



## CASE REPORT

# Taurodontism and Klinefelter syndrome: case report and review of the literature

Fatih Oznurhan, DDS, PhD

Cumhuriyet University School of Dentistry, Department of Pediatric Dentistry, Sivas, Turkey

### ARTICLE INFO

*Article history:*

Received 01 April 2013

Accepted 05 June 2013

*Keywords:*

Taurodontism

Klinefelter

Pediatrics

Dentistry

### ABSTRACT

This paper presents a seven-year old patient with Klinefelter Syndrome with dental treatment needs. The patient had mild mental retardation, learning and speaking difficulties, taurodontic teeth, missing premolars and incisor teeth. This paper emphasizes the importance of regular dental visits with panoramic radiography for early recognition of the systemic disorder and to improve quality of patient's life.

## INTRODUCTION

Klinefelter's syndrome (KS) is a disorder first described by Dr. Klinefelter et al. in 1942 that is characterized by an extra X chromosome in human cells.<sup>1-3</sup> The patient with KS have two X chromosomes with Y.<sup>2,4-8</sup> Approximately one in 1000 boys is born with an additional X chromosome and the most common type of this syndrome is 47,XXY, which represents 82% of these cases and the remaining cases show a variety of aneuploidies like 48,XXXY; 48,XXYY; 49XXXXY and mosaic patterns like 46,XY/47,XXY, 47,XXY/48XXXXY.<sup>9-11</sup> KS is probably the most frequent cause of male hypogonadism. The presence of two X chromosomes, one carrying a small

translocated fragment of Y chromosome, makes men with sex reversal syndrome very similar to men with KS. Both syndrome groups are characterized with eunoid habitus, but men with sex reversal syndrome are shorter than men with KS.<sup>5,6,12</sup> The diagnosis of KS is rare because the men with KS do not differ from healthy men till puberty.<sup>2,6,13</sup> The characteristic clinical findings of this syndrome may vary but most of the patients have infertility, tall, slender body habitus with long legs and shorter torso, a coarse face, skeletal anomalies, hypogonadism, osteoporosis, mental retardation, aspermatogenesis, variable gynecomastia, small testis and penis. In addition to these findings, language

Corresponding author at: Dr. Fatih ÖZTURHAN, Cumhuriyet University School of Dentistry, Department of Pediatric Dentistry Kampüs-Sivas/ Turkey, Tel: +90.346.2191010.3103, Fax: +90.346.2191237, E-mail: fatihozn@hotmail.com

delays, learning problems, behavioral difficulties and cardiovascular problems may occur.<sup>4,6,7,11,14,15</sup>

Oral findings of KS reported in many cases include cleft lip and palate, taurodontism, larger pulp chamber and crowns, maxillary and mandibular prognathism, congenital absence of permanent teeth, bifid uvula, shovel incisors and enamel defects.<sup>2,9,16,17</sup>

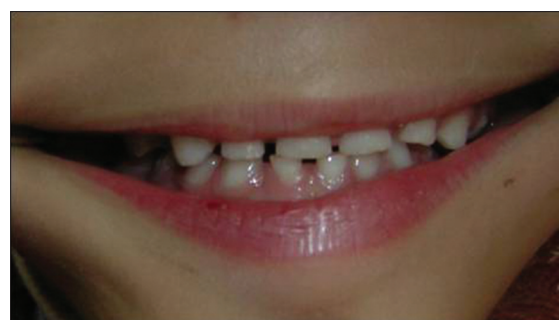
The most common dental finding of KS is taurodontism. Taurodontism is observed with several syndromes and anomalies including amelogenesis imperfecta, ectodermal disturbance, Down syndrome, Klinefelter syndrome, trichodonto-osseous syndrome, Mohr syndrome, Wolf-Hirschhorn syndrome and Lowe syndrome.<sup>2,8,18,20</sup> Tauorodontism was first proposed by Sir Arthur Keith<sup>18</sup> in 1913. The teeth were classified as normal, hypotaurodont, mesotaurodont or hypertaurodont by Shaw in 1928<sup>2,19</sup>. Hypotaurodontism is the least pronounced form, in which the pulp chamber is enlarged. Mesotaurodontism is the moderate form in which the roots are divided only at the middle third. Hypertaurodontism is the most severe form in which bifurcation or trifurcation occurs near the root apices.<sup>20</sup> Tauorodontism leads to constriction of the cemento-enamel junction, which results in vertically elongated pulp chambers, apical displacement of the pulpal floor, and bi or trifurcation of the root. This trait can be seen both in primary and permanent teeth, in single or several molars of the same quadrant and can be seen unilateral or bilateral.<sup>12,20</sup>

The aim of this case report was to describe a case with Klinefelter's syndrome and to discuss the oro-dental findings with current literature.

