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CLINICAL SECTION

Kabuki syndrome: a case report

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This article reports the case of an 8-year-old female with Kabuki syndrome and the oral/dental implications of this syndrome, namely hypodontia with interdental spacing, abnormal tooth morphology, malocclusion and a defect in the anterior midline of the palate. The oral findings will aid the clinician in diagnosing this syndrome, which was once thought to be seen exclusively in the Japanese population.

Key words: Kabuki syndrome, hypodontia, cleft lip, cleft palate, microdontia, micrognathia

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Introduction

Kabuki syndrome (KS) is also known as Kabuki make-up syndrome, as well as Niikawa–Kuroki syndrome. This is a rare disorder, which was initially described in 1981 in Japan.[1,2] It was named due to patients' characteristic facial features, which is said to reminiscent of the make-up of actors performing in the Japanese traditional Kabuki theatre, hence the name Kabuki syndrome. However, the term 'make-up' has been discarded due to concern that it causes parental confusion or offence.

The syndrome describes a person with characteristic facial features with other manifestations, such as skeletal and dermatoglyphic anomalies, learning difficulties and postnatal growth retardation.[1–3]

The facial features typically show long palpebral fissures, eversion of the outer third of the lower eyelids, arched and sparse eyebrows, prominent and misshapen ears, and a depressed nasal tip.[1,2]

The dental aspects of this syndrome include hypodontia, abnormal tooth morphology, enlarged pulp chambers, external root resorption, delayed tooth eruption, and abnormalities of the lip and palate including clefting.[4,5]

The purpose of the paper is to increase the awareness amongst clinicians of Kabuki syndrome together with its oral/dental findings, which was once thought to be confined to the Japanese population. It also illustrates the importance of multidisciplinary team management of the KS patient.

Case study

An 8-year-old Afro-Caribbean female was referred to Community Dental Service, from her General Dental Practitioner for routine dental care. Her medical history revealed that she had Kabuki syndrome. This was diagnosed at Great Ormond Street Hospital, London, when the patient was 3 years old. The Kabuki syndrome accounted for her learning difficulties and scoliosis, the latter currently being corrected with a back brace. In her earlier years she suffered from bronchitis because of an increased susceptibility to respiratory infections. On questioning the mother reported no family history of any affected family members.

On general examination the patient was of short stature with learning difficulties. Extra-oral examination (Figure 1) showed long palpebral fissures, ectropium of the lateral third of the lower eyelids, arching eyebrows with sparse lateral halves, depressed nasal tip, large and prominent ears, micrognathia, mid-facial hypoplasia and prominent finger tip pads.

Intra-oral examination

Soft tissue examination showed a palatal soft tissue anomaly in the anterior midline of the hard palate. It appeared symmetrical about the midline and was developmental in aetiology (see Figure 2). This defect was previously examined at Royal London Hospital where, according to the mother, no treatment was performed and no arrangements were made for any review appointments. There was no evidence of an oro-nasal communication or fistula. There was a high palate and satisfactory oral hygiene.

In the upper arch the teeth present were permanent central incisors, canines, first and second deciduous molars, and first permanent molars.

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Figure 1 Extra-oral view showing features of Kabuki syndrome, i.e. long palpebral fissures, ectropium of lateral third of the lower eyelids, arching eyebrows with sparse lateral halves, depressed nasal tip, micrognathia and mid-facial hypoplasia

In the lower arch all the permanent lower incisors were present except for LR1. Permanent canines, first premolars, second deciduous molars and first permanent molars were also present.

The patient was in the mixed dentition stage. Carious cavities were seen in the mandibular second deciduous molars. The patient exhibited hypodontia with missing maxillary lateral incisors and a mandibular central incisor. There was interdental spacing with 'screw-driver-shaped' maxillary incisors. The maxillary incisors

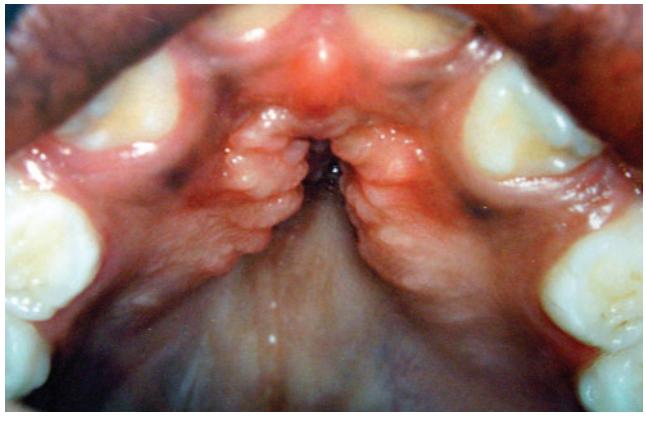

Figure 2 Soft tissue anomaly on the anterior region of the hard palate

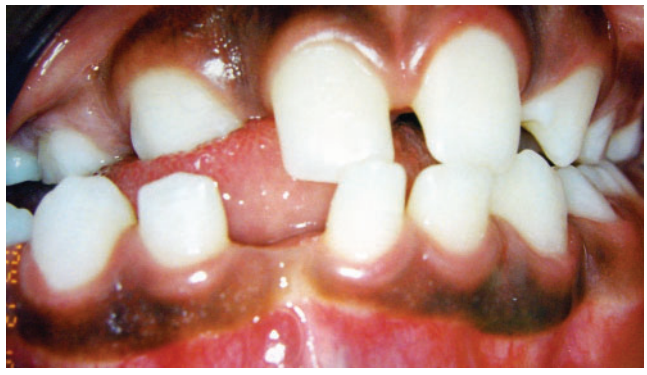

Figure 3 Intra-oral view of anterior teeth showing spacing with 'screwdriver-shaped' maxillary central incisors and Class III incisor relationship

appeared upright. The dental arch and tooth size showed evidence of microdontia (Figure 3).

