Case reports

Research Council, Ministry of Health, Hungary, 5-12-1101-02-1/L, 6-09-1102-01-1/Gá.

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References


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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

Received for publication 26 November 1981

FIGURE Aberrant X chromosome in situ (RBA banding).

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examined. 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-bromodeoxy-
uridine 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. 1–8 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. 8,9

In our patient, as in most cases of deleted Xq,
the proximal part of the long arm of the X chromo-
some (with its inactivation centre) 12 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 dys-
function. 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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References

1 Hecht F, Jones D, Delay M, Klerit M. Xq— Turner’s
syndrome: reconsideration of hypothesis that Xp— causes

2 Bocian M, Krmpotić E, Szego K, Rosenthal L. Somatic
stigmata of Turner’s syndrome in a patient with

3 de la Chapelle A, Schroder L, Hoahtela T, Aro P. Deletion
mapping of the human X chromosome. Hereditas

4 Seabright M. A rapid banding technique for human

5 Dutrillaux B, Laurent C, Couturier J, Lejeune J. Colora-
tion des chromosomes humains par l’acridine orange après
traitement par le 5 bromodeoxyuridine. C R Acad Sci (D)

6 Summit RL, Tipton RE, Wilroy RS Jr, Martens PR,
Phelman JP. X-autosome translocations: a review.
Birth Defects 1978;14(6C):219–47.

7 Bozekowski K, Mikkelson M. Fluorescence and auto-
radiographic studies in patients with Turner’s syndrome
and 46,XXp— and 46,XXq— karyotypes. J Med Genet

8 Fraccaro M. DNA replication and sex chromosome
function in the first stages of gametogenesis. Vortrag
Gehalten bei der Gesellschaft für Antropologie und

Daume E. Short arm deletion of an X chromosome,

10 Branković S, Lača Z, Dramušić V, Ivanović M, Morić-
Petrović S. A case of long arm deletion of the X chromo-
some in a patient with secondary amenorrhoea. Hum
Genet 1979;48:139–42.

11 Ruther U, Maschik S, Fredrich F, Breitencker G.
Partial long arm deletion of one X chromosome in a
patient with secondary amenorrhoea. Hum Genet 1979;

12 Theran E, Sarto GE, Patau K. Center for Barr body
condensation on the proximal part of the human Xq: a

13 Mattei GM, Mattei FJ, Vidal L, Giraud F. Structural
anomalies of the X chromosome and inactivation center.

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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,XXt(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. 1

Received for publication 16 November 1981
X long arm deletion with oligomenorrhoea.

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doi: 10.1136/jmg.19.4.305

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