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THE MEGACYSTIS – MICROCOLON – INTESTINAL HYPOPERISTALSIS SYNDROME*

Report of a Case and Review of the Literature

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SUMMARY: Yiğit Ş, Barlas Ç, Yurdakök M, Önderoğlu L, Zafer Y, Saltık İ. (Department of Pediatrics, Hacettepe University Faculty of Medicine, Ankara, Turkey). The megacystis-microcolon-intestinal hypoperistalsis syndrome: report of a case and review of the literature. Turk J Pediatr 1986; 38: 137-141.

The megacystis-microcolon-intestinal hypoperistalsis syndrome is part of a spectrum of intestinal motility disorders and is characterized by abdominal distension, lax abdominal musculature, incomplete intestinal rotation, microcolon, megacystis, bilious vomiting and decreased or absent intestinal peristalsis. In this report a newborn girl with megacystis-microcolon-intestinal hypoperistalsis syndrome is reported.
Key words: megacystis-microcolon-intestinal hypoperistalsis syndrome, prenatal diagnosis.

The megacystis-microcolon-intestinal hypoperistalsis syndrome (MMIHS) is a congenital disorder characterized by urinary bladder distension and hypoperistalsis throughout the entire gastrointestinal tract. This uniformly fatal condition appears to be caused by functional disturbances of bladder and intestinal motility, for which a specific cause has not yet been recognized. The syndrome was first described in 1976 by Berdon et al[1]. Since then more than 40 cases with this syndrome have been reported in the medical literature[2], two of which were reported from Turkey[3,4]. We present a new patient with the megacystis-microcolon-intestinal hypoperistalsis syndrome with the aim of considering prenatal diagnosis of this rare syndrome.

Case Report

A twenty-nine-year old pregnant woman was referred to the maternity department of Hacettepe University Hospital at 33 weeks gestational age because of polyhydramnios. There was no consanguinity between the parents, who also had a nine-year-old boy with Down syndrome (47, XY, + 21) Ultrasonographic

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examination showed a large cystic structure in the fetal abdomen (Fig. 1). Because of accelerating polyhydramnios and respiratory distress of the mother, amniocentesis was performed twice to reduce the amniotic fluid volume.

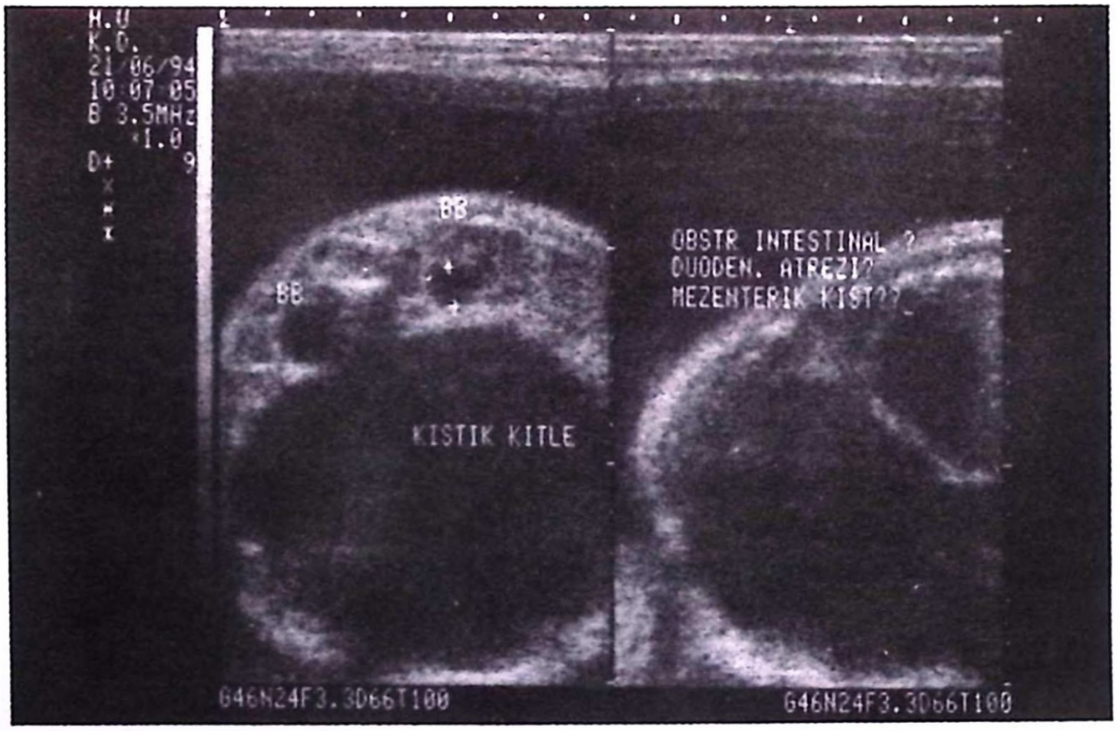

Fig. 1: Prenatal ultrasonographic appearance of large cystic structure in the fetal abdomen.

