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Requests for reprints to Dr Barbara F Crandall, Neuropsychiatric Institute, 760 Westwood Plaza, Los Angeles, California 90024, USA.

A severely retarded 18-year-old boy with tertiary partial trisomy 14

SUMMARY An 18-year-old severely mentally and physically retarded boy was found to have an unbalanced chromosome complement 47,XY,+14q-. He had the characteristic facial dysmorphism, abnormal hands, and other features described previously in cases of partial trisomy 14, but appears to be the oldest case reported. His mother is a reciprocal translocation carrier, and lack of other carriers in the family is noteworthy.

Subjects with partial trisomy 14 have similar phenotypic abnormalities associated with markedly delayed motor and mental development.¹⁻³ In the majority, a balanced translocation was present in the mothers. This report outlines another case of partial trisomy 14, in an 18-year-old boy, where the mother was a balanced translocation carrier.

Case report

The proband was the third child born to healthy unrelated parents when the mother was 39 and the father 38 years of age. His sibs were aged 9 and 7.5 years at this time and there had been one spontaneous first trimester abortion. He was born at term after a normal delivery, birthweight 2.33 kg. There were respiratory and feeding difficulties with failure to thrive as an infant. Feeding problems have remained to the present time. All milestones were delayed and he is now severely retarded. Fits began at 17 months of age and he has been on continuous anticonvulsant medication since that time.

When seen here at 18 years of age, his height was 114.5 cm, weight 22 kg (on the 50th centile for a 5 to 7 year-old), head circumference 49 cm (on the 50th centile for a 2-year-old) (fig 1). He had a slightly asymmetrical skull with the left side more prominent, a low hair line on the forehead and two abnormal vortices of hair posteriorly, low set on each side of the neck. He had a short neck, sloping shoulders, and a slightly asymmetrical chest. The face was asymmetrical with a dimple on the left side of the chin, a flat nasal bridge, deep set small eyes, small slit-like normally orientated palpebral fissures, and flat supraorbital ridges. The nose was large and bulbous, and there was a small thin carp-like mouth with retrognathia. His hands showed tapering fingers, thumbs anteriorly placed and low set, a simian crease variant on the left, and all fingers and the thumb were hyperextensible. Both feet showed pes cavus and talipes equinovarus and all the toes were hyperextensible. The abdomen was protuberant. Both testes were descended and small and there was no sign of puberty. There was a sacral dimple.

Investigations included routine serum haematological and biochemical parameters and urinary studies, which were normal. Ophthalmic examination under anaesthesia showed hypermetropia and astigmatism, and a suspicion of early keratoconus, with irregular cornea. An electroencephalogram was abnormal.

CYTOGENETIC ANALYSIS

Chromosome analysis of standard peripheral blood cultures⁴ gave a modal number of 47 chromosomes in each of 62 cells, the extra chromosome being morphologically indistinguishable from a normal G group chromosome. G banding⁵ showed this extra fragment to be a deleted D group chromosome. All other chromosomes in the complement were normal in length and structure. Dermatoglyphic analysis showed absence of *d* triradius bilaterally,

FIG 1 Patient aged 18. The mouth and nose are characteristic even at this age. Note also the extreme growth retardation and lack of pubertal development.

axial t'' on the left palm, and seven ulnar loops with three arches on the fingers (total count 62).

FAMILY STUDIES

The father, mother, two older sibs, the maternal grandmother, the three maternal uncles, and a maternal aunt were phenotypically normal and healthy adults and chromosome analysis was normal in each case except the mother. In the mother, chromosome analysis of 40 cells, including G, C, and R banding,⁶ gave a modal number of 46 chromosomes, with a deleted No 14 (compatible with that seen in the proband) and extra chromosome material on the end of chromosome 3 (fig 2). The mother was the carrier of a reciprocal translocation, karyotype 46,XX,t(3;14)(p25;q21) and hence the karyotype of the proband was 47,XY,+der(14),t(3;14)(p25;q21)mat.

Discussion

Since the first report of proximal partial trisomy 14 in a mentally retarded patient,⁸ seven patients with partial trisomy 14 have been described in separate reports, reviewed by de Grouchy and Turleau,⁹ and a further four cases from one family were recently reported by Miller *et al*,³ who also reviewed the previous cases. It is apparent from these reports that cases of proximal partial trisomy 14 show mental and motor retardation with similar facial and other

FIG 2 Partial G, C, and R banded karyotypes from (A) the proband, showing D and G group chromosomes and (B) his mother, showing A and D group chromosomes. C banding⁷ showed normal Y heterochromatin.

phenotypic abnormalities. The most characteristic features are microcephaly, a thin downturned mouth, prominent nose, micrognathia, small deep set eyes, short neck, simian crease, anomalies of the hands and feet, with generally normal ears and genitalia. Also commonly present are a low birthweight at term and the later development of epilepsy with an abnormal electroencephalogram. All these features were present in our patient, who appears to be the oldest recorded case. An adolescent girl was reported by Muldal *et al*¹⁰ and she did have early puberty.

The chromosome complement seen in this patient arose at maternal oogenesis by 3:1 disjunction at the first meiotic division, the segregation resulting in the normal chromosomes 3 and 14 and the 14q- fragment moving to the same pole at anaphase I. At fertilisation, the father's normal chromosomes 3 and 14 made his complement 47,XY,+14q-. The error leading to this chromosome complement could not have arisen in any other way. However, we do not know if the translocation arose in the mother *de novo*, or was inherited from her father (who was dead). It would seem likely that the translocation arose *de novo* in the mother as none of her sibs carried it, which would be unusual if the father was a carrier. Other families reported in which an unbalanced proband had arisen by 3:1 disjunction from a carrier mother showed many carriers in the sibships of both the proband and the carrier mother. For example, Simpson and Zellweger² found five carriers and only one normal sib of the mother of their proband, whose sib was also a carrier, and in their case the translocation was also onto chromosome 3. In our family, the abnormality will have 'died out' after one generation.

In their very extensive study of unbalanced offspring derived from 3:1 meiotic disjunction in reciprocal translocation carriers, Lindenbaum and Bobrow¹¹ showed that there were certain characteristic features in these cases. There was no evidence of infertility among the carriers, there was a high incidence of spontaneous abortions (38% overall), which was higher in these 3:1 cases than in the offspring of reciprocal translocation carriers in general, there was no advanced maternal age effect, and frequent involvement of the acrocentric chromosomes was noted. The family described here shows all these features, except that a maternal age effect was evident.

We wish to thank Dr Ong and Mr E Ozzals for referring the patient, Eunice Simpson for the dermatoglyphic data, Robin Murray for photographic assistance, and the parents for their co-operation.

ARABELLA SMITH, GESINA DEN DULK, AND
GAE ELLIOTT

Cytogenetics Unit, Oliver Latham Laboratory,
Health Commission of New South Wales,
North Ryde, Australia

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Requests for reprints to Dr A Smith, Cytogenetics Unit, Oliver Latham Laboratory, Health Commission of NSW, PO Box 53, North Ryde, NSW 2113, Australia.

Partial trisomy of the short arm of chromosome 8 resulting from balanced maternal translocation

SUMMARY Extra chromosomal material on the long arm of chromosome 15 was found in an infant with growth retardation, dysmorphic features, skeletal abnormalities, and congenital heart defect. The phenotypically normal mother had a balanced translocation between the short