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Case Report

A case of Kabuki syndrome with a large palatal fistula and malocclusion

Shuji Yoshida*, Shion Saito, Isao Kamiyama, Aki Ito, Jun-ichi Tanaka

Department of Stomatology, Tokyo Metropolitan Ohtsuka Hospital, 2-8-1 Minami-Ohtsuka Toshima-ku, Tokyo 170-8476, Japan

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ABSTRACT

Kabuki syndrome (KS) is a multiple congenital anomaly syndrome characterized by distinctive facial appearances, skeletal anomalies, abnormal dermatoglyphic patterns, mental retardation, and delayed growth. Patients with KS may show cleft palate and malocclusion that require treatment. However, active treatment procedures such as orthognathic surgery often cannot be performed because of mental retardation. Herein, we present a case involving a female KS patient who was 16 years old at the time of her first visit, had a cleft palate at birth, and underwent a palatoplasty at the age of 1 year and 6 months and pharyngeal flap surgery at the age of 10 years at another hospital. At her first consultation, she had a large palatal fistula, marked maxillary retrusion, mandibular protrusion and anterior open bite. When she was 16 years old, a tongue flap was used to close the large palatal fistula in 2007. In 2011, at the age of 20, she underwent a maxillomandibular orthognathic surgery involving a Le Fort I osteotomy and bilateral sagittal split ramus osteotomy. Both surgical procedures were followed by a favorable postoperative course, with no problems during her inpatient care. As this patient had mental retardation, perioperative management was expected to be difficult. However, our efforts to communicate frequently with the patient during hospitalization led to improvements in her motivation and activity level. The findings obtained in this case suggest that active surgical procedures can be performed on KS patients with mental retardation as long as a careful treatment evaluation demonstrates communication improvements.

1. Introduction

Kabuki syndrome (KS) is a multiple congenital anomaly syndrome that was first reported by Niikawa et al. and Kuroki et al. in 1981 [1,2]. A previous study showed that the incidence of KS in Japan is 1 out of 32,000 persons and that KS has a dominant inheritance pattern [2]. Currently, no consensus exists regarding the cause of KS, despite extensive genetic research. KS is characterized by distinctive facial appearances, skeletal anomalies, abnormal dermatoglyphic patterns, and mental retardation. Some of the major characteristics include arched eyebrows, eversion of the lower eyelids, and long palpebral fissures. The term "Kabuki syndrome" is derived from these facial features. Congenital anomalies in the oral cavity, such as tooth anomalies [3,4] or motor dysfunction of the soft palate [5], are occasionally observed. Cleft palate has been reported to be present in 41% of the cases [6]. Unfortunately, mental retardation makes active treatment difficult for KS patients, and few case reports exist regarding treatment methods, even in the presence of masticatory dysfunction associated with jaw deformities. Herein, we present the findings obtained from a patient, with KS and mild mental retardation, who was admitted to our hospital for improvement of her masticatory function. The treatment course involved closure of a large palatal fistula by using a tongue flap and subsequent orthognathic surgery.

2. Case report

2.1. Clinical history

A 16-year-old female patient was admitted to our hospital after undergoing the following procedures at a different hospital: a

☆ Asian AOMS: 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.

* Corresponding author at: Department of Stomatology, Tokyo Metropolitan Ohtsuka Hospital, 2-8-1 Minami-Ohtsuka, Toshima-ku, Tokyo 170-0005, Japan.
Tel.: +81 3 3941 3211; fax: +81 3 3941 9557.
E-mail addresses: yoshidashuji@tdc.ac.jp (S. Yoshida), sion1978410@yahoo.co.jp (S. Saito), isao_kamiyama@tmhp.jp (I. Kamiyama), itoa-o@ohtsuka-hospital.toshima.tokyo.jp (A. Ito), jtanaka-o@ohtsuka-hospital.toshima.tokyo.jp (J.-i. Tanaka).

