

MEGACYSTIS-MICROCOLON-INTESTINAL HYPOPERISTALSIS SYNDROME - report of a very rare pathology in a neonate

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ABSTRACT

We are reporting a baby who presented with features of neonatal intestinal obstruction, with an x-ray picture suggestive of hold up at the duodenal level. On laparotomy he had malrotation of the gut which was corrected. He also had a dilated urinary bladder, which later on was treated with vascostomy. The baby did not move his bowel inspite of no mechanical obstruction and even bypass procedure (gastrojejunostomy). She had all the features of Megacystis-Microcolon-Intestinal Hypoperistalsis Syndrome (MMIHS).

MMIHS (also called hollow viscus myopathy) is a neonatal condition causing a severe form of pseudo (functional) intestinal obstruction. This is an autosomal recessive disorder more common in females. The various components of the syndrome are dilated urinary bladder, decreased motility of the gut, microcolon, intestinal malrotation and lax abdominal musculature. All these components of the syndrome were found in our case.

KEY WORDS: Megacystis-Microcolon-Intestinal Hypoperistalsis Syndrome, Intestinal motility disorder

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INTRODUCTION

Megacystis-microcolon-intestinal hypoperistalsis syndrome is a functional intestinal obstruction of neonates and infants char-

acterized by decreased motility of the gut, microcolon, megacystis (enormously dilated urinary bladder). It is also called hollow visceral myopathy. Other components are malrotation of the gut and lax abdominal wall. The condition is more common in female babies.

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CASE REPORT

A 3 days old female baby presented with bilious vomiting, non-passage of meconium and upper abdominal distension since birth. On examination the baby was otherwise normal and maintaining her vitals. There was fullness in the epigastrium.

On rectal examination the anal canal was found patent with no evidence of meconium. X-ray abdomen showed a double bubble sign with the rest of the abdomen found gasless suggestive of duodenal obstruction. On exploration she was found to have intestinal malrotation with bands of Ladd's. The distal patency

of the gut was confirmed by instilling saline. Urinary bladder was found enormously distended. A Ladd's procedure was performed correcting malrotation and a small feeding tube was left in the bladder for decompression. After the procedure she did not pass stools even after 7 days. Postoperative x-rays showed an enormously distended stomach with no evidence of gas in the rest of the abdomen. Mic-turating cystourethrogram showed a dilated bladder with no evidence of bladder outlet obstruction. Barium enema showed microcolon. The patient was re explored on the 7th postoperative day. A hugely dilated stomach was found with no mechanical obstruction as confirmed by instilling air into the gut. The urinary bladder was again found very much distended. Once again she did not pass any stools after another 7 days. X-ray showed a dilated stomach shadow with no gas lower down. So a third laparotomy was performed. There was some adhesive obstruction that was corrected by dividing the adhesions. Patient did not pass stools even after a fortnight of previous exploration. Repeat x-ray showed again a gasless abdomen with enormously dilated stomach shadow. So the baby was explored for the 4th time. Again no mechanical obstruction was found and a gastrojejunostomy was performed as a bypass procedure. And vasicostomy was done to divert the urine because there was persistent megacystis. Specimens taken from the stomach and the jejunum for histopathology showed evidence of ischemia. Ganglion cells pattern was found normal. Total parenteral nutrition could not be administered due to lack of facility and the baby died on the 40th day of life.

DISCUSSION

Megacystis-Microcolon-Intestinal Hypoperistalsis Syndrome (MMIHS) was first described by Berdon et al in 1976¹. Also called as hollow visceral myopathy², it affects female babies more often than males (4:1)³. The pattern of inheritance is autosomal recessive. It is a rare congenital condition causing severe form of functional intestinal obstruction in the

newborn. MMIHS is characterized by abdominal distension due to distended but non-obstructed urinary bladder, microcolon, decreased or absent intestinal peristalsis and malrotation of gut.⁴

Although more than 100 cases have been reported in the literature, the etiology of this syndrome is not yet fully understood.⁵

The characteristic feature of MMIHS is hypoperistalsis in the presence of ganglion cells. In one study it has been proposed that perhaps the initial event in the pathogenesis of MMIHS is an intramural inflammatory process that affects the gastrointestinal and urinary tracts. This leads to extensive fibrosis, which destroys the intestinal neural network, producing hypoperistalsis. The same process causes neuromuscular incoordination in the bladder wall, resulting in irregular bladder contractions against a "closed sphincter" leading to bladder distension (megacystis). The enlarged bladder then interferes with the rotation of the intestine causing malrotation.⁶

