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Neurofibromatosis type I presenting as elongation of neck of femur - an unusual presentation

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ABSTRACT

A 16 years old male presented with pain in right hip and swelling in right thigh since one year. The radiograph of pelvis with both hips revealed elongation of neck of right femur with overlying soft tissue swelling shadow. MRI brain revealed hyper intense foci on T2W images in left parietal lobe. After evaluation the Radiological diagnosis of Neurofibroma of right thigh with elongation of neck of right femur, with hamartoma in left parietal lobe was kept. On basis of clinical and radiological features the final diagnosis of Neurofibromatosis I (NF I) was made. Neurofibromatosis type I is a neuro cutaneous syndrome which present as multi system manifestations. Focal lengthening of bone is an unusual manifestation of neurofibromatosis type I, so whenever there is focal lengthening of bone associated with an overlying mass lesion one should suspect NF I and should look for its cutaneous manifestations.

INTRODUCTION

Among the various neurocutaneous disorders Neurofibromatosis type 1 (NF1) is a relatively common disease which has manifestations of multiple system specially the skin, nervous system and musculoskeletal system[1]. The pattern of inheritance of this disease is autosomal dominant; however it is seen in only 50% of patients, in rest it is due to new mutations. The prevalence rate this disease is 1/3,000 individuals[2]. Plexiform neurofibromas are the most common mode of presentation of this disease. Diffuse neural enlargement and formation of multiple neurofibromas along the course of peripheral nerves is the pathology lying behind the plexiform neurofibromas. It commonly involves the fifth cranial nerve; however any other nerve can be involved[1]

CASE HISTORY

A 16 years old male presented with pain in right hip and swelling in right thigh since one year. On local examination of right thigh region there was a single firm swelling of size 10 x 6 cms on lateral aspect of thigh which was fixed to underlying tissue but the overlying skin was free. There was no deformity; however mild tenderness was present over the mass. On general examination there were cutaneous stigmata of neurofibromatosis type 1 (NF-I) in form of café u lait spots, axillary freckling and cutaneous neurofibromas. The radiograph of pelvis with both hips (fig 1) revealed elongation of neck of right femur with overlying soft tissue swelling shadow. The soft tissue mass lesion was causing remodelling of iliac bone in the form of notching over the acetabulum. The skeletal survey was done to detect other stigmata of neurofibromatosis type 1 however it was unremarkable. CT pelvis (fig 2) revealed well defined hypodense mass lesion below the right gluteus minimus muscle abutting the iliac bone, extending from iliac bone till the neck of femur.

On MR pelvis (fig 3) there was well defined mass lesion below the right gluteus minimus showing heterogenous signal intensity predominantly hyperintense on T2 W images and isointense on T1 W images, abutting the right iliac bone and neck of right femur. The lengthening of neck of right femur was also evidenced. On MR brain (fig 4 and 5) there was well defined mass lesion in left parietal lobe at grey white matter interface appearing hyperintense on T2 W and FLAIR images and hypointense on T1 W images. There was no associated edema. After evaluation the radiological diagnosis of Neurofibroma of right thigh with elongation of neck of right femur with hyperintense lesion in left parietal lobe likely to be Hamartoma was made. The final diagnosis of Neurofibromatosis I was made.

Finally, FNAC of the thigh mass revealed, benign spindle cell neoplasm with myxoid changes.

