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Down syndrome with XO/XX mosaicism

By

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A 2-month-old girl with Down syndrome was found to have 47, XX, +21/46, X, +21 mosaicism. No symptoms indicative of infantile Turner syndrome were observed.

The proposita was the first child of a 17-year-old woman and her 19-year-old husband. The family history was negative, physical and dermatoglyphic examination of the parents revealed nothing unsual. Birthweight was 2600 g, length 48 cm. At 2 months of age the infant's appearance was characteristic of Down syndrome (Fig. 1).

She had a flat face and occiput, bilateral epicanthic folds, upward obliquity of the palpebral fissures, Brushfield-spots on the iris, small ears, big tongue, extra posterior cervical skin, muscular hypotonia. No pterygium, low posterior hairline or oedema of the hands and feet could be observed.

Dermatoglyphics showed, a right

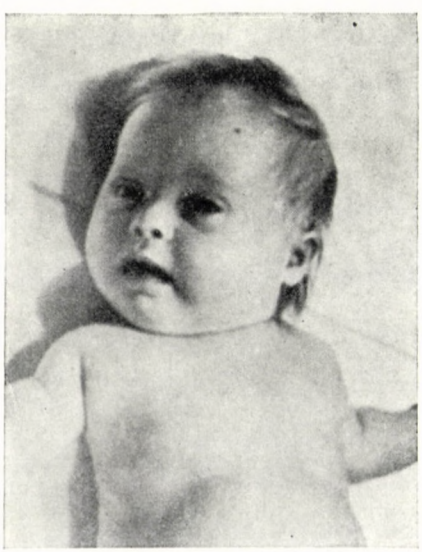

FIG. 1. Patient showing Down syndrome phenotype

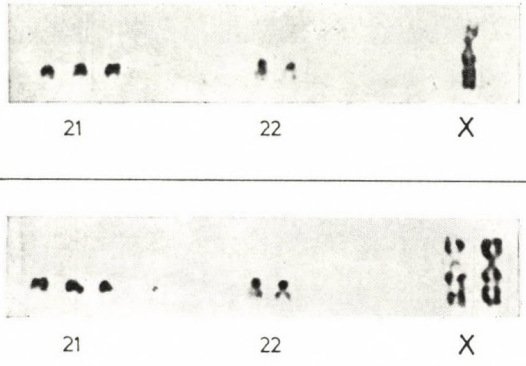

FIG. 2. Giemsa banded G and X chromosomes demonstrating XO, +21 and XX, +21 cell lines

hand with 5 digital ulnar loops and Sidney-line of the palm; left hand with a t' axial triradius and 5 digital ulnar loops. Total ridge count, 142.

Biochemical data were within normal limits, including serum thyroxin, TSH and amino acid concentrations. An electrocardiogram showed normal curves. No sign of a heart malformation could be detected.

CHROMOSOME STUDIES

Of a total of 50 cultured lymphocytes analysed from two blood samples, 34 contained 47, and 16 only 46 chromosomes. In Giemsa banded preparations trisomy 21 was present in each of the cells, and one X was missing in the mitoses with 46 chromosomes (Fig. 2). Thus, the karyotype of the patient was 47, XX, +21/46, X, +21.

Buccal smear X-chromatin, 36% positive.

Drumstick, 15/1000 positive.

No fibroblast cultures were done.

Karyotype and sex chromatin pattern of the parents and of the paternal grandparents proved to be normal.

DISCUSSION

Double aneuploidy, with or without mosaicism, has been reported in many instances. At the same time, combination of mongolism with Turner syndrome seems to be very rare. The number of cytogenetically proved Down—Turner patients does not exceed ten, including the cases reviewed by Villaverde and Da Silva [5], and Townes et al. [4], and the present observation. All the patients mentioned had mosaic karyotypes. In addition, X chromosome structural aberration associated with trisomy 21 was described in a few subjects [2, 3].

As in the previously observed patients, in our case too, the features of Down syndrome dominated the clinical picture; the additional XO cell line was discovered by chance.

As in similar cases, investigation of the parents and grandparents gave no hints at increased non-disjunction in this family. Thus, for counselling, in agreement with Jenkins et al. [1], we think that for practical purposes the recurrence risk is the same as that for primary trisomy 21.

References

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