A Report of a Rare Case of Multiple Fibro adipose Venous Anomaly

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Learning Point of the Article:

Fibro adipose venous anomaly is a separate entity with distinguishable clinical, radiological, and pathological picture, where conservative and radiological interventions have poor outcome and increased morbidity.

Abstract

Introduction: Although not described more than a decade ago, fibro adipose vascular anomaly has grown of increasing importance as conventional management of arterio venous malformation with interventional radiology measures carry little success and cause significant morbidity in pediatric age groups like the case report we present here. Surgical resection, even though requiring significant loss of muscle bulk, is the mainstay of the treatment.

Case Report: An 11-year-old patient presented with equinus deformity and intensely tender calf and foot swellings in the right leg. Magnetic resonance imaging showed presence of two distinct lesions, one involving the gastrocnemius and soleus, and the other in the Achilles tendon for which en bloc resection of the tumor was performed. Histopathology of the specimens confirmed the diagnosis of fibro adipose venous anomaly.

Conclusion: As per our knowledge, this is the first case of multiple fibro adipose venous anomaly confirmed by clinical features, radiology, and histopathology.

Keywords: Vascular anomaly, fibrous, adipose, pediatric, lymph anomaly.

Introduction

DFibro-adipose vascular anomaly is a distinct, benign, and painful condition that was first described by Alomori et al. in 2014 [1]. It is classified under "Provisionally undiagnosed vascular anomalies" according to the International Society for the Study of Vascular Anomalies (ISSVA) [2]. Diagnosis of Fibro adipose vascular anomaly (FAVA) is made on the basis of clinical features, radiological features, and confirmed by histopathology. Patients typically present with unusual pain, contracture of the affected extremity, swelling, and limitation of range of motion [3]. Histologically, there is presence of fibro fatty infiltration into the muscle along with lymphoplasmacytic aggregates, atrophied skeletal muscle, and ectatic blood vessels [1].Treatment options available are sclerotherapy, intralesional steroids, cryotherapy, and surgical resection. Although no specific guidelines for the treatment have been established [4]. Sclerotherapy provided only temporary pain relief [5] and had very little overall benefit for most patients [6]. Cryotherapy showed significant relief in pain, increased patient satisfaction, and even reduced limitation of movement [7]. Although with cryotherapy, several recurrences were noted and had to be treated by surgical resection. Surgical intervention appears to be the best mode of intervention [5]. Persistent pain, muscle contracture, limitation of range of motion, and failure of other methods of treatment are the indications for surgical intervention [6]. Surgical resection with appropriate

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Author's Photo Gallery


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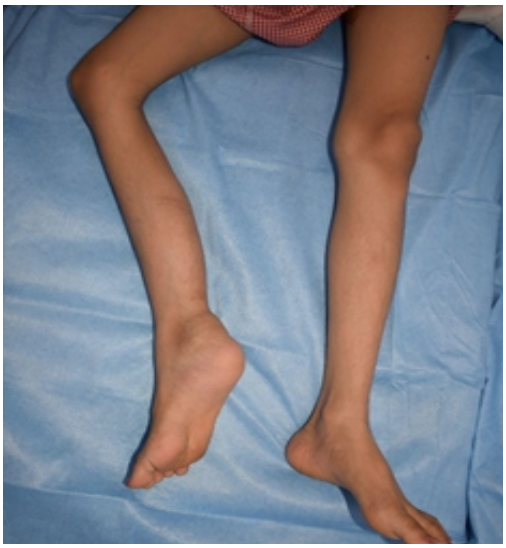

Figure 1: Right knee kept in flexion for ease. Mild fullness in the calf, Ankle in fixed equinus deformity.

