$t(2q-; Dq+)$ in a Mentally Retarded Female Child

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This paper reports the clinical, biochemical, and psychological findings in a 3-year-old mentally retarded girl who was found to have a chromosomal abnormality.

Family History

The child is the younger of two sibs born to non-consanguineous parents. The father and mother were aged 35 and 34, respectively, at her birth, and are of average height (father is 173 cm. and mother is 168 cm.). The height and weight of the patient's 14-year-old sister have been within normal limits. There was no family history of mental retardation or epilepsy.

Case History

The patient was born at term weighing 3.4 kg. after a normal delivery, but the last four weeks of the pregnancy were complicated by mild pre-eclamptic toxæmia. She thrived and developed normally until 8 months, when she was admitted to hospital with pyrexia and inability to move her left leg. Osteomyelitis of the left femur was diagnosed, the lesion resolved satisfactorily with antibiotics, and there was no residual disability 6 months later.

At 1 year her parents suspected that she was retarded and she did not begin to walk until 17 months. Though she had frequent respiratory infections, none necessitated hospital treatment. There was no history of convulsions. At the age of 3 she was still not able to talk, put everything in her mouth, was not toilet trained, and so she was referred for psychiatric assessment.

Physical examination. At age 3 years and 8 months she was 106.5 cm. tall (above 97th percentile), had an arm span of 103 cm., and a head circumference of 50 cm. She weighed 19.6 kg. (between 90–97th percentile). This child's greater than average development has been a gradual process as supported by her weight chart. There was no evidence of any congenital abnormality.

Investigations. Psychological assessments at 3 years and 9 months: on the Merrill Palmer scale, mental age was 23 months; on the Vineland social maturity scale, social age was 28 months. EEG examination showed that no single rhythm was dominant and the over-all voltage was within the low-medium category.

X-ray of the skull: no lesion of the pituitary fossa detected.

Urinalysis: normal amino acid chromatogram.

Levels of fasting blood glucose, plasma proteins, transaminase, cholesterol, calcium, magnesium, electrolytes, acid and alkaline phosphatase, thymol turbidity, and zinc sulphate were all within normal limits.

Palm prints. Radial loops on both index fingers and ulnar loops on the remaining digits yielded a low total finger ridge count of 96 (normal female 127). The total *atd* angle was 96° and the combined a-b ridge count was 68.

Cytogenetic studies. Sex chromatin was present in epithelial cells from buccal smear preparations. Chromosome counts of metaphases from leucocytes of two peripheral blood cultures and fibroblasts from one skin culture revealed a modal number of 46 but there was one abnormal long acrocentric chromosome, one normal No. 2 chromosome, and an extra chromosome similar to a No. 3 chromosome (Fig.). This karyotype is most simply interpreted as an apparently balanced translocation $t(2q-; Dq+)$. Both parents had normal karyotypes.

Discussion

The chromosomal findings in the patient could have resulted from: (i) insertion of part of the long arm of a No. 2 chromosome into the long arm of a D group chromosome; (ii) translocation of part of a No. 2 chromosome onto a D group chromosome; or (iii) there could have been reciprocal translocation of a major part of the long arm of a No. 2 and a minor part of a D group chromosome.

There have been two other reports of translocation involving the long arms of a No. 2 and the long arms of a D group chromosome. Ricci, Dallapiccola, and Cotti (1968) reported finding an abnormal D chromosome ($Dq+$) in a child with

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FIG. Karyotype of the patient showing the apparently balanced 2/D translocation.

microcephaly, micrognathia, and hypertelorism, while her mother was a balanced translocation carrier $t(2q-;Dq+)$. They suggested that the patient had partial trisomy 2, or partial trisomy 2 with associated partial monosomy D. The patient reported by Lisco and Lisco (1967) had a chromosome complement similar to that of the present case ($2q-;Dq+$), but apart from infertility no other abnormality was detected and she was thought to be a balanced translocation heterozygote.

The mental retardation in this child who had no other abnormality at 8 months is probably due to her abnormal chromosomal findings, but the remote possibility of brain damage at 8 months due to pyrexia cannot be excluded. If the former is accepted, then it must be postulated that the struc-

tural rearrangement of the chromosomes has resulted in loss of genetic material too small to be detected by present techniques.

Summary

The clinical, biochemical, and psychological findings in a 3-year-old mentally retarded girl with a chromosomal abnormality are described.

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