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Case Study

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A RARE CASE REPORT OF NEUROFIBROMATOSIS TYPE 1 IN HARD PALATE REGION

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ABSTRACT

Neurofibroma is a benign peripheral nerve sheath tumor comprising variable mixture of Schwann cells, perineurial-like cells, and fibroblasts. Neurofibroma may occur as solitary lesion or as part of a generalised syndrome of neurofibromatosis or very rarely as multiple neurofibromas without any associated syndrome. Here, we present a case of a 42-year-old male patient with NF-1. Patient have generalized freckling and café au lait spots throughout the body since 10-12 years and a diffused swelling measuring about 3 cm × 2 cm, extending from the right maxillary hard palate region to the midpalate since 1year. The diagnosis of NF-1 was made according to the presence of two or more diagnostic criteria of the National Institute of Health Consensus Development Conference. No recurrence was observed in a 6-month follow-up after extensive surgical ablation.

INTRODUCTION

According to the 2017 World Health Organization Classification of Head and Neck Tumors, neurofibromas are benign peripheral nerve sheath tumors that consist of a mixture of Schwann cells, perineurial cells, fibroblasts, and axons.[1] It mostly seen as skin lesion but also neurofibromas present in oral cavity not uncommon.[2] Neurofibromatosis (NF) mostly involves 5th cranial nerve and upper cervical nerves, with common sites being tongue, gingiva, major salivary glands and jaw bones.[3] Neurofibromas resemble schwannomas and are often seen as elastic, hard, well defined masses with no capsule. The World Health Organization has classified neurofibromas into 2 types: dermal and plexiform. Dermal neurofibromas arise from a single peripheral nerve, while plexiform neurofibromas are associated with multiple nerve bundles.[4]

Neurofibromatosis type 1 was first described by Frederich von Recklinghausen in 1882.[5] Neurofibromatosis type 1 (NF-1) or von Recklinghausen disease is an autosomal dominant disorder with a basic defect in the embryonic neural crest cells which give rise to ectodermal and mesodermal derivatives that affect one in 3000 live births.[2] Solitary neurofibromas rarely occur in the oral cavity.[6] The frequency of solitary neurofibromas in the oral cavity is reportedly 6.5%, particularly for lesions that are not associated with NF-1, and their occurrence on the hard palate is extremely rare.[7] Treatment for both NF-1 and NF-2 is directed toward controlling symptoms and managing the complications.

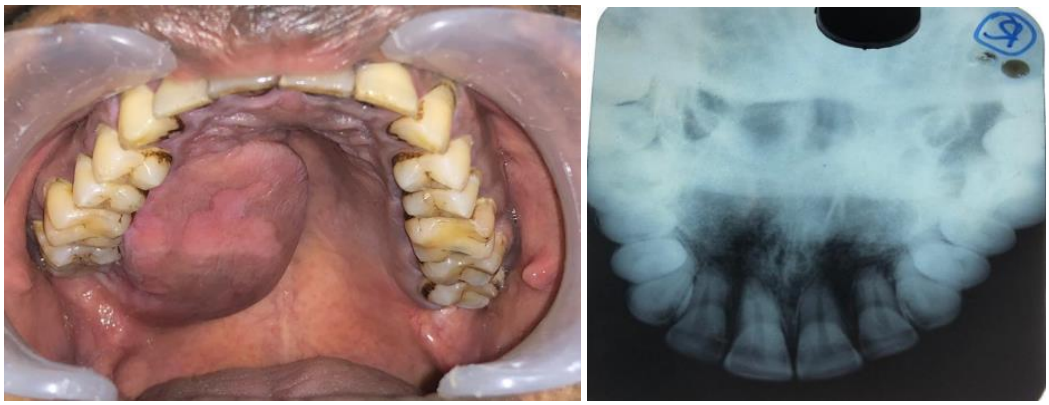
We report an unusual case of a palatal neurofibroma with well-demarcated radiolucency diagnosed using intra oral periapical radiograph (IOPAR) and histopathological examination with other findings like several café au lait pigmentation and freckling spots on the patient's upper body region and palms.

CASE REPORT

A 42 year old male patient reported to our department of oral and maxillofacial surgery with a painless swelling on the right side of the hard palate since 15-20 days. It had become noticeable for the past 2 years which gradually increased over a period of time and attained its present size. There was a positive family history and no relevant medical history. On physical examination, several café au lait pigmentation and freckling spots on the patient's upper body region and palms were evident.

