Skeletal Radiology
https://doi.org/10.1007/s00256-024-04617-3

TEST YOURSELF: ANSWER

A painful mass infiltrating the quadriceps compartment of a young female

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Received: 13 December 2023 / Revised: 30 January 2024 / Accepted: 5 February 2024

Diagnosis

Fibro-adipose vascular anomaly (FAVA).

Discussion

There have been a few isolated studies or reports published in the literature since the identification of fibro-adipose vascular anomaly (FAVA) as a distinct clinical, radiological, and pathological soft tissue lesion in 2014. This condition is characterized by intramuscular replacement with fibro-fatty tissue, complex vascular malformation, phelbectesia, venous thrombosis, and lymphatic involvement [1, 2]. This lesion was evaluated for a possible FAVA, given the higher predisposition of young females with painful swelling and contractures due to intramuscular mass lesions (Figs. 1, 2 and 3) [3].

Although the more prevalent neoplasm in an arteriovenous malformation was the initial consideration prior to presentation to our institute, a re-evaluation of the diagnosis was necessary due to the persistent pain that resisted conservative management and the imaging findings of a diffuse vascular mass lesion that was not suitable for sclerotherapy due to the absence of tumour-specific feeding or draining vessels.

A core needle biopsy confirmed the presence of a benign intramuscular mass, characterized by perivascular fibrous and adipose tissue, as well as abnormal arterial, venous, and lymphatic channels within the lesion (Fig. 4). Although the factors that contribute to myosteatosis remain largely unacknowledged, recent evidence suggests that the accumulation of intramyocellular lipid is associated with a decrease in muscle strength.

The option of surgical removal remains a viable long-term curative strategy, especially in patients with contractures and large mass lesions limiting the functional range of motion, as was the case with this patient (Fig. 5) [4]. The knee joint was primarily affected by flexion, with a limited range of movement ranging from 10 to 40 degrees, encompassing a 30-degree arc. To limit the difficulty or disability resulting from large muscular resections due to the intramuscular tumour location, near-total excisions may be performed [5]. Following the surgical excision, we were able to relieve her of the contracture from the painful mass and restore functional movement to the knee joint.

Sirolimus has recently been used as an alternative to surgery in the treatment of lesions in children under expert supervision [4]. The utilization of minimally invasive ablative procedures, such as radiofrequency or cryoablation, has been demonstrated to have varying degrees of success in alleviating pain, either with or without embolization or sclerotherapy treatment options, in selected lesions [6].

Investigation performed at Kokilaben Dhirbhai Ambani Hospital and Medical Research Institute, Mumbai, India.

The case presentation can be found at https://doi.org/10.1007/s00256-024-04616-4.

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Published online: 20 February 2024

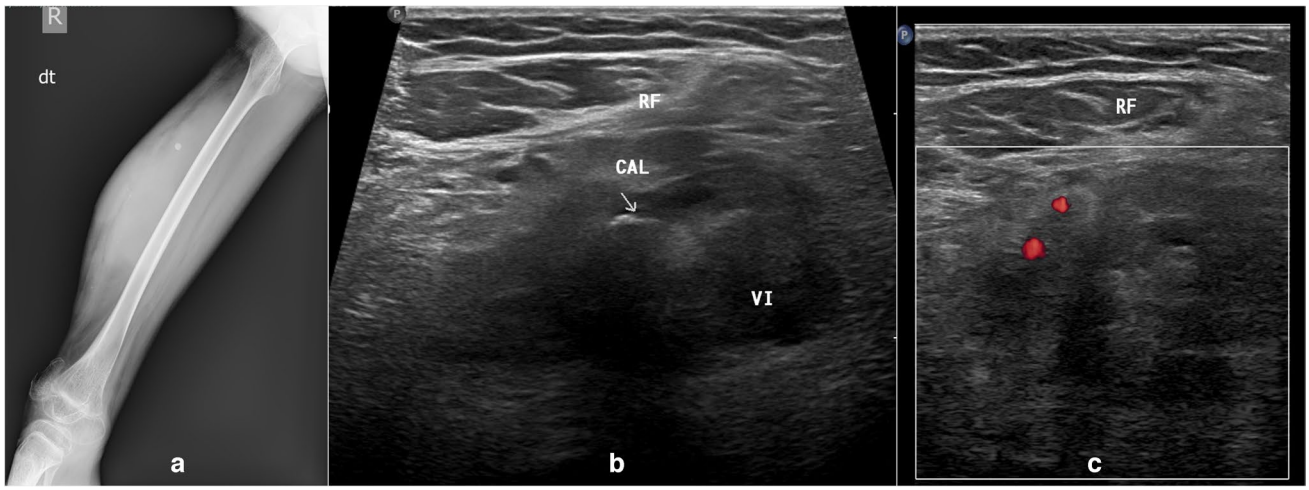

Fig. 1 **a** A lateral view radiograph of the thigh reveals an irregular soft tissue swelling with a few phleboliths in the mid-thigh. **b** Additionally, an ultrasound examination of the anterior thigh reveals an intramuscular echogenic lesion involving the vastus musculature, predominantly the vastus intermedius (VI) with calcification (CAL), with a sparse presence of the rectus femoris (RF) and **c** there is a minimal venous flow within the lesion, which has vascular channels on Color Doppler

