Megacystis, microcolon, intestinal hypoperistalsis syndrome and bilateral streak gonads

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ABSTRACT

Megacystis, microcolon, intestinal hypoperistalsis is an uncommon condition presenting in neonatal age with features of intestinal obstruction and bladder evacuation abnormalities. We present here an infant girl with the diagnosis consistent with this entity.

Key words: Intestinal hypoperistalsis, megacystis, microcolon

Introduction

Megacystis, microcolon, intestinal hypoperistalsis syndrome is a rare condition predominantly affecting females and characterized by a large urinary bladder and generalized intestinal hypoperistalsis. Most patients would present with features of functional intestinal obstruction. Many other defects like omphalocoele, cardiac lesions, undescended testis, rhabdomyomata have been reported to occur with this condition. We present here an infant with typical features of this uncommon condition along with bilateral streak gonads – a feature that has not been reported previously.

Case Report

A 1-month-old baby presented with a history of progressive abdominal distension. She had hematuria, lethargy, poor feeding, and bilious vomiting for 1 day. On examination she was lethargic and dehydrated (weight 2 kg) with a pulse rate of 132/min, respiratory rate 48/min, and capillary fill time < 3 s. The liver was palpable 2.5 cm below the costal margin, and the bowel sounds were sluggish. Investigations showed leucocytosis with a shift toward polymorphic counts and normal platelets. The blood urea and creatinine were 34 and 0.8 mg/dL, respectively, serum sodium was 135 mEq/L, and potassium 3.6 mEq/L. The CRP was 70 mg/dL and prothrombin time 17 s. The venous blood gas was suggestive of metabolic acidosis. A plain X-ray of the abdomen revealed multiple dilated bowel loops. The urine routine examination showed turbid urine with sheets of pus cells, and the culture grew *Escherichia coli*. A diagnosis of urosepsis with paralytic ileus was made; the child was started on appropriate antibiotics and supportive therapy including catheter drainage.

The baby had been born to a 25-year-old primigravida of a nonconsanguineous marriage after 36 weeks of gestation. The birth weight was 2890 g. An antenatal ultrasound at 35 weeks of gestation had revealed polyhydramnios and a large cystic mass (11 × 10 cm) in the fetal abdomen with nonvisualization of the urinary bladder. A postnatal ultrasound scan on day 2 showed a cystic anechoic structure displacing the bowel loops and bilateral hydroureteronephrosis. A differential diagnosis of the mesentric or ovarian cyst with bilateral gross hydronephrosis was made and surgery was advised but the relatives refused.

During the current admission, sonography showed hepatomegaly (5.5 cm with normal echotexture) and bilateral mild hydronephrosis. The urinary bladder was collapsed with a catheter *in situ*.

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After 1 week of appropriate therapy, the baby continued to have abdominal distension and bilious aspirates. In view of persistent symptoms, a barium enema was done, which revealed a featureless sigmoid colon and rectum with proximal microcolon [Figure 1]. An exploratory laparotomy revealed a large urinary bladder extending up to the xiphisternum [Figure 2]. The colon was small in caliber (microcolon). There were streak gonads on both sides in place of ovaries. A vesicostomy was performed after excising a disc of the bladder wall at the dome. A rectal biopsy was also taken.

The histology of the bladder showed focal denudation of the lining transitional epithelium, transmural edema, vascular congestion, and chronic inflammation. The rectal biopsy showed a normal muscularis propria with splaying of muscle fibers by edema. The nerve bundle and ganglion cells were identified with an increase in elastin fibers and fibrosis in the subepithelial region.

The baby continued to have biliary aspirates and total parenteral nutrition was started. In view of a large bladder and a small colon and persistent biliary aspirates, a possibility of megacystis, microcolon, intestinal hypoperistalsis syndrome (MMIHS) was considered. The parents refused further surgical interventions and decided to take the child home.

