Jpn. J. Human Genet. **29**, 387–389, 1984

A 7q− SON OF AN XYY FATHER

Tadashi KAJII and Ichiro MURANO

Department of Pediatrics, Yamaguchi University School of Medicine, Ube, Yamaguchi 755, Japan

Summary A 47,XYY man had a son with deletion of the distal segment of 7q and a karyotypically normal daughter.

INTRODUCTION

XYY men, when fertile, overwhelmingly produce normal children. A minority of their children, however, may have chromosomal or phenotypic abnormalities. Those so far reported included XYY, trisomy 21, mosaics, anencephaly and spina bifida (cf. Stoll *et al.*, 1979, for references).

We wish to describe a 47,XYY man and his son with deletion of distal segment 7q, a hitherto undescribed association.

CASE REPORTS

S.N. was born on October 3, 1949. Apart from his large stature (173 cm, 83 kg) he had no particular health or medicolegal problems. He married on March 11, 1979, and a son was born on August 9, 1980 after a normal term pregnancy. When seen by us at age 2.5 years, the son was mentally retarded (DQ 63), measured 85.5 cm (−1.4 SD), weighed 12.75 kg (−0.3 SD) and his head circumference was 46.4 cm (−1.8 SD). He had a wide and flat face with a prominent forehead, heavy cheeks, a bulbous nose with a fleshy, flattened tip, thick lips, a small and round chin (Fig. 1). Also noted were a flattened occiput, a high-arch palate, a short neck, wide-spaced nipples, and protruding heels. Intravenous pyelography revealed left double ureters and double pelves. He developed epileptic seizures controllable by anticonvulsants. His G-banded karyotype was 46,XY,del(7) (q32 → qter) (Fig. 2). This chromosomal abnormality led to the study of the chromosomes of the father, whose karyotype was 47,XYY, and of the mother, whose karyotype was normal. His younger sister, born in September, 1983, was chromosomally normal.

Received July 16, 1984

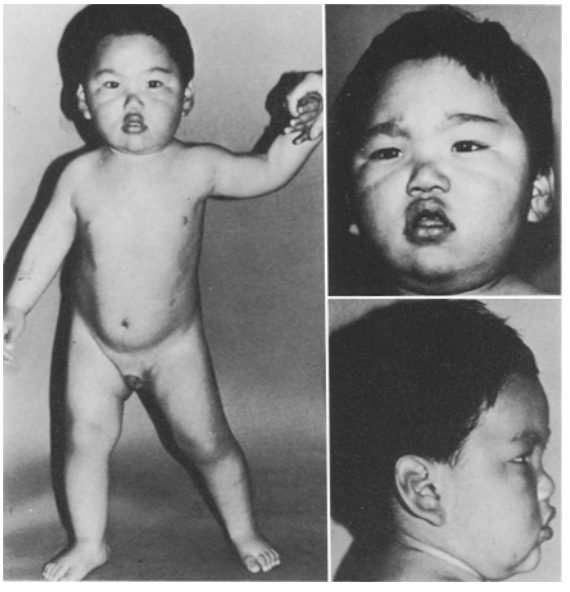

Fig. 1. The son at age 2.5 years.

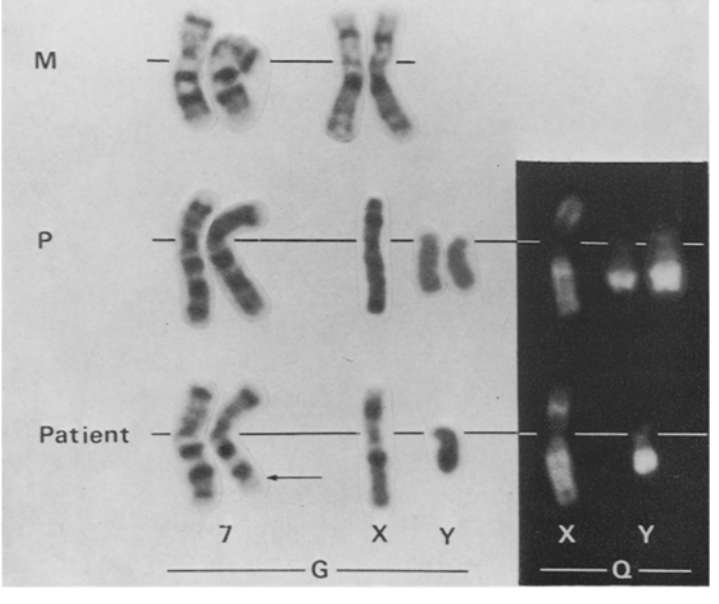

Fig. 2. Partial G- and Q-banded karyotypes from the mother (top), father (middle) and the son (bottom). The arrow indicates the 7q− chromosome.

DISCUSSION

We are aware of 57 children born to 28 XYY men (cf. Stoll *et al.*, 1979, for references). They included 44 apparently normal children (23 boys, 19 girls, and 2 children of undisclosed sex), 19 of whom were karyotyped and normal, three with trisomy 21, three with XYY, two with mosaics and five deaths soon after birth. Many of the latter children were ascertained in view of their abnormal phenotype, and therefore influenced by both ascertainment and publication bias. To this we may add our two children, a boy with *de novo* 7q3 monosomy and a chromosomally normal girl.

Monosomy 7q3, as observed in the boy we described, has been reported in at least 14 instances (Bernstein *et al.*, 1980). Most of the 7q3 monosomies occurred *de novo*, with the breakpoint usually being in q32. The phenotype in our boy agreed well with that in the previously described patients.

The correlation between the XYY karyotype in the father and 7q3 monosomy in his son remains to be determined. The association may well be ascribed to chance. It has been known that there is a strong bias towards paternal errors in *de novo* rearrangements that are not of the Robertsonian type and that do not have additional chromosomes (Chamberlin and Magenis, 1980). Therefore, it seems reasonable to assume that the 7q deletion in the son was of paternal origin. Interchromosomal effects are thus a possibility, in the sense that the presence of an extra chromosome may influence during meiosis the behavior of other chromosomes. This is different from the usual situation in which balanced structural rearrangements or aneuploidy for sex chromosomes appear to influence the segregation of other chromosomes (Lejeune, 1963; Grell and Valencia, 1964).

REFERENCES

Bernstein, R., Dawson, B., Morcom, G., Wagner, J., and Jenkins, T. 1980. Two unrelated children with distal long arm deletion of chromosome 7: clinical features, cytogenetic and gene marker studies. *Clin. Genet.* **17**: 228–237.

Chamberlin, J. and Magenis, R.E. 1980. Parental origin of *de novo* chromosome rearrangements. *Hum. Genet.* **53**: 343–347.

Grell, R.F. and Valencia, J.I. 1964. Distributive pairing and aneuploidy in man. *Science* **145**: 66–67.

Lejeune, J. 1963. Autosomal disorders. *Pediatrics* **8**: 326–337.

Stoll, C., Clavert, F.A., Beshara, D., and Buck, P. 1979. Abnormal children of a 47,XYY father. *J. Med. Genet.* **16**: 66–68.