120. An Additional Case of Partial Trisomy 22 with Multiple Clinical Malformations

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Even with banding techniques, an extra small acrocentric element constituting partial trisomy of either no. 13 or no. 22 is generally difficult to be conclusively identified. Coloboma and microphthalmia have not been detected in patients with trisomy 22, while such eye defects are known to be usually associated with trisomy 13 (Smith 1970; de Grouchy and Turleau 1977).

We wish to describe in this paper an additional case with an extra small acrocentric chromosome in which we failed to define coloboma, microphthalmia and anal atresia.

Case reports. The propositus was a five-year and eight-month-old female child, 14.5 kg in weight, with circumferences of 48.2 cm in head and 54.0 cm in breast, and 105 cm in length. She was born to 30-year-old mother and 41-year-old father as a third children. There was no history of abortion, stillbirth, congenital malformations, exposure to atomic bomb, and consanguinity in this couple. Clinical examinations revealed that her parents, and elder brother and sister were phenotypically normal without evidence for mental deficiency. The birth weight of this patient was 2650 g at 40 weeks of gestation.

Remarkable clinical signs of this child were mental retardation, muscular hypotonia, frontal bossing, low posterior hair line, mongoloid slant of eyes, epicanthus, inophthalmos, blindness, short and beaked nose, long and convex upper lip, deep philtrum, microretrognathia, long, posteriorly rotated and low-set ear, periauricular skin tag and pit on the right ear, suspiciously auditory disturbance, high-arched and narrow palate, incomplete bifid uvula, bilateral simian crease with proximally implanted thumb, slender feet with long toes, and congenital right hip dislocation. The most significant was cardiac malformations, such as pulmonary stenosis, interventricular septal defect, and right ventricular enlargement. No signs for coloboma, microphthalmia, atresia ani and abnormalities of external genitalia.

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were observed in this patient (Fig. 1).

![Fig. 1. Phenotypic profiles of the proband. 1: The face showing mongoloid slant of eyes, epicanthus, inophthalmos, short and beaked nose, long and convex upper lip, deep philtrum, and micromegadactyly. 2: Large ears, posteriorly rotated and low-set. Preauricular skin tag and pit are noted on the right ear. 3: A thumb, proximally implanted. A finger remarkable by hyperlaxity of articulation. 4: Slender feet with long toes.](image)

Cytological findings. Chromosome preparations were made from leucocyte cultures established in the propositus and her parents. As differential staining, G-banding procedure (Seabright 1971) was applied to air-dried slides. Chromosome counts were done on 50 well-delineated metaphases in each sample. Karyotypes were analysed on 20 cells based on conventional Giemsa and G-banding preparations.

Karyotypes of the proband prepared from conventional Giemsa- and differential staining specimens showed that the number of chromosomes was 47 by the presence of an additional element of small size, and that the extra chromosome was identified as being due to the partial deletion of the long arm of no. 2 (Fig. 2). Her mother and father were chromosomally normal without slight evidence for anomalous phenotype (Figs. 3–4).

Remarks and conclusion. The literature refers to several infor-
Fig. 2. G-banding karyotype of +22q- patient.

Fig. 3. G-banding karyotype of the mother of the proband.
mation dealing with complete and incomplete cat-eye syndromes, and
the partial trisomy 22 syndrome with an extra small acrocentric chromosone (Zellweger et al. 1975; Zellweger et al. 1976; Hsu and
Hirschhorn 1977; Bofinger and Soukup 1977).

The major clinical features of the complete cat-eye syndrome are
generally shown by the combination of coloboma and anal atresia. In
the incomplete cat-eye syndrome, only one of the two major abnormal-
ities, i.e., either coloboma or anal atresia, was observable. Except
for the eye anomalies such as coloboma and microphthalmia, the partial
trisomy 22 do share several clinical features which are preauricular
skin tag or sinus, antimongoloid slant of eyes, and skeletal abnor-
malities and so on. It is well known that coloboma and microphthalmia
are associated with trisomy 13, but not with a partial trisomy 22
(Smith 1970; de Grouchy and Turleau 1977).

One of the reasons why the status of trisomy 22 and cat-eye
syndrome remains complex is that in many cases the extra small
acrocentric chromosome is not conclusively identified even with
banding techniques.

Obviously the present case lacked signs of coloboma, micro-
phthalmia and anal atresia. The clinical feature of this patient was
slightly mild in comparison with that of the complete trisomy 22
(Kadotani et al. 1978). Similar picture was found to occur in a
reported case with the partial trisomy 22 by Kadotani et al. (1978).
In the light of this clinical findings it is very probable that the extra
acrocentric chromosome is not a partially trisomic 13 element, but corresponds to a chromosome 22 with partial deletion of its long arm.

Summary. A female infant with multiple malformations is described. She has 47,XX,+22q-(q13) karyotype based on G-banding analysis. Her mother and father are chromosomally and clinically normal.

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References