Case Report

Solitary Neurofibroma over Lower Lip: A Rare Manifestation

Shishira R. Jartarkar, B. Spoorthy, Sruthi Kareddy

Department of Dermatology, Venereology and Leprosy, Vydehi Institute of Medical Sciences and Research Centre, Bengaluru, Karnataka, India

Abstract

Neurofibromas are common nerve sheath tumors, occurring either sporadically or associated with Von Recklinghausen's disease. Only 6.5% of solitary lesions are seen to involve the oral cavity without any features of neurofibromatosis type 1 (NF-1). It presents as a soft, skin-colored nodule with a characteristic buttonhole invagination. Histologically, it is an unencapsulated lesion consisting of proliferated neural elements, with a background of mucin and mast cells. Surgical excision is the treatment of choice. A 49-year-old female patient presented with a 35-year history of single, asymptomatic lesion over the lower lip, with no features of NF-1. On the basis of the history, histopathological findings, and dermoscopy, a diagnosis of neurofibroma was made and the lesion was excised, with no recurrence over a period of 1 year. The present case report has been reported for its interesting presentation and unusual site of involvement.

Keywords: Lower-lip, neurofibroma, solitary

Introduction

Neurofibromas are the most common benign peripheral nerve sheath tumors, composed of Schwann cells, perineural-like cells, and fibroblasts.[1] They can occur either as localized lesions or a part of generalized syndrome of neurofibromatosis type 1 (NF-1). Solitary neurofibromas develop as a well-circumscribed mass along a peripheral nerve but are unencapsulated. The skin is the most commonly involved site, rarely occurring in the oral mucosa.[2] Most of the neurofibromas present in the second to third decade of life, with no sex predilection.

Only 25% of cases are seen to be involving the head and neck regions. Out of which 6.5% of patients present with oral lesions. Tongue is the most common site of involvement, with rare presentation over the lips.[3]

Microscopically, the neurofibromas of sporadic or syndromic origin are similar, composed of spindle cells with myxoid stroma and scattered mast cells. They are immunopositive for S-100 protein.

Here we report a case of solitary neurofibroma over the lower lip in the absence of any signs of NF-1.

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Quick Response Code:

Website: www.jcasonline.com

DOI: 10.4103/JCAS.JCAS_100_21

Case Report

A 49-year-old female patient presented with a 35-year history of solitary swelling over the lower lip nonprogressive in size or number. It was not associated with any pain, itching, or bleeding. No similar lesions were noted elsewhere on the body.

There was no spontaneous resolution seen over years. There was no significant medical or family history.

Physical examination revealed a solitary skin-colored, well-defined, firm, compressible, nontender nodule of 0.5 cm × 0.5 cm size present over the left side of lower lip, with no secondary changes. No café au lait spots, axillary freckling, skeletal abnormalities, and Lisch nodules were noted. The findings of systemic examination were unremarkable [Figure 1].

On dermoscopy, a yellowish-white mass with an overlying vascular network in a reticular pattern was seen [Figures 2 and 3].

Address for correspondence: Dr. B. Spoorthy, Department of Dermatology, Venereology and Leprosy, Vydehi Institute of Medical Sciences and Research Centre, 82, Near BMTC 18th Depot, Vijayanagar, Nallurhalli, Whitefield, Bengaluru 560066, Karnataka, India.
E-mail: bspoorthy94@gmail.com

How to cite this article: Jartarkar SR, Spoorthy B, Kareddy S. Solitary neurofibroma over lower lip: A rare manifestation. J Cutan Aesthet Surg 2022;15:189-92.

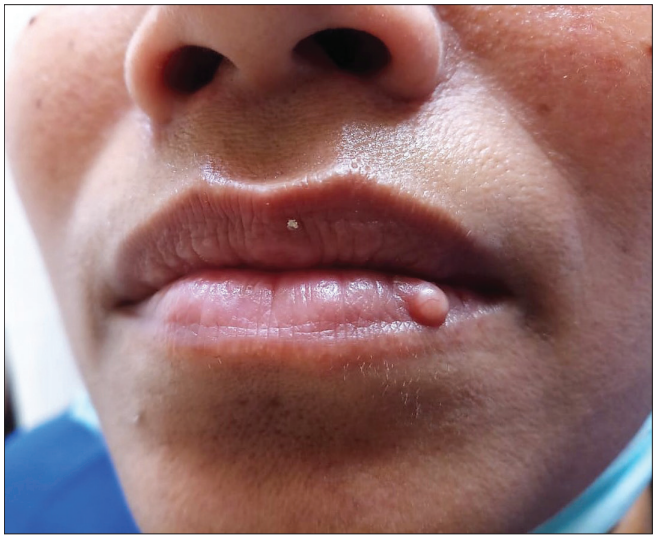

Figure 1: A solitary, well-defined soft, skin colored nodule, smooth surface of size 1 cm × 1 cm over the left side of lower lip

