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A RARE CASE OF DIFFUSE NEUROFIBROMA – A CASE REPORT

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ABSTRACT

Diffuse neurofibroma is the rarest form of neurofibroma which is more common among children and young adults. The lesions are ill defined and infiltrative in nature and usually affects the skin and subcutaneous tissue. We present a case of 10 year old boy whose initial clinical and radiological evaluation did not reveal the diagnosis and it was only after the histopathological study we concluded the case as neurofibroma

Key Words: Diffuse neurofibroma, Café au lait patches, Spindle cells, Meissners corpuscles

INTRODUCTION

Neurofibroma is a benign nerve sheath tumour of the peripheral nervous system. It has three distinct types namely localized, diffuse and plexiform type. This case report is of a patient who presented with diffuse neurofibroma, which is also the rarest form. The aim of presenting this case of diffuse neurofibroma is to emphasize the importance of clinical and histopathological examination in its diagnosis. Also, this condition when detected early can be monitored on a periodic basis to detect complications of the same and improve patient outcome.

CASE REPORT

A 10 year old boy presented with swelling in the right leg for seven years. The swelling was initially small and gradually got bigger to attain the present size. There was pain for two days after being hit over the swelling while playing. There was no history of bleeding or discharge from the swelling. There was no history of fever or other similar swelling elsewhere.

It was interesting to note that over the past seven years patient has been thoroughly investigated for recurrent pain in his leg. In 2010, patient underwent medial epiphysodesis of right ankle for calcaneo valgus deformity correction in a government hospital. CT and MRI taken in a private hospital showed periosteal based lesion along the tibial diaphysis possibly sub periosteal hematoma or

sub periosteal abscess. The patient continued to have pain inspite of an extended treatment for infection. A repeat MRI in 2012 showed small resolving abscess with haemorrhagic contents. No other new clinical finding was noted that time.

On this occasion, the general condition was fair. The swelling on the right leg was 15x7cms on the anterolateral aspect extending from 3cms below the knee to 4cms above the medial malleolus. The swelling was not warm and non tender. The surface was smooth and firm in consistency. It was not mobile and fixed to the underlying bone. There was no scars and no dilated or engorged veins. There was limb lengthening which measured 2cms. The limb girth on the right side was 3cms more than the left. There was full range of movements at both the knee and ankle joints. Examination did not reveal any neurovascular deficit.

Routine blood investigations were normal. X-ray of the leg showed sclerosis, periosteal thickening, anterolateral bowing and deformity of the bone. CT angiography revealed residual hematoma probably infective in aetiology.

Excision biopsy was planned. Per-operatively, a soft tissue mass was found and was sent for histopathological examination along with a piece of bone.

Histopathological examination of the soft tissue mass showed a neoplasm composed of elongated, spindle-

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shaped cells with wavy, buckled nuclei and (Fig A,C) some with oval nuclei arranged in a haphazard fashion (Fig B) entrapping lobules of mature adipose tissue and skeletal muscle fibres. Some areas show differentiation towards Meissner type of tactile corpuscles and scattered ectatic blood vessels were also seen (Fig D). Neurofibromatous tissues show immunoreactivity with S-100 protein (Fig E,F). It is a sensitive, but non-specific marker of benign nerve sheath tumors.¹

Sections from the bone showed normal mature lamellated bone. (Fig G) Based on these findings, it was reported as diffuse neurofibroma.

Following the tissue diagnosis, the patient was examined retrospectively for café au lait patches and multiple café au lait spots were seen over the trunk. (Fig H) .An ophthalmology opinion was sought to detect Lisch nodules which was however absent. CT brain and trunk showed no abnormalities. The post operative course was uneventful.

DISCUSSION

Neurofibromas are benign nerve sheath tumors in the peripheral nervous system, which frequently occur as a part of Neurofibromatosis 1. Neurofibromatosis is a genetic disorder that may be associated with multiple neurofibromas. They arise from the non myelinating type of Schwann cells that exhibit biallelic inactivation of the NF1 gene that codes for the protein neurofibromin. Neurofibromas are classically described as localized, diffuse, and plexiform types² depending on the growth pattern.

