Cytological Findings of 10 Cases with i(Xq) and One with dic(X)(qter–cen–p22 : : p11–qter)

Hiroko Fujita¹,², Yoko Tanigawa², Yoko Yoshida¹, and Yoshiaki Okada³

¹ Department of Child Health, Science of Living Faculty, Osaka City University, Osaka
² Chromosome Laboratory, Hyogo Prefecture Tsukaguchi Hospital, Hyogo
³ Department of Medicine, Yodogawa Christian Hospital, Osaka

Summary. Cytological findings in 11 girls with a structurally abnormal X chromosome are presented. All patients were referred either as probable Turner's syndrome or as short stature. Of 10 patients with an i(Xq) chromosome, 6 had symmetrical G-bands in both arms and one C-heterochromatin, and 4 had an extra G-band in the proximal portion of one arm and double C-bands. The iso-X chromosome of the former was described as i(Xq) (qter–cen–qter) and that of the latter as idic(Xq)(qter–p11–qter). Karyotype of the one remaining was identified by G- and C-banding patterns as 46,X,dic(X)(qter–cen–p22 : : p11–qter).

Introduction

Several reports dealing with the structure of the iso-X chromosome have been presented. De la Chapelle and Stenstrand (1974) found that human i(Xq) chromosomes can be divided into at least two groups from C-banding findings. Priest et al. (1975) proposed seven types of i(Xq), including translocation of t(X;X). In the present study we also recognized two types of iso-X chromosome and one translocation of X-X in the 11 cases of short stature and amenorrhea.

Materials and Methods

The patients were referred as either possible Turner's syndrome or short stature to our cytogenetic clinic in Tshukaguchi Hospital from the endocrine clinic of Yodogawa Christian's Hospital.

Buccal smears for X-chromatin were stained with crecylecht violet and chromosome examination was made from blood lymphocytes. G-staining was performed with a slightly modified method of ASG (Sumner et al., 1971) and trypsin technique (Wang and Fedoroff, 1972). A flame-dried slide is incubated in 2 × SSC for 1 h at 60° and then rinsed briefly with water and 95% ethanol. After being air-dried, the slide soaks in 0.5% trypsin for 5 to 10 s at 0° and is stained with 5% Giemsa diluted by pH 6.8 phosphate buffer. C-staining was done according to Bobrow et al. (1972).
Results

Clinical findings in the patients are listed in Table 1. All patients are of short stature and delayed in secondary sexual development. Some of them have Turner's stigmata, including cubitus valgus, short neck, and shortened metatarsal bone. They complain of congenital anomalies such as strabismus, dyschromatopsia, heart disease, and hip joint dislocation, respectively. Details of their clinical and laboratory findings will be presented elsewhere.

Parental ages at birth of the patients are listed in Table 1. Mean age is 28.6 for the mothers, and 34.1 for the fathers (28.7 and 32.1 respectively for the general population in Annual Vital Statistics of 1955). There is no indication of increased maternal age and although there is a slight increase in paternal age, these samples are too small to draw a conclusion.

Cytogenetic data are summarized in Table 2 and in Figures 1 and 2. Their X-chromatins were positive and larger than normal. Of the 11 patients, 5 had a mosaic karyotype accompanied by a 45,X cell line. In cells having 46 chromosomes, one of the C-group chromosomes was replaced by a No. 2-like or No. 3-like chromosome. In order to identify these abnormal karyotypes, we used both G- and C-banding techniques.

According to findings of the G- and C-banded cells, the abnormal X chromosomes in the 11 cases can be divided into at least three types:

1. Iso-X chromosome having a symmetric G-banding in both arms and a single C-band at the centromere (Fig. 1). Included in this group are five patients with a modal cell line and one mosaic. Their iso-X chromosome was metacentric and resembled a No. 3 chromosome on conventionally stained cells. With G-banding, this chromosome was identified as an iso-X chromosome, consisting of one centromere and two complete long arms. With C-banding, we found single heterochromatin in various sizes in each case at the centromere. This iso-X chromosome was described as i(Xq)(qter-cen-qter).

2. Iso-X chromosome having an extra G-band on the proximal portion of the longer arm and double C-bands (Fig. 2, 7—10). Included in this group are three mosaics and one patient with a modal cell line. The iso-X chromosome of the mosaics was relatively metacentric but one arm was a little longer than other (Fig. 2, 7—9). In C-banded cells, one C-band with centromeric constriction was close to another C-band without constriction. In G-banded cells, an extra G-band appeared in the same position as the C-band without constriction. We believe that this extra G-band corresponds to the C-band. The iso-X chromosome of the patient with the modal cell line was distinctly submetacentric and a secondary constriction appeared on the longer arm of conventionally stained cells. With the G- and C-banding, an extra G-band or bipartite heterochromatin appeared in the position of the secondary constriction. The iso-X chromosome must consist of a double chromatid from qter to p11 and should be described as idic(Xq) (qter-p11-qter).

3. Abnormal chromosome consisting of an extremely asymmetric G-banding pattern on both arms and two C-bands (Fig. 2, 11). This included only one mosaic. Her abnormal X chromosome was submetacentric and longer than a