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. Garau et al. 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. 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.

The patient of Buckton and Barr 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.

References


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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 ventriculoaortic defect, and a patent ductus arteriosus; bilateral talipes calcaneovalgus with vertical talus; bilateral radial hypoplasia, camptodactyly...

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and abnormal palmar creases; ambiguous genitalia with microphallus, a left hydrocele and left inguinal hernia; and a small, proximally placed anus.

At the age of four and a half months, in addition to the above congenital malformations, the patient had marked postnatal growth retardation, microcephaly, and striking craniofacial dysmorphism (fig 1) with prominent frontal bossing and trigonocephaly, upswept frontal hair pattern with marked hirsutism of the forehead, low set, posteriorly rotated ears with a thickened helix and markedly prominent antihelix, a 'beaky' nose, a thick, wide philtrum with a small notch in the vermilion border of the lip lateral to the right pillar of the philtrum, a high palate with thick palatine ridges, micrognathia, and a short neck.

High resolution chromosome analysis was performed on peripheral blood. One hundred GTG banded cells showed a 46,XY,−9,+der(9),t(6;9)(p11;p24) chromosome complement. This karyotype indicates a duplication of the majority of the short arm of chromosome 6 (6p11→pter) and a deletion of a distal region of the short arm of chromosome 9 (9p24→pter) (fig 2). Subsequent parental
The megacystis-microcolon-intestinal hypoperistalsis syndrome: a fatal autosomal recessive condition

SUMMARY We report the cases of two sibs with the megacystis-microcolon-intestinal hypoperistalsis syndrome. The parents are first cousins. These cases further support the view that this syndrome is inherited in an autosomal recessive fashion.

Case reports

Cases 1 and 2 are the second and fourth pregnancies of a consanguineous couple of Indian origin. Both parents were normal and the family history was unremarkable. The first pregnancy resulted in a 12 week spontaneous abortion and the third in the birth of a normal male infant at term.

CASE 1

Prenatal. Uneventful until 31 weeks' gestational age when the mother was admitted in preterm labour. Initially controlled with ritodrine, but labour became established.

Birth. Lower segment caesarian section was performed because of the preterm breech presentation. There was difficulty in delivery and resuscitation because the foetal abdomen was distended.

Clinical examination. A liveborn female infant, weighing 2050 g, was delivered and 475 ml of clear fluid was drained from a suprapubic puncture. On examination the child appeared to have deficient anterior abdominal wall.

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table: Comparison of dysmorphic characteristics.

<table>
<thead>
<tr>
<th>Clinical features</th>
<th>Dup(6p)</th>
<th>Del(9p)</th>
<th>Present patient</th>
</tr>
</thead>
<tbody>
<tr>
<td>Mental retardation</td>
<td>+</td>
<td>+</td>
<td>+</td>
</tr>
<tr>
<td>Growth retardation</td>
<td>+</td>
<td>-</td>
<td>+</td>
</tr>
<tr>
<td>Microcephaly</td>
<td>-</td>
<td>-</td>
<td>-</td>
</tr>
<tr>
<td>Trigonocephaly</td>
<td>+</td>
<td>+</td>
<td>+</td>
</tr>
<tr>
<td>Hydrocephalus</td>
<td>+</td>
<td>+</td>
<td>-</td>
</tr>
<tr>
<td>Prominent forehead</td>
<td>+</td>
<td>+</td>
<td>+</td>
</tr>
<tr>
<td>Prominent nasal bridge</td>
<td>+</td>
<td>-</td>
<td>+</td>
</tr>
<tr>
<td>Short nose</td>
<td>+</td>
<td>-</td>
<td>-</td>
</tr>
<tr>
<td>Flat nasal bridge</td>
<td>+</td>
<td>+</td>
<td>-</td>
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<tr>
<td>Low set ears</td>
<td>+</td>
<td>-</td>
<td>+</td>
</tr>
<tr>
<td>Low set ears</td>
<td>+</td>
<td>+</td>
<td>NR</td>
</tr>
<tr>
<td>Posteriorly rotated ears</td>
<td>-</td>
<td>+</td>
<td>-</td>
</tr>
<tr>
<td>Club feet</td>
<td>+</td>
<td>+</td>
<td>+</td>
</tr>
<tr>
<td>Scoliosis</td>
<td>-</td>
<td>-</td>
<td>NR</td>
</tr>
</tbody>
</table>

chromosome studies were normal, indicating a de novo rearrangement in the patient.

Duplications of 6p are exceedingly rare and are generally secondary to a familial translocation. Our patient has a duplication involving a greater portion of 6p than those previously reported. He has the classical facial dysmorphism associated with 6p duplications, including flat facies with a high forehead, broad, prominent nasal bridge, short nose, small mouth with thin lips, and low set ears.

Cases with a deletion of the short arm of chromosome 9 generally present with a clinically defined phenotype. These distinctive features, which our patient also shows, include trigonocephaly, flat nasal bridge, long philtrum, low set, posteriorly rotated ears, micrognathia, and a short neck.

The table outlines the features seen with 6p duplications and 9p deletions and the features of our patient are compared to these phenotypes. As mentioned above, he has characteristics of both conditions. He clearly has additional anomalies which may be secondary to the size of his 6p duplication.

A detailed review of published reports suggests that our case is the first reported de novo rearrangement resulting in duplication of 6p and deletion of 9p. It also involves the largest region of 6p reported in a liveborn infant to date.

References