Requests for reprints to Dr C Estévez de Pablo, Servicio de Genética Médica, C E Ramón y Cajal, Madrid 34, Spain.

Reference

Interstitial deletion of the long arm of chromosome 5 in a deformed boy: 46,XY,del(5)(q13q15)

Summary A boy with mental retardation and physical abnormalities had an interstitial deletion of one chromosome 5: 46,XY,del(5)(q13q15).

Deletion of the short arm of chromosome 5 is a well known syndrome. Interstitial deletions, however, are uncommon. We had the opportunity to study a patient with an interstitial deletion of the long arm of a chromosome 5.

Case report

The proband was born after a normal pregnancy. A caesarean section was performed for fetal distress. He was the second child of the family. The parents, 21 and 25 years old, were in good health and had normal intelligence. A sister was born prematurely 2 years previously and died at 6 days of age. She had no malformations. The family history was otherwise unremarkable and there had been no abortions. Birthweight was 2500 g, length 45 cm, and head circumference 34.5 cm. The Apgar score was 9.

On physical examination the following abnormal findings were present (fig 1): a small and narrow forehead, a small, broad, upturned nose, a flat nasal bridge, hypertelorism, upward curving eyelashes, a large prominent metopic suture, a triangular shaped mouth, a large philtrum with a deep groove, retrognathia, large ears, short neck, short upper limbs, syndactyly of the big toe and the 3rd and 4th toes, and clinodactyly of the 5th finger. A cardiac murmur was also heard. The rest of the physical examination was normal.

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Variants and Anomalies in Man, 32 interstitial deletions are known\(^1-^3\) and Kucerova and Polivkova\(^4\) found three cases with del(5)(q15q23), (q21q23), and (q15q22). The segment of the chromosome 5 that was lost in these three cases was different from the one in our patient. The clinical abnormalities of the patient of Pescia et al\(^3\) also differed from those of our patient.

Studies attempting to map genes on the fragment which was lost in our patient were inconclusive.

C Stoll, J-M Levy, and Marie-Paule Roth
Institut de Puériculture, CHU, Strasbourg, France

References


Requests for reprints to Dr C Stoll, Institut de Puériculture, 23 rue de la Porte de l’Hôpital, 67000 Strasbourg, France.

Neurological and neuropathological findings in ring chromosome 4

SUMMARY Despite the fact that mental retardation, microcephaly, seizures, and hyperactivity are common in patients with ring chromosome 4, little has been written about the underlying neuropathology. We describe a 6-year-old girl whose neuropathological findings included low brain weight, abnormal gyral development, and heterotopic neurons. The significance of these findings in regard to other retardation syndromes is discussed.

Microcephaly, mental retardation, seizures, and hyperactivity are frequently present in patients with

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C Stoll, J Levy and M P Roth

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