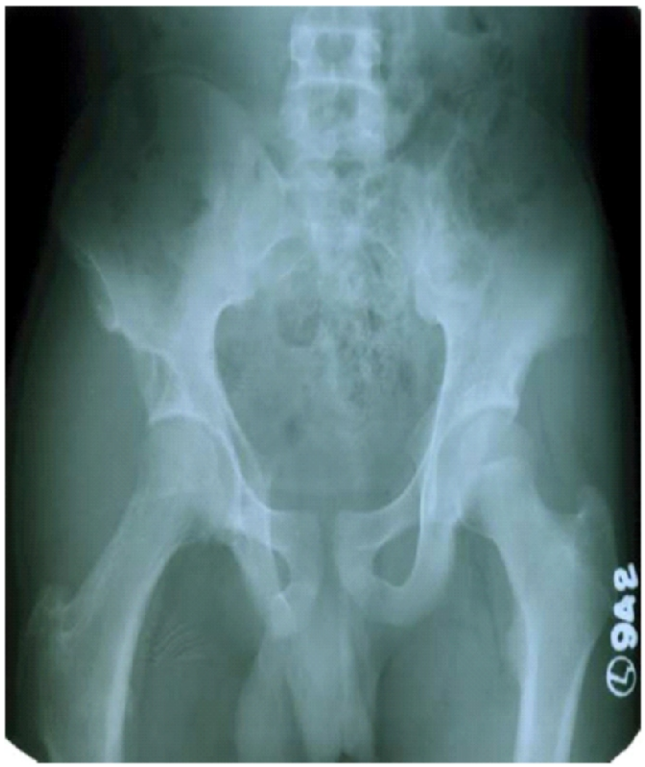

Fig 1 : Radiograph of pelvis with both hips showing elongation of neck of right femur with overlying soft tissue swelling shadow

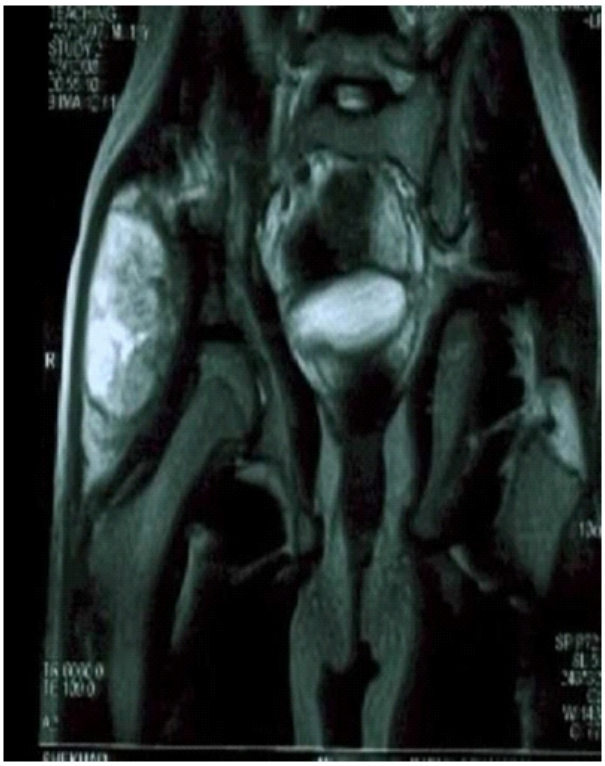

Fig 3 : MR pelvis showing well defined mass lesion below the right gluteus minimus having heterogenous signal intensity predominantly hyperintense on T2 W images and isointense on T1 W images, abutting the right iliac bone and neck of right femur.