In occlusion, the incisor relationship was Class III, the overjet and overbite both being reversed. The buccal segments showed a right unilateral posterior crossbite with displacement.

Radiographic findings

An orthopantomogram (Figure 4) and bitewing radiographs were taken. These showed carious mandibular second deciduous molars, and enlarged pulp chambers of the maxillary incisors and first permanent molars. Apart from the maxillary third molars and the teeth previously noted as absent, all other permanent teeth appeared to be present, although unerupted.

Treatment

The patient and parent were given oral hygiene instruction, as well as dietary advice. The first molars were all fissure sealed and the mandibular second

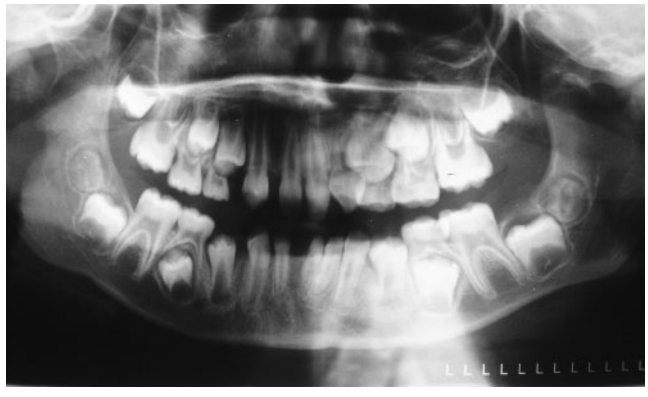

Figure 4 Orthopantomogram showing enlarged pulp chambers of the maxillary incisors and first permanent molars

deciduous molars were restored with radio-opaque glass ionomer cement, which was difficult, due to the patient's anxiety and limited compliance.

Photographs and impressions were taken of the anterior region of the hard palate for baseline records in order to monitor any future changes. The quality of these was, however, compromized due to the patient's learning difficulties. The patient will be later referred for joint orthodontic/restorative clinic with regard to the hypodontia and arch discrepancy.

The patient will be monitored with regard to her moderate risk of caries with preventive advice reinforced. Regular monitoring will enable the patient to be acclimatized to the dental environment to hopefully improve compliance.

Correction of the Class III incisor relationship and right unilateral crossbite with mandibular displacement may require an upper removable appliance. However, this would need compliance from a child with moderate learning difficulties.

The hypodontia requires joint orthodontic/restorative planning to effect either space closure or replacement of missing (incisor) teeth with fixed or removable prosthodontics.

Discussion

Kabuki syndrome is a rare disorder and has an estimated frequency of 1 in 32,000[6], which could mean around 50 cases a year in the UK. The aetiology of the syndrome is unknown and is thought to be possibly due to sporadic mutation with no familial history.[7] Inherited transmission has been reported with some facial resemblance in mothers of KS patients suggesting autosomal dominant inheritance with variable expressivity.[8] This variable expression may account for its under-diagnosis. KS has equal sex predilection and is found in all ethnic groups. Initially thought to be more prevalent in Japan, where it was first reported, an increasing number of KS patients have been recognized in non-Japanese children therefore indicating that this syndrome is not as rare as once previously thought.[6]

There is no prenatal screening, genetic test or consensual diagnostic criteria to confirm this condition. The patients are diagnosed according to the recognizable facial features.

The typical facial features are present from an early age aiding the clinical diagnosis. Clinical recognition of the syndrome in the neonate is difficult, as the phenotype appears to evolve with time. The diagnosis is, on average, made by the age of 2 years.[9] One study described the phenotype becoming less striking in adults.[10]

Cardiovascular abnormalities such as septal defects, co-arctation of the aorta and bicuspid aortic valve have been reported in some studies in association with KS.[7,11] This may have implications in regard to antibiotic prophylaxis.

KS may be an under-diagnosed condition in the cleft lip and palate population.[12] Approximately one-third of KS patients have cleft lip/palate,[3,4,6] and a high arched palate has been seen in almost two-thirds of patients.[3] KS patients may also exhibit ptosis, an expressionless face and a drooping lower lip.[6] Dysarthria may also be a common feature.[12]

Dental abnormalities have been reported in over 60% of patients with KS[4] and include most commonly hypodontia (particularly of central/lateral incisors and premolars)[5,13] associated with interdental spacing and microdontia. KS patients have a tendency of mid-face hypoplasia possibly predisposing them to the development of a small dental arch and malocclusion (associated arch discrepancy).[4] Abnormal tooth morphology[4,13] has been seen in this syndrome with teeth being described as conically-shaped and the upper incisors as 'flat head' screwdriver-shaped, which may aid diagnosis.

Petzold *et al.*[5] described dental radiographic features of enlarged pulp cavities of molars with pulp stones, widened pulp chambers of incisors, external root resorption of incisors, incomplete root formation and root division of a normally single-rooted tooth. These radiographic features together with the clinical features assist in diagnosing the condition.

The prognosis of survival to adulthood is relatively good as KS is not typically associated with severe medical complications.

Conclusions

The dental features may aid the diagnosis of this rare syndrome and form part of the diagnostic criteria. It is important that dental professionals are aware of this syndrome with its variable expressivity as it is not as rare as once thought. They may be able to identify children or adults who maybe mildly affected by this disorder. Dental management is important for the overall care of KS patients, as well as the early involvement of a multidisciplinary team.

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