

Venous malformation associated with skin hyperelasticity and subcutaneous atrophy

Malformação venosa associada a hiperelasticidade cutânea e atrofia do tecido subcutâneo

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Abstract

In syndromes that involve reduced quantities of elastin, the rigidity of vein walls may be increased, causing venous malformations, even in people who have mosaicism for these syndromes. There are no previous descriptions in the literature of collagen diseases presenting in specific, delimited areas. The patient described here presented with a lesion restricted to a well-defined area of the anteromedial thigh, in which elasticity was increased and vessels were tortuous, in the area of the lesion only, and with no other signs of collagen syndromes. A biopsy was conducted and the findings included changes to the normal arrangement of the elastic fibers and proliferation of blood vessels. The venous malformation was treated satisfactorily by embolization. Collagen diseases can cause cutaneous hyperextensibility, provoking flaccidity and a propensity to traumatism. Connective tissue diseases restricted to well-delimited areas are rare, since this group of diseases usually has systemic involvement. Vascular malformations can be seen in many different collagen diseases, but with generalized rather than localized presentation. One possible explanation for the case described here is genetic mosaicism.

Keywords: skin abnormalities; vascular diseases; vascular dermatopathies; vascular malformations; collagen diseases.

Resumo

A rigidez da parede venosa pode aumentar em síndromes em que há uma redução da quantidade de elastina, ocasionando malformações venosas mesmo em indivíduos que possuem mosaicism para tais síndromes. Casos com apresentação de afecções colagenosas em áreas delimitadas não foram descritos na literatura. O paciente apresentava lesão bem delimitada em região anteromedial da coxa com aumento de elasticidade e presença de vasos tortuosos apenas no local da lesão, não apresentando nenhuma síndrome colagenosa. Foi realizada uma biópsia que evidenciou alterações em relação ao padrão das fibras elásticas e proliferação de vasos sanguíneos. A malformação venosa foi tratada satisfatoriamente com embolização. As doenças do colágeno causam hiperextensibilidade cutânea, o que provoca flacidez e propicia traumas. As colagenoses bem delimitadas são raras, pois geralmente esse grupo de doenças envolve acometimento sistêmico. As malformações vasculares podem ocorrer em diversas doenças do colágeno, mas de forma generalizada e não localizada, e uma explicação para isso seria o mosaicism genético.

Palavras-chave: anormalidades da pele; doenças vasculares; dermatopatias vasculares; malformações vasculares; doenças do colágeno.

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■ INTRODUCTION

Venous malformations are congenital vascular anomalies caused by diffuse errors during embryonic development that result in inadequate development of the venous and/or arterial vascular systems.¹ A significant reduction in elastin content can cause increased venous wall rigidity and this sometimes occurs in collagen abnormalities.² Genetic mosaicism is defined as the presence of countless cell populations with different genotypes found in the same person.³ There are no previous descriptions in the literature of collagen diseases presenting in specific delimited sites.

■ CASE REPORT

A 25-year-old male patient presented with a well-delimited lesion in the anteromedial region of the thigh with atrophy of subcutaneous tissue. The lesion had a diameter of 20 cm and was related to increased skin elasticity, restricted to the affected site only. Blood vessels were dilated and tortuous, but there were no signs of Marfan syndrome or other diseases of the connective tissues, such as Ehlers Danlos syndrome, subcutaneous T-cell lymphoma, osteogenesis imperfecta or pseudoxanthoma elasticum (Figure 1).

Doppler ultrasonography revealed reflux in the great saphenous vein, varicose dilation of superficial collateral veins of the thigh and arterial abnormalities. Magnetic resonance showed that the thighs were asymmetric, with thinning of the subcutaneous tissue

in the left thigh, in the same place that the lesion was visible on physical examination, and morphology of the left gracilis muscle was longer and thinner than in the right thigh. On arteriography, the distal third of the femoral artery was blurred at the medial surface and the superficial femoral artery had a larger number of branches. No early venous filling was seen. In view of the inconclusive results, skin and vein biopsies were performed.

The biopsy histology findings were as follows: skin with an abnormal elastic fiber pattern in the reticular dermis, with a predominance of fine, pre-elastic type fibers; connective tissues exhibited dilated and tortuous venous vascular structures; changes to the usual pattern of elastic fibers and rarefaction of coarse elastic fibers; and proliferation of small caliber dermal blood vessels (Figure 2).

The venous malformation at the anterior surface of the thigh was embolized with 3% polidocanol foam. Three months after the treatment, the patient reported an 80% subjective improvement in the appearance of the lesion. The month after this reassessment, embolization with 3% polidocanol foam (2 mL) was performed once more, with a significant improvement in the vein's appearance, according to the patient, who was satisfied with the results (Figure 3).

■ DISCUSSION

Connective tissue diseases are characterized by destruction of the elastic properties of collagen fibers and by abnormalities of connective tissue synthesis



Figure 1. (a) Venous malformation in the anteromedial thigh; (b) Hyperelasticity in the region of the venous malformation.