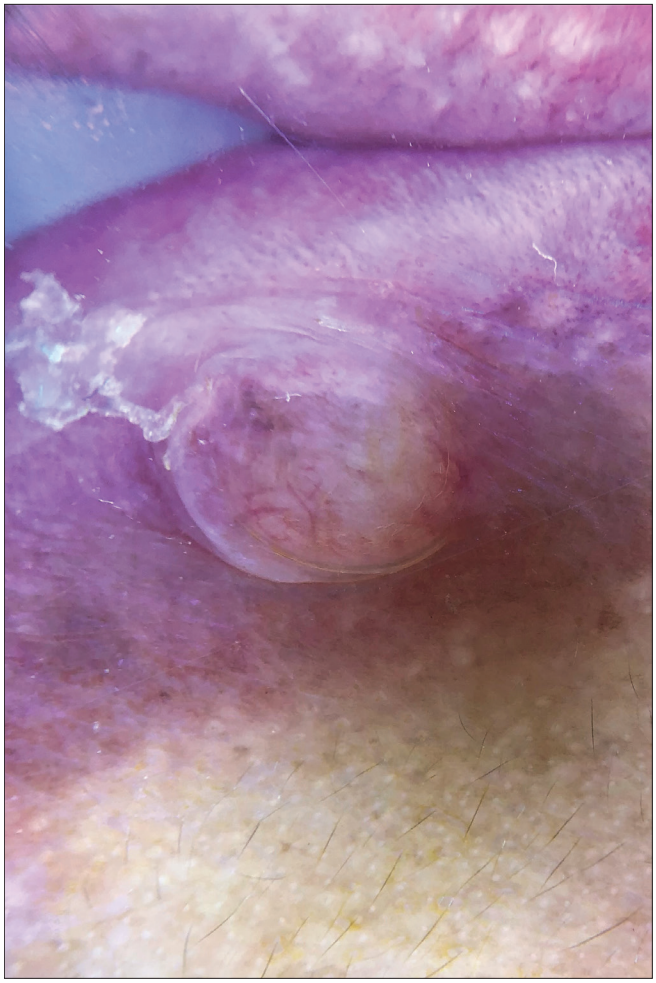

Figure 2: Dermoscopy showing a white-to-yellow homogenous lesion with normal surrounding skin

A provisional diagnosis of mucocele, fibroma, or lymphatic hemangioma was made, and the lesion was excised completely.

