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A rare case of neurofibroma present since birth.

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ABSTRACT Neurofibromas are benign spindle cell tumour of peripheral nerve sheath origin. They are composed of cells, stromal mucosubstances, mast cells, Wagner-Meissner corpuscles, Pacinian corpuscles, axons, fibroblasts and collagen. It occurs mostly between the second and third decade of life and are very rare in infants and children. In head and neck about 25 % of all neurofibromatosis (NF) are seen. We here report a rare case of neurofibroma in an one year old male infant who had swelling in the nape of neck more on right side since birth which was gradually increasing in size. He was operated for the same and had no evidence of recurrence.

KEYWORDS : Neurofibroma, infant

Introduction

Neurofibromas are considered common amongst all neurogenic tumors, presenting as soft to firm swelling occurring anywhere in the body. The common site of involvement is skin and soft tissues. They may present as part of neurofibromatosis (von Recklinghausen's disease) or as a component of multiple endocrine neoplasia syndrome (MEN). The subtypes of neurofibroma includes : Plexiform , diffuse cutaneous , focal cutaneous and intraneural . In plexiform neurofibroma irregularly expanded nerve bundles having nodular appearance with prominent myxoid matrix and are associated with NF1. In diffuse cutaneous neurofibroma there is adnexal entrapment with infiltration into fat. While in focal cutaneous type there is no adnexal or fat infiltration. Neurofibromas show S-100, EMA and CD 34 positivity on immunohistochemistry.

Age of presentation is second and third decade of life. However, its incidence ranging from 10 months to 70 years of age is known, though rare. No sex preponderance is present.

We present a rare case of neurofibroma in one year old male infant who presented with swelling in nape of neck right side since birth, which was gradually increasing. He was successfully surgically treated without recurrence till date.

Case report :

An one year old male infant was brought to the oncology outdoor patient department (OPD) by his parents with complains of swelling in the nape of neck right side since birth. But since last 2 months it was gradually increasing in size. On examination a firm swelling was present in the nape of neck right side which measured 7 x 5 cm, with overlying skin being normal (Fig 1). The infant became irritable on pressing the swelling. Various investigations were advised which included complete blood count, fine needle aspiration cytology(FNAC) and computed tomography (CT) scan. Fine needle aspiration cytology was reported as neural tumour . However , excision was advised for confirmation. CT scan showed a well circumscribed mass below skin without extension into surrounding structures. Patient was posted for surgery after anaesthetic fitness. Following surgery the mass was removed and subjected to histopathological assessment . On gross a well circumscribed tumour tissue was received measuring 7 x 6 x 3 cm, lobulated (Fig 2). Cut section of which shows a grey white homogenous areas with glistening. Various sections from the tumour mass was taken for Haematoxylin and Eosin staining (H&E). Histopathology described it as nerve fibres in sheets having serpentine wavy nuclei with indistinct cytoplasm (Fig 3) . No areas of mitosis or necrosis seen, was finally labelled it as neurofibroma, focal type. Patient improved well and is on regular follow up since last one year without evidence of recurrence (Fig 4).

Discussion :

Neurofibromas are peripheral nerve sheath origin tumours benign in nature, are associated frequently with neurofibromatosis type 1. Their occurrence in the body is not rare 1 but present since birth makes it rare. In literature no such cases have been described in infant since birth.

The cause is unknown of solitary neurofibroma. Neurofibromatosis are autosomal dominant disorders. Multiple neurofibromatosis shows neurofibroma to be present on both muosal and skin. They can be classified by many ways one being diffuse and nodular forms. Another feature may be presence of diffuse large pigmentation also known as café au lait.2

Neurofibroma needs to be distinguished from schwannoma as well as malignant peripheral nerve sheath tumors (MPNST). Schwannoma have hypercellular as well as hypocellular areas described as Antoni types A with Verocay bodies and Antoni B areas.3 Schwannomas are also S-100, EMA and CD34 positive tumours. While MPNST also show hypercellular and hypocellular areas with bullet shaped nuclei along with areas of atypia and pleomorphism. Mitosis and necrosis are seen along with heterologous elements like bone, cartilage. Apart from above two tumours neurofibroma needs to be distinguished from palisaded encapsulated neuroma (PEN) , traumatic neuroma and perineurioma4 too. Traumatic neuroma are encapsulated tumours and shows proliferation of both Schwann cells and axons. They are S-100 and CD 34 positive. Perineurioma are non-encapsulated tumours with elongated spindle cells presenting with bipolar cytoplasmic processes, fusiform nuclei and eosinophilic cytoplasm disposed embedded in a mixed collagenous and myxoid stroma. They show EMA positivity.

Neurofibroma are seen between second and third decade of life and are very rare in infants. But in our case neurofibroma was seen in an infant of one year old, was present since birth.

Gold standard treatment is surgical excision with proper margins. Total surgical excision may be difficult if tumour is of infiltrative nature especially in the region of head and neck.5

Our is rare case of neurofibroma in an infant present since birth. Following excision, no recurrence was reported.

Conclusion:

Neurofibroma presents with features soft tissue swelling or skin pigmentations makes its diagnosis easy. However, difficulty arises if it is present in an infant. Proper diagnostic tools may help in exact diagnosis and treatment.

Fig 1 : Swelling in nape of neck right side.

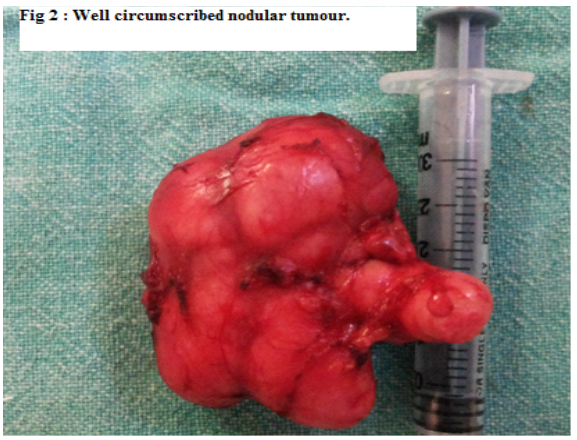

Fig 2 : Well circumscribed nodular tumour.

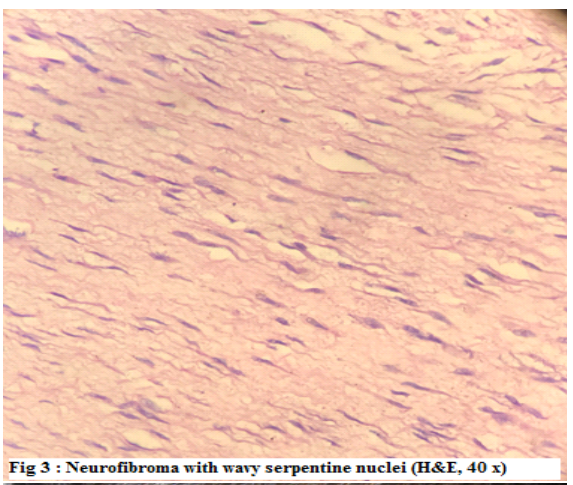

Fig 3 : Neurofibroma with wavy serpentine nuclei (H&E, 40 x)

Fig 4 : Post operative follow up

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