

Prepubertal diagnosis of Klinefelter syndrome in a patient with taurodontic teeth

PA Darbyshire, DDS, MS CJ Witkop, Jr., DDS, MS J Cervenka, MD, PhD

Abstract

A 9-year, 10-month-old male presented for dental treatment planning for multiple missing permanent teeth. Panoramic radiographs revealed taurodontic permanent first molars and primary second molars. The patient was of slim build with a long lower body and moderately long fingers. Because of the presence of taurodontic teeth, chromosomal analysis was done and revealed 47,XXY - Klinefelter syndrome. Patients with meso- or hypertaurodontic teeth who do not have a syndrome known to be associated with taurodontic teeth should be considered for chromosome analysis because of the high association of taurodontic teeth with X-chromosome aneuploidy syndromes.

Introduction

First described in 1942 (Klinefelter et al.), Klinefelter syndrome is a chromosomal aneuploidy with or without mosaicism. The most common types are 47,XXY, 46,XY/47,XXY, and 46,XX/47,XXY (Optiz 1979). The minimal diagnostic criteria are an X-chromatin-positive male with small, soft testes and karyotypic abnormality (Optiz 1979).

Most patients are phenotypically inconspicuous. Diagnosis in infancy is rare, and it is suspected only when the testes are significantly smaller than normal. Multiple minor anomalies usually are overlooked until puberty, when hypogonadism is sometimes associated with tall stature, approximately 10 cm taller than XY males (Optiz 1979). Associated minor anomalies include brachycephalic skull, low nuchal hairline, minor defects of ears, clinodactyly of the 5th fingers, 1 flexion crease on some fingers, and a decrease in the total ridge count of fingertip dermatoglyphics (Optiz 1979).

There is a deficient maturation of male secondary sex characteristics at puberty. Gynecomastia is variably present (Klinefelter et al. 1942). Disturbances in sexual function appear later and include infertility, impotence, and lack of libido. Also, obesity of the trunk and

osteoporosis are occasionally late features of the syndrome. While the majority of patients have normal intellectual development, in some patients the average intelligence is reduced, and 15% of patients are mentally retarded (Optiz 1979). Personality and character trait disturbances, emotional and behavioral troubles, and other psychological aberrations are common (Optiz 1979; Wiedemann et al. 1985). The frequency of occurrence of Klinefelter syndrome is between 1.18 and 2 in 1000 newborn males (deGrouchy and Turleau 1984; Wiedemann et al. 1985). Life expectancy is normal. Eunichoidism and other manifestations of hypogonadism possibly are due to progressive testicular sclerosis with loss of germinal and endocrine tissue (Optiz 1979).

In addition to the previously listed minor anomalies, Klinefelter syndrome and patients with X-chromosome aneuploidy generally exhibit taurodontic teeth (Keeler 1973; Stewart 1974; Feichtinger and Rossiwall 1977; Gardner and Girgis 1979; Jaspers and Witkop 1980; Farge et al. 1985; Witkop et al. 1988). Studies other than case reports that have investigated the presence of taurodontism in patients selected for X-chromosome aneuploidies have indicated a high but variable prevalence of the trait associated with X-chromosomal aneuploidies: Keeler (1973), 6 patients of 6 investigated; Feichtinger and Rossiwall (1977), 1 of 7; Gardner and Girgis (1978), 2 of 3; Farge et al. (1985), 4 of 4; and Witkop et al. (1988), 16 of 17. Taurodontic teeth have the furcation of the roots displaced toward the apex of the root. Consequently, the pulp chambers are enlarged and have a greater apico-occlusal height than the pulp chambers in cynodontic teeth (Witkop et al. 1988).

Shaw (1928) subclassified taurodontic teeth into hypo-, meso-, and hypertaurodontic types to reflect the severity of the trait. Taurodontic teeth have been reported to occur in from 2.5 to 3.2% of the chromosomally normal caucasian population; most have hypotaurodontic teeth (Keene 1966; Blumberg et

al. 1971; Witkop et al. 1988). Four studies have shown no differences in the prevalence of taurodontism in males and females (Blumberg et al. 1971; Shifman and Chanannel 1978; Ruprecht et al. 1987; Witkop et al. 1988), and one study indicates that taurodontism in males was twice as frequent as in females (Holt and Brook 1979).

Case Report

A 9 year, 10-month-old male presented at the University of Minnesota, Department of Pediatric Dentistry for long-term treatment planning regarding congenitally missing mandibular second premolars and central incisors. The patient's panoramic radiograph revealed the presence of unerupted small or peg permanent lateral incisors and taurodontic permanent molars and primary second molars (Fig 1). The second primary molars and maxillary first permanent molars were hypertaurodontic, while the mandibular first permanent molars were hypotaurodontic. The patient had a slim build, a long lower body, and moderately long fingers (Fig 2). There was no apparent mental impairment. Since these body features and the taurodontic teeth suggested that he might have an X-chromosome aneuploidy, a 10 mL blood sample was drawn for karyotyping. Twenty-three of 25 cells counted had 47,XXY chromosomes (Fig 3). The two cells with fewer than 47 chromosomes also had 2 Xs and 1 Y. The patient was diagnosed as having Klinefelter syndrome—47,XXY, with no indication of mosaicism.

