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A 47,XXq-Y Klinefelter Male*

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During a continuing survey of chromosome anomalies in Bangalore we had occasion to study an 18-year-old Muslim boy (case no. IGS 12H) who had been clinically diagnosed as a Klinefelter. This report summarizes the results of chromosomal, dermatoglyphic, and other studies on this patient.

Case Report

The patient was first seen by one of us (G.N.R.) when he was in St Martha's Hospital being treated for viral hepatitis. He is a dull-looking boy with a rather large build for his age. The lower jaw is prominent, the hands are large, and there is gynaecomastia on both sides. He also shows sinus bradycardia. Body hair has a feminine distribution. The testes are small and soft and a biopsy showed atrophy of the seminiferous tubules.

He is the 3rd of 5 children. His mother, two older sisters, and a younger brother and younger sister are apparently normal. His father is no longer alive.

Sex Chromatin and Chromosome Analyses. Cells from his hair roots and buccal mucosa were studied twice. On both occasions one sex chromatin body was seen in 40–60% of the cells examined. In many cells the sex chromatin body appeared to be distinctly smaller than usual.

Peripheral blood was cultured on 4 different occasions using the method of Hungerford (1965). H^3-thymidine radioautographs were prepared according to standard methods to study the late-replicating chromosomes. Approximately 5 hours before harvest, tritiated thymidine (specific activity: 5·5 c/mmole; purchased from the Bhabha Atomic Research Centre, Bombay) was added to the cultures at a final concentration of 1 μc/ml of medium. The remaining procedures were essentially similar to those described by Chandra and Hungerford (1967).

Nearly all the cells had 47 chromosomes. There were 15 chromosomes in the C group and an additional chromosome resembling chromosome 16 in morphology. This extra chromosome was somewhat larger than 16 and could be distinguished from chromosome pair 16 in all good preparations (Fig. 1). This chromosome was distinctly late-replicating in over 80% of the labelled metaphases.

The extra chromosome was interpreted as a deleted X because of (1) the presence of sex chromatin; (2) an apparent reduction in the size of at least some of the sex chromatin bodies; (3) the presence of 47 chromosomes —but only 15 chromosomes in the C group; and (4) late replication of the extra chromosome. A little less than half the long arm appears to have become deleted from one of the X chromosomes. The karyotype was thus interpreted as being 47,XXq-Y.

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TABLE I

DERMATOGLYPHIC DATA FROM THE PATIENT

	Left Hand					Right Hand					No. of Digital Triradii 20			
	v	iv	iii	ii	i	i	ii	iii	iv	v	Summed Ridge-count			
Pattern type	W	W	W	W	W	W	W	W	W	W	Ulnar loop	Radial loop	ARC	TRC
Ulnar loop count	19	19	18	17	19	21	17	17	18	19	184			
Radial loop count	14	14	10	11	10	13	12	5	16	20		125	309	185
Maximal *atd* angle	38°					35°					Sum 70°			
a–b ridge-count (n)	45					36					Sum 81			
a–b ridge distance (mm) (d)	23½					21½					Sum 45			
Mean ridge breadth (mm)											(dL + dR)/(nL + nR + 2)45/83 = 0·542			
Palmar triradii	*abcdt*					*abcdt*					Total number 10			

