Case Report / Olgu Sunumu

Solitary plexiform neurofibroma of the buccal region unassociated with neurofibromatosis type 1

Nörofibromatöz tip 1 ile ilişkili olmayan bukkal bölgenin izole pleksiform nörofibromu

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Neurofibroma is a benign neural tumor. Plexiform type of this tumor is rarely seen in oral cavity in solitary form. In this article, we present an 18-year-old male case with an isolated plexiform neurofibroma localized at buccal region without any other manifestation or family history of neurofibromatosis type 1.

Key Words: Buccal region; neurofibroma; neurofibromatosis; oral cavity; plexiform; solitary.

Nörofibrom, benign nöral bir tümördür. Bu tömürün pleksiform tipi, nadiren tek başına oral kavitede görülmektedir. Bu yazıda nörofibromatöz tip 1'in diğer bulguları veya aile öyküsü olmadan, bukkal bölgede izole olarak ortaya çıkan pleksiform nörofibrom izlenen 18 yaşında erkek bir olgu sunuldu.

Anahtar Sözcükler: Bukkal bölge; nörofibrom; nörofibromatozis; oral kavite; pleksiform; izole.

A neurofibroma is a poorly-circumscribed benign tumor arising from neurons and perineural cells.[1] The most commonly-affected site is the trunk, followed by head, neck and limbs. Although the head and neck region is often affected[1] because of diffuse neural innervation of this area,[2] neurofibromatosis (NF) is rarely seen in the oral cavity. We describe a solitary plexiform neurofibroma of the buccal region in an 18-year-old male with no other manifestation or family history of neurofibromatosis type 1 (NF1) and discuss the importance of differentiating between solitary and NF1 - associated solitary or multiple neurofibromas.

CASE REPORT

An 18-year-old male was referred to our ear nose throat (ENT) clinic with a mass in the right cheek of approximately 10-12 years' duration. There had been mild pain for six months but no paresthesias or numbness. There was no history of trauma, regression or enlargement in size or any discharge. The mass did not cause any aesthetic problem and was not obvious on inspection. A 3.5x1.5x2 cm

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firm, nontender, mobile, nonpulsatile mass was palpable. Ultrasonography (USG) confirmed a 3x2x2 cm solid, well defined subcutaneous mass anterior to the masseter muscle. The rest of the medical history was unremarkable and routine laboratory tests were within normal limits. Fine needle aspiration (FNA) biopsy suggested an epidermoid cyst. The patient underwent surgical excision of the mass under local anesthesia. Histopathological and immunohistochemical analysis revealed multinodular lesions composed of spindle-shaped schwann cells aligned in a plexiform manner, forming bundles embedded in a loose myxoid stroma (Figure 1, 2). The diagnosis was plexiform neurofibroma. The patient had no family history of NF1. On physical examination, no other cutaneous and subcutaneous lesions were found. Neurological assessment was normal and no lesions were found on magnetic resonance imaging (MRI) scan of the brain. Lisch nodules were not noted on eye examination. As a result, we diagnosed a solitary plexiform neurofibroma of the buccal region without NF1. The patient was discharged uneventfully and no recurrence was seen during one-year follow-up.

DISCUSSION

Benign nerve sheath tumors may be divided into neurofibromas and schwannomas.[3] Schwannomas arise from schwann cells. They are usually solitary and encapsulated. Neurofibromas originate from schwann cells and fibroblasts.[1] They are nonencapsulated and can be solitary or multiple. They may occur as a part of the syndrome of NF1[2] of which they are the most common benign tumors. Neurofibromatosis is the term used to describe a group of genetic disorders that primarily affect the cell growth of neural tissues. The most common form is NF1, often known as Von Recklinghausen's Disease. This accounts for about 90% of cases. It is one of the most common autosomal dominant inherited disorders with an incidence of one in 3000 births. The National Institutes of Health (NIH) Consensus Development Conference has proposed the criteria for diagnosis of NF1 in 1988.[4] The diagnostic criteria for NF1 are met if a patient has two or more of these features. Using NIH criteria, a diagnosis of NF1 can be made in 94% of patients by age six years. This rate is 100% by age 20. There is a positive family history in half of patients and the remaining represent spontaneous mutations.[5] Three types of neurofibromas exist: cutaneous, subcutaneous and plexiform. Plexiform neurofibroma is an unusual variant of the common neurofibroma.[2] Cutaneous and subcutaneous varities are not specific for NF1. Some studies and several respected textbooks published after 1990 have claimed that the multiple plexiform neurofibroma is pathognomonic for NF1.[2,6] Plexiform neurofibroma is uncommon and occurs almost 5-15% of patients with NF1. But there are few articles that report the occurrence of solitary plexiform neurofibroma affecting peripheral nerves without any other stigmata of NF1, as in our patient.[1,2,7-12] Neurofibromas are most commonly skin lesions that are rarely seen in the oral cavity.[1] Also, neurofibromas associated with NF1 are generally encountered as multiple lesions and rarely occur alone in the oral cavity. The real frequency of isolated neurofibromas

