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Case Report- A Rare Case of a 8 year Old Child with Facial Plexiform Neurofibroma

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Abstract:
Plexiform neurofibroma is a very rare variant (5-30%) of type 1 neurofibromatosis. It is a non-circumscribed, benign irregular thick tumor arising from the peripheral nerve sheath. It is often compared to bag of worms due to its soft diffuse nature. It also involves connective tissue and skin folds. Here we present a case of 8years old boy who had facial dysmorphism noted from early childhood, progressively increasing in nature, had been evaluated and diagnosed clinically as plexiform neurofibromatosis.

Introduction

Neurofibromatosis(NF) type 1 is a rare autosomal dominant genetic condition that occurs in one in 3000-5000 births.[1] It is caused by mutations of the NF1 gene which is located at chromosome 17q 11.2, characterized by multiple skin lesions called neurofibromas.[2] It is a significant cause of morbidity and until recently no effective medical treatment was available.[3] only conservative line of management and long term monitoring of tumor growth are essential.[4,5,6] We report a rare case of NF in a 8year old boy who was having clinical features suggestive of plexiform variant, which is an uncommon form of type 1 NF.

Case Report:

A 8 year old child visited the outpatient department of pediatrics with chief complaints of swelling of left side of face noted since 2 years of age, swelling over occiput and multiple hypopigmented skin lesions noted since one year of age. The swelling over the face was gradual in onset, progressively increasing in size, spreading towards nose, ear and chin and slowly causing disfigurement of face. Child also had difficulty in swallowing noticed as the size of the swelling started increasing. There was no history of any discharge, pain, numbness, paresthesia in

association with swelling. No history of any similar swelling noticed in other parts of the body. No significant past history, medical history, family history was noted.

General physical examination showed moderately nourished 8year old male child with a steady gait and normal vital signs. On detailed head to toe examination it was found that over the right side of scalp, a linear scar with soft swelling was noted measuring around 5x1cm in size, also over left side of face there was a soft swelling with indistinct borders extending from lower eyelids downwards till the lower border of mandible measuring around 8x7cm in size and an irregular linear scar was noticed over the swelling. Over the tongue increased papillomatosis was found and misaligned teeth was noticed. On chest and back, multiple hypopigmented macules were noted. On the right leg, a depigmented linear macule was noticed, on the right axilla, freckling was noticed.

On palpation of the swelling, there was no local raise of temperature but mild tenderness was present. The swelling also had a varied consistency, soft in most of the areas with few firm nodular areas, pitting was absent. On auscultation bruit was absent. Examination of other systems were within normal limits.

Routine laboratory tests were normal, histopathological examination and genetic test had not been done as consent for the same had not been given by patient's relatives.

Ultrasound examination of face was done which was suggestive of diffuse adipose tissue infiltration in the inter and intra-muscular plane of the face. Cranio-facial CT scan showed bone, fat and soft tissue hemi-hypertrophy on left side of face. Ophthalmology opinion was taken to rule out optic glioma & lisch nodules. Two diagnostic criteria for NF 1 were met.[7] This patient was advised to come for follow up once in 6months.

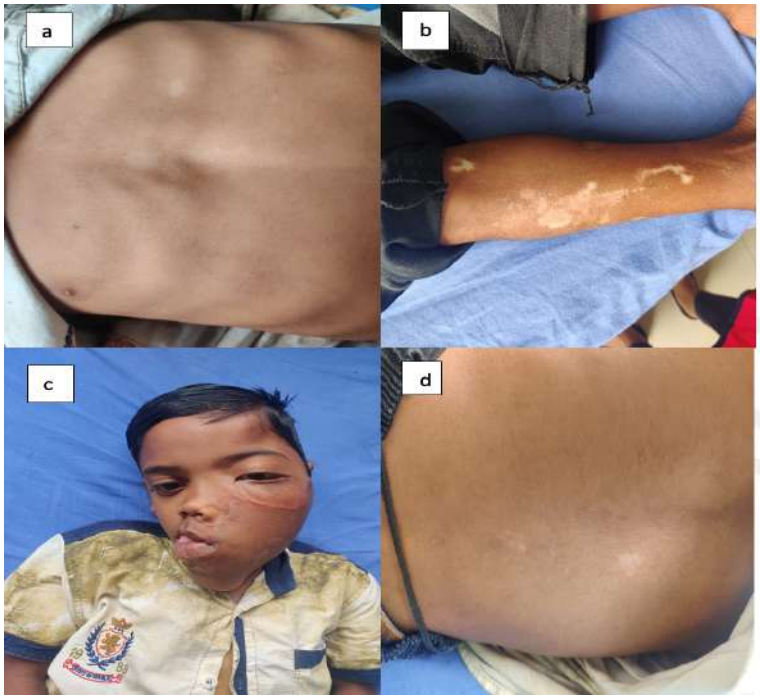

Figure 1 a and d. Showing on chest and back multiple hypopigmented macules.
Figure 1 b Showing a depigmented linear macule on the right leg.
Figure 1 c Showing over left side of face, a soft swelling with indistinct borders extending from lower eyelids downwards till the lower border of mandible measuring around 8x7cm in size and an irregular linear scar was also noticed over the swelling.

