Journal of Pediatric Surgery Case Reports 30 (2018) 48–51

Contents lists available at ScienceDirect

Journal of Pediatric Surgery Case Reports

journal homepage: www.elsevier.com/locate/epsc

An atypical case of megacystis microcolon intestinal hypoperistalsis syndrome with extended survival and consistent bowel function

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1. Case presentation

This patient is a baby girl born to two healthy unrelated parents from South America. There is no familial history of consanguinity, and the couple has an older daughter who remains healthy. Early in the pregnancy, the patient's mother declined cell-free DNA analysis and amniocentesis. The patient's anatomy appeared normal on ultrasound at 18–19 weeks timed by last menstrual period. The patient's mother was admitted to an outside hospital at 32 weeks due to subjective contractions and newly diagnosed polyhydramnios and fetal megacystis. The patient's mother was transferred to our institution and underwent amnioreduction of approximately 1 L. She experienced significant improvement in frequency of painful contractions. An amniotic fluid sample was sent for karyotyping, and this showed 46 chromosomes (46,XX) with a balanced translocation of t (4; 7) (q21; p15). A repeat ultrasound performed at 33 weeks showed a normal amount of amniotic fluid, a persistently dilated fetal bladder with bilateral hydronephrosis, and dilated loops of what appeared to be small bowel (Fig. 1). The fetal bladder measured approximately 9.6 cm × 6.4 cm x 7.7 cm (Fig. 1). A normal fetal bladder size at 33 weeks would measure approximately 2.8 cm in sagittal or longest diameter (gestational age in weeks – 5 mm) [1]. A fetal echocardiogram was normal, and fetal activity was reassuring. A repeat fetal ultrasound performed 3 days later showed a slight increase in the dimensions of the fetal bladder, persistent hydronephrosis, and amniotic fluid at the upper limit of normal. The patient's mother was discharged home with close follow-up. A repeat fetal ultrasound performed at 36 weeks again showed amniotic fluid at the upper limit of normal, fetal bladder measuring 10.7 cm × 8.0 cm × 7.3 cm, bilateral hydronephrosis, and dilated loops of small bowel (Fig. 2). Fetal movements were still reassuring at that time. The patient was given a prenatal diagnosis of megacystis microcolon intestinal hypoperistalsis syndrome (MMIHS) based upon her ultrasound findings. Two days later, the patient's mother presented with premature labor and confirmed rupture of membranes.

The patient was born at 35 weeks and 3 days of gestation via emergent C-section, and she weighed 2.94 kg. The patient was cyanotic at first with a heart rate less than 100 bpm and a distended abdomen. With non-invasive positive pressure ventilation and 40% fraction of inspired oxygen, her oxygen saturation rose to 100% by 1 min of life. Her APGAR scores were 4, 8, and 9. The patient passed meconium and voided urine spontaneously within the first few minutes of life. A urinary catheter was placed in the delivery room despite the spontaneous voiding because of the known diagnosis of megacystis. Gross hematuria was observed, but the exact post void residual volume was not recorded. The patient was transferred to the Neonatal Intensive Care Unit (NICU) per protocol for prematurity and anticipated complex course given her diagnosis of MMIHS. High throughput genetic sequencing revealed a heterogeneous missense mutation R257C in the ACTG2 gene, which encodes gamma-2 smooth muscle actin, a fairly ubiquitous visceral intracellular smooth muscle protein. A repeat cytogenetics analysis confirmed the balanced translocation between chromosomal bands 4q21 and 7p15 [t (4; 7) (q21; p15)], but this translocation was interpreted as non-pathogenic.

On her initial lab studies, the patient had a mild coagulopathy with an INR of 1.37, and in the setting of gross hematuria, she received a transfusion of fresh frozen plasma. Her complete blood count (CBC) and chemistry results were unremarkable. Her coagulopathy resolved completely within a few days after birth. A retroperitoneal ultrasound performed on the first day of life revealed severe bilateral hydroureteronephrosis. Echoencephalogram on the second day of life revealed no abnormalities. Due to abdominal distention, a diluted iohexol contrast enema study was performed on the second day of life, and this revealed a small caliber colon throughout its entire length, or “microcolon” (Fig. 3). A voiding cystourethrogram performed on the fifth day of life revealed a trabeculated bladder, but no evidence of vesicoureteral reflux. The patient was transitioned from an indwelling Foley urinary catheter to intermittent catheterization.

