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A rare case of Turner's Syndrome with 45,X/46,X,Dic (X) (qter→p11.1::p11.4→qter)

GÜVEN LÜLECI[1], ORHAN HOŞGÖR[2], SIBEL KARAÜZÜM[1] and MINE ÜNER[2]

[1]*Department of Medical Biology and Genetics,* [2]*Obstetrics and Gynecology, Medical Faculty, Akdeniz University, Antalya, Turkey*

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This paper presents a female patient with clinical features of Turner's Syndrome and the chromosomal constitution of 45,X/46,X,dic (X) (qter→p11.1::p11.4→qter). This abnormal X chromosome was compared with similar-structured dic(X).

Güven Lülecí, Department of Medical Biology and Genetics, Medical Faculty, Akdeniz University, Antalya, Turkey

Isodicentric X/X and dicentric X/X translocations have been discovered recently (VAN DEN BERGE and FRYNS 1973; DE LA CHAPELLE and STENSTRAND 1974; FUJITA et al. 1977; MATTEI et al. 1977; DAVIDENKOVA et al. 1978; LAĆA et al. 1979; OTTO et al. 1981). The morphology of these chromosomes, their origin, replication, and inactivation have been extensively studied (THERMAN and PÄTAU 1974; SILLESEN et al. 1976; MARASCHIO et al. 1977; MIRZAYANTS and BARANOVSKAYA 1978; CHEN et al. 1980; MUTCHINIK et al. 1981; FRYNS et al. 1983; RIVERA et al. 1984; THERMAN et al. 1986; SARTO et al. 1987). Structurally abnormal X chromosome appears to be functionally inactive, the phenotype of patients with structural rearrangements being close to the phenotype of patients with X monosomy. In the present study we report clinical and cytogenetic findings of a female patient with a dicentric X chromosome, in whom part of the cells are found to have bipartite Barr Bodies.

Case report

A woman aged 30, married for one year, was referred because of amenorrhea and infertility to the Department of Gynecology. Proband was the first child born to healthy and unrelated parents. Family history was negative with regard to congenital malformations, infertility, miscarriages, and mental disease. She was a teacher at the primary school. She had no menstruation till 16, after which hormonal therapy was initiated. She had regular menses until she stopped the therapy four years ago. She had irregular bleeding for the following two years and has amenorrhea since the last two years. Now she is under cyclic oestrogen-progesteron treatment and has normal menses. Physical examination revealed a height of 143 cm, weight of 51 kg, cubitus valgus, small breasts, female type pubic and axillary hair, and small uterus.

Laboratory findings: Blood, urine and other biochemical analyses were normal.

Hormonal findings: Serum; Thyroid-Stimulating Hormone 0.61 uIU/ml, Follicle-Stimulating Hormone 55.24 mIU/ml, Luteinizing Hormone 39.40 mIU/ml, Prolactin 230.89 mIU/ml, Dehydroepiandrosterone Sulfate 108.16 ng/ml, Estradiol 85.98 pg/ml, Progesterone 0.26 ng/ml.

Laparoscopy findings: Small uterus, normal fallopian tubes, bilateral streak gonads.

Radiological findings: Horse-shoe kidney.

Cytogenetic findings

Peripheral blood leukocytes for the routine cytogenetical studies were cultured in Mc Coy's 5A supplemented with 15 % fetal calf serum (MOORHEAD et al. 1960). The chromosomes were banded by GTG and CBG banding techniques (SEABRIGHT 1971; SUMNER 1972). 100 cells were counted from

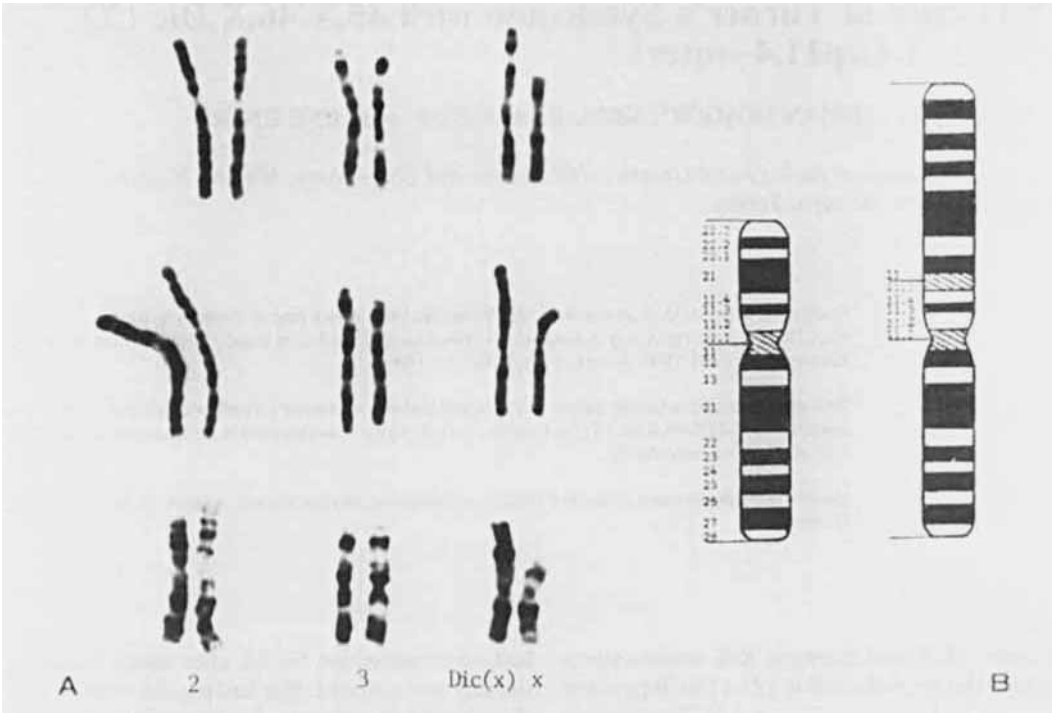


Fig. 1A and B. Partial karyotypes of the proband with GTG banding from different cells (**A**). Diagrammatic illustration: Normal X and Dic(X), (**B**).

