



# Evaluation of Intraosseous Neurofibromatosis of the Mandible (type 1) Using Cone Beam Computed Tomography Images: A Case Report

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## ABSTRACT

Neurofibromatosis type 1 is an autosomal dominant neurogenic disorder, which is occurring one in every 3000 of population. We present a 37-year-old man with complaint of hard speech and several masses in his tongue. Neurofibromatosis rarely occurs as intraosseous lesion in its primary form that in this case, it has appeared as unilacunar and well-defined radiolucency in panoramic view. Cone beam Computed tomography (CBCT) was carried out for better evaluation of the extension of mandibular lesions. The aim of this article is to report a sporadic case of NF1 in a family, with multiple soft and hard tissue lesions, and eliminate the complications and functional impairments.

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## Introduction

Neurofibromatosis type 1 is an autosomal dominant neurogenic disorder, which first described in 1882 by von Recklinghausen.<sup>1,2</sup> It is occurring one in every 3000 of population. However, about 50 percent of cases have no family history and represent new mutations. This group has one of the highest rates of mutations among genetic disease.<sup>3,4</sup> NF1 affects the bone, the nervous system, soft tissue, and the skin.<sup>5</sup> It characterized primarily by café-au-lait pigmentation of the skin, multiple neurofiromas, and a wide variety of other possible abnormalities including Lisch nodules, central nervous system tumors, mental deficiency and seizures.<sup>6</sup> The most common location of NF1 is the post portion of the mandible, although a few cases have been reported in the maxilla.<sup>8,7</sup> The radiographic features of the jaw with NF1 can be characteristic and include enlargement of coronoid notch, increasing the angle between the body and the ramus, deformity of condylar head, lengthening of the condylar neck, an elongated coronoid process, and lateral bowing and thinning of the ramus, as seen in basal skull views. Other findings are enlargement of mental and mandibular foramina and the inferior alveolar canal, increased incidence of branched mandibular canal, erosive changes of outer contour of mandible, unerupted teeth and a cystic lesion.<sup>9</sup> We report a case of mandibularneurofibroma in a NF1 patient with panoramic view and cone beam computed tomography (CBCT) features of the lesion.

## Case Report

A 37-year-old man with complaint of hard speech because of several masses in his tongue was referred to the department of oral and maxillofacial medicine. The masses had become noticeable since the last 2 years and had started interfering with his speech. There was no history of pain. Physical examination showed manifestations of neurofibromatosis. Clinically, there were 30 café-au-lait patches and more than 6 spots larger than 1.5cm and cutaneous neurofibromas at the face, neck,

thracolumbar and hand region (fig1). ThereforeNF1 was diagnosed based on these clinical criteria.

The patient had no familial history of NF1. Intraorally, some nodules measuring 3×2×2 Cm were present in left ventral surface of the tongue. On palpation, it was nontender, nonfluctuant and rubbery in consistency. The surface changes were lobulated with multiple corrugates(fig 2). The panoramic view showed, deep sigmoid notch, deformed condylar head, elongated condylar neck, and enlarged mandibular canal on the left side. Furthermore there was a well-defined unilocular radiolucent lesion in the angle of left ramus, which was suggested central neurofibromatosis. It seems the mass interfered with normal eruption of third molar (fig 3). Cone beam Computed Tomography (CBCT) was carried out for better evaluation of the extension of mandibular lesions. Cross-sectional views of the left ramus revealed a large defect on the lingual wall of the mandible, which caused thinning in buccolingual dimension. Enlargement of mandibular foramen and posterior portion of inferior mandibular canal (from distal of third molar to mandibular foramen) wasevident(fig 4).

The excisional biopsy was performed from left ventral surface of the tongue. In histopathology examination, tongue stratified epithelium was found with evidences of hyperplasia and hyperchromatism in basal and parabasal layer. In subconnective tissue spindle cell with wavy nuclear shape, scattered in fibrous connective tissue. There was no evidence of malignant transformation. Immunostaining using anti-S 100 protein was positive, which confirmed to be neurofibroma(fig 5).

## Discussion

The most common form of neurofibromatosis is its type 1 form (NF1), which is known as a skin Von Recklinghausen's disease.<sup>1,2</sup> The disease can be inherited as a dominant autosomal condition, although new mutations are responsible for the disease in half of the cases.<sup>10</sup> The case reported has no family history of the disease and is the only person with the disease in

his family. The age range to develop NF1 is about 14 to 45 years.<sup>11</sup> In addition, the disease has no gender dependency. The recent case is a 37-year-old man who has found about his condition 10 years ago. Pigmented lesions are one of the most common manifestations in NF1 that usually appear during the first years of life or at birth time as Café-au-lait spots or Freckles. The lesions may occur anywhere on the skin; however, they are less common on facial skin. Axillary and inguinal freckles (Crow's sign) are often seen in these patients. In the case patient, more than 6 café-au-lait spots and bilateral axillary and inguinal freckles were reported.

Neurofibromatosis may occur in all intraoral soft and hard tissues. However, the most common intraoral involvement site is tongue. The mentioned case had also multiple neurofibromas lesions on his tongue. Involvement of NF lesions is slow and painless, and the condition may be aggravated during growth, puberty or pregnancy.<sup>7, 10, 11</sup> Partial or complete resection of NF lesions is performed to relieve aesthetic and functional problems; however, it is wise to delay the treatment until the end of growth to reduce the risk of lesions recurrence. Radiotherapy and chemotherapy approaches are not recommended for the treatment.<sup>12</sup> In the case, the skin and tongue lesions at age 37 were removed surgically to improve the patient's beauty and speaking.

