Case Report

Plexiform neurofibroma with nevus of ota-rare presentation

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Abstract

Plexiform neurofibroma (PNF) is a rare form of neurofibromatosis type 1 which is rarely seen isolated. This generally spreads along the peripheral nerve and may affect some nervous rami. This is a poorly circumscribed and locally invasive tumor. About 21% of patients with NF-I are affected with PNFs. The nevus of Ota also called oculodermal melanocytosis is a macular discoloration of the face. It is most commonly found in the Japanese and very rare in the Indian subcontinent. It is unilateral oculodermal melanosis along the first two branches of the trigeminal nerve. We hereby present a very rare case of occurrence of isolated PNF (not associated with neurofibromatosis type 1) along with nevus of ota of the left side of the face in a 28-year-old female with thorough radiographic work up.

Keywords: Neurofibroma, nevus of ota, plexiform, trigeminal nerve

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INTRODUCTION

Neurofibroma is a benign peripheral nerve sheath tumor that arises from Schwann cells and perineural fibroblasts. Neurofibromatosis-I, also called Von Recklinghausen's disease, is a neurodermal dysplasia, it was first described by the pathologist Friederich Daniel Von Recklinghausen in the year 1882. It is the most frequent genetic human disease, affecting 1:3000 newborn and one of every 200 inhabitants with mental retardation. Plexiform neurofibroma (PNF) are elongated neurofibromas and spread along nerves involving multiple fascicles or large branches of the major nerve.[1] The cranial nerve most commonly involved are 5th, 9th and 10th nerve and it may appear anywhere on the face, orbit and globe. Majority of time it is associated with neurofibromatosis type 1.[2] About 21% of patients with NF-I are affected with PNFs.[3] The morbidity of PNFs in NF-I is high since they tend to grow until reaching a great size and producing disfigurement and their malignant transformation rate is 2-5%.[4]

Nevus of Ota, a dermal melanocytic nevus, is extremely uncommon in the Indian subcontinent. It was first described by Ota and Tanino in 1939, involves the skin along the distribution of the first and second division of the trigeminal nerve. It is a rare condition that affects only 0.014%–0.034% of the Asian population and is rare among males.[5] We hereby present a very rare case of an isolated PNF along with nevus of Ota of the left side of the face in a 28-year-old female.

CASE REPORT

A 28-year-old female patient reported to the department

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with a chief complaint of difficulty in eating food because of swelling in the left upper gums and cheek for the last 2 years. The patient first noticed it around 8 years back when it was of a size that was barely noticeable and didn't hamper her mastication and then slowly it increased to the present size. It was otherwise asymptomatic but for the last 2 years because of the increase in its size, it was getting impinged between the teeth. On asking about obvious facial asymmetry and discoloration with respect to the left side of the face patient gave a history of being operated which was present since childhood and was initially 1–2 cm and continued to increase in size with age. Therefore, surgery was done to remove the mark and a skin graft from the thigh was placed over the left cheek area. After few years of surgery, the patient noticed swelling over the left side of the face, the upper part of the nose and around the left eye which slowly increased over a period of time to the present size. On extraoral examination a well-defined periorbital growth of size 4 cm × 2.5 cm was noticed on the left side which was extended laterally in a reversed c shape curve, demarcated by a surgical line [Figures 1 and 2]. The overlying skin was bluish-black in color. On palpation, it was nontender and doughy in consistency with a firm nodular growth appreciated in relation to the infraorbital region having bag of worm consistency, no bruit or pulsation were present. Graft with hair growth along surgical lines was present on the left middle third of the face with respect to the cheek, zygomatic and postauricular region. The bluish-black pigmentation with irregular margin was present with respect to left side bridge of nose and left corner of the mouth. Drooping of the left corner of the mouth present because of pressure from the swelling. The facial nerve examination was normal. Intraoral examination revealed mucosal growth with respect to attached gingiva of left upper canine (23) to left second molar (27) which was approximately 4.5 cm × 1.5 cm in size almost covering occlusal aspect of teeth, having pink in color and smooth surface with normal-appearing surrounding mucosa [Figure 3]. The growth was non-tender with mixed soft to firm consistency giving bag of worm feel. Similarly, two mucosal growth seen from left buccal mucosa, one 1cm posterior to left retrocommisure area and 2nd on anterosuperior aspect corresponding to 23 measuring 0.7 cm × 0.4 cm and 0.3 cm × 0.2 cm respectively. The left upper first molar (26) was missing.

On the basis of the above clinical findings differential diagnosis for pigmentation includes mongolion spot, melasma, blue nevus, drug-induced hyperpigmentation and for growth PNF, congenital melanocytic nevus (as hyperpigmentation and hypertrichosis common in both), vascular malformation was given.

