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. Psycho-
Nevertheless, more data are necessary for a final conclusion.

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**References**


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