Plexiform Neurofibroma: An Unusual Diagnosis of Lip Swellings

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Neurofibromas (NF) are seen either as a solitary lesion or as part of the generalized syndrome of NF (NF-1, also known as Von Recklinghausen disease of the skin). In plexiform neurofibroma (PN), there is proliferation of Schwann cells from the inner aspect of the nerve sheath, thereby resulting in an irregularly thickened, distorted, tortuous structure. Oral involvement by a solitary and peripheral PN in patients with no other signs of NF is rarely seen. It is reported that only 4-7% of patients affected by NF display oral manifestations. A solitary PN in a patient with no other symptoms is a diagnostic challenge, more so when the location of the lesion is one of the rarest sites.

Keywords: Lip, Neurofibroma, Plexiform, Von recklinghausen disease

INTRODUCTION

Neurofibromatosis (NF) is an autosomal dominant (AD) disorder that affects the bone, the nervous system, soft tissue and the skin,1 they are seen either as a solitary lesion or as part of the generalized syndrome of NF (usually NF-1, also called von Recklinghausen disease of the skin).2 The solitary form differs from disseminated form in not being associated with systemic and hereditary factors. The World Health Organization has subdivided NF into two broad categories: dermal and plexiform. Dermal arise from a single peripheral nerve, while the plexiform are associated with multiple nerve bundles. Other clinicopathological subtypes include localized NF (sporadic NF), diffuse NF, plexiform neurofibroma (PN), and epithelioid NF.2

PN is a type of NF, representing an anomaly rather than a true neoplasm, in which the proliferation of Schwann cells occurs from the inner aspect of the nerve sheath, thereby resulting in an irregularly thickened, distorted, tortuous structure. They are characterized by diffuse enlargements of multiple fascicles of the nerves and its branches. It is a slow growing and locally infiltrative benign tumor extending along the nerves into the surrounding tissue, however, have a greater chance of malignancy when located deeply.

Despite their occurrence in the head and neck region, neural sheath tumors are rarely encountered in the oral cavity. Oral involvement by a solitary and peripheral PN in patients with no other signs of NF is rarely seen.3 It is reported that 4-7% of patients affected by NF display oral manifestations.3 The oral lesions are known to occur as discrete, non-ulcerated nodules, which tend to be of the same color as the normal mucosa, usually occurring on the buccal, palate, alveolar mucosa, vestibule, and the tongue.2 Hereby, we report an interesting case of PN of the lip swelling, an unusual and an extremely rare site.

CASE REPORT

A 6-year-old boy presented with a single diffuse swelling on the left upper lip extending to the buccal mucosa, which was progressively increasing in size. There was no history of any pain or bleeding. The swelling measured about 5 cm × 4 cm in size, irregular in shape and ill-defined margins. It was of the mucosal color and there were no secondary changes. On palpation, the swelling was firm, non-tender, irreducible, non-pulsatile, and there was no fluctuation or discharge from it. The external surface was lobulated and there were folding present on the swelling. No café-au-lait spots were present over the body.

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A clinical diagnosis of hemangiofibromatosis was made. The patient was subjected to reduction plasty and multiple grayish white soft tissue bits were sent for histopathology. On gross examination, the largest tissue piece was 3.0 cm × 0.8 cm × 0.7 cm with skin attached. Cut section was homogenous and grayish white in color while rest of the tissue pieces were aggregating to 1.5 cm × 1.5 cm × 0.3 cm in dimensions. Microscopic examination showed an overlying stratified squamous epithelium of normal thickness, while the underlying subepithelial tissue showed myxoid NF lobules with central nerve bundles (Figures 1 and 2). The adnexal tissue was unremarkable. On the basis of histopathology, a diagnosis of PN was made.

