References

- ¹ Shapiro SD, Hansen KL, Pasztor LM, *et al.* Deletions of the long arm of chromosome 10. *Am J Med Genet* 1985;20:181-96.
- ² Turleau C, de Grouchy J, Ponsot G, Bouygues D. Monosomy 10pter. *Hum Genet* 1979;47:233-7.

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De novo inv(5)(p15q22), del(5)(p15) in a boy with cri du chat syndrome

The proband was born weighing 3030 g after a normal pregnancy to a 20 year old primigravida mother and an unrelated 30 year old father. The family history was negative. A cat-like cry was evident neonatally. Psychomotor development was delayed and at 14 months of age he was still unable to sit and had no speech. Physical examination at this age (fig 1) showed height 77 cm, head circumference 43 cm (below the 3rd centile), hypotonia, and a round facies with hypertelorism, epicanthic folds, and down turned corners of the mouth.

Cytogenetic analysis on peripheral blood lymphocytes using GTG banding showed a rearranged chromosome 5 (fig 2) that seemingly resulted from two pericentric breaks, one located at the proximal edge of p15 and the other at q22, with inversion of the centric segment and deletion of the p15 band. The karyotype was interpreted as 46,XY,inv(5),del(qter→q22::p15→q22:). Parental chromosomes were normal.

FIG 1 The patient aged two years.

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FIG 2 Three chromosome 5 pairs from the patient. The rearranged chromosome is on the left and matched with the normal homologue along the centric p15→q22 segment. Also note the absence of the p15 band.

Since the clinical picture was fairly typical of the cri du chat syndrome, this observation agrees with the current notion that the monosomy 5p15, probably a narrow area around the 5p15.2 sub-band, is responsible for the phenotype.¹ Clearly, application of modern molecular genetic techniques will help to define the critical segment at the DNA level.²

The three known instances of pericentric inversion with deletion concern chromosomes 1, 4, and 5.⁵ The inversions and deletions of chromosomes 1 and 4 were three break rearrangements with interstitial deletions. In the only previous report of inv del(5) there were also two break points (at p15 and q31 or 33) with terminal deletion of p15. This may suggest, as in the present case, the formation of a neotelomere. These two examples may add further evidence for the occurrence of true terminal deletions.

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References

- ¹ Niebuhr E. The cri du chat syndrome. Epidemiology, cytogenetics and clinical features. *Hum Genet* 1978;44:227-75.
- ² Carlock LR, Wasmuth JJ. A molecular approach to analyzing the human 5p deletion syndrome, cri du chat. *Somatic Cell Mol Genet* 1985;11:267-76.
- ³ Kaiser P. Pericentric inversions. Problems and significance for clinical genetics. *Hum Genet* 1984;68:1-47.

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