Occlusal guidance of two Kabuki make-up syndrome patients: case reports

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Kabuki make-up syndrome (KMS) has been reported since 1981 by Niikawa et al. Complications of KMS were moderate mental retardation, skeletal and dermatoglyphic abnormalities. A 7 year-old boy, who had severe permanent tooth deficiency, anterior open bite, tongue thrust and mild mental retardation, was referred to our clinic. Oral characters of another patient were an anterior open bite, transposition of maxillary central and lateral incisor. Orthodontic treatment in two patients is reported and suggested future treatment plans in these patients is given.
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INTRODUCTION

The features of Kabuki make-up syndrome (KMS) are: mental retardation, characteristic facial dysmorphic features, skeletal abnormalities, dermatoglyphic abnormalities, mild to moderate deficiency, and postnatal physiological growth retardation.[1,2] Although the absence of any proven familial case suggests the likelihood of fresh dominant mutation or chromosomal defect in several cases, the etiology is obscured.[3-5]

Oral anomalies are common in KMS (over 60%) and include abnormal dentition, widely spaced dentition, cleft palate or lip, high palate, hypodontia, conical incisors, screwdriver-shaped incisors and ectopic upper first molars.[3,6-10] Although these dental findings have been reported in many patients with the syndrome, Mhannni[10] reported dental findings of 8 patients, the tooth size, dental arch form, and the maxillomandibular relationship have studied by Matsune.[11]

We experienced two KMS patients in our clinic, 7 year-old boy (patient 1) had eight missing permanent teeth and early loss of primary incisors. In the 11year-old boy (patient 2), he had one missing permanent tooth, anterior crowding in maxillary with transposed left lateral incisor and central incisor. These patients had no remarkable general disorder except mental retardation. Oral issues were revealed KMS features, the aim of this study was to detail with the oral and dental features including occlusal guidance of Kabuki make-up syndrome.

CASE REPORT 1

A 7-year-old boy (patient 1), who was diagnosed with Kabuki make-up syndrome (KMS), was referred to our clinic due to deficient dentition. The past medical history revealed that he was born after a 36-week gestation, with no complication during pregnancy. The parents were healthy and non-consanguineous issue. An elder sister, aged 16 years, was also healthy, and she has been treated for maxillary protrusion in division of orthodontic dentistry. He was 110.3 cm tall and weighed 18.4 Kg, at birth the weight was 2774g and height was 49cm. Pediatric examination revealed slight mental retardation. Postnatal growth retardation was not present.

The most striking manifestation was his peculiar face, characterized by long palpebral fissures with ectropion of the lateral third of the lower eyelids, arched eyebrows with sparseness of the lateral half, a