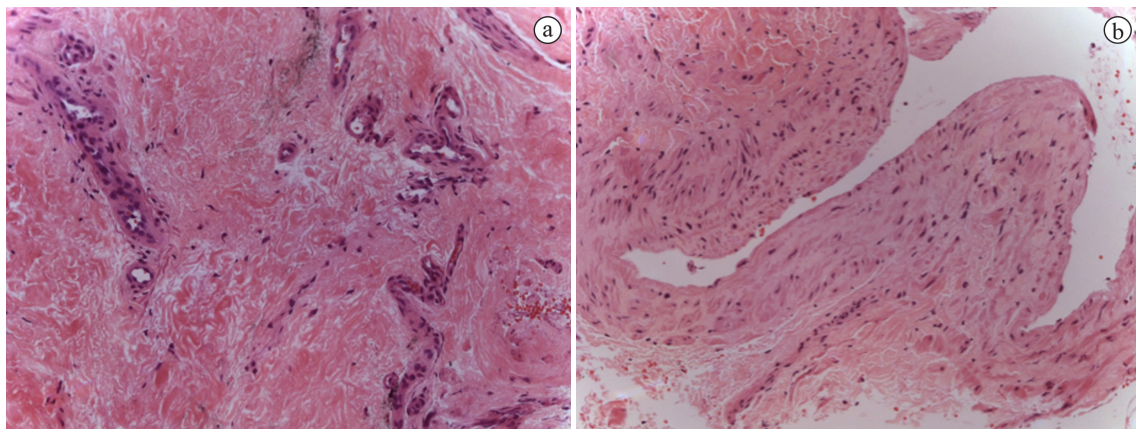


Figure 2. Slides from biopsies of skin and the venous malformation. (a) Microscopy of the reticular dermis: changes to the normal pattern of elastic fibers with predominance of fine, pre-elastic type fibers and areas of rarefaction of the coarse elastic fibers. There is also discrete proliferation of small caliber dermal blood vessels; (b) Microscopy of the vein: connective tissue exhibiting venous vascular structures with dilated and tortuous vessel walls.



Figure 3. Postoperative result of embolization of the venous malformation.

and structure. These conditions are made possible by genetic mutations that are specific to each disease. Cutaneous hyperextensibility, with consequent flaccidity and vulnerability to traumas, is one of the classic symptoms of connective tissue diseases.⁴ Other causes of elastic skin are pseudoxanthoma elasticum,⁵ Ehlers Danlos syndrome,⁶ Marfan syndrome,⁷ osteogenesis imperfecta⁸ and subcutaneous T-cell lymphoma. The patient described here only exhibited signs of connective tissue disease in a specific delimited area, which is not characteristic of systemic diseases.

The symptoms of a venous malformation are related to its size and distribution – those most often observed

are pain, swelling and thrombosis.^{1,9} Delineation of malformations is achieved using magnetic resonance and direct-injection venography. Treatment may be surgical, non-surgical, a combination of both or by sclerotherapy.¹⁰ Sclerotherapy is employed to reduce the size of the lesion and can be used as a preoperative support or as a supplement during the postoperative period. The sclerosing agent most widely used to treat venous malformations is polidocanol, which is beneficial because of its low complication rates.

CONCLUSIONS

Collagen abnormalities with cutaneous hyperextensibility, atrophy of subcutaneous tissue and vascular malformations can occur in several genetic diseases, but in a generalized, rather than localized, pattern. For these signs to be precisely delimited, the tissue abnormalities must be restricted to a small area and genetic mosaicism is a possible explanation for this presentation.

REFERENCES

1. Cabrera J, Cabrera J Jr, Garcia-Olmedo MA, Redondo P. Treatment of venous malformations with sclerosant in microfoam form. *Arch Dermatol.* 2003;139(11):1409-16. <http://dx.doi.org/10.1001/archderm.139.11.1409>. PMID:14623700.
2. Gandhi RH, Irizarry E, Nackman GB, Halpern VJ, Mulcare RJ, Tilson MD. Analysis of the connective tissue matrix and proteolytic activity of primary varicose veins. *J Vasc Surg.* 1993;18(5):814-20. [http://dx.doi.org/10.1016/0741-5214\(93\)90336-K](http://dx.doi.org/10.1016/0741-5214(93)90336-K). PMID:8230568.
3. De S. Somatic mosaicism in healthy human tissues. *Trends Genet.* 2011;27(6):217-23. <http://dx.doi.org/10.1016/j.tig.2011.03.002>. PMID:21496937.
4. Callewaert B, Malfait F, Loeys B, De Paepe A. Ehlers-Danlos syndromes and marfan syndrome. *Best Pract Res Clin Rheumatol.*

- 2008;22(1):165-89. <http://dx.doi.org/10.1016/j.berh.2007.12.005>. PMID:18328988.
5. Mayo Clinic [site na Internet]. Ehlers-Danlos syndrome. Rochester: Mayo Clinic; 2015 [citado 2015 out 03]. <http://www.mayoclinic.org/diseases-conditions/ehlers-danlos-syndrome/basics/definition/con-20033656>
 6. National Heart, Lung, and Blood Institute – NHLBI [site na Internet]. What is Marfan syndrome? Bethesda: NHLBI; 2010 [citado 2015 out 03]. <https://www.nhlbi.nih.gov/health/health-topics/topics/mar/>
 7. OrthoInfo [site na Internet]. Osteogenesis imperfecta. Illinois: American Academy of Orthopaedic Surgeons; 2015. [citado 2015 out 03]. <http://orthoinfo.aaos.org/topic.cfm?topic=a00051>
 8. Genetics Home Reference [site na Internet]. Pseudoxanthoma elasticum. Bethesda: U.S. National Library of Medicine; 2015 [citado 2015 out 03]. <http://ghr.nlm.nih.gov/condition/pseudoxanthoma-elasticum>
 9. Rabe E, Pannier F. Sclerotherapy in venous malformation. *Phlebology*. 2013;28(Supl 1):188-91. <http://dx.doi.org/10.1177/0268355513477282>. PMID:23482557.
 10. Gopal B, Keshava SN, Moses V, et al. Role of percutaneous sclerotherapy in venous malformations of the trunk and extremities: a clinical experience. *Biomed Imaging Interv J*. 2013;9(3):1-6.

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