Short reports

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Behaviour disorder in monosomy 10qter

Monosomy for the terminal segment of the long arm of chromosome 10 has been reported in 11 cases as summarised recently by Shapiro *et al.*¹ The phenotype of these patients has tended to conform to a consistent pattern with triangular face and moderate mental retardation. We now report a child who shows these features in association with severe behavioural disturbance.

The seven year old male proband was the only child of healthy unrelated parents. His mother and father were aged 25 and 34 years, respectively, at the time of his birth. He was delivered at 41 weeks' gestation weighing 3.00 kg and had mild meconium aspiration at birth.

He sat at seven months and began walking at 21 months when concern first arose because of speech delay and short stature. Hearing assessment indicated bilateral mixed sensory and conductive loss. On assessment aged five years, height (97 cm), weight (14 kg), and head circumference (47.5 cm) all fell below the 3rd centile. He had a triangular face (fig 1) with dolichocephaly, prominent nasal root with beaked nose and flared nares, long philtrum, small pointed jaw, and convergent strabismus. Other findings included bilateral fifth finger clinodactyly, limitation of extension of both elbows, and undescended testes.

FIG 1 The patient aged five years.

FIG 2 Partial GTG banded karyotype of the proband showing the normal (right) and the deleted chromosomes 10.

Formal developmental assessment at the age of 67 months showed a mean mental age of 39 months with highest scores being obtained on visuomotor tests. Noted by all who worked with him was his hyperkinesis and aggressive behaviour, with limited attention span and diminished need for sleep. He was often deliberately provocative and destructive, being prone to spit when thwarted or reprimanded. He could also be very affectionate. Following admission to residential school aged six years there was only minimal improvement in overall behaviour. An educational psychologist concluded that the pattern of behaviour was primarily intrinsic to the patient rather than the result of parental interaction or frustration associated with deafness.

Cytogenetic analysis from cultured lymphocytes showed a deletion of approximately the whole of the distal band (q26) of the long arm of one chromosome 10 (fig 2). There was also a pericentric inversion of one chromosome 9 which was present in the karyotype of the mother. Parental chromosomes were otherwise normal. The karyotype of the proband was interpreted as 46,XY,inv(9)(p1q21)mat, del(10)(q25-3 or 26-1).

The features of growth and mental retardation, triangular facies, strabismus, and cryptorchidism present in this boy are characteristic of other cases of monosomy 10qter.¹ Though developmental delay appears to be significant in all cases, details of behaviour are included less frequently. Two of the patients reported by Shapiro *et al.*¹ were described as hyperactive and the 11 year old girl described by Turleau *et al.*² was noted to be restless and agitated. We therefore suggest that behavioural disturbance may be a primary feature, hitherto unstressed, of this chromosome abnormality.

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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→p15→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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