case of partial trisomy 14q an adolescent girl had
d 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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References
1 Smith A, den Dulk G, Elliott G. A severely retarded 18 year old
2 Smith GF, Berg JM. Down’s anomaly. 2nd ed. Edinburgh:
3 Smith A, Silink M, Ruxton J. Trisomy 18 in an 11 year old girl. J
4 Muldal S, Enoch BA, Ahmed A, Harris R. Partial trisomy

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Familial transmission of
autosomal whole arm
translocation

Centric fission followed by centromeric fusion of either
heterologous or homologous non-acrocentric chromo-
somes is a very rare chromosome rearrangement leading
to whole arm translocations in man.1 Familial trans-
mission has been reported by Breg et al2 and Schober and
Fonatsch.3 Breg et al2 reported an apparently balanced
t(11p17q;11q17p) in a five year old girl with 18q deletion
and her phenotypically normal mother. Schober and
Fonatsch3 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

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.

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reccurrent fetal wastage in the translocation carriers. As in
the two other reported families, the balanced transmission
of the heterologous whole arm translocation appeared to
be harmless for the carrier offspring in the present family.

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References

1 Kleczkowska A, Fryns JP, Van den Berghe H. Autosomal
whole arm translocations in man. A patient with t(5p7p:5q7q)
type rearrangement and review of the literature. Clin Genet
1986;30:72-5.

2 Breg HW, Miller DA, Allderice PW, Miller OJ. Identification
of translocation chromosomes by quinacrine fluorescence. Am J

3 Schober AM, Fonatsch C. Balanced reciprocal whole-arm
translocation t(1;19) in three generations. Hum Genet 1978;42:
349-52.

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