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Case Report

Radiology Section

Elephantiasis Neuromatosa in Neurofibromatosis Type I Mimicking Venocapillary Malformation

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ABSTRACT

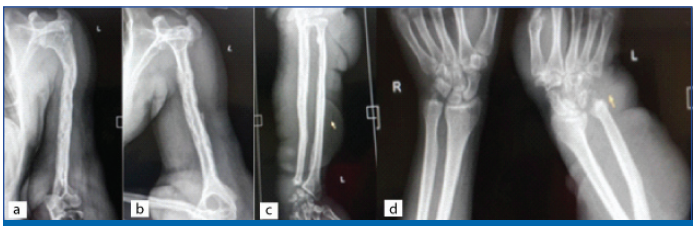
Elephantiasis neuromatosa is a rare clinical manifestation associated with the Neurofibromatosis Type 1 (NF1). A 14-year-old male presented with progressive non-tender swelling of left arm and forearm since birth, axillary freckling, café-au-lait macules and lisch nodules in left iris on slit lamp. X-ray, USG and MRI showed abnormality involving areas of the subcutaneous tissue, superficial and deep muscles and underlying bones of left arm and forearm with diffuse hypertrophy of subcutaneous fat with fatty and fibrotic tissue. Multiple vascular structures were seen throughout the arm and forearm with dilated subcutaneous, superficial and deep veins without evidence of arterial feeders. Possibility of diffuse neurofibroma or veno-capillary malformation was given. USG guided TRUcut biopsy confirmed neurofibroma.

Keywords: Gigantism, Hypervascular neurofibroma, Haemangioneurofibroma Neurocutaneous syndrome

CASE REPORT

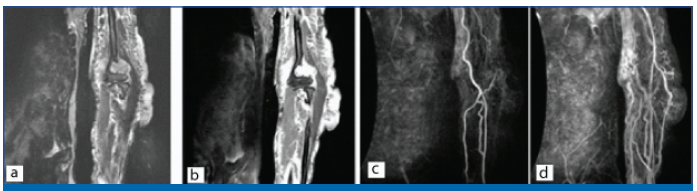
A 14-year-old male presented with progressive non-tender swelling of left arm and forearm since birth. No family history of NF1 or any other neurocutaneous syndrome. Axillary freckling was present. Café-au-lait macules were present, which were >6 and >15 cm in size. Slit lamp examination showed lisch nodules on left side. X-ray showed soft tissue hypertrophy with bony remodeling, sclerosis, lamellar periosteal reaction at humerus, radius and ulna and posterior dislocation of ulna.

Plain X-ray left arm, forearm and wrist showed diffuse increase in soft tissues, lateral bowing at proximal shaft of humerus, irregular, thickened and sclerotic cortex with thinning at places and lamellar periosteal reaction at meta-diaphyseal region of humerus, radius and ulna with enlarged olecranon fossa and posterior dislocation of ulna. The zone of transition was wide [Table/Fig-1a-d].



[Table/Fig-1 (a-d)]: X-ray AP arm, X-ray lateral arm, X-ray AP forearm, X-ray AP bilateral wrists, showed diffuse soft tissues hypertrophy, bony remodelling, cortical irregularity and lamellar periosteal reaction involving humerus, radius and ulna.

USG showed diffuse increase in amount of fatty and fibrotic tissue with numerous vessels in subcutaneous areas as well as coursing through the lesions and areas of loculated collection suggesting lymphedema with secondary fatty and fibrotic hypertrophy. Contrast enhanced MRI left arm and forearm showed altered signal intensity involving areas of the subcutaneous tissue, superficial and deep muscles and underlying bone of left arm and forearm with diffuse hypertrophy of subcutaneous fat with fatty and fibrotic tissue within, more on medial side. Altered marrow signal intensity at humerus, radius and ulna with irregular margin at places and lamellated periosteal reaction was noted. Post contrast study showed diffuse heterogeneous enhancement of the hypertrophied components. The olecranon fossa of humerus was enlarged and filled with moderate enhancing pocket of collection, extending along long bones. Multiple flow voids was seen throughout the arm and forearm, more in posteromedial compartment [Table/Fig-2a,b]. TRICS, phase 2 and TRICS phase 7, show dilated basilic, cephalic, brachial veins and subcutaneous veins without evidence of arterial feeders [Table/Fig-2c,d]. USG guided TRU-cut biopsy showed features of neurofibroma.



