The Prader-Willi syndrome with a 15/3 translocation

SUMMARY A de novo translocation of 15q to 3p with complete monosomy of 15p and partial monosomy of 15q was detected by trypsin banding on peripheral lymphocytes of a 5-year-old boy with Prader-Willi syndrome (severe mental retardation, dyslalia, cryptorchidism, and muscular hypotonia). The pathogenic role of chromosome 15 abnormalities in the aetiology of this syndrome is discussed.

About 10% of patients with Prader-Willi syndrome show cytogenetically detectable abnormalities of a D chromosome, usually the translocation of one of the D chromosomes to another chromosome. Hawkley and Smithies (1976) suggest that it is chromosome 15 which is involved in the pathogenesis of this syndrome.

Using the trypsin banding method (Burkholder and Comings, 1972), we have detected a de novo unbalanced translocation of the distal part of the long arms of chromosome 15 to the short arms of chromosome 3 in a patient with Prader-Willi syndrome.

Case report

The boy was the first child of healthy young parents; his birthweight was 2750 g, length 50 cm. Psychomotor retardation was obvious from the first months of his life. He started to walk at the age of 20 months and to speak at 3 years. Now, at 5 years old, his mental ability corresponds to debilitas gravis. Other clinical symptoms are obesity (+4-5 sigma), muscular hypotonia, genu valga, cryptorchidism, and dyslalia (Fig. 1).

His parents and younger brother are healthy with a normal karyotype.

CYTOGENETIC STUDIES

There was complete monosomy of the short arms and partial monosomy of the long arms of chromosome 15, and probably also monosomy of a small part of the short arms of chromosome 3 (Fig. 2).

The karyotype was 45,XY,-3,-15,+t(3;15) (p25;q15) or 45,XY,-3,-15,+t(3;15) (3qter→3p25:15q15→15qter).

Discussion

The unbalanced chromosomal translocation described is quite rare; we have failed to find a similar abnormal karyotype in published reports. The clinical signs of our patient support the hypothesis of Hawkley and Smithies (1976) about the role of the short arms of chromosome 15 in the Prader-Willi syndrome. Some of the other karyotypes of reported patients are also in agreement with this hypothesis.
Case reports

Nevertheless, more data are necessary for a final conclusion.

Maria Kučerová, Marta Straková, and Zdenka Polívková
Pediatric Department of the Postgraduate Medical Institute, Thomayer’s Hospital, Prague, Czechoslovakia

References


Requests for reprints to Dr M. Kučerová, Pediatric Department of the Postgraduate Medical Institute, Thomayer’s Hospital, Budějovická 800, Prague 4, Czechoslovakia.