Dental Manifestations of Kabuki Syndrome: A Case Report

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Introduction

Kabuki Syndrome (KS) is a congenital multi-system disorder caused by mutations in the KMT2D and KDM6A genes. These genes are crucial in regulating chromatin structure, which determines how genes are turned on or off during development. The disorder effects males and females equally and has a prevalence of 1 in 32,000-86,000 births. It is characterized by developmental delays, short stature, skeletal deformities, distinct facial features, and congenital anomalies such as heart defects. Oral manifestations reported in patients with KS include hypodontia, malocclusion, high-arched palate, delayed tooth eruption, small dental arches, and microdontia. In this case, the dental rehabilitation of a patient with Kabuki Syndrome is discussed.

Case Description

A 4–year-old male presented to University Hospitals Rainbow Babies & Children's Hospital dental clinic in March 2023 with a chief complaint of dental pain. Mother reports the patient has been signaling pain by pointing at his posterior teeth. The patient's medical history is significant for Kabuki Syndrome, congenital laryngomalacia, failure to thrive, feeding complications, global developmental delay, non-verbal status, recurrent upper respiratory infections, and wheelchair dependency. The patient is taking Azithromycin for recurrent URI with no reported drug allergies.

A comprehensive exam was completed in the clinic. Extra oral exam reveals a convex facial profile, micrognathia, and long palpebral fissures. Intra oral exam reveals a buccal crossbite, high-arched palate, and full primary dentition except for unerupted tooth J (Fig 1 B). Dental defects including a gray, mottled appearance of enamel were present on all anterior teeth and first molars (Figure 1 A-D). Clinical caries were present on the maxillary first molars and all anterior teeth with the exception of tooth M (Fig. 1 D). Due to the extent of treatment and the patient's inability to cooperate for radiographs, the case was scheduled for comprehensive oral rehabilitation under general anesthesia in July of 2024.

A radiographic examination was completed in the OR and confirms the clinical findings with decay on teeth B, C, D, E, F, G, H, I, O, P, N ,Q, and R, blunted and underdeveloped roots of all maxillary (Figure 2 D-F) and mandibular teeth (Figure 2 G-I), enlarged pulps (Figure 2 D-I), amorphous permanent maxillary centrals (Fig. 2 C), and the absence of multiple mandibular succedaneous teeth including: teeth 23,24, and 26 (Fig. 2 G-I).

Due to extensive decay and poor prognosis, teeth D, E, F, G, I, O, P, and Q were extracted. Sealants were applied to the occlusal surfaces of teeth A, K, L, S, and T as a preventive measure. Teeth B, C, H, N, and R were restored with composite resin. Notably, tooth B received an indirect pulp cap with Theracal to preserve pulpal vitality. The visit concluded with a prophylaxis and application of fluoride varnish to support overall oral health.

The patient will remain under the care of pediatric dentistry for ongoing routine follow-up. At the recall visit in February 2025, a panoramic radiograph was attempted to evaluate the status of the developing dentition; however, the patient was unable to cooperate for the imaging. Clinically, no new carious lesions were observed on the remaining dentition at that time.