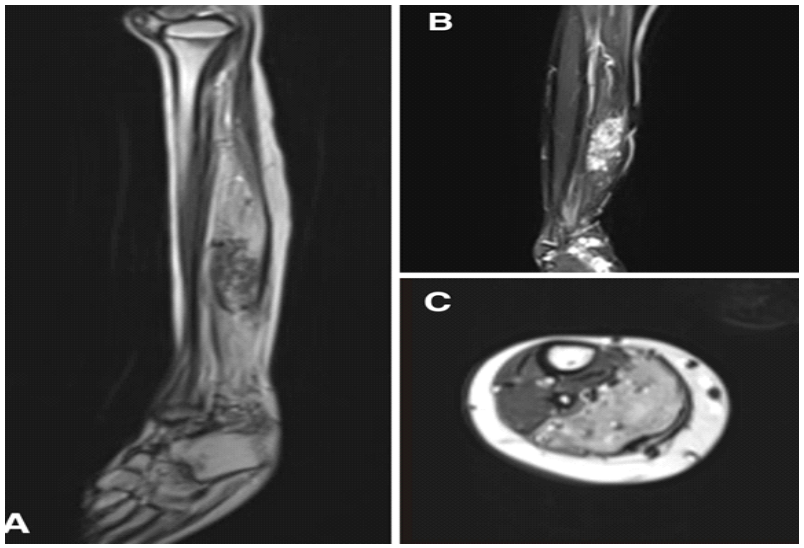

Figure 2: (a) T2- weighted sagittal image showing lesion in the gastrocnemius complex, (b) T2- weighted TIRM (Turbo inversion recovery magnitude) coronal image showing the lesions in calf and ankle, (c) T1 weighted axial image through the mid leg.

reconstruction may be helpful in salvage of limb function [8]. Mutations in phosphatidylinositol-4,5-bisphosphate 3-kinase catalytic subunit alpha (PIK3CA) genes were elucidated in some of the FAVA cases. These gain of function mutations lead to excessive activation of the mammalian target of rapamycin (mTOR) pathway, which promotes angiogenesis and lymphangiogenesis [3].

Case Report

An 11-year-old girl was referred from the interventional radiology department of our institution with a history of the right calf and foot swelling which was intensely painful to touch. Child had been keeping the knee in flexion and ankle in equinus to get relief from pain. She was a toe walker and had a fixed deformity in her right ankle. There were no such lesions elsewhere in the body. For the same complaints, she had undergone sclerotherapy 2 years back. However, the lesion recurred and symptoms failed to improve. Hence, she underwent an embolization procedure 1 year back. There was no resolution of symptoms following the second procedure as well. On examination, there was fullness in the calf and medial aspect of the ankle. Dilated veins could be seen all over the right thigh, leg, and foot. The child would not allow any sort of palpation in the area. The knee was kept in flexion for ease of pain but range of motion was full. Ankle was fixed in the 60 degree equinus (Fig. 1). There were no neuro vascular deficits in the leg. Other limbs had no abnormality. A contrast-enhanced magnetic resonance imaging (MRI) was done for the lower limb (Fig. 2a-c). It showed 4 × 2 × 7 cm enhancing lesions involving gastrocnemius and soleus muscles showing early arterial enhancement with multiple branches from posterior-tibial, common peroneal artery, and early venous drainage which was suggestive of venous malformation. Another similar lesion was found in the distal Achilles tendon measuring 2. 5 × 2. 1 × 3 cm and medial to the calcaneus measuring 4 × 4 × 2.5 cm. Lesions were stable in comparison to MRI done 8

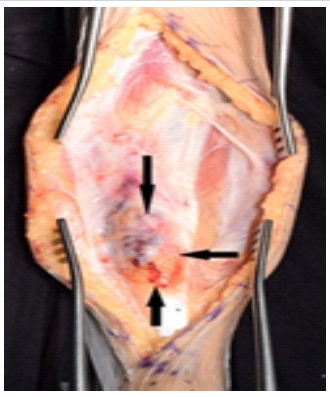

Figure 3: Intraoperative image showing the lesion in the belly of gastrocnemius.

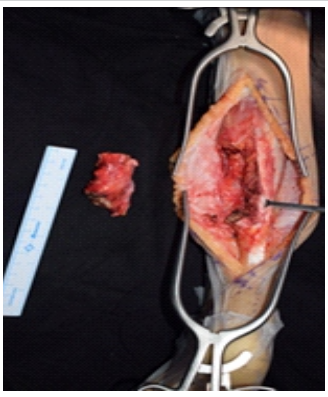

Figure 4: Intraoperative image showing resected specimen and muscle defect.

