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Case report

Von Recklinghausen disease in tongue: A case report

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ABSTRACT

Von Recklinghausen's disease, neurofibromatosis type 1 (NF1), is an autosomal dominant neurogenetic disorder that is known to have characteristic symptoms such as peripheral nerve sheath tumor, neurofibromas, café-au-lait macules and pigmented hamartomas of the iris, called Lisch nodules. Cases of NF1 accompanied by radiological abnormalities of the jaw bones have been reported. In this article we present a case of NF1 with a tongue mass of neurofibroma accompanied on both sides by enlargement of the mandibular canals and mental foramens.

1. Introduction

Neurofibromatosis (NF) is an autosomal dominant neurogenetic disorder that includes three forms: neurofibromatosis type 1 (NF1), neurofibromatosis type 2 (NF2) and schwannomatosis [1]. NF1, also known as von Recklinghausen's disease, is known to display characteristic symptoms such as peripheral nerve sheath tumor, neurofibromas, café-au-lait macules and pigmented hamartomas of the iris, called Lisch nodules. NF1 is caused by mutation of a gene located on chromosomal segment 17q11.2, which encodes a protein known as neurofibromin that plays as a negative regulator of the Ras signal transduction pathway [2,3]. There are cases of NF1 that arise due to spontaneous mutation. NF1 is a relatively common genetic disorder and is present in approximately 1 in 3500 live births [4]. In NF1 of oral region, the soft tissue manifestations have been often described and there have been cases that present radiological abnormalities of jaw bones [4–11]. But the cases that affected the soft tissue and jaw bones concomitantly are rare. In this article we present a case of NF1 involving the tongue in which enlargement of the mandibular canals and mental foramina on both sides was observed.

2. Case report

A 41-year-old male visited our clinic for precise examination of a tongue mass on the left side. He had already been diagnosed as NF 1 in childhood. There was no family history of NF1. He had scattered café-au-lait macules on his trunk, upper and lower extremities, and some cutaneous nodules of different sizes, which had already been biopsied and diagnosed as neurofibroma. He had a slight cognition impairment and was taciturn, but other NF1 associated disease such as optic disturbances (Lisch nodules), macrocephaly, scoliosis, cardiovascular abnormalities and hypertension were not detected. He had received left side tongue mass resection when he was 6 years old and periodic follow-up examination was continued over the next ten years. Two months previously his mother noticed that he was complaining of discomfort in his tongue. When she eventually looked at his tongue she found that the left side of his tongue was enlarged. She convinced that the swelling occurred in the same site previously treated. Subsequently, since there was no improvement in this enlargement of the tongue, the patient was introduced to us by his family dentist for a close inspection.

On oral examination a sessile lobulated mass with irregular borders was found in the left side of the tongue body that had spread from the midline to the left margin and from the dorsal to the ventral surface of the tongue (Fig. 1). The mass was painless and soft, and the covering mucosa was a normal color without ulceration

☆ AsianAOMS: Asian Association of Oral and Maxillofacial Surgeons; ASOMP: Asian Society of Oral and Maxillofacial Pathology; JSOP: Japanese Society of Oral Pathology; JSOMS: Japanese Society of Oral and Maxillofacial Surgeons; JSOM: Japanese Society of Oral Medicine; JAMI: Japanese Academy of Maxillofacial Implants.

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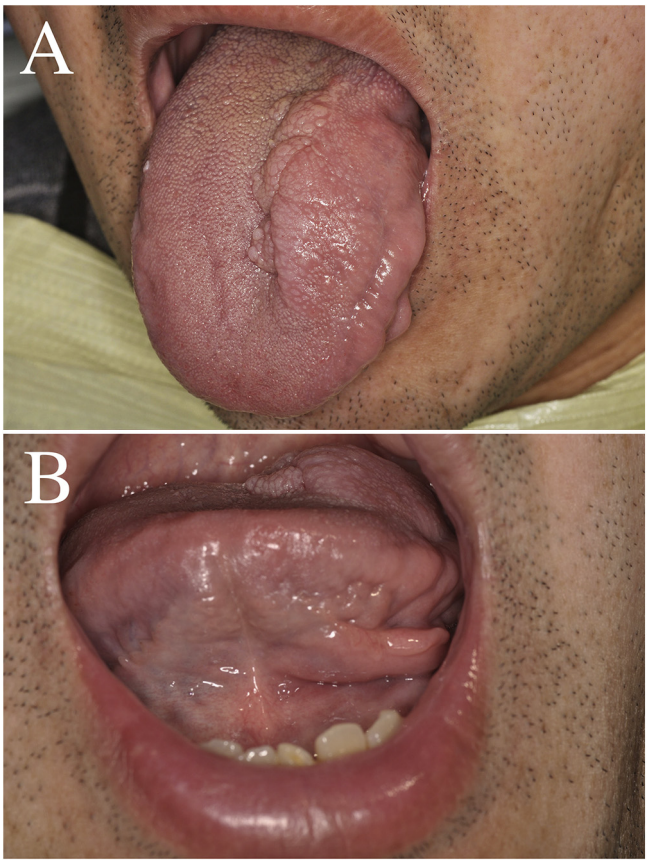

Fig. 1. Photograph of the oral examination. A mass with an irregular margin was found on the left side of the tongue. (A) The dorsal surface, (B) The ventral surface of the tongue.

