

Clinical consequences of deletion 1p35

SHARON L WENGER*, MARK W STEELE*, AND DOROTHY J BECKER†

*Division of Medical Genetics, and †Department of Endocrinology, Children's Hospital of Pittsburgh, Department of Pediatrics, School of Medicine, University of Pittsburgh, Pennsylvania, USA.

SUMMARY Few cases of deletion 1p have been reported. We report a case of terminal deletion 1p35 in a patient with psychological and neurological dysfunction.

Case report

The patient was the 3500 g white male product of a normal, term, uneventful pregnancy and delivery to a 38 year old gravida 2, para 2, white female and 40 year old white father. His infancy appeared normal (other than undescended testes repaired at three years of age) but by the age of two and a half years language delay, short attention span, hyperactivity, and social difficulties were apparent. By school age the patient showed significant psychiatric problems which were treated first with methyl phenidate (Ritalin), then dexamphetamine (Dexedrine), and finally thioridazine (Mellaril). In school the patient regressed from learning disability/mental retardation classes to trainable mental retardation classes, finally ending up in a special class for autistic children after several psychiatric hospital admissions. At the age of nine the patient developed a remarkable increase in appetite and his weight went from the 50th centile to over the 90th centile within a year. Evaluation in the Endocrine Clinic at the age of 10½ years for suspected Prader-Willi syndrome showed height at the 50th centile, weight above the 90th centile, eyes slightly almond shaped, mild epicanthic folds, small hands with incurving of the fifth fingers, small chin, and unusual facies. Sexual development was Tanner stage I to II. Bone age on x ray was 1 to 2 SD above the mean. Neurological evaluation found some intention tremors in the upper extremities on fine motor activity and also

mild motor incoordination with mild ataxia. Speech showed poor pronunciation and articulation of words with intermittent stuttering. Otherwise, the neurological findings were unremarkable. The patient's family history was unremarkable. He had an older brother of superior intelligence.

CYTOGENETIC STUDIES

Peripheral blood lymphocytes were cultured for 72 hours and then harvested for metaphase preparation. Chromosomes were analysed by trypsin Giemsa banding. The patient's karyotype was 46,XY,del(1)(p35) (figure). The patient's parents refused genetic evaluation.

Discussion

Deletions of chromosome 1p are very rare. Only a few such cases have been reported involving the terminal end by either ring formation¹⁻⁴ or translocation⁵⁻⁷ and our case is the first deletion 1p35 reported. The clinical findings in the other reported cases have included mental and growth retardation, some facial dysmorphism, and cleft palate, but all in all, a diagnostic clinical phenotype has not been clearly defined.

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Correspondence and requests for reprints to Dr Sharon L Wenger, Children's Hospital of Pittsburgh, One Children's Place, 3705 Fifth Avenue at DeSoto Street, Pittsburgh, Pennsylvania 15213-3417, USA.

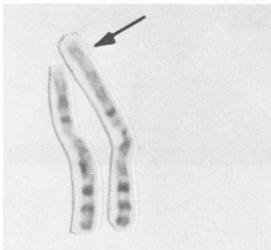


FIG Chromosome 1 pair from the patient showing the deletion 1p35.

Received for publication 6 February 1987.

Revised version accepted for publication 21 April 1987.