proximal to our breakpoint in 1q22 or in 1q21. Our UGPP assay results might suggest that UGPP lies in the same region but this should be interpreted with caution.

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C ESTÉVEZ DE PABLO, J M GARCÍA SAGREDO, M T FERRO, P FERRANDO, AND C SAN ROMÁN
Servicio de Genética Médica, C E Ramón y Cajal, Madrid, Spain

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

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, retro-micrognathia, 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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FIG 1 The patient with del(5)(q13q15).
According to 5 chromosome abnormalities, the obvious of the Discussion showed an Arylsulphatase B, hexosaminidase of the groups. Blood preparations Chromosome orcein and treated and located there were seven -3 SD.

-3 SD, Chromosome preparations from short term peripheral blood leucocyte cultures were stained with orcein and treated for RHG banding, CBG banding, and QFQ banding. In one culture flask BRedU was added at a concentration of 0.09 mmol/l for the last 8 hours of cultures. Mitoses were photographed with UV after acridine orange staining. These techniques showed an interstitial deletion of the long arm of chromosome 5 del(5) (q13q15) (fig 2).

The karyotypes of the parents were normal. The following genetic markers of the parents and of the child were studied but were not informative: hexosaminidase B, arylsulphatase B, HLA antigen, blood groups.

Discussion

The abnormalities found during clinical examination of the proband and his mental retardation are obviously the result of chromosomal abnormality. Interstitial deletions are rare chromosomal mutations resulting from two breaks with loosening of the intermedial segment and rejoining of the two parts. According to the Repository of Chromosomal Variants and Anomalies in Man, 32 interstitial deletions are known1-3 and Kucerova and Polivkova4 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 al5 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

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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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