Partial trisomy 8 (trisomy 8q2106→8qter)

SUMMARY A case of trisomy for part of the long arm of chromosome 8, confirmed by G-banding analysis, in a white male infant is described. The mother carried a reciprocal translocation between chromosome 8 and chromosome 13 (46,XX,t(8;13)(q21;q34). The patient had inherited the translocated chromosome 13 and was thus trisomic for the distal half of the long arm of chromosome 8. He had many of the clinical features of the full trisomy 8 syndrome. As compared with previously reported cases with trisomy of the distal end of chromosome 8, he was more dysmorphic and showed greater developmental retardation.

In 1974, Sanchez and Yunis reported 2 sibs with partial trisomy 8 and suggested that the distal end of the long arm of chromosome 8 might be responsible for the bulk of the trisomy 8 phenotype. We wish to report a patient with partial trisomy of a slightly longer segment of the long arm of chromosome 8 (8q2106→8qter), who also had distinct phenotypic abnormalities. The findings tend to confirm this hypothesis.

Case report

A.A. was born at term at Women and Infants Hospital of Providence, Rhode Island, on 21 July 1975 to an 18-year-old, unmarried primigravida, who took diazepam, 5 to 10 mg daily, throughout gestation because of anxiety. She smoked marijuana daily but denied other drug usage. Presentation was frank breech, Apgar scores 7 and 9, and birthweight 2580 g. Oxygen was given for 2 minutes because of mild cyanosis. Physical examination disclosed a dysmorphic male infant, with multiple anomalies (Fig. 1). The skin appeared thickened and was tight and shiny over the extremities. Head circumference was 33 cm with an anterior fontanelle of 4 × 3 cm. The cranial vault had a square-shaped, asymmetrical appearance, with flattening of the top and a slightly prominent occiput. There was apparent ocular hypertelorism, exotropia, and a double skin crease below the lower eyelid. The pinnae were small with mild posterior angulation. The nose was pugged and the philtrum long. The upper lip was partially everted, revealing a thick midline frenulum connecting to a notched upper alveolar ridge. There was moderately severe micrognathia and a short neck with redundant skin folds posteriorly. The nipples were normally spaced but small. A grade 2/6 systolic murmur was noted at the upper left sternal border. The pelvis appeared narrow. The scrotum was normally rugated, but hypoplastic, without palpable testes. Both patellae were present. Slight resistance to flexion and extension of the proximal interphalangeal joints of the 4th and 5th digits was present, as well as mild clinodactyly of both 5th fingers. Both thumbs were short and curved inward. The toes were crowded, with deviation of both large toes inward and under the second toe. The plantar areas had deep, V-shaped fissures (Fig. 2). Neurological examination disclosed a generalised increase in muscle tone and an intermittent coarse tremor of the upper extremities. He was unable to suck effectively and required gavage feedings. Chest x-ray film revealed short blunt ribs, and a cardiac silhouette at the upper limit of normal. At 2 days of age, he had a generalised tonic seizure lasting a few seconds. He was placed on phenobarbitone and no further seizures occurred.

Because his mother was unable to care for him, he was transferred to a chronic care facility. At the age of 11 months, his growth and development were considered to be poor, in spite of an intensive stimu-
Fig. 2  Left foot of propositus showing deviation of large toe and deep fissures in plantar area.

Fig. 3  Karyotype of mother of propositus: 46,XX,t(8;13),(q21;q34). Trypsin banding and Giemsa stain.