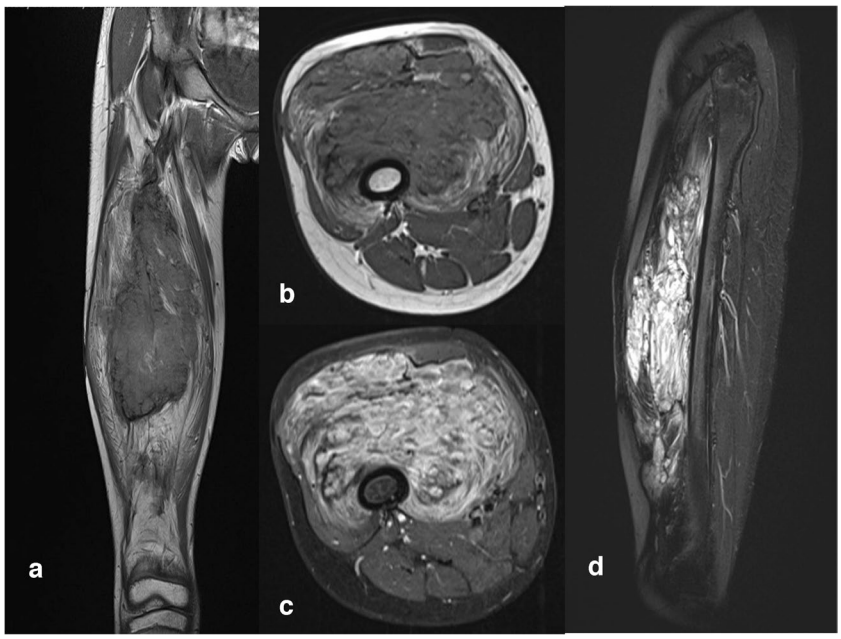

Fig. 2 T1W MRI pre-contrast in **a** coronal and **b** axial and MRI T1W-FS post-contrast in **c** axial and **d** sagittal shows a large heterogeneous intramuscular lesion in the quadriceps compartment of the entire right thigh involves the vastus muscles. The lesion is characterized by regions of intralesional fat and fatty atrophy of the vastus muscles surrounding it, as well as serpiginous enhancement of the lesion on contrast-enhanced images

The next-generation sequencing and molecular testing were employed to identify a clinically relevant PIK3CA gene-associated anomaly in this case [7], as it may be used to distinguish FAVA from close differentials of soft tissue hamartomas or vascular malformation syndromes [8, 9].

FAVA is a very rare and challenging type of vascular tumor, requiring a collaborative effort from multiple specialists to diagnose and manage it. Although intramuscular lesions may result in the loss of muscle bulk through surgical intervention, the correction of deformities and the surgical removal of painful lesions can enhance function.

FAVA is characterized by intramuscular replacement with fibro-fatty tissue, complex vascular malformation, phelbectesia, venous thrombosis, and lymphatic involvement. The diagnosis involves a combination of clinical, radiographic, and pathological suspicion, augmented by genetic mutation analysis for FAVA. This is due to the fact that FAVA has not yet been recognized as a distinct

