**120. A 45,X/46,XY,del(Y)(q11) Mosaicism in a
Phenotypically Normal Male Adult**

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In a chromosome survey of a phenotypically normal male adult, the 45,X/46,XY,del(Y)(q11) mosaicism was found. In spite of the large proportion of 45,X-cell, he had the normal male external genitalia and no stigma of Turner's syndrome. The clinical symptom suggesting a sex chromosome aberration was sterility alone.

Case reports. The patient was a 36-year-old man. After a normal pregnancy, he was born at term as the first child of three children between a 19-year-old mother and a 26-year-old father. There was no history of abortion, stillbirth, congenital malformations, mental retardation, exposure to the atomic bomb, radiation, and consanguinity in this couple. His parents, younger brother and younger sister were phenotypically normal.

At birth he was 2750 g in weight. His postnatal course was smooth and normal. There was no sign of developmental retardation or mental retardation. He was graduated from the university and got a job. He married at 29 years of age, but he had no child. He has suffered from recurrent hematological disorders since 23 years of age and psychosis; schizophrenia since 31 years of age. He was admitted to our sanatorium for the treatment of leukopenia. At admission in his peripheral blood examination, the counts of white blood cell were 1600/mm³.

Based on the cytological analysis, as one of the examinations, the chromosome aberration; 45,X/46,XY,del(Y)(q11) was found. He had the normal external genitalia showing a normal sized testicle and a normal sized penis. He had also the normal secondary sex characters; pubic hair, axillary hair, beard, the Adam's apple and so on. There was no Turner-like stigma on his body. He had no complaint in the sexual function. The hormone level was given in accompanying table (Table I).

Cytological findings. The chromosomal preparations were made on the leucocyte cultures from the patient. The G- and Q-banding differential stainings were also applied for the chromosome identification. The chromosome counts were made with 112 well-delineated metaphases. The karyotype was analysed in 100 cells by the conventional Giemsa staining, in 12 cells by the G-banding, and in 12 cells by the Q-banding analyses. The chromosome analysis of the patient based on the conventional Giemsa staining method showed the cells having 45 chromosomes and those having 46 chromosomes. The G- and Q-banding analyses revealed that the missing chromosome in 45,X-cell was Y chromosome and that

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Table I. Hormone levels of the patient

Material	Hormone	Value	Normal range
Urine	17 OHCS	7.4 mg/24 H	3.1~8.7
	17 KS	7.6 mg/24 H	4.2~12.4
	Estrogen	23.0 mcg/24 H	8~45
	Testosteron	20.3 mcg/24 H	12.0~60.0
Serum	LH	34.5 mIU/ml	6.5~34.5
	Progesteron	0.73 ng/ml	0.10~0.40
	Testosteron	327 ng/dl	300~850

Fig. 1. The partial karyotype of the patient. A: Conventional Giemsa staining. B: G-banding staining. C: Q-banding staining. (45,X/46,XY,del(Y) (q11).)

Table II. Cytogenetic findings in the leucocyte cultures of the patient

Specimen	Chromosome counts		Total cells counted
	45,X	46,XY,del(Y) (q11)	
Conventional Giemsa staining	65	35	100
G-banding differential staining	11	1	12
Total cells counted	76	36	112

Y chromosome in 46,XY-cell showed unusually shortened long arm deleted at q11→qter (Fig. 1, A, B). The proportion of 45,X-cell to 46,XY,del(Y) (q11)-cell was 76 to 36 (Table II). The Y-chromatin was not observed in the examination of 100 cells stained with the quinacrine mustard (Fig. 1, C). Then his chromosome formula was 45,X/46,XY,del(Y) (q11). The chromosomal examinations of his parents and siblings were not cooperated.

Remarks and conclusion. The development of the secondary male sex characteristics is strictly dependent on the testicular differentiation of the embryonic gonad (Jost, 1970). And the cytogenetic investigations have demonstrated that, for such a differentiation, the presence of the Y chromosome is necessary (Hamerton, 1968). However the exceptions; cases of phenotypically

male with the absence of a detectable Y chromosome were found, and have been mentioned as "XX-male" and "XO-male". Since De la Chapelle *et al.* (1964) reported the first case, more than 70 cases of "XX-male" have been recorded in reference to Bauch *et al.* (1980). Those cases showed usually the Klinefelter-like clinical features and had the small testicle. About "XO-male", as far as known to date, only three cases have been reported (Lo Curto *et al.* 1974; Forabosco *et al.* 1977; Kadotani *et al.* 1985). All these cases had also the small male external genitalia and the Turner-like stigmata.

Our case had 45,X/46,XY,del(Y)(q11) karyotype and proportion of 45,X-cell was 67.85%. Several authors reported on the mosaicism between 45,X and 46,XY (Puleo *et al.* 1983; Cheng *et al.* 1983). These cases showed also the clinically Turner's syndrome. And the previous case reports on the mosaic typed chromosome abnormalities have demonstrated that the clinical features depended on the proportion of component-cells.

In spite of the large proportion of 45,X-cell, our case had clinically normal external genitalia, with the normal size and the normal form, and no stigma of Turner's syndrome. Therefore, the application of findings in the previous case reports to our case is not suitable.

We expect further investigations which explain the contradiction between the karyotype and the phenotype of our case.

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