Discussion

MMIHS was first described by Berdon *et al*. in 1976.[1] MMIHS is congenital condition characterized by a large bladder, small colon, and intestinal hypoperistalsis throughout the gastrointestinal tract. About 180 cases with female preponderance have been reported worldwide.[2]

The presence of vacuolar degenerative changes in the smooth muscle cells of bowel and bladder suggest that it may be due to a visceral myopathy. Immunohistochemistry staining on a rectal and urinary bladder biopsy specimen show deficiency of cytoskeletal proteins and increased collagen.

Characteristic findings in the antenatal period are the presence of an enlarged urinary bladder with polyhydraminos. An enlarged bladder in the fetal period may occur in posterior urethral valves, neurogenic bladder, and urethral agenesis associated with oligohydraminos. In MMIHS, the kidneys are functionally normal and an increase in the amniotic fluid is probably due to decreased peristalsis of the gastrointestinal tract.

Other associated abnormalities that can occur with the condition are omphaloceles, cardiac malformations, multiple rhabdomyomata, and intra-abdominal testis. However, streak gonads have not been reported till date.

The management of the condition is difficult. Most complications arise due to severe malnutrition consequent to gastrointestinal involvement. Total or partial parenteral nutrition is required. Continuous or intermittent bladder drainage improves the bladder function and may reduce the reflux and associated hydroureteronephrosis. MMIHS is a lethal condition with less than 10% survival beyond 1 year of age. Palliative surgeries like colostomy and feeding gastrostomy are only of marginal benefit. Multivisceral transplantation can provide prolonged survival.[3]

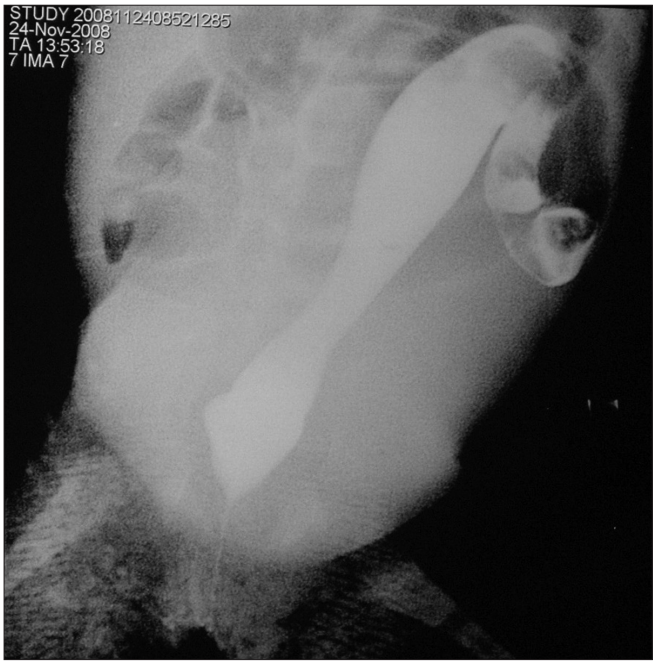

Figure 1: Barium enema showing featureless sigmoid colon and rectum with proximal microcolon

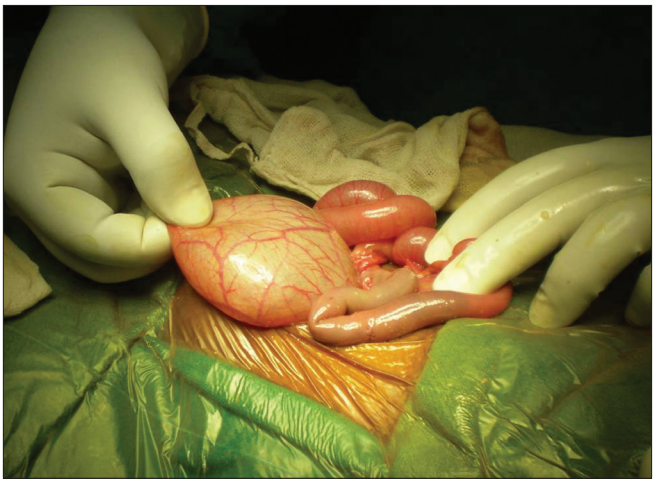

Figure 2: Peroperative findings showing a large, thin-walled distended bladder

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