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CASE REPORT

Fetal Magnetic Resonance Imaging of Megacystis microcolon intestinal hypoperistalsis syndrome

Dewi Asih Wirasasmita[1*], Alexandra Sachlan[2]

Abstract

Megacystis microcolon intestinal hypoperistalsis syndrome (MMIHS) or Berdon syndrome is a rare congenital anomaly, involve urinary tract and gastrointestinal tract. The prognosis is dismal, the death is caused by infection. In general, the diagnosis is made based on ultrasonography (US) examination during the antenatal period due to the presence megacystis in the female fetus. Fetal Magnetic Resonance Imaging (MRI) has excellent tissue contrast can depict colon caliber and meconium distribution, hence definitive diagnosis can be established, useful for counseling and post-natal management.

Keywords: megacystis microcolon intestinal hypoperistalsis syndrome, definitive diagnosis, ultrasound, fetal MRI

Introduction

Megacystis microcolon hypoperistalsis syndrome (MMIHS) or Berdon syndrome was first described in 1976 by Berdon et al.[1] MMIHS consists of massive dilated bladder, malrotated of the microcolon and functional obstruction of the gastrointestinal tract. The disease is more common in female infants and the evidence for autosomal recessive in heritance has been reported from several journals.[1] The prevalence of MMIH in Indonesia has not been reported due to complexity of diagnosis and lack of data. However, there is only one reported case of MMIH in Indonesia where the diagnosis was only confirmed after birth.[2]

This rare disorder is associated with significant morbidity and mortality. Intestinal dysfunction leads to nutritional compromise and is ultimately fatal without continual parenteral nutrition. Infants with MMIHS have myopathic dysfunction of bladder as well and associated urologic comorbidities due to urinary tract infection and vesicoureteral reflux.[1,3–5] Prenatal diagnosis is required for counseling and to anticipate the postnatal management.

Prenatal ultrasound (US) suspected of MMIHS based on a distended urinary bladder with bilateral hydronephrosis with normal or increased of amniotic fluid in female fetus.[3] Normal meconium in the bowel can be easily identified by US at the age of 18-20 weeks gestation, but US is difficult to differentiate between small and large bowel, consequently hypoperistaltic cannot be determine by US. In contrary small and large bowel can differentiated clearly by MRI due to excellent tissue contrast resolution which assists in the evaluation of complicated fetal anomalies. Therefore, more information can be acquired for definitive diagnosis.

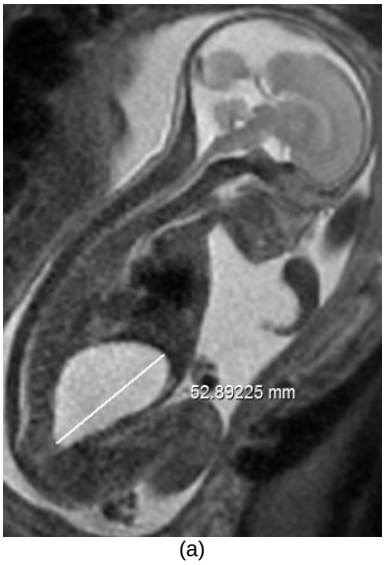
We reported a case of MMIHS that was diagnosed as megacystis by US. The aim of this case report is to demonstrate the role of fetal MRI in the prenatal diagnosis of MMIHS. Fetal MRI was performed with Siemens magnetom Avanto 1.5T system with Body Matrix coil. Axial, sagittal and coronal orientations were acquired with 4-5mm slice thickness, 0.5mm gap. Sequences used are T2HASTE (TR 10000ms, TE 120ms), and T1 Turboflash (TR 3, TE 1,1ms).

