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 hydrenephrosis. 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 malformation 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,8 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 hypoplastasia'. We await further histo-chemical receptor studies on specimens from our two cases.

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

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References


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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.

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In this form of jejunal atresia, because of agenesis of the mesentery, the distal part of the small intestine comes off the cecum and twists around the marginal artery, suggesting a maypole, a Christmas tree, or apple peel at operation. This specific condition has mainly been reported as a sporadic occurrence and rarely in sibs.1-11

In Kuwait, we recently observed another family with apple peel syndrome in sibs.

Case reports

CASE 1
A male infant was born in 1986 after an uneventful term pregnancy, weighing 2800 g. He was the first child of phenotypically normal, distantly related Palestinian parents. At birth he looked normal but he showed signs of intestinal obstruction nine hours later. Laparotomy showed upper jejunal atresia about 7 cm from the duodenojejunal flexure, a mesenteric gap, an absent superior mesenteric artery, the small bowel coiled around a branch of the ileocolic artery, and the presence of a mesenteric cyst. The baby died 60 days after the operation.

CASE 2
A male sib was born in 1987 at 36 weeks’ gestation weighing 2250 g. He was suspected of having bowel atresia after ultrasonographic examination in utero. Laparotomy performed a few hours after birth showed jejunal atresia 10 cm from the duodenojejunal flexure and a corresponding V shaped mesenteric artery with small bowel loops coiled around a branch from the ileocolic artery with a classical apple peel configuration. The child died 30 days after the operation. The parents accepted the high recurrence risk (1 in 4) given to them and decided to have another child. The mother now is in her first trimester of pregnancy.

The reported sibs bring the number of familial cases to 15 (seven families), seven of whom (three families) are Arabs with consanguineous parents. Mishalany and Najjar6 observed three affected out of 16 liveborn offspring of first cousin Lebanese parents, and Al-Awadi et al.10 observed two affected out of three offspring of first cousin Saudi Arabian parents.

We think that such familial occurrences, together with other familial forms of intestinal atresias, reported from Lebanon and Kuwait, whose populations contain a variety of subgroups, are probably the consequence of a high average inbreeding coefficient, a tendency to large family size, and the relative lack of family limitation after genetic counselling in Arabs.

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Note added in proof
The latest pregnancy ended at term with the delivery of another girl with apple peel jejunal atresia; she was operated upon on the first day and died 40 days later.