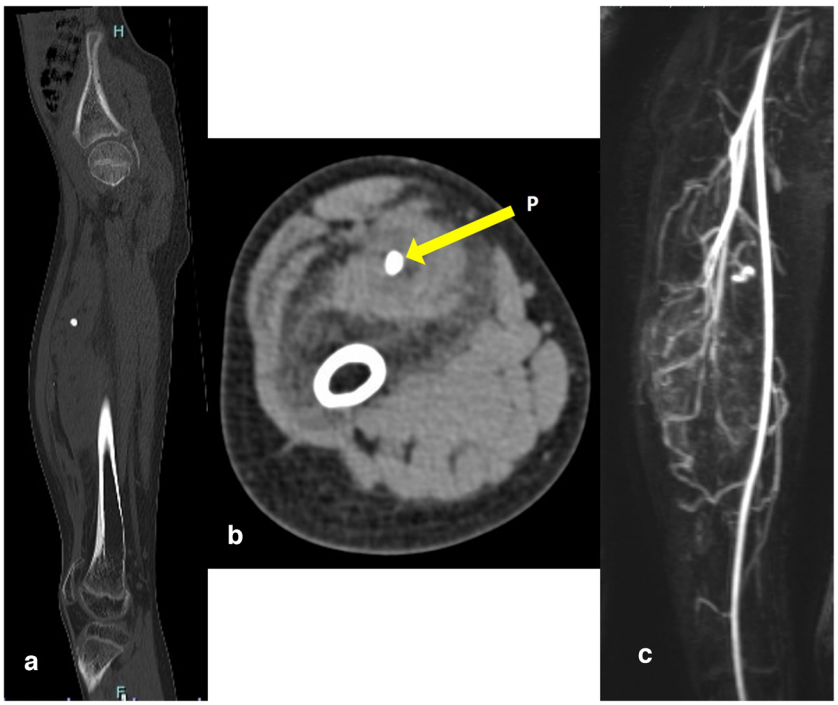

Fig. 3 CT scan in **a** axial and **b** sagittal with **c** post-contrast angiography with a calcific focus which appears to be a phlebolith (P) along with marked fatty atrophy of the surrounding muscles. On angiography, few dilated arterial branches from the profunda femoris artery supply the lesion with small areas of early enhancement without evidence of an early draining vein or discrete nidus

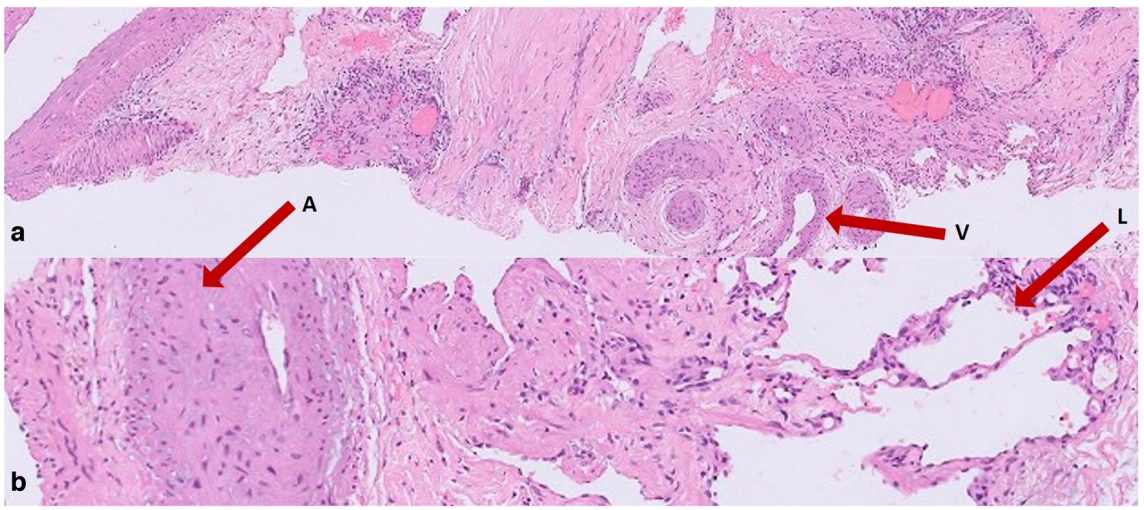

Fig. 4 Biopsy tissue in hematoxylin and eosin section in **a** 10×and **b** 40×magnification depicting an intramuscular mass with fibro-fatty matrix with many variable sized arteries (A), muscularised veins (V) and lymphoid aggregates (L) without evidence of atypia or necrosis

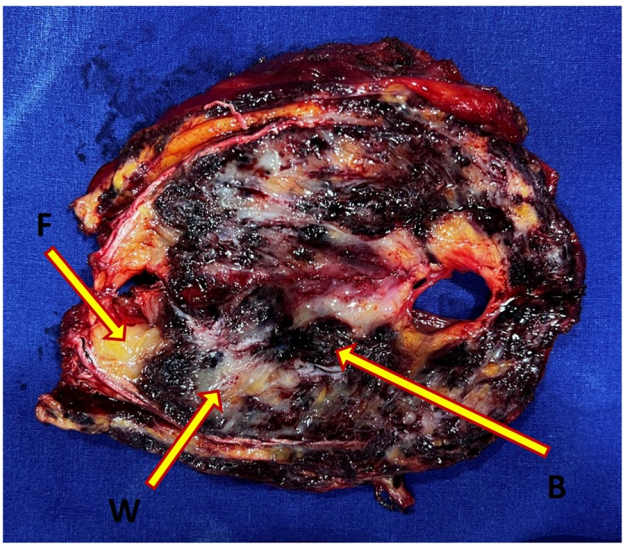

Fig. 5 Cross-section of the surgical specimen after resection with gross description that comprises of a circumscribed mass within the excised muscle and variegated appearance on cut section with firm rubbery white fibrous areas (W) interspersed with blood-filled reddish brown vacuolated cavities (B) along with yellow fatty areas (F) within the lesion

entity within the pathology classification of soft tissue neoplasms. Furthermore, the treatment of FAVA differs from its close differential in an intramuscular arteriovenous malformation.

Declarations

Competing interests The authors declare no competing interests.

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