Affiliation

[1]Department of Radiology, Universitas Prima Indonesia, Medan, Indonesia
[2]Department of Surgery, Harapan Kita Women and Children Hospital, Jakarta, Indonesia

Correspondence

dewi_wirasasmita@yahoo.com



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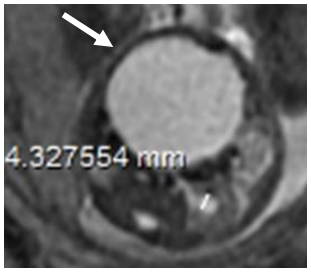

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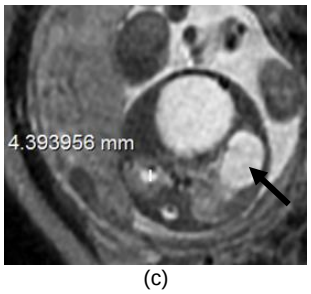

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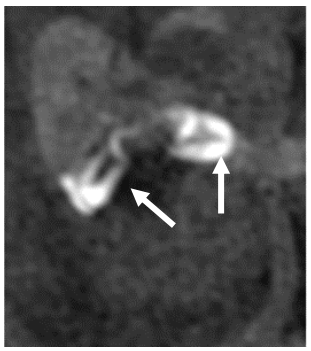

(d)

Figure 1. Female fetus at 24 weeks of gestation

a) Sagital T2 HASTE showed a distended urinary bladder
b) Axial T2HASTE showed normal pelviocalyceal of left kidney and distended urinary bladder (white arrow) and small caliber colon
c) Axial T2HASTE showed normal pelviocalyceal of right kidney and dilatation of stomach (arrow)
d) Coronal T1 showed distribution of meconium in the small bowel (white arrow)

Case Report

A 25-year-old primigravid patient was referred to radiology department due to abnormal prenatal US. Sonographic evalu-ation at 22 weeks gestation revealed a distended urinary bladder without pelvicali-ectasis and normal amniotic fluid in female fetus. Fetal MRI at 24 weeks of gestation confirmed sonographic findings (figure 1a,b) with additional information of gastric dilata-tion (figure 1c), small caliber of colon and the distribution of meconium is still in the small bowel (figure 1e). These findings suggest MMIH. Long term survival is unlikely. After counseling, the parents decided to continue the pregnancy.

The parent is a non-consanguineous couple and no record of genetic disease in the family. The baby was delivered by spontan labour at 36 weeks of pregnancy with a birth weight of 2500 g and Apgar scores of 10 at 1 min and 5 min. A few hours after birth, the baby presented food intolerance and massively abdominal distention. The baby has not passed stool and urinated for the first 24 hours. At the abdominal radiography revealed huge dilatation of the stomach without air distri-bution in the small and large bowel (figure 2). Since of these symptoms, intestinal obstruction was suspected and the baby underwent exploratory laparotomy on 2nd day of life. During the operation, the small bowel showed dilatation and contained meconium, while the caliber of the colon was small with ab-ent meconium. The location of appendix at the left side (figure 3). An appendectomy and ileostomy were performed. The distended urinary bladder was also presented without hydronephrosis. Biopsies of ileum and colon were performed and histological findings were nonspecific.

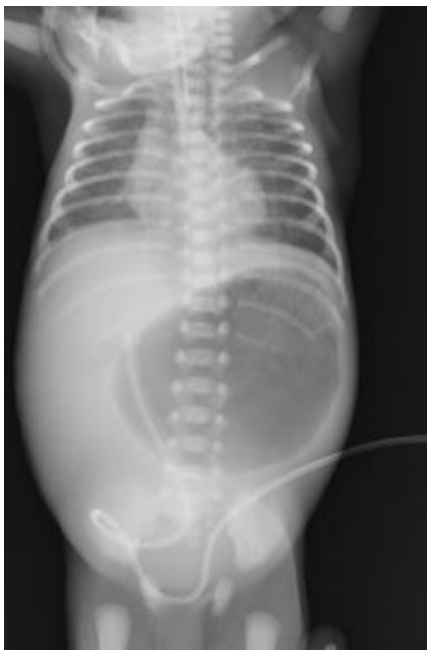

Figure 2. Plain radiograph showed distended stomach without air distribution at the small bowel.

