Case Report

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Megacystis microcolon intestinal hypoperistalsis syndrome complicated by perforation

Vishal K. Jain, Saurabh Garge[1], Sangram Singh[1], Brijesh Lahoti[2]

ABSTRACT

Megacystis microcolon intestinal hypoperistalsis syndrome (MMIHS) is a quite rare congenital and fatal disease. The disease is three to four times more prevalent in females than males. Most of the cases die within the early months of their lives. We present the case of a newborn male with antenatal ultrasound suggesting the diagnosis of MMIHS. The case was admitted for perforation complicated with prematurity after birth. This case is the only one where MMIHS presented as perforation.

Key words: Hypoperistalsis, megacystis, microcolon, perforation

INTRODUCTION

Megacystis microcolon intestinal hypoperistalsis syndrome (MMIHS) is a rare autosomal recessive disorder that was first described in 1976 by Berdon *et al.*, in five newborn girls.[1]

Our case fulfils the criteria originally defined by Berdon *et al.*,[1] for the diagnosis of MMIHS i.e. abdominal distension, non-obstructed bladder, microcolon, and normal distribution of ganglia cells in the intestine. We present an antenatally diagnosed male neonate with the syndrome presenting to us with pneumoperitoneum. The sex and the presentation both were peculiar to our case as compared to the available literature.

CASE REPORT

The patient was a preterm male infant, spontaneously delivered at 27 weeks gestation to a 24-year-old mother gravida II, with a birth weight of 1.5 kg. Ultrasound examination at 24week-gestation revealed bilateral renal pelvis and hugely distended urinary bladder. A foetal urination cycle was not shown; therefore, we suspected the absence of muscle tone in the bladder and a diagnosis of MMIHS. The patient did not pass meconium and urine and presented with abdominal distension.

Radiographs taken on the second day showed no gas in the distal bowel or rectum. On the fourth day the neonate developed abdominal distension. A babygram performed showed pneumoperitoneum. An abdominal operation was performed on the same day because of the presence of pneumoperitoneum. Exploration revealed a microcolon, malrotated intestine, short intestinum, and dilated ileum. Mild hydronephrosis was noted bilaterally without hydroureter along with small caecal perforation near the ileoceacal junction [Figure 1]. There was no evidence of mechanical obstruction in the urinary and intestinal tracts. The surgical procedure included a Ladd operation, restoration of incomplete intestinal rotation, appendectomy, bowel perforation

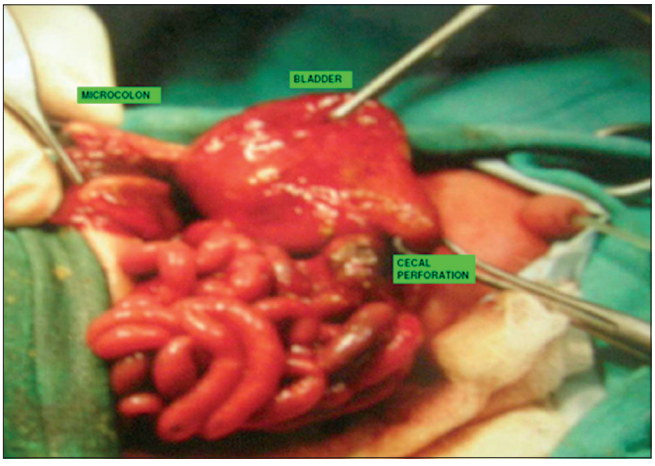


Figure 1: Intra operative photograph showing a male patient with megacystis microcolon and a cecal perforation.

Departments of Surgery and [1]Paediatric Surgery, SAIMS, Indore, [2]Department of Paediatric Surgery, M.Y. Hospital, M.G.M. Medical College, Indore, India.

Address for correspondence:
Dr. Saurabh Garge,
SAIMS, Indore, India.
E-mail: saurabhgarge8@gmail.com

repair and proximal ileostomy and bowel biopsy. On pathologic examination, a full component of normal, mature ganglion cells was shown, and MMIHS was diagnosed on the basis of these surgical pathologic findings and clinical features. The baby died two days after surgery due to sepsis and cardiopulmonary dysfunction.

DISCUSSION

MMIHS, which is a rare reason of neonatal intestinal obstruction, is a congenital disease with high mortality rates.[2,3] It is characterized by hypoperistalsis or aperistalsis of the gastrointestinal system, non-obstructive bladder distension, malrotation, dilate proximal ileum, and colon.[2,3]

MMIHS is part of the spectrum of intestinal motility defects.[2] Autosomal recessive transition has been defined in most publications which report family history and sibling cases.[4-6] The disease has been observed to be three to four times more frequent in girls than in boys. Of the 182 total cases reported only 43 were males with the majority being females.[7] In our case, the patient was a male, which is rare.

Various theories have been advocated by various authors for the pathogenesis of MMIHS, including imbalance between several kinds of intestinal peptides, absence of the interstitial cell of Cajal (ICCs), intestinal myopathy suggested by degenerative vacuolar degenerations, reduction in smooth muscle actin and other contractile and cytoskeletal proteins in the smooth muscle layers of MMIHS bowel, a defect of glycogen-energy utilization. Recently, the role of certain nicotinic acetylcholine receptor (AChR) subunits has also been described in the literature.[7]

Our case was admitted to our clinic because of mass in the abdomen due to globe vesicale and hydronephrosis in antenatal and postnatal ultrasonography (USG). Presentations and surgical findings as given in the review by Puri *et al*., showed that short-bowel syndrome was found in 37 cases, dilated proximal small bowel in 19, segmental stenosis of the small bowel in three, duodenal web in one, and Meckel's diverticulum in one. Malrotation was found in a total of 81 cases.[7] Our case is the first reported presentation of the disease in the form of a caecal perforation. Caecal distension due to visceral myopathy may interfere with the perfusion of its wall, resulting in ischemia, and increased caecal pressure which could lead to perforation. Furthermore, the prematurity may also be associated with a component of necrotizing enterocolitis which may have contributed to the pathogenesis of caecal perforation.

The prognosis is poor and treatment has been shown to be ineffective. Transuterine-transfoetal bladder taps have been used to relieve bladder pressure *in utero*.[7] Recommendations for surgical management include palliative surgery. Considering the poor prognosis, termination of pregnancy may be offered to the patient. Because intestinal dysmotility of MMIHS is difficult to manage and has a high mortality rate, intestinal transplantation is currently the only therapeutic option that would allow these patients to survive.

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