At 34 weeks gestational age a 2600 g female infant was delivered by cesarean section. The baby was noted to have marked abdominal distension and lax abdominal musculature. Most of the abdomen was occupied by a soft, slightly mobile mass. A plain radiograph of the abdomen showed a dilated stomach and no bowel gas, suggesting a high intestinal obstruction. Ultrasound examination demonstrated that the cystic mass was due to an enlarged bladder. The bladder was catheterized easily, and 700 ml of clear urine was obtained.

Intravenous pyelography and voiding cystourethrogram demonstrated a megacystic bladder without reflux (Figs. 2 and 3). There was pelvicalyceal and ureteral dilatation of the left kidney. Barium enema examination revealed microcolon (Fig. 4). The baby began to vomit bile-stained material on the first day of life. A nasogastric tube drained bilious fluid. No meconium was observed until three days after delivery, although a small rectal tube was inserted. Serum electrolytes, creatinine and BUN were found to be normal.

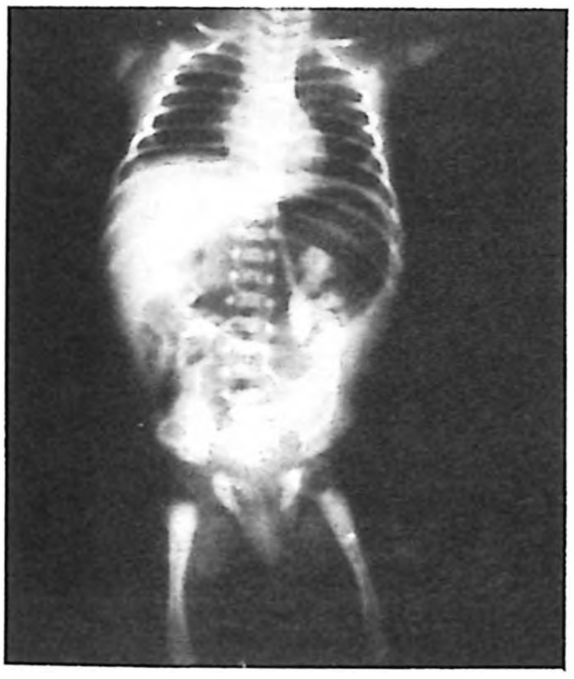

Fig. 2: The intravenous pyelography of the patient showing pelvicalyceal and ureteral dilatation of the left kidney.

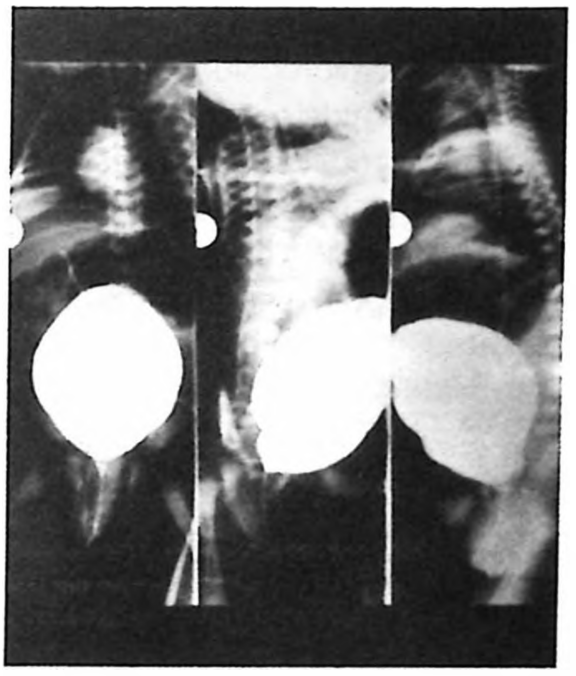

Fig. 3: The voiding cystourethrogram of the patient showing megacystic bladder without reflux.

