Neurofibromatosis Type 1 in the Mandible

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Abstract

Neurofibromatosis type 1 (NF1) was first described in 1882 as a hamartomatous disorder of neural crest derivation. We present the imaging of a 65-year-old woman with NF1. Computed tomography revealed that there were three major findings presented: skeletal deformity, an area of fat (probably related to mesodermal dysplasia), and benign neoplasm within the masticator space. Moreover, masticatory muscles were hypoplastic.

Keywords: Computed tomography imaging, mandible, masticatory muscle, neurofibromatosis type 1

Introduction

Neurofibromatosis type 1 (NF1) was first described in 1882 as a hamartomatous disorder of neural crest deviation.[1] NF1 is a well-documented heredity condition characterized by multiple café-au-lait spots, neurofibromas, and skeletal change.

NF1-related craniofacial findings include macrocephaly and sphenoid wing dysplasia.[2] The facial skeletal malformations are results of genetic and epigenetic factors, such as tumor growth.[2] The presence of impacted, displaced, or missing teeth, particularly in the mandible, and overgrowth of the alveolar ridge were recognized as oral manifestation of NF1.[3] In this report, we describe computed tomography (CT) images of a female with NF1.

Case Report

A 65-year-old woman visited the authors' dental hospital for prosthetic treatment in February 2013. In her 20s, she had been diagnosed with dermal NF1 of the face, extremities, and trunk. The patient had no family history of NF1. Excision of subcutaneous neurofibroma on the left cheek was done in her 30s, which recurred in 2011 and was again surgically removed. The patient otherwise has had no symptomatic lesions requiring management.

Physical examination demonstrated that there was diffuse swelling on the angle of the right mandible but no pain at the dental hospital in 2013. The surface mucosa was normal [Figure 1]. Routine panoramic radiography for dental prosthetic treatment revealed a concavity of the right posterior border of the ramus, a deep coronoid notch, and prominent antegonial notching [Figure 2].

CT was performed using Light Speed VCT (GE Healthcare, Milwaukee, WI, USA). Scan conditions were as follows: 120 kVp, 200 mA, 512 × 512 matrix size, 2.5 mm slice thickness, and ϕ25 cm FOV. The contrast enhancement was done by infusing 100 ml of iohexol. Coronal, sagittal, and three-dimensional reformatted images were made after examination. CT revealed that the right condylar head was small and deformed [Figure 3a]. The right coronoid process and lateral pterygoid processes were hypoplastic [Figure 3b]. There was a corticated, irregularly shaped depression of the lateral aspect of the right ramus [Figure 3c]. The right mandibular canal appeared widened [Figure 3d]. The ascending ramus was bowed [Figure 3e]. The right temporal muscle and lateral pterygoid muscles were hypoplastic [Figure 4a and b]. There were enhanced lesions within the right masticatory muscle space [Figure 4b and c], and there were no contrast-enhanced masses, instead hypodense but fat-like tissue adjacent to the bone deformity of the mandible [Figure 4d]. Mandibular asymmetry was evident and reinforced the changes of the right mandible previously

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Access this article online

Quick Response Code:

Website: www.amsjournal.com

DOI: 10.4103/ams.ams_135_17

How to cite this article: Uchiyama Y, Sumi T, Marutani K, Takaoka H, Murakami S, Kameyama H, *et al*. Neurofibromatosis Type 1 in the Mandible. Ann Maxillofac Surg 2018;8:121-3.

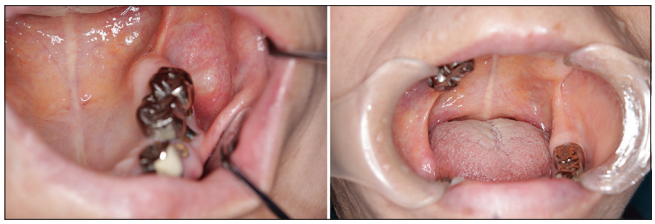

Figure 1: There is diffuse swelling on the angle of the right mandible. The surface mucosa is normal. There is diffuse swelling on the angle of the right mandible. The surface mucosa is normal