## CASE REPORT

A seven-year old boy was referred to the Department of Pediatric Dentistry with a

history of pain in his teeth. The patient had diminutive speech, sluggish movement. The mother of the child reported that he was premature (34 weeks) with 1680gr birth weight and had the chromosome anomaly KS, mild mental retardation, learning and speaking difficulties. He was having special education for his complaints. Mother was 25, the father was 31 year old and they have consanguinity. They were healthy, made intermarriage and had one another boy who was healthy. There was no other person in the family that had chromosome anomaly. A visual examination of the dentition revealed multiple caries, redness and swelling of the vestibulum of the left mandibular second primary molar (75) and right first primary molar (54). The maxillary arch was larger than mandibular arch and the right mandibular lateral incisor was missing (82) (Figure 1) and attrition of maxillary and mandibular anterior teeth was noted. In the panoramic radiograph examination (Figure 2), congenital absence of four permanent teeth; maxillary second premolar (15), mandibular left second premolar (35), mandibular right lateral incisor (42) and mandibular right second premolar was noticeable (45). Maxillary second (55) and mandibular first primary molars (74, 84) were hypertaurodont; maxillary left first primary molar (54) and mandibular right second primary molar (85) were mesotaurodont. The roots of the permanent molars are not yet formed



**Figure 1.** Large maxiller arch, attrition of the both mandibular and maxillary teeth, absence of the primary lateral can be seen.

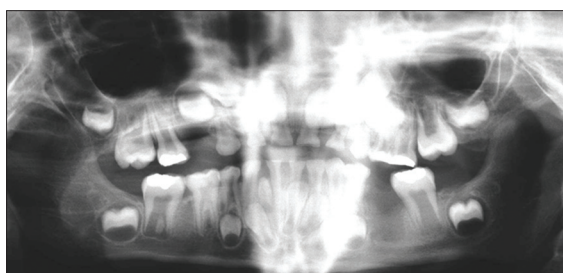
but four permanent molars had a large pulp chamber. Because of the difficulties in behavior guidance of the patient, the treatment was performed under general anesthesia. The tooth 54, 75 extracted, 64, 65 were restored with compomer and the teeth 55, 36, 46 were restored with amalgam (Figure 3).

## DISCUSSION

The relationship between an extra chromosome and taurodontism has been widely described. Taurodontism is an isolated anomaly and is detectable before puberty. It can be associated with several syndromes and anomalies like KS and trisomy 21 or Down syndrome.<sup>18</sup> The endodontic treatment of taurodontic teeth may be complicated. The shaping and obturation of the root may be difficult because of the canal orifices.<sup>2,18</sup> Identification of the patients with multiple taurodontic teeth could lead to early recognition of systemic disorder and improve quality of life. If the treatment of these teeth is delayed, the teeth could



**Figure 2.** Panoramic view of the patient before dental treatment.



**Figure 3.** Panoramic view of the patient after dental treatment.

be extracted. Many of the primary teeth, particularly second primary molars, have absence of their successors as in this case and the extraction of these teeth could lead to orthodontic malformation. And also the extraction of the teeth is difficult because of the dilated apical third.<sup>18</sup>

Children with KS have some dental anomalies like hypodontia. In the present case, four permanent teeth were absent. As described by some authors<sup>13,18</sup> that the patients with KS have mental retardation as in this patient. Most of the authors described prognathism about the jaws but only a large maxillary arch was seen in this patient.<sup>2,5,8,10,13,14</sup>

The patients with KS are undiagnosed because the clinical findings of the KS are often varied. It was found that nearly three quarters of estimated cases in the general population were undiagnosed.<sup>2,9</sup> The patients with meso- or hypertaurodontic teeth who do not have a syndrome known to be associated with taurodontic teeth should be consulted for chromosome analysis because there is a high association of taurodontic teeth with X-chromosome aneuploidy syndromes and the children may benefit from early diagnosis of KS.<sup>2,7,8</sup>

## CONCLUSION

Although taurodontism is a dental rarity, the patient should be examined carefully if there are other clinical findings related with this syndrome and the dentist should refer the patient for chromosome analysis to provide predictable long-term oral health care and improve the life quality.

### Why this paper is important for pediatric dentist?

The patients with KS are undiagnosed because the clinical findings of the KS are often varied.

Taurodontic teeth could be a clinical sign of this syndrome and the pediatric dentist should refer for chromosome analysis.

