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Oral Manifestations Associated with Kabuki Syndrome: Moroccan Case Report

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1. Abstract

Kabuki syndrome, also known as "Niikawa-Kuroki Syndrome", is a rare genetic disorder first discovered in Japan. Its frequency is estimated at 1 in 32,000 with an autosomal dominant mode of transmission. This polymalformative syndrome is usually associated with an intellectual deficit and morphological variations specific to the syndrome located in the eyes, nose, and ears. The oral manifestations associated with the syndrome are multiple: cleft palate, hypodontia, microdontia, delayed tooth eruption, diastemas, enamel hypoplasia and taurodontism.We report the case of a 6-year-old male child with Kabuki syndrome who consulted the pedodontics department at the department of pediatric dentistry in Rabat for a mouth cavity repair. The diagnosis was made by the team belonging to the medical genetics department at the National Institute of Hygiene in Rabat. This is the first case of Kabuki reported in Morocco.

2. Introduction

Kabuki syndrome is a rare sporadic congenital anomaly, first described in Japan in 1981 by Niikawa et al. [1] and Kuroki et al. [2]. The patients with the syndrome have a facial appearance similar to that of traditional Japanese actors, hence the name Kabuki.The frequency of Kabuki syndrome is estimated to be 1 in 32,000 [3] [4] with an autosomal dominant mode of transmission, which has been confirmed by the identification of point mutations in the KMT2D (56% to 75% of cases) [5] and KDM6A (5% of cases) [6,7] and located on chromosome Xp11.23 [8].Indeed, Kabuki syndrome presents a strong clinical, radiological and biological heterogeneity. It includes multiple congenital anomalies on which the diagnosis of the syndrome is based, combining syndrome, associating a characteristic facial dysmorphia, growth retardation, skeletal anomalies, persistence of skeletal abnormalities, persistence of fetal-like pads and mild to moderate intellectual deficit. These diagnostic criteria are difficult to determine before the age of 1 year and also after the age of 16 years of age, due to the evolution of morphological variations that become less striking in adult age [3]. Molecular analysis is essential to confirm the diagnosis of the syndrome.

3. Case Presentation

Y.D, Moroccan, 6 years old, male from a second degree consanguineous marriage (Figure 1). On examination, the mother reported that the pregnancy had not been monitored by a physician, no medication had been taken during gestation and that the delivery was completed without complications. No family history was reported.Further to the learning difficulties and the staturo-ponderal delay; the parents brought the child for consultation. The genetic diagnosis of the syndrome was made by the team belonging to the Department of Medical Genetics at the National Institute of Hygiene in Rabat. The general examination (Figure 2) showed that the patient presents a delay in weight and height with a difficult behavior with an autistic character. The patient presents other alterations related to the syndrome including prominent pads (Figure: 3) on the fingers with clinodactyly of the fifth finger. For oral

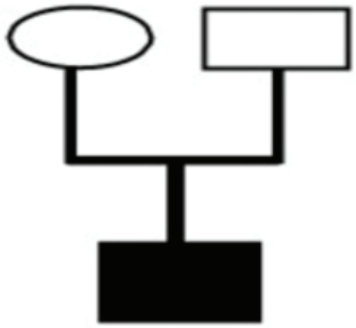

Figure 1: Genealogical diagram consanguinity second degree.

Figure 2: Front view of the patient showing a staturo-ponderal delay.

cavity rehabilitation, the patient consulted the pedodontics-prevention department within the Centerof consultations and dental treatments of Rabat. We found:

3.1. Extra Oral Examination

Frontal extra-oral examination showed an ovoid face with unevenness of the facial stages.The frontal lobe is prominent, the middle and lower layers are diminished. The patient presents a facial asymmetry, long palpebral fissures, ectropion of the eyelids of the left eye, arched and sparse eyebrows, flat nasal tip with large protruding ears, thin upper lip and micrognathia (Figure 4).Profile examination revealed a slightly convex profile with a notion of hypodivergence in vertical direction.

