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Abdominal Radiograph with Intravesical Air and Possible Small Bowel Atresia

Michael S. Stewart, MD,

Section of Neonatology, Department of Pediatrics, University of Colorado School of Medicine, Aurora, Colorado

Robert M. Dietz, MD, PhD,

Section of Neonatology, Department of Pediatrics, University of Colorado School of Medicine, Aurora, Colorado

Matthew P. Landman, MD, MPH,

Department of Surgery, University of Colorado School of Medicine, Aurora, Colorado

Steven L. Moulton, MD, and

Department of Surgery, University of Colorado School of Medicine, Aurora, Colorado

Clyde J. Wright, MD

Section of Neonatology, Department of Pediatrics, Children's Hospital Colorado, University of Colorado School of Medicine, Aurora, Colorado

A male infant of 35 weeks' gestational age was born due to maternal preeclampsia with a prenatal diagnosis of Eagle-Barrett or prune belly syndrome (PBS) based on 19-week fetal ultrasound and confirmatory fetal magnetic resonance imaging. The antenatal course was complicated by polyhydramnios requiring a therapeutic amniocentesis and megacystis requiring vesicocentesis. The infant was born via cesarean delivery with meconium-stained amniotic fluid and had respiratory compromise necessitating intubation in the delivery room. Physical examination was noteworthy for a normal thoracic diameter and shape with abdominal dilation and laxity of musculature. On day of life 5, he developed significant abdominal distention and bilious drainage from his orogastric tube. An abdominal radiograph (Figure 1) showed air in the stomach and proximal duodenum with a paucity of distal gas. There was marked dilation of the bladder, outlined by incidental introduction of air from urinary catheter placement. A contrast enema (Figure 2; available at www.jpeds.com) was performed showing a microcolon concerning for small bowel atresia. Subsequent exploratory laparotomy revealed a massively dilated bladder, complete bowel patency with inspissated meconium, and hindgut malrotation, suggestive of megacystis-microcolon intestinal hypoperistalsis syndrome (MMIHS). Direct injection of a hyperosmotic, water-soluble contrast material into the lumen of the small bowel using a 3-mL syringe and 30-gauge needle softened the inspissated meconium, allowing it to be milked into the large bowel and later passed from below.

Eagle and Barrett characterized PBS as the congenital absence or deficiency of abdominal wall musculature, genitourinary outlet obstruction, and bilateral cryptorchidism in males.¹ The overall incidence is 3.8 per 100 000 live births,² and the pathogenesis remains unknown.

Recessive X-linked inheritance has been proposed due to the predominance of male affected patients. Recently, however, an autosomal recessive, muscarinic acetylcholine receptor mutation was documented in a consanguineous family with a PBS-like phenotype.³

MMIHS is an extremely rare congenital syndrome first reported in 1976⁴ with approximately 200 cases reported in the literature.⁵ It is characterized by abdominal distention, dilated nonobstructed urinary bladder, and microcolon with decreased or absent peristalsis. The pathophysiology and etiology remain unknown. It has a female predominance and a proposed autosomal recessive inheritance due to an increased incidence in consanguineous families.⁶ Treatment is supportive, and multivisceral transplantation has had varying success. Ultimately, MMIHS has a poor prognosis with an average life expectancy of 3 months.⁷

PBS and MMIHS have a phenotypical overlap with a key difference of bladder outlet obstruction in the former. Gastrointestinal anomalies, either functional or anatomic, are commonly observed in both syndromes. Malrotation, intestinal volvulus, small bowel atresia, and anorectal malformations have been reported in patients with PBS.^{8,9} Hypoperistalsis and microcolon are pathognomonic for MMIHS. Without definitive gene testing, clinical differentiation may remain elusive. Therefore, we suggest diagnostic imaging including upper and lower intestinal contrast studies in all patients diagnosed with PBS or MMIHS due to their large clinical overlap.

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Figure 1. Abdominal radiograph shows a “double bubble” with air present in the stomach and duodenum (*white arrow*). The large hypolucent region (*black arrow*) overshadowing these structures is a megacystic bladder with intravesical air incidentally introduced from urinary catheter placement.

Figure 2.
Contrast enema showing diffuse microcolon with contrast filling the appendix (*white arrow*).