using routine methods and analysed after trypsin G banding. In addition, lymphocytes were cultured with BrdU in order to demonstrate the late replicating X chromosome.

Both parents had normal karyotypes, but the proband's karyotype revealed an apparently balanced X;13 translocation: 46,X,t(X;13) (Xpter→Xq13::13p11→13pter;13qter→13p11::Xq13→Xqter) (figure). BrdU incorporation showed the normal X to be late labelling in all of 100 cells examined. As the normal X was late replicating in each cell, it was not possible to detect any spread of inactivation from the X to the 13 in the 100 cells examined.

Discussion

Although it is easy to see why unbalanced X; autosome translocations cause phenotypic abnormalities due to deletion or duplication of chromosomal material, carriers of balanced translocations are often phenotypically normal. However, there are several well established classes of abnormality found in association with X;autosome translocations. Firstly, ovarian dysgenesis is associated with breakpoints on the X within the region Xq13 and Xq26.² Secondly, evidence is accumulating that there can be mutation damage by gene disruption at the breakpoint on the X chromosome. Examples supporting this theory include the cases of Duchenne muscular dystrophy occurring in girls with de novo X; autosome translocations with breakpoints at or near Xp21 (listed in Hodgson and Bobrow³), corresponding to the position allocated to the DMD locus by linkage analysis.⁴

Thirdly, the position effect can cause phenotypic

abnormalities in balanced X;autosome translocations, due to dissociation of genes from regulator sites or the spread of inactivation from an area of heterochromatin to a portion of the neighbouring translocated chromosome which would not normally be inactivated.

Five of seven previously reported patients with X; autosome translocations with breakpoints at Xq13² were infertile, but none had other dysmorphic features. Of seven patients reported with deletions of Xq with breakpoints at Xq13, none had phenotypic abnormalities other than the Turner stigmata.⁵ Our patient is thus the first reported case of a balanced X; autosome translocation with a breakpoint at Xq13 and an abnormal phenotype, other than gonadal dysgenesis.

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Terminal deletion of the long arm of chromosome 10

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SUMMARY A de novo chromosome abnormality interpreted as a terminal deletion of chromosome 10, del(10)(pter→q25·2:), was ascertained in a newborn female with multiple malformations. The clinical features observed at birth and on follow up at 10 months of age

are described and compared with previously reported cases.

Three patients with monosomy for the chromosome region 10q25→qter have been described previously, two with a de novo terminal deletion^{1 2} and one with an unbalanced familial translocation.³ In addition, five reports describe slightly more distal deletions with the breakpoint in band 10q26,⁴⁻⁸ and a single

FIG 1 The patient (a) at birth and (b) at 10 months of age.

report describes monosomy for the region 10q23→qter.⁹

Case report

The proband (fig 1a) was the first child of unrelated parents aged 18 and 19 years, born by normal vaginal delivery at 42 weeks' gestation after induction of labour. At birth she was covered with vernix and was noted to be below the 3rd centile for all parameters: occipitofrontal diameter 30.5 cm, weight 2.15 kg, length 44 cm. She had microcephaly and brachycephaly. The eyes were prominent owing to shallow orbits and there was marked conjunctivitis. The mandibles were hypoplastic (right more than left), the nose was large and broad with a prominent bridge, and the ears were large but not low set. A unilateral simian crease and bilateral clinodactyly were noted, as well as severe bilateral talipes equinovarus. She had hypoplastic female external genitalia. There was no evidence of cardiac anomalies. A skeletal survey confirmed microcephaly and revealed small facial bones, particularly in the mid-face. Twelve pairs of ribs were present with splaying of the lower ones. The long bones were gracile in the mid-portion with some linear layering sclerosis.

At 10 months of age (fig 1b) her development was

FIG 2 G banded homologues of chromosome 10 from three cells, the abnormal chromosome to the right, and a diagrammatic representation indicating the breakpoint (arrowed).

severely delayed (less than 3 months). Weight and length remained below the 3rd centile and the head circumference was only 38.5 cm, 2 cm below the 3rd centile. Feeding was predominantly by nasogastric tube.

CYTOGENETIC STUDIES

Chromosome analysis was performed on a specimen of peripheral blood obtained shortly after birth. G banding using a trypsin-Leishman protocol revealed a terminal deletion of chromosome 10 (fig 2). The karyotype was thus 46,XX,del(10)(pter→q25.2:). Parental karyotypes were normal.

Discussion

Clinical features of the patient common to the majority of earlier cases with a similar chromosome abnormality included low birth weight, microcephaly, a broad prominent nasal bridge, large or otherwise abnormal ears, developmental delay, and growth retardation.⁸ Her facies (fig 1) were strongly reminiscent of some of the previous patients,^{2,4} although prominent eyes resulting from shallow orbits have not been apparent previously and hypoplasia of the mandibles is not a consistent feature.

So far there is an excess of females (9/12 including

the present patient) with monosomy for the terminal portion of the long arm of chromosome 10, and overall the clinical presentation of this patient is compatible with those previously reported.

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Announcement

CLINICAL GENETICS SOCIETY

The next meeting of the Clinical Genetics Society will be held at the Royal College of Physicians, London, on 5 and 6 December 1986 following immediately after the Royal College of Physicians' Conference 'New Prospects in Genetic Disease' on 3 and 4 December 1986. Those intending to present

papers or posters at the Clinical Genetics Society meeting should submit abstracts (about 150 words) before 17 October 1986 to the Secretary of the Society, Professor N C Nevin, Department of Medical Genetics, Institute of Clinical Science, Grosvenor Road, Belfast BT12 6BJ, Northern Ireland, from whom forms are available on request.