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Megacystis-Microcolon-Intestinal Hypoperistalsis Syndrome

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We report a case of megacystis-microcolon-intestinal hypoperistalsis syndrome (MMIHS) in a newborn female infant who presented with an abdominal mass, absent bowel sounds, and feeding intolerance with bilious emesis. MMIHS is a rare congenital bowel and bladder defect requiring surgery and chronic total parenteral nutrition in an attempt to sustain life. With few exceptions, it is predominately fatal within the first six months of life. We describe the relevant clinical and radiologic findings with ultrasound correlation of this case followed by a brief review of literature included in the discussion.

Introduction

Megacystis microcolon intestinal hypoperistalsis syndrome (MMIHS) was first described in 1976 by Berdon et al. [1] and has been found predominantly in females with an approximate 4:1 female to male ratio [2]. It is characterized by abdominal distention caused by a grossly enlarged, non-obstructed bladder (megacystis). The small intestine is malrotated and often short and distended followed by a hypoperistaltic microcolon. It is a rare and predominantly fatal disease that presents clinically at birth with bilious vomiting, an overdistended abdomen, and absent or decreased bowel sounds. Further findings include a failure to feed or to pass stool despite many attempts to stimulate bowel activity with various cathartic medications. Initial catheterization often reveals greater than 200 ml of urine followed by a rapid refill of the bladder. Chronic intermittent catheterization is often necessary.

Infants afflicted with this syndrome invariably require extensive abdominal surgery to correct the malrotation, remove adhesions, and perhaps visceral transplantation. They require near constant central venous access to deliver TPN due to a failure to stimulate sufficient bowel function. Secondary liver failure often develops as a result of chronic TPN and gallbladder cholestasis. This is followed by renal failure due to the dysfunctional bladder and repeated urinary tract infections (UTIs). Infants are usually overcome by sepsis and multiple organ failure as a result of these sequelae of events.

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Abbreviations: MMIHS, megacystis-microcolon-intestinal hypoperistalsis syndrome; TPN, total parental nutrition; UTI, urinary tract infection; KUB, kidney-ureter-bladder; ISH, in-situ hybridization; nAchR, n-acetycholine receptor

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Case Report

An eight-hour-old infant female delivered by an uncomplicated cesarean section to a gravida 3, para 3 mother presented with feeding intolerance, bilious emesis, and a distended abdomen. Physical exam revealed a responsive child, with normal physical findings with the exception of a palpable mass in the abdomen with hypoactive bowel sounds. Nasogastric suctioning was performed with the evacuation of approximately 32 ml of bilious fluid.

The abdominal radiograph revealed several mildly dilated loops of bowel within the right upper quadrant with a large soft tissue density occupying much of the left hemi-abdomen which exerted a mass-like effect on the stomach (Fig. 1). Abdominal ultrasound (US) revealed a massively dilated bladder that extended into the left upper quadrant, which corresponded to the mass seen on the frontal abdominal radiograph (Fig. 2). A severe hydrouretonephrosis was seen on the left. Although there was no right ureteral dilation, there was a right ureteropelvic obstruction seen that was causing moderate hydronephrosis. Both kidneys demonstrated marked hydronephrosis (left greater than right) but were normal in size and the parenchyma was of normal echogenicity (Fig. 3).

The bladder was decompressed via catheterization, which revealed 210 ml of urine. After bladder decompression, a follow-up radiograph revealed increased dilatation of proximal loops of bowel without any gas distally (Fig. 4). Afterwards, a Barium enema, upper GI series, and a small bowel follow through were performed (Fig. 5). These demonstrated a duodenojejunal junction (DJJ) distal to the duodenal bulb level and overlying the mid-portion of the spine with a small caliber colon throughout its entirety. This was suspicious for malrotation of the bowel without

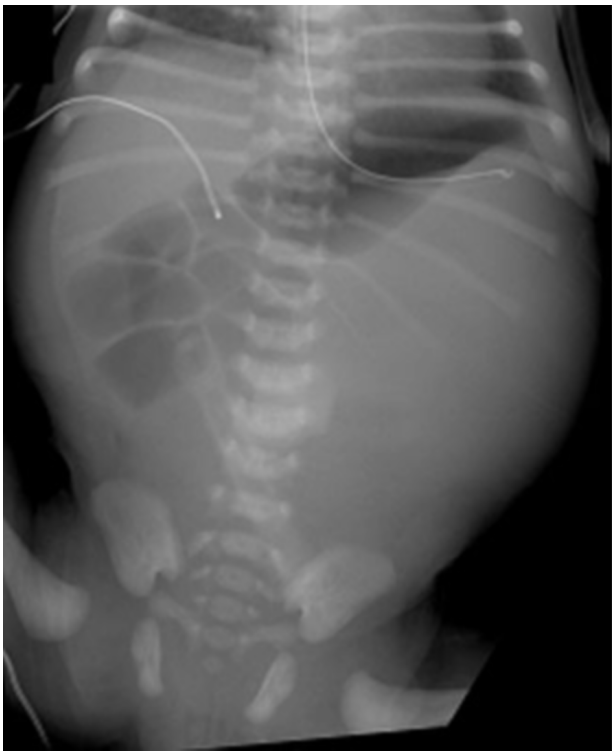

Figure 1. Abdominal radiograph demonstrates a large soft tissue density within the left hemi-abdomen with several loops of dilated bowel displaced towards the right upper quadrant.