chromosomal preparation of blood lymphocytes. There was only one X chromosome (45,X0) in 68 cells, 32 cells contained an aberrant X chromosome. This chromosome resembled a chromosome 2 in size and morphology. GTG banding pattern indicated that this abnormal chromosome consisted of a long and part of the short arm of one and a long arm of the other X chromosome, including a centromere (Fig. 1). CBG-banding showed that this chromosome was dicentric with centromeric areas of the same size (Fig. 2B). Her karyotype is 45,X/46,X,Dic (X) (qter→p11.1::p11.4→qter). RBG technique showed that the abnormal X chromosome was late replicating in all cells (Fig. 2A) (Dutrillaux and Viegas-Pequignot 1981). Sex chromatin bodies were observed as large or bipartite (Fig. 3) (Klinger and Ludwig 1957).

Comments

Gonadal dysgenesis, short stature and the other features of Turner's Syndrome are often seen in cases carrying a dicentric X chromosome formed by the fusion of the short arms (Sillesen et al. 1976; Mattei et al. 1977; Davidenkova et al. 1978; Laća et al. 1979; Otto et al. 1981; Rivera et al. 1984). But some reports have described cases in which the stature was within the normal range (Barnes et al. 1987). In our case short height and other characteristics of Turner's Syndrome, bipartite sex chromatin and dic(X)(qter→p11.1::p11.4→qter) karyotype were observed. In the translocation X chromosome, breakage points on the short arms often occur in the regions of p22, p21 and p11 (Sillesen et al. 1976; Fujita et al. 1977; Mattei et al. 1977; Davidenkova et al. 1978; Otto et al. 1981; Therman et al. 1986). In these cases with an abnormal chromosome, chromosome shape, size and clinical symptoms change according to the different breakage points. Since the inactive dicentric X chromosome is not stable, it is lost in some cells and 45,X0 cells are observed as mosaic. Reporting of different cases will make possible the classification of X chromosome translocations and the corresponding clinical entities.

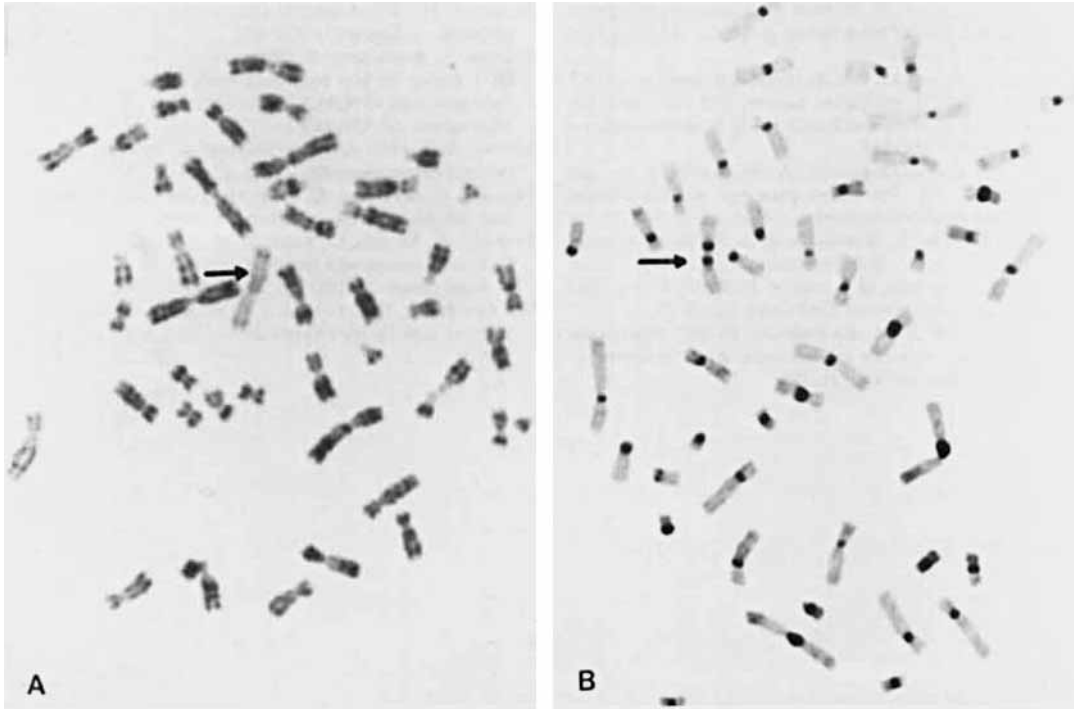


Fig. 2A and B. RBG banded cell showing the late replicating dic(X), (**A**). CBG banded cell showing the centromeres on dic(X), (**B**).



Fig. 3. Cells with large and bipartite Barr bodies from proband.

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