Isolated congenital megacystis without intestinal obstruction: A mild variant of chronic intestinal pseudoobstruction syndrome?

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Isolated congenital megacystis without intestinal obstruction: a mild variant of chronic intestinal pseudo-obstruction syndrome?

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Abstract

Megacystis is frequently involved with chronic intestinal pseudo-obstruction syndrome; however, isolated megacystis without intestinal obstruction is extremely rare. We present the case of a female patient with isolated congenital megacystis without severe intestinal obstruction. In this case, barium enema did not reveal any significant findings; however, histological evaluation of her rectum showed hypoganglionosis of the submucous and myenteric plexuses. These findings indicate that this case may be a mild variant of chronic intestinal pseudo-obstruction syndrome. The presence of megacystis should alert the physician to the possibility of chronic intestinal pseudo-obstruction syndrome.

Megacystis microcolon intestinal hypoperistalsis syndrome (MMIHS) was originally described in 1976 as a cause of intestinal obstruction in newborns [1]. It is a severe subtype of the chronic intestinal pseudo-obstruction syndrome, a rare clinical entity in which recurrent symptoms of intestinal obstruction are present in the absence of an anatomical or mechanical obstructive lesion [2,3,4]. Megacystis is frequently involved with chronic intestinal pseudo-obstruction syndrome; however, isolated megacystis without severe intestinal obstruction is extremely rare. Here we present the case of a female patient with isolated congenital megacystis without intestinal obstruction.

Case report

The patient was a female infant with an antenatal diagnosis of intra-abdominal mass born at a gestational age of 38 weeks via cesarean section. Prenatal ultrasound examination showed no oligohydramnios. Physical examination showed a distended abdomen. Abdominal ultrasound imaging and computed tomography revealed an enlarged urinary bladder without hydronephrosis (Fig.1A). Barium enema did not reveal microcolon or malrotation. She had no

significant symptoms of intestinal obstruction and voiding was achieved by abdominal pressure. Her development was normal. However, at the age of 4, she had her first urinary tract infection. She was admitted to our hospital for re-evaluation of her condition. DMSA scintigraphy revealed no defects and cystography showed a massively enlarged bladder without vesicoureteral reflux. MR urography showed megacystis without ureterohydronephrosis (Fig.1B). Urethroscopy showed no obstructive lesions. Urodynamic study showed a hypocontractile detrusor and increased compliance. Reevaluation by barium enema did not reveal any significant findings (Fig.2A). A positive rectoanal reflex was obtained in an anorectal manometry. Histological examination of her rectum showed hypoganglinosis of the submucous and myenteric plexuses (Fig.2B). Thinning of the longitudinal muscle and connective tissue proliferation were not observed. Increased acetylcholinesterase activity was not observed. The MRI of sacral region of spinal cord showed no significant findings. Mutational analysis of the CHRNA3 gene, which encodes the nicotinic acethylcholine receptor (nAchR) α3 subunit, revealed no mutations. The protein expression of the nAchR α3

subunit was normal in her colon. She had mild, laxative-responsive constipation and voiding was achieved by intermittent catheterization.

Discussion

MMIHS is a rare cause of intestinal obstruction in the newborn, characterized by abdominal distension caused by a distended, non-obstructed urinary bladder, microcolon, incomplete intestinal rotation, decreased or absent intestinal peristalsis, and failure to pass meconium [1]. This is a severe subtype of the chronic intestinal pseudo-obstruction syndrome, a rare clinical entity in which recurrent symptoms of intestinal obstruction are present in the absence of an anatomical or mechanical obstructive lesion [2,3,4]. Chronic intestinal pseudo-obstruction syndrome represents a challenging disease in children. Survival of patients with chronic intestinal pseudo-obstruction syndrome has been possible due to recent advances in gastrointestinal management, particularly improvements in parenteral nutrition. However, prognosis for this syndrome remains poor with deaths occurring because of nutritional abnormality,

sepsis, cardiac arrythmia, complications of total peripheral nutrition, and renal failure [5,6].

Urological involvement is common in chronic intestinal pseudo-obstruction syndrome and the incidence varies from 33% to 92% [3,7,8,9]. Megacystis is a popular urological manifestation in this syndrome [7]. Prenatal diagnosis of megacystis is made in about 60% of cases with this syndrome [7]. The concomitant finding of megacystis and the absence of oligohydramnios may lead the physician to suspect the diagnosis. Because of the information available from sonography, appropriate investigations can be undertaken immediately after delivery.

