Oral Aspects of 49, XXXXY Syndrome: A Case Report

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ABSTRACT

The 49, XXXXY syndrome is a rare chromosomal disorder. The purpose of this paper was to present the case of a 9-year-old boy with this karyotype. The most remarkable skeletal and dental anomalies were the absence of 9 permanent teeth, taurodontism of the permanent first molars, and the conical shape of permanent maxillary lateral incisors. Third molars were not visualized. Cephalometric analysis revealed a well-positioned maxilla, a slightly protruding mandible, and retroclined mandibular incisors. The surgery and dental rehabilitation are described. Given the specific characteristics of this syndrome, it is important to reassess the patient's skeletal and dental development and promote good dental hygiene practices. (J Dent Child 2007;74:136-9)

KEYWORDS: CHROMOSOME ABNORMALITIES, XXXXY KARYOTYPE, TAURODONTISM

The 49, XXXXY karyotype, first described by Fraccaro and Lindsten[1] in 1960, is a variation of the Klinefelter's syndrome rarely found in the general population. About 100 case reports have been published since its first description in the literature.[2,3] This syndrome shows great phenotypical variation, and predominant characteristics are: dysmorphic facial features, mental retardation, and skeletal anomalies. The most frequent anomalies are radioulnar synostosis and marked hypogenitalism.

Another typical finding is infertility due to impaired spermatogenesis.[4,5] Although the general characteristics of this syndrome have been described in the literature, little is known about its oral aspects of interest to dental professionals.

The purpose of this case report was to describe the general conditions and the skeletal and dental anomalies in a 9-year-old boy with 49, XXXXY chromosomal disorder.

CASE STUDY

A 9-year-old Caucasian with a diagnosis of 49, XXXXY syndrome was seen in the pediatric dental clinic of the Hospital Universitário de Brasília (HUB), Brasilia, Brazil. At presentation, severe decay of all primary molars and caries extending into the pulp of permanent maxillary first molars were observed. Poor dental hygiene was evident from the large amount of plaque and intense cariogenic activity.

The boy's medical history revealed that he was the first child of a young healthy nonconsanguineous couple. The mother's karyotype was normal (46,XX), pregnancy and delivery were normal, and the boy was born at term at 40 weeks. Weight and height at birth were 2,800 g and 47 cm. From an early age, psychomotor delay and low weight gain were observed, and development revealed: hypogenitalism, hypogonadism, discrete gynecomastia, centripetal obesity, myopia, poor pronation of forearms, and other skeletal anomalies. The most remarkable facial anomalies were: hypertelorism, a flattened nasal bridge, and slanting eyes. The patient has a diagnosis of moderate retardation.

Dental evaluation was complemented by a panoramic radiograph that detected the congenital absence of all second premolars, both mandibular first premolars, permanent mandibular central incisors, permanent mandibular left lateral incisors, and third molars. Two cystic lesions were visualized, the smaller in the maxilla surrounding the right primary maxillary canine, and the larger in the mandible, involving primary second and first right molars.

Moreover, taurodontism of the permanent maxillary

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and mandibular first molars was seen, and both permanent maxillary lateral incisors were conical (Figure 1).

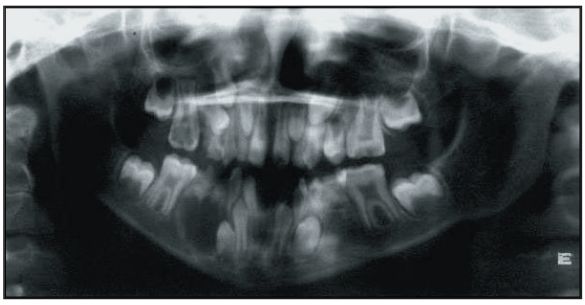

Figure 1. Panoramic radiograph of patient at 9 years and 2 months of age. Severe caries, congenital absence of 9 permanent first molars. Third molars were not visualized.

Initially, restorative treatment was attempted under local anesthesia. Since the patient's behavior was uncooperative, and his psychological conditioning was limited due to the mental deficiency, dental treatment had to be conducted under general anesthesia. Permanent mandibular first molars were restored with resin modified glass ionomer. Some teeth were extracted (all primary teeth and both permanent maxillary first molars) and marsupialization was the technique used for the treatment of the dentigerous cysts.