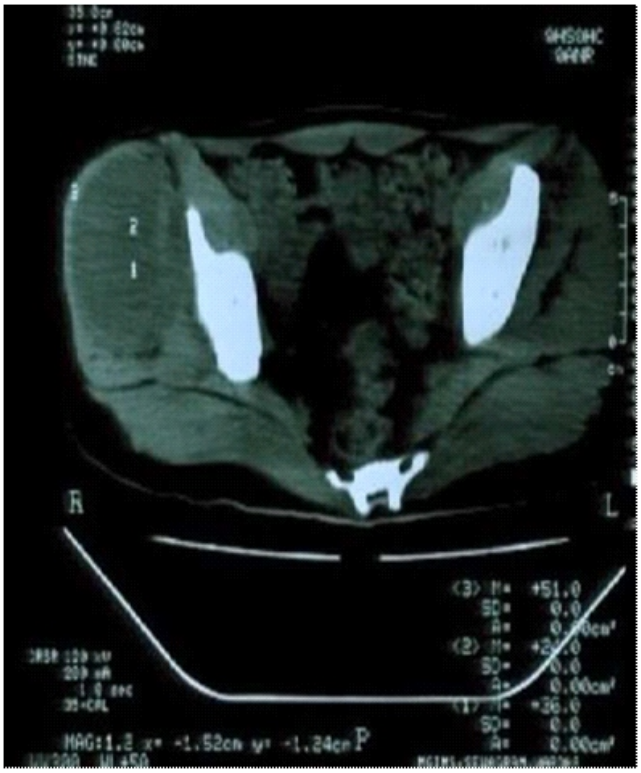

Fig 2 : CT pelvis showing well defined hypodense mass lesion below the right gluteus minimus muscle abutting the iliac bone, extending from iliac bone till the neck of femur.

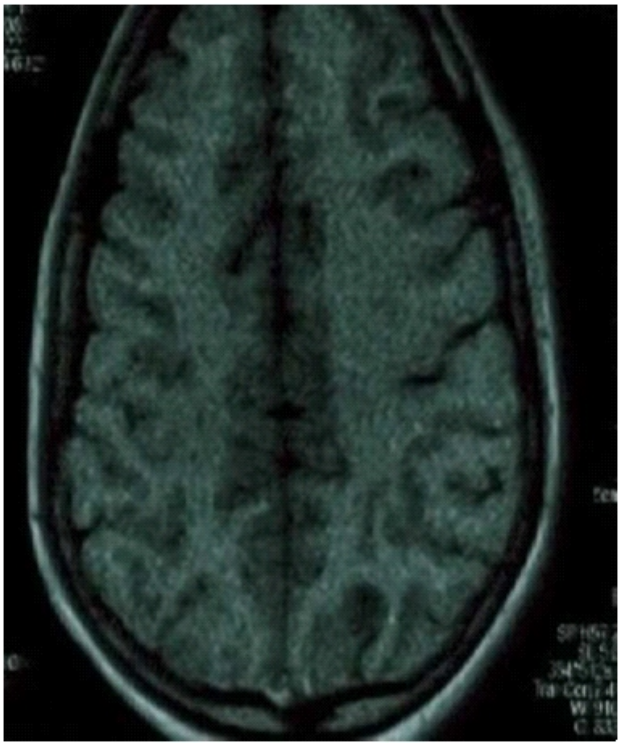

Fig 4 : MR brain showing well defined mass lesion in left parietal lobe at grey white matter interface appearing hyperintense on T2 W and FLAIR images

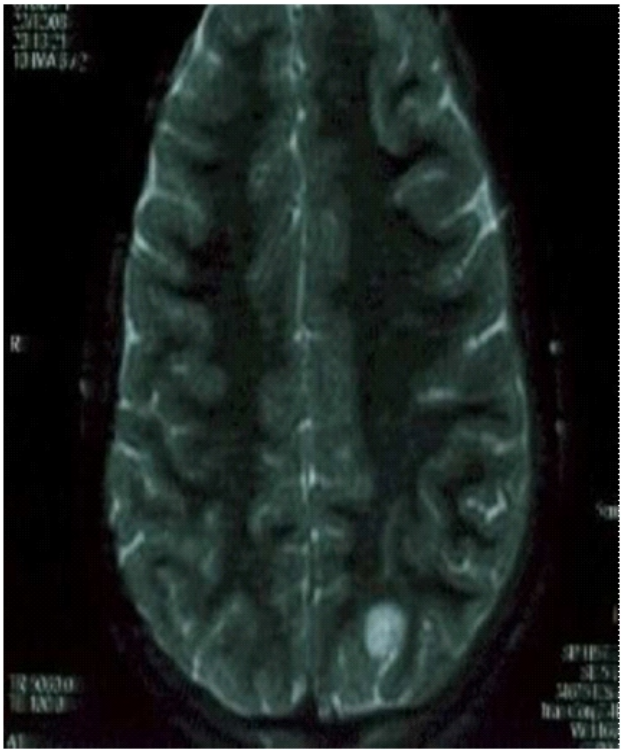

Fig 5 : MR brain showing well defined mass lesion in left parietal lobe at grey white matter interface appearing hypointense on T1 W images with no associated oedema.