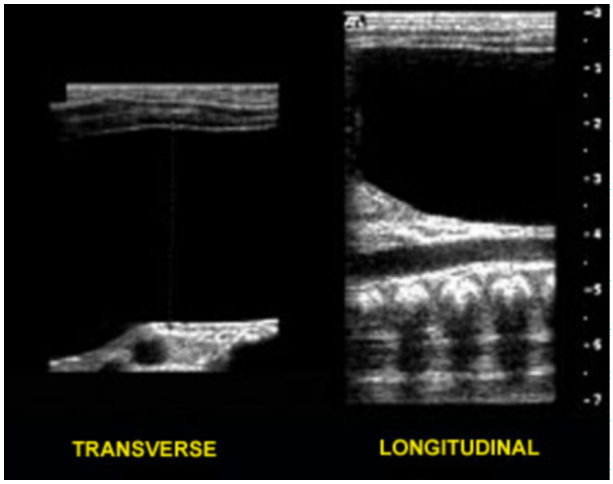

Figure 2. Transverse and sagittal abdominal ultrasound images reveal a massively dilated bladder extending into the left upper quadrant.

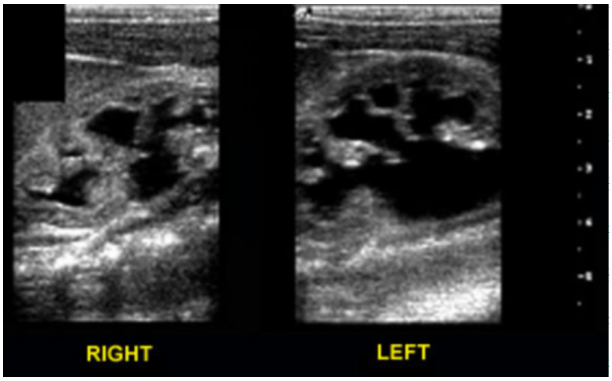

Figure 3. Ultrasound images of both kidneys show moderate hydronephrosis on the right and severe hydronephrosis on the left. Normal parenchymal echogenicity is present in bilateral renal cortices.

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any evidence of a midgut volvulus. However, this study must be viewed with caution due to the dilated loops of bowel which can appear to displace the DJJ below the first part of the duodenum.

On the first day, an exploratory laporotomy was performed for reduction of the malrotation and gastrostomy tube placement. At 20 days of age, a diverting loop ileostomy was performed to bypass the colon to allow for outflow of oral feeds. After discharge the parents decided not to pursue the option of small bowel transplantation and wished to keep their child as comfortable as possible. At six months, the child was jaundiced from TPN cholestasis but had few repeat hospitalizations for major complications.

Discussion

MMIHS is a severe congenital disease of infancy that presents uniformly with abdominal distention secondary to an enlarged, non-obstructed bladder, and failure to feed secondary to a hypoperistaltic bowel. Preferential diagnostic tests include abdominal US, UGI and contrast enema that together with the clinical history provide the diagnosis. Laproscopic examination reveals a shortened small bowel, malrotation, bladder distention, and microcolon. Afflicted patients must endure chronic TPN feedings and intermittent catheterization with a predominantly poor prognosis. The unique feature of the case we present is the ultrasound correlation with the radiographic abdominal mass representing a severely distended bladder or megacystis.

