Solitary plexiform neurofibroma: An unusual clinical presentation on the lip

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Abstract

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 a proliferation of Schwann cells from the inner aspect of the nerve sheath, thereby resulting in an irregularly thickened, distorted, and 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 characteristically an autosomal dominant (AD) disorder affecting the bone, the nervous system, soft tissue, and the skin.[1] It is usually seen 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 is not associated with systemic and hereditary factors whereas the generalized form is associated with the same. World Health Organization 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 usually represents an anomaly rather than a true neoplasm, where the proliferation of Schwann cells is seen from the inner aspect of the nerve sheath and thus resulting in an irregularly thickened, distorted, and tortuous structure. This NF usually shows diffuse enlargements of multiple fascicles of the nerves and its branches. Commonly, it is a slow-growing and locally infiltrative benign tumor, however, have a greater chance of malignancy when located deeply.

Nerve sheath tumors are rarely encountered in the oral cavity. PN in patients with oral involvement, but without other signs of NF is rarely seen.[2] Around 4-7% of patients affected by NF display oral manifestations.[3] Usually, oral lesions occur on the buccal, palate, alveolar mucosa, vestibule, and the tongue, as discrete, non-ulcerated nodules, tending to be of the same color as the normal mucosa.[2] Hereby, we report an interesting case of PN of the lip swelling, an unusual, and an extremely rare site.

Case Report

A female patient aged 23 years reported to the Department of Oral and maxillofacial surgery Sri Rajiv Gandhi College of Dental Sciences, Bengaluru, with a chief complaint of single diffuse swelling on her upper lip at the midline level [Figure 1]. There was no history of any pain or bleeding. The swelling measured about 0.8 cm × 0.6 cm in size, regular in shape, and
well-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 was no folding present on the swelling. Café au lait spots were not identified over the body.

A clinical diagnosis of mucocele was made. The patient was subjected to excisional biopsy, and a single grayish white soft tissue bit was sent for histopathological examination. On gross examination, the largest tissue piece was 0.7 cm × 0.5 cm with skin attached. Cut section was homogenous and grayish white in color [Figure 2]. Microscopic examination showed an overlying parakeratinized stratified squamous epithelium of reduced thickness representing epithelial atrophy, while the underlying subepithelial tissue showed encapsulation of myxoid NF lobules with central nerve bundles [Figures 3 and 4]. The adnexal tissue was unremarkable. On the basis of histopathology, a diagnosis of PN was made [Figure 5].

Figure 1: Clinical picture of the solitary diffuse swelling present on the upper lip at the midline

Figure 2: Macroscopic picture of the excised specimen measuring 0.7 cm × 0.5 cm × 05 cm in size

Figure 3: Microphotograph showing stratified squamous epithelium of reduced thickness, while the underlying subepithelial tissue showed encapsulation and myxoid neurofibromas lobules with central nerve bundles (H and E, ×4)

Figure 4: Microphotograph showing subepithelial tissue comprising myxoid neurofibromas lobules with central nerve bundles (H and E, ×10)

Figure 5: Microphotograph showing numerous Schwann cells with wavy nuclei (H and E, ×40)
Discussion

NF also known as von Recklinghausen disease is a genetically inherited disease, which was first described in 1882.[2] NF occurs in two distinct variants, and they 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.[1,3] However, NF Type-2 (central NF) is an AD disease, accounts 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 which present in three forms: Local discrete, generalized NF, and PN.[4] The tumor per se is 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.[4]

It is extremely rare to find a plexiform neurofibroma over the lip. It is seen quite less commonly 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 are quite variable.[6] It runs in the form of tortuous cords growing centripetally along the segments and branches of a nerve. The diffuse enlargements of multiple fascicles of the nerves and its branches lead 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 usually described as a network-like growth of tumor which involves multiple fascicles of a nerve, forming 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 NF-1 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 literature studied.[1]

Close review of literature of this lesion showed only on reported case on the gingival making this report even more rare which was found as a lip swelling. Till date, very few cases of NF of the lip have been found to be published.[2]

Skin lesions appear as soft, drooping and doughy masses, often resembling “a bag of worms.”[5] Oral lesions are discrete, non-ulcerated nodules, which tend to be of same color of normal mucosa. The most common sites of presentation are buccal mucosa, palate, alveolar ridge, vestibule and tongue others being, lips, and gingiva.[6] It usually appears as a nodular, well defined, mobile, and sessile mass with slow growth.[6]

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.[6]

Conclusion

The presence of PN in a patient with the generalized syndrome of NF-1 is relatively an easy diagnosis, but the diagnosis of solitary PN in a patient with no other symptoms becomes 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.

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References
