

CASE REPORT

Dental Treatment of 49,XXXXY Syndrome: A Case Report

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ABSTRACT

A 12-year-old male patient with 49,XXXXY syndrome is presented, who was treated under general anesthesia due to mental retardation and uncooperative behavior. The child was treated as an emergency case in two occasions due to severe pain and abscesses. Several teeth were severely decayed and had to be extracted resulting in extensive anodontia as there was no secondary dentition to follow. This case emphasizes the importance of regular dental care, good nutrition and monitoring of dental development in children with 49,XXXXY syndrome.

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INTRODUCTION

The sex chromosome abnormalities (structural or numerical) are the most common chromosomal abnormalities. They are characterized by the presence of supernumerary X or Y chromosome or the absence of an X. More than two X chromosomes rarely are identified and the individuals have 48 or 49 chromosomes.¹ The 49,XXXXY syndrome was first reported in 1960 by Fraccaro et al.² It is a rare sex chromosome abnormality that occurs in a frequency of approximately 1 in 85000 male births.³ Approximately 100 cases of the 49,XXXXY syndrome have been published to date.⁴⁻⁸

One of the features of 49,XXXXY syndrome is the possible absence of permanent teeth. This fact, in combination with the mental retardation which severely impacts the patients' oral hygiene, may result in early and extensive tooth loss. Hence, the dental care providers need to be very vigilant about teaching oral hygiene. Also, there must be excellent cooperation with parents and caregivers so as to prevent severe damages in dentition. A case is presented which highlights the importance of oral and dental care in patients with 49,XXXXY syndrome with a hope to contribute to the limited current literature on this rarity and provide some guidance in the dental management of these patients.

CASE REPORT

A 12-year-old male patient with 49,XXXXY syndrome was firstly admitted at a regional pediatric hospital in 2008 at age 8 years, due to extensive caries which caused

ABBREVIATIONS

IgA = immunoglobulin A
OPG = orthopantomogram

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dental abscess and fever. The morphological features of the syndrome were present including mandibular micrognathia, high palate, broad based nose, scaphoid thorax, hypoplastic fifth finger, and radius-ulnar synostosis bilaterally.¹⁰⁻¹³

Patient's family history was unremarkable for genetic diseases. Relevant medical history included severe mental retardation (IQ <60), speech problems, no apparent heart defects (even though considered common in this syndrome), malabsorption syndrome which was possibly due to celiac disease based on serological and histological examination and a mild form of IgA immunodeficiency.^{5,9}

At this initial visit, the patient was treated as an emergency case under general anesthesia. An orthopantomogram (OPG) was not possible to be obtained preoperatively as the patient was very agitated. We proceeded according to clinical findings. The deciduous teeth with severe caries were extracted. Oral hygiene instructions were provided and follow-up visits were scheduled, but unfortunately the patient failed to comply.

The patient was referred again to the dental department 4 years later (in 2012) at the age of 12 complaining for pain on chewing and reduction in food intake. Clinical examination on admission revealed tooth caries, extensive tooth loss (Fig. 1) and poor oral hygiene. The child was unable to brush correctly and also the caregivers did not assist him with his daily oral hygiene. Tartar and food deposits were present and the gingiva were hemorrhagic. As several teeth were severely decayed and the pain was not responsive to analgesics, it was considered appropriate to schedule an immediate treatment plan under general anesthesia. Due to severe pain the child was uncooperative and an OPG could not be performed. As it was evident that the child was unable to maintain proper oral hygiene, it was decided to extract all the teeth with extensive caries. The procedure was carried out successfully. The postoperative course was uncomplicated and the patient was discharged after 24 hours.



FIGURE 1. Extensive tooth loss is apparent (arrow).

On re-evaluation three weeks later, the patient was cooperative and we managed to obtain an OPG. We identified absence of permanent premolars (Fig. 2). Again poor oral hygiene was obvious (Fig. 3) and the caregivers were provided with instructions and advice for orthodontic adjustment of occlusion to assist the eruption of the remaining permanent teeth.

DISCUSSION

The 49,XXXXY syndrome is often described as "variant of the Klinefelter syndrome". However, the clinical features are not the same as in Klinefelter. People with 49,XXXXY polyploidy often have congenital heart defects,^{5,9} hypogonadism, cryptorchism, radioulnar synostosis, clinodaktyly, and mental retardation.¹⁰⁻¹³

The oral cavity manifestations are taurodontism of the permanent molars, congenital absence of permanent teeth, delayed tooth eruption, prognathism of the mandible, shovel-shaped incisors and enamel defects.¹⁴

When a child with 49,XXXXY polyploidy is in the mixed dentition stage it is difficult for a dental care provider to de-

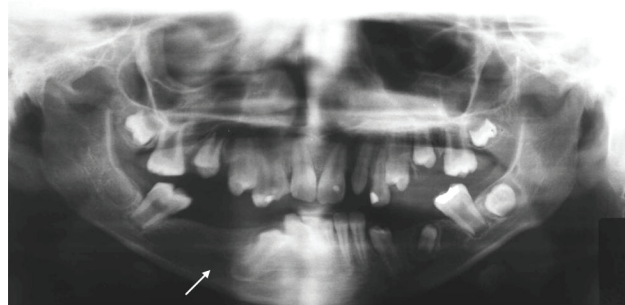


FIGURE 2. The patients' final orthopantomogram. We identified absence of permanent premolars (arrow).



FIGURE 3. On last visit the patient's hygiene was still poor (arrow).

side on the optimal treatment plan, so multiple frequent visits should be scheduled. The dentist must determine the permanent teeth that can erupt without orthodontic assistance. It is also advisable to maintain for as long as possible the primary dentition that will not be replaced by permanent teeth.

This 49,XXXXY patient is a typical example of how mental retardation and lack of adequate support by the family may result in poor oral hygiene and subsequent multiple extractions. In addition, the syndrome caused congenital absence of permanent teeth. These factors resulted in extensive secondary anodontia in our patient which affects his quality of life. If the child had been referred to us at an earlier age prior to the beginning of dental caries possibly he might have had a chance to maintain more healthy teeth following the same oral health care protocol we follow for children with special needs. Moreover, parents and caregivers should be aware of the importance of child's oral hygiene and proper nutrition.

There is limited experience concerning the correct dental treatment of children with 49,XXXXY syndrome. Our experience with one such patient could provide some guidance concerning the oral care of patients with 49,XXXXY syndrome.

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