Localized and plexiform neurofibromas are well known subtypes. The localized form is the most common and they usually present as skin-colored dome-shaped or pedunculated papules that display the pathognomonic “buttonhole” sign. They can be sporadic or Neurofibromatosis associated. Plexiform neurofibromas are larger and more extensive tumors that are usually associated with Neurofibromatosis.

Diffuse neurofibroma is an uncommon but distinctive subtype of neurofibroma. They are usually solitary lesions and are progressively enlarging and infiltrating type, simulating malignant tumors. They occur most commonly among children and young adults. They typically involve the skin and subcutaneous tissues of the head and neck^{3,4,5} . Trunk and extremities are also commonly involved and constitute more than one third of the lesions.⁶

They are poorly defined lesions that spreads extensively along connective tissue septa and between fat cells. They surround rather than destroy adjacent normal structures

^{3,4,5}. This lesion has also been termed “paraneurofibroma” as the tumor extends beyond the confines of the perineurium⁷.

They differ from conventional neurofibromas by the presence of uniform matrix of fine fibrillary collagen, less elongated schwann cells and clusters of meissner body-like structures which are the characteristic feature of this lesion.

The incidence of neurofibromatosis among patients with diffuse neurofibroma has been reported to be approximately 10%⁴ . For the diagnosis of Neurofibromatosis type 1, two or more of the cardinal signs are required:

- 6 or more brownish discolorations on the skin, more than 5mm in diameter in pre-adolescents and greater than 15 mm in adolescents and adults.
- 2 or more neurofibromas.
- Freckling in the axilla or groin.
- Optic glioma
- 2 or more Lisch nodules, or small masses on the iris.
- Bone lesions like sphenoid dysplasia.
- A first degree relative with Neurofibromatosis 1.

In our case, as solitary neurofibroma and multiple café au lait spots are present there is a high potency for the development of Neurofibromatosis I.⁸

Malignant transformation rarely occurs in solitary diffuse neurofibromas⁹. Pain or enlargement of a neurofibroma may herald malignant transformation. Surgical excision is not mandatory as malignant transformation is rare². Resection is performed when the tumor is severely disfiguring or severely compromises function¹⁰.

Partial or complete surgical excision is done for large neurofibromas. The extensive and infiltrative nature of the lesions precludes complete surgical resection.^{2,3,10} Recurrence may develop even after complete excision because of its infiltrative growth pattern in Diffuse neurofibroma. As there is a relatively high incidence of recurrence and the potential development of neurofibromatosis, yearly follow-up is warranted.^{3,7,10}

CONCLUSION

Most often neurofibromas are of histopathological diagnosis especially in solitary lesions because of the indefinite clinical characteristics. Relevant history taking, clinical examination and ophthalmic and radiological investigations are needed for the preoperative suspicion. In order to exclude neurofibromatosis, special attention is needed for the detection of café au lait spots.

