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.)

associated 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.

HALLA HAUKSDÓTTIR*, ÁSTRÓS ARNARDÓTTIR*,
MARGRÉT STEINARSDÓTTIR*,
ELÍN GUÐMUNDSDÓTTIR*, SÆVAR HALLDÓRSSON†,
AUÐÓLFUR GUNNARSSON‡, AND
MARGARETA MIKKELSEN§

*The Chromosome Laboratory, Department
of Pathology, University of Iceland, †Department
of Paediatrics, St Joseph's Hospital,
‡Department of Gynaecology and Obstetrics,
Landspítalinn, 101 Reykjavík, Iceland; and
§John F Kennedy Institutttet, G1 Landevej 7-9,
Glostrup, Copenhagen, Denmark.

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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.¹ 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).

F PRIETO AND L BADIA

Sección de Genética,
Servicio de Hematología y Hemoterapia,
Hospital Infantil,
Ciudad Sanitaria de la Seguridad Social 'La Fe',
Valencia, Spain.

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Pyloric stenosis: children vs sibs

SIR,

We have reported¹ 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