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Case Report

Von Recklinghausen's or Plexiform Neurofibromas - A rare case

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Abstract

Neurofibromas are benign peripheral nerve tumors composed of proliferating Schwann cells and fibroblasts. It has been estimated that approximately 25% of all neurofibromatosis (NF) are found in the head and neck. Patients with NF type 1 of the head and neck suffer from one of the most dramatic cosmetic disabilities, which may lead to psychological troubles and social segregation. There is no known complete cure for NF. Solitary neurofibroma is a rare tumor of the head and neck region. It is more common in viscera, where it is associated with features of NF1. It occurs most often between the third and fourth decade. These lesions are extremely rare in children.

We report a case of giant, solitary neurofibroma presenting as a progressive mass in neck region in an child. This case is unique in its age of presentation 8 years, over neck on left side, size 12*11*10 cms without any symptoms or neurological deficit. We are presenting our experience in treating this case, with a trial to establish a protocol for dealing with such case as regards; collection and sorting them, assessment of the lesion, dealing with the complications and the plan of treatment. Difficulties encountered during surgery were discussed in particular the management of intraoperative bleeding and post operative brachial plexus neuropraxia.

Key words: Schwann cells, Solitary neurofibroma, brachial plexus, neuropraxia, plexiform neurofibromas

1. Introduction

The aetiology behind solitary neurofibroma is still unknown. Marocchio *et al.*[1] considered solitary neurofibroma to be hyperplastic hamartomatous malformations rather than neoplastic. Neurofibromas are the hallmark of von Recklinghausen's or Plexiform neurofibromas. pigmented lesions of the skin called café au lait spots, freckling in non-sun-exposed areas such as the axilla, hamartomas of the iris termed Lisch nodules, and pseudoarthrosis of the tibia. Neurofibromas are benign peripheral nerve tumors composed of proliferating Schwann cells and fibroblasts. They present as multiple, palpable, rubbery, cutaneous tumors. They are generally asymptomatic; however, if they grow in an enclosed space, e.g., the intervertebral foramen, they may produce a compressive radiculopathy or neuropathy. Aqueductal stenosis with hydrocephalus, scoliosis, short stature, hypertension, epilepsy, and mental retardation may also occur. Plexiform neurofibromas (PNF) are benign tumors which originate from nerve sheath cells, subcutaneous, or visceral peripheral nerves that can involve multiple fascicles. Because of the involvement of multiple fascicles of nerves and tissues and the spread of PNF, there is high risk of neurological and functional destruction when surgical resection is carried out. The surgical interventions are frequently postponed as long as possible from the early childhood. Most cases require repeat surgery since they are limited to debulking as PNF often re-grow later.[2]

Patients with NF1 are at increased risk of developing nervous system neoplasms, including plexiform neurofibromas, optic pathway gliomas, ependymomas, meningiomas, astrocytomas, and pheochromocytomas. Neurofibromas occasionally undergo malignant degeneration to become malignant peripheral nerve sheath tumors.

NF-I is estimated to occur in one of every 3000 births with no sex predilection. PNF usually occur in as much as 30% of patients with neurofibromatosis type I (NF-I) – an autosomal dominant disorder caused by defect of one allele of the tumor suppressor gene, NF1 on 17q and encodes neurofibromin, a tumor-suppressor protein with GTPase-activating activity that inhibits RAS function.

2. Case report

8 year male child presented to OPD complaining of swelling over left side of neck since 4 years which was gradual in onset and progressive in nature at the time presentation swelling measurement was 12*11*10 extending from 4 cms behind chin anteriorly till nape of neck posteriorly from mastoid process superiorly till acromion angle of left shoulder.

On physical examination moderate to severe pallor was present, Café-au-lait spots present but freckles were absent, lisch nodules were present on slit lamp examination.