New panoramic radiograph were obtained 28 days and 5 months after surgery and showed the involution of the cystic lesions. One year after the surgery, impressions for models, panoramic (Figure 2), hand and wrist radiograph, and cephalometric radiograph (Figure 3) were taken. Table 1 shows the cephalometric study of the patient at 10 years and 10 months. Cephalometric analysis revealed a well-positioned maxilla (SNA), mandibular prognathism (SNB), normal anteroposterior relationship (ANB), and well-positioned maxillary incisors (11.NA). Mandibular incisors, however, were retroclined (41.NB).

Table 1. Cephalometric Analysis

	Patient	Norm
Angular measurements	(°)	(°)
Facial angle	94.08	88
SNA	84.23	82
SNB	83.39	80
ANB (SNA - SNB)	0.84	2
SND	80.02	76
11.NA	22.85	22
41.NB	9.57	25
Linear measurements	**(mm)**	**(mm)**
11-NA	3.84	4
41-NB	0.98	4

Improvement in the direction of eruption of the permanent maxillary right canine was observed. Permanent maxillary second molars moved mesially, which favored the closure of spaces generated by tooth losses and avoided inclination of crowns. Oral examination revealed the eruption of the permanent maxillary right lateral incisor in crossbite. Inclined or rotated were: (1) all permanent second molars; (2) maxillary left first premolar; (3) mandibular right first premolar; (4) permanent maxillary right central incisor; (5)

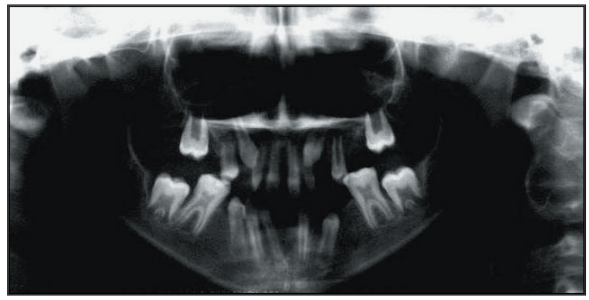

Figure 2. Panoramic radiograph at 10 years and 10 months of age.

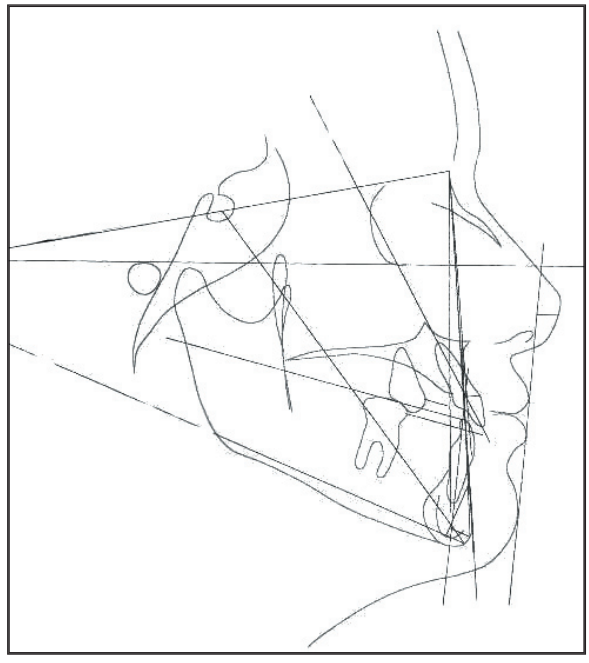

Figure 3. Cephalometric tracing at 10years and 10 months of age.

