A Structurally Abnormal Chromosome (46,XX,?17p+) Associated with Mental Deficiency and Congenital Malformations

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A chromosome anomaly, consisting of the absence of a chromosome in the 17-18 group, which was substituted by an abnormal medium-sized one, with median centromere, was observed in a female patient who had been selected for cytogenetic investigations because of mental retardation and congenital malformations.

Case Report

The patient, a girl, the only child of normal non-consanguineous parents, born on February 19, 1951, in Biella (Italy), is an inmate of an institution for mentally handicapped patients.

At conception the mother was 33 and the father 31 and neither had been exposed to environmental or radiation hazards. The patient was delivered after a normal pregnancy, but her weight at birth was only 1700 g. She walked at 2 years and began to speak simple words at 2½. When she was 12, her IQ was 39 (Binet-Simond scale). Menarche began at 15 years, with scanty bleeding. At this age the following data were recorded: height 138 cm.; weight 35.5 kg.; arm span 130 cm.; chest circumference 68 cm.; head circumference 52 cm.

She presented with peculiar facies (Fig. 1), depressed nose bridge, blepharochalasis, open mouth, protruding lower lip, high arched palate, irregular teeth, hypertrophic and easily bleeding gums, low-set ears, masculine implantation of the hairs on the neck, moderate joint hyperflexibility, short fifth fingers, bilateral genu valgum. The breasts and the external genitalia were normally developed. Routine haematological data were within the normal range. The leucocyte alkaline phosphatase was slightly raised, with a score of 135 (normal range 30–100).

Blood Types.

Patient B, CCDee, M+ N- P- S- K+ k+ Fy-
Mother O, CCDee, M+ N+ P+ S- K- k+ Fy-
Father B, CCDee, M+ N- P- S- K+ k+ Fy+

Dermatoglyphs (Fig. 2). The total finger ridge count was only 30. Both palms presented a slightly raised axial triradius (r'); the atd angle was 36° right, and...
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32° left; the a-b ridge count was 45 right, and 40 left. On the right palm a vestige was present in the third interdigital area, and was associated with absence of the c triradius and displacement of the b triradius. On the left palm the c triradius was absent and the d triradius was displaced into the fourth interdigital area. Flexion creases were normal.

Cytogenetic Investigations. 24 Barr bodies were found in 100 oral mucosa nuclei, and 6 drumsticks were found in 2000 granulocytes. Chromosomal investigations on peripheral blood cultures (see Table) revealed the constant absence of a 17-18 chromosome which was substituted by a medium-sized one with symmetrical arms (Fig. 3). A comparison of the short arms of group mid was added for the last 90 minutes. Slides were coated with Kodak AR 10 and exposed for 7 days. The study of the autoradiographs revealed the normal presence of a late-replicating C group chromosome (‘hot’ X), while the abnormal chromosome was either unlabelled or slightly and often asymmetrically labelled (Fig. 5).

Discussion

The clinical pattern presented by our patient does not correspond to any of the presently known chromosomal syndromes: it presumably results from the duplication of some chromosome material. Indeed, the abnormal chromosome could have

| TABLE |
| CHROMOSOME COUNTS (IN PARENTHESES ANALYSED METAPHASES) |
|<45 | 45 | 46 | 47 | Total | Remarks |
| Patient: | | | | | |
| Culture 1 | 2 | 4(4)* | 25(9) | — | 31 | Abnormal 17p+ |
| Culture 2 (with 3H-TdR) | 5 | 5(5)* | 20(16) | — | 30 | Normal |
| Mother | 3 | 2(2)* | 22(3) | — | 27 | Normal |
| Father | 2 | 2(2)* | 17(5) | — | 21 | Normal |

* Random losses.

E chromosomes from several metaphase plates (Fig. 4) did not allow us to decide with certainty whether the abnormal chromosome derived from a 17 or from an 18.

The parents had apparently normal chromosome sets.

Autoradiography. Four hours before harvesting, tritiated thymidine (Amersham Radiochemical Centre, specific activity 14.8 Ci/mM) was added at a concentration of 0.4 µCi/ml. to a peripheral blood culture. Colce-
The presence is reported of a structural karyotypic abnormality, consisting of a probable translocation on the short arm of a 17-18 chromosome, in a girl with severe mental retardation and slight congenital malformations.

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**References**


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FIG. 5. Autoradiographic image of a metaphase; the arrows point to the 'hot' X and to the abnormal chromosome, identified on photograph taken before autoradiography (see 6th row of Fig. 4b).