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Case report

Widespread deformation of the tongue in a neurofibromatosis type 1 patient: A case report

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ABSTRACT

Neurofibromatosis type 1 (NF1) is an inherited, autosomal-dominant disorder, also known as von Recklinghausen's disease. This pathology was first described from a series of patients displaying a combination of nervous system tumors and findings such as café-au-lait spots, eye lesions, and bone lesions. Oral mucosal tumors are common in association with NF1, but are relatively rare on the lateral border of the tongue. We describe the case of a 17-year-old boy who presented to the Department of Oral and Maxillofacial Surgery at Tokyo Women's Medical University Hospital with the chief complaint of swelling and pain involving the right lateral border of the tongue. Medical history included NF1 and developmental disorder. Several café-au-lait spots were observed on the back, but cutaneous neurofibromas were not apparent elsewhere on the body. Examination of the oral cavity revealed a lobulated lesion involving the right anterior two-thirds of the tongue. Excisional biopsy was performed for an exophytic portion of the right lingual edge of the lesion. Histopathological analysis of the tongue lesion confirmed the diagnosis of neurofibroma. Because the tongue lesion was widespread on the right side and the patient was 17 years old, we performed conservative treatment comprising surgical resection of only the dysfunctional, painful part of the lesion. Three years have elapsed with no sign of exacerbation.

1. Introduction

Neurofibromatosis type 1 (NF1) is an inherited, autosomal-dominant disorder, also known as von Recklinghausen's disease. Neurofibromatosis (NF) was described from a series of patients with a combination of cutaneous lesions and peripheral and central nervous system tumors in the late 1800s [1], and subsequently distinguished into type 1 and type 2 in 1981 [2]. NF1 and NF2 are very different diseases, both in terms of clinical features and the location of the responsible genes. NF1 predominantly affects the skin, bone, and nervous system, but complications are widespread, unpredictable and variable, even within families with the same germline NF1 mutation [3–8]. Oral mucosal tumors occasionally develop in association with NF1 [9,10]. Various manifestations on the tongue have been documented in NF1 patients [10–14], but most have been localized to small parts of the tongue. We present an uncommon case of NF1 with widespread deformation of the entire right border of the anterior two-thirds of the tongue in an adolescent patient in which stable improvement was achieved using conservative treatment.

2. Case report

A 17-year-old boy presented to the Department of Oral and Maxillofacial Surgery at Tokyo Women's Medical University Hospital with chief complaints of swelling and pain involving the right lateral border of the tongue. Medical history included NF1 and developmental disorder. Several café-au-lait spots were observed on the back, but cutaneous neurofibromas were not recognized anywhere on the rest of the body (Fig. 1). The patient had no family history of NF. Examination of the oral cavity revealed a lobulated lesion measuring 37 × 25 mm in greatest dimensions, involving the right side of the tongue (Fig. 2). This lesion had a smooth surface and pinkish-red coloration. On palpation, the lesion was confirmed as soft tissue with mild tenderness. Magnetic resonance imaging (MRI) revealed a hypervascular mass on the right side of the tongue consistent with lymphangioma or hemangioma (Fig. 3).

Excisional biopsy was performed on an exophytic portion of the right lingual edge of the lesion. Histopathological examination

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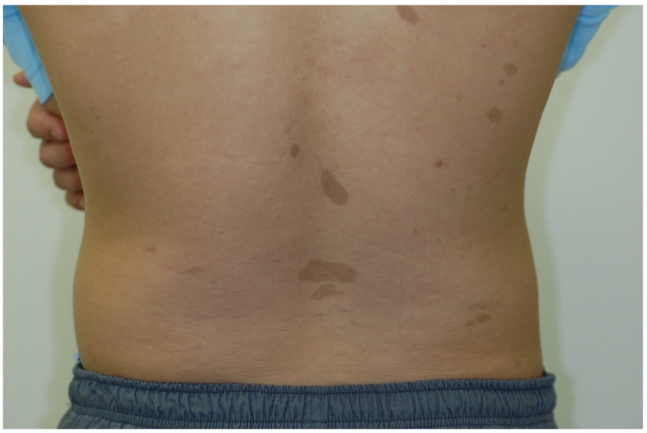

Fig. 1. Multiple café-au-lait spots on the back.

exhibited connective tissue that was rich in spindle cells and lined by stratified squamous epithelium (Fig. 4A). Concentric circular structures resembling nerve endings were scattered in the connective tissue (Fig. 4B, C). Immunohistochemical analysis for S-100 protein was positive in central cells of the structures resembling nerve endings and some spindle cells (Fig. 4D). Histopathological analysis of the tongue lesion confirmed the diagnosis of neurofibroma.

After excisional biopsy, the patient was followed up for 6 months without any events (Fig. 5). The excisional biopsy was restricted to the exophytic portion of the tongue lesion, but 3 years has elapsed with no sign of exacerbation or progression.

