Clinical Article
Radiology

ROLE OF MRS TO CHARACTERISE POPLITEAL CYSTIC LESION IN A KNOWN CASE OF NF1

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Abstract:

There are multiple differential diagnoses for cystic lesions around the knee. While USG and conventional MRI are sufficient for diagnosis, there are situations where a diagnostic dilemma can occur. We present a case of a cystic popliteal lesion in a child with neurofibromatosis 1. With clinical symptoms of mild pain and swelling, the differentials of neurofibroma and abscess were considered.

We demonstrate the usefulness of MR spectroscopy in diagnosing the lesion as an abscess, rather than a neurofibroma with cystic degeneration.

Key-words: *Spectroscopy, abscess, lactate*

Introduction:

Neurofibromatosis type 1 is a hereditary disease involving the skin, nervous system, bones, endocrine glands and other organs. The classic tumour in this condition is neurofibroma.[1] Neurofibromas have a tremendous heterogeneity of appearance and can mimic other disease entities. This is due to primary germ cell layer abnormalities and the ubiquity of peripheral nerve fibers.[2]

Cystic lesions are ideally evaluated by ultrasound, followed by MRI. In certain cases, even contrast enhanced MRI might not help in arriving at a confident diagnosis. In these situations, the use of MR spectroscopy might be warranted.

We present a case of a child who is a known case of neurofibromatosis type 1, who presented with a swelling in the left popliteal fossa, for whom the final radiological diagnosis was made using MR spectroscopy.

Clinical History:

An 8 year old boy with neurofibromatosis type 1 previously diagnosed with multiple cafe au lait macules and bilateral optic nerve glioma, presented with a history of swelling in the left popliteal fossa since 10 days. No history of fever or past history of trauma to the site .

Physical examination:

The child was afebrile.On local examination a mildly tender ,relatively firm swelling, measuring approximately 1.5x1.5 cm with no local inflammatory changes noted in the left popliteal fossa.The child was unable to extend the left knee.Cafe au lait macules were noted over the chest .A left inframammary swelling measuring 2 x 2 cm was noted which was a biopsy-proven peripheral neurofibroma.

Based on the history and physical findings in a known case of NF1,a clinical suspicion of subcutaneous neurofibroma of the popliteal fossa was made.

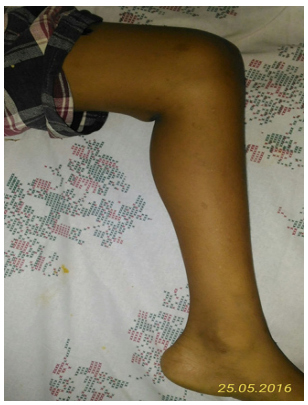
Method:

Anteroposterior and lateral radiographs of left knee, an ill defined soft tissue density in the popliteal fossa. No calcification noted. Bones around knee appear normal. No joint effusion noted.

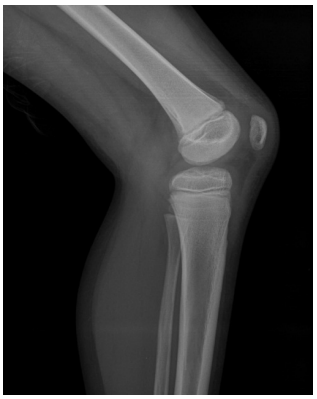
Ultrasound of the popliteal fossa showed an ill defined heterogenously hypoechoic lesion measuring 1.5x1.5 cm in the subcutaneous plane. The lesion showed cystic areas within. No vascularity noted. Popliteal vessels appeared normal.

Contrast enhanced MRI showed a T1 isointense, T2 & STIR hyperintense, peripherally enhancing lesion in the popliteal fossa with surrounding STIR hyperintensity. Close relation of the lesion to the tibial nerve was noted. There was no communication with the knee joint. Multiple smaller well defined lesions showing similar signal characteristics and homogenous enhancement were noted adjacent to the neurovascular bundle in the popliteal fossa.

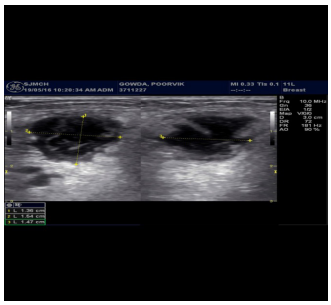
Based on ultrasound and MR imaging features, differential diagnosis of abscess versus neurofibroma with cystic degeneration was proposed. To differentiate between the two, single voxel MR spectroscopy was performed from the centre of the lesion. This showed a single large peak at 1.3 ppm, corresponding to lactate peak. No choline peak was seen. From this, we were confident in placing a diagnosis of an abscess. This was proved on incision and drainage of the collection.



