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References

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Case report
A 35-year-old female patient with oligomenorrhea 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.

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examined. GTG banding\(^4\) 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\(^5\) 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,\(^6\) 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.\(^1\)\(^-\)\(^3\)\(^7\) The critical point for these two phenotypes lies within the band q22 or q21.\(^8\)\(^-\)\(^10\) Female patients with deletions distal to the critical band have amenorrhoea without any stigmata of Turner's syndrome.\(^8\)\(^-\)\(^11\)

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)\(^12\)\(^-\)\(^13\) 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 serum 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\(^1\) 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.\(^1\)\(^-\)\(^3\)

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