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Case Report

Oral plexiform neurofibroma not associated with neurofibromatosis type I: case report

Luciana S. Marocchio[1)], Michele C. Pereira[1)], Cléverson T. Soares[2)] and Denise T. Oliveira[1)]

[1)]Department of Stomatology, Area of Pathology, Bauru School of Dentistry, University of São Paulo, São Paulo, Brazil
[2)]Department of Pathology, Lauro de Souza Lima Research Institute, São Paulo, Brazil

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Abstract: An unusual case of isolated plexiform neurofibroma arising in the oral cavity without other clinical manifestations or family history of neurofibromatosis-1 (NF-1) is described. The tumor was histopathologically analyzed and an immunohistochemical panel comprising S-100 protein, epithelial membrane antigen (EMA), collagen IV, and CD34 was performed. Typical features of plexiform neurofibroma characterized by enlarged nerve fascicles composed of elongated nuclei and scant cytoplasm cells were identified. Subjacent to the oral epithelium, tactile-like bodies were also detected. On the basis of this report, we would like to emphasize that plexiform neurofibroma can occur in the oral cavity as a benign isolated tumor in patients without other stigmata of NF-1. (J. Oral Sci. 48, 157-160, 2006)

Keywords: neurofibroma; isolated plexiform neurofibroma; diagnosis.

Introduction

Plexiform neurofibroma is a poorly circumscribed, diffuse enlargement of neural sheets that typically involves major nerve trunks of the head and neck region because of the rich innervation of this area (1-4). Although this benign tumor has long been recognized as a pathognomonic criterion of neurofibromatosis type 1 (NF-1 or von Recklinghausen's disease), it may also occur as a solitary lesion arising in a nerve root (1,2,5). When solitary plexiform neurofibroma occurs in patients without stigmata or family history of NF-1, the tumor probably represents the segmental form of NF-1 caused by a later somatic mutation (6-9). A review of the literature showed that, particularly within the oral cavity, solitary plexiform neurofibroma not associated with NF-1 is exceedingly rare (4,7,10-12). This article describes an unusual case of isolated plexiform neurofibroma in a female patient with no other manifestation or family history of NF-1 and discusses the importance of differentiating between isolated and NF-1-associated neurofibromas.

Correspondence to: Dr. Denise Tostes Oliveira, Faculdade de Odontologia de Bauru, Área de Patologia, Alameda Octávio Pinheiro Brisolla, 9-75 CEP 17012-901, Bauru, São Paulo, Brazil
Tel: +55-2114-32358251
Fax: +55-2114-32234679
E-mail: d.tostes@fob.usp.br

Case Report

A 24-year-old woman was referred to Bauru Dental School for evaluation of a painless swelling in the right cheek. Intraoral examination revealed a well-circumscribed mass, firm in consistency, measuring 1.8 × 1.1 cm and covered by normal mucosa. Inflammatory fibrous hyperplasia was clinically suspected, but no history of trauma at this site was obtained. The lesion was surgically excised under local anesthesia and the specimen was submitted to routine histopathological analysis. In addition, immunohistochemical studies including S-100 protein (polyclonal antibody, dilution 1:800, Dako, Carpenteria, CA, USA), EMA (polyclonal antibody, dilution 1:80, Dako, Carpenteria, CA, USA), collagen IV (polyclonal antibody, dilution 1:80, Dako, Carpenteria, CA, USA), and CD34 (polyclonal antibody, dilution 1:50, Dako,

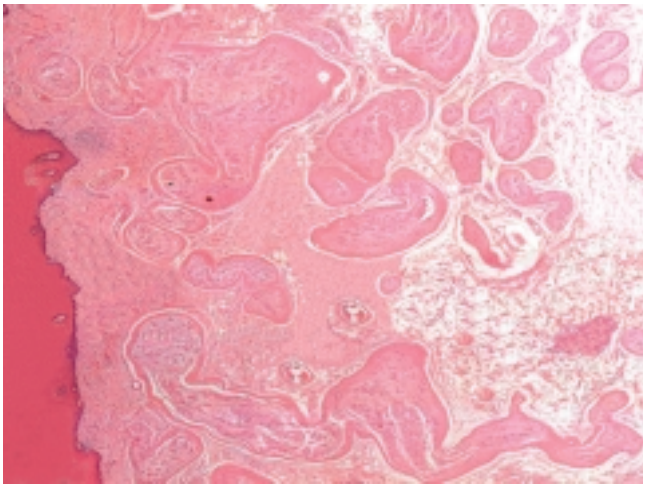

Fig. 1 Panoramic view of the plexiform neurofibroma composed of spindle-shaped cells, delicate wavy collagen fibrils, and a considerable amount of mucin within expanded nerve fascicles (H&E stain; magnification ×25).

