



Maxillofacial Surgery

SOLITARY PIGMENTED NEUROFIBROMA OF BUCCAL MUCOSA: A RARE CLINICAL ENTITY

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ABSTRACT Neurofibroma is a benign tumour of peripheral nerves composed of schwann cells and endoneural fibroblasts. It may occur as solitary lesion or a syndromic condition as Von-Recklinghausen's disease or very rarely as multiple neurofibromas without any association with neurofibromatosis syndrome. Solitary neurofibromatous lesions that occur in the buccal mucosa mimic the appearance of traumatic fibroma unless other extra-oral features indicate the diagnosis towards the former. We are reporting the case of solitary melanotic neurofibroma without any tell-tale signs of neurofibroma extra-orally but proved to be so with the assistance of histopathological and immunohistochemical analysis.

KEYWORDS : Neurofibroma, Melanocytes, Immunohistochemistry, Neurofibromin

INTRODUCTION:

Neurofibroma (NF) is a benign tumour of peripheral nerves that is composed of Schwann cells and endoneural fibroblasts.¹ Neurofibroma of the oral cavity are presented as a submucosal, nontender, discrete mass. It may occur as solitary lesion or as part of a generalised syndrome of neurofibromatosis (von Recklinghausen's disease) or very rarely as multiple neurofibromas without any association with neurofibromatosis syndrome.² This condition has been found to be linked to the mutation of the NF1 tumor suppressor gene located on chromosome 17, locus q11.2. The latter gene encodes a cytoplasmic protein called neurofibromin, which negatively regulates the proto-oncogene Ras.³ Pigmented neurofibromatosis of oral cavity is often a very rare finding and very few are documented in literature. Here we are discussing a case of solitary pigmented neurofibroma of the buccal mucosa with diagnostic outline, histopathologic and immunohistochemical findings and management.

CASE REPORT

A 46-year-old female was referred to the Department of Oral and Maxillofacial Surgery, K.L.E dental college, Bangalore, with a chief complaint of painless growth in the lower left cheek mucosa region for 6 months. There was no positive family history and no relevant medical history. It started initially as a small growth few months ago which steadily increased to the current size. Chewing and normal oral function have been hampered by the growth. Extra-orally, no neurofibromatous changes were noted, however melanotic pigmentation was noted over right and left arms and cheeks bilaterally [Fig 1 and 2] A comprehensive clinical examination was performed in our department, which revealed a well circumscribed, smooth, nontender, firm and lobulated pink swelling measuring about 2cm * 2 cm in its greatest diameter in relation to the tooth number 38 on the lower side of buccal mucosa. [Fig 3]. A provisional diagnosis of Irritational fibroma was given and a differential diagnosis of Pyogenic granuloma and Neurofibromatosis were suspected. The treatment plan of surgical excision of the lesion, histopathological examination and Immunohistochemistry of the lesion was made and executed with the consent of the patient.

MANAGEMENT

Routine blood investigations were performed and were in normal limits. The surgical site was prepared with antiseptic solution (betadine), Local anaesthesia with adrenaline (2% Lignocaine HCl with adrenaline 1:80,000) was infiltrated around the growth. The growth was excised completely [Fig 4] from its base using a 15-no. blade, haemostasis was achieved and the surgical site was closed with two interrupted silk sutures. Post-operative medications such as Tab Oflox OZ (Ornidazole 500mg and Ofloxacin 200mg), Tab Imol plus

(Ibuprofen 400mg and Paracetamol 325mg) were prescribed for a period of 5 days from the day of surgery. Post-operative instructions were given to the patient that include maintenance of surgical site, soft diet and management of oral hygiene. A follow-up after 3 days, 1 week, 1 month [Fig 5] and at the end of 6 month [Fig 6] was done. The surgical site healed uneventfully with no recurrence.

