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A RARE CASE OF OF GIANT PLEXIFORM NEUROFIBROMA

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Abstract : Plexiform neurofibroma is a benign tumor of peripheral nerves arising from a proliferation of all neural elements, pathognomonic of neurofibromatosis type 1 (NF1). It involves single or multiple nerve fascicles that arises from major nerve branches(1,2). Plexiform neurofibromas are among the most common and debilitating complications of neurofibromatosis type 1 (NF1)(3). They account for substantial morbidity, including disfigurement, functional impairment, and may even be life threatening. (4)

Keyword :PLEXIFORM NEUROFIBROMA, LOWER EXTREMITIES

CASE REPORT: A 40yrs old female patient came with complaints of mass in the left thigh for 30yrs. Patient was apparently normal at the age of 10 followed by which patient developed multiple swellings all over the body, one of the swellings in the left thigh that gradually increases in size for past 30yrs and attained present size, patient had difficulty in walking and day today's active due to the large mass. Patient had no H/O pain over the mass, no H/O of ulceration over the mass, no H/O sudden rapid increases of size. Patient also had H/O of multiple hyper pigmented patches all over the body and her daughter also having similar swelling all over the body. Patient was not diagnosed with diabetes mellitus, hypertension, asthma and tuberculosis. On examination patient general condition was fair and her vitals are stable PR -76,BP- 130/80 mm Hg,weight-45kg,height-145 cm, no pallor, not jaundice, kyphoscoliosis present, more than 10 café au lait macule present all over the body with largest of size 4cm,multipl neurofibromas present all over the body **(fig.2)**. On local examination a large pedunculated mass of size 45*30*25 cm extending from posteromedial aspect left upper 1/3rd of thigh up to the level of mid leg**(fig.1)** ,dilated veins are present all over the mass, no decubitus ulcer or scars. On palpation no warm, not tender, smooth surface with variable consistency, skin was pinch able all over the mass. There was no distal neurovascular deficit. No regional lymphadenopathy. Clinical diagnosis was made as neurofibromatosis type 1 with giant Plexiform neurofibroma and the case was subjected for further investigations to find other congenital abnormalities.

Giant Plexiform neurofibroma left thigh

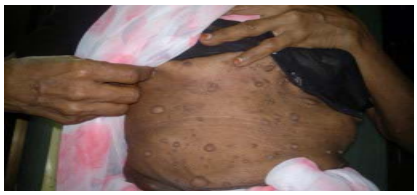

Fig.2

Patient had normal baseline blood investigation, chest X ray shows kyphoscoliosis. MRI left thigh a heterogeneous mass of size anteroposterior length 12cm,transverse length 42cm extending from left gluteal region to the level mid leg, at proximal thigh the mass is infiltrating deep into the muscles and intramuscular plane on posteromedial aspect**(fig.3&4)**

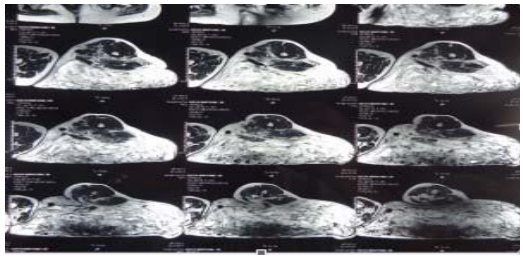

Fig.3

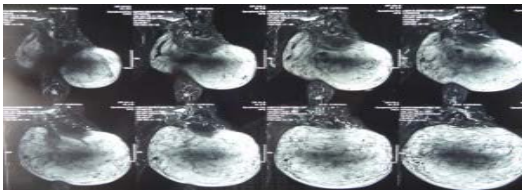

Fig.4
CT chest shows severe kypho scoliosis, multiple neurofibroma in para spinal location with widening of neural foramen with extra spinal extension adjacent to D4 to D9 vertebra **(fig.5)**

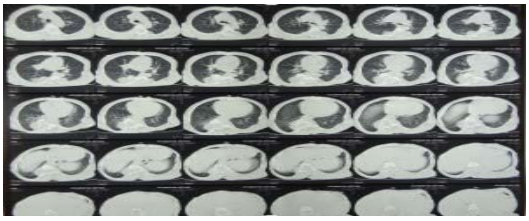

Fig.5
Due to the difficulty in day today activities of the patient because of the mass we planned for wide excision of mass **(fig.6&7)** after clearance from Neurosurgeon, orthopedician. Intra operatively mass was found to be infiltrating deeply in to the semimembranosus muscle which was removed along with the mass. The size of the mass was 45cm in length,35cm in breath, 25cm in transverse length, weight 12.5kg about 31% of total body weight **(fig.8)**. Post operatively patient mobilized on second pod, patient was on regular physiotherapy, patient developed wound infection followed by wound gaping which are treated conservatively and patient discharged on 20th post of period **(fig.9)**. Biopsy report came as Plexiform neurofibroma.