DISCUSSION:

After observing the solid cutaneous lesions, Frederick Daniel von Recklinghausen coined the term "neurofibromatosis" for this disease in 1882; since than other various NF manifestations have been recognized. Primarily there is involvement of neuro ectodermal and mesodermal tissue in NF I leading to abnormalities of nervous, musculoskeletal and the cutaneous issues[3].

In our case the neurofibromatosis I presented with musculoskeletal, CNS and cutaneous manifestations in the form of neurofibroma over neck of femur, causing its elongation, hamartoma in the left parietal lobe , café u lait spots and axillary freckling . Some of the above findings resemble the classic peripheral manifestations of NF1which include limb hemi hypertrophy and peripheral nerve neurofibromas. Sometimes NF I mimics other diseases e.g. large peripheral nerve tumour resembling primary soft-tissue sarcoma and multiple neurofibromas resembling rare hypertrophic neuropathies such as Charcot-Marie-Tooth disease or Dejerine-Sottas disease. But the cutaneous findings of NF I easily differentiate NF I from above diseases[1].

Neurologically the NF I can involve the cerebellar peduncles, globus pallidus, pons, midbrain, thalami and uncommonly, the cerebral white matter, appearing hyperintense on T2W images; however they usually represent asymptomatic lesion[4]. In our case we had one such lesion involving the parietal lobe white matter on left side. Shigeki et al detected foci of prolonged T2 without significant mass effect in 32 patients. The characteristics findings of these so-called hamartomas were that they were found only in patients with NF-1 and not in those with NF-2, they were found only in young patients and the lesions in the globus pallidus may be different from the other foci of abnormal signal, because they were normally larger than the other lesions and sometimes hyperintense on short TR/TE images[4]. When these hyperintense foci seen on T2-weighted MR images were evaluated pathologically they consisted of areas of vacuolar or spongiotic change with predominant water content. The hyper intense signals on T2-weighted images occurred because of predominant fluid content of these lesions[2].

Post contrast MR imaging is usually not required in patients with NF I however it can be useful in cases of residual or recurrent acoustic neuroma or meningioma, multiple lesions (e.g., neurofibromatosis or meningiomatosis) and inconclusive or equivocal non enhanced MR imaging [5]. Post contrast MR was not performed in this patient as there was no mass effect and surrounding peri lesional edema.

These neurofibromas appear as hypodense mass lesion on non enhanced CT scans with ill-defined margins. The high water content of the mucinous matrix, the lipid nature of Schwann cells, and the cystic degeneration and incorporation of surrounding adipose tissue into the masses may lead to their hypodense appearance on CT scans. The same factors are responsible for hypointense appearance onT1-weighted, spin-echo MR images and marked hyperintensity is seen on T2-weighted images. Sometimes neurofibromas present a target appearance on T2-weighted images i.e. central areas of decreased signal intensity and a peripheral hyperintense signal. The central hypointensity corresponds to dense collagenous tissue. This target appearance helps to differentiate neurofibromas from other soft-tissue tumors[6]. The CT and MR findings of peripheral neurofibromas in our case were consistent with above mentioned findings.

CONCLUSION

Neurofibromatosis type I is a neuro cutaneous syndrome which present as multi system manifestations. Focal lengthening of bone is an unusual manifestation of neurofibromatosis type I, so whenever there is focal lengthening of bone associated with an overlying mass lesion one should suspect NF I and should look for its cutaneous manifestations.

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