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MICROSCOPIC PICTURES

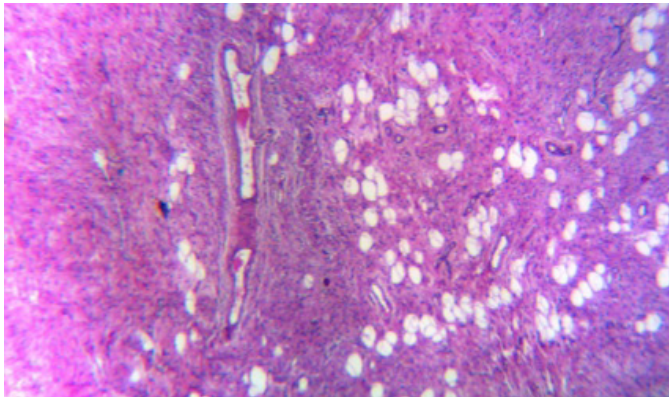


Figure A: Spindle shaped cells entrapping adipose tissue

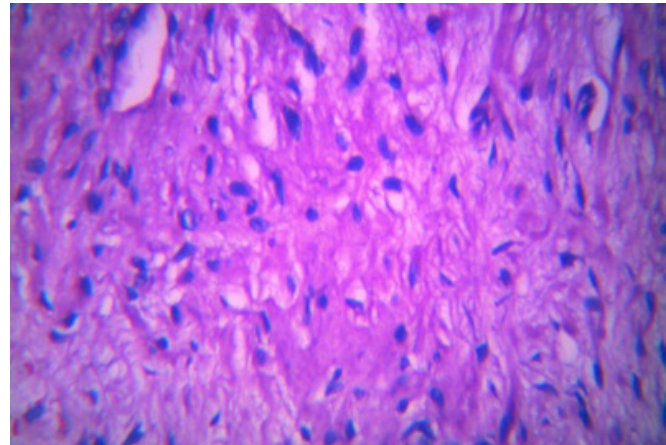


Figure C: Spindle cells with wavy buckled nuclei

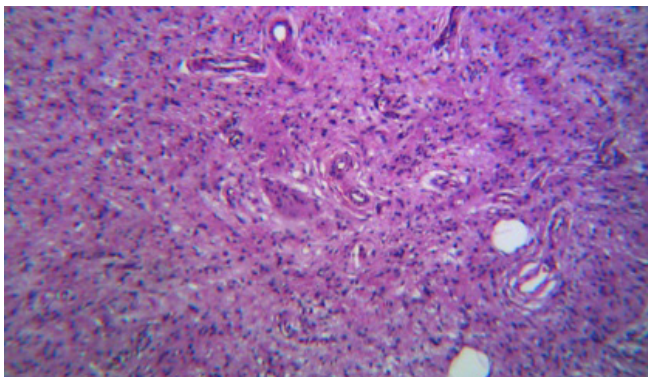


Figure B: Spindled cells in haphazard fashion

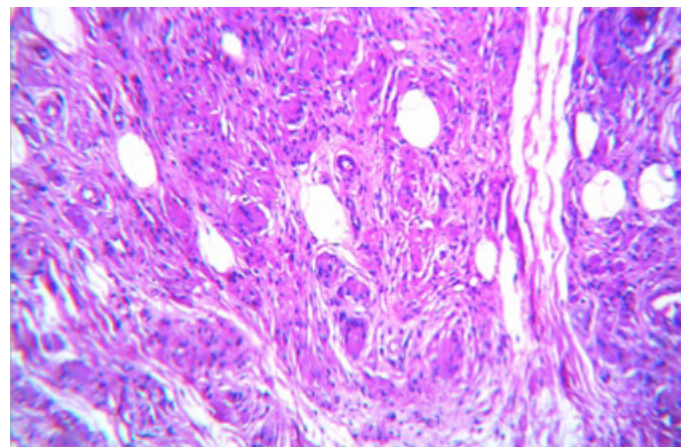


Figure D: Areas showing Meissner type of tactile corpuscles

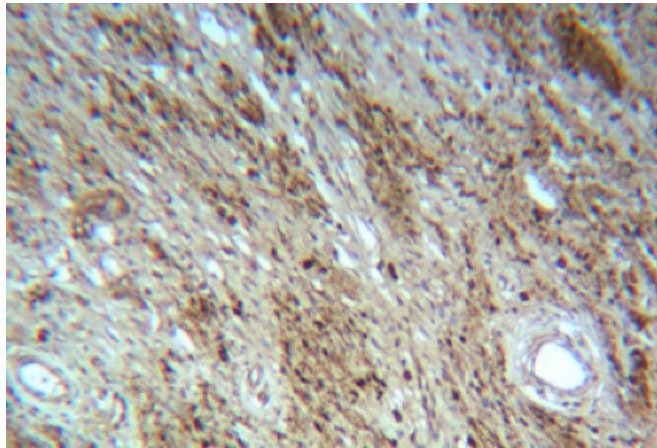


Figure E: S100 positivity

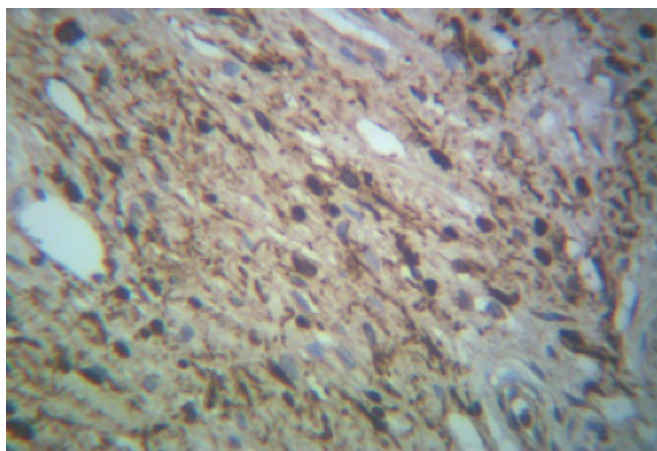


Figure F: S100 Positivity



Figure G: Normal mature bone

