TABLE Comparison of dysmorphic characteristics.

Clinical features	Dup(6p)	Del(9p)	Present patient
Mental retardation	+	+	+
Growth retardation	+	-	+
Microcephaly	-	-	+
Trigonocephaly	-	+	+
Hydrocephalus	+	-	-
High forehead	+	-	+
Prominent nasal bridge	+	-	+
Short nose	+	+	-
Flat nasal bridge	-	+	-
Long philtrum	-	+	-
Low set ears	+	+	+
Posteriorly rotated ears	-	+	+
Short, broad neck	-	+	+
Widely spaced nipples	-	+	NR
Congenital heart defects	+	+	+
Renal abnormalities	+	-	-
Ambiguous genitalia	-	-	+
Hypospadias	-	+	-
Cryptorchidism	-	+	-
Hernias	+	+	+
Radial abnormalities	-	-	+
Club feet	+	+	+
Scoliosis	-	+	NR

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.¹⁻⁶ 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.⁷⁻⁸ 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.

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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 fetal abdomen was distended.

Clinical examination. A liveborn female infant, weight 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.

Received for publication 8 December 1987.
Revised version accepted for publication 13 April 1988.

Short reports

musculature and a very distended bladder. Ultrasound showed megacystis and bilateral hydronephrosis and an initial diagnosis of prune belly syndrome was made. It was proposed to proceed to bilateral open nephrostomies but the infant developed a cardiac arrhythmia and resuscitation was unsuccessful. She died aged seven hours.

Necropsy. The abdominal wall muscle was found to be normal in quantity and structure. On opening the abdomen there was a hugely distended bladder with only a moderately thinned wall, bilateral megaureters, distended renal pelvis with extremely thin walls, and hydronephrosis. The urethra, anal canal, and rectum were patent.

Although the duodenum and small intestine were contracted, the stomach was distended with gas, but no obstruction was found at the pylorus or in the duodenum. The small intestine was shortened and both this and the large intestine were empty. The ileocaecal junction lay free in the mid-abdomen. All other systems were grossly normal.

Histology of the gastrointestinal and urinary tracts showed normal innervation. Thus, necropsy ruled out mechanical obstruction or absent innervation of the gastrointestinal and urinary tracts.

CASE 2

Prenatal. Ultrasound examination at 16 weeks confirmed a single viable fetus. Both fetal kidneys were hydronephrotic and the bladder was distended; the fetal stomach also appeared distended with no duodenal fluid observed. Liquor volume was normal. It was felt that this represented a recurrence of the syndrome observed in case 1 and termination was offered and accepted by the parents. This was undertaken the next day by extra-amniotic prostaglandin infusion and resulted in abortion of a female fetus.

Necropsy. This showed a female fetus, weight 170 g, with a thin upper abdominal wall. The bladder, stomach, duodenum, and ureters were grossly distended. The urethra, anal canal, and rectum were empty and probe patent from below. There was malfixation and malrotation of the gastrointestinal tract with shortening of the small intestine. Histological examination showed apparently normal innervation of the gastrointestinal and urinary tracts. All other systems were normal.

On review of both cases the diagnosis was revised to that of the megacystis-microcolon-intestinal hypoperistalsis syndrome (MMIHS).

Cell culture in case 1 became infected and failed to show any results but case 2 showed a normal female karyotype on a fully banded preparation.

Discussion

There have now been five sets of affected sibships (including this report) of the MMIHS reported.¹⁻⁴ Our set and one other,⁴ as well as a single case,⁵ occurred to consanguineous parents. Several authors have commented on the similarity in appearance between the MMIHS and the prune belly syndrome,^{4,6} and it has been proposed that the apparent female preponderance seen in the MMIHS may be the result of the misdiagnosis of male cases as cases

of prune belly syndrome.⁴ The abnormalities in the MMIHS include functional obstruction of both urinary and gastrointestinal systems. Although the urinary tract distension appears to resolve once open drainage has been established, the gastrointestinal tract never manages to function properly despite the use of various surgical and pharmacological manoeuvres and despite apparently normal innervation; hence death invariably occurs at an early age.^{1,6}

These cases further support the view that the MMIHS is an autosomal recessive condition.⁴ We therefore propose that this syndrome is the result of an autosomal recessive end organ receptor defect confined to the smooth muscle of the urinary and gastrointestinal tracts, the upper gastrointestinal distension being caused by continuing function of the oesophageal striated muscle and the urinary tract distension by an inability of the bladder to void urine. The intestinal shortening and malrotation may be the result of 'disuse hypoplasia'. We await further histochemical receptor studies on specimens from our two cases.

We thank Dr Eleanor Allibone for the detailed and painstaking pathology.

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Apple peel syndrome in sibs

SUMMARY We report an Arab sibship of two brothers with apple peel jejunal atresia. The parents are consanguineous. Other reported familial cases are briefly reviewed.

Received for publication 24 February 1988.
Accepted for publication 26 April 1988.