of late replication. In the 13q portion of the der(X), band 13q22 shows faint staining, suggesting late replication and hence inactivation. The phenotype of our patient supports the replication pattern in that she has some but not all the features associated with trisomy 13q.¹

A 50% increase in her ESD activity, compared to that of the normal parents, confirms, as previously shown by Mohandas *et al.*,² non-inactivation of the ESD loci in the 13q portion of the der(X) chromosome.

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Is the expression of fra(2)(q13) age dependent?

In 1987 Keskiaho *et al.*¹ described the inherited folate sensitive fragile site fra(2)(q13) in three unrelated, mentally retarded children, two of them with different forms of epilepsy. Repeated chromosome studies in the six parents failed to detect the same fra(2)(q13) in any of them, except for one cell in one of the fathers. The authors concluded that fra(2)(q13) can be transmitted by persons not expressing it and that fra(2)(q13) expression may be age dependent.

In the fragile X screening programme of the Leuven Centre for Human Genetics we had a similar experience.²

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We detected a fra(2)(q13) in two dystrophic and profoundly retarded male sibs in 30% and 20% respectively of the cells in three different cultures. The boys were three months and 18 months old at the time of examination and presented with recurrent convulsions, failure to thrive, and similar dysmorphic craniofacial features with narrow, sloping forehead, long occiput, exophthalmia, and micrognathia. Both died before the age of two years and, apart from the craniofacial dysmorphism and a large interhemispheric cerebral hygroma, no other anomalies were found.

These brothers were the fourth and fifth born children of a 34 year old moderately mentally retarded mother and a 32 year old alcoholic father. Three other children of this family, one boy and two girls, aged between three and seven years, are physically normal with borderline mental development. We were unable to find the fra(2)(q13) in three separate lymphocyte cultures from the parents and these three children.

The findings in the present family confirm the data of Keskiaho *et al.*¹ that persons not expressing the fra(2)(q13) can transmit it and that its expression may be age dependent.

On the other hand, large scale studies of autosomal fragile sites in the mentally retarded versus a control population failed to show any relationship between the expression of autosomal fragile sites and clinical abnormalities or mental retardation.

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Simultaneous occurrence of heritable t(3;7) and t(14;21) in two sibs

Cytogenetic analysis was performed on a one year old boy with features of Down's syndrome. Results showed a

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