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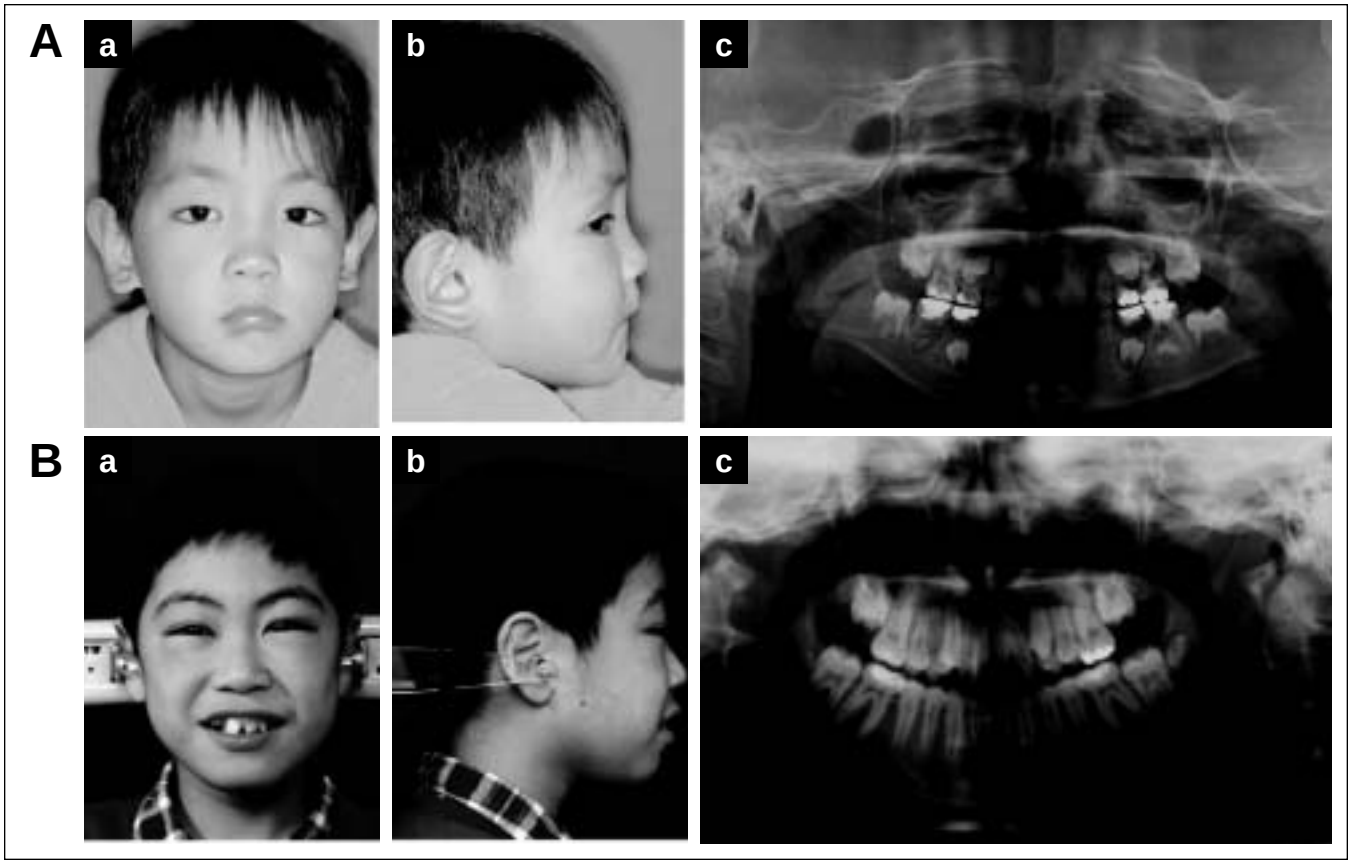

Figure 1. Feature of KMS in patient. (Aa,b) Facial appearance of a patient 1. (Ac) Panoramic radiograph shows severe hypodontia. (Ba,b) Facial appearance of patient 2. (Bc)Panoramic radiograph shows absence of right lateral incisor.

bulbous nose with depressed nasal tip, and prominent ears, all features consistent with the so-called Kabuki make-up syndrome (Table 1, Figure 1a,b). Laboratory data of blood examination and karyotype indicated no abnormalities.

Oral findings consisted of anterior open bite, left posterior cross bite, high arch palate and V-shaped maxillary arch as summarized in Table 1. The anterior open bite was due to a tongue thrusting habit and failure to outgrow the infantile sucking pattern. Panoramic radiographs revealed anomalies of both sides of lower primary central, lateral incisors and canines. The apical roots of these teeth were recognized as slightly resorbed with mobility, which were rounded-shaped (Figure 1Ac). Although these findings may be the results of physiological root resorption or tongue habit, it needs to be noted as a possible oral finding associated with this case. All primary molars are infected and have resorbed apices of the roots. Eight permanent teeth were absent; all canines, both sides lower central and lateral incisors. The teeth sizes that were involved had mean size of normal Japanese teeth. Cephalometric analysis and profilographic data enhancing the angle of convexity that obtained from the lateral cephalograms were presented Table 2 and Figure 2A.[12,13] The data obtained from the lateral cephalograms shows convexity -2.3SD, slightly small SNA, SNB angle indicated middle face hypoplasia. Large L1 to mandibular plane angle revealed –0.95SD means rotated mandible.

The lower first and second primary molars were extracted due to periodontitis. Therefore, a space maintainer was made to gain masticatory function and preserve space. Two months later the lower right primary lateral incisor exfoliated. So we added artificial teeth on the space maintainer (Figure 3Ad). After 2 months, lower left primary canine and lateral incisors showed remarkable mobility (Figure 3B). We trimmed the mobility region of these teeth on the space maintainer. We planned that if the remaining incisor teeth exfoliated, then adaptation or restructure of the appliance will be needed.

