เนื้องอกหลอดเลือดชนิด Multiple Familial Glomangioma: รายงานผู้ป่วย

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Abstract: Multiple Familial Glomangioma: A Case Report

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(Received: 22 November, 2022; Revised: 28 December, 2022; Accepted: 1 May, 2023)

Glomuvenous malformations (GVMs), also known as glomangiomas, are benign localized tumors of the skin, that often appear during infancy and childhood. Diagnosis of glomangiomas is based on clinical and histological features. This was a report of a 31-year-old woman who presented with violaceous plaques on the right upper chest since birth. New lesions continued to develop on the other sites including the right shoulder, right arm, right palm and right leg. Her brother had similar lesions on his left thigh. Incisional biopsy showed multiple dilated cavernous-like, thin-walled vascular spaces surrounded by multiple layers of glomus cells and red blood cells in the lumen. Her clinical and histological features were consistent with the diagnosis of multiple familial glomangioma. The treatment was not required for asymptomatic glomangiomas but the larger and extensive glomangiomas could be treated with laser therapy or sclerotherapy. This patient was treated with Nd: YAG (Excel V) for a reduction in glomangioma size.

Keywords: Glomuvenous malformations, Glomangiomas, Venous malformations

บทคัดย่อ

Glomuvenous malformations (GVMs) หรือ glomangiomas เป็นเนื้องอกที่พบได้ตั้งแต่เด็ก รอยโรคมักอยู่ที่ผิวหนัง การวินิจฉัย glomangiomas ใช้ลักษณะทางคลินิกและผลการตรวจทางพยาธิวิทยา รายงานผู้ป่วยรายนี้เป็นผู้หญิงอายุ 31 ปี มาพบแพทย์ด้วยมีผื่นนูนสีคล้ำที่หน้าอกขวาตั้งแต่แรกเกิด ต่อมามีขนาดโตขึ้น และมีรอยผื่นใหม่ที่ไหล่ขวา ฝ่ามือขวาและต้นขาขวา พี่ชายของผู้ป่วยมีรอยผื่นแบบเดียวกันที่ต้นขาซ้าย ผู้ป่วยได้ทำการตัดชิ้นเนื้อผลทางพยาธิวิทยาพบ multiple dilated cavernous-like, thin-walled vascular spaces surrounded by multiple layers of glomus cells, red blood cells in the lumen จากลักษณะทางคลินิกและผลทางพยาธิวิทยาเข้าได้กับ multiple familial glomangioma การรักษา glomangiomas อาจไม่มีความจำเป็นในคนไข้ที่ไม่มีอาการ ในกรณีที่รอยโรคขนาดใหญ่หรือมีอาการอาจให้การรักษาด้วยเลเซอร์ หรือการฉีดสาร sclerotherapy สำหรับผู้ป่วยรายนี้เลือกใช้เลเซอร์ Nd: YAG เพื่อลดขนาดของ glomangiomas

คำสำคัญ: เนื้องอกหลอดเลือดชนิด Glomuvenous, Glomangiomas, Venous malformations

Introduction

Glomuvenous malformations (GVMs), also known as glomangiomas, are benign localized tumors of the skin comprising glomus bodies with predominant blood vessels.[1, 2] Glomangiomas are often present at birth but may also appear later in life and are most commonly found in the skin or subcutaneous tissue of the upper extremity but occasionally can be presented on the lower extremities, head, or neck. The glomangiomas are multifocal, rather than localized, frequently hyperkeratotic with a cobblestone-like appearance, and their color varies from pink to purplish dark blue.[3-5] It is often confused with venous malformations (VMs) but can be distinguished by clinical and histopathological examination. The treatment is not required for asymptomatic glomangiomas. The small, superficial lesions can be treated with surgical excision, however the larger and extensive glomangiomas may be treated with laser therapy or sclerotherapy.[13, 14]