[Table/Fig-2 (a-d)]: (a,b) Coronal T2WI, coronal T1FS post contrast shows olecranon fossa of humerus enlarged and filled with moderate enhancing pocket of collection, extending along long bones. Multiple flow voids seen throughout the arm and forearm, more in posteromedial compartment; (c,d) TRICS phase 2 and TRICS phase 7 show altered signal intensity areas involving subcutaneous tissue, superficial and deep muscles and marrow of underlying bone of left arm and forearm with diffuse hypertrophy of subcutaneous fat, cortical irregularity and lamellated periosteal reaction.

DISCUSSION

Neurofibromatosis (NF) is a neurocutaneous syndrome with autosomal dominant inheritance with a birth incidence of 1 in 2500 and a prevalence of 1 in 4000 [1], and is classified into two distinct types that differ in clinical and genetic characteristics, NF-1, a peripheral NF and NF-2, a central NF.

The National Institute of Health (NIH) in 1987 established diagnostic criteria of patients with NF-1, which are neurofibromas (two or more simple, or one plexiform neurofibroma), café–au–lait spots (six or more, >5 mm in greatest diameter in children and >15 mm in adults), Lisch hamartomas in iris (two or more), axillary or inguinal freckling, skeletal abnormalities (sphenoid dysplasias or cortical thinning, with or without pseudoarthrosis), optic gliomas and first degree relative with NF-1. Presence of two or more of these seven criteria establishes the diagnosis of NF-1 [2]. In our patient three of these seven criteria were present.

Elephantiasis neuromatosa is a rare clinical manifestation associated with the NF1. Three types of neurofibroma are classically associated with NF1: localized, diffuse, and plexiform [3]. Plexiform neurofibromas can occasionally lead to gigantism, with massive hypertrophy of the skin, soft tissues and underlying skeleton of

an extremity, known as elephantiasis neuromatosa as named by Virchow [4].

Localised neurofibromas are the most common type of neurofibroma and more frequently involve large deep nerves and are multiple in number. It shows a fusiform growth-pattern. There are certain other like diffuse neurofibromas which are not distinct lesions and extend along connective tissue septa. They encircle rather than destroying the nearby normal structures.

Plexiform neurofibromas are essentially pathognomonic of NF1, affecting approximately 30% of patients and it subcutaneously feels like a "bag of worms" clinically. Plexiform neurofibroma may be also of two types depending on its vascular nature: a normal vascular variety and the hypervascular variety with abnormal ectatic vessels which are more prone for bleeding due to myxomatoid degeneration known as haemangio-neurofibroma [5].

Neurofibromas show characteristically low signal intensity on T1 weighted images and heterogeneous high signal intensity on T2 weighted images and show heterogeneous enhancement following gadolinium administration. Furthermore, MRI study of involved limb may demonstrate the target sign, fascicular sign, and split-fat sign, typical features of NF [6].

Elephantiasis neuromatosa is an uncommon clinical expression of NF1 phenotype and its features are early and excessive growth of the affected limb and it occurs due to neoplastic proliferation of the perineural connective tissue [7]. Signs appear early due to lymphostasis and lymphedema resulting in adipocyte metaplasia and chronic hyperemia induced bone overgrowth and focal gigantism [8]. The differential diagnosis are filariasis, macrodystrophia lipomatosa, lymphangiomatosis, vascular malformation [6], Proteus syndrome, Klippel–Trenaunay Weber syndrome [5]. CT is helpful in evaluation of the bone lesion and extent of the soft tissue involvement. However, MRI is the modality of choice due to better contrast resolution and tissue plane visualisation [6].

Dynamic contrast-enhanced MRA delineates the arterial and venous supplies, it helps in visualising the atypical changes of the vasculature in the affected limb [6]. Post-contrast images show extensive contrast throughout the soft tissue lesion and the large ectatic veins [9].

In our case the lesion was a hypervascular neurofibroma with ectatic vessels that showed heterogenous hyperintensity in T1W and postcontrast enhancement and extensive bony remodeling in agreement with previous literature.

CONCLUSION

Elephantiasis neuromatosa is a rare clinical manifestation of NF1 and MRI can be efficacious tool in the diagnosis and clinical characterisation of early onset cutaneous, subcutaneous and skeletal anomalies.

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