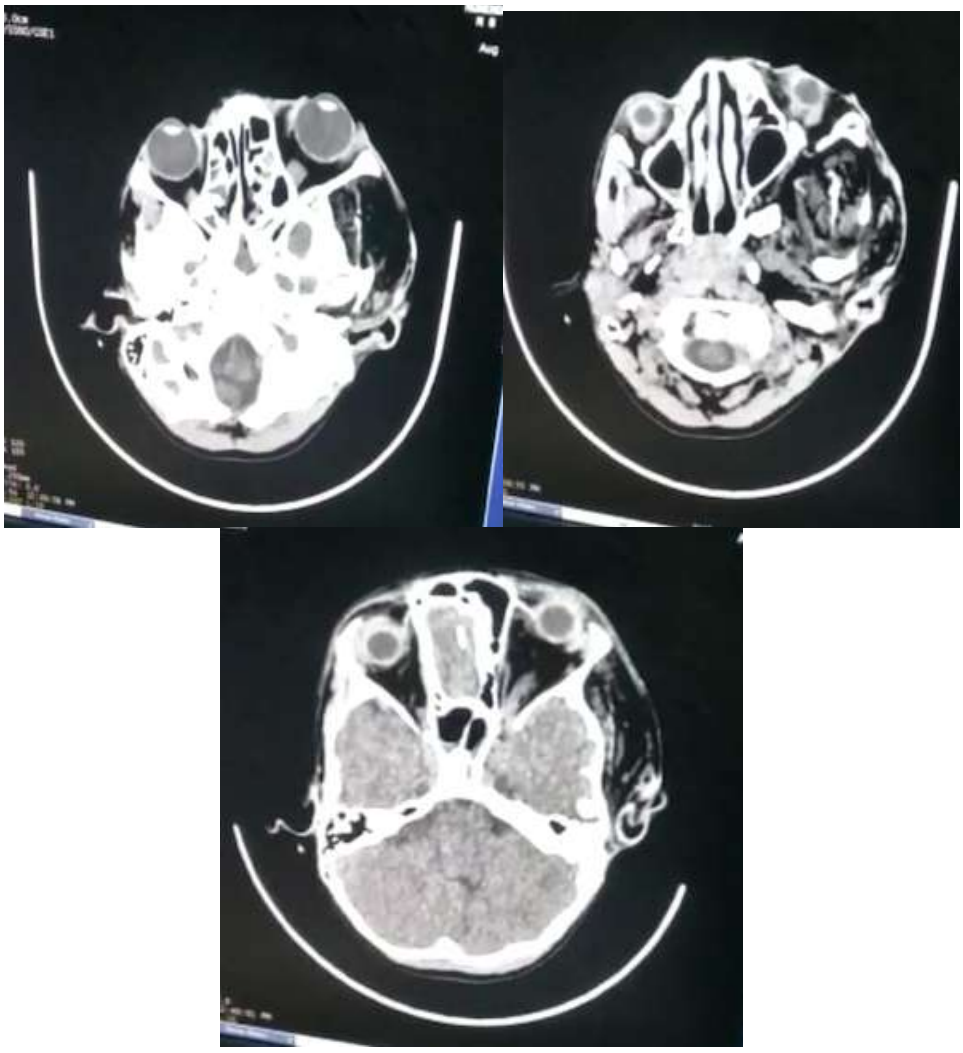

Figure 2: showing CT scan findings of face which is suggestive of fat and soft tissue hemi hypertrophy on the left side of the face.

Discussion:

Neurofibromas are benign, slow growing tumors with nerve sheaths as their site of origin and also consisting of fibromas, schwann cells and perineural cells. Within the spectrum of NF 1 pathology, plexiform NF is a very distinct presentation and can be seen either as a solitary lesion or in association with other tumors.[8] Two types of plexiform NF have been identified: Nodular type and Diffuse type.[9]

A diagnosis of NF can be made if at least two out of following criterias are met:

1. Six or more café au lait macules over 5mm in greatest diameter in prepubertal persons and over 15 mm in greatest diameter in post pubertal persons .
2. Two or more neurofibromas of any type or one 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 long bone cortex with or without pseudoarthrosis.

7. A first-degree relative (parent, sibling or offspring) with NF type 1, based on the above criteria.[10,11]

Prenatal testing may be used to identify the existence of NF 1 mutation in the fetus.[12] There is no specific therapy of NF 1. Prevention or management of complications are the only two feasible treatment modalities. In view of greater chance of injury and bleeding, resection of benign plexiform NF is done with great caution. There is high chance of recurrence of these lesions after removal.[13,14,15]

Conclusion:

Plexiform neurofibromas are usually benign. Sometimes might cause pain, disfigurement and might turn malignant. The most characteristic feature is its progressive nature and majority of cases it might worsen over a period of time. Mainstay in approach of plexiform neurofibroma case lies in its early diagnosis and prevention of its complications. Long term follow up is also mandatory.

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