The patient's caloric needs were managed with a combination nasogastric, parenteral, and oral feeding. Over the course of the first few weeks of life, she transitioned off of parenteral and nasogastric feedings, and she was taking oral feeds ad lib by the third week of life. During her hospital stay, the patient had persistent hypercalcemia with

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https://doi.org/10.1016/j.epsc.2017.11.010
Received 18 May 2017; Received in revised form 12 November 2017; Accepted 12 November 2017
Available online 21 November 2017

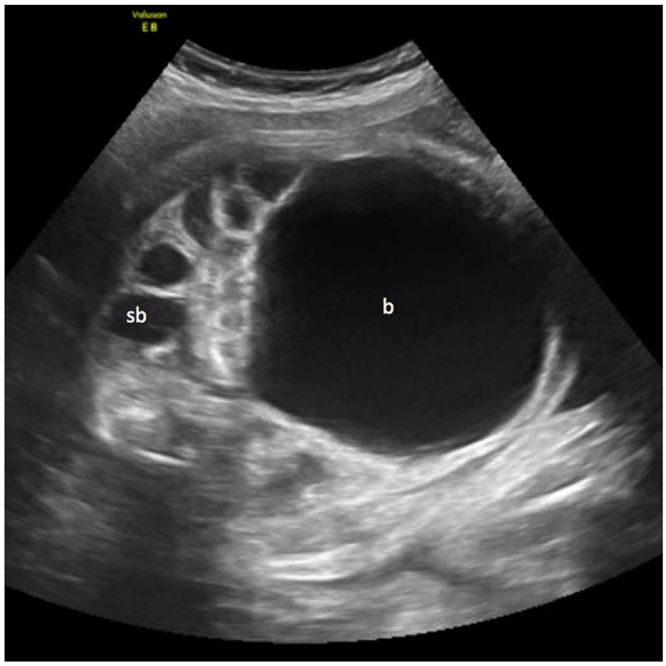

Fig. 1. Prenatal ultrasound at 33 weeks showing megacystis (*b)* and dilated loops of small bowel (*sb)*.

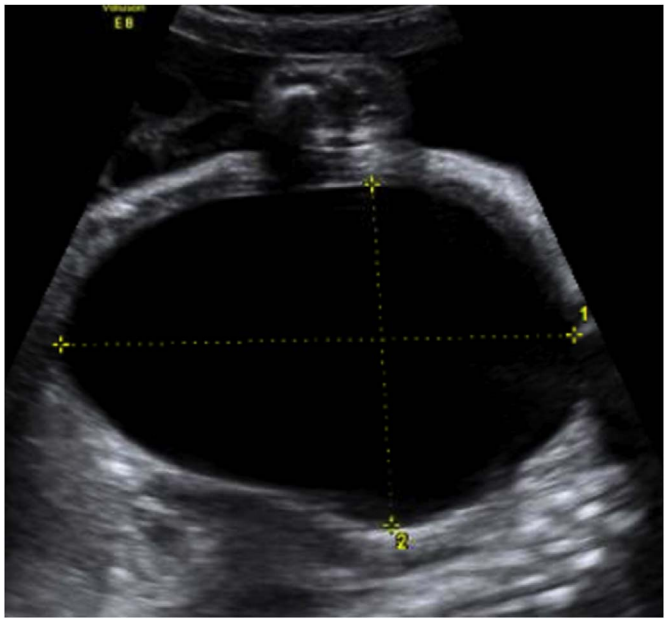

Fig. 2. Prenatal ultrasound at 33 weeks showing severely dilated bladder measured at 9.6 × 6.4 × 7.7 cm.

pseudo-hyperparathyroidism and low vitamin D levels. During the fourth week of life, the patient was discharged to home in good, stable condition with appropriate vitamin supplements. She was having soft frequent bowel movements and tolerating oral feedings well.