3. Discussion

NF1 is characterized by findings such as pigmented nevus, neurofibromas, certain bony abnormalities, and eye lesions. NF1 is one of the most common autosomal dominant inherited diseases. The birth incidence of NF1 is 1 in 2500–3000, with a minimum prevalence of 1 in 4000–5000 [1,3,4]. No significant differences in sex or ethnicity have been identified in terms of the NF1 morbidity rate. In general, NF1 is a hereditary disease, but more than half of cases are sporadic by nascent mutation of the NF1 gene. [15]. In the present case, the parents were negative for NF1 and the patient had no siblings. We therefore speculate that NF1 in this patient represented sporadic disease caused by a new mutation.

Diagnosis of NF1 was based on clinical criteria developed by the National Consensus Conference in 1988, and later reaffirmed by another group of experts [6–8]. NF1 is defined by principal cutaneous features (caféi-au-lait spot and cutaneous and subcutaneous neurofibromas) in addition to central nervous system tumors, freckling, Lisch nodules, and certain bony abnormalities (Table 1). NF1 usually presents during childhood with the development of cutaneous manifestations [1]. In the present case, more than six café-au-lait spots over 15 mm diameter were observed on the back (Fig. 1). In addition to the neurofibroma on the right side of the tongue, a similar lesion that seems to be a neurofibroma was also recognized on the right side of the floor of mouth (Fig. 6A, B). The patient was diagnosed NF1 based on these clinical criteria.

Neurofibromas are benign tumors that normally develop on the skin, and are discernible as focal cutaneous, subcutaneous, or spinal

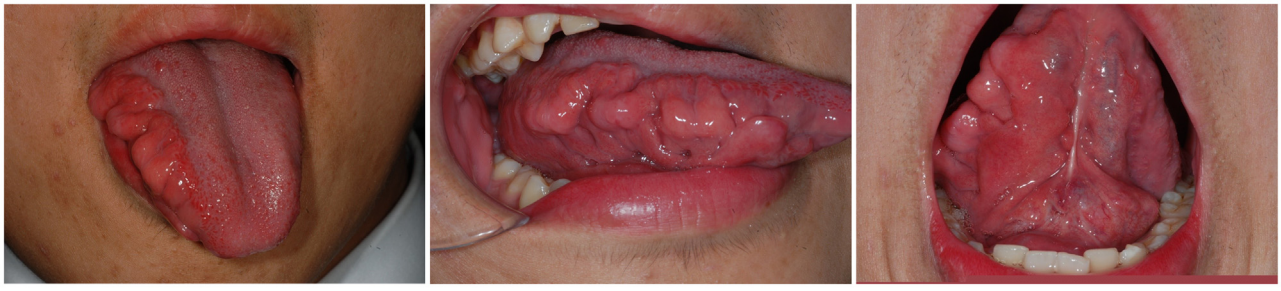

Fig. 2. Intraoral findings during the first inspection. The lobulated lesion involves the right half of the tongue.

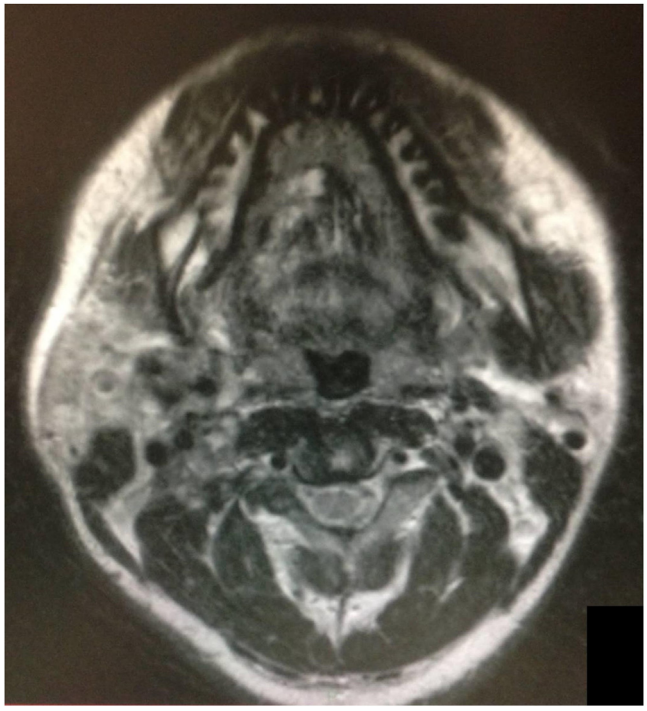

Fig. 3. Preoperative non-contrast T2-weighted magnetic resonance imaging. Mild signal hyperintensity is evident on the right side of the tongue, and small cystic lesions are apparent in the right parotid gland.

Table 1
Diagnostic criteria for neurofibromatosis type 1.

1. Six or more café-au-lait macules with diameter
≥5 mm in pre-pubertal individuals
≥15 mm in postpubertal individuals
2. Two or more neurofibromas of any type, or one or more plexiform neurofibromas
3. Axillary or inguinal freckling
4. Optic glioma
5. Two or more Lisch nodules of the iris
6. A distinct osseous lesion
Dysplasia of the sphenoid bone
Dysplasia of the thinning of long bone cortex
7. A first-degree relative with NF1

Diagnostic criteria for NF1 are based on clinical findings. Subjects must fulfill at least 2 of the 7 features listed [8].

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