Megacystis microcolon intestinal hypoperistalsis syndrome: a report of a variant

Ahmed H. Al-Salem

Megacystis microcolon intestinal hypoperistalsis syndrome is a very rare cause of functional intestinal obstruction in newborns. It is associated with nonobstructed distended urinary bladder, microcolon, and decreased or absent intestinal peristalsis. The prognosis is poor and most patients die early because of sepsis or total parental nutrition-related complications. This report describes a new case of megacystis microcolon intestinal hypoperistalsis syndrome associated with meconium ileus, dilated stomach, and megaesophagus. ***Ann Pediatr Surg*** **10:57–60 © 2014 Annals of Pediatric Surgery.**

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Department of Pediatric Surgery, Maternity and Children Hospital, Dammam, Saudi Arabia

Correspondence to Ahmed H. Al-Salem, MD, PO Box 61015, Qatif 31911, Saudi Arabia
e-mail: ahsalsalem@hotmail.com

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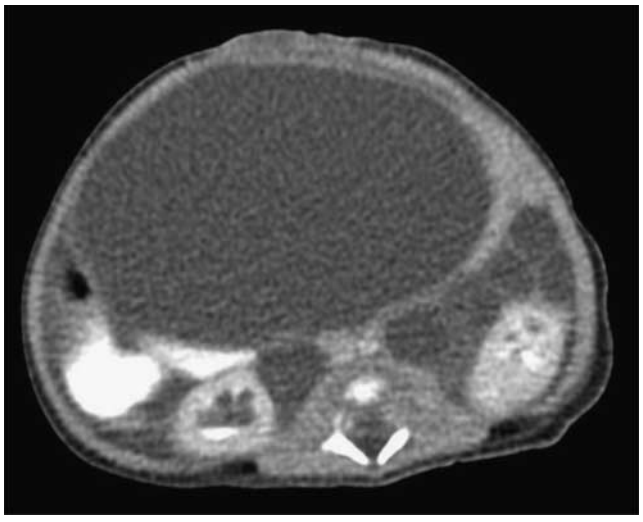
Introduction

Megacystis microcolon intestinal hypoperistalsis syndrome (MMIHS) is a very rare congenital condition of unknown etiology. It is characterized by abdominal distension caused by a markedly distended, nonobstructed urinary bladder, microcolon, and intestinal hypoperistalsis with functional intestinal obstruction [1–3]. The exact etiology of MMIHS is not known, but the most commonly accepted etiology is that MMIHS is a form of visceral myopathy [4]. The main manifestation is intestinal obstruction in a newborn, with other associated abnormalities. This report describes a new case of MMIHS associated with meconium ileus, dilated stomach, and megaesophagus.

Case report

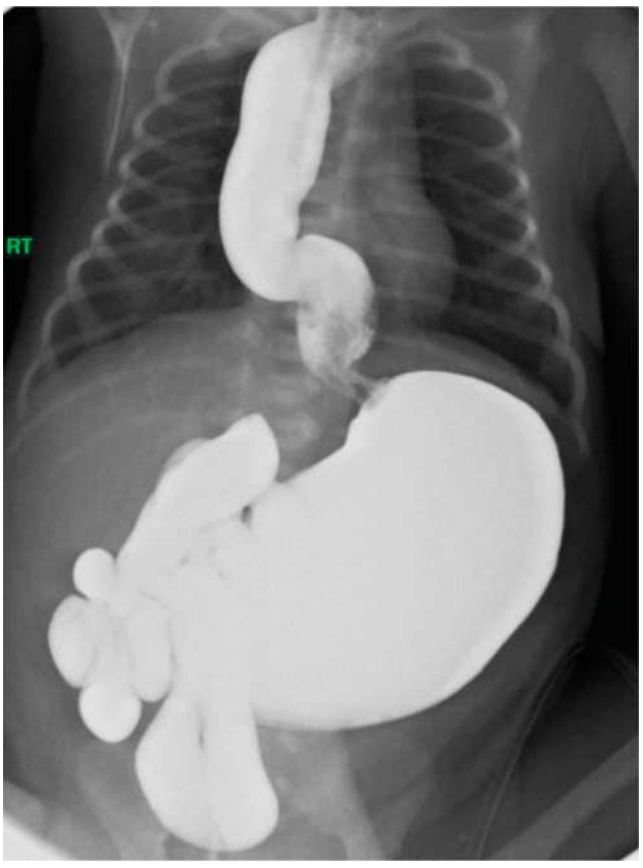
A newborn female was diagnosed with a huge abdominal mass and bilateral hydronephrosis on antenatal ultrasound. At week 35 of gestation, she was delivered by cesarean section. Her birth weight was 2.9 kg and Apgar score was 6 and 9 at 1 and 5 min. She did not pass meconium and clinically she was found to have abdominal distension with a palpable mass filling most of her abdomen. A plain abdominal radiograph showed a dilated stomach with a double bubble sign. Abdominal ultrasound and computed tomography scan indicated a hugely dilated urinary bladder filling most of the abdomen as

Fig. 1



Abdominal computed tomography scan showing a markedly dilated urinary bladder.