The etiology of this syndrome remains unclear. The pathogenesis is varied with possible associations with an abnormal autonomic nervous system [10,11,12,13], degenerative muscle disease [14], imbalance of gut peptides [15], and dysganglinosis or inflammation [16]. Histological studies of the myenteric and submucosal plexuses of the bowel of patients with this syndrome have found normal ganglion cells in the majority of patients, decreased in some,

hyperganglionosis and giant ganglia in others [17]. It has been reported that transgenic mice lacking the nAchR α3 subunit have a phenotype similar to that of MMIHS [18]. In situ hybridization and immunohistochemistry studies to examine α3 mRNA or α3 protein showed a lack of α3 staining in most cases with MMIHS [19]. These findings indicate that the absence of the nAchR α3 subunit may provide a possible explanation for the underlying pathogenesis of MMIHS. However, mutational analysis of the CHRNA3 gene revealed no mutations in our case, similar to the results by Lev-Lehman et. al [20]. Further studies are warranted to clarify the genetic basis for MMIHS.

Our patient had congenital megacystis; however, she had no significant symptoms of intestinal obstruction and achieved prolonged survival. Barium enema did not reveal any significant findings; however, histological examination of her rectum showed hypoganglionosis of the submucous and myenteric plexuses. These findings indicate that this case may be a mild variant of chronic intestinal pseudo-obstruction syndrome. To our knowledge, this is the first report to present a case of isolated congenital megacystis without intestinal obstruction.

The presence of megacystis should alert the physician to the possibility of chronic intestinal pseudo-obstruction syndrome.

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Figure legends

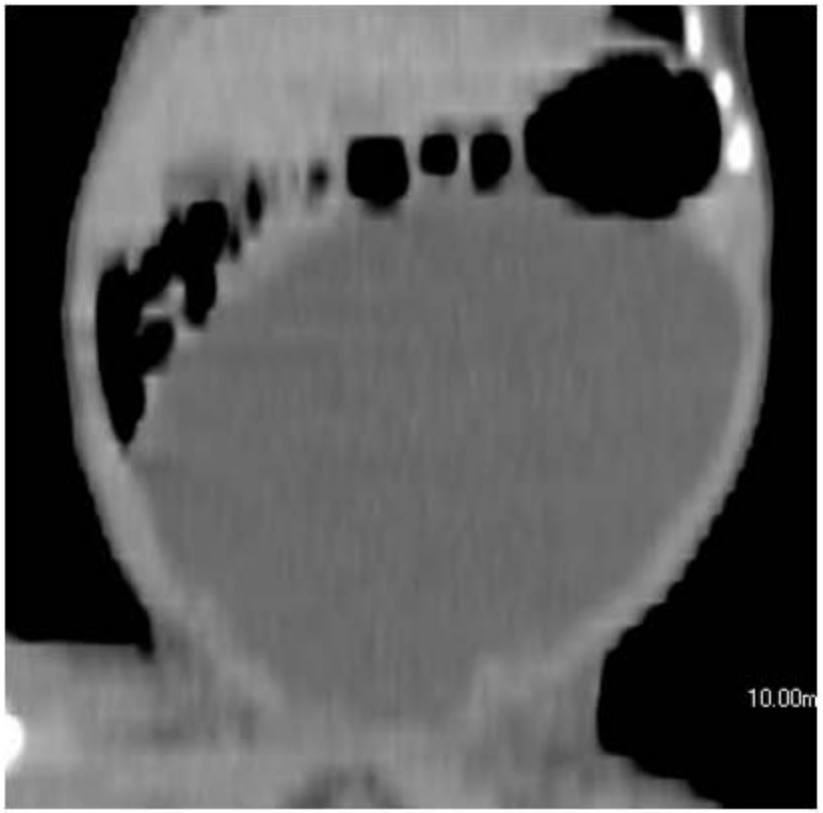
Fig.1. Congenital megacystis without hydronephrosis and vesicoureteral reflux. A: Abdominal three-dimensional CT imaging, axial view. B: MR urography imaging, axial view.

Fig.2. Radiological and histological evaluation for gastrointestinal tract in this case.

A: Barium enema imaging. There were no significant pathological lesions.

B: Histological findings of the patient's rectum. Rectal biopsy showed hypoganglinosis of the submucous and myenteric plexuses. Thinning of the longitudinal muscle and connective tissue proliferation were not observed.

