patient has ‘a new form of ectodermal dysplasia’ and that he has ‘not seen a combination like this before’ (personal communication to J.M.O., 28 February 1974).

We are impressed that this patient has a genetic condition, however, the exact aetiology remains unknown. The senior author would be grateful for information about similar patients.

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REFERENCES

A case of partial (9p) trisomy in a family with a balanced translocation 46,XX,t(1p+9q—)

Summary. A case of partial trisomy 9 is described, confirming that this will produce a recognizable syndrome of a characteristic facies with deep-set eyes and an unusual shape of the nose. Failure of secondary sexual characteristics to develop appears to be a feature in adults. In this case the mother had a balanced translocation between chromosomes 1 and 9 and must have, in addition, had a non-disjunction of her normal and her deleted No. 9 in order to produce the unbalanced state in her daughter.

A syndrome associated with trisomy of the short arms of chromosome No. 9 was first identified by Rethore et al (1970). This report describes a further example of the syndrome in an adult female, born to a mother who was a balanced translocation carrier 46,XX,t(1p+9q—).

Case report

The subject is a young Caucasian woman born on 30 August 1951 to non-consanguineous mother and father aged 24 and 25 years, respectively, at her birth. Birth weight was 2 900 g. There was a history of threatened abortion at 3 and 6 months. She had jaundice at 3 weeks of age and appeared to be mentally retarded by the age of 1 year. She was sitting up at 3½ years and standing at 5 years. She could say single words at 4 years. She had an operation for ptosis at the age of 6 and attended a special school until her admission to a hospital for the mentally handicapped on 13 March 1964. Since her admission to hospital there has been little change in her physical and mental state.

Physical state (Figs. 1 and 2). She is of small stature; height 134.0 cm and weight 3 600 g. She has fair hair, blue eyes, and rather small ear lobes. Cranial circum-
ference is 51 cm, cranial length 17 cm, and cranial width 13 cm. There is a convergent strabismus, ptosis, low hair line anteriorly and posteriorly, and the end of the nose has a bulbous appearance. The palate is high arched. Blood pressure is 100/50. She has short thumbs, clinodactyly, and single palmar creases on both hands. Her left hand finger print pattern is as follows: ulnar loop on the thumb, arch on the index finger, and ulnar loops on the middle, ring, and little fingers. On the right hand she has an ulnar loop on the thumb, arches on the index and middle finger, and ulnar loops on the ring and little fingers. The axial triradii are in the normal position. On both palms the c and d digital triradii are replaced by one interdigital triradius between digits IV and V. There is a complete failure of development of the secondary sexual characteristics with absent pubic and axillary hair and no breast development. Because of the amenorrhoea, apparent webbing of the neck, and her small stature, a possible diagnosis of Turner’s syndrome was initially made.

**Mental state.** She is in the profoundly mentally retarded category with a mental age of 3 years and an intelligence quotient on the Terman Merrill tests of less than 30. Her conversation is limited to ‘Daddy’ and ‘Hallo’. She is ambulant and can feed herself.

**Investigations.** Radiological skull examination shows slight hypertelorism and a thick cranial vault.

Radiology of thorax, spine, and limbs shows no abnormality. Amino-acid excretion is normal. Ketosteroid excretion is 3.33 μmol/24 h (0.96 mg/24 h); considerably below normal. Electroencephalogram shows an unusual abnormality in the form of paroxysmal runs lasting about 2 s of bilateral and diffuse 8 Hz waves of low to medium voltage. The latter is of greater amplitude in the frontal rather than the occipital areas. She has no history of epilepsy.

**Cytogenetic techniques**

Short-term lymphocyte cultures were set up using a modification of the method of Moorhead *et al* (1960). Cells were harvested after 72 h incubation with Colcemid (Ciba) treatment during the last 2 h. The cultures were subjected to treatment with a hypotonic solution of 0.9% trisodium citrate at 37°C for 10 min, and fixed in three changes of methanol:acetic acid (3:1). Slides were prepared by a flame-drying technique, Giemsa-banded metaphase spreads were prepared by a modification of Seabright’s method (1971). Pre-warmed, freshly-made slides were flooded with a solution of 5% trypsin (Difco) diluted 1:14 in isotonic saline for about 20 s. The slides were rinsed twice in isotonic saline then stained for 5 min in 10% Giemsa (Gurr) in pH 6.8 phosphate buffer. The slides were rinsed in buffer, dried, and mounted in DePex.