After being discharged from her initial hospitalization, the patient was followed by Pediatric Urology, Gastroenterology, Endocrinology, Nutrition, and Primary Care. She continued to have frequent soft bowel movements and required intermittent straight urinary catheterization three times per day. Routine urine cultures grew Klebsiella, Enterobacter, and Enterococcus species over a span of about 4 months, and the patient was treated successfully with oral antibiotics. A follow-up renal ultrasound at 8 months of age showed stable moderate right-sided hydronephrosis, a decrease in the degree of left-sided

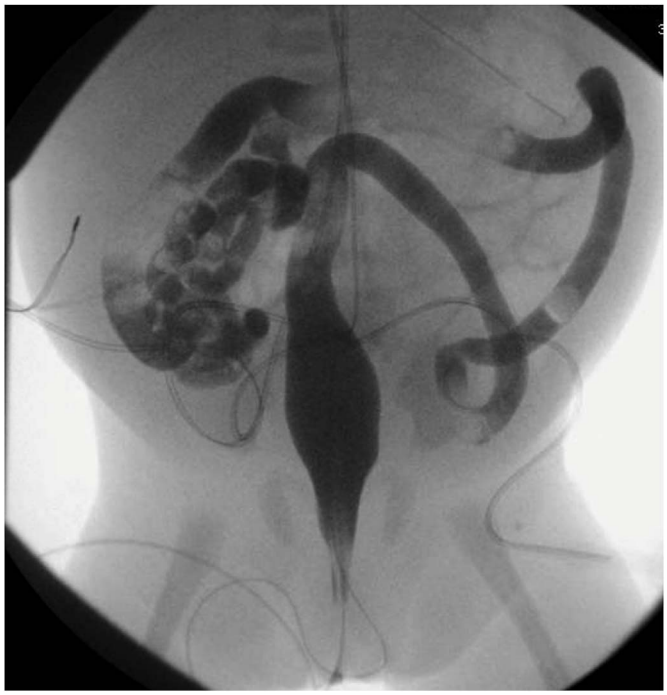

Fig. 3. Hypaque™ enema on day of life 2 showing small caliber of entire colon, consistent with the diagnosis of microcolon.

hydronephrosis, adequate bilateral renal growth, and persistent bladder wall trabeculations The patient also was found to have persistent mild hypercalcemia with elevated parathyroid hormone levels, the exact cause of which has yet to be identified. By 11 months of age, the patient had adequate weight gain, was tolerating formula feeding, and was having frequent soft bowel movements with over-the-counter suppositories used only once per month. She still required straight bladder catheterization several times per day for complete bladder emptying. A barium enema performed at 11 months of age revealed a normal diameter colon throughout its entire length (Fig. 4). At 12 months of age, the patient was in the 20th percentile for weight and 5th percentile for

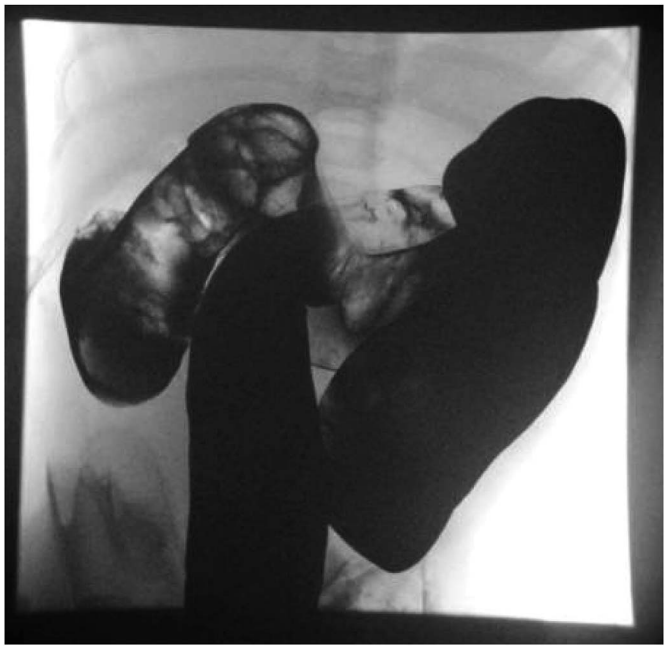

Fig. 4. Barium enema at 11 months of age revealing normal colon caliber, a direct contrast with the initial Hypaque™ images from day of life 2.