A



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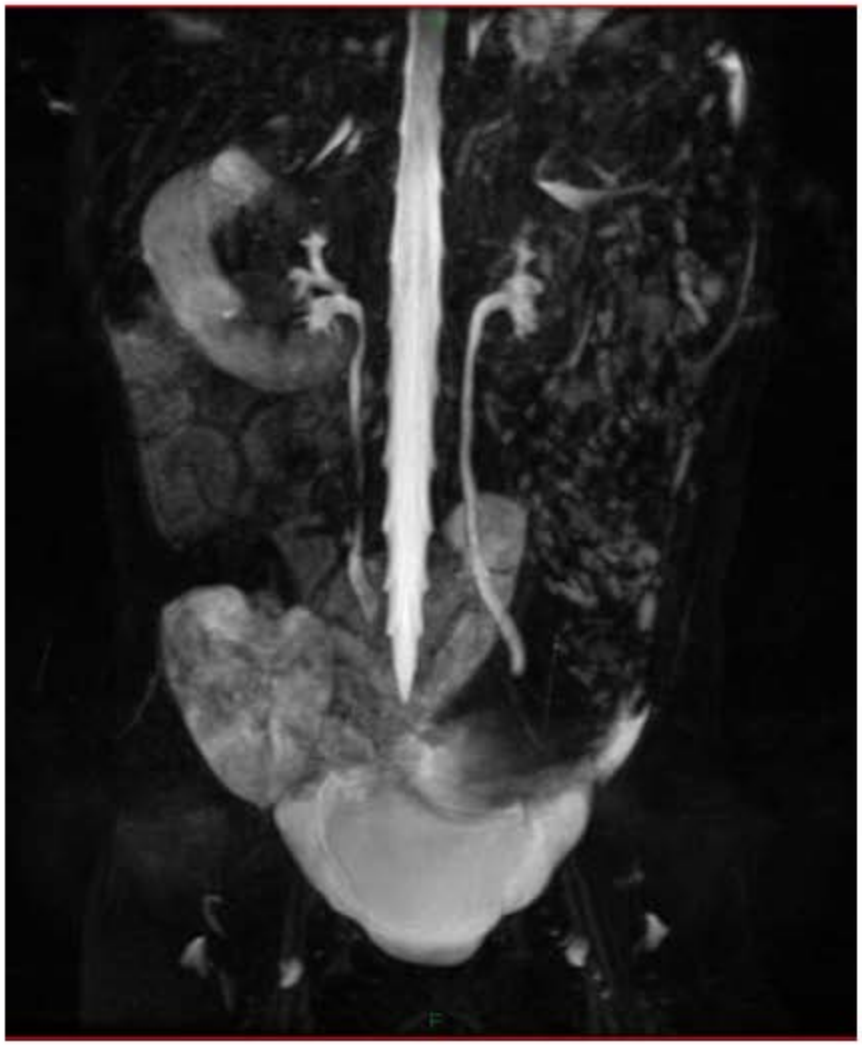

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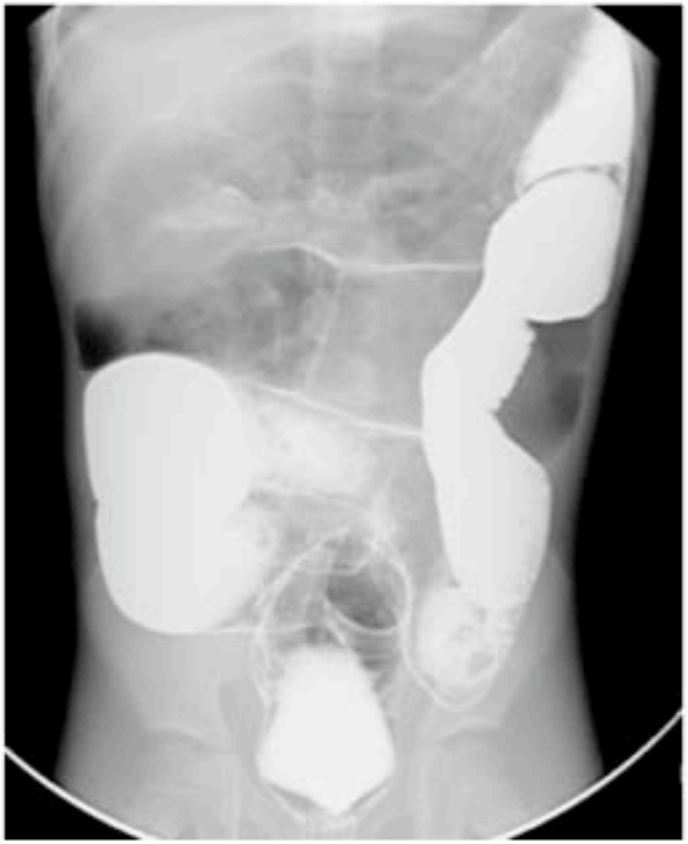

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