A comprehensive clinical examination was performed in our department, which revealed diffused swelling measuring about 3 cm × 2cm approximately, extending from anteriorly 1 cm distance from palatal mucosa w.r.to 12; posteriorly up to maxillary tubersity region; medially crossing the mid palatine raphe and laterally up to palatal mucosa w.r.to right upper premolars ad molars teeth. The palatal tissue of the upper right quadrent was described

as fluctuant when palpated, no tenderness was present(Figure 1a).



a) **b)**

Figure 1: (a) Intraoral view showing the swelling involving the right side of the hard palate and extending to mid palate. (b) Intraoral periapical radiograph of a space occupying lesion.

In intra oral periapical radiograph showed a solitary, well-defined, homogenous space occupying lesion in the right maxillary region (Figure 1b).

An incisional biopsy under local anesthesia from right anterior maxillary region of that swelling was done. Biopsy revealed densely packed collagen bundles interspersed with wavy bundles of nerve tissue having spindle nuclei and fibroblasts were plump and multiple with spindle shaped fibrocytes were evident. No dysplastic features were reported in the specimen given (Figure 2).

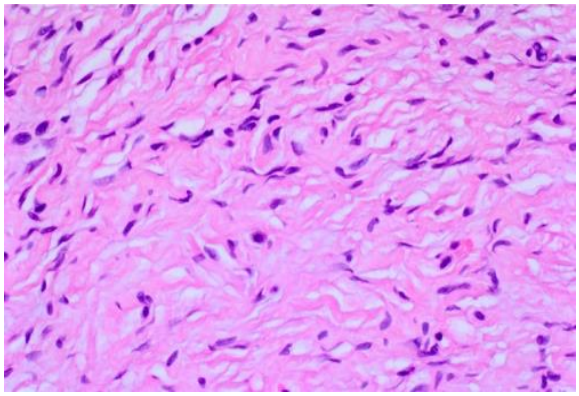
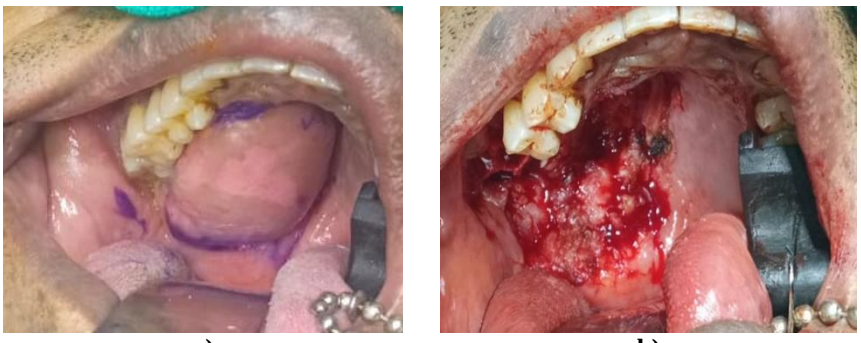


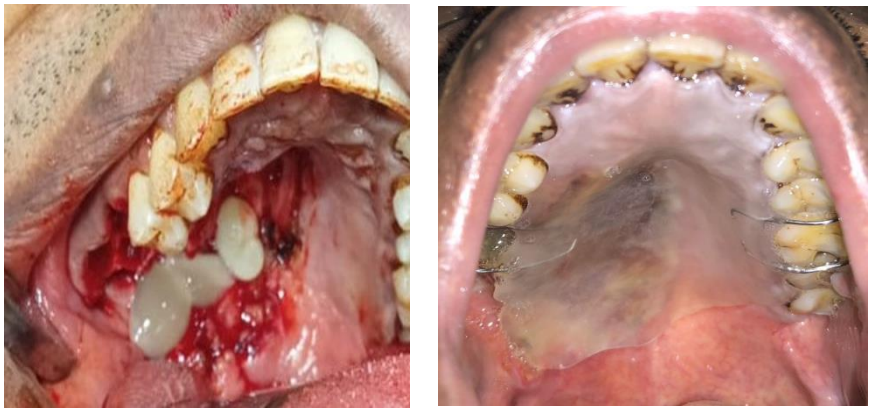
Figure 2: Histopathological examination showed spindle nuclei and fibroblasts.

An immunohistochemical test for the S-100 protein was positive (detection system used was the horseradish peroxidase polymer) confirming the neural origin of the tumor. The epithelial membrane antigen, however, was negative, hence ruling out von Recklinghausen's disease. The histological findings were consistent with diagnosis of neurofibroma.

A decision was taken to carry out a wide local excision under general anaesthesia followed by the primary closure.