Discussion

MMIHS or Berdon syndrome is a rare congenital syndrome that was first described in 1976 by Berdon et al.[1] It represents the most severe form of functional intestinal and bladder obstruction in newborn due to smooth muscle disorder. This syndrome is characterized by a massively dilated urinary bladder, incomplete intestinal rotation, microcolon and small bowel dilatation with hypoperistalsis throughout the intestinal tract.[1]

Prenatal diagnosis of MMIHS is a still challenging and the data on fetal MRI features of MMIHS is limited. The main clue for prenatal diagnosis MMIHS by US is megacystis.[1–5] Megacystis anomaly in fetus is nonspecific, with several differential diagnosis including prune belly syndromes, mechanical lower urinary tract obstruction (LUTO) and cloaca malformation. Therefore, it is neces-

sary to perform fetal MRI examination to confirm the definitive diagnosis and to rule out differential diagnosis.

Fetal MRI has ability to differentiate between the large and small bowel. Stomach and small bowel have bright signal intensity on T2W due to amniotic fluid, large bowel has hypointense signal on T2W and hyperintense on T1W due to high protein content in meconium. Normally, at 13 weeks of gestation meconium migrates distally from the small bowel and at 17 weeks gestation it accumulates in the distal colon.[6] In this case, fetal MRI showed distribution of meconium is still in the small bowel, no hyperintense signal at distal colon on T1W indicating intestinal hypoperistaltic. Fetal MRI can also measure caliber of small and large bowel. In this case, the caliber of large bowel is small with gastric dilatation. Therefore, diagnosis of MMIHs can be established not only based on the enlarged urinary bladder but also based on meconium distribution and caliber of the bowel. Fetal MRI can assist the diagnosis of this rare entity. Exploratory laparotomy confirmed the diagnosis of MMIHS.

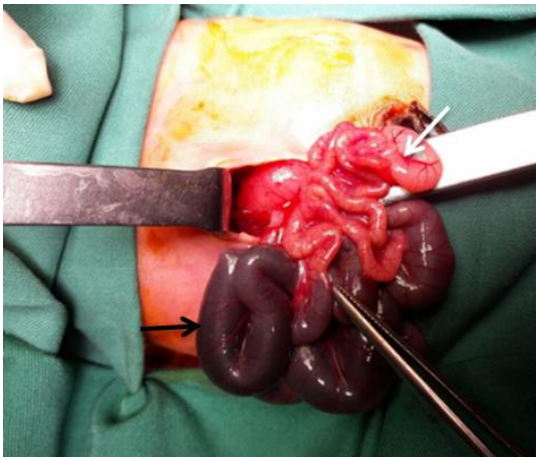

Figure 3. Intraoperative image showed intestinal malrotation, enlargement caliber of small bowel (black arrow) and contain meconium. Microcolon (white arrow) with absence mechanical obstruction.

In MMIHS the amount of amniotic fluid volume varies, with the tendency normal volume or oligo hydramnios up to 25-30 weeks gestation, after 34 weeks shows polyhydramnios due to the effect functional bowel obstruction becomes more evident.[3] For that reason the amount of amniotic fluid in case of MMIH cannot be use as a reference to assist in confirming the diagnosis. However, conversely in case of mechanical LUTO the amniotic fluid is typically reduced due to stenosis or atresia in the urethra. Prune belly syndrome is characterized by week abdominal wall, undescended testicle or cryptorchidism and hydrouteronephrosis is always present. Determination of sex is very important since MMIHs is almost exclusively in female.[1,3–5]

Prognosis in MMIHS is poor. Death occurs due to sepsis with multiorgan failure, malnutrition and complications of parenteral nutrition. As in this case, the baby has recurrent abdominal distention, so nutrition can only be administered parenterally. According to Prathapan et al, multiorgan transplantation can increase survival of patients with MMIHS.[7]

Conclusions

MMIHS is characterized by megacystis with absence of mechanical obstruction, microcolon, colon malrotation and intestinal hypoperistalsis in female fetus. Although fetal US is still modality of choice in prenatal diagnosis of complex congenital malformation, this has significantly changed after the advent of fetal MRI. Fetal MRI has superior in depiction of bowel loop caliber, meconium distribution, distended bladder and collecting systems in MMIHS. Fetal MRI can narrow the diagnosis and even establish a definitive diagnosis in case of MMIHS which is not possible with US.

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