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Case Report

Imaging findings of a twin male neonate with megacystis microcolon intestinal hypoperistalsis syndrome ☆

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ABSTRACT

Megacystis microcolon intestinal hypoperistalsis syndrome (MMIHS) is a rare congenital defect of the bowel and bladder that is especially rare in males. We present a case of MMIHS in a male nonidentical twin neonate who presented with abdominal distention, urinary retention, and hypoplastic bowel. The voiding cystourethrogram included in this report displays rare image of MMIHS in a male urogenital system. The constellation of clinical and imaging findings presented in this case are characteristic of MMIHS and may aid the early diagnosis of male neonates affected by this disease.

Introduction

In patients with MMIHS, smooth muscle motility is impaired in the bladder and intestine. Megacystis results from impaired detrusor motility, presenting as urinary retention, poor emptying, and abdominal distension secondary to an overfilled bladder. Microcolon results from impaired intestinal smooth muscle peristalsis, presenting as poor digestion, nutritional compromise, and failure to pass meconium. Many patients remain dependent on catheterization for the bladder and Total Parenteral Nutrition (TPN) for nutrition for life. It occurs predominantly in females, with a 4:1 female to male ratio [1–3].

Our patient presented with a complicated clinical picture at birth, and imaging studies were instrumental in determining his diagnosis. Initial evaluation included renal ultrasound, voiding cystourethrogram (VCUG), barium enema, and fluoroscopic evaluation of the gastrointestinal system. Because this disease is exceedingly rare, particularly in males, we present this case to aid recognition and diagnosis of this disease. Early diagnosis is important for creating treatment plans as well as guiding conversations to counsel the family.

☆ Competing Interests: None.

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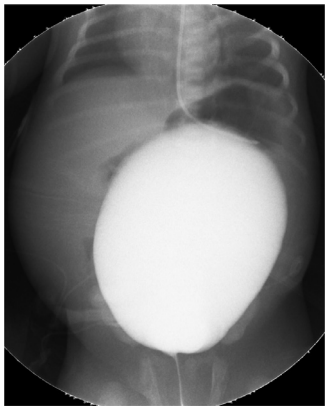

Fig. 1 – Voiding cystourethrogram shows a severely distended bladder with no spontaneous voiding. No vesicourethral reflux.

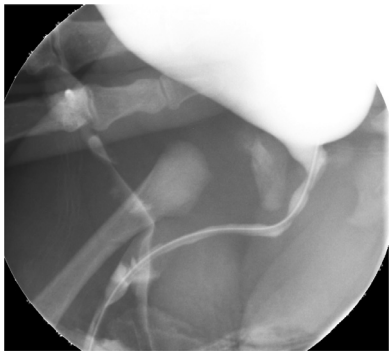

Fig. 2 – Following a Crede maneuver, a normal appearance of the urethra was demonstrated, excluding PUV.

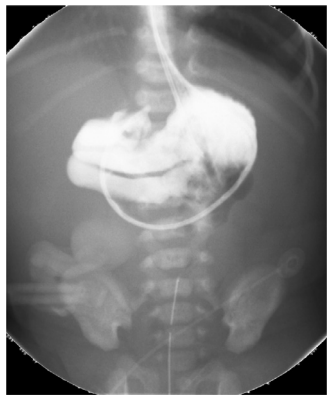

Fig. 3 – Upper GI series shows normal sized duodenum, with low duodeno-jejunal junction. Small bowel peristalsis absent.

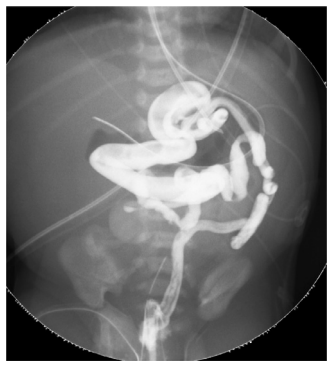

Fig. 4 – Fluoroscopy with contrast enema shows meconium filled microcolon, predominantly situated in the left hemiabdomen. Contrast refluxed only into the distal ileum.

Case Report

A 38-year-old G2P0, with a history of previous miscarriage at 8 weeks, carried a set of nonidentical twins without any major pregnancy complications. On fetal ultrasound, Twin A, the subject of this study, was found to have intermittent hydronephrosis and incomplete bladder emptying. Distended bladder was never noted, but the bladder never emptied completely at any time. Caesarean section was performed at 37 weeks. Delivery was complicated by footling breech. Initial APGAR scores were 4 at 1 minute and 9 at 5 minutes. Renal ultrasound was done to assess abdominal distension, which showed bilateral hydronephrosis and hydroureter with dilated urinary bladder. A urinary catheter was placed, and the bladder was decompressed, yielding 75 mL of urine. Twin B has no medical issues and is currently healthy.