**Cytogenetic findings**

**Proposita.** Chromosome analysis of peripheral lymphocyte cultures revealed a modal number of 47.
An additional small metacentric chromosome was present, which was similar in appearance to the members of pair E16, but which could be readily identified in the majority of metaphase spreads by the possession of a prominent secondary constriction. The other 46 chromosomes were of normal female pattern. A single Barr body was seen in 45% of cells in a buccal smear. In view of these results chromosomal investigations of her parents and of her younger sister, age 15, were carried out. Her sister is of normal appearance and attends an ordinary school. The parents are phenotypically normal.

Parents and sister. Analysis of the father’s chromosomes revealed a normal male pattern: 46,XY. Both the mother and normal daughter, however, showed an abnormal karyotype. All the cells examined contained 46 chromosomes. One member of the C group was apparently missing and was replaced by a small metacentric chromosome bearing a secondary constriction which was also seen in the proposita. In addition, one arm of the chromosome 1 was found to be unusually long.

These findings were tentatively interpreted as resulting from a balanced reciprocal translocation between a No. 1 chromosome and a member of the C group, thought to be a No. 9 because of the presence of the secondary constriction.

Giemsa-banded preparations were made from fresh cultures which confirmed this interpretation (Figs. 3, 4, and 5). It could be seen that two breaks had occurred: one at band p36 on chromosome 1 and the other at q21 on chromosome 9. Reciprocal translocation of the distal portions of these chromosomes had taken place. Both the mother and the phenotypically normal daughter are, therefore, balanced translocation carriers, with a karyotype:

![Diagram of chromosome bands](http://jmg.bmj.com/)

**Fig. 3.** Banding studies on the proposita, confirming trisomy of the short arm of the No. 9 chromosome.
Case reports

46,XX,t(1;9)(1qter→1p36::9q21→9qter;9pter→9q21::1p36→1pter) (shortened designation: 46, XX,t(1;9)(p36;q21)). Giemsa-banded metaphase spreads from the proposita confirmed that the additional chromosome was the No. 9 chromosome involved in this reciprocal translocation, i.e., der(9). The karyotype of the propositus is therefore 47,XX,+der(9),t(1;9)(p36;q21)mat.

Discussion

Rethoré et al (1970) described four mentally retarded patients with an unusual appearance, notably microcephaly and brachycephaly with small eye sockets, deep set eyes, and a convergent squint. The nose was large and globular with a broad base. Clinodactyly was also featured. Failure of development of secondary sexual characteristics was seen in the older cases. The present case fits this descrip-
tion accurately. Rethoré and his co-workers were able to identify the chromosome concerned as the No. 9 because of the secondary constriction. In the present case, as well as a secondary constriction being present, banding studies further confirmed the identity of the chromosome. Previous reports of the C-group trisomies had not further identified the particular chromosome involved but nevertheless certain of these reports described patients with comparable abnormalities to those given here. These were reviewed by Deminatti et al (1969), and it seems likely that in some of these cases the No. 9 was also concerned. The syndrome has been further described by Hoehn et al (1971), Baccichetti and Tenconi (1973), Podruch and Weisskopf (1974), and Rethoré et al (1973).

Of particular interest is the dual event which has taken place. The mother, as well as being a balanced translocation carrier, must in addition have had a non-disjunction of her normal and her deleted No. 9 chromosome in order to produce the trisomic state in the daughter. Such a dual event is rare in man but has been described by Insley et al (1968). In a number of the previously reported cases of trisomy 9p the children have been born to a parent with a balanced translocation and a similar dual event must also have taken place in these patients. In the present case the question arises as to the risk of a similar non-disjunction affecting the younger sister with the balanced translocation. Amniocentesis will undoubtedly be indicated if she becomes pregnant.

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