

of three, and behavioural abnormalities such as perseveration, patterned movements, easy distractibility, and autistic tendencies.

The proband (fig 1), born at term, was the 4600 g, 51 cm product of a first pregnancy of a 32 year old primigravida. The father was 34 years old. The pregnancy was complicated by gestational diabetes and, during the first trimester, exposure to doxylamine-dicyclomine HCl-pyridoxine HCl (Bendectin^R). The family history was unremarkable. The patient had hypotonia and delayed developmental milestones and speech. A borderline abnormal EEG was documented after spells of staring or left arm posturing with abnormal vocalisation. He had frequent respiratory infections despite normal serum immunoglobulin levels and obesity beginning at the age of three years without compulsive eating or aberrant choice of foods. Behavioural abnormalities included perseveration, patterned movements, easy distractibility, and autistic tendencies, poor concentration, and an IQ of 50 which necessitated special educational facilities. Physical examination (aged seven) showed a height of 134 cm (above the 97th centile; at the 50th centile for nine and a half years), a weight of 59.5 kg (far above the 97th centile; 50th centile for age 15½ years), a head circumference of 56 cm (50th centile for 17 years), normal craniofacial morphology, bilateral myringotomy tubes in place, a normal phallus (6 cm, 60th centile) and testes, dermatoglyphic patterns of RUUUU on the left hand and UUUUU on the right, and mild hypotonia. The patient was withdrawn but responsive to commands and interacted with the examiner.

Prometaphase karyotyping with Giemsa-trypsin (fig 2)

was performed on peripheral blood lymphocytes using standard methods. The absence of a dark band at q12.1 or 12.3 was noted in one of the 18 homologues of the proband. The normal distance from the centromere to the remaining dark band suggests that band q12.3 was deleted. Parental karyotypes were normal.

Patients with the 18q syndrome have characteristic facial dysmorphism and behavioural abnormalities such as hyperactivity, aggressiveness, and irritability with easy frustration.¹ Wilson *et al*² reported seven patients with the typical 18q- phenotype who had common deletion of band 18q21. A male patient with deletion of band 18q12.1 or q12.3 had a different phenotype with epicanthic folds, prominent upper lip, micrognathia, cryptorchidism, left simian crease, weight below the 5th centile, seizures, and a developmental quotient of 30 to 40. While Wilson *et al*² also favoured the interpretation of del(18)(q12.2q21.1) in their patient, the different phenotype in our case argues against identical breakpoints. Two patients with a larger interstitial deletion of 18 (q11.2q21.3) had some characteristics of the 18q- syndrome, but shared obesity and deletion of band q12.3 with our proband.³ Further patient studies are needed to refine the phenotypic/cytogenetic correlations on 18q, since this is a region where psychological testing and high resolution cytogenetics may identify several loci relevant to cognitive function.

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Partial trisomy 16q secondary to a maternal 9;16 translocation

A six month old dysmorphic female was found to have partial trisomy for the long arm of chromosome 16 owing to a maternal translocation: 46,XX,t(9;16)(p24;q21). The

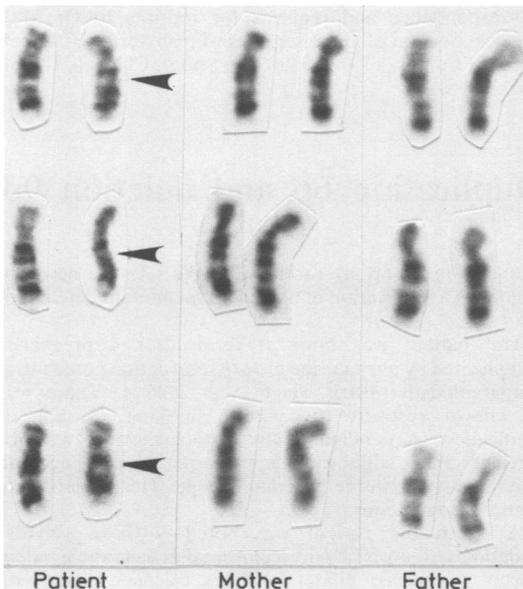


FIG 2 Partial karyotypes showing chromosomes 18 in the patient, mother, and father. The presumed deletion at band q12.3 is indicated by an arrow.