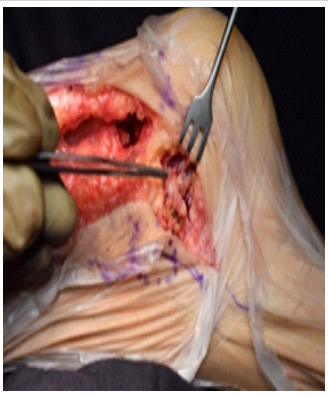

Figure 5: Intraoperative image showing lesion in the ankle.

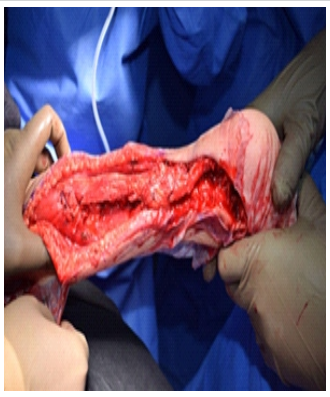

Figure 6: Complete repair of gastrocnemius muscle with Z lengthening of tendo achilles

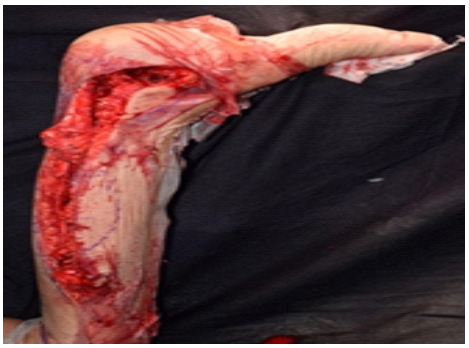

Figure 7: Neutral ankle was achieved.

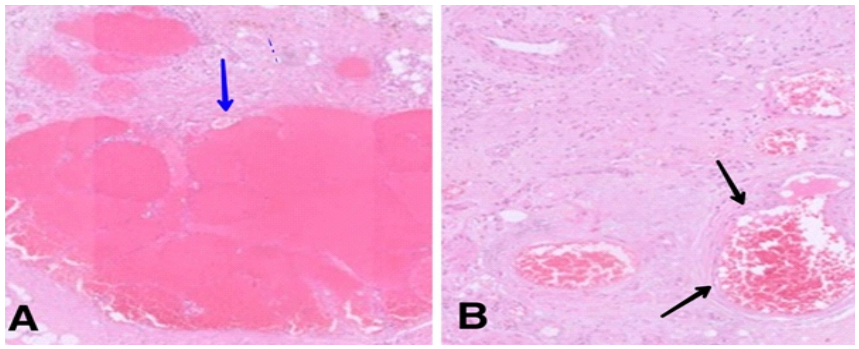

Figure 8: (a) Section showing lobules of adipose tissue with small capillaries in myxoid stroma with small to medium thick walled vessels and (b) section showing fibroadipose and fibrocartilaginous tissue with variably sized vascular channels and cavernous spaces filled with hemorrhage.

months before. Multidisciplinary team meeting involving the interventional radiologist, orthopedic oncologist, vascular surgeon, and pathologist was convened. The clinical correlation to a fibro-adipose venous anomaly was made and because of the poor outcomes of interventional procedures in such cases [6] in the literature, an en bloc resection of the tumor was planned. Pediatric orthopedic opinion was also sought for correction of the fixed equinus deformity in the ankle. Incision was planned mid-posterior over the swelling. The lesion was well distinguishable from the normal tissue. It was firm in consistency (Fig. 3). Resection was done en-bloc. There was a residual muscle gap of 6 cm (Fig. 4). Lesion was excised from the ankle by extending the same incision. The lesion in the ankle was friable and difficult to locate as its location was not intramuscular (Fig. 5). The tendo achilles was lengthened by Z-plasty to achieve a neutral ankle (Figs. 6 and 7). Child was put in an above knee cast post operatively to counter the action of gastrocnemius muscle. Wound inspection was done on day 5 through a window in the cast. Child was made to ambulate on the fiber-cast. Histopathology section of the gastrocnemius lesion revealed fibroadipose and fibrocartilaginous tissue with variably sized vascular channels and cavernous spaces filled with hemorrhage. There was no evidence of malignancy (Fig. 8b). The ankle lesions showed lobules of adipose tissue with small capillaries in myxoid stroma with small to medium thick walled vessels (Fig. 8a). All the findings were in agreement with the fibroadipose venous anomaly. At 1-year follow-up, the repeat scans showed no recurrence. The foot was plantigrade and the child was walking with a slight limp. She was referred for gait training.