CBC was normal except for low Hb, urine routine was normal, other all blood investigation were within normal limit. X-ray of the cervical spine AP and Lateral shows increased atlanto axial distance and prevertebral soft tissue swelling noted. Ultrasonography of the neck examined by 14 megahertz frequency linear probe shows 56*55*33mm ill defined hypoechoic solid mass is noted in left side, posterior lateral to the left carotid vessels. Inferiorly the lesion is seen extending upto superior mediastinum. FNAC of cervical lymphnode report shows polymorphus population of the lymphoid cells composed of mature lymphocytes, immunoblasts, centroblasts and centrocytec. FNAC reveals reactive lymphadenitis. Same FNAC is sent to other laboratory which shows moderate cellular material with few clusters of round to oval cells having bland nuclei. Back ground shows frank haemorrhagic material with scattered lymphocytes which are the features of mascular mass or haemartoma. Biopsy of the lesion which was sent to two different laboratories which gave opinion that may be a begin spindle cell tumor with

differential diagnosis of juvenile fibromatosis, fibrous hemartoma of infancy. CT scan of the neck both plain and contrast has been performed which showed large well defined homogenously hypodense lesion measuring 12*11*10cm is seen in the soft tissue planes of the neck on the left side. The lesion is deep to the sternocleido mastoid muscle and posteriolateral to the neurovascular bundle. The carotid and jugular vessels seem to be displaced anteriorly. Most likely suggestive of soft tissue neoplasm. MRI neck without contrast gives evidence of 8.2*12.3*10.2cm size well defined lobulated T2/STIR hyperintense to muscle,T1 isointense solid mass is seen involving anterior and posterior triangle of the neck on left side. Superiorly it extends to the level of C2 vertebra (base of the skull) involving left parapharyngeal space. inferiorly it extends to the D3 level, lies anterior to left clavicle..

The swelling was excised under GA intraoperatively profuse bleeding was present,haemostasis achieved and post operatively patient had neuropraxia of the brachial plexues which recovered spontaneously with physiotherapy.

Fig 1: Pre operative clinical image

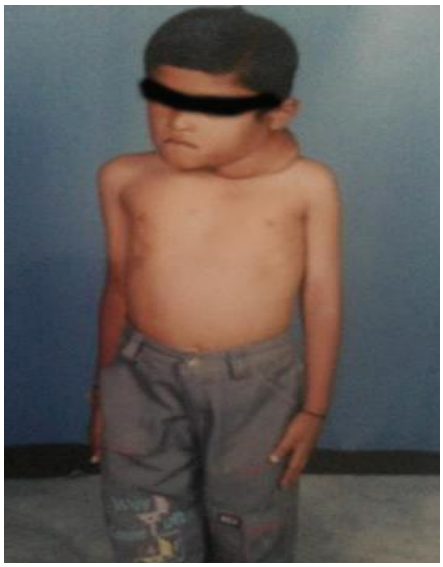


Fig 2: Computed tomography image

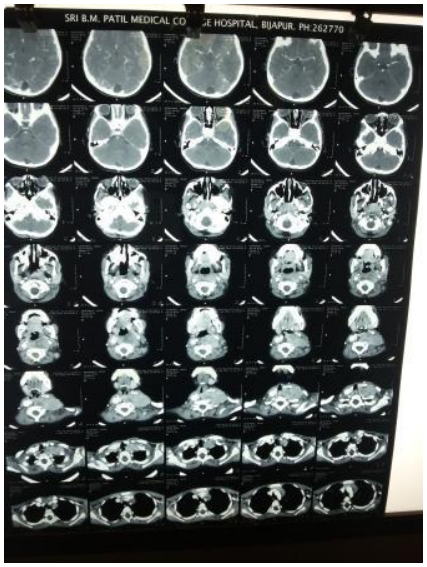


Fig 3: Intraoperative image

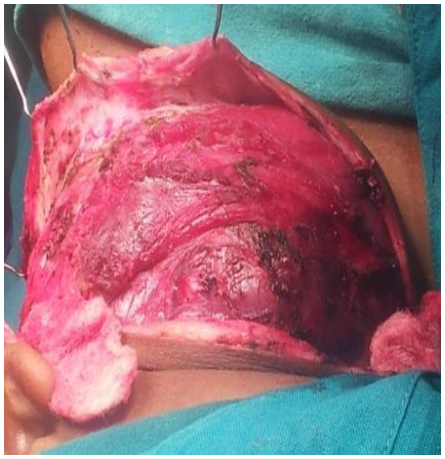
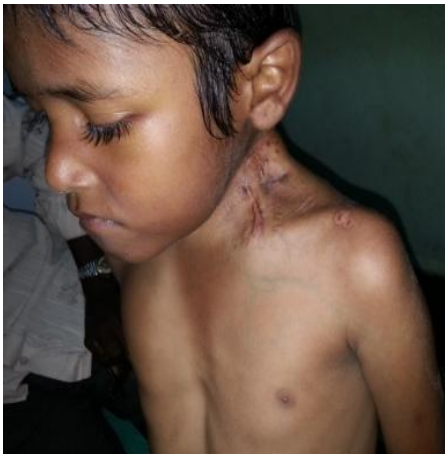