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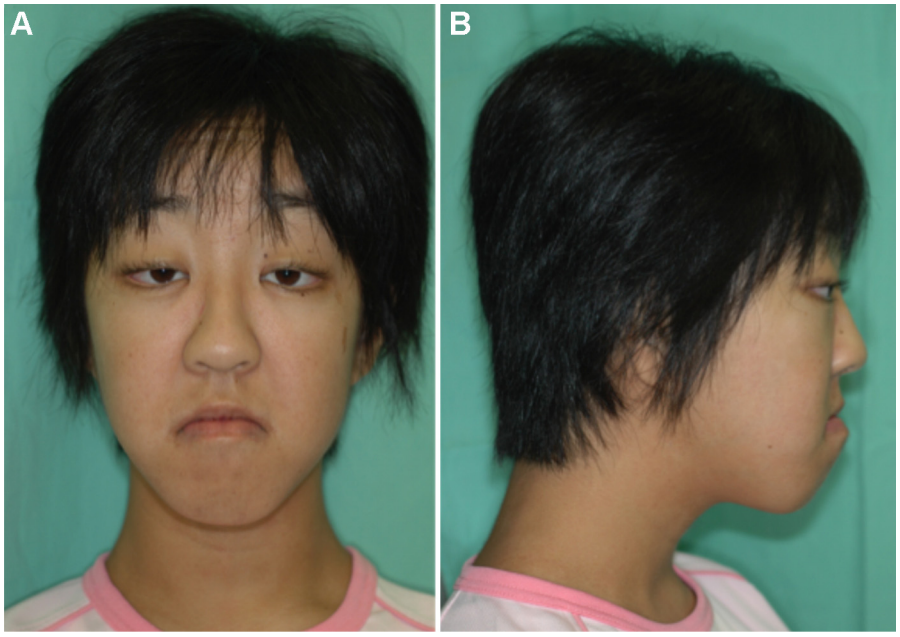

Fig. 1. Facial appearance at the first visit. The facial findings were characteristic of a patient with KS, such as arched eyebrows, eversion of the lower eyelids, and long palpebral fissures.

palatoplasty at the age of 1 year and 6 months, surgery for exotropia at the age of 9 years, and a pharyngeal flap surgery at the age of 10 years. She had no notable family history. The patient had short stature and hirsutism but did not have any apparent abnormal dermatoglyphic patterns or organ impairments. As for facial findings (Fig. 1), we observed findings characteristic of KS, such as arched eyebrows, eversion of the lower eyelids, and long palpebral fissures. In addition, a midfacial recession and lower lip protrusion were present. In addition, we obtained the consent from the patient's parents in showing identifiable personal information to show the characteristic facial appearances.

2.2. Oral findings

The patient had the jaw deformity (maxillary retrusion, mandibular protrusion, and anterior open bite) accompanied by an open bite (Fig. 2). At the center of the hard palate, a large oronasal fistula was observed with an associated rhinolalia aperta and leakage of masticated food into the nasal cavity. Preoperative speech showed instability dysarthria associated with mental retardation and mild rhinolalia aperta due to palatal fistula.

2.3. Assessment of the patient's communication and learning abilities

In addition to the above-mentioned findings, mild mental retardation was present. Furthermore, upon general consultation, her communication and learning abilities demonstrated a level equivalent to that of the upper grades of primary school. Her intellectual level (IQ) score was 50–70, such that moderate social support was required.

2.4. Closure of the palatal fistula by using a tongue flap

In order to prevent the entry of food residue into the nasal cavity and for the treatment of rhinolalia aperta, we carried out closure of the palatal fistula (diameter, 15 mm) with the tongue flap. With the use of a local flap, closure of the large palatal fistula was assessed to be difficult; therefore, a tongue flap was used (Fig. 3).

The surgery was performed under general anesthesia. Nasal intubation was readily performed since the pharyngeal flap that was previously attached had undergone atrophy. The nasal floor was reconstructed by everting the mucosa around the fistula toward the nasal cavity side, and an anteriorly based tongue flap

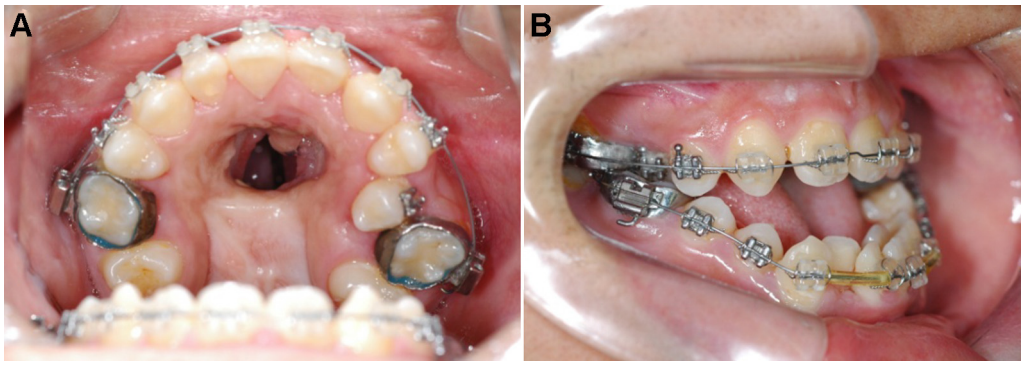

Fig. 2. Oral findings at the first visit. An anterior crossbite with an open bite and a large oronasal fistula were observed.