CASE REPORT 2

Identical 11-year-old boy (patient 2) diagnosed with KMS was referred to our department of orthodontic division for deficient dentition. The past medical history revealed that he had been born at a 39-week gestation, with neonatal jaundice and the weighted 2870g with a height of 48cm at birth. The parents were healthy and non-consanguineous issue. A pediatric

Table 1. Manifestations of the KMS in two patients and Oral anomalies +, Findings present, -, findings absent.

	patient	
	1	**2**
Gender	Male	Male
Age	6	13
Short stature	+	+
Mental retardation	+	+
Facial findings		
Lower palpebral eversion	+	+
Short nasal septum	+	+
Arched eye brows	+	+
Prominent ears	+	+
Depressed nasal tip	+	+
Malformed ears	+	+
Skeltal abnormalities		
Short 5th finger	+	+
Dermataglyphic abnormalities		
Finger pads	+	+
Other abnormalities		
Cardiovascular anormaly	-	-
Kidney / urinary tract malformation	-	-
Knee joint luxation	-	-
Precocious puberty	-	-
Tumor of cranial bone	-	-
High arched palate	+	+
Cleft palate	-	-
Malocclusion	+	+
Deep bite	-	-
Open bite	+	+
Crowding	-	+
Median diastema	+	+
Mandibular protraction	-	-
Posterior cross-bite	+	-
Wide-spaced dentition	-	-
Malformed teeth	+	+
Conial teeth	-	-
Microdontia	-	-
Small dental arch	+	+
Hypodontia	+	+
Enamel hypoplasia	-	-

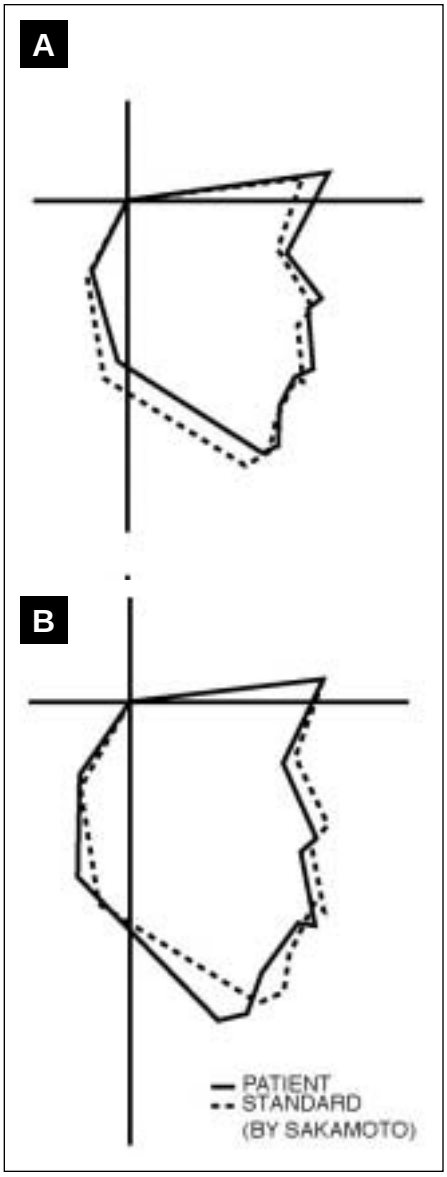

Figure 2. Profilograms of feature of KMS in patients. (A) Patient 1. (B) Patient 2. solid line: patient's profilogram dash line: Standard profilogram.

intelligence quotient (IQ) test revealed mild to moderate mental retardation. He was 129.0 cm tall and weighed 25.0 Kg. Postnatal growth retardation was not present. His anomalies included a short fifth finger due to brachymesophalangy V. Dermatoglyphic abnormalities include pad-like swelling of all fingers. Laboratory data of blood examination and karyotype indicated no abnormalities. The patient exhibited maxillofacial features, which were similar to patient 1, facial photographs showed as Figure 1Ba,b and all features as summarized in Table 1.

Oral findings consisted of Hellman's dental age was IIIc, Angle Class two malocclusion with an anterior open bite, median separation, high arch palate, V-shaped maxillary arch. Upper right incisor and lower right incisor revealed cross bite occlusion (Figure 4A). Panoramic radiographs revealed missing lower left lateral incisor and left upper incisor and lateral incisor was migrated (Figure 1Bc). The teeth sizes were involved mean size of normal Japanese teeth. Cephalometric analysis and profilographic data enhancing the angle of convexity that obtained from the lateral cephalograms were presented Table 2 and Figure 2B.[12,13] Large mandibular plane and GZN means that mandible rotated clockwise. U1 to FH=+2.08SD, U1 to SN =+1.35SD revealed upper incisors were labioversion.