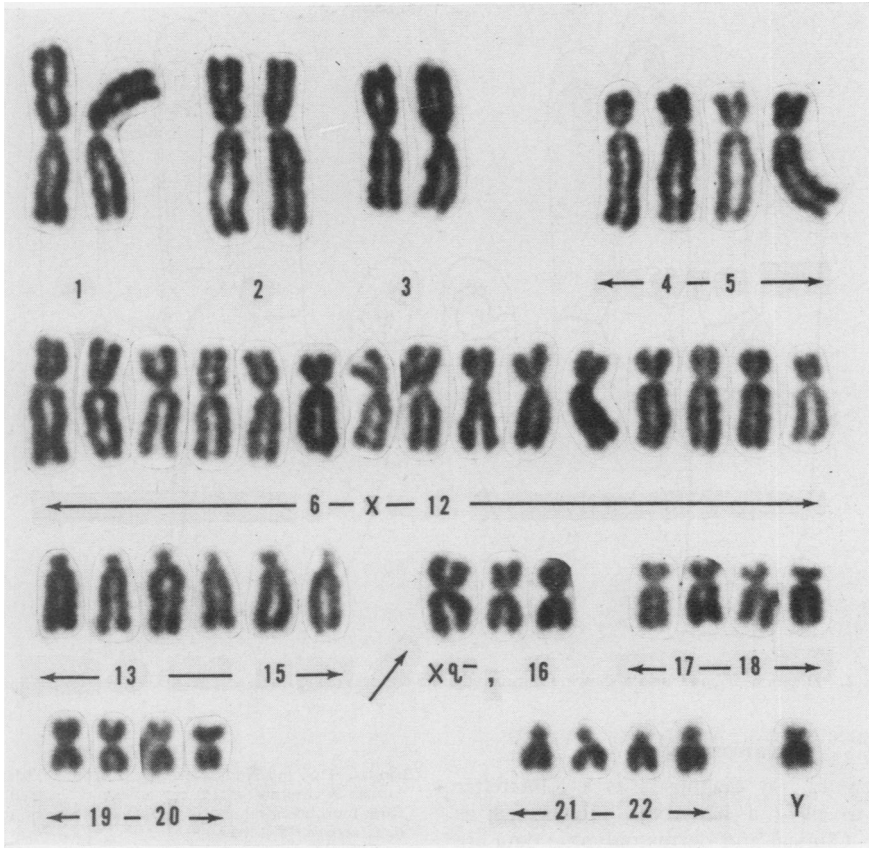

FIG. 1. Karyotype from a lymphocyte culture showing the deleted X chromosome (arrow).

Chromosome counts and karyotype analyses gave no evidence of mosaicism.

Karyotypes of the mother of the patient and those of his younger brother were apparently normal.

Dermatoglyphs (Professor Penrose). The data of the palm and finger prints of the patient and his brother are summarized in Table I. Professor Penrose states that 'the patterns are nowhere abnormal but the comparison between the two is very interesting. In particular, the total ridge count for the patient is 185, just 14 less than that of the brother. In Europeans, for Klinefelter males, the average count is 133 as compared with 145 for XY males. The total reduction here is brought about by the patient's smaller whorls even though he has 10 of them as compared with his brother's 8. Moreover the palms of the patient show low pattern intensity, compared with his brother who has a loop in each hypothenar region. This low pattern intensity is characteristic of Klinefelter cases.'

Xg^a Blood Grouping (Drs R. R. Race and Ruth Sanger). Blood samples from the patient, his sister, and mother were all found to be Xg(a+). The Xg groups were thus uninformative as to the source of the patient's deleted X chromosome.

Colour Vision (Dr S. G. Ramnarayan Rao). Both the patient and his younger brother had normal colour vision.

Discussion

Deletions in the long arm of the X have been reported in several phenotypically male individuals (eg, Crawfurd, 1961; Valencia *et al*, 1964). However all these individuals were mosaic with only one or two of the cell lines containing the deleted X. In fact, the only apparently non-mosaic case we are aware of is the one reported by Nielsen (1966). He reported a Klinefelter patient who was then 54 years old and whose karyotype is very similar to that of the present case. However, in Nielsen's case the deletion appears to involve a major portion of the long arm.

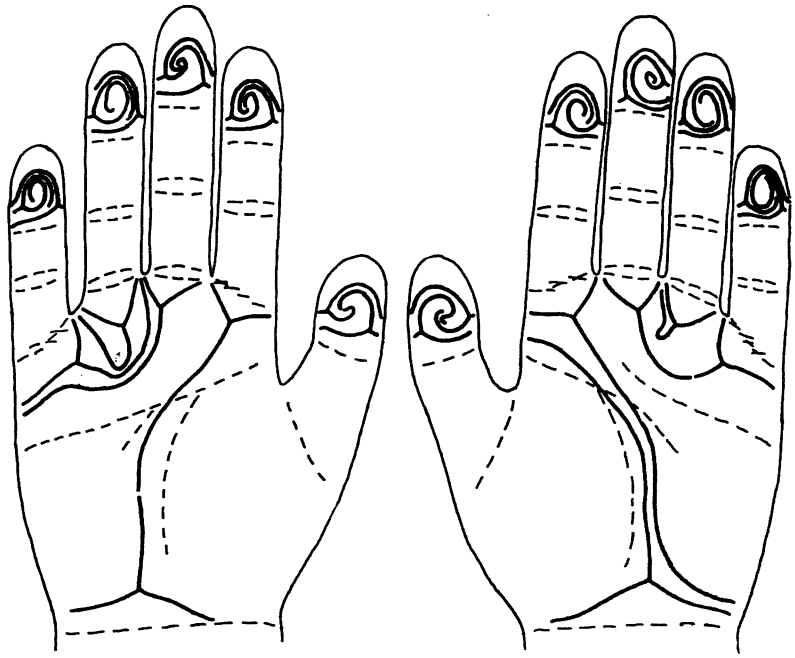

FIG. 2. Diagram of hand prints showing dermatoglyphic features (solid lines) and flexion creases (dotted lines).

Summary

An 18-year-old boy diagnosed as a Klinefelter was found to have a karyotype interpreted as 47,XXq-Y. Clinical and dermatoglyphic data are also given.

We wish to thank Dr S. R. V. Rao and his associates at the Department of Zoology, Delhi University for preparing the radioautographs, Professor L. S. Penrose for the dermatoglyphic analysis, Dr R. R. Race and Dr Ruth Sanger for the Xg data, and Dr S. G. Ramnarayan Rao for the colour vision tests.

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