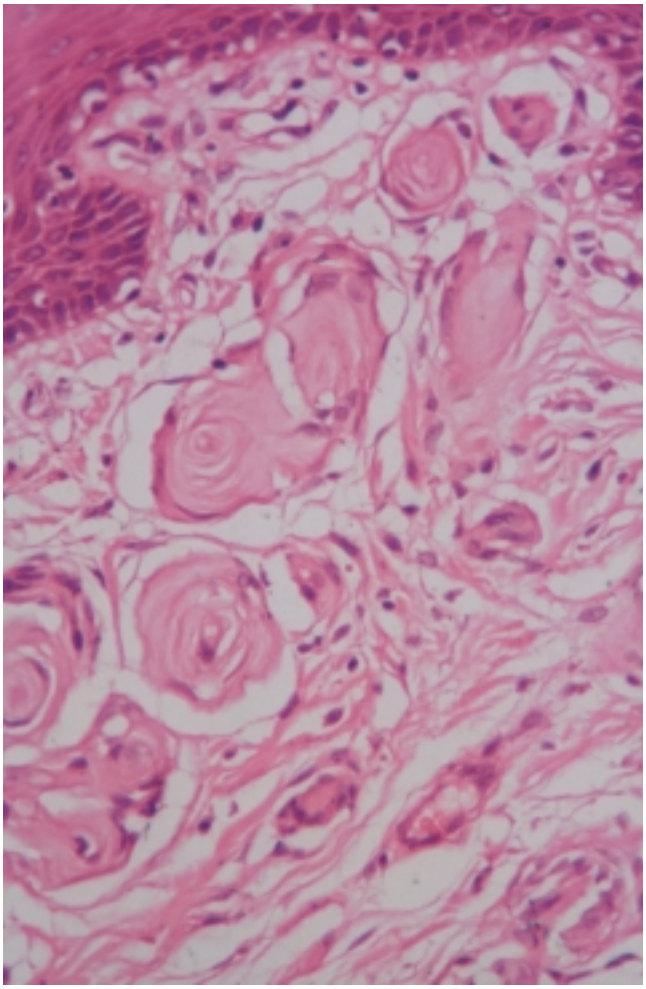

Fig. 2 Detailed appearance of the tactile-like bodies resembling pacinian corpuscles (H&E stain; magnification ×400).

Carpenteria, CA, USA) were performed.

Histopathological examination revealed multiple enlarged fascicles of nerve cells with elongated nuclei and scant cytoplasm embedded in a stromal mucin and collagenous matrix within fibrous connective tissue (Fig. 1). Tactile-like bodies were detected subjacent to the oral epithelium (Fig. 2). Immunohistochemical studies showed that the most of the cells within the nerve fascicles as well as the tactile-like bodies were strongly positive for S-100 (Fig. 3). The perineurium surrounding the nerve fascicles expressed a cellular phenotype that was EMA positive (Fig. 4) and S-100 negative. Collagen IV expression was typically pericellular and involved some cells in the stroma (Fig. 5). CD34-positive cells were observed within the connective tissue surrounding the lesion and in the blood vessels (Fig. 6). On the basis of the histopathological features and strong positive staining for S-100, a diagnosis of plexiform neurofibroma was made. The patient was investigated for

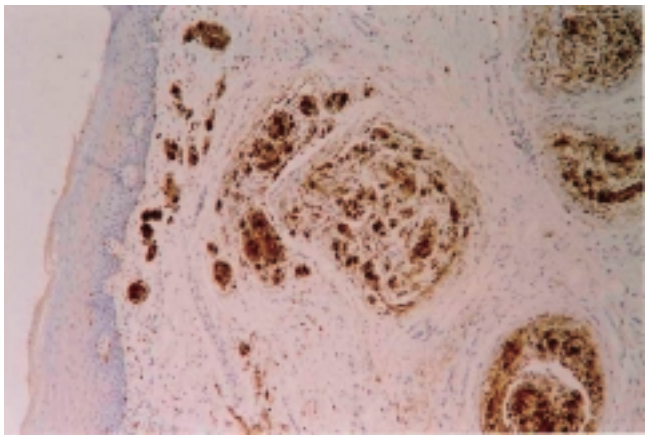

Fig. 3 Photomicrograph of the lesion showing Schwann cells and tactile corpuscle-like structures positive for S-100 (anti-S-100; magnification ×50).