Case report

The 31-year-old Thai woman presented with violaceous plaques on the right upper chest since birth. New lesions continued to develop on the other sites including the right shoulder, right arm, right palm, and right leg. Some of the lesions were painful and were precipitated by pressure. She denied a history of anemia or bleeding either from the skin lesion or gastrointestinal tract. There were no other systemic symptoms. Her elder brother had a similar lesion on his left thigh. Physical examination revealed multiple violaceous non-blanchable plaques with the nodular surface on the upper chest extended to the right arm, right shoulder, right palm, and right leg (Figure 1). There was no thrill or bruit. She had normal extremities without limb-length discrepancy. Magnetic Resonance Imaging (MRI) of the chest wall, right arm, and right forearm showed low-flow type vascular malformation involving skin and subcutaneous tissue along the right upper extremities, right shoulder, and right-sided anterior upper chest wall.

An incisional biopsy was performed on the right arm shows multiple dilated cavernous-like, thin-walled vascular spaces surrounded by multiple layers of glomus cells, red blood cells in the lumen, no significant finding of arteriovenous malformation and no atypical cell (Figure 2, 3). The clinicopathology confirms the diagnosis of multiple familial glomangioma.

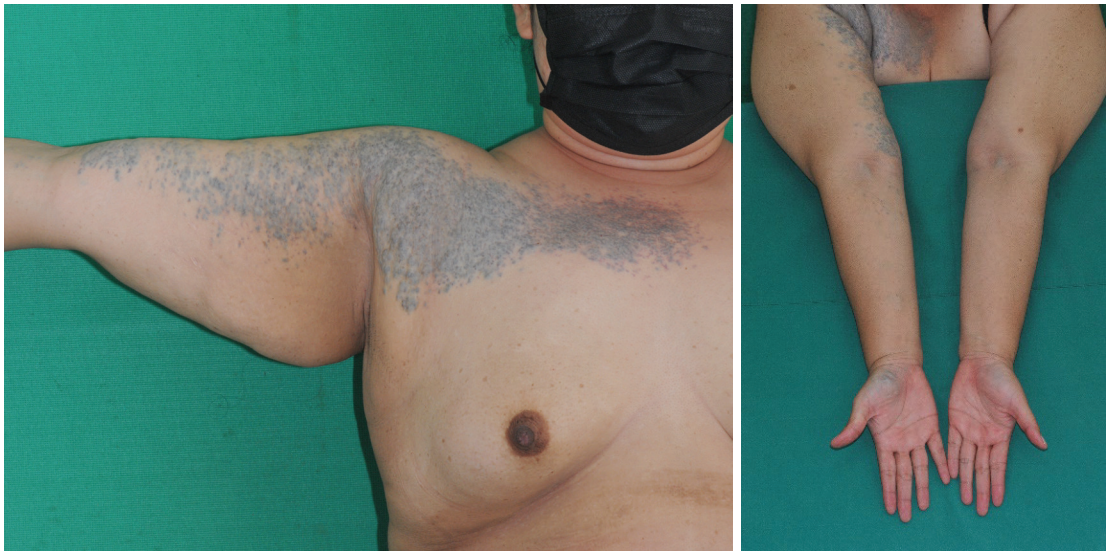

Figure 1. Multiple violaceous non-blanchable plaques with the nodular surfaces on the upper chest extended to the right arm and right palm.

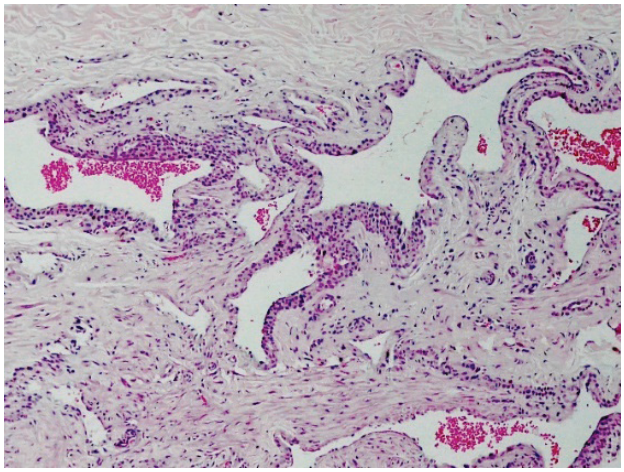

Figure 2. Multiple dilated cavernous-like, thin-walled vascular spaces surrounded by multiple layers of glomus cells. H&E, x100

