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SHORT COMMUNICATION

XYY KARYOTYPE IN A MENTALLY RETARDED MAN WITH PROGNATHISM AND MALFORMATION OF HIS HANDS AND TOE NAILS

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ABSTRACT

We report on a 25-year-old man who was referred for evaluation of possible Fragile X syndrome on the basis of mild mental retardation and malformation of his hands and toe nails. He was found not to have this syndrome but to have a 47,XYY karyotype. Only one other case of XYY syndrome with prognathism and malformation of hands has been reported.

Key words: Fragile X syndrome (FXS); Mental retardation; Karyotype

CASE REPORT

The incidence of XYY karytotype is about 1 in 1,000 human males [1]. Other than being slightly taller than normal these males have no striking phenotypical abnormality. They may also have behavioral problems [1].

We have investigated a 25-year-old male who was referred to our laboratory because of mild mental retardation and who was suspected to have the Fragile X syndrome (FXS). His height was 194 cm. His face was mildly dysmorphic and showed prognathism. He often showed an excessively negative mood and aggressiveness. He suffered from muscle cramp. Although he had a repaired hydrocoele, his genitalia were otherwise normal. He had hypoplastic toe nails and short hands. He had completed his primary school education and at the time of diagnosis he worked as a mechanic.

Blood lymphocyte cultures from the patient were set up in RPMI 1640 medium supplemented with 20% FBS and proliferation was stimulated with phytohemagglutinin. The cells were harvested after a 72-hour culture time [2]. In addition, a cytogenetic test was performed for FXS using standard protocols [2]. The karyotypte derived from these cells showed an additional chromosome Y and was thus ascertained as 47,XYY. No Fragile X syndrome was observed after screening over 100 cells.

In the last decade there has been a significant increase in the proportion of XYY males detected prenatally [3,4]. Therefore, it is very important to obtain a clear idea of possible problems that may arise during further development of boys with the 47,XYY karyotype, especially in cases where this diagnosis is made prenatally.

In general, XYY males are relatively tall [5]. A study of 38 XYY males found that their height, weight and head circumference was usually above normal [5]. The craniofacial dimensions in eight

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adult 47,XYY males were larger than those of normal male and female controls [6]. At least two studies have indicated that 47,XYY males had longer tooth roots than normal male and female controls [7,8].

Prenatal diagnoses of the XYY karyotype accompanied with some clinical anomalies have been published. Phupong and Sittisomwong [3] reported a fetus with posterior cervical cystic hygroma with no other structural anomalies who was found to carry the 47,XYY karyotype. Brain anomalies associated with the 47,XYY karyotype has also been detected prenatally [4].

The occurrence of the XYY syndrome with apparently unrelated diseases has been reported. For example, bilateral cryptorchidism has been found in an 11-month-old boy with the 47,XYY karyotype [9]. Asano *et al.* [10] presented a case of myotonic dystrophy with 47,XYY [10]. An XYY karyotype male has been diagnosed as having Prader-Willi syndrome [11].

In some reported cases, mental retardation and/or delayed language and/or motor development have been seen in XYY patients [11,12]. For example, Geerts *et al.* [12] have found that most of the XYY children attend kindergarten in the normal education circuit, but in 50% of cases psychosocial problems are documented. From primary school age on, psychiatric disorders such as autism are seen more frequently in XYY males than XY males.

A significantly higher frequency of antisocial behavior has been reported in XYY karyotype males in adolescence and adulthood. This was believed to be mediated mainly through lowered intelligence [13]. A 9-year-old boy with 47,XYY and behavioral 'internalizing' and 'externalizing' symptoms and deficient spatial memory has been reported [14]. There is still dispute on whether or not XYY men are more likely to indulge in criminal and violent behavior than 46,XY males.

Freyne and O'Connor [15] reported two cases of XYY males who had committed murder. Briken *et al.* [16] found that in 13 men, perpetrators of sexual homicide, three had the XYY karyotype [1.8%].

We could find only one published article concerning a XYY karyotype male with prognathism and hand malformation such as the one reported here [17]. To the best of our best knowledge, no XYY male has been reported with toe nail malformations. This combination of XYY male and hand and toe nail deformities may be coincidental. More information on the mental and physical characteristics of XYY karyotype males is required so that parents found to have a fetus with the XYY karyotype can be guided in making a well-informed decision.

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