Glomuvenous malformations in a young man

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Abstract

A young man presented with blue nodules on the trunk, face, and extremities that gradually increased in number and size. His mother had similar lesions. Initially, blue rubber bleb nevus syndrome was suspected, but histological findings confirmed the diagnosis of hereditary glomuvenous malformations. Making the correct diagnosis spares the patient unnecessary evaluation for the arteriovenous malformations of the gastrointestinal tract associated with the former diagnosis.

Keywords: vascular malformation, hemangiomas/vascular tumors

Introduction

Glomuvenous malformations (GVM) are benign abnormal vascular proliferations arising from glomus cells. They appear in early childhood and continue to grow throughout adolescence. The lesions are most often sporadic but familial forms exist. It is often confused with blue rubber bleb nevus syndrome (BRBNS). The two entities can be distinguished by histological examination.

Case Synopsis

A 17-year-old boy presented for evaluation of blue nodules on the skin. He was born with a single blue lesion with a smooth surface in the right axilla that was elevated a few millimeters above the adjacent skin. He had no evidence of anemia or gastrointestinal bleeding. The patient's mother reported having a similar lesion on her left flank. Two months prior to his presentation, the lesion had increased in size. Physical examination revealed two right axillary lesions that were composed of three, confluent dark blue, compressible plaques forming a 3×1 cm plaque and another 2×1 cm plaque (**Figure 1**). He also had a 3× 4 mm dark blue compressible papule on his right foot, and smaller 1-2 mm dark blue macules on the right arm, right earlobe, and right cheek (**Figures 2, 3**).

An excisional biopsy was performed on the right axillary lesion. The histopathology demonstrated large endothelial-lined branching vessels, which extended from the superficial dermis to the subcutaneous fat. Two to ten layers of cuboidal cells were noted in the wall of these enlarged vessels (**Figures 4, 5**). These findings were consistent with GVM. BRBNS was initially favored on clinical grounds,

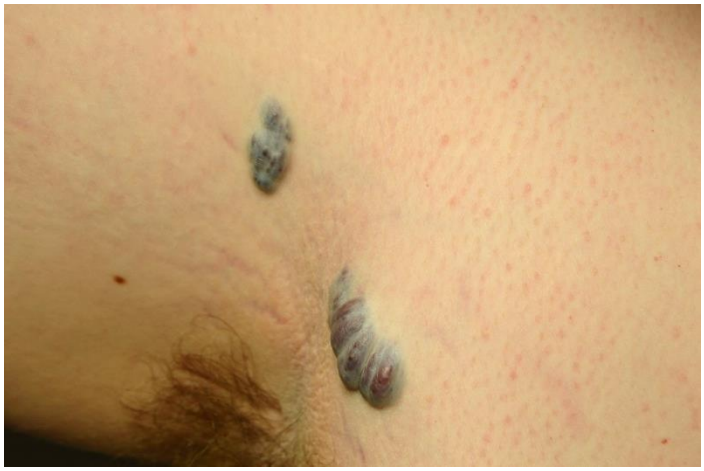

Figure 1. *A 3×1 cm dark blue compressible lesion composed of 3 lobules and another 2×1 cm plaque were present in the right axillary area.*

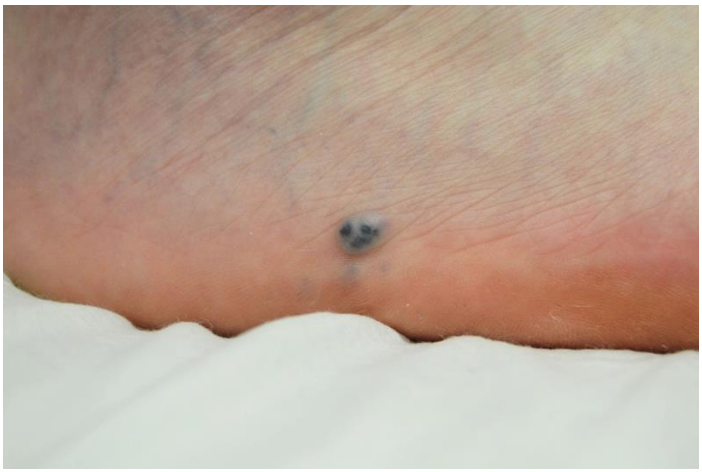

Figure 2. *A 3×4 mm dark blue compressible papule on the right foot.*

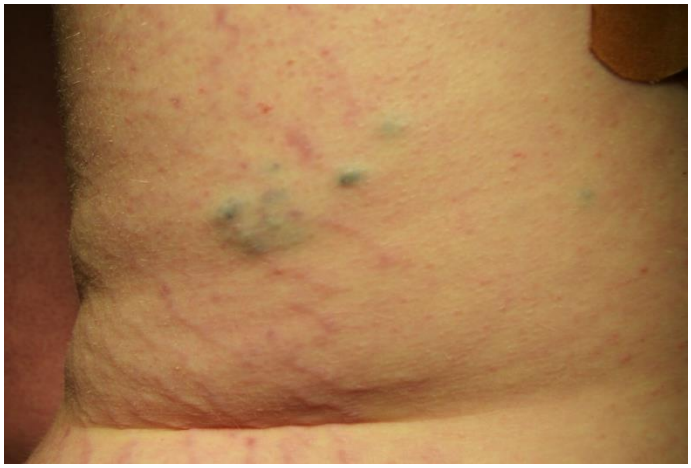

Figure 3. *Multiple 1-2 mm blue papules on the right arm.*

but the histopathology confirmed the diagnosis of multiple hereditary GVM.