Fig. 2



Barium swallow and meal showing a dilated esophagus, stomach, and upper part of the small intestines.

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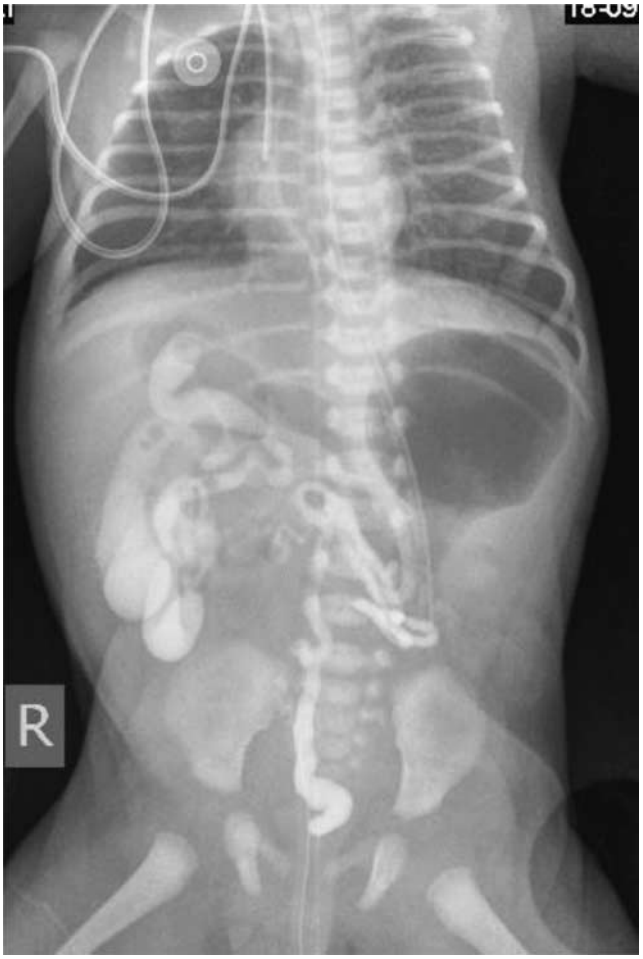
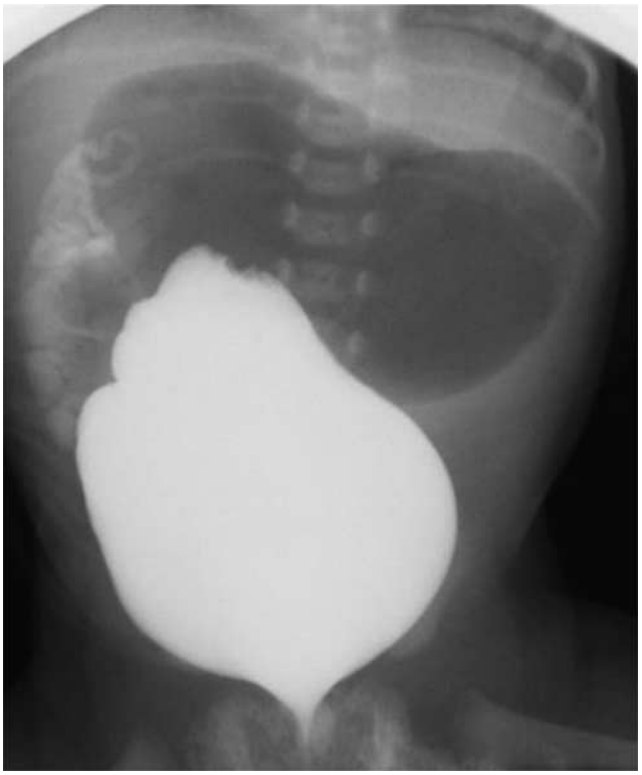
Fig. 3



Barium enema showing small unused microcolon.