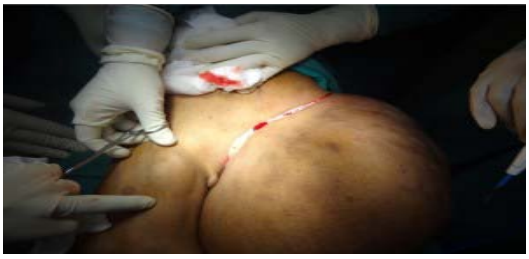

Fig.6

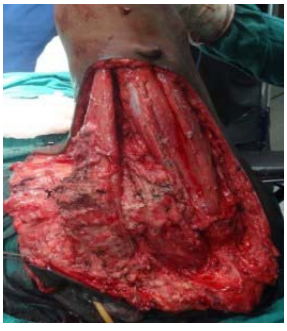

Fig.7

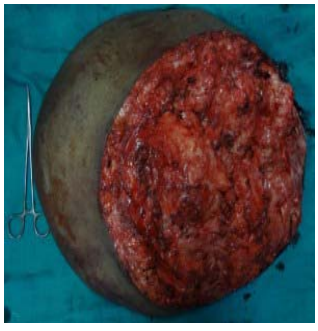

Fig.8

Fig.9

DISCUSSION

The neurofibromatoses are genetic disorders of the nervous system that primarily affect the development and growth of neural (nerve) cell tissues. These disorders cause tumors to grow on nerves and produce other abnormalities such as skin changes and bone deformities The neurofibromatoses occur in both sexes and in all races and ethnic groups.

Types

Neurofibromatosis type 1 (Von Recklinghausens disease)
Neurofibromatosis type 2 Schwannomatosis

Neurofibromatosis type 1

NF1 is the more common type of the neurofibromatoses, occurring in about 1 in 4,000 individuals. Although many affected persons inherit the disorder, between 30 and 50 percent of new cases arise spontaneously through mutation (change) in an individual's genes(5-6). Once this change has taken place, the mutant gene can be passed on to succeeding generations. Previously, NF1 was known as peripheral neurofibromatosis (or von Recklinghausen's neurofibromatosis) because some of the symptoms--skin spots and tumors--seemed to be limited to the outer nerves, or peripheral nervous system, of the affected person. This name is no longer technically accurate because central nervous system tumors are now known to occur in NF1. Neurofibromatosis type I (NF1) is caused by mutation in the neurofibromin gene. An autosomal dominant neurogenetic disorder. Characterized by the presence of multiple benign neurofibromas. Affects the bone, the nervous system, soft tissue, and the skin. Clinical symptoms increase over time. Neurologic problems and malignancy may develop

DIAGNOSTIC CRITERIA for **NF 1** includes

1.Six or more café au lait macules larger than 5 mm in greatest diameter in prepubertal individuals and those larger than 15 mm in greatest diameter in postpubertal individuals
2. Two or more neurofibromas of any type or 1 plexiform neurofibroma
3.Freckling in the axillary or inguinal regions
4.Optic glioma
5.Two or more Lisch nodules (iris hamartomas)
6.A distinctive osseous lesion, such as sphenoid dysplasia or thinning of the long bone cortex, with or without pseudoarthrosis
7.A first-degree relative with NF-1 according to the above criteria Diagnostic criteria are met if 2 or more of the features listed. Symptoms, particularly those on the skin, are often evident at birth or during infancy, and almost always by the time a child is about 10 years old. Neurofibromas become evident at around 10 to 15 years of age. In most cases, symptoms are mild and patients live normal and productive lives. In some cases, however, NF1 can be severely debilitating. Symptoms and severity of the disorder may vary among members of affected families. Plexiform neurofibromas (pNFs) are the second most common type of tumor in individuals with NF1. It is

estimated that up to 50% of patients with NF1 will develop a pNF. pNF are histologically benign tumors that are made up of a variety of cell types including neuronal axons, Schwann cells, fibroblasts, mast cells, macrophages, perineural cells and extracellular matrix materials such as collagen. They grow along the nerve sheath and may involve multiple fascicles and branches of nerve. pNFs can occur in any part of the body and can grow throughout a person's lifetime, often becoming disfiguring, disabling or deadly via compression of vital structures or conversion to a malignant sarcoma, malignant peripheral nerve sheath tumor.

Giant neurofibroma is a poorly defined term used to describe a neurofibroma that has grown to

a significant but undefined size. There are a number of case reports and series found in the literature

discussing giant neurofibromas.(7-15)

PNFs are difficult to manage surgically as they are extensively infiltrative, highly vascularized and

tend to recur. Surgical treatment must be decided judiciously and individualized for each patient.Major

complications of neurofibromas include malignant differentiation and potentially life-threatening

hemorrhage(12-15),fortunately these are quite rare. On the other hand, minor complications such as

local infections or wound problems are very common.(16,17)

Clinical management for the PNF requires a multidisciplinary approach. However, current

treatment options for PNF are limited to surgical intervention. The surgical experience of giant

neurofibromas is limited to case reports.Resection is performed when the tumor is severely

disfiguring or severely compromises functionality.Complete resection is often difficult because of the

extensive and infiltrative nature of these lesions.(18)

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