The patient had hypotonia and delayed lingual development when evaluated at the age of two years. He had frequent head movements, easy distractibility, and autistic tendencies. 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.

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

References


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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.
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, microglossia, 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. Garau et al1 postulated that trisomy 16q is mainly responsible for prenatal lethality of trisomy 16. While partial trisomy 16q arises as the result of balanced parental translocations, the involvement of chromosomes in the translocations is not random. In a review of 12 known cases of partial trisomy 16q, 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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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 talus; bilateral radial hypoplasia, camptodactyly.

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