## REFERENCES

1. Giedd JN, Clasen LS, Wallace GL, Lenroot RK, Lerch JP, Wells EM, et al. XXY (Klinefelter syndrome): A pediatric quantitative brain magnetic resonance imaging case-control study. *Pediatrics* 2007 Jan;119(1):e232-40.
2. Joseph M. Endodontic treatment in three taurodontic teeth associated with 48,XXXXY Klinefelter syndrome: A review and case report. *Oral surgery, oral medicine, oral pathology, oral radiology, and endodontics* 2008 May;105(5):670-7.
3. Lee le Y, Quek SC, Chong SS, Tan AS, Lum JM, Goh DL. Clinical report: A case of Williams Syndrome and Klinefelter Syndrome. *Annals of the Academy of Medicine, Singapore* 2006 Dec;35(12):901-4.
4. Ross JL, Zeger MP, Kushner H, Zinn AR, Roeltgen DP. An extra X or Y chromosome: Contrasting the cognitive and motor phenotypes in childhood in boys with 47,XYY syndrome or 47,XXY Klinefelter syndrome. *Developmental disabilities research reviews* 2009;15(4):309-17.
5. Scepan I, Glisic B, Markovic E, Babic M. Craniofacial complex specificities in five men with sex reversal syndrome. *Clinical oral investigations* 2008 Sep;12(3):265-9.
6. Wattendorf DJ, Muenke M. Klinefelter syndrome. *American family physician* 2005 Dec 1;72(11):2259-62.
7. Bojesen A, Gravholt CH. Morbidity and mortality in Klinefelter syndrome (47,XXY). *Acta Paediatr Jun*;100(6):807-13.
8. Schulman GS, Redford-Badwal D, Poole A, Mathieu G, Burleson J, Dauser D. Taurodontism and learning disabilities in patients with Klinefelter syndrome. *Pediatric dentistry* 2005 Sep-Oct;27(5):389-94.
9. Hur M, Cho HC, Lee KM, Park H, Lee SY, Kim KN, et al. Cleft palate in a rare case of Variant Klinefelter syndrome with 48,XXXXY/46,XY mosaicism. *Cleft Palate Craniofac J* 2009 Sep;46(5):555-7.
10. Hunter ML, Collard MM, Razavi T, Hunter B. Increased primary tooth size in a 47,XXY male: A first case report. *International journal of paediatric dentistry/the British Paedodontic Society [and] the International Association of Dentistry for Children* 2003 Jul;13(4):271-3.
11. Fruhmesser A, Kotzot D. Chromosomal Variants in Klinefelter Syndrome. *Sex Dev* Apr 29.
12. Lahdesmaki R, Alvesalo L. Root lengths in the permanent teeth of Klinefelter (47,XXY) men. *Archives of oral biology* 2007 Sep;52(9):822-7.
13. Gorlin RJ, Redman RS, Shapiro BL. Effect of X-Chromosome Aneuploidy on Jaw Growth. *Journal of dental research* 1965 Jan-Feb;44:SUPPL:269-82.
14. Celebic A, Brkic H, Kaic Z, Vojvodic D, Poje Z, Singer Z. Occlusal plane orientation in Klinefelter syndrome (47,XXY males). *Journal of oral rehabilitation* 1997 Dec;24(12):942-6.
15. Yilmaz YF, Titiz A, Yurur-Kutlay N, Ozcan M, Unal A. Congenital bilateral parotid gland agenesis in Klinefelter syndrome. *J Craniomaxillofac Surg Jun*;38(4):248-50.
16. Hata S, Maruyama Y, Fujita Y, Mayanagi H. The dentofacial manifestations of XXXXY syndrome: A case report. *International journal of paediatric dentistry/the British Paedodontic Society [and] the International Association of Dentistry for Children* 2001 Mar;11(2):138-42.

- 17.** Alvesalo L, Tammissalo E, Townsend G. Upper central incisor and canine tooth crown size in 47,XXY males. *Journal of dental research* 1991 Jul;70(7):1057-60.
- 18.** Yeh SC, Hsu TY. Endodontic treatment in taurodontism with Klinefelter's syndrome: A case report. *Oral surgery, oral medicine, oral pathology, oral radiology, and endodontics* 1999 Nov;88(5):612-5.
- 19.** Varrela J, Alvesalo L. Taurodontism in 47,XXY males: An effect of the extra X chromosome on root development. *Journal of dental research* 1988 Feb;67(2):501-2.
- 20.** Marques-da-Silva B, Baratto-Filho F, Abuabara A, Moura P, Losso EM, Moro A. Multiple taurodontism: The challenge of endodontic treatment. *Journal of oral science*;52(4):653-8.

**How to cite this article:** Fatih Oznurhan. Taurodontism and Klinefelter syndrome: case report and review of the literature. *Cumhuriyet Dent J* 2014;17(Supp 1):21-25.