Discussion

Klinefelter syndrome, with a risk of occurrence of up to 1 in 500 males, is not an uncommon finding. Generally, it is not discovered until puberty, when testicular atrophy is observed. Only infrequently has

the presence of taurodontic teeth in a patient led to diagnosis of Klinefelter syndrome. Chromosomal analysis is indicated in patients with the extreme forms of taurodontism (i.e., meso- or hypertaurodontic teeth), who, in addition, have other morphometric anomalies associated with X-chromosome aneuploidy (Stewart 1974; Jaspers and Witkop 1980).

There is no primary treatment for a chromosomal anomaly such as Klinefelter syndrome. Genetic counseling was provided to the parents by the Department of Oral Pathology and Genetics. Counseling involved description of the syndrome and its prognosis. Supportive treatment can include treatment of hypogonadism, surgical treatment of marked gynecomastia, psychotherapy for emotional complications, and prevention of obesity (Optiz 1979; Wiedemann et al. 1985). There is no apparent significant increase in risk for the patient's siblings (Optiz 1979).

The finding of multiple missing teeth, and small or peg permanent lateral incisors may be related to Klinefelter syndrome, as in general, as the number of chromosomes deviates from

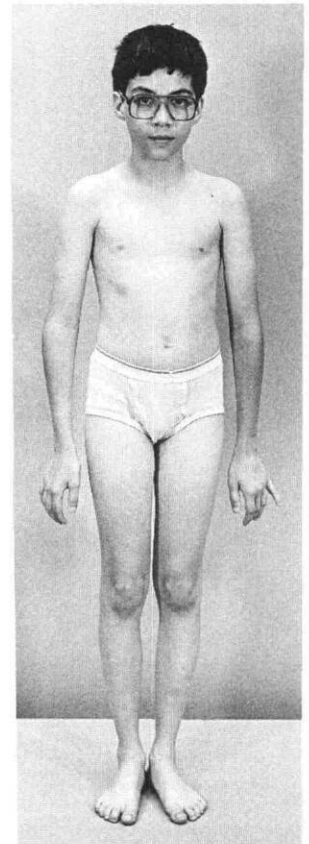


Fig 2. Grossly normal appearance with slim build and long lower body.



Fig 1. Panoramic radiograph showing taurodontic molars, congenitally missing mandibular second premolars and central incisors, and unerupted maxillary small or peg permanent lateral incisors.

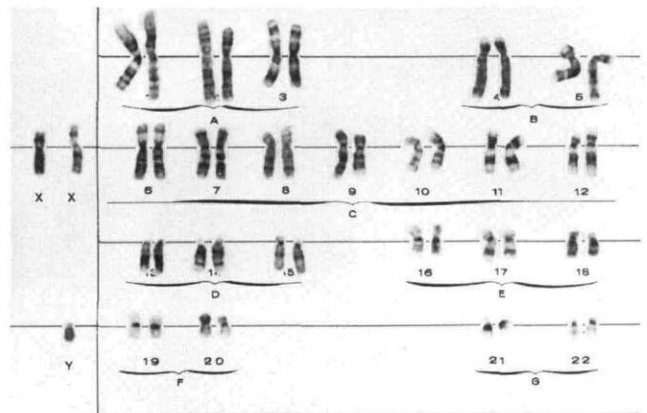


Fig 3. Karyotype - 47,XXY.

normal diploidy, the number of morphometric anomalies can be expected to increase (Shapiro 1983).

There is a controversy in the literature considering the etiology of taurodontism in the chromosomal aneuploid state. Epstein (1986) argued that there are certain genes on particular chromosomes which cause morphologic alterations. Shapiro (1983) argued in favor of additional chromosome material leading to a disruption in developmental homeostasis. Analysis of these factors by Witkop et al. (1988) is compatible with the latter concept.

Dr. Darbyshire is a pediatric dentist, private practice, in Kingston, Ontario, Canada. Dr. Witkop and Dr. Cervenka are professors, department of oral pathology and genetics, School of Dentistry, University of Minnesota, Minneapolis. Requests for reprints should be sent to: Dr. Peter A. Darbyshire, 773 Blackburn Mews West, Kingston, Ontario K7P 1Y5 Canada.

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Operation smile brings smiles

A 30-member group of medical professionals recently treated 100 children in Vietnam, as part of a project call Operation Smile. Plastic surgeons, pediatricians, anesthesiologists, dentists and nurses operated on the children, some with cleft lip and palate deformities. Other children received reconstructive surgery for burns and scars.

Another Operation Smile team, composed of 120 volunteer health professionals, traveled to the Philippines to operate on more than 600 children in three cities.

The organization, founded in 1982, has chapters in seven U.S. cities, and has aid programs in the Philippines, Africa, Southeast Asia and South America.