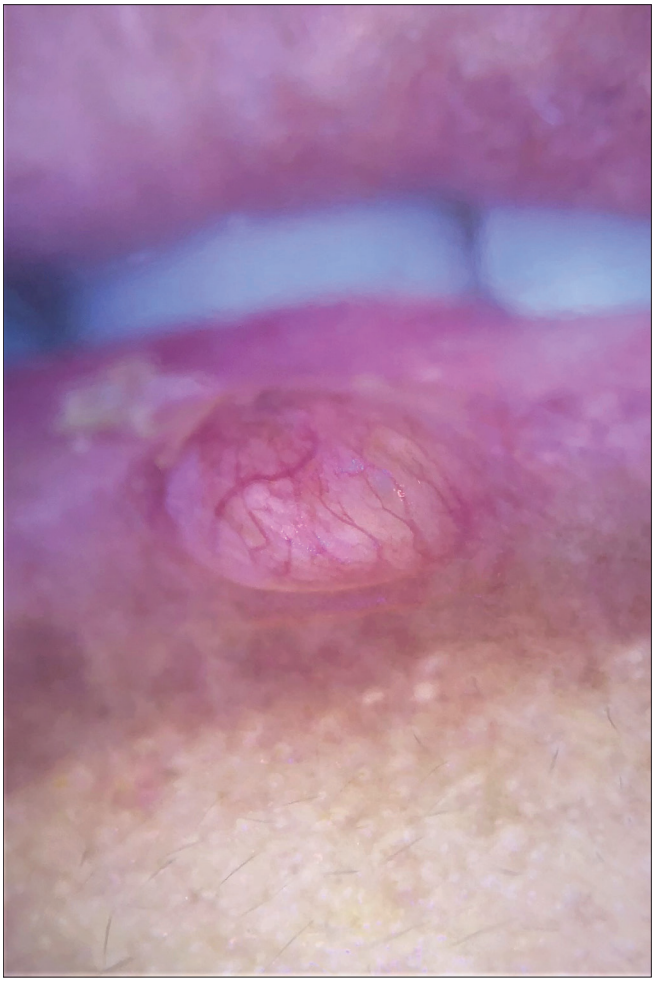

Figure 3: Focal dermoscopy showing a reticular arrangement of vessels overlying the lesion, which is skin-colored

On histopathological examination, an encapsulated lesion composed of bland spindle cells arranged in a storiform pattern was seen in the dermis. It was interspersed with collagen bundles and scattered mast cells, with hyalinized thickened blood vessels [Figures 4 and 5].

On the basis of clinicopathological correlation, a diagnosis of neurofibroma (a spindle cell neoplasm) was made.

Discussion

Neurofibromas are benign tumors of the nerve sheath, which can occur sporadically or as a component of neurofibromatosis. When one or two such lesions are found, they are usually sporadic in nature without any systemic manifestations. However, more than three neurofibromas point toward neurofibromatosis, with other cutaneous, skeletal, and neural manifestations.[4]

Neurofibromas in the head and neck region can be either cutaneous, mucosal, or deep neural bundles, usually related to facial, glossopharyngeal, or vagus nerves.

Oral cavity neurofibromas usually occur as a part of syndrome, that is, NF-1 or NF-2. Solitary neurofibromas

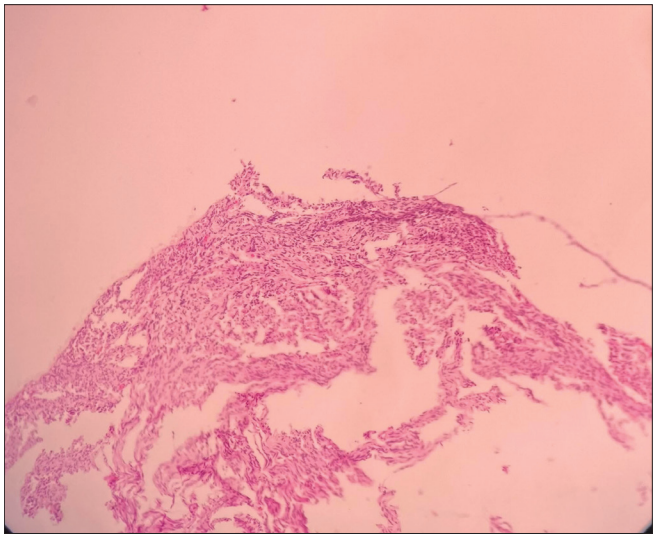

Figure 4: Histology on 10× showing encapsulated lesion composed of neural tissues in a storiform pattern in the dermis

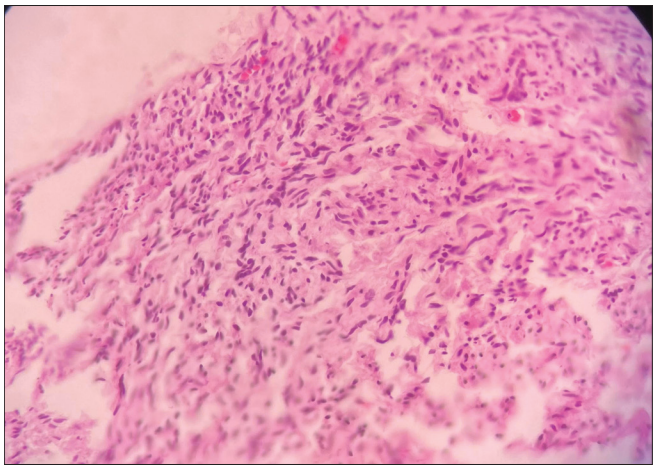

Figure 5: Histology on 40× showing cells with a wavy serpentine nuclei, interspersed with collagen bundles and mast cells

are rare in the oral cavity with a prevalence of 4%–7%. On the basis of the literature, tongue is the most common site followed by palate, mandibular ridge, maxillary ridge, buccal mucosa, and gingiva (in the order of frequency). They appear as pedunculated or sessile nodules, characterized by slow growth and asymptomatic nature.[4,5]

Solitary cutaneous neurofibromas are not associated with axillary freckling, café au lait spots, Lisch nodules, scoliosis, or any family history of NF-1.