4. Intra-Oral Examination

4.1. The Endo-Buccal Examination Showed

On the bone level: an ogival palate indicating maxillary endognathy, atresia of the anterior third of the palate, and outgrowth on both sides of the median suture with a vestibular exostosis in the incisivo-canine sector. On the dental level: an inverted articulation at the anterior sector and at the right and left lateral sectors.The teeth show microdontia, ameloid hypoplasia and diastemas. In the maxilla, all the teeth are temporary. In the mandible, we note the presence of permanent central incisors and the absence of the left lower lateral incisor.The temporary molars are too dilapidated; consequently, there is a loss of posterior wedging and therefore a collapse of the vertical dimension of occlusion, resulting in of an erased lower level. The occlusal examination showed an inverted articulation with a class 3 canine and a loss of posterior wedging. The first lower permanent molars are erupting (Figure 5).

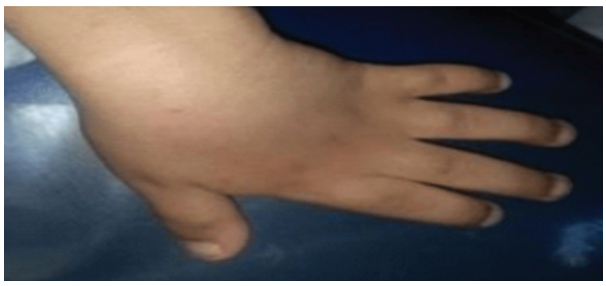

Figure 3: View of a left hand showing prominent finger pads and a short little finger with clinodactyly of the fifth finger.

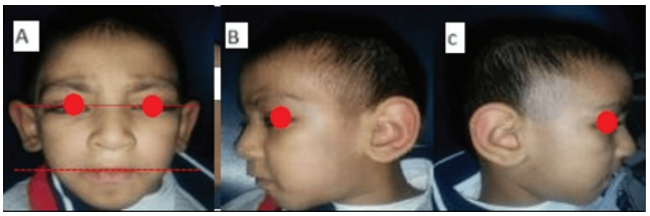

Figure 4: A- Front view showing the distinctive craniofacial features of Kabuki syndrome. B et C - Right and left lateral view of the patient's face showing depressed nasal tip, thin upper lip and protruding ears.

4.2. Radiographic Examination

The panoramic was not taken; the child is claustrophobic and refuses contact with any stranger. The retroalveolar assessment showed significant coronal decay of the temporary teeth and presence of teeth in root state. The temporary lower left lateral incisor is missing and the agenesis of the germ of its successor. In the upper temporary central incisor, there is an apical lesion as well as furcation invasion on the molars. Amelar hypoplasia is apparent on the retroalveolars (Figure 6).

5. Discussion

Niikawa-Kuroki syndrome, more commonly known as kabuki syndrome, is a rare autosomal dominant congenital disease, due to mutations in the KMT2D gene [5] or in the KDM6A gene [6,7]. These patients present a wide and varied clinical picture, craniofacial dysmorphia, postnatal growth retardation, neurosensory disorders such as neurosensory disorders such as hearing loss secondary to chronic otitis media, congenital cardiac malformations, an immune deficiency at the origin of autoimmune diseases and increased susceptibility to infections, as well as mild to moderate intellectual deficits associated with autistic features [8-10].On the oral side, this syndrome is associated with cleft lip and palate, an ogival palate and various dental anomalies (number, shape and structure).The patient reported in this work presents the majority of these anomalies, namely, a staturopunderal delay, an intellectual deficit and a scoliosis. He also presents a characteristic facial dysmorphia, arched eyebrows, elongated palpebral slits, eversion of the lower eyelid, short nasal septum, convex profile, prominent ears, hypoplastic nails and prominent finger pads and brittle hair. No heart problems were diagnosed, yet he had recurrent respiratory infections.