An intraoral periapical radiograph (IOPA) with respect to 25–28 region and Orthopantograph (OPG) was advised for the patient. IOPA showed distal inclination of 24, 25 and mesial inclination of 27 and missing 26 along with

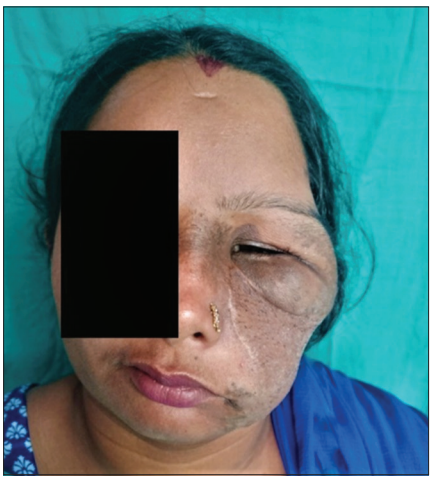

Figure 1: Clinical Image shows extraoral view-front view

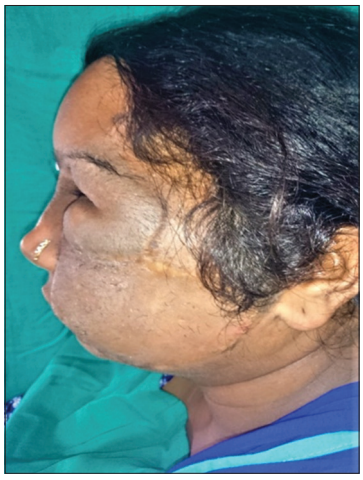

Figure 2: Clinical Image shows Left lateral profile of the patient

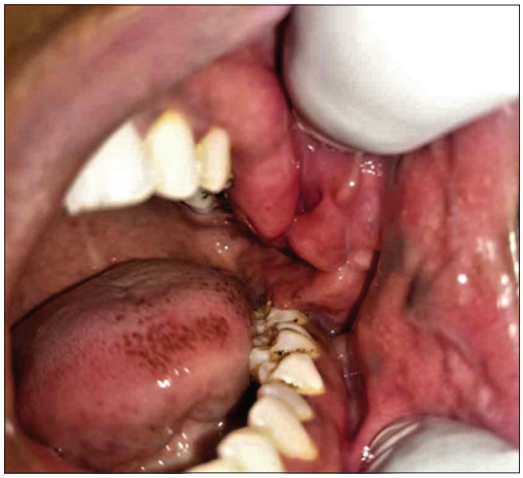

Figure 3: Clinical image shows Intraoral findings

the erosion of crest of alveolar bone with respect to 25, 27 [Figure 4].

OPG was advised for the patient which showed thinning of the left zygomatic arch, along with deepening of the sigmoid notch with elongated coronoid process and obliteration of left maxillary sinus with multiple septation was observed [Figure 5].

Ultrasonography revealed soft tissue thickening with respect to left temporal, infratemporal, orbital and buccal mucosa with vascularity seen on color Doppler with areas of subtle erosion of bone at places. Further for full bony extent contrast-enhanced computed tomography (CT) Scan was advised. CT finding suggested a heterogeneously enhancing soft tissue thickening involving the left side of the face infiltrating temporal, infratemporal fossa and masticatory space [Figure 6]. There was subtle erosion of underlying bone and thickening with expansion (remodeling) at the lateral wall of orbit, zygomatic arch and anterior surface of maxilla of the left side. There was the widening of the inferior orbital fissure. The soft-tissue thickening seems to be infiltrating into the left buccal mucosa and lateral aspect of the left orbital, however the globe appears normal. There was remodeling of the left maxillary sinus causing it to decrease in size in the transverse direction. Minimal mucosal thickening seen with respect to bilateral maxillary sinus. The radiographic impression was for PNF with differential diagnosis of low flow vascular malformation was given. Incisional biopsy was done. The surgical sample was subjected to histopathological evaluation. H & E section showed interlacing bundles of spindle cells exhibiting wavy nuclei [Figure 7]. These cells were in association with delicate collagen bundles and variable amount of myxoid matrix and foci of mast cells which confirmed the diagnosis of plexiform neurofibroma.

Further dermatological and ophthalmic consultation was done for the patient to rule out Neurofibromatosis 1. No café au lait spots or any other neuromas were detected and ophthalmic examination was normal.

Thus we concluded it to be the rare case of isolated PNF with nevus of ota. The patient underwent debulking procedure under general anesthesia as because of the massive extent total excision was not possible. The patient was informed about symptoms and chances of its malignant transformation and was advised regular follow-up. Six-month postsurgery, patient did report the recurrence.