The first treatment plan was to improve the crowding and space maintenance of the maxillary canine. The first molars were banded. A lingual arch was placed with support wire in maxillary to improve cross bite of lateral

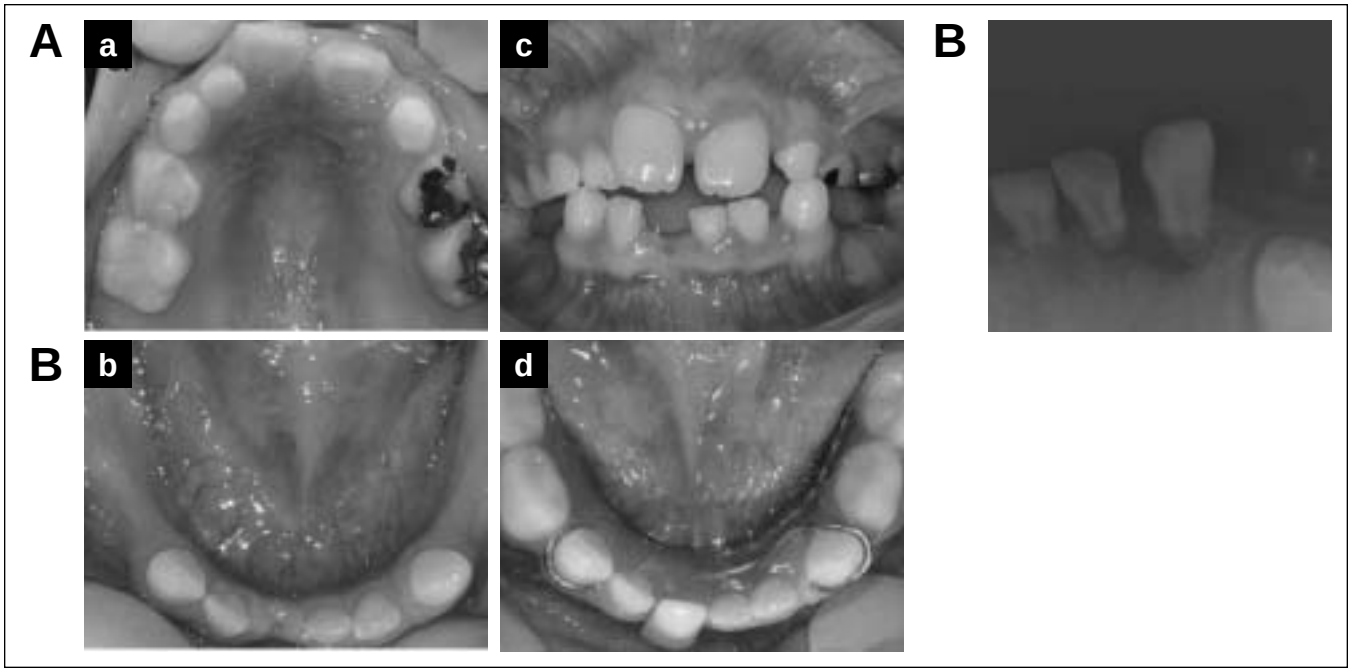

Figure 3. Oral photograph of patient 1. (Aa-c) Intra-oral viewing of after extraction of lower primary molar. (Ad) Inserted the space maintainer. (B) Dental radiograph indicated alveolar bone was resorbed remarkably.

incisors. Therefore, brackets were placed on the maxillary incisors to improve anterior crowding (Figure 4B). After anterior teeth leveling, edgewise brackets were placed on all remaining maxillary teeth except both sides of canine, due to the fact that they were not fully erupted. Open coil spring wire was used to expand the canine space. After finishing the space expansion, brackets were placed on both sides of canines. (Figure 4C).

Table 2. Lateral Cephalometric Analysis in two patients.

patient		1	2
Skeletal	Facial angle	0.00	+0.63
	Convexity	-2.30	+0.60
	A-B Pl.	-0.12	+0.38
	Y axix	-2.14	-0.17
	FH to SN	+1.38	+2.62
	SNA	-2.32	-1.40
	SNB	-2.16	-3.05
	ANB	-1.41	+0.31
	Mandibular pl.	-0.95	+1.08
	Gonial angle	+1.92	+0.90
	GZN	-0.61	+2.02
Denture	U-1 to FH Pl.	+0.05	+2.08
	U-1 to SN Pl.	+0.51	+1.35
	L-1 to mandibular Pl.	+1.64	-0.77
	Interincisal	-0.84	-1.17
	Occlusal Pl.	+0.54	-0.49

Unit: SD by Sakamot, Iizuka et al.

