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Case Report

Plexiform neurofibromatosis of left upper arm: a rare case report and review of the literature

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ABSTRACT

Giant plexiform neurofibromatosis is a relatively rare manifestation of Type 1 neurofibromatosis. This condition leads to gross disfiguration along with functional disability. Due to the unpredictable natural history and high vascularity of these lesions, the optimal management is still not well defined. We are presenting a 20 year male of Plexifrom Neurofibromatosis of left upper limb. The aim of this rare case report is also to discuss the management difficulties encountered.

Keywords: Plexiform Neurofibromatosis, Cfe-au-lait spot

CASE REPORT

A 20 years short statured old male presented to us with a gradually progressively increasing swelling over his left upper limb in whole length for last 10 years. Patient has positive family history (plexiform neurofibromatosis) in the right lower leg of the grandfather's brother.

Patient had a small hyper pigmented area with unusual long hairs at the lateral aspect of the left arm since birth.10 year back, this tuft of hair shed off and followed by an increase in size and volume of the hyper pigmented area. Since then, the swelling gradually has been increased to the present size.

Patient is normotensive with normal behaviour and examination of the patient's left arm revealed a soft, non-tender, diffuse swelling from shoulder joint to proximal fingers level, freely mobile over the underlying tissues. The largest diameter of this swelling is 15 cm at elbow level (Figure 1).There was grade-2 power in all the extensor and flexors of the arm without sensory deficit.

In addition, patient has multiple hyper pigmented macules with smooth margins over the trunk (café au lait macules). He also had multiple soft nodules in the skin multiple cutaneous turnours (mollusca fibrosa) which were widely dispersed over the back (Figure 2).

There were no Lisch nodules of iris in slit lamp examination of both eyes and the patient has no freckle like macules in both axillae and inguinal areas (Crowe's sign). A clinical diagnosis of Neurofibomatosis type I with plexiform variant of left arm was made on the basis of NIH guideline mentioned below.

In the investigations, X-ray right arm revealed pseudoarthosis with radial and ulner bowing (Figure 3). No adrenal abnormality in ultrasound/ CT abdomen was found. A biopsy from lesion on right hand showed a

whorled proliferation of spindle shaped cells consistent with neurofibroma.

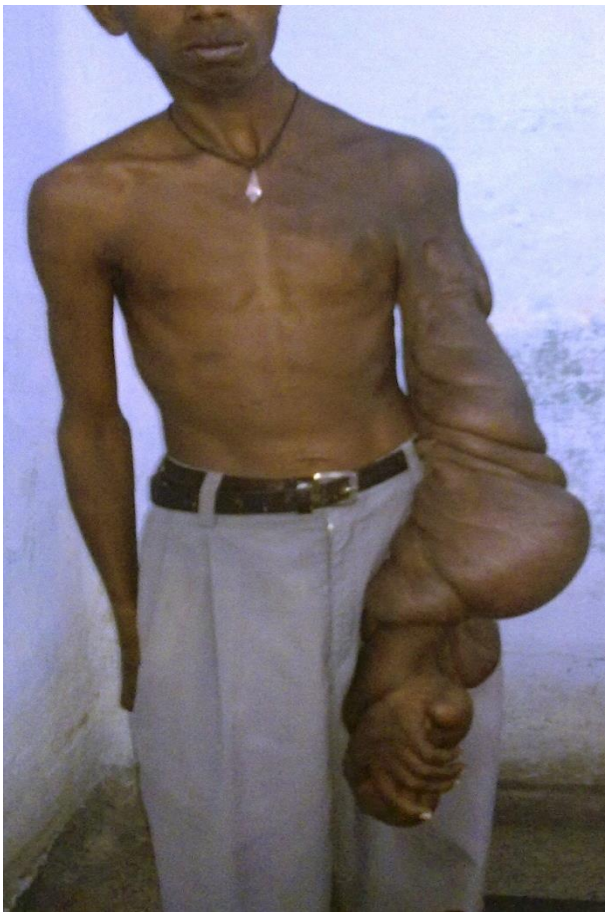

Figure 1: Left arm plexiform neurofibromatosis

Staged surgical excisions were done at an interval of 2 months. In first operation done for diffuse swelling of left hand area, there was profuse bleeding which was managed by electrocoutry and tourniquet with difficulty. In the next operation, we used large vascular clamps along with tourniquet and electro-coutry for the control of bleeding with great success.