Discussion

FAVA was defined as a distinct entity which had unique clinical, radiological, and histopathological features, only in 2014 before which it was incorrectly termed "cavernous hemangioma" [9]. FAVA was classified under the "unclassified vascular anomalies" category by the ISSVA in 2018. Other conditions in this category are: Intramuscular hemangioma, angiokeratoma, sinusoidal hemangioma, acral arteriovenous "tumor," multifocal lymphangioendotheliomatosis with thrombocytopenia/cutaneous visceral angiomatosis with thrombocytopenia, and phosphatase and tensin homolog hamartoma [2]. The most of the patients are older children or teenagers. Khera et al. [4] found that females were predominantly affected by FAVA with a female to male ratio of 4:1. The lower limbs were affected more than the upper limbs. The calf was the most commonly involved site, followed by the thigh. As in the most cases of FAVA, our patient too is a young female presenting with equinus deformity and toe walking but with multiple tender masses in the lower limb. The pain in FAVA is continuous as opposed to the intermittent pain seen in other types of vascular abnormalities [5]. One of the masses was in the right calf while the other was present at the right ankle, medial to the calcaneus. The equinus deformity and the toe walking are said to be the result of fibrotic process occurring in the gastrocnemius muscle, while pain may be due to the contracture of muscle itself or additional causes such as thrombophlebitis, phlebectasia, and involvement of nerves. Post-operative outcome was observed to be worse in cases with direct nerve involvement [10]. Due to common features, FAVA shares with other vascular disorders such as hemangiomas and soft-tissue tumors, diagnosis is often delayed or incorrect. Hence, awareness regarding its distinct clinical and imaging features is crucial to initiate early treatment. MRI is useful to elucidate FAVA [11]. In our case, hyperintense masses were seen in both the calf and the ankle on T2. Histopathology of the specimen is necessary for confirmation of diagnosis which demonstrates presence of fibrofatty and lymphoplasmacytic infiltration in the muscle along with atrophied skeletal muscle. Mosaic gain of function mutations of PIK3CA were identified in 62.5% of patients by Hori et al. although they might not necessarily be involved in the onset of FAVA. p.E542K,

p.E545K, and p.Q546K in the helical domain and p.H1047R in the kinase domain were some of the mutations that were discovered. These mutations activate the phosphatidylinositol-3-kinase/mTOR pathway which phosphorylate 4EBP1 and S6K1 leading to excessive angiogenesis and lymphangiogenesis. Activation of 4EBP1 and S6K1 through mTOR independent pathways may be the root cause of abnormal vessels in FAVA [3].

Sclerotherapy has been increasingly reported as being less effective in cases of FAVA as it acts only on the venous component but not the fibrofatty component. While it may reduce the pain, it has no effect on the muscle contracture, hence, making surgical resection a more appropriate method of treatment as in our case [9]. Cryoablation was found to reduce symptoms of swelling, pain, and skin hyperesthesia without any major complications. Sirolimus, an mTOR inhibitor, was found to be useful in rapidly reducing symptoms of pain and improving quality of life in two patients by Erickson et al. [12]. While surgical resection improves deformity, movement restriction, and pain relief, it has a corresponding risk of procedure related morbidity due to extensive removal of tissue. Damage to surrounding tissue is imminent as it is difficult to find the optimal surgical plane for excision of the mass.

Conclusion

As per our knowledge, this is the first documented case with multiple masses of FAVA in a single As per our knowledge, this is the first documented case with multiple masses of FAVA in a single patient. The clinical and radiological findings correspond with the diagnosis of FAVA. Histopathology of all the masses fits the confirmatory diagnostic criteria of FAVA which include the presence of fibrofatty infiltration into the muscle along with vascular abnormality.

Clinical Message

Fibroadipose venous anomaly is a separate entity which clinicians are not well aware of. It has a well distinguishable clinical, radiological, and pathological picture. Conservative and interventional radiology measures have poor outcomes and increase the morbidity and final outcomes of such patients.

Declaration of patient consent : The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient has given the consent for his/ her images and other clinical information to be reported in the journal. The patient understands that his/ her names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

Conflict of interest: Nil **Source of support:** None

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