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ORIGINAL ARTICLE

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Megacystis – microcolon – intestinal hypoperistalsis syndrome

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Abstract Two non-related female neonates with the megacystis-microcolon-intestinal hypoperistalsis syndrome are described. One presented with a family history of a similar condition. In one child no intestinal peristalsis was observed, while in the other decreased peristalsis with occasional rectal evacuation occurred. Both had fatal outcomes. These two cases demonstrate the wide variation in the clinical course of this rare and fatal syndrome.

Key words Megacystis – microcolon – intestinal hypoperistalsis syndrome · Family history · Female cases

Introduction

The megacystis – microcolon – intestinal hypoperistalsis syndrome (MMIHS) is a rare cause of functional intestinal obstruction in the newborn that was first described by Berdon et al. [1] in 1976. Puri and Tsuji [2] reviewed all 59 cases of MMIHS reported to the date of their study in 1992.

The syndrome is characterized by the following findings: (1) a distended, non-obstructed bladder (in some instances with associated hydroureter and hydronephrosis); (2) a microcolon (an unused colon); (3) malrotation; and (4) decreased or absent intestinal peristaltic movement. This report describes two new cases, both in girls, who shared the same morphologic characteristics of the syndrome but had differing clinical courses.

Case reports

Case 1. A female baby was born at term following a normal gestation and delivery with a birth weight of 2.9 kg. She was the first child of young, healthy parents. A single meconium stool was passed immediately after birth, followed by abdominal distension and bile-stained vomiting. No intestinal peristalsis could be heard; most of the abdomen was occupied by a mass, later found to be a dilated bladder. Plain abdominal radiographs showed isolated dilated small-intestinal loops in the upper abdominal quadrants with opacification of the central and lower segments. An upper gastrointestinal (GI) contrast study demonstrated the presence of intestinal malrotation with gross delay in the onward passage of contrast. A presumptive diagnosis was made of intestinal obstruction associated with and due to intestinal malrotation.

At laparotomy, a large bladder was found causing displacement of other abdominal organs. A small-intestinal malrotation without volvulus and not causing intestinal obstruction was present. A microcolon and bilateral mild hydronephrosis was detected. No intestinal peristaltic activity was seen during the operation. An appendectomy was done, accompanied by repositioning of the bowel in the nonrotated position, by opening the C-loop of the duodenum and placing the cecum in the left hypochondrium. A catheter emptied 0.5 l clear urine from the bladder. On the basis of the clinical, radiographic, and operative findings, a diagnosis of MMIHS was made.

Following the operation, the baby was treated by total parenteral nutrition (TPN), cisapride, and an indwelling urinary bladder catheter. She died at the age of 1 month due to TPN catheter-related sepsis. During this period the patient's intestine remained obstructed; no clinical evidence of bowel motor function was demonstrated.

Case 2. A female baby born at term with a birth weight of 3.2 kg was the second child of a father whose first child died in similar circumstances shortly after birth. On physical examination a very distended bladder was found. A urinary catheter was inserted and drained 0.5 l urine.

An emergent IV urogram revealed bilateral hydroureter and hydronephrosis with an associated large bladder. The urinary catheter was removed within 4 days and the baby managed to pass urine spontaneously, although she showed some degree of urinary retention.

Over a period of 2 months following delivery, the baby developed feeding difficulties associated with abdominal distension and the evacuation of occasional small-sized stools. Because of this and during an episode of gross abdominal distension, an emergency colostomy was carried out on the premise that short-segment Hirschsprung's disease (HD) was present. However, rectal biopsies disproved this possibility as mature ganglion cells were seen, and the colostomy was subsequently closed. Following this procedure the patient developed intestinal obstruction. A barium enema study was normal, and an upper GI contrast study with follow-through showed that the second and third parts of the duodenum were dilated and that very slow progression of contrast material occurred beyond this area into the small intestine. However, over a number of days it was established that the intestinal tract was not mechanically obstructed.

Parenteral feeding was initiated and because of ongoing intestinal obstruction the patient was resubmitted to surgery, during which a

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gastrotomy, a Santulli ileostomy, and an appendectomy were carried out. A Broviac catheter for TPN was placed. Following this procedure, an intensive bowel washout program was initiated. Pharmacological manipulation of intestinal peristaltic activity by cisapride proved ineffectual. However, gastrostomy tube drainage gradually decreased and occasional stools were passed spontaneously. Oral feeding was initiated in a graded manner and the child was able to leave the hospital for a period of 3 months, during which time she gained 1.5 kg while maintained on oral feedings only. She was readmitted after this period with intestinal decompensation that was progressive in nature. She developed complete intestinal obstruction and died some 2 months later at the age of 7 months as a consequence of catheter-related sepsis and associated progressive deterioration in her renal function.

Discussion

The MMIHS involves two organ systems. In the urinary tract a megacystis without demonstrable mechanical cause is present, which may be seen on sonography (US) prenatally. Upper tract dilatation involving the renal collecting systems is also encountered. In the GI tract a microcolon (unused colon) is noted, accompanied in some cases by malrotation of the intestine. An absence of intestinal peristalsis or hypoperistalsis results in functional intestinal obstruction. Thus, the clinical presentation and findings in these cases are related to the functional obstruction involving the bladder and bowel.

In establishing the diagnosis of the syndrome, the clinical presentation, physical examination and findings, combined with the findings noted on abdominal US and an intestinal contrast study confirm its presence. Most of the reported patients have been girls. A family history is present in many instances; this has suggested autosomal recessive inheritance, which indicates a chromosomal abnormality. The pathogenesis of the syndrome remains obscure. A variety of abnormalities that involve the nerve supply of the bladder and intestine and that resemble HD have been described in patients with MMIHS. Focal hypoganglionosis [3], an increase in the number of nerve fibers in the myenteric plexus, and the presence of scarce, shrunken, or immature ganglion cells have been reported. However, most histopathologic specimens show normal-appearing nerve-cell and fiber components.

The interpretation of findings such as axonal dystrophy of the entire nervous system [4] and intestinal neuronal dysplasia [5] is not clear. In vitro studies on specimens obtained from the intestinal tract have shown decreased autonomic inhibitory input to intestinal smooth muscle [6] and an imbalance of gut peptides [7]. As can be expected, manometric studies [8], have demonstrated disturbed motility of the small intestine. In a more conclusive study during which electron microscopy of specimens was used by Puri et al. [9], degenerative changes involving smooth-muscle cells with an increase in connective tissue between cells was seen. These authors regard MMIHS as a visceral myopathy based on these findings. It has also been suggested that MMIHS could be placed together with prune-belly syndrome, although intestinal motility in the latter is always adequate. A recent study [10] proposed that the pathogenesis could be related to an intramural inflammatory process affecting the GI and urinary tracts, resulting in fibrosis and destruction of the intestinal neural network, followed by hypoperistalsis. This neural insult would cause bladder distention, which interferes with the rotation of the intestine.

Most patients with MMIHS die within the 1st year of life. Long-term survivors are dependent upon parenteral nutrition. It is stressed that patients with this syndrome show a spectrum of decreased peristaltic activity; completeness of the intestinal obstruction is therefore, in exceptional instances, variable, as illustrated by the second case reported in this paper. It should be noted that the intestinal hypomotility has been reported to improve with passage of time [11].

No effective long-term pharmacological treatment is currently available. Surgical procedures performed on the intestine have not been successful, and most authors regard them to be contraindicated. Intestinal transplantation, once it has become a clinically viable option in early life, will be the therapeutic route to follow in the future for patients with this syndrome.

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