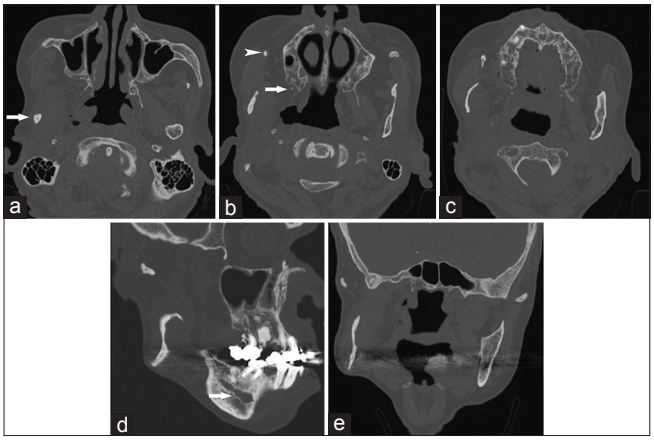

Figure 3: (a) Axial bone computed tomography imaging demonstrates that the right condylar head is small and deformed (arrow). (b) The right coronoid process (arrow head) and lateral pterygoid processes (arrow) are hypoplastic. (c) There is a corticated, irregularly shaped depression of the lateral aspect of the right ramus. (d) The right mandibular canal appears widened (arrow). (e) The ascending ramus is bowed

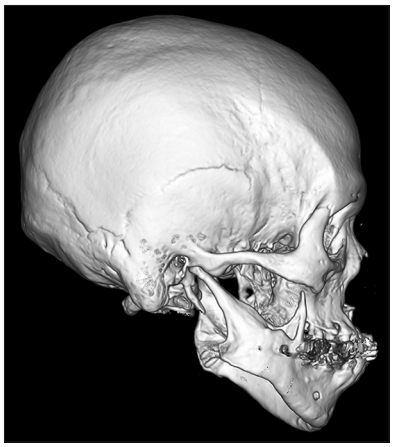

Figure 5: Three-dimensional computed tomography imaging demonstrates mandibular asymmetry is evident, and reinforced the changes of the right mandible. There are concavity of the right posterior border of the ramus, elongated condylar necks, prominent antegonial notching, and hypoplastic appearance of the condyle and coronoid process

noted on the panoramic radiography and bone window CT, including concavity of the right posterior border of the ramus, elongated condylar necks, prominent antegonial notching, and hypoplastic appearance of the condyle and coronoid

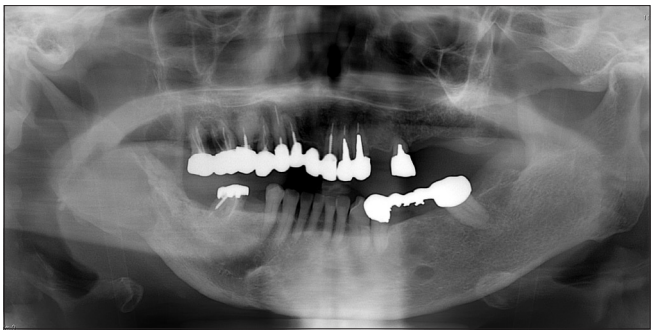

Figure 2: Panoramic radiography reveals a concavity of the right posterior border of the ramus, a deep coronoid notch, and prominent antimonial notching