DISCUSSION

The purpose of this report was to identify anomalies of the oral cavity and teeth in these patients with Kabuki make-up syndrome (KMS). The findings confirm the increased occurrence of a cleft palate, as described in the literature.[6,9,14,15] Patient 1 and 2 did not confirmed cleft palate clearly, but both patients had similar characteristics with a narrow maxillary arch and high palate. Remarkable characterization of patient 1 had numerous missing teeth and early loss of primary incisors. Patient 2 had transposed teeth, anterior cross bite, maxillary arch crowding and absence of right lower lateral teeth.

KMS includes multiple dental abnormalities. One of the severe dental problems is hypodontia. Several reports have represented the absence of upper lateral and lower central and lateral incisor teeth in KMS.[16-19] Matsune *et al*.[11] reported that hypodontia appeared in 3 of 5 patients. Mhanni *et al*.[10] have reported that all 8 patients have hypodontia. Hypodontia may arise as a familial condition: a high proportion of affected individuals are members of families with a previous history of the condition.[20] The nature of the inheritance is complex and not well understood.[21] Hypodontia is also a common presenting feature in a number of systemic conditions, such as hypohidrotic ectodermal dysplasia, Down syndrome, and chondroectodermal dysplasia. In KMS Kuroki *et al*.[2] reported an autosomal dominant disorder in which every patient represents a fresh mutation. Hypodontia is defined as the develop-