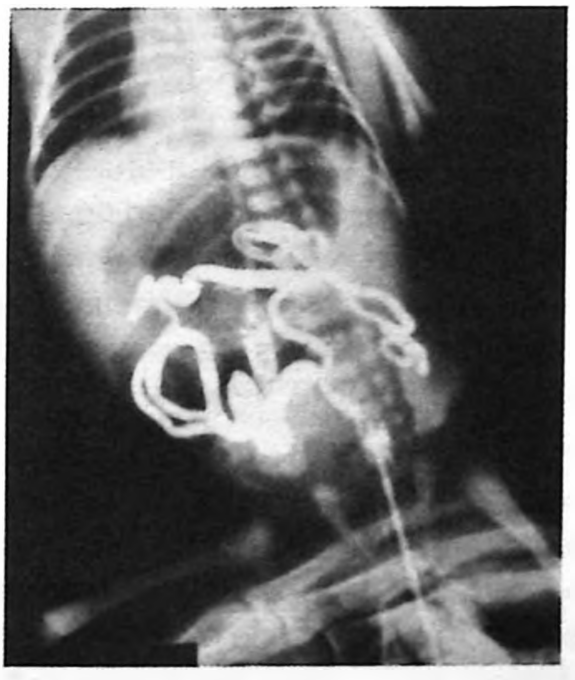

Fig. 4: The barium enema showing microcolon.

All these clinical and radiological findings suggested the diagnosis of MMIHS. Total parenteral nutrition via peripheral veins was initiated. Histological examination of the colon could not be performed. The patient was discharged from the hospital at the parents' request.

Discussion

Megacystis-microcolon-intestinal hypoperistalsis syndrome is part of a spectrum of intestinal motility disorders and is characterized by abdominal distension, lax abdominal musculature, incomplete intestinal rotation, microcolon, megacystis, bilious vomiting and decreased or absent intestinal peristalsis. It is generally incompatible with long-term survival, and death usually occurs within a year due to gram-negative septicemia. Rare cases surviving more than a year have been reported[5, 6]. Although there is a female preponderence, reports of male patients with MMIHS are increasing[7].

The etiology of this syndrome is unknown, but based on families with multiple siblings affected and consanguinity, autosomal recessive inheritance has been postulated[7, 8]. A relationship between maternal drug ingestion (clomiphene citrate, trimethoprim-sulfadiazine, dyprone, scopolamine) and MMIHS has also been suggested[3]. In the case of our patient there was no consanguinity or maternal drug ingestion, but the history of a sibling with Down syndrome was notable.

Similar to some of the cases reported in the literature[1, 5, 9-11], our patient's pregnancy was complicated by polyhydramnios. The etiology of polyhydramnios is unclear. It may be speculated that diminishing reabsorption of amniotic fluid from the intestine due to gastrointestinal dysfunction leads to polyhydramnios.

The pathophysiology and mechanism of the neuromuscular function disturbance in the gut and bladder remain unclear in this syndrome. While earlier reports have mentioned a full complement of normal mature ganglion cells in all parts of the bowel[5], recently inconsistent microscopic findings such as fewer and shrunk neurons in some parts of the bowel have been reported[12]. One of the most recent reports about the pathogenesis of MMIHS suggested that the intial event is an intramural inflammatory process that affects the gastrointestinal and urinary tracts[6]. Another new hypothesis is that MMIHS may be an unusual symptomatic form of neonatal axonal dystrophy[13].

Surgical intervention was not planned for the treatment and diagnosis of our patient. Since MMIHS represents a functional intestinal obstruction, surgical intervention has not been helpful and deaths due to septicemia in the post-operative period are frequent in patients with MMIHS[1, 3-5, 9, 14, 15]. Also, the disorder is resistant to pharmacotherapy[14]. Total parenteral nutrition is necessary for patients with MMIHS because of dysfunction of the gastrointestinal system.

Prenatal diagnosis of this condition by ultrasound gives information for appropriate investigation to be undertaken immediately after delivery. Prenatal ultrasound in subseguent pregnancies might reveal an enlarged bladder in early pregnancy, and a selective termination may be an alternative[2]. Our patient's sonographic examination revealed megacystis, mild left hydronephrosis and polyhydramnios.

Similar findings may lead to the diagnosis of MMIHS. Prenatal diagnosis of this syndrome by ultrasound has been reported as early as 16 weeks of gestation[8]. Therefore, in high risk cases any change in the urinary collective system should perhaps be considered to be a sign of MMIHS.

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