

REPRODUCTIVE FAILURE IN 45X/46XX FEMALE

by

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Turner's syndrome and its variants occur in 0.1 per 1000 live births. These patients mainly fall into chromatin positive and chromatin negative groups (Levine-1971). The chromatin positive patients are mosaics and they often menstruate but are very rarely fertile. The present paper reports reproductive failure in 45X/46XX mosaic Turner.

CASE REPORT

Mrs. P., 28 year old, was referred to the clinic for reproductive failure. Her obstetric history showed that she had given birth to 2 full term babies—a boy and a girl (Fig 1). Both the children died within 24 hours of birth. The cause of death was some heart abnormality. Patient's third pregnancy (Fig. 1) ended in a 4½ months abortion. The fetus was malformed and the exact sex was not specified.

General examination of both husband and wife was done. Husband was normal. Patient was short statured (4 feet 10 inches) had short neck and low spina bifida at L₅ L₆ level. She was otherwise in good physical and mental health. The patient had attained menarche at 13 years of age and has regular cycles.

Material and Method

The buccal smears of both husband and wife were taken. Husband was X-chromatin negative and therefore, normal. Wife was X-chromatin positive with 30%

count. The size of the X-chromatin was within normal range ($0.071 \mu-0.1/\mu$).

The peripheral leukocyte culture was performed on the couple by method of Moorhead *et al* (1960). Husband had normal karyotype (46XY). Wife had two cell lines. In this case 100 cells were counted and 5 karyotyped. 70% of the cells had 46XX cells and 30% had 45X (Figs 2 and 3). In the 45X karyotype additional chromosomal material was observed on chromosome 18 (18 P+).

Discussion

Mosaicism (XX/XO) is the most common finding in patients with chromatin positive gonadal dysgenesis and is next to XO chromosomal constitution (Levine, 1971). These women usually exhibit few associated somatic anomalies. The women may menstruate and even be fertile.

Very few cases with 45/46XX chromosomal constitution being fertile have been reported (Liber, 1973), Beishun and Morton (1968) reported a woman with karyotype 45X/46XX who had repeated abortions and malformed fetuses. It has been known that patients with abnormal chromosomal constitution like this can become pregnant but the outcome of pregnancy is often unsuccessful. Hence it is very important that patient with bad obstetric history should undergo chromosomal analysis so that it can help us to offer better genetic counseling.

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See Figs on Art Paper III