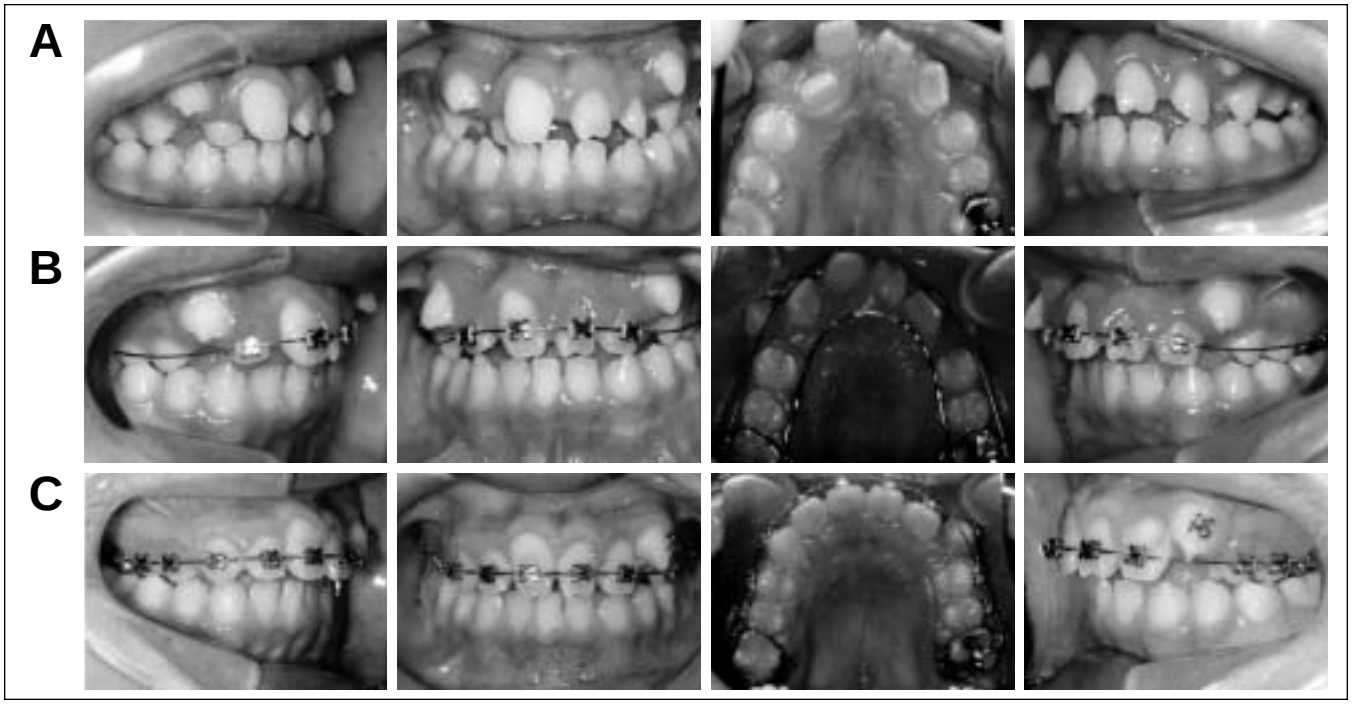

Figure 4. Steps of orthodontic treatment were shown in patient 2. (A) Initial oral findings (B) Partially fixed appliance in maxillary arch. (C) Fixed multi brackets in maxillary arch.

mental absence of one tooth or more. The absence of all teeth (anodontia) is rare. Hypodontia may present with varying degrees of severity, and severe hypodontia has been defined as the absence of six teeth or more, excluding third molars.[20,22,23] The lack of teeth is often associated with a developmental failure of the alveolar bone, resulting in an apparent atrophy of the ridge and lack of posterior support. The age of the patients is a most important consideration during any treatment plan.

In patient 2, there was no inheritance problem in both families. Case 1 belonged to severe hypodontia, were fresh mutation was caused by absence of eight permanent teeth: both sides of mandibular centrals, laterals, canines and maxillary canines.

The age of the patient is a most important consideration during treatment planning. Although, case 1 is not suitable for treatment by orthodontic methods, we suggested that pre-restorative orthodontic intervention would be needed to move the teeth to a favorable position. All lower anterior primary teeth roots have begun to resorp and the lower right lateral primary teeth have already fallen out. We considered that in this case a positional retainer would be inserted in the patient, after extrusion of posterior primary teeth.

Short root anomaly is, in itself, an unusual finding. It has been reported in many dentin and pulp dysplasias; indeed dentin dysplasia is often referred to as 'rootless teeth'. There have been case reports of short stature and short roots associated with bird-headed dwarfism of Seckel[24]and inpatients with Rothmund-Thompson syndrome[25] as well as sporadic cases with microcephaly[26] and learning difficulties. Our case was going to deciduous term, nevertheless, the patient had genetic back ground, congenitally missing teeth and short stature. We suggested that these issues included short root anomalies. In case 1, we are planning for this problem, lower anterior missing teeth lesion will recover with prosthodontic treatment. For the missing maxillary canine, one suggestion included that restoration with an implant retained prosthesis. The most common treatment in the case of missing teeth in children is dentures as used in the present case. Recently, implant-bone total telescopic denture have described as a possible treatment. But high costs, difficulties in placement and high failure rate make use questionable. The following treatment goals were established after discussion with the patient and his family members.

Patient 2 had tooth transposition, an anomaly involving the positional interchange of two teeth. There are numerous reports, which described transposition of the maxillary canine and the first premolar.[27,28] If transpositions are detected early enough, interruptive treatment may be possible. Movement to the normal positions in the arch is a possible alternative, but this may prolong orthodontic treatment, with compromised results because of difficulties in root movement.[29] In this case, the maxillary central incisor and lateral incisor transposed position these teeth were already erupted. It is high risk for the patient, the prolong treat-

ment time and difficult to change the teeth position. We planned orthodontic treatment in that position.

The behavior management (tell-show-do) is important for the success of the treatment concerning the compliance and the prosthetic outcome. In KMS cases, for esthetic, phonetic treatment to rehabilitate the tongue habit and the vertical occlusion, especially for a better mastication function and facial appearance, is of great importance. In most KMS cases, it is difficult to treat and communicate with the patient due to the mental retardation, so little information has been reported with dental treatment. Our patient was not cooperative for dental treatment the first time. We must establish good cooperation for us to perform dental care. We made an effort to get used to the environment of our clinic several times before we attempted to get an oral impression or take intraoral photographs. Good communication was achieved slowly. Patient 1, most of the alveolar bone surrounding the premolar had been resorbed due to periodontitis, so extraction was done.

In the future, hypodontia problems will occur in permanent dental arch. Management of patients with hypodontia has improved considerably; treatment planning should involve careful consideration of the behavior, oral hygiene, socioeconomic background, and ability to follow maintenance instructions. Finally, in this case we suggested how to deal with dental problems in KMS patients. The dental anomalies we presented and guide for occlusion of our results may be helpful in establishing the diagnosis of KMS patients.

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