A severely retarded male with deletion of chromosomes 15 (pter→q13) and 10 (q26→qter)

The proband (fig 1), aged 15 years, was the second child (of three) born to non-consanguineous healthy parents, when the mother was 31 and the father 29. His height, weight, and head circumference were all below the 3rd centile, there were no signs of puberty, and the testes were undescended. He had an ataxic wide based gait, hyperreflexia, increased and spastic tone, a high bossed forehead, short nose, flattened nares, a very high palate, continually open mouth, hypoplastic maxillae, and small mandible. A convergent strabismus had previously been operated upon. Convulsions began at the age of 14 years and a recent EEG was abnormal.

Cytogenetic studies on peripheral blood revealed the karyotype 45,XY,-10,-15,+t(10;15)(q26;q13) (fig 2). There was no evidence of mosaicism with any other cell line. A fragment consisting of the short arm region, centromere, and proximal long arm of chromosome 15 plus the terminal band of 10q was not detected in any cell examined from three peripheral blood cultures. There was no NOR staining in the abnormal derivative chromosome which was also monocentric on C banding. Parental chromosomes were normal.

The most likely explanation for this boy's karyotype is that a de novo balanced reciprocal translocation had occurred resulting in the large abnormal chromosome and a small fragment, which could have been lost in an early stage of cell division, or could still be present in a small circumscribed area of a tissue other than peripheral

FIG 1 Patient aged 15.

FIG 2 Partial karyotypes from two G banded cells. Arrow points to 10;15 translocation chromosome.

blood. Thus, the mental retardation and phenotype of the patient could result from the same mechanisms considered possible for other retarded de novo balanced reciprocal translocation carriers.¹ The abnormalities in this patient could also result from 10q loss, even though this monosomy is only of the small terminal band. We could not find any other cases of monosomy 10q.²

Finally, the abnormalities could be related to monosomy of chromosome 15, involving the short arm, centromere, and proximal long arm to band q13. There has been one similar case described of 15q monosomy, with loss of 15pter→q14,³ resulting from a balanced parental translocation. This patient at 3 years showed severe motor and mental delay, hypotonia at birth, seizures, small nose, narrow palate, and narrow dolichocephalic head, all features found in our patient. However, she had marked skeletal abnormalities not found in our patient. More cases are needed before a proximal 15q— syndrome can be delineated.

We wish to thank Dr R Balandin, Stockton Hospital, for referring the patient.

ARABELLA SMITH and GESINA DEN DULK
Cytogenetics Unit, Oliver Latham Laboratory,
Health Commission of NSW, Australia

References

- 1 Funderburk SJ, Spence MA, Sparkes RS. Mental retardation associated with "balanced" chromosome rearrangements. *Am J Hum Genet* 1977;29:136-41.
- 2 de Grouchy J, Turleau C. *Clinical atlas of human chromosomes*. New York: Wiley, 1977:153-4.
- 3 Wyandt HE, Magenis RE, Hecht F. Abnormal chromosomes 14 and 15 in abortions, syndromes and malignancy. In: Yunis JJ, ed. *New chromosomal syndromes*. New York, London: Academic Press, 1977:325-8.

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