Case Discussion

Glomuvenous malformations, formerly called glomangiomas [1], are tumor-like structures arising from glomus cells. Glomus cells are thermo-regulatory cells derived from smooth muscle cells. They are comprised of glomus bodies, which are usually present in acral skin; however, these tumor-like structures can be found in many locations on the body [2]. In contrast to the more common glomus tumors, which are also comprised of glomus cells, GVM are typically painless [3]. Glomuvenous malformations come in many shapes and sizes, ranging from pink to blue in appearance. The nodules appear primarily on the skin and only rarely on mucosal surfaces and internal organs. The histopathology of GVM shows thin-walled cavernous vascular spaces surrounded by one or more layers of glomus cells [2]. Glomuvenous malformations often appear sporadically but familial cases have been reported. The inheritance pattern is believed to be autosomal dominant with incomplete penetrance [3]. Mutations in the glomulin gene on chromosome 1p21-22 have been found to contribute to the formation of GVM [4].

The diagnosis of hereditary GVM is often challenging with a median time to diagnosis of almost 15 years

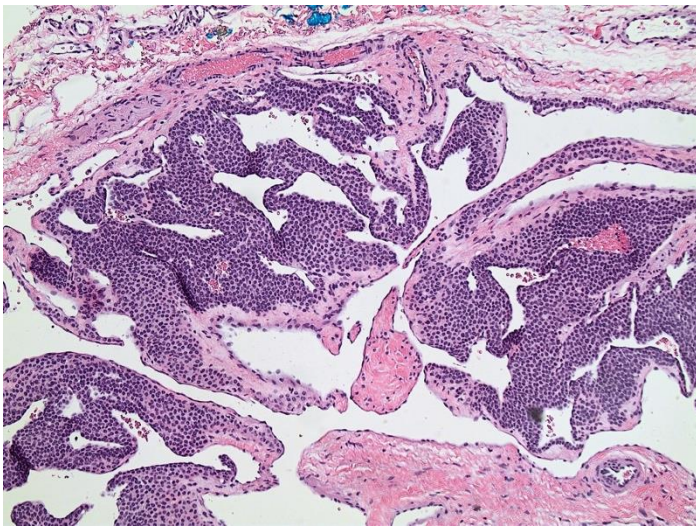

Figure 4. *Branching vascular spaces are noted, lined by endothelial cells and a perivascular proliferation of multilayered cuboidal cells with uniform round to ovoid nuclei. H&E, 100×.*

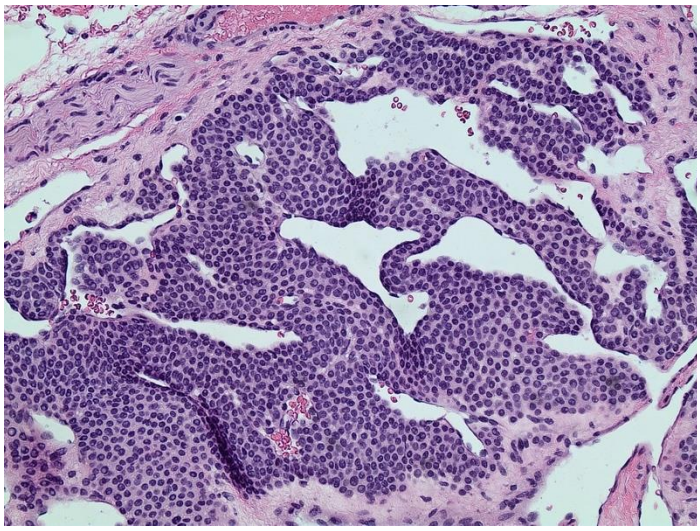

Figure 5. *Branching vascular spaces are noted, lined by endothelial cells and a perivascular proliferation of multilayered cuboidal cells with uniform round to ovoid nuclei. H&E, 200×.*

[3]. Clinically, GVM appears identical to BRBNS. In fact, it is believed that many cases of GVM have been misdiagnosed as blue rubber blebs in the past [5]. The distinguishing features of GVM include: 1) lack of mucosal and gastrointestinal involvement; and, 2) glomus cells on histopathology [2]. Although GVM are benign, they can become irritated and bothersome to patients. Successful treatment modalities include excision and laser therapy [1].

Conclusion

Distinguishing BRBNS from GVM is important. A diagnostic workup searching for vascular lesions of the bowel should be initiated to avoid bleeding, intussusception, and bowel necrosis associated with BRBNS. This can be avoided in GVM since it is not associated with lesions of the gastrointestinal tract.

References

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