Lower Extremity Fibro-Adipose Vascular Anomaly (FAVA): A New Case of a Newly Delineated Disorder

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The acronym FAVA (Fibro-Adipose Vascular Anomaly) has been recently given to a distinct vascular entity that is characterized by fibrofatty infiltration of muscle, unusual phlebectasia with pain, and contracture of the affected extremity. We report a new case of FAVA in a 10-year-old girl with pain in her right lower leg and equinus contracture. As in our case, FAVA typically presents in young females with calf involvement and limited ankle dorsiflexion with local pain. FAVA should be considered as a differential diagnosis when evaluating vascular anomalies in the lower extremities.

Keywords: Fibro-Adipose Vascular Anomaly, vascular anomalies, venous malformation

Introduction

A cohort of patients with a unique intramuscular lesion of the extremities comprising dense fibrofatty tissue and slow-flow vascular malformations have been recently identified. The acronym FAVA (Fibro-Adipose Vascular Anomaly) has been given to a distinct entity that is characterized by fibrofatty infiltration of muscle, unusual phlebectasia with pain, and contracture of the affected extremity. Description of new vascular entities with characteristic clinical, radiological and histopathological features helps to better define the management of these challenging patients. Vascular surgery consultation is often necessary in FAVA patients, therefore vascular surgeons should be aware of this new entity to differentiate from the classical venous malformation and avoid unnecessary treatment. As in our case, FAVA typically presents in young females with calf involvement and limited ankle dorsiflexion with local pain. FAVA should be considered as a differential diagnosis when evaluating vascular anomalies in the lower extremities.

Case Report

A 10-year-old girl was referred to us for a painful mass in the right calf and equinus contracture of the right foot that presented progressively in the last year. Upon examination at our institution, there was a visible, tender mass in the lateral aspect of the right lower leg and a toe-walking in the right leg secondary to an equinus deformity which was not passively correctable to neutral. Her neurologic examination showed normal patellar tendon and Achilles tendon reflexes, full strength (5/5) of the surrounding musculature, and normal sensation. No skin findings were
observed. We obtained radiographs, right calf mass ultrasound and lower extremities magnetic resonance imaging (MRI) which showed a $5 \times 4.4$ cm complex mass in the right calf consistent with a venous malformation located in the lateral head of the gastrocnemius muscle (Fig. 1A). After discussion of the case in our Vascular Anomalies Clinic, a percutaneous sclerotherapy with etoxisclerol was indicated. This procedure showed a diffuse calf venous malformation with marked phlebectasia (Fig. 1B). Three months after sclerotherapy, the pain had improved but the toe-walking was remarkable and surgery was recommended. A marginal resection of the mass was performed through a right lateral calf incision (Fig. 2A). The mass had two components, a venous malformation within the gastrocnemius muscle surrounded by normal muscle tissue and a fatty component (Fig. 2B). A complete percutaneous Achilles tenotomy was also performed. The patient’s orthopaedic course was unremarkable and she began full weight bearing 3 weeks postoperatively. Follow-up examination 2 months after operation revealed normal ankle dorsiflexion and the patient reported no pain.

Histological exam (Fig. 3) revealed the lesion was predominantly composed of prominent dense fibrous tissue, adipose tissue and cluster of thin walled and back to back venous channels with anomalous lymphatic vascular component. Additionally lymphoplasmacytic aggregates, grouped atrophy of skeletal muscle and entrapped nerves were observed.

**Discussion**

In 1982, Mulliken and Glowacki classified congenital vascular lesions into vascular tumors (infantile hemangioma, rapidly involuting congenital hemangioma, non-involuting congenital hemangioma, kaposiform hemangioendothelioma and tufted angioma) and vascular malformations (arteriovenous malformation, venous malformation, lymphatic malformation, lymphatic-venous malformation, and capillary malformation). In 1996, the International Society for the Study of Vascular Anomalies (ISSVA) approved this classification system to establish a common language for the many different medical specialists who are involved in the management of these lesions. New vascular entities have been described and the ISSVA classification is routinely reviewed. Alomari, et al., from the Vascular Anomalies Center in Children’s Hospital Boston, have recently described a new vascular disorder called FAVA (Fibro-Adipose Vascular Anomaly) which constitutes a distinct clinical, radiologic, and histopathologic entity comprising fibrofatty infiltration of muscle, unusual phlebectasia with pain, and contracture of the affected extremity. As most of patients reported by Alomari, our patient was a pre-adolescent girl who presented with a painful calf mass, diagnosis of venous malformation (VM) on MRI/ultrasound with gastrocnemius muscle involvement, and firstly treated with sclerotherapy that showed phlebectasia (dilation of the veins) over the spongyform pattern seen in common VM. Predilection for lower extremities in FAVA is also reported and the overlying skin is usually unaffected. Eight of the 12 reported patients with calf lesions developed limited ankle dorsiflexion (i.e., equinus contracture) of variable severity as in our case, therefore orthopaedic consultation and physical therapy are often necessary.

A great variety of vascular anomalies are incorrectly referred to as ‘cavernous hemangiomas’ in the medical literature that is frequently used to name a venous malformation. As described in Alomari’s article, several cases reported as intramuscular cavernous hemangiomas might correspond with FAVA, since the lesions were typically located within the gastrocnemius muscle and dense fibrous-adipose tissue was
Fig. 2  Intraoperative findings.  (A) Gastrocnemius muscle infiltrated by venous malformation (black arrow).  (B) Specimen with venous malformation (black arrow) and fibroadipose tissue components (asterisk).

Fig. 3  Histological findings.  (A) Cross section consisted of a variable admixture of dense fibro-fatty tissue and vascular clusters. The fibrous component was particularly prominent.  (B) Clusters of vascular channels with a extensive fibro-fatty tissue, separating bundles of skeletal muscle.  (C) Cluster of thin walled, back to back and empty or blood filled venous channels.  (D) Anomalous lymphatic vascular component, supported by endothelial D2-40 immunopositivity.

found at surgical exploration. Histological findings in our case showed both components, fibrofatty tissue and slow-flow vascular malformations including venous and lymphatic subtypes. The lymphatic component was supported by endothelial D2-40 immunopositivity. The presence of entrapped nerves may be associated with local pain which is very common in FAVA and disproportionately severe.3)
Differential diagnosis of calf masses should include soft tissue sarcomas and traumatic intramuscular hematomas. A malignant tumor masquerading as hematoma should be suspected when the mechanism and the energy of the trauma do not justify the clinically detected severity of the injury, or the lesion does not follow the expected clinical course of resolution after initial conservative management.

Alomari\(^3\) describes that therapeutic options for FAVA patients include sclerotherapy and surgical resection, but the dominant solid fibrofatty component over the venous malformation makes FAVA less amenable to sclerotherapy. In our case, percutaneous sclerotherapy was temporally helpful with decrease in pain sensation, but the equinus contracture showed no improvement and required surgical resection and Achilles tenotomy.

**Conclusion**

To our knowledge, this is the second article about FAVA, a newly delineated disorder described in the field of vascular anomalies that typically presents in young females with calf involvement and limited ankle dorsiflexion with local pain. Description of new vascular entities with characteristic clinical, radiological and histopathological features helps to better define the management of these challenging patients. Vascular surgery consultation may be necessary in FAVA patients, therefore vascular surgeons should be aware of this new entity to differentiate from the classical venous malformation and avoid unnecessary treatment.

**Disclosure Statement**

Israel Fernandez-Pineda serves as a consultant to Virgen del Rocio Children’s Hospital and has no conflict of interest. All authors have no conflict of interest.

**References**