X long arm deletion with oligomenorrhoea

SUMMARY A 35-year-old female patient with oligomenorrhoea had a deletion of the long arm of the X chromosome. The breakpoint at band q23 caused infertility in spite of excessive pituitary stimulation. The aberrant X chromosome was inactivated in all cells analysed.

Deletion of the long arm of the X chromosome is one of the most frequent structural aberrations of this chromosome. Fertility and reproduction are affected by these chromosomal anomalies.1-3 This paper presents a patient with a deletion of the long arm of the X chromosome and oligomenorrhoea.

Case report

A 35-year-old patient was admitted to the Clinic of Gynaecology and Obstetrics for examination because of oligomenorrhoea. Menarche had occurred at the age of 11, but menstruation was irregular at 1 to 3 month intervals; these intervals becoming progressively longer. Her weight and height were 51 kg and 160 cm, respectively. The breasts were well developed and pubic and axillary hair was normal. Laparoscopy showed that the uterus, fallopian tubes, and ovaries were normal. Gonadotrophins were raised (FSH 76 mIU/ml, LH 68 mIU/ml) and the oestradiol level was high (241 pg/ml). Laparoscopy showed that ovulation was remarkable on the right ovary.

CYTOGENETIC FINDINGS

Forty-two metaphases from peripheral blood were

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Requests for reprints to Dr A Tóth, Department of Obstetrics and Gynaecology, Postgraduate Medical School, Szabolcs u35, H-1135 Budapest, Hungary.
examine. GTG banding showed a deletion of the long arm of the X chromosome at band q23. Six hours before the harvest, 100 mg/l of 5-bromodeoxyuridine was added to the culture. RBA banding showed that in 50 cells analysed the aberrant X chromosome was inactivated (figure). The chromosomes of the patient's parents could not be examined.

Discussion
A critical role of the X chromosome in primary determination of sex and fertility has recently been revealed. Studies of deletions of the X chromosome showed that in most cases of Xq aberrations the breaks occur within the critical region Xq13–q27, which is responsible for normal ovarian and female sex development. Female patients with deletions of the long arm of the X chromosome have two different phenotypes: one with characteristics of Turner's syndrome, and the other with a picture of pure gonadal dysgenesis. The critical point for these two phenotypes lies within the band q22 or q21. Female patients with deletions distal to the critical band have amenorrhoea without any stigmata of Turner's syndrome.

In our patient, as in most cases of deleted Xq, the proximal part of the long arm of the X chromosome (with its inactivation centre) was left intact and the deleted X chromosome was inactivated. In our patient, the breakpoint occurred at band q23 within the critical region of the X chromosome, resulting in infertility. The deletion of genetic material of one X chromosome is a frequent cause of hypergonadotrophic amenorrhoea and ovarian dysfunction. In spite of normal laparoscopy findings the high level of gonadotrophins in our patient and progressive oligomenorrhoea showed altered ovarian function. Serum oestradiol was high owing to the excessive pituitary stimulation.

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Cd banding studies in a homologous Robertsonian 13;13 translocation

Summary A phenotypically normal female with a history of two miscarriages was found to have the karyotype 45,XX,t(13p;13p). C banding showed the translocation to have two regions of centromeric constitutive heterochromatin, silver staining showed an active NOR in 60% of the cells screened, and Cd banding studies showed a single Cd band with absence of the Cd band at the suppressed centromere.

Eiberg produced a method which clearly showed in metaphase two dot-like bodies situated in the area where the centromere should be. These centromeric dots are considered to be the kinetochores and appear to be equal in size.

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X long arm deletion with oligomenorrhoea.

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