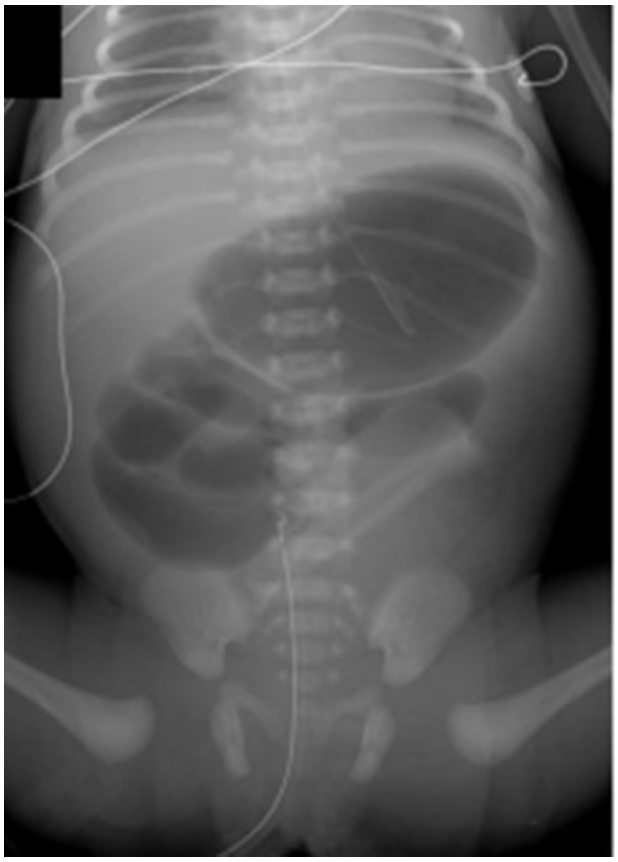

Figure 4. Repeat abdominal radiograph following catheterization that drained 210 ml of urine demonstrates increased dilatation of proximal loops of bowel without any gas seen distally.

In a review of 72 reported outcomes of infants with MMIHS, 61% died within the first six months and 72% within the first year of life [2]. After repeated attempts to feed, TPN related cholestasis, and repeated UTIs, infants are usually overcome by secondary liver failure, renal failure, and septicemia. Since this is such a severe disease of infancy, much has been done to determine its etiology and pathogenicity.

A prevalence exists among MMIHS patients towards female infants with a 4:1 female to male ratio [2]. It has been demonstrated that MMIHS is an autosomal recessive disorder with an increased risk associated with consanguinous parents [3].

The apparent cause of MMIHS is the functional disruption of the bladder and intestinal motility. To determine the cause of this dysmotility, intestinal and bladder biopsies are often taken. Histologic examination has frequently revealed findings consistent with smooth muscle intestinal myopathy [2, 4]. These findings include extensive transmural fibrosis, thinning of the longitudinal smooth muscle layer, connective tissue proliferation within intestinal smooth muscle, vacuolar degeneration, reduced SMA (anti- -smooth muscle-actin) immunoreactivity in the circular smooth muscle layer and absent immunoreactivity in the longitudinal layer [4, 5].

Srikanth et al. [4] proposed that the destruction of visceral smooth muscle and neuronal abnormalities are the initial event that leads to bowel and bladder wall fibrosis. The fibrosis of the bladder results in dysfunctional contractions of the bladder wall against a closed sphincter resulting in bladder distention. They further theorize that the distended bladder eventually interferes with the developmental rotation of the intestine resulting in malrotation.

In contrast to repeated findings of intestinal myopathy, neuronal biopsy results have been inconsistent. Studies of the myenteric and submucosal plexus have revealed

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giant ganglia, hypoganglionosis, and hyperganglionosis, although the majority of ganglion cell findings were normal [2]. This should not imply, however, that the intestinal innervation in MMIHS is normal. In addition to the fact that cholinergics have been shown to be ineffective [1, 2], In-Situ Hybridization (ISH) has revealed a significant difference between acetylcholine receptor (nAchR) subunits of normal intestinal specimens and MMIHS intestinal specimens [6].

Richardson et al. [6] used ISH to examine the expression of the alpha-3 nAchR subunit between 12 control samples of small bowel tissue and similar specimens from 10 patients with MMIHS. ISH revealed that the alpha-3 subunit mRNA was widely distributed in control specimens. However, there was no evidence of the alpha-3 mRNA in the specimens from MMIHS patients. This does much to explain the reason for cholinergic failure as well as display a possible explaination for the smooth muscle myopathy described earlier. Without successful autonomic stimulation, it seems plausible that the enteric smooth muscle cells might not develop sufficient contractile fibers during maturation to maintain a functioning gut. Perhaps with these recent genetic links to MMIHS, the disorder can be detected earlier and followed with interventional strategies that target enteric smooth muscle function and development.

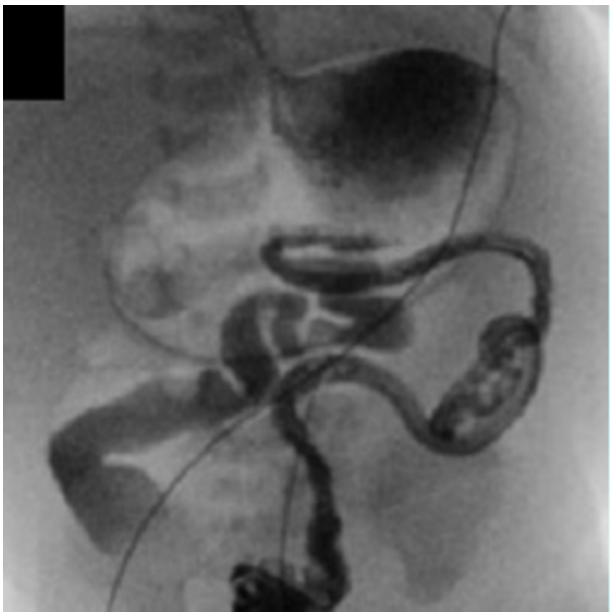

Figure 5. Barium enema with residual contrast seen from upper GI series and small bowel follow through reveal a microcolon with malrotation.

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