**DISCUSSION**

NF also known as von Recklinghausen disease is a genetically inherited disease, which was first described in 1882.² It includes two distinct variants that differ from each other genetically, histologically, and clinically. NF-1, is one of the most common AD inherited disorders, the prevalence being 1 in 2500-3300, characteristic feature being peripheral nerve sheath tumors and NF, leading to symptoms, and disfigurement.¹ However, NF Type-2 (central NF) is an AD disease, accounting for an extremely small percentage of the total cases of NF. The hallmark being the presence of bilateral vestibular schwannoma.¹

NF are benign tumors of neural origin or nerve sheath tumors which present in three forms: Local discrete, generalized NF, and PN.⁴ They are composed of neurites, Schwann’s cells, and fibroblasts within a collagenous or myxoid matrix. It is generally associated with a generalized syndrome of NF, but a few cases of the solitary intraoral lesions have also been reported.⁵

PN is an extremely rare tumor and more so of a lip swelling. It is much less common than the conventional NF and taking two distinct forms; a relatively small type lesion seen in the dermis or superficial subcutis; and a larger, usually deeply situated type, which often involves voluntary muscles or visceral structures. The anatomical distribution and depth of the lesion is quite variable.⁶ They form tortuous cords along the segments and branches of a nerve with a tendency to grow centripetally. Diffuse enlargements of multiple fascicles of the nerves and its branches, leads to thickening of nerves. They are poorly circumscribed, slow growing, and locally infiltrating benign tumors.⁴ This tumor is said to be indicative of virtual retinal display even though it may be the only manifestation of the disease.² It is used to describe a network-like growth of tumor involving multiple fascicles of a nerve, leading to a diffuse mass of thickened nerve fibers surrounded by proteinaceous matrix.¹

However, it spreads along the peripheral nerve and may affect some nervous rami. About 21% of patients with NF-1 are affected with PNs. Morbidity of PNs in NF1 is high since they tend to grow until reaching a considerable size and produce disfigurement. Besides, the risk of malignancy is between 2% and 5%. Due to its diffuse involvement/appearance and soft consistency, palpation of NF is similar to that of lipoma, vascular malformation, lymphangioma or rhabdomyoma. Similar diagnostic dilemma was seen in our case which was provisionally diagnosed as hemangiofibromatosis due to its diffuse involvement and consistency. However, in our case also it was poorly circumscribed and locally invasive extending from the lip up to the buccal mucosa, the findings consistent with the literature studied.¹

Head and neck region is the most common site of involvement but only 4-7% of patients affected display oral manifestations.⁸ Close perusal of literature on this lesion
revealed only one another report of its occurrence in the gingiva, making this report even more rare which was found as lip swelling with extensions in the buccal mucosa and gingival. Mobile tongue is the most common site involved, followed by buccal mucosa, floor of the mouth, palate, lips, and gingival.

Till date, very few cases of NF of the lip have been found to be published. Involvement of superficial soft tissue is more frequent than deeper lesions. It is an important lesion for two reasons: First, at whatever site, it is pathognomonic of NF; second, it runs a small but significant risk of undergoing malignant change, particularly if deeply located.

Skin lesions appear as soft, drooping and doughy masses, often resembling “a bag of worms” oral lesions are discrete, non-ulcerated nodules, which tend to be of same color of normal mucosa. Buccal mucosa, palate, alveolar ridge, vestibule, and tongue are the common sites of presentation others being, lips, and gingiva. It usually appears as a nodular, well defined, mobile, and sessile mass with slow growth. Although painless, but pain or paraesthesia may occur due to nerve compression, concordant with our findings.

PN predominantly affects children as reported by Yamada et al. a 7-month-old infant presented with respiratory distress and a sublingual mass. The patient died due to respiratory failure and an autopsy revealed laryngeal submucous PN nodules as well as extensive PN involving the vagal, recurrent laryngeal, and phrenic nerves. However in our patient, we did not encounter any lesion in the upper airway. Another case of solitary PN which manifested as a double lip and without systemic or familial involvement has been reported in a 4-year-old girl.

**CONCLUSION**

The presence of PN in a patient with generalized syndrome of NF-1 is relatively an easy diagnosis, but a solitary PN in a patient with no other symptoms is a diagnostic challenge, more so when the location of the lesion is one of the rarest sites. However, thorough clinical examination, gross and microscopic features help to achieve a diagnosis of PN after ruling out the conventional differential diagnosis of lip swellings. Therefore, in an asymptomatic lip swelling a differential diagnosis of PN should be kept in mind and the patient be evaluated on these lines.

**REFERENCES**


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