It has also been observed that marked reduction of contractile and cytoskeleton proteins in Smooth Muscle Cells combined with reduced expression of intramuscular interstitial cells of Cajal in the gut may be responsible for the motility dysfunction in MMIHS.⁷ Detailed histo-immuno- and ultrastructural pathology assessment has revealed neuronal dysplastic changes associated with increased laminin and fibronectin.⁸

Due to severe motility disorder of the gut there is absence of stool in the colon. Surgical bypass procedures for the intestine are usually not effective but the associated megacystis usually responds to drainage procedures.⁹ The mortality is reported 87% and those who survive are dependent upon parenteral nutrition.¹⁰

Our patient was also a female baby who presented in the early neonatal age with symptoms of intestinal obstruction. She had all the components of the syndrome along with malrotation of the gut. On first exploration the malrotation was corrected with the hope that all is well. As the baby did not pass stools, second and later on third exploration was done

with the idea to divide any adhesions that caused gut obstruction. The fourth exploration was done to do a bypass procedure as the dilated stomach persisted. When even the bypass procedure did not work we got alarmed that perhaps we are dealing with some severe functional obstruction. We went into the literature and found all the components of the syndrome were present in this patient.

There was megacystis, which responded to drainage operation. There was severe hypomotility, which even did not respond to bypass procedure. There was microcolon. Stools were never found in the colon inspite of no mechanical obstruction. The final diagnosis in our case was delayed but even with early diagnosis surgery has little role. The condition is generally fatal with a high mortality, no matter what treatment is offered.^{4,10} Total parenteral nutrition does give some hope and children have been seen getting better with TPN and living up to 11 years of age⁸ but the overall mortality is very high.¹⁰ Though we have no facility of total parenteral nutrition for the newborns but we did administer partial parenteral nutrition. Our patient succumbed at the 40th day of life. She had lost most of her body fat and died because of overwhelming sepsis.

REFERENCES

1. Berdon WE, Baker DH, Bland WA. Megacystis-microcolon-intestinal hypoperistalsis syndrome: a new case of intestinal obstruction in the newborn. Report of radiologic findings in five new born girls, *Ann Roentgenol* 1976;126:957.
2. Puri P. Hirschsprung's Disease. Clinical and experimental observation. *World J Surg* 1993;17:374.
3. Puri P, Tsuji M. Megacystis-microcolon-intestinal hypoperistalsis syndrome (a neonatal hollow viscus myopathy). *Pediatr Surg Int* 1992;7:18.
4. Rolle U, O'Briain S, Pearl RH: Megacystis-microcolon-intestinal hypoperistalsis syndrome: Evidence of intestinal myopathy. *Pediatr Surg Int* 2002;18:2-5.
5. Puri P. Megacystis-microcolon-intestinal hypoperistalsis syndrome. *Hirschsprung's Disease and Allied Disorders* in, Holschneider W, Puri P (ed2). Harwood, Amsterdam, The Netherlands, 2000, pp 185-95.
6. Srikanth MS, Ford EG, Isaacs Jr H, Mahour GH. Megacystis microcolon intestinal hypoperistalsis syndrome: Late sequelae and possible pathogenesis. *J Pediatr Surg* 1993;28(7):957-9.
7. Piaseczna Piotrowska A, Rolle U, Chertin B, Caluwé D, Bianchi A, Puri P. Alterations in smooth muscle contractile and cytoskeleton proteins and interstitial cells of Cajal in megacystis microcolon intestinal hypoperistalsis syndrome. *J Pediatr Surg* 2003;38:749-55.
8. Ciftci AO, Cook RC, Velzen DV. Megacystis microcolon intestinal hypoperistalsis syndrome: Evidence of a primary myocellular defect of contractile fiber synthesis. *J Pediatr Surg* 1996;31(12):1706-11.
9. Teitelbaum DH. Hirschsprung's disease and related neuromuscular disorders of intestine. In: O'neill JA. *Pediatric Surgery* 5th Edition. USA Mosby -Year Book, Inc. 1998;1417-8.
10. Goldberg M, Pruchniewski D, Beale PG, et al: Megacystis-microcolon-intestinal hypoperistalsis syndrome. *Pediatr Surg Int* 1996;11:246.