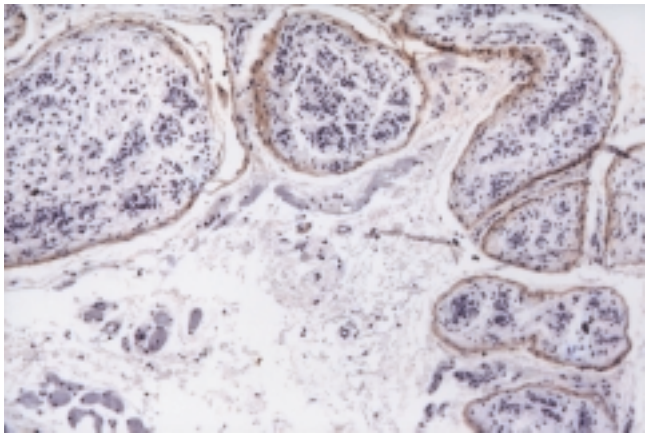

Fig. 4 Immunostaining for EMA in the oral plexiform neurofibroma (anti-EMA; magnification ×50).

the other manifestations of NF-1 and no typical features of this syndrome were detected. No recurrence of the tumor has been detected in three years' follow-up.

Discussion

Most neurofibromas in the head and neck region tend to be solitary tumours, but the occurrence of isolated plexiform neurofibroma affecting peripheral nerves without any other stigmata of NF-1, as in the case reported, is unusual in the oral cavity. Microscopically, as illustrated in Fig. 1, the plexiform neurofibroma exhibited typical features characterized by the presence of multiple relatively well demarcated fascicles of spindle-shaped nerve cells, most of them positive for S-100 (Fig. 3). An uncommon microscopic feature in the presented case was the presence of tactile-like bodies resembling pacinian corpuscles (Fig. 2) detected subjacent to the oral epithelium. These tactile-like bodies exhibited strong S-100 positivity.

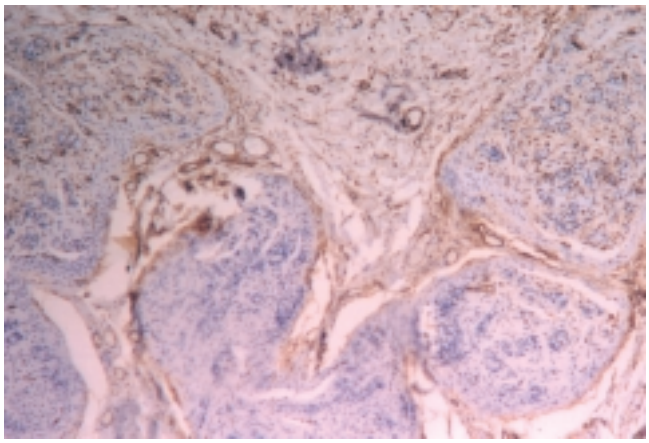

Fig. 5 Pericellular and stromal expression of collagen IV (anti-Collagen IV; magnification ×100).

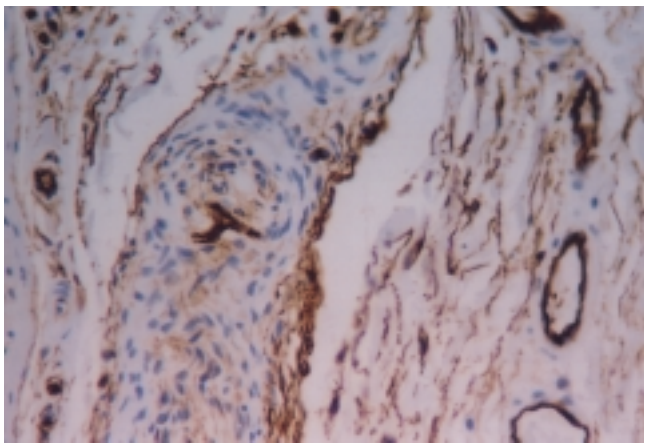

Fig. 6 Intense positivity for CD34 protein in the fibroblastic and endothelial cells of the oral plexiform neurofibroma (anti-CD34; magnification ×100).

The type of plexiform neurofibroma seen in the present case could be a clinical manifestation of segmental neurofibromatosis resulting from mosaicism of NF-1. Although genetic testing for some of the mutations of the NF-1 gene is available, there is no evidence that such testing is helpful in diagnosing NF-1 in patients with isolated plexiform neurofibroma (8). Moreover, the length of the locus and the heterogenicity of the DNA sequencing of the NF-1 gene mutation has, to date, precluded the availability of genetic testing except on a research basis (5). The reported patient did not undergo genetic evaluation, however, the fact that neither recurrence nor other clinical manifestations of NF-1 have been detected on long-term follow-up suggests that this plexiform neurofibroma is likely hyperplastic in nature.

Distinguishing between isolated neurofibromas and those associated with NF-1 is important because the treatment and prognosis differ greatly. Furthermore, neurofibromas associated with NF-1 are more likely to recur or undergo malignant transformation (1,2,8). The present case report intends to emphasize that plexiform neurofibroma can occur in the oral cavity as a benign, isolated, and superficial tumour in patients with no family history or other features of NF-1. Moreover, it reinforces the literature suggesting that in this case, the tumour could be hyperplastic or hamartomatous rather than neoplastic in nature (1,2,5,9).

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