Fig 4: Post operative clinical image after 6 months of follow up



3. Discussion

Plexiform neurofibroma is a rare type of generalised neurofibromatosis, which occurs due to overgrowth of neural tissue in the subcutaneous fat or deeper in the body. It is usually considered to be a hamartoma rather than a typical tumour.They originates from nerve sheath cells; subcutaneous or visceral peripheral nerves and can involve multiple fascicles. PNF are uncommon and occur almost exclusively in about 5-15% patients with neurofibromatosis type 1.[3] Malignant changes in 2.4-29% of patients with neurofibromatosis have been reported[4]

The condition is autosomally dominant, with variable penetration, and presents as multiple nodules of various sizes, which are firm and non-ender, often associated with *café au lait* spots and spindle deformities. There is evidence that only 50% of PNF patients have a positive family history of the disease and the remaining represent spontaneous mutations [5-7] this evidence is consistent with the case being reported. Neurofibromas are characterized by wavy and spindle-shaped nuclei as in the case with our patient.

Café au lait macules develop during the first 3 years of life and they are not present at birth and as our patient is of 8 years skin lesions were observed . Lisch nodules, hamartomas of the iris were noted on slit lamp examination, help to confirm the disease. Axillary freckling and inguinal freckling were not present in our patient as they often develop during puberty.[8]

Patients with NF-1 (25-40%) often have below average intelligence while 5-10% may have mental retardation[9,10]which were not observed in our patient.

The criteria for neurofibromatosis type I are met if a patient has two or more of the following features[10]

a) Six or more *café au lait* macules over 5 mm in greatest diameter in prepubertal persons and over 15 mm in greatest diameter in post-pubertal persons;
b) Two or more neurofibromas of any type or one plexiform neurofibroma;
c) Freckling in the axillary or inguinal regions;
d) Optic glioma;
e) Two or more Lisch nodules (iris hamartomas);
f) A distinctive osseous lesion such as sphenoid dysplasia or thinning of long bone cortex with or without pseudoarthrosis; and
g) A first-degree relative (parent, sibling or offspring) with neurofibromatosis type I,

Imaging is frequently employed in the confirmation of diagnosis of PNF. Superficial ultrasonography may show homogeneous hypoechogenicity or slight echogenicity. Contrast enhanced computed tomography is useful in predicting resectability, detecting metastasis, and evaluating response of the treatment. Magnetic resonance imaging often reveals peripheral hyperintensity and central hypointensity on T2-weighted sequences and marked contrast. Histopathologic distinction of the lesion may not be always easy but provides a confirmatory diagnosis. The application of 67Ga citrate scintigraphy as primary investigation in patients with neurofibromatosis and suspected malignant change has been reported[11].

Although there is no specific therapy for PNF, treatment is often directed towards prevention or management of the disease. Though surgery is the mainstay of treatment for solitary neurofibromas, but recurrence rate is high.

For solitary plexiform neurofibroma of the head and neck, however, radiotherapy may have a role in shrinking these lesions and controlling their growth. Intensity-modulated radiotherapy was shown to be effective in controlling extensive or recurrent juvenile angiofibroma[12] and it should be combined with steroid, retinoic acid, multivitamins, and chemotherapeutic agents to prevents metastasis and post operative symptoms.

4. Conclusion

Bleeding is one of the main concerns in surgery of massive neurofibroma in the head and neck. Preoperative preparations, together with meticulous surgical techniques, can minimize bleeding. Temporary external carotid artery clamping helps to secure haemostasis when other measures have failed to control the bleeding. The disfiguring nature of neurofibromatosis can be psychologically traumatic for most patients and often require good counseling. The patient being reported dropped out of school, after surgery patient is attending his school.

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