Histopathology and Immunohistochemistry

Fig 1: Melanotic Pigmentation On Cheeks



Fig 2: Melanotic Pigmentation On Right And Left Arm



Fig 3: Firm Lobulated Swelling On The Left Buccal Mucosa



Fig 4: Excised mass of swelling for histopathological examination and Immunohistochemistry



Fig 5: Post operative view of the operated site after 1 month



Fig 6: Post-operative View Of The Operated Site After 6 Months

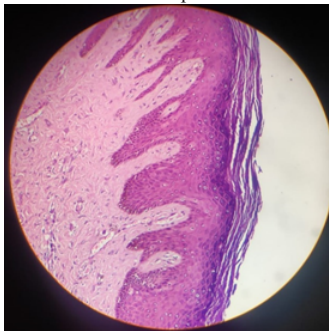


Fig 7: Histopathological View Of Spindle-shaped Cells With Wavy Nuclei Are Seen

The excised tissue was sent for histopathological diagnosis which revealed the presence of appearance of para to ortho keratinised stratified squamous epithelium exhibiting focal areas of hyperplasia with connective core entrapment. Prominent granular cell layer is evident at few areas. The superficial layers of the epithelium at foci show spongiosis. The basal cell layers at many areas are laden with melanin pigmentation. The underlying connective tissue stroma is predominantly mature and fibrous with collagen arranged in dense fibres and bundles in reticular, linear, whorled and interlacing patterns associated with plump fibroblasts and fibrocytes. The fibroblasts subepithelially present are plump and giant cells with few cells showing multiple nuclei in them. Spindle-shaped cells with wavy

nuclei are seen. [Fig-7] Melanin incontinence and melanin pigment laden cells are seen in the connective tissue stroma subepithelially. At focal areas the collagen shows “Shredded Carrot Appearance”. The stroma shows even distribution of mast cells throughout the lesion with few of them degranulating. Neurites are present among the fibrous connective tissue stroma at focal areas which was suggestive of “Neurofibroma- Collagenous, Pigmented Variant” Immunohistochemical analysis of the lesion revealed positive reaction to S-100 proteins [Fig 8]

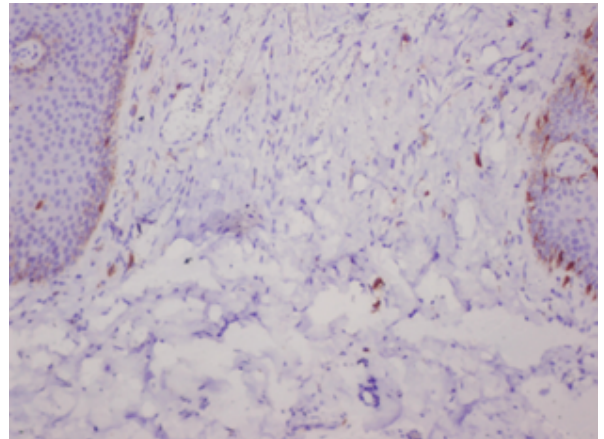


Fig 8: Immunohistochemical Analysis Of The Lesion Revealing Positive Reactions To S-100 Proteins

DISCUSSION:

The mutation of NF1 tumor suppressor gene located on chromosome 17, locus q11.2, results in haploinsufficiency which in-turn results in lack of neurofibromin that results in failure of downregulation of RAS.⁴ Uninterrupted transmission of mitogenic signals to the nucleus would result in uncontrolled proliferation of fibroblasts. It is very rare to encounter a solitary neurofibromatous lesion that is also melanotic. As per the previous literatures, the hyperpigmentation of the skin in NF-1 is attributed to the conjectures of schwann cells and Melanotic cells and atavism of the schwann cells and melanotic cells that share similar origin. The other reason could also be the entrapment of atypical melanotic cells that get trapped in these tumours.⁵ The diagnostic feature of neurofibromatosis type I includes multiple café-au-lait macules, skin fold frecklings, iris lisch nodules and tumours of the nervous system. However, in the case presented to our centre did not show any pathognomonic signs of NF-1 clinically which led to the provisional diagnosis of traumatic fibroma attributing to the site of occurrence. Histopathologic picture was in consistency with the those reported cases of neurofibromatous lesions of oral cavity.^{6,7,8} and Immunohistochemistry revealed positive reaction to S-100 protein which is confirmatory investigation for neurofibromatosis. Melanin deposition was noted in the histopathological sections of the lesion which was in accordance with previous literatures.^{9,10}

CONCLUSION

Based on various literature analysis, reviews and reports of various cases of NF-1, It is prudent to histopathologically assess all masses that develop in the oral cavity irrespective of solitary lesion or clustered and Immunohistochemical assessment as per indications. With the above discussed case we can arrive at a conclusion that not all pathognomonic signs and clinical features of NF-1 is noted all the time.

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