Additional informed consent was obtained from the patient for which identifying information is included in this article.

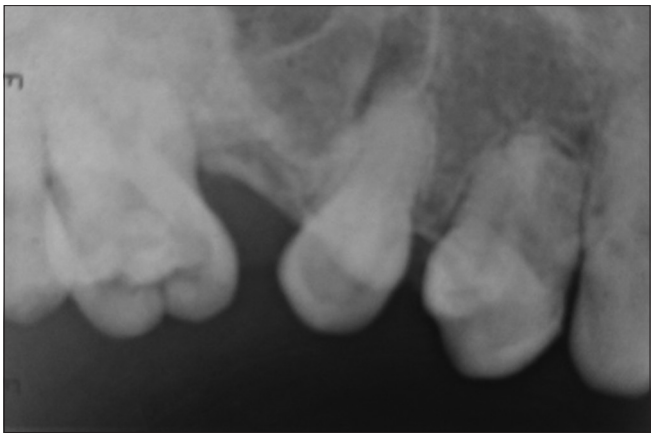

Figure 4: Image showing intraoral periapical radiograph with respect to 26, 27 region

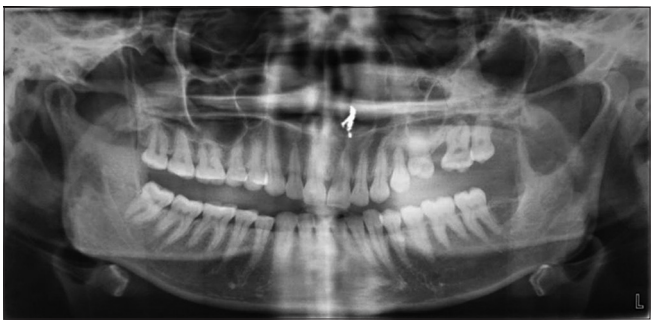

Figure 5: Orthopantomograph showing thinning of the left zygomatic arch, deepening of the sigmoid notch, elongated coronoid process

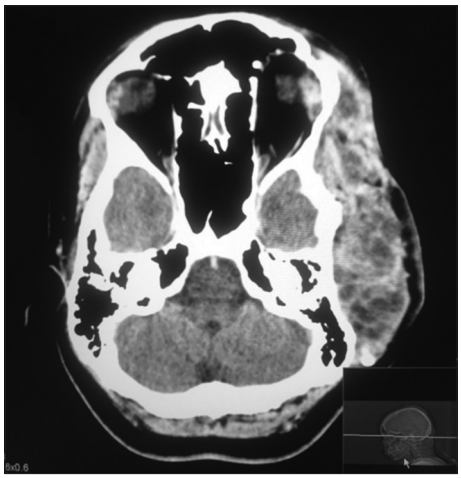

Figure 6: Axial section computed tomography scan showing a heterogeneous soft-tissue thickening involving the left side of face infiltrating temporal, infratemporal fossa and masticatory space

DISCUSSION

The word plexus means the combination of interlaced parts or networks. PNF can be quiet disfiguring and may present as hemifacial hypertrophy. Impingement on surrounding structure may cause functional compromise and soft tissue and bone hypertrophy. They may be present

since in relation to but most of them usually appear during the first 2 years of life and if not present by then rarely develop after adolescence.[6] Gingival involvement as seen in our case present as a unilateral nontender growth with the consistency of "bag of worms". Normally the swellings on the gingiva present as diffuse unilateral gingival enlargement of attached gingivae. In some cases, interproximal gingiva may also display enlargement.[7] These swellings are fibrous and do not exhibit signs of inflammation.[8] While 6.5% of patients with NF 1 have reported with gingival neurofibroma. Solitary PNF has been reported in patients with no relation to NF1.[9,10] The natural history of PNF is associated with the period of rapid growth followed by period of quiescence. The rapid growth associated with persistent associated unexplained pain, change in consistency along with neurological deficit can be alarming sign for complication of malignant peripheral nerve sheath tumors (MPNST).[11]

The condition can be quite disfiguring, as observed in the case being presented, and hemifacial hypertrophy can occur. Complications include bleeding from trauma due to excessive neovascularization, neurological deficits and psychological disturbance because of abnormal anatomy.[12] Radiographic manifestation [Figure 8] can be lengthening, narrowing of coronoid process with deepening of the sigmoid notch as seen in our case. Intraosseous neurofibroma may cause enlargement of mandibular canal, mental foramen or mandibular foramen.[13]