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patient was trisomic for the region 16q21→qter with a karyotype of 46,XX,-9,+der(9),t(9;16)(p24;q21)mat (figure). The father's blood chromosomes were normal.

The child was born at term after an uncomplicated pregnancy to unrelated Polish parents (mother 32, father 35). Birth weight was 2325 g, length 46 cm, and head circumference 32.5 cm. The newborn period was complicated by respiratory distress, a right pneumothorax, persistent fetal circulation secondary to meconium aspiration, and hypoglycaemia. The mother had a history of two previous miscarriages, each at three to four months' gestation. An older brother was reported to be in good health and the family history was negative for mental retardation and congenital malformations.

Abnormal features noted at birth and in subsequent examinations were a long face, coarse hair, prominent forehead with bossing, prominent nasal bridge with beak shaped nose, bilateral epicanthic folds, short palpebral fissures, strabismus, thin upper lip, micrognathia, high narrow palate with ridges, low set and posteriorly rotated ears, short fingers with deep, abnormal palmar creases, clinodactyly of the fifth fingers, widely spaced nipples, hypotonia, and psychomotor retardation. No other anomalies of organ systems were detected. Echocardiograms, skull x rays, head and abdominal ultrasound studies, TORCH titres, and CMV culture were normal.

Partial trisomy 16q is a rare disorder with significant dysmorphism, psychomotor retardation, and reduced survival in liveborn infants.^{1,2} Garau *et al*¹ postulated that trisomy 16q is mainly responsible for prenatal lethality of full trisomy 16. While partial trisomy 16q arises as the result of balanced parental translocations, the involvement of chromosomes in the translocations is not random.³ In a review of 12 known cases of partial trisomy 16q,² and in our own patient, translocations involved chromosomes 15, 22, 18, 21, 9, and 11. The trisomic segment in six patients, including ours, was 16q21→qter. Of the remaining seven

cases, four were trisomic for the region 16q11 or q13→qter, one was trisomic for the complete long arm of chromosome 16, and two were undefined. Patients with a longer trisomic segment tended to have shorter survival and a higher incidence of congenital heart disease (80%) than did those who were trisomic for the distal half of the long arm, heart defects being present in 17% of the latter cases. Our case has many of the clinical features that appear to make partial trisomy 16q a recognised phenotype.²

The patient of Buckton and Barr³ was similar to ours in that the translocation involved chromosome 9 with a possible monosomic segment of 9p24→pter. However, in contrast to our patient, theirs had a two-thirds distal trisomic segment, a more severe phenotype, including flexion deformities and heart and cerebral defects, and died at eight days of age. The length of the trisomic segment appears to influence clinical severity and longevity among patients with partial trisomy 16q.

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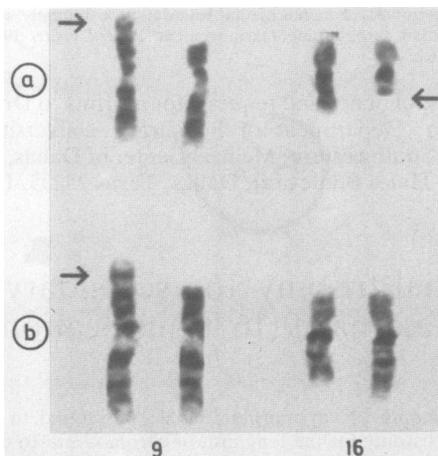


FIGURE G banded partial karyotype showing chromosomes 9 and 16 from (a) the patient's mother and (b) the patient.

Duplication 6p and deletion 9p

Duplications of 6p are rarely seen. We report a newborn male with a duplication of 6p and a concomitant deletion of 9p.

The patient was born at term after a pregnancy complicated by intrauterine growth retardation noted ultrasonographically. Birth weight was 2126 g. There was no known exposure to cigarette smoking, alcohol, or medication. A detailed family history showed two miscarriages and a stillborn male in the maternal lineage and one miscarriage in the paternal lineage. The parents were non-consanguineous.

At birth the patient was noted to have valvular pulmonary stenosis, a ventriculoseptal defect, and a patent ductus arteriosus; bilateral talipes calcaneovalgus with vertical tali; bilateral radial hypoplasia, camptodactyly,

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