Two pericentric inversions, *inv(2)(p11q13)* and *inv(5)(p13q13)*, in a patient referred for psychiatric problems

SUMMARY Two pericentric inversions were found in the karyotype of a male patient referred for psychiatric problems. Cytogenetic analysis, using conventional Giemsa staining and G and C banding techniques, revealed a pericentric inversion in chromosome 2, *inv(2)(p11q13)*, and chromosome 5, *inv(5)(p13q13)* (fig 1). Subsequent family studies showed that the proband's father carried both inversions also (fig 2), while other members were found to be carriers of either the inverted 2 or the inverted 5. To our knowledge, this is the first report of two pericentric inversions to be found in the human genome.

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The clinical and genetic counselling problems associated with pericentric inversions are not clear cut.¹⁻⁴ It is generally accepted, however, although exceptions do occur, that small inversions have a much better genetic prognosis than large inversions.

Case report

The patient was first described as being a potential psychopath with possible XYY syndrome. He is phenotypically normal with no congenital abnormalities. The family pedigree shows a history of alcoholism in the males of two generations accompanied, in some instances, by various mental disturbances (fig 3). The proband's father, although not unintelligent, has a background of antisocial behaviour, violent outbursts, and severe alcoholism, which is now at a terminal stage.

The reproductive history of the family shows only three miscarriages, but there was difficulty in conceiving in the marriage of III.7 and in the two marriages of III.1. Seven members of the family have died of cancer, three at an early age (III.1, III.4, and III.7).

FIG 1 G banded karyotype from the proband showing 46,XY,*inv(2)(p11q13)* and *inv(5)(p13q13)*.

FIG 2 Partial karyotypes from proband's father showing (a) conventional staining, (b) G banding, (c) C banding of *inv*(2) and *inv*(5).

III.3 suffers from manic depression, but is not a carrier of either the inverted 2 or 5. The youngest son of III.2 is dyslexic.

Discussion

With the exception of the cases of Crawford and Mason,⁵ it can be seen from the nine reported cases of pericentric inversions of chromosome 5 that the general rule of large inversions giving rise to recombinant chromosomes and greater risk of abortion holds true.

In our particular family, the inverted 5 is observed to involve slightly less than one-third of the total chromosome length, reducing the chances of producing unbalanced offspring.

Pericentric inversions of chromosome 2 are possibly more common than inversions in any other autosome, with the exception of chromosome 9.

In comparison with other autosomal inversions, no recombinant products have been found in published reports, but the 23 reported cases of inverted 2 have shown additional chromosomal structural aberrations, aneuploidy, miscarriages, abortions, infertility, and associations with mental retardation and congenital abnormalities.⁶ Considering the uncertainties and limited data available on the behaviour of pericentric inversions, their association with our family's clinical history cannot be ruled out.

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FIG 3 Pedigree of family

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Partial deletion of the long arm of chromosome 4: a clinical syndrome

SUMMARY Partial deletion of the long arm of chromosome 4 at q31 results in a clinical syndrome of mental retardation, characteristic ears, facial bone hypoplasia, cleft palate very prone to scarring on repair, and specific hand abnormalities. A female, aged 9 years, is described and compared with six other reported cases.

Case report

The proband is the 9-year-old daughter of a 30-year-old father and 25-year-old mother. The parents are

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Argentinian emigrants of Spanish descent. There are two female sibs, aged 7 and 4, who are normal. There is no history of maternal or paternal radiation or miscarriage in the mother. The proband was delivered normally at term and had a birthweight of 3.1 kg. A cleft palate was noted at birth. She was described as a quiet baby, who needed to be woken for feeds. There was hard evidence of developmental retardation by the age of 1 year. Three attempts were made to repair the palate in Argentina at the age of 3, 5, and 6 years, all resulting in palatal fistula and scarring. Her speech is particularly retarded and indistinct, seemingly out of proportion to her general developmental level. There were only half a dozen distinguishable words and the child was frustrated by her inability to be understood.

CLINICAL EXAMINATION

Her height was 122 cm (10th centile), and her head circumference was 48 cm (less than the 3rd centile). She had prominent and small ears coming to a rounded point superiorly (fig 1). She had mid-facial and mandibular hypoplasia and a broad nasal bridge (fig 2), small 5th metacarpal bones and small distal phalanges of the 5th digits (fig 3). In addition there was some irregularity of the outline of the distal

FIG 1 Ear abnormality, pointed and prominent.

FIG 2 The proband. Note small jaw, facial bone hypoplasia, and prominent ears.