Plexiform neurofibroma with limb hypertrophy
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Section: Musculoskeletal system
Area of Interest: Musculoskeletal system
Musculoskeletal bone Musculoskeletal soft tissue
Procedure: History
Procedure: Diagnostic procedure
Imaging Technique: Experimental
Imaging Technique: Conventional radiography
Imaging Technique: MR
Special Focus: Hyperplasia / Hypertrophy Neoplasia
Case Type: Clinical Cases
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Patient: 4 years, male

Clinical History:

A 4-year-old boy presented for right lower limb mass with lower limbs discrepancy. The mass increased gradually in size causing intermittent throbbing pain. Physical examination showed a solid, mobile, mildly tender mass of the right calf. The boy and his brother have multiple pigmented macules (Fig. 1).

Imaging Findings:

On X-ray the right leg is longer than the left one. Minimal bowing of the right tibial and fibular shafts was noted, as well as an increased soft tissue density. (Fig. 2)

MRI showed a soft tissue mass of the posterior compartment of the right leg. The mass was infiltrating, showing lobulated margins and extending from the distal thigh and popliteal fossa to the plantar aspect of the foot. It demonstrated an intermediate signal on T1-weighted images, a high signal on T2 and minimal enhancement after gadolinium administration. Axial T2-images showed multiple target appearances within the mass. (Fig. 3-4)

Discussion:

Neurofibromatosis is an autosomal dominant multisystem genetic disorder, resulting from deletion/mutation of the NF1 gene responsible of production of a tumour suppressor protein: "neurofibromin". Lack of neurofibromin induces malformations of the skin, skeleton and nervous system. The prevalence of clinically diagnosed cases ranges from 1/2000 to 1/5000. [1]

The skeleton is commonly involved in NF1: spinal deformities, sphenoid and tibial bone dysplasia, excessive bone and soft-tissue growth. [2]

Dysplasia of long bones, most frequently the tibia, is seen in 7.1% of cases. [2]

In general it presents as tibial bowing with cortical thinning, increased risk of fracture and pseudarthrosis. In such cases limb discrepancy results from shortening of the affected limb, however, limb overgrowth may occur without dysplasia as in our case where the long limb demonstrates a soft tissue mass. It is postulated that the tumour-associated hyperaemia somehow induces limb overgrowth. [3]

The diagnosis of NF is clinical. [4] It is made in our case by the presence of three of seven clinical criteria (cafe-au-
lait spots, tibial dysplasia and a first degree relative with cafe-au-lait spots); this clinical context and the above described MRI findings lead to the diagnosis of plexiform neurofibroma (PN).

PN is pathognomonic of NF1, found in 27% of cases. [5] It is an ill-defined infiltrating mass involving the nerve, muscles and adjacent fat, consequently often non totally resectable. [6] PN typically involves the trunk and extremities but also may affect the head, neck, or urinary bladder or even the mesentery. [7]

On MRI it presents as an infiltrative mass of fascicular aspect, with hypo/iso T1 signal, iso/hyper T2 signal with subtle or no enhancement after gadolinium administration. A characteristic MR finding is the "target sign" on axial T2 images: a low signal centre of collagen fibres and a high signal rim of myxomatous material. [8]

Besides the clinical picture, MRI is the diagnostic modality of PN. Angio-CT or MR also help assess the vascularization for any pre-surgical embolisation. [9]

Clinical context is the most important clue for diagnosis. When large, the neurofibroma causes hypertrophy of the affected member giving a presentation called “elephantiasis neuromatosa”. [1]

Differential diagnoses include proteus syndrome, a sporadic disease of tissue and bone with limb overgrowth, vascular malformations and muscular asymmetry [11, 12] and Kasabach-Merritt syndrome that is thrombocytopenia with large infiltrating soft tissue haemangiomas. [13] The first syndrome is unlikely since the patient's disease is familial, the second includes bright T2 significantly enhancing lesions unlike our case.

**Differential Diagnosis List:** Plexiform neurofibroma, Venous or lymphatic malformation, Sarcoma, Proteus syndrome and Kasabach-Meritt syndrome

**Final Diagnosis:** Plexiform neurofibroma

**References:**


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Figure 1

Description: Right lower limb overgrowth with brown skin macules in the right thigh. Origin: elie barakat, radiology department, lebanese university
**Description:** Lower limbs discrepancy with mild medial bowing of the right tibial and fibular shafts and increased soft tissue density at the right leg. **Origin:** Elie Barakat Radiology Department Lebanese University
Description: A- Coronal T2-weighted image shows a high signal soft tissue lesion in the posterior compartment of the right leg.
B- Axial T2-weighted image shows the characteristic target sign within the mass (red arrows). Origin: elie barakat radiology department, lebanese university
Description: A- Pre and B- Post-contrast administration coronal T1-weighted images show a minimally enhancing low to intermediate signal intensity mass of the right leg posterior compartment. Origin: elie barakat, radiology department, Lebanese university