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Megacystis microcolon intestinal hypoperistalsis syndrome presenting as acute intestinal obstruction in 4-days male neonate: A rare case report

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We report a case of megacystis microcolon intestinal hypoperistalsis syndrome in a 4-days old male neonate. The patient presented with abdominal distension due to intestinal obstruction and massive enlargement of the urinary bladder and was misdiagnosed as the mesenteric cyst on ultrasonography abdomen. The aim of this report is to highlight the fact that due to the rarity of this syndrome, it can be missed in the emergency room and a high index of suspicion should be kept for timely intervention.

Key Words: Neonate; megacystis; microcolon; intestinal obstruction; hypo peristalsis.

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Introduction

Described first by Berdon in 1976; megacystis, micro colon intestinal hypo peristalsis syndrome (MMIHS) is a rare congenital condition with unknown etiology [1]. It is characterized by massive non-obstructive urinary bladder distension, micro colon, absent or hypo peristalsis of bowel segment [2]. Many theories had been proposed to explain underlying pathogenesis like the imbalance between intestinal peptides (Interstitial cells of Cajal), vacuolar degeneration of intestinal smooth muscles, abnormality in cytoskeleton

protein and nicotinic receptor subunit defect; but exact mechanism still remains obscure [3]. Most of the patients are managed with supportive treatment and surgical interventions like bowel transplantation whenever possible. However, MMIHS still carry poor prognosis and affected patients usually die within the early months of their lives [4].

Case report

A full term, 2400 g male baby was delivered vaginally to a primigravida mother. The perinatal period was uneventful and breast feed was started soon after birth. Baby had passed urine with adequate stream at the 12 hour of life but did not pass meconium. There was progressively increasing distention of abdomen and development of bilious vomiting during last 12 hours. The baby was referred at

our center on 4th day of life with massively distended abdomen [Fig. 1], not passing faeces and decreased urine output. Initial investigations revealed Hb 14 g, total leukocyte count 11000/mm³, blood urea 44 mg/dl, serum creatinine 0.9 mg/dl and serum sodium 136 meq/dl.



Fig. 1. Male neonate with massively distended abdomen.

In abdominal radiograph multiple air fluid levels were seen with no evidence of pneumoperitoneum and a large cystic mass was occupying almost whole of the abdominal cavity. Ultrasonography (USG) of abdomen revealed a large cystic lesion suggestive of mesenteric cyst along with dilated bowel loops and 1+ free fluid in peritoneal cavity. Nasogastric aspiration drained bilious secretions. Based on above findings, a presumptive diagnosis of intestinal obstruction with the mesenteric cyst was made and surgical laparotomy was planned after initial intravenous fluid resuscitation. On shifting the patient to operation table, urinary bladder was

catheterized. Surprisingly, it drained about 650 ml of clear urine leading to decrease in abdominal distension. On surgical exploration of the abdomen, papery thin urinary bladder was identified [Fig. 2A,B] and urinary catheter was kept *in situ*; which continued to drain clear urine.



Fig. 2. Peroperatively, (A) Massively distended urinary bladder without dilatation of ureters, (B) papery thin urinary bladder after draining urine.

On further exploration, the colon was found of smaller caliber. Biopsies were obtained from the colon and ileostomy was performed. No

other associated gut anomalies were detected per-operatively and the abdomen was closed in layers. Histological examination of resected colonic segment revealed the presence of mature ganglion cells. Computed tomography (CT) scan of abdomen did not reveal any vertebral deformity. Postoperatively, urine output (2.1 ml/kg/hour) was adequate with normal renal function tests and the neonate was managed conservatively by nil per orally (NPO) along with broad spectrum antibiotics. But bilious aspirates reappeared and signs of sepsis developed next day. We could not start oral feeds in view of hemodynamically instability in the baby. The baby continued to deteriorate and got expired on 3rd postoperative day due to septic shock. The clinical autopsy could not be done as parents did not consent for the same.

Discussion

Also known as Berdon syndrome, MMIHS is rare congenital malformation with high case fatality. Although no genetic locus had been identified, but autosomal recessive and dominant pattern of inheritance had been suggested as genetic basis [5]. In a recent review, Halim et al [6] had suggested that mutations in actin gamma 2 (ACTG2) and loss of *leomodin* 1 (LMOD1) genes will lead to impaired intestinal smooth muscle contractility. Female neonates are more commonly affected with reported male to female ratio 1:4. Puri et al [7] had observed only 43 male neonates in their review of 182 affected patients. Screening with USG and fetal magnetic resonance (MR) may prove helpful in antenatal diagnosis. Enlargement of bladder was the most common finding on prenatal USG and can be detected as early as 16th week of gestation [8]. Although no radiological finding is pathognomonic,

prenatal diagnosis is mandatory for optimal counseling and instituting early postnatal treatment of MMIHS. Abdominal enlargement can lead to difficult labor; sometimes Caesarean section may be indicated. However, our case was male neonate and delivered vaginally with no evident clinical finding at the time of birth.

MMIHS can involve any organ system, but genitourinary and gastrointestinal systems are the most commonly affected. Clinically presentation is similar to features of the intestinal obstruction [9]. Abdominal distension with bilious vomiting is most frequently reported finding. Non obstructive distension of urinary bladder may also contribute in abdominal distension. Other associated anomalies like malrotation, short bowel, a dilated proximal small bowel, bilateral streak gonads, and a bilateral duplicated urinary system had been described [9]. MMIHS may have overlapping clinical features with Prune Belly syndrome (PBS), but the early differentiation is required between the two owing to different treatment approach and prognosis. As in index case, failure to pass urine was not evident from birth. After draining the urine by catheterization, bladder was papery thin which rules out antenatal obstructive lesions like posterior urethral valve (PUV). On abdominal radiograph, we failed to find any abnormality in vertebral column which rules out neurogenic cause leading to bladder obstruction. Another unique feature of our case was absence of other anomalies like malrotation, hydronephrosis and dilatation of ureters.

Radiological investigations can be helpful in suggesting the diagnosis. An abdominal radiograph may show dilated bowel loops of massively distended cystic structure (urinary bladder) displacing bowel loops towards one

side in abdominal cavity. Ultrasonography (abdomen) can reveal anatomy of involved organs better and can also pick up other associated anomalies like hydronephrosis and dysplastic kidneys. Barium studies can be performed to rule out micro colon and malrotation of gut. In index case due to less suspicion, massively distended urinary bladder was misdiagnosed as mesenteric cyst. As there were obvious signs of intestinal obstruction, we had not performed barium contrast studies laparotomy was planned. CT/MR can be helpful in defining the anatomical details better than USG. As general condition of index case was rapidly worsening and preliminary investigations were in favor of intestinal obstruction, so we could not shift the baby for CT scan or MRI abdomen.

The available therapeutic options for MMHIS are prokinetic drugs, total parenteral nutrition (TPN) due to intestinal hypo motility, repeated bladder catheterization and surgical interventions like multi-organ transplantation, but none of them is curative [4,7]. However with use of TPN, metabolic complications, hepatic failure and overwhelming sepsis are common which may prove fatal. Bladder function may not improve after transplantation and patient may need clean intermittent catheterizations [10]. Palliative procedures like ileostomy or colostomy can be done in some cases to decompress the intestine.

In conclusion, MMIHS is a lethal congenital malformation presenting with features of intestinal obstruction and dilated bladder. One should aware of presentations for timely diagnosis and appropriate treatment. Importantly, urinary bladder catheterization should always be performed before laparotomy in every neonate with intestinal obstruction and abdominal distension.

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