Case reports

CYTOGENETIC STUDIES

Chromosome preparations were made on the patient and his mother from peripheral blood lymphocyte cultures. G-banded metaphase spreads were prepared by treatment of the slides with trypsin solution and staining with Giemsa. The mother was found to have an apparently balanced reciprocal translocation involving the distal portion of the long arm of chromosome 8 and the telomeric portion of chromosome 13, with break points at 8q2106 and 13qter. Her karyotype was thus 46,XX,t(8;13),(q21;q34) (Fig. 3). Her parents (ages 40 and 37 when she was born) were found to be cytogenetically normal; her translocation was probably a de novo event. Studies to confirm paternity were not carried out. The propositus had the translocated chromosome 13 in an unbalanced form, and was thus trisomic for the distal half of the long arm of chromosome 8 (Fig. 4). According to the revised Paris Conference nomen-
clature (1975), his karyotype was 46,XY,der13,t(8;13)(q21;q34)mat.

Discussion

This patient had many of the clinical features of the full trisomy 8 syndrome (American Journal of Diseases of Children, 1975; Cassidy et al., 1975; Fineman et al., 1975), including asymmetrical head shape, micrognathia, slender trunk, cryptorchidism, incurved toes, thickened skin with deep furrows, hypertonia, and mild restriction of joint mobility.

Trisomy of the distal end of the long arm of chromosome 8 was originally described in 3 patients by Lejeune and Rethoré (1973). These patients were only mildly dysmorphic and had I.Q.s of about 70. The patients described by Sanchez and Yunis are more severely affected, even though the chromosome abnormality appears similar to that in the cases of Lejeune and Rethoré. In all 5 of these previously reported patients, who belonged to 3 different families, the translocation involved chromosome 8 and 22. Though Lejeune did not report break points (and used a different banding technique), the amount of chromatin material from chromosome 8 present in the extra G-like chromosome appeared to be similar in both reports. Our patient was also more seriously affected than those of Lejeune and Rethoré, both in the severity of his physical abnormalities and in the pronounced degree of retardation he showed, and more closely resembled the patients of Sanchez and Yunis (Table). The chromosome abnormality in our case, however, involved translocation of a larger

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<td>Clinical findings</td>
<td>Lejeune and Rethoré</td>
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<tr>
<td>Mental retardation</td>
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<tr>
<td>Thickened skin</td>
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<td>Dysmorphic facies</td>
<td>Mild</td>
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<td>Short neck</td>
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<td>Lowset or dysplastic ears</td>
<td>+</td>
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<tr>
<td>Micrognathia</td>
<td>−</td>
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<tr>
<td>Slender trunk or pelvis</td>
<td>+</td>
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<tr>
<td>Restricted articular function</td>
<td>−</td>
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<tr>
<td>Fifth finger camptodactyly</td>
<td>+</td>
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<tr>
<td>Large, incurved toes</td>
<td>+</td>
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<tr>
<td>Cryptorchidism</td>
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| Break points | Not stated | 8q24 | 8q21 |
segment of the distal end of chromosome 8 to chromosome 13, and it might be that deletion or inactivation of a small portion of the telomeric end of chromosome 13 could contribute to the more severe form of physical and developmental abnormality. It is also possible that drug ingestion during pregnancy might have exerted an additive effect.

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References


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Absence of distal interphalangeal fold causing difficulty in extending fingers

SUMMARY A 13-year-old girl sought medical advice, saying that for two years it had been increasingly difficult for her to extend her little finger. An examination revealed that all her fingers, with the exception of her thumbs, had no interphalangeal fold. Her mother had less pronounced signs of the same type. This abnormality seemed to be the result of an autosomal gene with dominant transmission.

Fig. 1 Absence of distal interphalangeal fold.

This 13-year-old girl had been playing the piano for 5 years. For a few months it had been more and more difficult for her to extend some of her fingers. When she was at rest, her little finger remained in a state of continuous flexion; it was no longer flush with the other fingers and could not be extended completely. At first it occurred to the physician that she was afflicted with Dupuytren's disease but the girl's age and the absence of other signs did not support such a diagnosis.

On examination, it was found that all her fingers, with the exception of her thumbs, had no interphalangeal fold. Her mother, who could move her fingers perfectly well, exhibited less pronounced signs of the same abnormality. No information
Partial trisomy 8 (trisomy 8q2106 leads to 8qter).

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