Eruption delay in a 47 XXY male: a case report

**ABSTRACT**

**Background** The 47,XXX syndrome, or Klinefelter syndrome, though it is a rare occurrence, it is the most common sex chromosome disorder affecting male subjects. This syndrome is underdiagnosed and seldomly before puberty. In this case, diagnosis was made before birth, through chorion villus sampling.

**Case report** A 16 month-old Italian male with 47 XXY syndrome showed the absence of primary teeth, with a delay of about 8-10 months, whereas during the first 15 months of life the auxological development has been normal both in weight and height (about 50th percentile). We assumed that this delay may be linked with Klinefelter syndrome, as sexual chromosomes play an important role in the dental development.

**Keywords** Dental eruption delay; Klinefelter syndrome; Primary dentition.

**Introduction**

Klinefelter syndrome is the most commonly occurring sex-chromosome disorder. It is characterised by the presence of one or more extra X chromosomes. Several X-aneploidy variants exist, including 47 XXY, 48 XXXY, 48 XXXXY, and 49 XXXXY [Hunter et al., 2003] but the karyotype 47 XXY is the most prevalent, occurring in approximately 80% of the cases [Gorlin, 1977]. The birth prevalence of chromatin-positive males is approximately 2:1000.

The “classical” Klinefelter syndrome is associated with the 47,XXX karyotype and is characterised by narrow shoulders, broad hips, sparse body hair, gynecomastia at late puberty, hypogonadism (small testes, azoospermia/oligospermia), androgen deficiency, hyalinization and fibrosis of the seminiferous tubules and elevated urinary gonadotrophins [Bojesen and Gravholt, 2007]. This means that these subjects do not produce enough of the testosterone hormone before birth and during puberty; as a consequence during puberty, the normal male sexual characteristics do not develop fully. Affected males may exhibit psychosocial problems and minor developmental and learning disabilities, including delayed speech and language acquisition.

However, a less distinct phenotype has been described.

Klinefelter syndrome is an underdiagnosed condition; only 25% of the expected affected patients are diagnosed, and of these only a minority are diagnosed before puberty [Hata et al., 2001]. Patients with Klinefelter syndrome should be treated with lifelong testosterone supplementation starting at puberty, to prevent the long-term deleterious consequences of hypogonadism and to secure proper masculine development of sexual characteristics, muscle bulk and bone structure [Bojesen and Gravholt, 2007].

Evidence accumulated over the last four decades supports the role of both X and Y chromosomes, independently of secondary hormonal influences, on growth and development of dental structures [Garn and Rohman, 1962]. In general, the results of measurements of enamel and dentine layers thickness in individuals with various types of sex chromosome abnormalities indicate that the X chromosome primarily influences enamel thickness, whereas the Y chromosome promotes both enamel deposition and dentine growth [Alvesalo 1981, 1985].

Cephalometric investigation reveals reduced calvarial size, reduced cranial base angle, and gonial angle wider than normal. Both maxillary and mandibular prognathism tend to occur [Ingerslev and Kreiborg, 1978]. In addition, various dental features have been observed, including taurodontism, [Stewart, 1974; Jaspers and Witkop 1980] congenital absence of permanent teeth [Stewart, 1974], shovel incisors [Gardener and Girgis 1978] and increased permanent tooth size [Townsend and Alvesalo, 1985].

**Case report**

A 15 month-old Italian male was referred to the Department of Special Care, School of Dentistry of Bologna University.

The Chorion Villus Sampling (CVS) that had been carried out at the 12th week of gestation revealed a male karyotype 47 XXY, consistent with Klinefelter syndrome. From the familiar medical history we learned that the mother had previously had two miscarriages, at the first month and at the 23th week of gestation because of chorioamnionitis.

The child was normally delivered at 36 weeks; birth
weight was 2,920 g and height 50 cm. Immediately after birth he was enrolled in a follow-up program at the Paediatric Department of the Sant’Orsola-Malpighi University Hospital of Bologna, Italy, in order to perform regular auxological evaluations.

New karyotype analysis confirmed the diagnosis of Klinefelter syndrome. The abdominal ultrasound was normal, while cerebral ultrasound showed a mild enlargement of LLVV and of the interhemisphere space.

During the first 15 months of life the child exhibited a good auxological development, both in weight and in height, with values in the 50th percentile (Fig. 1). His clinical conditions are good and the neuromotor development is age-appropriate as well. He underwent vitamin D implementation.

A clinical evaluation of the baby mouth revealed the total absence of primary dentition at the age of 16 months (Fig. 2).

**Discussion**

The child exhibited a severe delay in dental eruption of the primary teeth. Considering that the common timing of eruption of central lower primary incisors is around 6-8 months, we observed a delay of about 8-10 months. The eruption delay of primary mandibular incisors is an important clinical information because the auxological development of the child is otherwise normal (Fig. 1). In addition there are not changes in other organs of ectodermal derivation.

Though there is no evidence in literature that eruption delay of primary teeth is a typical Klinefelter syndrome feature, we suppose it may be linked with this syndrome because sexual chromosomes play an important role in the dental development.

Further studies are needed to investigate the relations between the number of sexual chromosomes and dental anomalies.

**Acknowledgements**

We would like to thank the Fondazione del Monte di Bologna e Ravenna for the support.

**References**