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Plexiform Neurofibroma in a Young Patient: A Case Report

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Abstract: Plexiform neurofibromas arise as bulging and deforming masses from multiple nerves, also involving connective tissue and skin folds. It represents an uncommon variant (30%) of neurofibromatosis type 1 (NF-1). Due to the soft and diffuse nature of plexiform neurofibroma, it is often difficult to distinguish it from a vascular malformation or a lymphangioma. Thus requires thorough clinical and histopathological examination and imaging of the lesion. Because of its consistency it is compared to 'a bag of worms'. We report a rare case of a 19-year boy who presented with a progressive skin deformity which initiated in early childhood. On clinical examination there were multiple neurofibromas and café-au-lait macules on the trunk and arms. Histopathological examination of biopsy sample showed overgrowth of peripheral nerve components and connective tissue. Two diagnostic criteria for NF-1 (plexiform variant) were met. The patient did not accept to undergo genetic testing. USG of the local lesion confirmed the presence of a deforming mass with mixed ecogenicity and increased vascularity. Surgery is the mainstay of the treatment. Excision of the lesion was done followed by skin grafting. Diagnosis of plexiform neurofibromas is usually made clinically, especially if classical hallmarks of NF-1 are present. Therapy is surgical, aiming at resecting deforming masses and cancerous tissue when malignant transformation occurs.
Keywords: Plexiform neurofibroma, Bag of worms, Café-au-lait macules.

INTRODUCTION

Neurofibromatosis type 1 (NF-1) is a rare autosomal dominant disease (1/3000 subjects). It is caused by mutations of the *NF1* gene, which is located at chromosome 17q11.2. It is characterized by multiple skin lesions such as café-au-lait macules and axillary freckling and tumor growth along nerves, called neurofibromas[1]. Plexiform neurofibromas is an uncommon variant of NF-1 in which neurofibromas arise from multiple nerves as bulging and deforming masses also involving connective tissue and skin folds. Hence the clinical description of lesions is as “bags of worms.”

We report a rare case of plexiform neurofibroma, which presented with classical hallmarks of NF-1 disease. Finally, we discuss clinical findings, diagnosis, and therapy of this rare deforming disorder.

CASE REPORT

A 19 year old boy was admitted for evaluation of a progressive deformity of skin over neck and left shoulder that began in early childhood (at around 5 years of age). His medical history was non-significant and none of the relatives was known to have similar skin disease.

On physical examination the left side of his neck was deformed by a bulging and soft mass involving the neck, left shoulder, left upper chest, left upper arm and left side of upper back, with sparing of the face and right side (Figure1 A&B). There was no restriction of neck movement or vision.

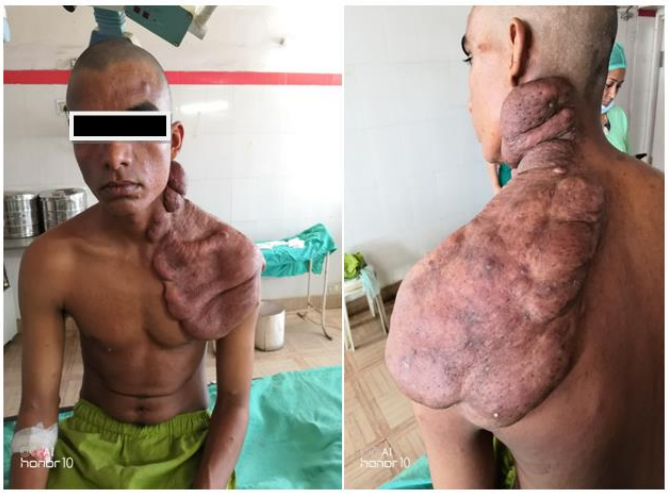

Fig-1 (A) **Fig-1 (B)**

Figure showing plexiform neurofibroma

Skin examination also revealed multiple neurofibromas and café-au-lait macules on the trunk and arms (Figure 2 A,B,C &D).

Routine laboratory tests were normal. USG of the local lesion confirmed the presence of a deforming mass with mixed ecogenicity and increased vascularity. Histopathological examination on biopsy samples showed overgrowth of peripheral nerve components and connective tissue (Figure 3).

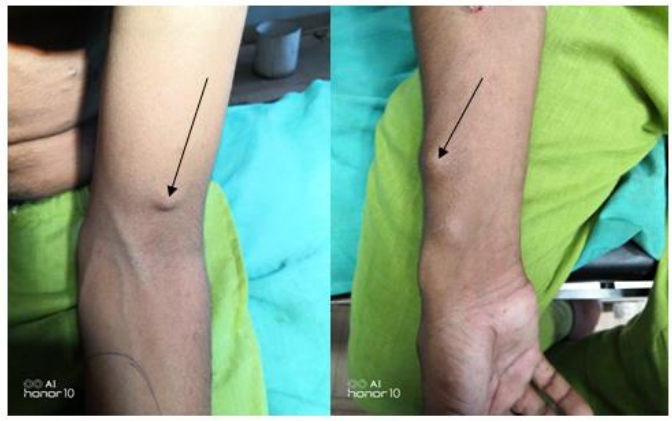

Figure 2 (A) **Figure 2(B)**

Figure 2(C) **Figure 2 (D)**

Fig-2(A&B): Showing multiple neurofibromas all over the body, (C&D) showing multiple café au lait spots

