negative region in the short arm proximal to the centromere. The region stained positively with CBG and GBG banding indicating that it is heterochromatic and late replicating. It does not fluoresce brilliantly with QFQ banding in contrast to the centromeric region and it is G11 negative, indicating that it is not translocated 9 heterochromatic material. Investigation of parental chromosomes revealed that the variant was maternally inherited. The mother had had three normal children and no evident miscarriages. The karyotype of the proband might be described as 46,XY, var(3)(p11+).mat.

The staining characteristics of the present variant region are unusual as it stains negatively with GTL banding but positively with CBG banding. The only G negative, C positive regions in the normal human karyotype are at 9p12 (which is G11 positive whereas the variant described here is not) and within 1q12, where alternating positive and negative G bands can be observed in prometaphase chromosomes.1

It is likely therefore that the observed GTL negative, CBG positive material results from amplification rather than translocation, as has also been suggested by Seabright et al with respect to a variant of chromosome 5.

No conclusions can be drawn regarding an association between this variant and the presentation of the proband, namely recurrent abortion in his wife, particularly as the variant is also present in the mother who reproduced normally.

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Pubertal development in partial trisomy 14q

In 1980 we described an 18 year old boy with severe mental and physical retardation who had partial trisomy 14q (pter→q21) resulting from tertiary trisomy segregation in meiosis from a t(3;14)(p25;q21) maternal carrier.1 At the time his height was 114.5 cm (on the 50th centile for a five year old), weight 22 kg, and head circumference 49 cm (on the 50th centile for a two year old). He had no pubertal development.

Follow up in 1987, at the age of 27 years, showed a length of 140 cm (on the 50th centile for a 10½ year old), bare weight 27.4 kg, and head circumference 50 cm (on the 50th centile for a three and a half year old). Hand length was 13 cm (with palm length 8.5 cm), foot length 16 cm, and chest circumference 74 cm. Pubertal development was complete with coarse skin, rough facial hair, adult male axillary and pubic hair (but no chest hair), a well developed penis, and normal adult testes. He had a thick, healthy head of hair and masculinised facial features. He was shaved every third day. The deformities noted previously were more pronounced, in particular the microphthalmia and closely set eyes (inner canthal distance 2 cm, outer canthal distance 7 cm) (figure), the pes cavus and talipes equinovarus of the feet, and the tapering fingers. He had also developed flexion contractures of the elbows and knees and was markedly hypertreflexic and hypertonic.

Studies included haemoglobin and blood film, renal and liver function tests, serum electrolytes, and thyroid function tests which were all within the normal male adult range, as was the serum testosterone (21 nmol/l), FSH (11.9 mIU/ml), and LH (6.0 mIU/ml).

Pubertal development in patients with autosomal chromosome abnormality is generally delayed, although in some cases precocious puberty occurs.2 Other than Down's syndrome, there is a paucity of reported cases detected in infancy or childhood whose subsequent development is followed. In one

FIGURE The patient aged 27 years.
case of partial trisomy 14q an adolescent girl had precocious puberty.\(^4\) In our case normal puberty occurred in a severely mentally and physically retarded male, approximately 10 years later than normal. He would appear to be the oldest case reported.

The main medical problem is the establishment of an adequate anticonvulsant medication regimen without causing undue sedation.

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**Familial transmission of autosomal whole arm translocation**

Centric fission followed by centromeric fusion of either heterologous or homologous nonacrocentric chromosomes is a very rare chromosome rearrangement leading to whole arm translocations in man.\(^1\) Familial transmission has been reported by Breg *et al\(^2\)* and Schober and Fonatsch.\(^3\) Breg *et al\(^2\)* reported an apparently balanced t(11p17q;11q17p) in a five year old girl with 18q deletion and her phenotypically normal mother. Schober and Fonatsch\(^3\) described seven balanced carriers of a t(1p19q;1q19p) in a large family without any evidence of reproductive failure or chromosomal imbalance.

We recently studied another type of heterologous whole arm translocation, t(6p10q;6q10p) (fig 1), in a 24 year old normal female (II.1, fig 2) who had three first trimester spontaneous abortions after the birth of a normal daughter. Chromosomal analysis was normal in her two sisters, but the same type of translocation was found in a maternal cousin (II.4) whose second and third pregnancies ended in spontaneous abortion. Prenatal diagnosis was performed in the fourth pregnancy and showed a male fetus with a normal chromosome complement.

The present family is the first in which a heterologous whole arm translocation was detected by the occurrence of

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**FIG 1** G banded partial karyotype of II-1 with the whole arm translocation t(6p10q;6q10p); the rearranged chromosomes are on the right.

**FIG 2** Pedigree of the family.