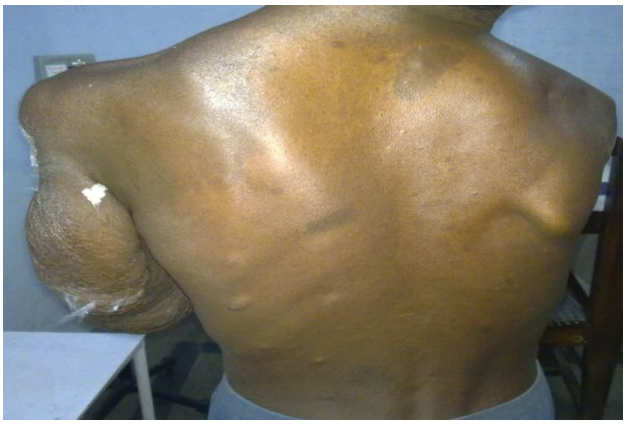

Figure 2: Back of patient of multiple cutaneous turnours (mollusca fibrosa).

For orthopedics and functional disabilities of the left arm, the patient is still being managed by our senior orthopaedician and Physiotherapist. We have also put the patient on vitamin D and calcium orally.

DISCUSSION

Neurofibromatosis is a genodermatosis of neuroectodermal origin characterized by multiple cutaneous turnours (mollusca fibrosa), pigmented 'cafe au lait' macules, axillary freckles, lisch nodules in iris and variable involvement of central nervous system.[1] The genetic defect is localised to chromosome 17 which encodes a protein known as neurofibromin which plays a role in intracellular signalling. The neurofibromin is a negative regulation of the Ras oncogene and is transmitted in an autosomal dominant pattern.[2] The incidence is 1 of 2500-3300 live-births regardless of race, sex or ethnic background.[3]

Plexiform neurofibroma is reported to occur in 26.7% of patients with type I neurofibromatosis.[5] Plexiform neurofibroma presents as a diffuse and elongated swelling along the course of a nerve trunk/plexus. These tend to infiltrate into deeper structures like fascia, muscles and bone. Plexiform neurofibromas present at, or soon after, birth as areas of hyperpigmentation, thickening of the skin and hair excess.[6] The optimal management of plexiform neurofibroma is not well defined and surgery is often delayed until significant disfigurement has occurred.[7]

The National Institute of Health (NIH) has created specific criteria for the diagnosis of NF-1.

Two of these seven "Cardinal Clinical Features" are required for positive diagnosis.[4]

- 6 or more café-au-lait macules over 5 mm in greatest diameter in pre-pubertal individuals and over 15 mm in greatest diameter in post-pubertal individuals
- 2 or more neurofibromas of any type or 1 plexiform neurofibroma
- Freckling in the axillary or inguinal regions
- Optic glioma
- 2 or more Lisch nodules (iris hamartomas)
- A distinctive osseous lesion such as sphenoid dysplasia or thinning of the long bone cortex with or without pseudarthrosis
- A first degree relative (parent, sibling, or offspring) with NF-1 by the above criteria.

The aim of this report is to discuss the difficulties occur in the surgical intervention of this rare condition. There were a lot of difficulties in managing the excess bleeding. We had used large vascular clamps along with the use of electro-coutry. We could not use Tourniquet because of fear of impending fracture of humerus bone of affected side.

The natural history of these lesions is variable with some remaining superficial and asymptomatic throughout life and some progressing into large invasive disfiguring lesions.[8] Friedrich et al, classified these lesions based on the magnetic resonance image (MRI) appearance-superficial, displacing and invasive.[8] Superficial plexiform neurofibromas remain within the upper layer of the skin, usually not involving major. The displacing type develops in deeper layers of the skin or within the body but does not invade adjacent muscles or skin. In contrast, invasive plexiform neurofibromas infiltrate multiple tissue planes to involve muscle and bone. These are much more difficult or impossible to resect. It is not known if a superficial lesion can progress into the invasive type and it is not clear if early surgical intervention can halt or slow the progression of these tumours. Friedrich et al.9 recommended MR delineation and early excision of superficial lesions thus preventing possible progression. In a series of 9 children there was no recurrence in 4 years of follow-up.[9] Resection and debulking of invasive plexiform neurofibromas is associated with a high rate of recurrence. In one paediatric series, complete resections developed recurrence in 20% and incomplete resections had a recurrence of up to 45%.[9] A common theme is the vascularity of these lesions and their abnormal propensity to bleed. Mukherji compared them to angiomas and also commented on the friability of the vessels.[10] We were also encountered with this bleeding problem which was controlled by use a tourniquet and large vascular clamps.

The life-long risk of malignant transformation for plexiform neurofibromas quoted in the more recent reviews is 2-5%.[5,6]

Due a lack of natural history data and the unpredictable growth patterns of plexiform neurofibromas, it remains difficult to decide about the best time to intervene surgically.[7] It would appear that timely intervention could limit the disfigurement and morbidity associated with large lesions.

Such rare cases need a sort of national and international data registry to facilitate patient monitoring and the development of appropriate treatment protocols along with long follow-up. The genetic counselling of these rare cases is also important.

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