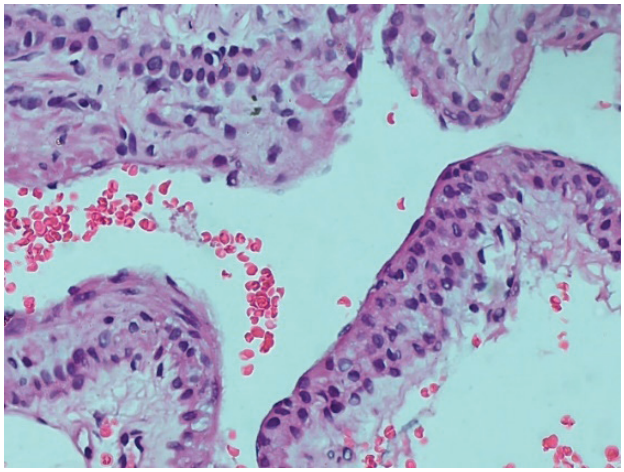

Figure 3. Multiple dilated cavernous-like, thin-walled vascular spaces surrounded by multiple layers of glomus cells. H&E, x400

Case discussion

Glomuvenous malformations (GVMs), also known as glomangiomas, are benign localized tumors of the skin comprising glomus bodies, the specific cutaneous structures that are involved in thermal and baroregulation.[1, 2] Glomangiomas are most commonly found in the skin or subcutaneous tissue of the upper extremity but occasionally can be presented on the lower extremities, head, or neck. Rarely, the tumors may involve mucous membranes and internal organs.[3]

The glomangiomas are multifocal, rather than localized, frequently hyperkeratotic with a cobblestone-like appearance, and their color varies from pink to purplish dark blue[4, 5]. Glomangiomas are different from venous malformations (VMs) in clinical presentation. Glomangiomas are nodular or segmental plaques. Color may vary from pink to purplish dark blue, mainly located on the extremities and involving skin and subcutis. They are generally asymptomatic, but some lesions may be tender by compression, and pain attacks may occur in association with menstruation and pregnancy. The venous malformations are soft, blue, often localized on vascular lesions, commonly affected muscles and joints, and painful on awakening or after activity. Therefore differentiating glomangiomas from VMs is important to the outcome and the treatment options.[6, 7, 8]

Glomangiomas are often present at birth but may also appear later in life and slowly expand during childhood. Solitary and multiple glomangiomas must be distinguished based on characteristic clinical and histologic findings, occurring sporadically or in a familial pattern. Nonfamilial solitary type glomangioma is more common than multiple types and frequently painful, histological findings are small endothelium-lined cavities and many glomus cells in 2-3 layers. However, multiple type glomangiomas are familial autosomal dominant inheritance, presented with localized, segmental, or disseminated patterns, in which histological findings are irregular large ectatic vessels and few glomus cells. They are asymptomatic and sometime may be slight tenderness. Approximately two-thirds of patients with glomangiomas have a family history of similar lesions. Autosomal dominant inheritance of glomangiomas results from a heterozygous germline mutation in the glomulin gene on chromosome 1p21-22.[9, 10] This generates a truncated glomulin protein that interferes with vascular smooth muscle differentiation and late maturation of vascular smooth muscle cells resulting in the formation of glomus cells. Four mutations in glomulin were identified in 70% of the GVM families (108C → A, 157delAAGAA, 554delA+556delCCT, and 1179delCAA) whereas the other 30% have unique mutations.[9, 11, 12]

Histological examination of glomangiomas consists of large, dilated, thin-walled veins in the dermis and subcutaneous tissue, histologically equivalent to those comprising venous malformation but surrounded by clusters of round or polygonal cells with plump nuclei and scant eosinophilic cytoplasm (glomus cells), These features were consistent with the diagnosis of glomangioma in our patient.[9, 12] The congenital origin, the scattering lesions, and the similar lesion of her sibling led to the provisional diagnosis of multiple familial glomangiomas with mosaicism.