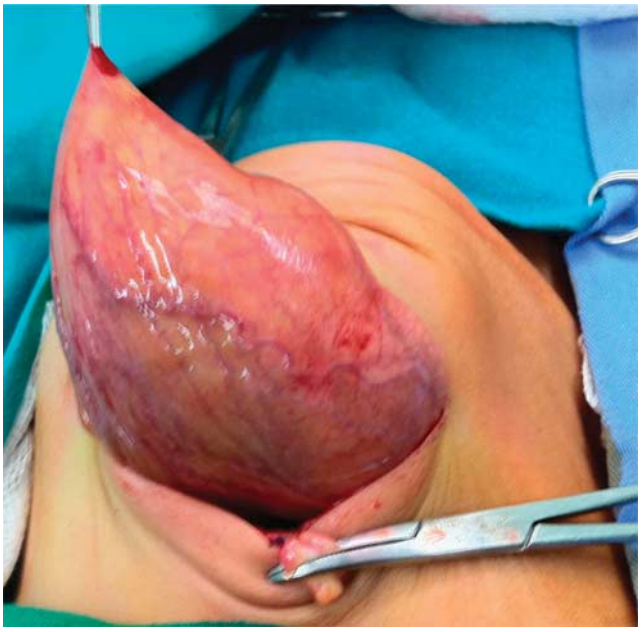
well as bilateral hydronephrosis more on the left side (Fig. 1). A barium swallow and meal indicated dilated esophagus, stomach, and part of the proximal small intestines (Fig. 2). A barium enema showed small unused microcolon and micturating cystourethrogram showed a markedly dilated urinary bladder that was also abnormal in shape (Fig. 3). There was no evidence of vesicoureteric reflux but significant postvoid residual urine. She underwent exploration laparatomy and insertion of a central line. Laparatomy indicated a hugely dilated urinary bladder and stomach (Fig. 4). The proximal small intestines were also dilated, but the colon and part of the terminal ileum were very small and unused. The distal ileum was filled with inspissated meconium. The distal end of the dilated terminal ileum was brought out as an end ileostomy and the distal part was brought out as a mucous fistula. The histology of the resected part of the small intestines and appendix showed congestion, hemorrhage, and prominent lymphoid cells focally in the mucosa and the presence of ganglion cells. Postoperatively, the ileostomy never functioned. She was started on total parental nutrition and several trials of nasogastric feeds were not tolerated and the ileostomy did not function. At the age of 97 days, she died because of sepsis and total parenteral nutrition liver failure (Figs 5–7).

Fig. 4



Micurating cystourethrogram showing a markedly dilated urinary bladder. There was no evidence of vesicoureteric reflux.

Fig. 5

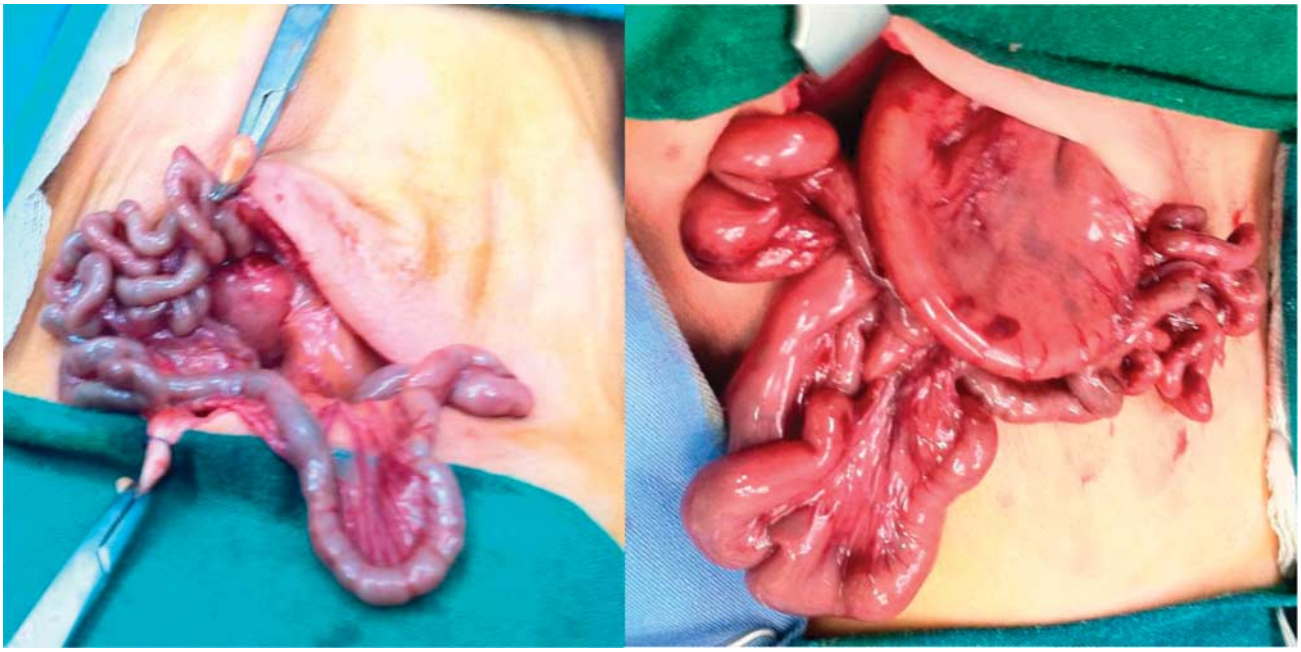


Intraoperative photograph showing a markedly dilated urinary bladder.

