47,X,i(Xq),Y Karyotype in Klinefelter's Syndrome

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Summary. This is a case report of 47,X,i(Xq),Y in a 24-year-old infertile male with Klinefelter's syndrome. C staining indicated that this isochromosome X had a single small centromere. BUdR incorporation revealed the isochromosome X to be late replicating.

Klinefelter's syndrome classically represents the aneuploid chromosomal complement 47,XXY with one extra X chromosome. Clinically, patients with this syndrome present azoospermia, gynecomastia, a variable degree of eunuchoidism, elevated urinary gonadotropins and small testes with atrophy and hyalinization of the seminiferous tubules, and preservation of interstitial Leydig cells.

Many numerical chromosomal variants of this syndrome have been described, such as 48,XXXY or 49,XXXXY. They have usually been associated with mental retardation. Very few structural variants, mainly deletions or rearrangements of sex chromosomes, have been reported in Klinefelter's syndrome (Chandra et al., 1971; Nielsen et al., 1966, 1976; Zang et al., 1969).

Case Report

Our patient was first seen with his (unrelated) wife at the Fertility Center, Royal Victoria Hospital, for investigation of primary infertility. They had been married for two years. There were no other known cases of infertility in the family. Basic investigation of the patient's wife showed no abnormality. The patient was 24 years old, 166.4 cm tall and weighed 77.7 kg. He had an average male habitus but a tendency to obesity. The upper to lower segment ratio was normal. Arm span exceeded height by 14.5 cm. He had a moderate amount of pubic and axillary hair growth (pubarche at 13 years of age) but complete absence of hair growth on his face, extremities, nostrils, external ears and back.

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The phallus was small, measuring approx. 5 cm in length and 2 cm in diameter. The scrotal sac was small and contained testes each approx. 1 cm in diameter.

Laboratory investigations showed raised pituitary gonadotropins with FSH 70 ng/100 ml (normal 5—30) and LH 10.4 (normal 2—10). Plasma testosterone, estrone and estradiol were low with testosterone 130 (normal 273—1200), estrone 2.2 (normal 4—10) and estradiol 0.7 (normal 2—7).

Dermatoglyphic analysis found nine digital ulnar loops and one whorl (right third finger) with a total digital ridge count of 117. Palmar axial triradii were proximally placed (t) bilaterally and palmar a—b ridge counts were 38 and 34 for left and right, respectively. A hypothenar pattern and third interdigital loop (1d) were present bilaterally. A whorl was present on the hallucal area of each foot.

Testicular biopsy was successful on the left side; the tissue section showed complete hyalinization, fibrosis and atrophy of the majority of tubules. The remaining tubules revealed marked peritubular fibrosis and thickening of the basement membrane. A few partially preserved seminiferous tubules were lined by Sertoli cells and rare germ cells were noted. There was no evidence of germ cell maturation or sperm formation. The prominent Leydig cells were present in clumps and clusters.

Fig. 1. G banding of sex chromosomes—normal XY and isochromosome X

Fig. 2. DNA replication pattern in normal XY and i(Xq)