The differential diagnosis of these lesions from other fibrosis lesions, such as juvenile fibromatosis and fibromyxoma in primary examination is difficult due to their histological view. It is better to prepare the biopsy of NF1 soft tissue swellings from its deep areas.<sup>11</sup> In this case; the biopsy was prepared deeply from the neck skin and ventral surface of tongue. According to obtained histopathological findings, presence of spindle cells with wavy nuclei within the loose fibrous stroma associated with positive antibody responses to protein S100 in immunohistochemical studies, the neurofibromatosis diagnosis was confirmed.

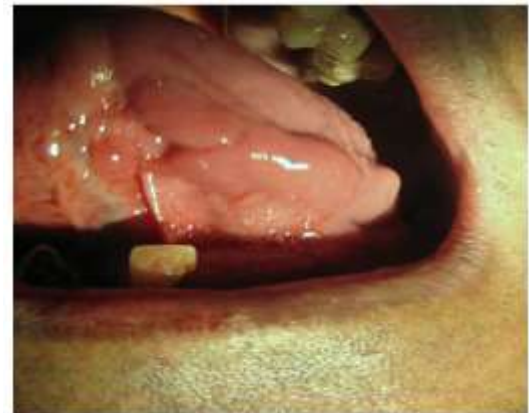
Intraosseous neurofibromatosis in the head and neck is relatively rare. The most common region in these areas is mandible. In this case, the lesions were found in the left side of the mandible. Higher incidence of mandibular NF lesions may be associated with thick and long bundles of inferior alveolar nerve.<sup>4, 8</sup>

Neurofibromatosis rarely occurs as intraosseous lesion in its primary form that in this case, it has appeared as unilacunar and well-defined radiolucency in panoramic view. The most common radiographic changes of NF1 in mandible include widening of mental and mandibular foramen, and mandibular canal. Also, branched mandibular canal has been observed in some cases.<sup>7,9,10</sup> In this case, the widening of mandibular canal and mandibular foramen was seen in panoramic radiographs and CBCT. In addition, radiographic signs suggesting the branched mandibular canal were not found. Other radiographic manifestation included deep coronoid notch, increased mandibular angle, deformed condylar head, lengthened condylar neck, thinning of the ramus, which were all found in one side of the reported patient's mandible.<sup>9</sup> Neurofibromatosis may cause dental malposition or impaction that in this case; the third molar has not completely erupted.<sup>7,10</sup> Neurological deficits and hemorrhage are the most common problems occurring during intraosseous NF lesions surgery.<sup>8</sup> Surgery on the reported case was avoided due to reasons mentioned and considering no change in lesion size over the last ten years. There was no evidence suggesting that surgery may cause malignant transformation. The malignancy change rate in NF1 neurofibromatosis lesions is 3 to 5 %.<sup>7,10,11</sup> NF malignant

transformation has a very bad prognosis, and distant metastases are possible. Some authors believe that relapse may occur after surgical resection, and recurrent relapses also increase the risk of malignancy. It is therefore important that the maxillofacial surgeon or the dentist of a patient with characteristic symptoms of NF1 keeps the patient under prolonged consideration care and follow-up visits.



**Figure 1. Physical examination showed 30 café-au-lait patches and more than 6 spots larger than 1.5cm and cutaneous neurofibromas at the thoracolumbar and arms region**



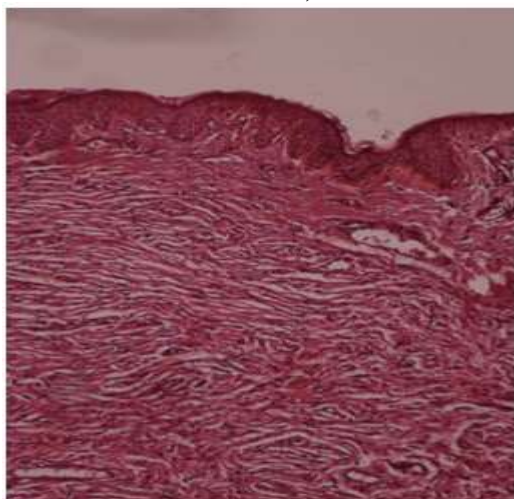
**Figure 2. Oral examination showed some nodules measuring 3x2x2 cm which were present in left ventral surface of the tongue**



**Figure 3. The panoramic view showed, deep sigmoid notch, deformed condylar head, elongated condylar neck, and enlarged mandibular canal on the left side. The black arrows outlining a well-defined unilacunar radiolucent lesion in the angle of left ramus, which was suggested central neurofibromatosis**



**Figure 4.** CBCT Cross-sectional views of the left ramus revealed a large defect on the lingual wall of the mandible, which caused thinning in buccolingual dimension. Enlargement of mandibular foramen and posterior portion of inferior mandibular canal (from distal of third molar to mandibular foramen) was evident



**Figure 5.** histopathologic examination, showed tongue stratified epithelium with evidences of hyperplasia and hyperchromatism in basal and parabasal layer. In sub connective tissue spindle cell with wavy nuclear shape, scattered in fibrous connective tissue. There was no evidence of malignant transformation. (Magnification  $\times 40$ )

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