Discussion

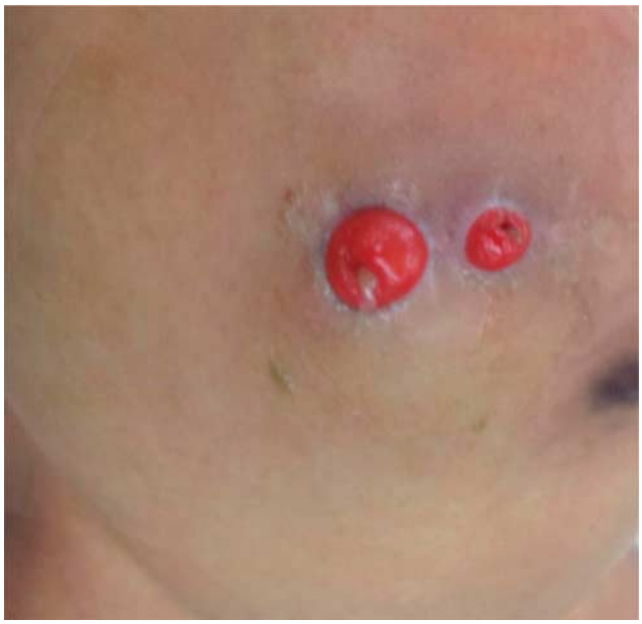
MMIHS was first described by Berdon *et al.* [5]. It is a rare congenital anomaly inherited as autosomal recessive that predominantly affects females (male : female, 1 : 4).

Fig. 6



Intraoperative photograph showing the dilated stomach and proximal upper small intestines on the right side and the distal small intestines filled with pellets of meconium on the left side.

Fig. 7



A clinical photograph showing the two stomas. The proximal stoma never functioned.

In an extensive review of the world literature, Gosemann and Puri [3] found only 227 MMIHS cases over a period of 35 years. MMIHS is characterized by the presence of a markedly distended urinary bladder without distal urinary tract obstruction, microcolon, and decreased or absent intestinal peristalsis [1,2]. MMIHS usually has a fatal prognosis and most of the cases die within the early months of their lives; nevertheless, there are some case reports of longer survival.

The exact etiology of MMIHS is not known. Histological studies suggest that the predominant intestinal manifestation is smooth muscle myopathy [4]. Puri *et al.* [6] suggested that MMIHS may be a degenerative disorder of smooth muscle cells by reporting vacuolar degenerative changes in the smooth muscle cells with abundant connective tissue between muscle cells. Subsequently, they suggested smooth muscle structural abnormalities [4]. Molecular observations have linked the disease to the neuronal nicotinic acetylcholine receptor, namely the absence of a functional $\alpha 3$ subunit of the nicotinic acetylcholine receptor, a de-novo deletion of the proximal long arm of chromosome 15 (15q11.2) [7]. Histological evaluation showed an appropriate light microscopic appearance of both the circular and the longitudinal layers of the small bowel muscularis propria. Immunohistochemical staining for smooth muscle actin, however, was selectively absent in the circular layer, indicating isolated absence in a unique and previously undescribed pattern. These observations raise the possibility that the proximal long arm of chromosome 15 (15q11) may be of clinical significance in MMIHS. MMIHS has been reported as an autosomal recessive disorder with female preponderance [7].

MMIHS is characterized by the presence of an unusually distended urinary bladder, microcolon and absent or decreased peristalsis. Other reported anomalies include malrotation, short bowel, segmental stenosis of the small bowel, a dilated proximal small bowel, bilateral streak gonads, and a bilateral duplicated urinary system [1,3]. In our patient, there was an associated megaesophagus and enlarged dilated stomach as well as an associated

meconium ileus. The distal small intestines were small and filled with thick inspissated meconium, whereas the proximal small intestines were dilated. The colon was classically small and unused. The associated megaesophagus and meconium ileus were not reported before.

There is no definite cure for MMIHS and the majority of patients die within the first year of life [3]. The most frequent cause of death was overwhelming sepsis, followed by malnutrition and multiple-organ failure. There are, however, reports of long-term survivors and these patients are on total parenteral nutrition, which is known to ultimately cause liver failure [8]. In 1999, the first multivisceral transplantation for MMIHS was performed for three patients; one of them was alive with almost complete dietary rehabilitation 17 months after transplant [9]. In 2008, a combined living-related liver and intestinal transplant was performed for a patient with MMIHS and maintained on total parenteral nutrition since birth [10]. Several attempts at multiorgan transplantation or combined liver and intestinal transplant in infants with MMIHS have been met with success. Currently, multivisceral transplantation is the only accepted treatment modality for these patients.

Acknowledgements

Conflicts of interest

There are no conflicts of interest.

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