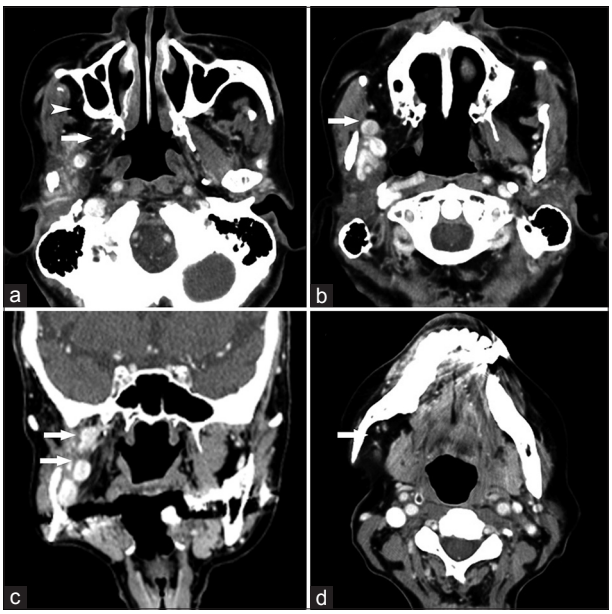

Figure 4: (a) Axial computed tomography imaging demonstrates that the right temporal muscle (arrow head) and lateral pterygoid muscles (arrow) are hypoplastic. (b and c) There are enhanced lesions within the right masticatory muscle space (arrows). (d) There are no contrast-enhanced masses, instead hypodense but fat-like tissue adjacent to the bone deformity of the mandible (arrow)

process [Figure 5]. The sphenoid bone was normal. There was no orbital involvement nor other central nervous system neoplasm, buphthalmos, and falx calcification.

An area of fat-like tissue adjacent to the bone deformity of the mandible which was consistent with a developmental defect would be a more appropriate characterization of this area. The heterogeneously, enhancing, well-defined round masses within the masticator space was consistent with benign neoplasm, most likely neurofibromas given the medical history.

There were no patient symptoms, clinical diagnosis of NF1, and absence of necrotic masses or any other radiographic indicators suggestive of malignancy on contrast-enhanced CT; biopsy was not indicated; and patient has been followed up radiographically in the outpatient clinic.

Discussion

NF1-associated craniofacial abnormalities include macrocephaly and sphenoid wing dysplasia.[2] The facial skeletal malformations occur secondary to genetic and epigenetic factors such as tumor growth.[2]

In classical NF1, skeletal defects occur secondary to abnormalities of derivatives of the neuroectoderm and mesoderm.[4]

Jaffe stated that the skeletal abnormalities seen in NF might represent (1) direct destruction from the proliferation of neurofibromatous tissue and (2) aberrations of skeletal development and growth, either localized or systemic.[5] The latter theory reflects a concept of NF as a disorder deeply rooted in the germplasm.[5] Hunt and Pugh reported that most skeletal defects are expressions of a mesodermal dysplasia, that is without direct association with neurofibromatous tissue.[6] Lee reported that the CT finding of tissue with fat density adjacent to the deformed bone supported the hypothesis that the osseous deformity was not necessarily the result of an immediately adjacent neurofibroma or neoplasm.[7] Instead, adipose tissue might play a role in its development, or the tissue might be the result of mesodermal dysplasia that includes bone.[5] Lee *et al*. suggested that there was no evidence that osseous changes of the mandible in patients with NF1 were the direct result of an adjacent neoplasm in their cases, but that the detection of adjacent tissue supported the hypothesis that changes seen in NF1 might represent manifestations of mesodermal dysplasia.[7] Cohen claimed that skeletal malformations of the face in patients with NF1 were genetic in origin and not secondary to neurofibromas.[8] In this case, fat-like tissue was observed lateral to the ramus related to mesodermal dysplasia and neurofibroma was seen within the masticator space. Thus, it was suggestive that fat-like tissue and neurofibroma occurred separately in this case. Mesodermal dysplasia was thought to be not related with neurofibromatous tissue. These findings coincide with the finding of Lee *et al*.

In this case, from the CT images, there were three major findings presented: skeletal deformity, an area of fat-like tissue (probably related to mesodermal dysplasia), and benign neoplasm within the masticator space.

In this case, the CT images demonstrated that the lateral pterygoid muscle, medial pterygoid muscle, and temporal muscle were hypoplastic. Conventional radiography could not demonstrate changes in these muscles, an area of fat and benign neoplasma. However, CT can reveal changes in the soft tissue. Thus, CT is thought to be useful for evaluation of bone as well as soft tissues in patients with NF1.

In conclusion, in CT findings of the face in this patient with NF1, there was skeletal deformity of the mandible, an area of fat (probably related to mesodermal dysplasia), and benign neoplasm within the masticator space and masticatory muscles were hypoplastic.

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.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

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