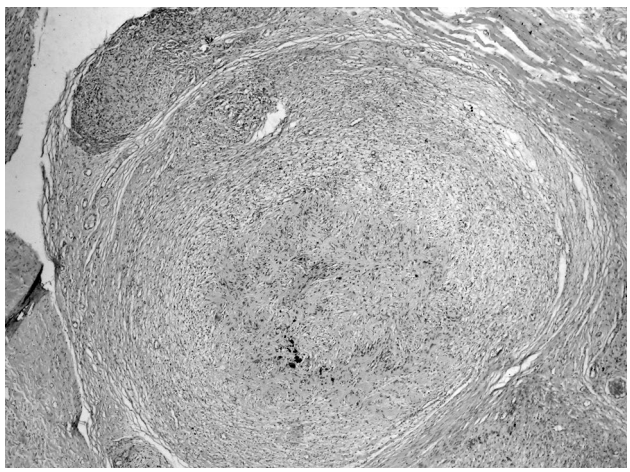

Figure 1. *Multinodular pattern of schwann cells (H-E x 40)*

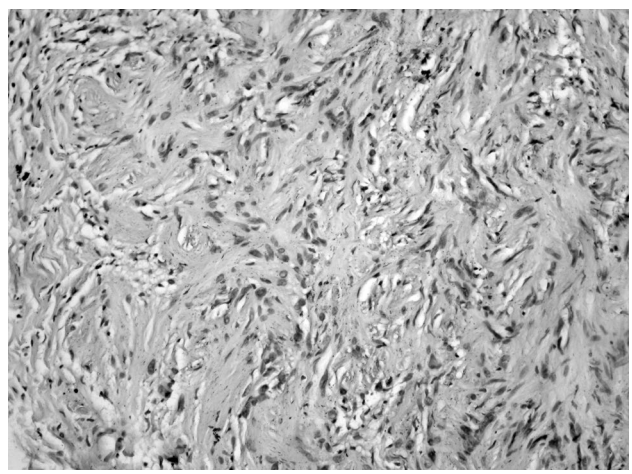

Figure 2. *Spindle shaped schwann cells alligned in a plexiform manner and form bundles embedded in a loose myxoid stroma (H-E x 200).*

unassociated with neurofibromatosis in the oral cavity is uncertain, but these tumors have been described in the tongue,[1,7,10,11,13] buccal mucosa,[13] floor of mouth[13] lips[13] gingiva,[13] palate, major salivary glands[14] and mandible.[8,13] The tongue is the most commonly involved site.

Plexiform neurofibromas may appear in a wide variety of sites including the trunk, limbs, head, neck, spinal cord, mediastinum, and abdominal viscera. Plexiform neurofibromas especially involve smaller branches of large nerves. In the head and neck, they most commonly involve the fifth cranial nerve, particularly the first and second divisions.

The natural history of plexiform neurofibroma may vary significantly; some lesions may be silent for many years, while others may grow rapidly, especially during puberty and pregnancy. They can cause cosmetic and functional deformities in the head and neck region. These tumors are known to cause symptoms ranging from minor discomfort to extreme pain. It is important to differentiate these tumors from schwannomas because neurofibromas have a potential of malignant transformation reported between 5-25% and malignant transformation of these benign tumors are the main cause of death in NF1 patients.[2]

Magnetic resonance imaging or CT gives useful information about the limits and extension of tumor. Although FNA has become an important technique in preoperative diagnosis of head and neck masses, the difficulty in obtaining adequate cellular smears for cytology in soft tissue can make he cytodiagnosis difficult or non-diagnostic as an our patient.

Surgery is the treatment of choice for these tumors due to the risk of malignant transformation, but complete resection might be difficult because of invasion into the surrounding soft tissue. Neurofibromas have extensive vascularity and tend to bleed during surgery. Therefore excessive bleeding should be kept in mind while attempting surgical removal. We were able to excise the mass totally without any complication. The consistency of the lesion has been compared to that of a bag of worms because of the presence of soft areas interspersed with firm nodular areas and this very consistency was well appreciable in our patient. On microscopy, plexiform neurofibromas have a loose myxoid background, multiple relatively well-demarcated fascicles of spindle-shaped nerve cells; mostly immunoreactive for S-100 protein.[15]

In conclusion, neurofibromatosis type 1 involves many structures of the head and neck region including the oral cavity, and ENT surgeons should be familiar with the diagnosis. In addition, rare cases of solitary plexiform neurofibroma may be detected unassociated with NF1. In this study we found only two previous reports of solitary plexiform neurofibroma in the buccal region making ours a very rare case. It emphasizes that plexiform neurofibroma may occur in the oral cavity as a solitary tumor in patients with no family history or other features of NF1. Detailed history, systemic examination and radiologic studies are necessary to diagnose solitary plexiform neurofibroma and exclude NF1. We should also remember that young patients may show other features of NF1 as they age. Therefore the patients should be examined and evaluated periodically.

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