Clinical and Radiographic Presentation

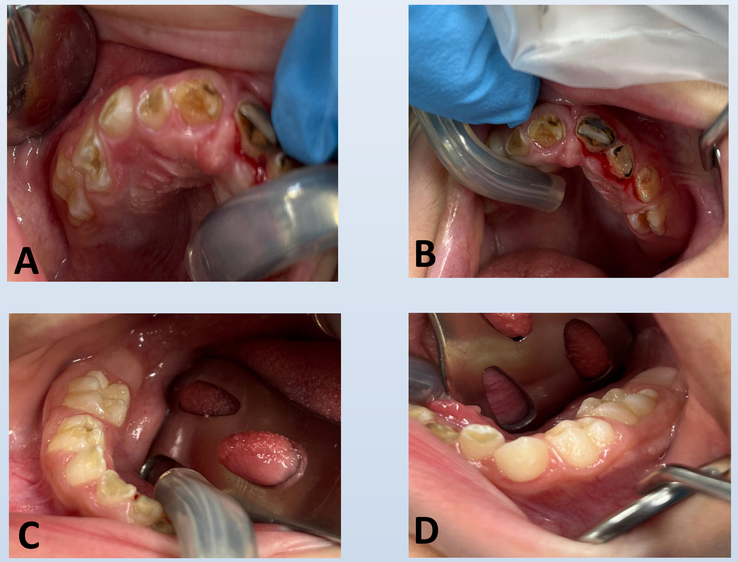

Figure 1: Intraoral clinical photographs of the patient on 7/24/24.

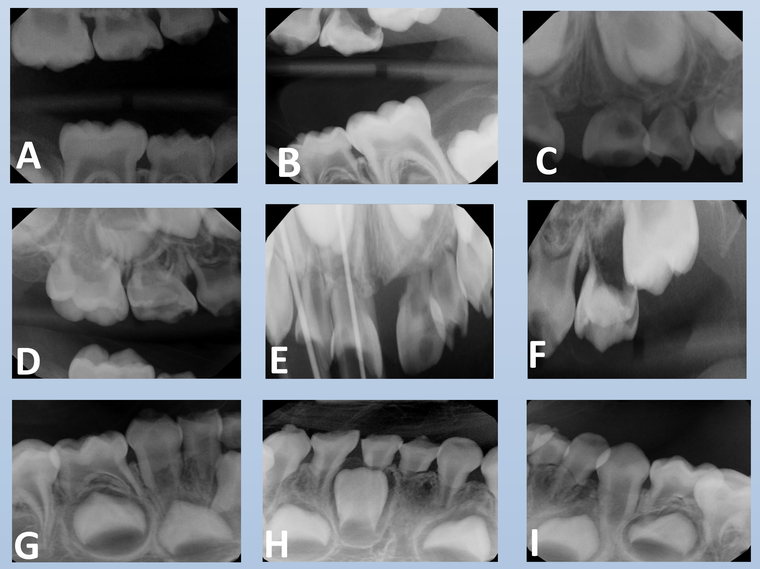

Figure 2: Radiographic presentation, images obtained on 7/24/24. A&B - Intraoral bitewings. C-I intraoral periapical radiographs on posterior and anterior regions.

Dental and Craniofacial Manifestations

The following dental and craniofacial anomalies were noted in our patient which is typical in a patient with Kabuki syndrome:

- Microdontia
- Hypodontia
- Enamel defects
- Delayed eruption
- Malocclusion
- Micrognathia
- High arched palate

Discussion

Abnormalities in the primary and permanent dentition are common in individuals that present with Kabuki Syndrome. The patient in this case experienced significant dental decay due to the enamel defects caused by the syndrome. The choice to restore tooth B with composite over a traditional stainless-steel crown was made to preserve as much tooth structure as possible due to the underdeveloped roots of the primary molar as seen in Figure 2 D. Sealants were placed on the remaining non-carious teeth to strengthen the enamel and attempt to prevent future caries.

Malocclusion is another commonly reported oral manifestation of KS due to the genetic abnormalities that affect facial development. This patient presented with micrognathia and a buccal crossbite. The premature exfoliation of primary teeth may only further exacerbate the malocclusion in the permanent dentition. The patient may also experience malformed or missing permanent teeth with delayed eruption. The absence of permanent teeth is strongly associated with KS, with the lateral incisors, canines, and premolars reported to be most commonly missing. Another characteristic dental abnormality often reported in patients with KS is the "screwdriver-shaped" appearance of the permanent maxillary incisors, which is seen in Figure 2 C.

This patient will likely require interdisciplinary management, including orthodontic and prosthodontic care to guide occlusal development and replace missing teeth. Speech and feeding therapy would also be beneficial to the patient due to difficulties caused by palate abnormalities. This case highlights the importance of recognizing the oral manifestations of Kabuki Syndrome early. Prompt, coordinated care can improve both oral function and quality of life for these patients.

References

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