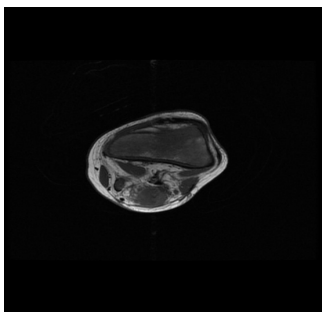
Swelling in popliteal region



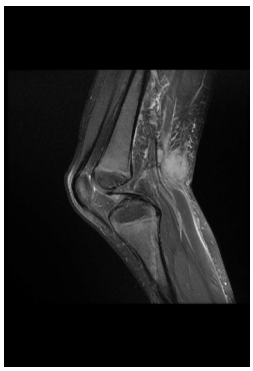
X ray shows soft tissue thickening in popliteal region. Underlying bones appear unremarkable



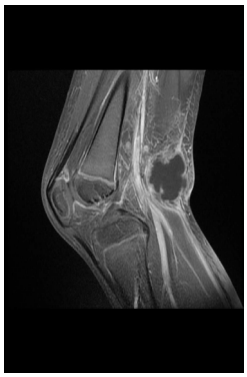
USG showed a complex hypo echoic lesion



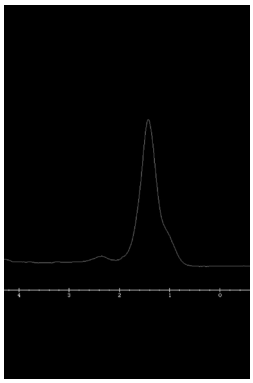
T1W images show a isointense signal focal mass lesion in popliteal fossa



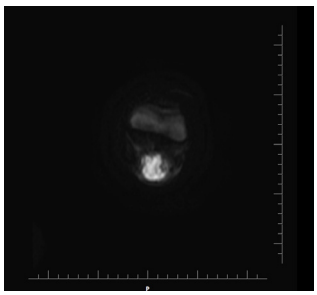
STIR shows hyperintense signal from the mass lesion



Post contrast images show peripherally enhancing mass lesion



MRS shows lactate peak at 1.3 ppm



DWI images show restricted diffusion with loss of signal on ADC images

Discussion:

An abscess can form in the popliteal fossa due to hematogenous spread, direct extension from adjacent tissues or breach of skin by surgery.[3]

Cystic lesions of the musculoskeletal system are evaluated with numerous radiologic modalities, including ultrasound and MRI. MRI is the preferred standard for characterisation and defining lesion extent. A significant limitation of conventional anatomic MRI sequences is the presence of overlap between some inflammatory, benign and malignant lesions. However, MR spectroscopy has the advantage of non invasively obtaining knowledge about biochemical processes within pathological lesions.[4]

Abscess is seen on MRI as a well defined fluid collection, T1 hypointense and T2 hyperintense. A classical finding is the appearance of an enhancing rim with absence of central enhancement. A major differential is tumor necrosis which cannot be differentiated by conventional MRI sequences alone.[5]

Superficial neurofibromas are found in dermis, epidermis as well as subcutaneous fat. The classical "target sign" of neurofibroma is seen only in 21% of superficial lesions.[6] More commonly, these appear as diffuse, poorly defined focus of T2 heterogenous signal.[7]

Intratumoral cystic change in a neurofibroma is a feature of malignant transformation of the neurofibroma. It represents necrosis or hemorrhage.[8] This is particularly common in deep seated neurofibromas in a patient with NF- 1.

Malignant transformation should be suspected in cases where there is rapid change in the size of a lesion or recent onset of pain in a previously painless mass.[9,10]

MRI is the investigation of choice in the evaluation of musculoskeletal lesion, however it is challenged in its ability to correctly characterize the lesions.[11-13] Proton MR spectroscopy provides the answer for characterizing lesion composition and malignant activity.

In the case of neurofibromas, presence of a choline peak is a marker of malignant histology. In abscesses, presence of a lipid-lactate peak at 1.3 ppm is diagnostic.

In our case, the patient was only mildly symptomatic and had other cutaneous neurofibromas, hence creating a dilemma about the nature of the cystic lesion. Axial diffusion weighted imaging showed true diffusion restriction in the centre of the lesion with no restricted diffusion in the walls. Single voxel MR spectroscopy showed a lactate peak at 1.3 ppm, hence confirming the diagnosis of abscess.

This case shows the limitation of convention MR in characterisation of cystic lesions and showcases the relevance of MR spectroscopy in problem-solving the biochemical nature of the lesion.

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