VCUG (Fig. 1) of the patient demonstrated a severely distended bladder filling most of the abdomen, asymmetric to the left. No vesicoureteral reflux was observed. Despite 500 mL of contrast distilled in the bladder, there was no spontaneous voiding. A Crede maneuver was needed to void contrast, and demonstrated a normal urethra (Fig. 2). No posterior urethral valves were seen. The catheter was left in place.

The patient had an unclear history regarding meconium passage and some bile stained return, so both upper and lower fluoroscopic evaluation was performed. An upper gastrointestinal series (Fig. 3) was performed through the nasogastric tube and showed a normal sized duodenum with a low duodeno-jejunal junction. Peristalsis of the bowel was absent. Water soluble contrast enema (Fig. 4) demonstrated a meconium filled microcolon, predominantly situated in the left hemiabdomen. Contrast refluxed only into the distal ileum.

At 3 weeks of age, the patient underwent ileostomy with gastrostomy tube placement to manage GI deficits. Circumcision, bilateral inguinal hernia repair, and right femoral central venous line placement were also done at the time of this surgery. The patient was discharged to home at 2 months old. He was briefly readmitted to the hospital for urinary tract infection caused by proteus mirabilis, which was successfully treated with ceftriaxone. The patient has continued clean intermittent catheterization (CIC) 4 times daily since discharge. At 4 months old, he remains dependent on TPN for nutrition. Genetic analysis demonstrates variant of uncertain significance on exon 8 of the ACTG 2 gene. Follow-up renal ultrasound again shows unchanged, moderate bilateral pelvocaliectasis.

Discussion

Though MMIHS is more common in females, the diagnosis should still be considered in a male newborn who presents with the clinical findings of abdominal distention and inability to void requiring catheterization, which are all exemplified by the male newborn presented in this report [1]. The imaging findings of intermittent hydronephrosis and incomplete bladder emptying on fetal renal ultrasound, large capacity bladder and absence of spontaneous voiding on VCUG, microcolon on fluoroscopy with barium enema, and impaired intestinal peristalsis on fluoroscopic upper GI series, all strongly support the diagnosis of MMIHS [4,5].

The initial differential diagnosis for this patient included posterior urethral valves and Prune Belly Syndrome (PBS). Both PBS and MMIHS should be considered in a male newborn who presents with urinary retention. PBS is also characterized by aplasia of the abdominal wall musculature and cryptorchidism [6]. The absence of these distinguishing features makes PBS a far less likely diagnosis for our patient. Additionally, microcolon and dilated loops of bowel, as seen in our patient, help distinguish MMIHS from PBS [6].

Treatment is largely supportive, with focus on management of functional deficits, primarily bladder decompression, bowel decompression, and adequate nutritional intake. Typical management options for bladder decompression include CIC or surgery to create urinary diversion, such as a vesicostomy [7]. Our patient's urinary retention is managed by CIC 4 times daily, which is commonly preferred for its less invasive nature. Clinical courses are often complicated by recurrent urinary tract infections [4].

Management options for GI symptoms can be categorized into both short-term and long-term goals. Temporizing surgeries such as ileostomy, gastrostomy tubes, and jejunal tubes are common. The only longer-term solution currently available is transplant, ranging from intestinal transplant to multivisceral transplants [4]. Our patient underwent ileostomy with gastrostomy tube placement at 3 weeks of age.

As for nutrition, most patients remain dependent on TPN [1]. Our patient relies on nourishment from TPN with supplements. Per parent reports, occasional drops of sucralose on a pacifier are offered to sooth the patient. Trophic feeds were attempted, but were held due to resulting emesis. Since discharge, ostomy output has been minimal, but slightly increasing. The gastrostomy tube is vented a few times a day, and output has been gradually decreasing. Increased ostomy output and decreased gastrostomy tube output are both signs of increased GI motility. If our patient shows marked improvement, enteral feeds may be attempted again. While prognosis for most neonates involves death in the first year of life, patients with less severe disease variations may live longer [3].

In conclusion, MMIHS is very rare, particularly in males [2,3]. Imaging is important in its diagnosis [5]. Our case is unique in that the newborn is a male of nonidentical twin gestation. A VCUG performed on this patient provides rare imaging of the male urethra in a patient with MMIHS. MMIHS is a probable diagnosis in a female neonate with a distended bladder, and should be evaluated with imaging. This case of a male with MMIHS serves as a reminder that males with this presentation should also be evaluated with imaging, despite the syndrome being markedly less common.

Consent

Patient consent was obtained for the paper, "I have read the information about this case and/or seen the photographs/images to be published. I give my consent for this material to appear in a scientific journal."

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