous cellular tissue and the muscular planes of the arm and the left thoracic wall, with extension to the neck. b and c) Axial and sagittal view in DPI sequence that allows the visualization of phleboliths.

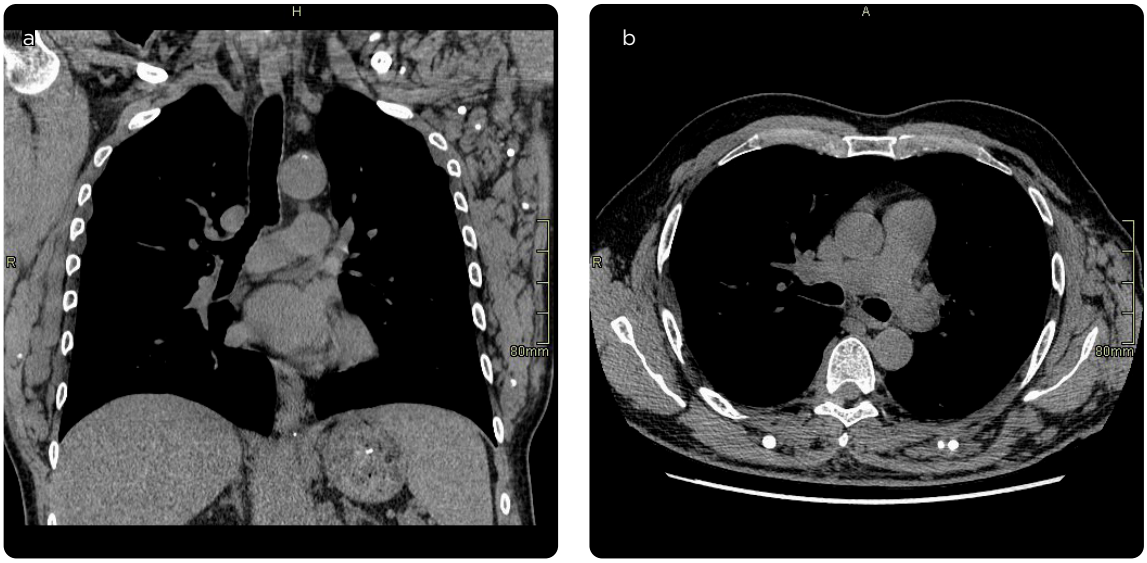


Figure 3. CT of the thorax (a) coronal and (b) axial: multiple calcifications in the soft tissues of the thoracic wall.



Figure 4. MRI of abdomen. a) Axial with T1 information, fat saturation and with contrast medium; b and c) axial and coronal MRI sequences with T2 information: mass of low se al in the left renal hilum that involves the vein and the renal artery.

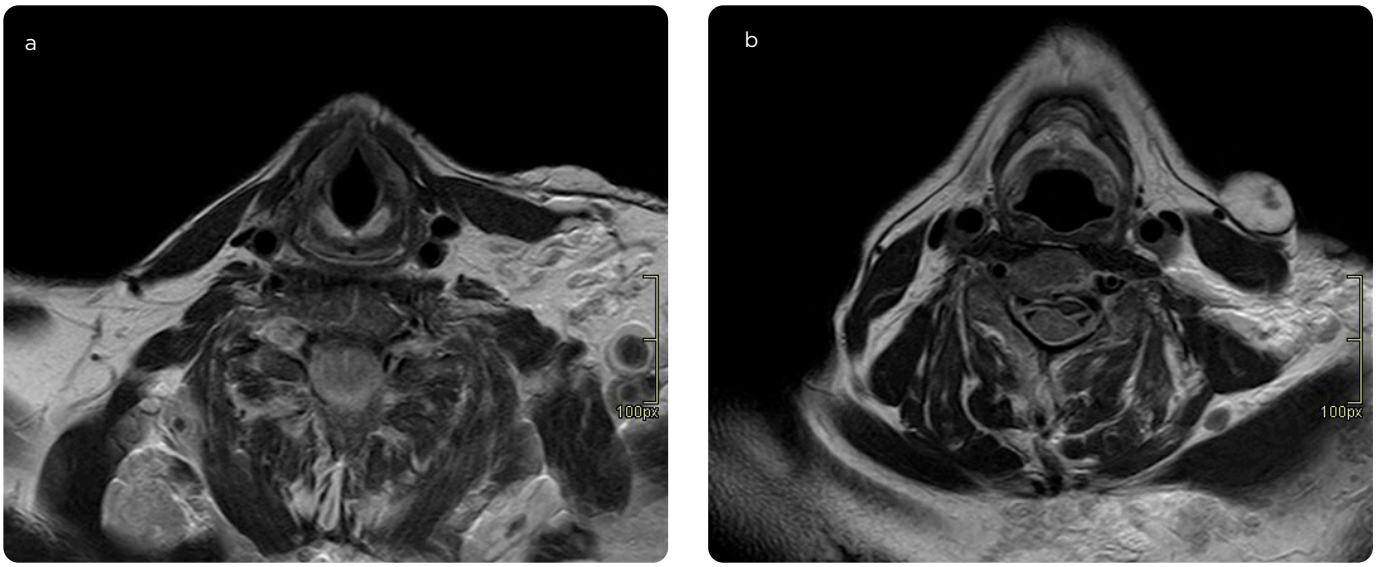


Figure 5. Neck MRI. a and b) Axial in sequence with T2 information: epidermoid cyst and areas of high se al in the paraspinal muscles. Phleboliths in soft tissues that originate artifice of magnetic susceptibility by calcium.

Sclerotherapy can also be offered as a treatment, which produces thrombosis and necrosis of the malformation, so it is suggested to inject contrast medium to better characterize the lesion, identify its flow and the drainage of other veins within the lesion. Finally, cryoablation has been described in the literature as a method to be performed under ultrasound guidance (7).

In conclusion, the patient in this case had not only the characteristic skin lesions, but also extensive soft tissue involvement, with vascular malformations in the chest wall, neck, arm and retroperitoneum. However, beyond the aesthetic manifestation, the patient had no additional manifestations.

Conclusion

Imaging plays an important role in blue nevus syndrome. They can be the starting point for incidental evidence of this pathology, since it is an infrequent disease and in some cases patients are asymptomatic; they are also useful for characterization of the lesions when they have already been identified on physical examination and, finally, interventional radiology can be used to provide management, such as sclerotherapy. Other treatments include surgical resection and cryoablation. It is important to remember that his main findings correspond to those characteristic of vascular malformations which, together with macroscopic skin lesions, make up the syndromic diagnosis. Although this patient had alterations in the soft tissues and in the retroperitoneal region, one of the most common sites of involvement is the gastrointestinal tract, which is usually visualized by endoscopy of the digestive tract or enteroscopy and also, in studies with barium or CT, when the malformations cause intussusceptions or any other complication and MRI, which evidences the lesions in organs such as liver, spleen and pancreas.

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