Craniosynostosis

Sir,

In their interesting paper on families with craniosynostosis, Professor Carter and co-workers suggest that our higher incidence of apparent autosomal dominant coronal synostosis may result from our inclusion of patients with Saethre-Chotzen and other syndromes among our study group. Certainly, if the minor hand anomalies to which we referred have the same aetiology as the craniosynostosis, then some families had 'private' syndromes. We agree that family number 1 has what the authors call the 'split face syndrome', and that it is like the family reported by Slover and Sujansky. However, in the interest of clarity, we would like to point out that we did not make a diagnosis of Saethre-Chotzen syndrome unless at least one family member had the typical syndactyly. Therefore, as the title implied, our families with Saethre-Chotzen syndrome were not included in the paper and we do not believe that families 4, 8, or 10 had this condition.

The degree to which the reported heterogeneity of the craniosynostosis syndromes represent true genetic heterogeneity must await biochemical or linkage markers for the genes or both. In the meantime it remains true that patients who have craniosynostosis in association with other dysmorphic features, notably of the hands, are more at risk to represent a single gene mutation than are those with isolated craniosynostosis.

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Pericentric inversion of chromosome 13

Sir,

In 1972 a paper from our laboratory described a large family with a pericentric inversion of chromosome 13, leading to a duplication deficiency

References


FIG 1 Pedigree of family.

[Diagram of pedigree with annotations: Normal, Inv (13) (p11 q22), Rec (13) dup p, inv (13) (p11 q22) type 1, Dead, not tested, Not tested, Spontaneous abortion, Proband, Congenitally abnormal, dead, not tested, Spontaneous abortion rec (13) dup p, inv (13) (p11 q22) type 2, Spontaneous abortion mosaop inv (13) (p11 q22), inv (13) (p11 q22), inv (13), Abortus provocatus, rec (13) dup q, inv (13) (p11 q22) type 1, Chromosome studies done on amniotic fluid]
Adjacent 2 translocation involving 13q and 21q

SIR,

The article in *Journal of Medical Genetics* entitled ‘Adjacent 2 translocation involving 13q and 21q’ (1982;19:314–5) states that this case is the first involving chromosomes 13 and 21 with an adjacent 2 disjunction in the infant and a balanced reciprocal translocation involving the long arms of chromosomes 13 and 21 in the mother.

We have studied a female carrier of a translocation in which the long arms of chromosomes 13 and 21 were involved. Identification with G banding (GTG) was not conclusive enough to enable us to establish definite breakpoints but, together with R banding (RBA), would suggest the following karyotype: 46,XX,t(13;21)(q21;q21).

The offspring of this woman suggest that this translocation carries a high risk. The first child died just after delivery in another hospital without cytogenetic study. The second child had dysmorphic features with partial trisomy 13 and partial monosomy 21 owing to an adjacent 2 meiotic disjunction. His karyotype was 46,XY,−21,+der(13),t(13;21)(q21;q21). The third child had the phenotype of Down’s syndrome because of a 3:1 segregation and his karyotype was 47,XY,+21,t(13;21)(q21;q21).

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Reference
1 Prieto F, Badia L, Asensi F, Roques V. Two reciprocal translocations t(9p+;13q−) and t(13q−;21q+). A study of the families. *Hum Genet* 1980;54:7–11.

Pyloric stenosis: children vs sibs

SIR,

We have reported findings in the relatives of patients with pyloric stenosis which showed, for female patients, more children affected than sibs. This is unexpected on a simple multifactorial threshold model and has led us and others to speculate whether there may be some direct maternal effect, though there is no indication, on the small series available, that maternal half-sibs are more often affected than paternal half-sibs.² We have continued to follow the children of the female patients born between 1933 and 1949 (but not those born between 1921 and 1932, who are not likely to have had further children) and the relatively high risk to children has now disappeared. The data on

adjacent with congenital malformations in three family members.¹

Since then all carriers have been followed and fetal cells cultured from all known pregnancies, including two spontaneous abortions (fig 1). The inversion and breakpoints have been confirmed with banding techniques (fig 2a, b).

The second type of recombinant chromosome postulated in our paper has been found in one spontaneous abortion (fig 2c).

We wish to add this new information and show the revised pedigree.

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Reference

![Chromosome 13 from a carrier. The inverted chromosome is shown with both G and NOR banding.](attachment:chromosome13.png)

**FIG 2** (a) Chromosome 13 from a carrier. The inverted chromosome is shown with both G and NOR banding. (b) The recombinant chromosome 13, type 1. (G banding.) (c) The recombinant chromosome 13, type 2. (Conventional, G, and NOR banding.)
Pericentric inversion of chromosome 13.

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doi: 10.1136/jmg.20.2.154-a

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