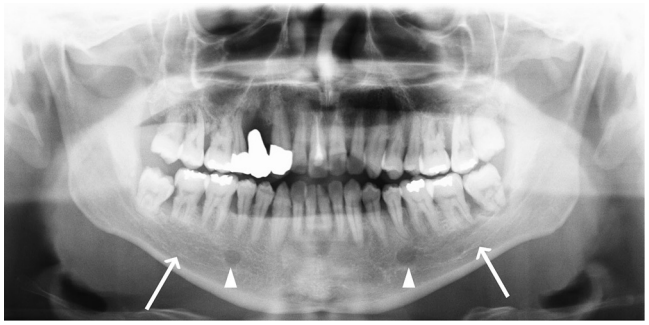

Fig. 2. Radiograph of orthopantomography. Enlargement of the mandibular canals (arrows) and the mental foramen (arrowheads) was found on both sides.

or bleeding. The function of the tongue was well preserved. There was no nodule or specks in other orofacial regions. X-ray examination revealed enlargement of the mandibular canals and mental foramens on both sides but other abnormalities were not found (Fig. 2). A biopsy of the mass was then performed. The histopathological findings showed that there was no capsule around the tumor and that the tumor was composed of a lot of small spindle cells dispersed in wavy delicate collagenous fibers (Fig. 3). There was no atypia or mitoses of the cells. Immunohistochemical examination indicated that the tumor cells were positive for the S-100 protein (Fig. 4). The pathological diagnosis of the biopsy specimen was neurofibroma (dermal neurofibroma). The patient was subjected to regular examination and there was no symptom of rapid increase or malignant transformation of the mass over the one year follow-up period.

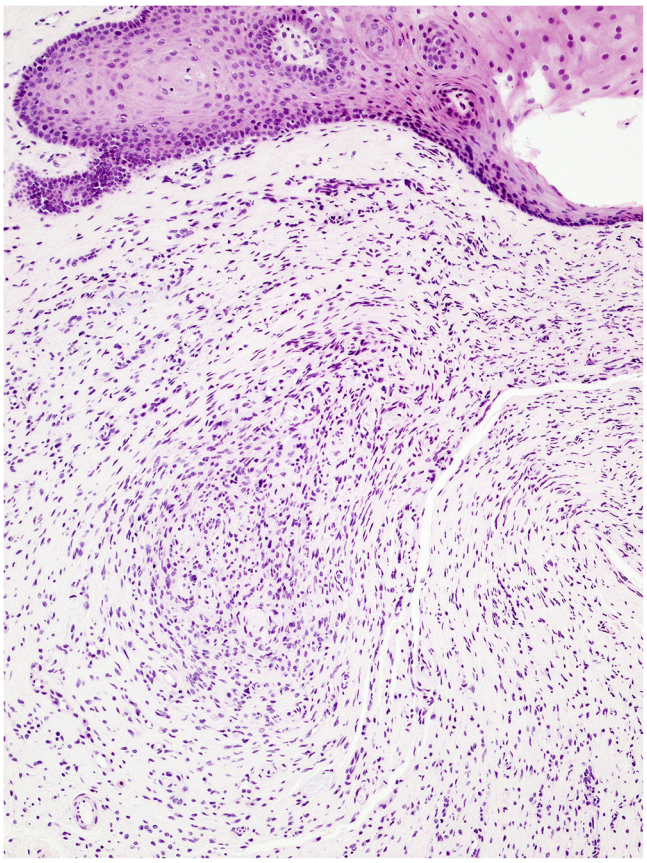

Fig. 3. Photomicrographs of H-E staining of the biopsy of the tumor mass. The tumor is composed of a lot of small spindle cells dispersed in wavy delicate collagenous fibers.

3. Discussion

The criteria for definitive diagnosis of NF1 (NIH consensus criteria for diagnosis of NF1) are shown in Table 1 [12,13]. Two or more clinical features are required for positive diagnosis. At birth, there may be only one symptom of several café-au-lait spots and subsequently, skin tumors of various sizes and in different parts of the body occur until puberty. In addition NF1 is commonly associated with other disorders such as glioma of the optic pathway, glioblastoma, malignant peripheral nerve sheath tumor, gastrointestinal stromal tumor, breast cancer, leukemia, phaeochromocytoma, duodenal carcinoid tumor, rhabdomyosarcomas, scoliosis, and neurological or cognitive impairment [13].

Oral soft tissues manifestations of NF 1 vary from 72% to 92% of NF1 s [4,5–8]. The oral masses are usually one to multiple nodules of various sizes and/or a relatively large mass of neurofibromas. The masses are usually asymptomatic and non-tender [4,7]. The masses of neurofibromas occur at all of the oral sites including the tongue, which is the most common site, buccal mucosa, the alveolar ridge, lips, palate, gingiva, nasopharynx, paranasalsinuses, larynx, floor of the mouth and salivary gland [7,14]. Discomfort is a sign of a tongue lesion [4,7]. Melanin pigmentation of the oral cavity is rare [15]. Histopathologically neurofibroma is often divided into two main types: common dermal neurofibroma and plexiform neurofibroma (PN) [16]. The occurrence of PN in the oral cavity is rare [17].

Of NF1 patients, 25% to 58% show radiological abnormalities of the jaw bones including impacted, displaced, supernumerary, or missing teeth, periapical cemental dysplasia, enlargement of the mandibular canal and mandibular foramen, increased size of the coronoid notch, lateral bowing of the mandibular ramus, inferior

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