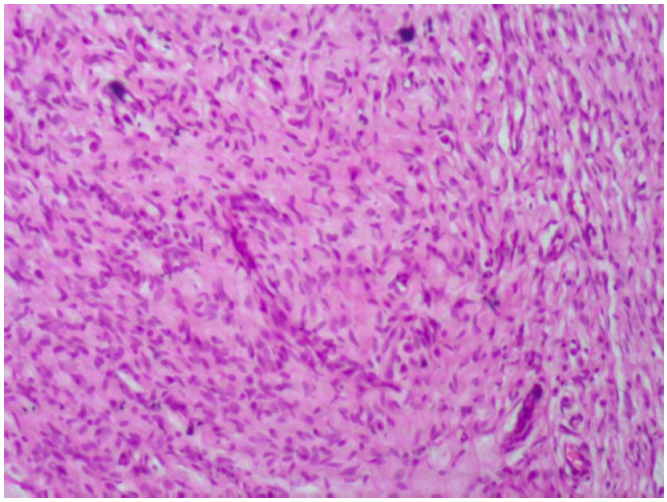

Fig-3: Histological features are suggestive of plexiform neurofibroma

Two diagnostic criteria for NF-1 (plexiform variant) were met. The patient did not accept to undergo genetic testing. Resection of the skin lesion was done followed by skin grafting (Figure 4). Post-operative period was uneventful.

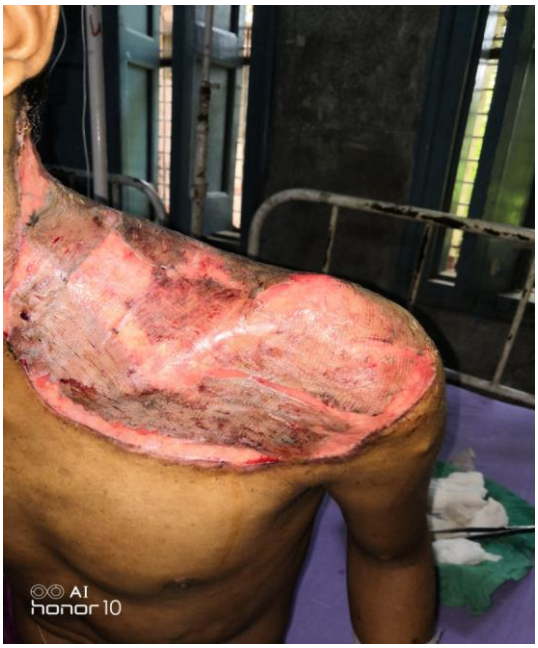

Fig-4: Postoperative image after resection and skin grafting

DISCUSSION

Plexiform neurofibroma is a pathognomonic feature of neurofibromatosis type I. It is uncommon and occur almost exclusively in about 30% of patients with neurofibromatosis-I. These lesions develop early in life and tend to transform to malignant peripheral nerve sheath tumors (MPNST) [2]. The most common cause of mortality in these types of neurofibromas is the malignant transformation occurring in 2% to 16% of cases [2].

These are benign tumors. It spreads out either just under the skin or deeper in the body. They originate from nerve sheath cells, subcutaneous or visceral peripheral nerves and can involve multiple fascicles. The term plexus means combination of interlaced parts or a network. Two types of plexiform neurofibromas have been recognized (i) Diffuse type/ elephantiasis neurofibromatosa and (ii) nodular type[3].

They have poorly defined margins and can arise anywhere along a nerve. They appear on the face, legs, or spinal cord and more frequently involve the cranial and upper cervical nerves. The fifth, ninth and tenth nerves are the most commonly involved cranial nerves in plexiform neurofibromas[4]. Hemifacial hypertrophy can occur secondary to a plexiform tumor involvement which is quite disfiguring [5]. The symptoms produced by the tumor ranging from minor discomfort to extreme pain [6, 7]. The consistency of

the lesion is like that of a 'bag of worms' because of the presence of soft areas interspersed with firm nodular areas and this very consistency was well appreciable in the lesion seen in our patient. They sometimes show increased vascularity causing dangerous bleeding during the surgery and may complicate the procedure. There appears to be an increase in the size of these tumors during puberty and pregnancy [3, 6].

Clinically it can be presented with different symptoms, depending on the locations. The main symptoms are restriction in vision due to ocular motility disturbances from eye involvement, dyspnea and respiratory failure due to upper airway compression, neurological deficits from cranial nerve involvement, depression and other mood disorders resulting from facial disfigurement [8].

When 2 or more criteria, developed by the National Institutes of Health (NIH), are met diagnosis of NF-1 is confirmed [9]. Plexiform neurofibromas are generally diagnosed clinically with appreciation of the typical features. Histopathology is useful to confirm the diagnosis and to exclude malignant transformation. Preimplantation genetic diagnosis could be used as screening method for NF-1 in embryos produced via in vitro fertilization, while chorionic villus sampling or amniocentesis can be used to detect NF-1 in the fetus[8].

Recently, two markers have been identified as potential early risk predictors of developing MPNST. These are insulin-like growth factor binding protein 1 (IGFBP1)) and regulated upon activation, normal T-cell expressed and secreted (RANTES) [10].

Treatment of plexiform neurofibromas is usually surgical, aiming at resection of deforming masses and cancerous tissue when malignant transformation occurs. However, recurrence rate of these masses is 20% despite an appropriate approach[11].

In unresectable and progressive lesions, administration of interferon-α has been reported with good results[12]. However, the prognosis is still unpredictable due to the high risk of progression of the disease and its variable expressivity[10].

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