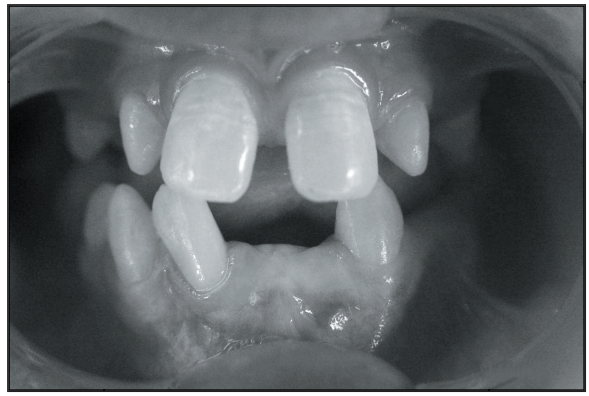

Figure 4. Frontal intraoral photo shows congenitally absent teeth and conical shape of permanent maxillary lateral incisors

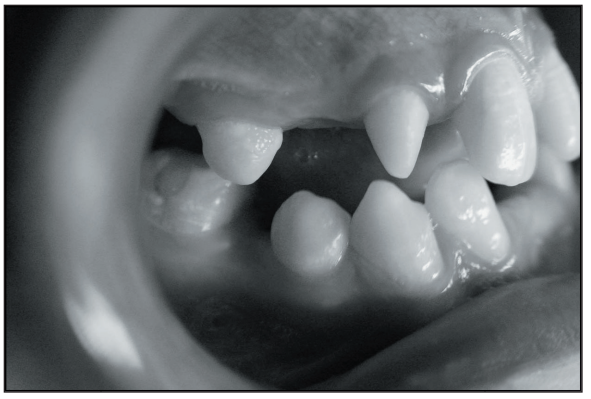

Figure 5. Right lateral occlusion.

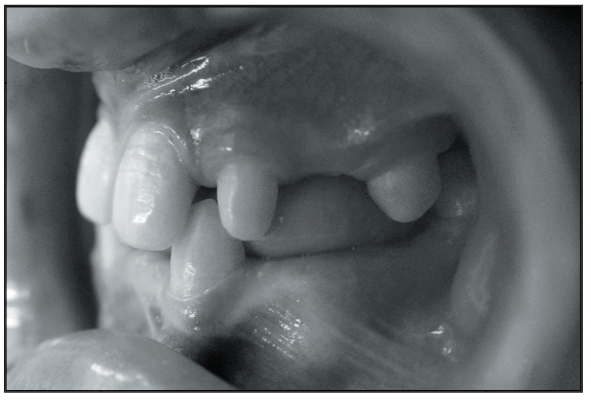

Figure 6. Left lateral occlusion.

permanent mandibular right lateral incisor; and (7) permanent mandibular right canine (Figures 4 through 6).

According to the bone maturation graph,[6] bone age was compatible with chronological age and the patient was 15 to 36 months from peak pubertal growth.

Oral hygiene improved significantly, bacterial plaque was controlled, and caries were not found after the intervention because parents had been instructed on brushing techniques and diet. Moreover, the child was more receptive to oral care.

DISCUSSION

Skeletal and craniofacial anomalies, as well as hypogonadism and mental deficiency associated with the extra X chromosomes, were the traits seen in this study's patient that matched descriptions in the literature [7-12]. Radioulnar synostosis, often associated with patients with 49, XXXXY syndrome, was not diagnosed in this patient.[12]

Taurodontism is a common finding in chromosomal disorders and is seen more frequently in X-chromosomal aneuploidy.[11,13] In this patient, all the permanent first molars were affected by taurodontism, confirm's the findings of other authors.[3,14-16] Taurodontism has also been reported in the primary dentition,[8,17] but could not be confirmed in this case study because of advanced dental decay. The mandibular first premolars, the maxillary first and second premolars, and the mandibular incisors were congenitally absent, a finding in agreement with reports in the literature.[3,17,18]

49, XXXXY syndrome patients usually have a retracted premaxilla and mandibular prognathism[3]. This study's patient, however, had a well-positioned maxilla and mild mandibular prognathism.

Because of the many clinical and radiographic findings of patients with this syndrome, it is important that their dental and skeletal development be monitored. The continuous incentive to oral hygiene practices is of paramount importance for the healthy general and psychosocial development of these patients.

CONCLUSION

A 9-year-old boy with 49, XXXXY syndrome with congenital agenesis of 9 permanent teeth. Taurodontism, the conical shape of lateral maxillary teeth, are common traits in this syndrome is reported.

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