Treatment is not required for asymptomatic glomangiomas. The small, superficial lesions can be treated with surgical excision, however the larger and extensive glomangiomas may be treated with laser therapy. Other alternatives include sclerotherapy and some studies demonstrate that sclerotherapy treatment in large facial glomangioma did not improve facial contour or discoloration.[13, 14] Our patient will be treated with Nd: YAG (Excel V) for a reduction in glomangioma size.

Conclusion

This is reported of a rare case. Glomangiomas are benign, arise from the glomus body, and are commonly found in the skin or subcutaneous tissue. Glomangiomas must be distinguished from venous malformations by clinical features and location. The diagnosis was based on the clinical aspect and confirmed with the histopathologic results.

References:

1. Jha A, Ramesh V, Singh A. Disseminated cutaneous glomuvenous malformation. Indian J Dermatol Venereol Leprol 2014; 80(6):556-8.
2. Abbas A, Braswell M, Bernieh A, Brodell RT. Glomuvenous malformations in a young man. Dermatol Online J 2018; 24(10):11.
3. Myers RS, Lo AKM, Pawel BR. The glomangioma in the differential diagnosis of vascular malformations. Ann Plast Surg 2006; 57(4):443–6.
4. Blume-Peytavi U, Adler YD, Geilen CC, Ahmad W, Christiano A, Goerdt S, et al. Multiple familial cutaneous glomangioma: a pedigree of 4 generations and critical analysis of histologic and genetic differences of glomus tumors. J Am Acad Dermatol 2000; 42(4):633–9.
5. Brouillard P, Ghassibé M, Penington A, Boon LM, Dompmartin A, Temple IK, et al. Four common glomulin mutations cause two thirds of glomuvenous malformations ("familial glomangiomas") : evidence for a founder effect. J Med Genet 2005; 42(2):13.
6. Mounayer C, Wassef M, Enjolras O, Boukobza M, Mulliken JB. Facial "glomangiomas": large facial venous malformations with glomus cells. J Am Acad Dermatol 2001; 45(2):239–45.
7. Cabral CR, Oliveira Filho Jd, Matsumoto JL, Cignachi S, Tebet AC, Nasser Kda R. Type 2 segmental glomangioma--Case report. An Bras Dermatol 2015; 90(3 sappl 1) :97-100.
8. Hughes R, Lacour J-P, Chiaverini C, Rogopoulos A, Passeron T. Nd:YAG laser treatment for multiple cutaneous glomangiomas: report of 3 cases. Arch Dermatol 2011; 147(2):255–6.
9. Calvert JT, Burns S, Riney TJ, Sahoo T, Orlow SJ, Nevin NC, et al. Additional glomangioma families link to chromosome 1p: no evidence for genetic heterogeneity. Hum Hered 2001; 51(3):180–2.
10. Dérand P 3rd, Warfvinge G, Thor A. Glomangioma: a case presentation. J Oral Maxillofac Surg 2010; 68(1):204-7.
11. de la Fuente S, Hernández-Martín A, Happle R, Torrelo A. Type 2 mosaicism in familial glomangiomatosis. Actas Dermosifiliogr 2014; 105(5):524–5.
12. Boon LM, Mulliken JB, Enjolras O, Vikkula M. Glomuvenous malformation (glomangioma) and venous malformation: distinct clinicopathologic and genetic entities: Distinct clinicopathologic and genetic entities. Arch Dermatol 2004; 140(8):971–6.
13. Parsi K, Kossard S. Multiple hereditary glomangiomas: successful treatment with sclerotherapy. Australas J Dermatol 2002; 43(7):43–7.
14. Shah A, Tassavor M, Sharma S, Tassavor B, Torbeck R. Surgical and non-surgical treatment modalities for glomuvenous malformations. Dermatol Online J 2021; 27(7):18.