Differential diagnosis includes mucocele, fibroma, dermatofibroma, lipoma, and lymphatic hemangioma. Mucoceles are cysts due to mucus accumulation, presenting as bluish, soft, and translucent swellings, often preceded by trauma. They usually resolve spontaneously. Lipomas are yellowish, soft swellings composed of fat tissue, which show the characteristic slip sign. Fibromas are nodules with hard consistency, sessile, and located often on the buccal mucosa.

Microscopically, they are unencapsulated spindle cell tumors with a bland appearance, restricted to corium or extending till subcutis. They are arranged in various patterns such as thin fascicles, storiform, or in haphazard fashion. Surrounding dermis shows myxoid changes with few scattered mast cells. This appearance has been called "shredded carrot" appearance. In 85%–100% of cases, immunostaining with S-100 confirms its neural origin.[6,7]

On dermoscopy, yellow-to-skin-colored background is the most commonly observed global pattern. However, focal dermoscopy shows exaggerated skin markings and pigment-like pattern frequently. Dermatofibroma shows comedo-like openings with irregular crypts.[7] Mucocele shows either reticular or hairpin-like vessels on an erythematous base.[8]

Solitary oral neurofibromas are stable in nature, which seldom shows a malignant transformation into a sarcoma. Treatment modalities include physical removal like surgery, radiofrequency ablation, CO_2 laser, and electrodesiccation. If admixed with nerves, preservation of nerve is of utmost importance.[9,10] Recently, FDA has approved Selumetinib (MEK inhibitor) for symptomatic neurofibromas.

Conclusion

Solitary neurofibromas over the lips are an unusual presentation and are one of the differential diagnoses of solitary nodules. They usually do not show any other features of NF-1 or NF-2. They are characterized by slow growth and asymptomatic nature. Excision is the treatment of choice, with minimal recurrence rates.

Providing insight into this cutaneous disorder is important for patient education and positive outcomes to affected individuals.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

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.

References

1. Broly E, Lefevre B, Zachar D, Hafian H. Solitary neurofibroma of the floor of the mouth: Rare localization at lingual nerve with intraoral excision. BMC Oral Health 2019;19:197.

2. Takahashi G, Matsuyama H, Nakahashi K. A rare solitary neurofibroma of the philtrum of upper lip. Dent Oral Maxillofac Res 2020;6:1-3.
3. Marocchio LS, Oliveira DT, Pereira MC, Soares CT, Fleury RN. Sporadic and multiple neurofibromas in the head and neck region: A retrospective study of 33 years. Clin Oral Investig 2007;11:165-9.
4. Roy P, Chakraborty S, Das S, Roy A. Solitary neurofibroma at the base of the tongue: A rare presentation. Indian J Dermatol 2015;60:497-9.
5. Maruyama M, Fushiki H, Watanabe Y. Solitary neurofibroma of the floor of the mouth: A case report. Case Rep Otolaryngol 2011;2011:967896.
6. Wick M. Cutaneous tumors and pseudotumors of the head and neck. In: Gnepp D, Bishop J, editors. Gnepp's diagnostic surgical pathology of the head and neck. 3rd ed. Amsterdam: Elsevier; 2020. p. 1012-97.
7. Behera B, Kumari R, Thappa DM, Gochhait D, Srinivas BH, Ayyanar P. Dermoscopic features of solitary neurofibroma: A retrospective analysis of 32 cases. Australas J Dermatol 2020;61:e406-9.
8. Ayhan E, Toprak SF, Kaya Ş, Akkaynak Ş. Dermoscopy of oral mucocele: Three types of extravasation mucoceles. Turk J Med Sci 2020;50:96-102.
9. Venkataswamy AR, Ranganath SP, Challapalli SM, Mohanthy L. Nonsyndromic solitary neurofibromas in the oral cavity: Case series and literature review. J Indian Acad Oral Med Radiol 2016;28:52-6
10. Chamseddin BH, Le LQ. Management of cutaneous neurofibroma: Current therapy and future directions. Neurooncol Adv 2020;2:i107-16.