Surgical excision is the line of treatment. It is associated with problems like the risk of neurological and functional deficit and challenges because of infiltrating and vascular nature. Multiple surgeries are often required and chances of recurrence are high i.e., 20%with complete resection and 45% with partial resection.[14] Until recently, neurofibromas management consisted of only surgical resection, associated with a high hemorrhagic risk, depending on functional and/or esthetic requirements of the patients, or in case of malignant transformation. The emergence of the oral selective mitogen-activated protein kinase inhibitor selumetinib has now become the current standard of care for problematic PNF, allowing their shrinkage in 68% of children.[15]

Complications included are NF1 patients present a higher lifetime cancer risk (59.6%) in contrast to the general population (30.8%). MPNST is the main malignancy which develops in 5% to 13% of NF1 patients with poor prognosis. Patients with NF1 are also at a risk of developing intraoral squamous cell carcinoma.[16]

Since our patient was having a frank bluish-black unilateral discoloration which was not classical of café au lait spot we further searched the literature for this irregular, macular pigmentation along the first and second division of the trigeminal nerve. We concluded it to be nevus of ota. Its a rare condition with a prevalence of 0.014% to 0.034% in Asians with higher incidence in females (80%) and unilateral presentation (90%–95%).[17] Its association with phakomatosis, pigmentovasculari nevus fammeus, Sturge weber syndrome and neurofibromatosis has been described in the literature.[18] It is associated with malignant melanoma

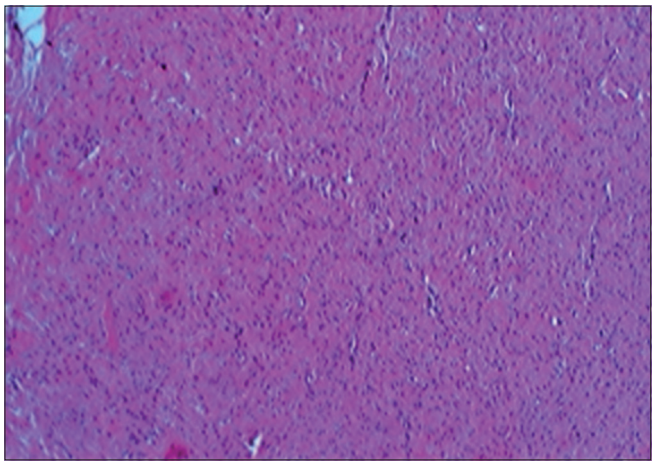

Figure 7: Histopathological Image shows interlacing bundles of spindle cells exhibiting wavy nuclei. Surrounded with delicate collgen bundles and variable amount of myxoid matrix and foci of mast cells (H & E, ×100)

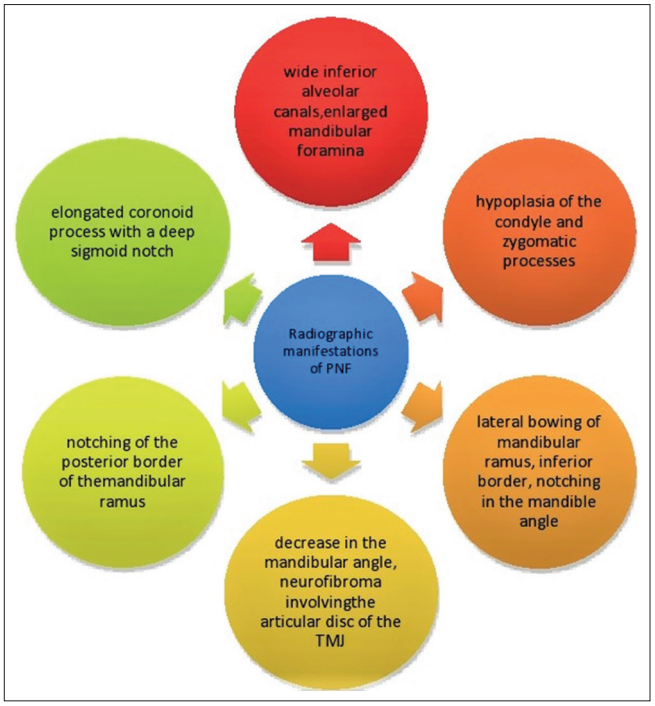

Figure 8: Image showing radiographic spectrum of Neurofibroma

so periodic reevaluation is recommended especially in cases with sudden increase in size of discoloration ulceration or paresthesia.[17]

CONCLUSION

Our case is a rare occurrence of isolated PNF not associated with neurofibromatosis type 1 along with nevus of ota. Striking radiological findings on orthopantomograph gave great insight into the diagnosis of the case. It is important that oral and maxillofacial physicians and general dentists must keep this disease under check when oral lesions characteristic of NF-I are present. These patients must be reviewed long term because of eventual complications, especially that of malignant transformation.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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