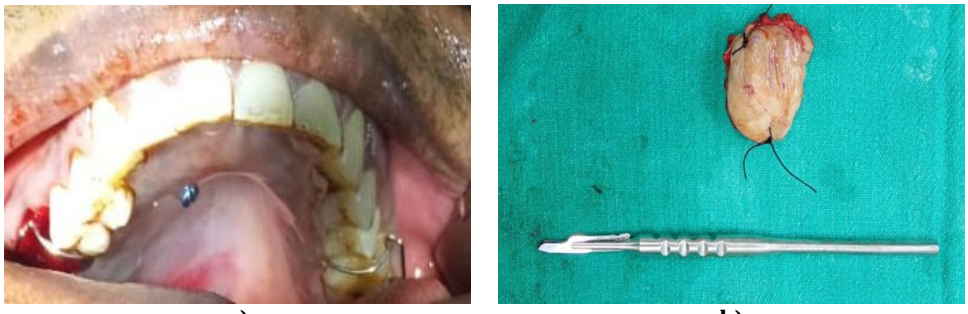
a) **b)**



c) d)

Figure 3: (a) incision marked by ink before surgery (b)wide local excision of soft tissue mass that results soft tissue defect (c) bone bleeding controlled by bone wax (d) soft tissue defect covered by palatal obturator.

Under general anesthesia, the lesion was excised, mucoperiosteal flaps were reflected from teeth #11-18 and greater palatine artery was identified, ligated and cauterized. The lesion was completely removed by the aggressive thinning of the palatal flap and upper right 1st, 2nd and 3rd molar was extracted. Bony bleeding controlled by applying bone wax (figure 3c). Soft tissue defect covered by platelet-rich fibrin (PRF) and Palatal soft tissue defect covered by palatal obturator (figure 3c & 3d) & it was fixed by 2x6mm titanium screw (figure 4a).



a) b)

Figure 4: (a)obturator fixed by titanium screw (b) excised tissue sent for biopsy.

Figure 5: follow up after 6 months.

The patient healed uneventfully following both the surgical procedures. At the 3rd and 6th month follow-up, there was no recurrence of such type of swelling (figure 6).

DISCUSSION

Oral and maxillofacial lesions of the neural origin are rare.[8] Neurofibroma is a benign peripheral nerve sheath tumor which arises from Schwann cells and peripheral fibroblasts. Fawcet and Dahlin reported seven such tumors in their review of 3987 primary jaw bone tumors, the most common site is the mandible.[9]

The National Institute of Health Consensus Development Conference in 1988 proposed diagnostic criteria for neurofibromatosis type 1, if a patient has two or more of the following findings:

1. Six or more café au lait macules
2. Two or more neurofibromas of any type or one plexiform neurofibroma
3. Freckling in the axillary or inguinal regions

4. Optic glioma
5. Lisch nodules
6. Distinctive osseous lesion such as sphenoid dysplasia
7. Family history of the first-degree relative with neurofibromatosis.[10]

In the present case, the patient reported with a positive family history along with palatal neurofibroma, café au lait spots and with generalized freckling.

Neurofibromas are benign lesions of the peripheral nervous system derived from Schwann and mesenchymal cells of the nerve sheath. Neurofibromas can appear as single tumors (solitary neurofibromas) or as NF-1 that presents as multiple tumors along with café-au-lait spots in von Recklinghausen disease. Neurofibromas are common in the tongue and buccal mucosa in the oral cavity, but approximately 20-60% of oral neurofibromas are associated with neurofibromatosis.[11] In 2019, Broly et al[12] reviewed 26 cases of isolated neurofibromas occurring in the oral cavity, and intraoral occurrences were very rare. However, because the oral cavity is composed of various tissues such as the tongue, buccal mucosa, gingiva, floor, and palate, the neurofibromas that develop there have different nerves of origin and different surrounding tissues that are removed during surgery.

There were no recurrences in any of the previously reported cases[13-19] or in our case. Recurrence can be avoided by providing a safety margin, including the periosteum, around the tumor at the point of resection. The current treatment of neurofibroma is complete resection. These tumors are nonradiosensitive and have limited benefit with chemotherapy.[20] Although the recurrence rate is low, there have been reports of a malignant transformation of these tumors, with the possibility that the presumed lesion may have been the first manifestation of von Recklinghausen's disease.[21]

The present case revealed a well-defined, unilateral neurofibroma without displaced anterior teeth and other signs of neurofibroma like café au lait spots and with generalized freckling on extremities with a positive family history. Surgical excision was performed, and it did not reoccur after 6 months of follow-up.

CONCLUSION

Considering other advanced investigations such as CBCT, immunohistochemistry (S-100 protein test) is often beneficial for an accurate diagnosis. Early detection and intervention with advanced technique and surgically complete removal of the tumor is the main stay of the treatment. Adequate margins should be established at the time of resection because the tumor often has no membrane. 1 year has passed since the surgery, and the patient has not developed any recurrences to date.

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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Nil.

Conflicts of interest

There are no conflicts of interest.

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