5.1. Various Oral Manifestations Were Described by Several Authors Namely

Tuna et al. [11], Teixeira et al.[7] and Schrander-Stumpel et al. [12] noted an agenesis of the upper lateral incisors or lower central incisors and canines whether temporary or permanent. Our patient has agenesis of the germ of the left lateral incisor.Unlike Rocha et al.[13] who reported the presence of supernumerary teeth in a nine-year-old boy.Silva-Andrade et al.[14], reported enamel hypoplasia, delayed eruption, microdontia and dental fusions in 8 patients aged 3 to 16 years. The teeth of both arches of our patient show microdontia and diastemas, as well as amelar hypoplasia.Mhanni et al [14]. Bescribed other anomalies such as ectopic eruption of the first permanent upper molars as well as enamel hypoplasia and gemination [15]. Dos Santos et al.[16] also found other anomalies such as gemination and dental fusion.Petzold et al.[17], described taurodontism in molars and incisors with pulp calcifications, external root resorption of incisors, incomplete root formation and root divisions of a single-rooted tooth.Abnormalities of the palate are also common [18, 19]. Indeed, a hollow palate is often observed in 58% of patients [7], as well as a short or hypotonic soft palate.Our patient presents an ogival palate indicating a maxillary endognathy, a short or hypotonic soft palate, and an atresia of the anterior third of the palate and an outgrowth on both sides of the median suture.Isolated cleft palate is more common than cleft lip in Kabuki syndrome [5,9].Petzold et al.[18] and Lung et al.[20] noted that KS patients tend to have a facial hypoplasia and thus hypo-development of the maxilla which contributes to micrognathia and favors the development of malocclusion such as the inverted bite.

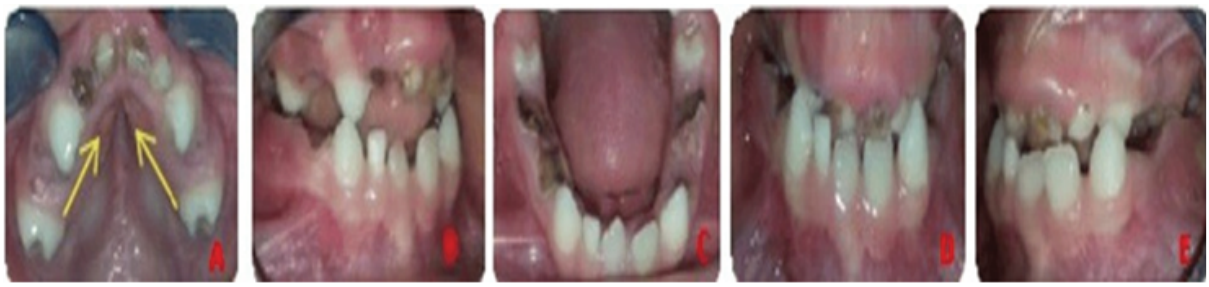

Figure5: intra -oral views:Atresia of the anterior 1/3 of the palate (A). Decayed, conical teeth (B).Absence of the 72 (C).An inverted bite (D). Class III canine and loss of posterior wedge (E).

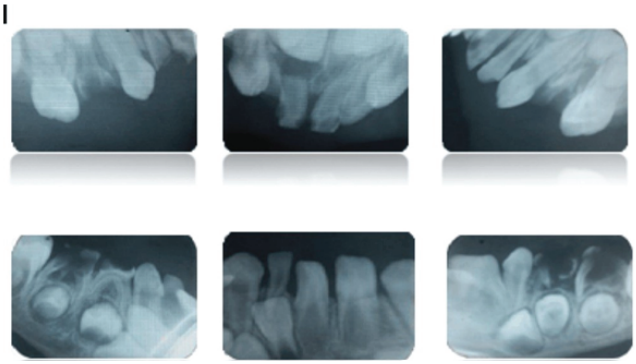

Figure 6: Retro-alveolars confirm the significant decay of the temporary teeth and the amelar hypoplasia.

In our case, the intra-oral examination showed endognathy and a class III with an inverted bite.

The management of this patient is multidisciplinary:

- Medically
- Management by a nutritionist
- Evaluation of immunology in case of recurrent infections
- Annual monitoring of hearing and vision
- A check-up of the oral cavity is recommended as well as a bi-annual follow-up.
- As far as we are concerned, a diagnosis and a global treatment plan have been established in order to:

Respond the initial reason for consultation. Nevertheless, the difficult behavior of our young patient with an autistic character led us to treat him under general anesthesia. Multiple dental extractions as well as primary impressions were performed in order to ensure a complete functional, aesthetic and psychological rehabilitation.

6. Conclusions

Kabuki syndrome is a rare congenital disease that requires early medical and oral care to prevent possible complications because life expectancy depends more on cardiac and immunological complications, thus contributing to the well-being of these patients who already present a heavy clinical and psychological picture.

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