E,F -S 100 Positivity
G - Normal mature bone

CLINICAL PICTURES



Figure H: Café au lait spots



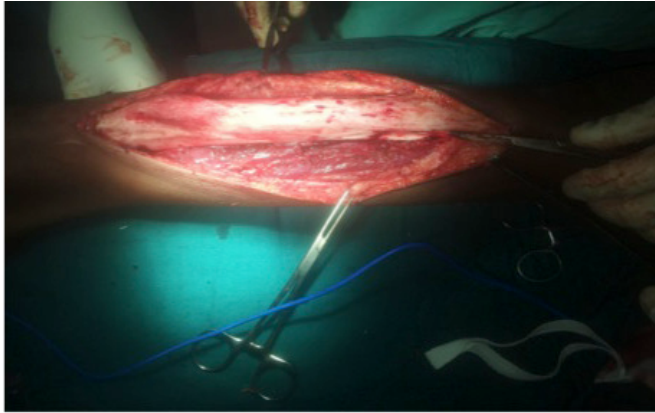
Figure I: Café au lait spots

LEGENDS FOR FIGURES

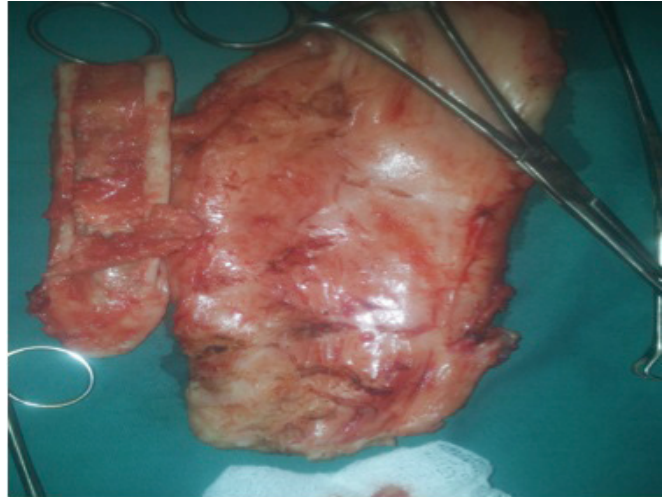
A, B, C-Photomicrograph shows diffuse neurofibroma cells with spindle shaped and buckled nuclei present within fibrillary collagen background. Scattered fat cells are present. **D** - Photomicrograph shows multiple laminated bodies that resemble Wagner-Meissner tactile corpuscles and are characteristic of diffuse neurofibroma.



CT ANGIO GRAPHY



PER OPERATIVE PICTURES



SOFT TISSUE MASS WITH BONE