Megacystis Microcolon Intestinal Hypoperistalsis Syndrome: Report of a Rare Case in Newborn

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Abstract- Megacystis Microcolon Intestinal Hypoperistalsis Syndrome (MMIHS) is a rare and the most severe form of functional intestinal obstruction in the newborn. The characteristic features of this congenital and fatal disease are abdominal distension, absent or decreased bowel peristalsis. Abdominal distension is a consequence of the distended, unobstructed urinary bladder with or without hydronephrosis. We present a case of female newborn with antenatal ultrasound revealing a large cystic mass in pelvic with urinary tract origin, abdominal distension, aperistalsis of the intestine and micro colon.

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Keywords: Megacystis; Microcolon; Hypoperistalsis; Newborn

Introduction

Megacystis microcolon intestinal hypoperistalsis syndrome (MMIHS) is a rare and fatal congenital disease firstly reported by Berdon and his colleagues in 1976 (1). It was characterized by abdominal distension caused by a large no obstructive urinary bladder, microcolon, aperistalsis or hypoperistalsis of the gastrointestinal system (1-3).

The prevalence of this rare disease is unknown. The disease has been reported in a total number of 227 MMIHS cases from 1976 to 2011(4).

Pathogenesis of this syndrome is not exactly recognized (5-7). Pathologic study shows vacuolar degenerative changes in the smooth muscle of bladder and bowel. MMIHS is a fatal disease, and most patients die within one year after birth due to severe sepsis, renal failure, liver failure, malnutrition and complication of TPN. Treatment is supportive. In this study, we report a case of MMIHS with typical manifestations in a newborn (3,4).

Case Report

During prenatal care of a 21-year-old pregnant woman, there was a sonographic report at 16 week of pregnancy that showed a live female fetus with a large cystic mass in pelvis with probability of urinary bladder origin without hydronephrosis. The fetus had longitudinal lie with cephalic presentation. The volume of amniotic fluid, heart rate and movement of the fetus were normal. Follow up sonography at 25 week of gestational age showed that the mass had occupied the majority space of abdominal cavity and had dilated the pyelocaliceal system and proximal part of ureters. At 28 week of gestational age 4-dimentional ultrasound and anatomical survey revealed the thin walled uni-loculated large cystic lesion with 100×71 mm in the abdominal and pelvic cavity had caused hydronephrosis in both kidneys, The amniotic fluid volume was normal (Figure 1). Screening tests for pregnancy including Downs's syndrome and Neural Tube Defect were normal. At 36th week of gestation, the female neonate was born. The birth weight was 3 kg., her Apgar scores at 1 and 5 min after delivery were 9/10. The neonate admitted to NICU. Immediately after birth, the neonate had severe abdominal distention without bowel sounds. Abdominal examination showed an irregular mass with defined margin in the middle part of the abdomen (Figure 2). Other examinations such as the head, neck, chest, limbs and spinal column were normal. The neonate had not urination on the first day of life. Postnatal sonography revealed severe dilatation of urinary bladder and bilateral hydronephrosis. Other organs of the abdomen

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were normal. After inserting an indwelling catheter in bladder about 600 ml urine, discharged and abdominal distention reduced to a great extent. In plain abdominal x-ray in the first day of life, a large gastric bubble with scanty air was seen in the intestine (Figure 3).

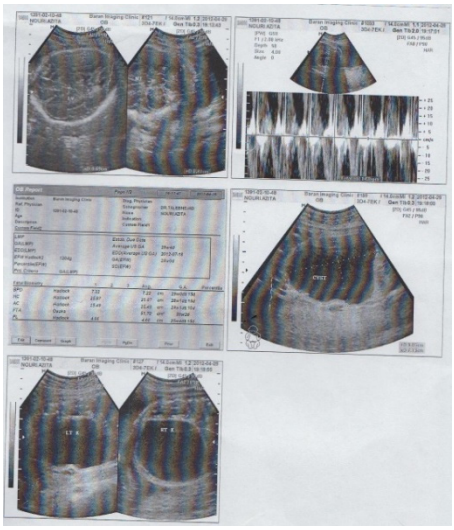

Figure 1. Thin walled uni-loculated large lesion with 100×71 mm in the abdominal and pelvic cavity in prenatal ultrasonography

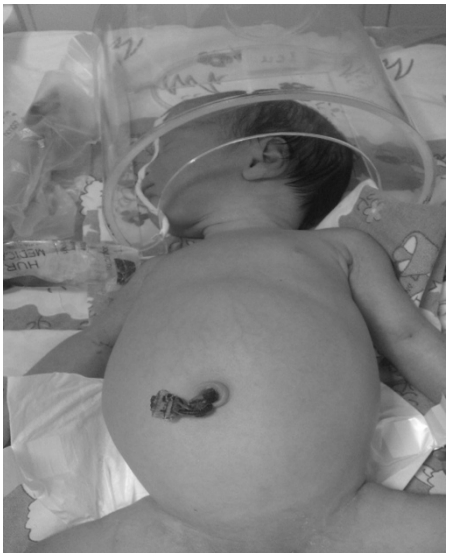

Figure 2. Severe abdominal distention in newborn

On the first day, Antibiotic therapy and TPN began. On the second day, the pelvic-abdominal x-ray showed no changes. On the third day of life, the abdominal distention occurred again. Barium enema showed micro colon (Figure 4) and echocardiography revealed ASD.On the 5th day of life, the neonate suffered from apnea which after proper primary management the stable condition occurred.

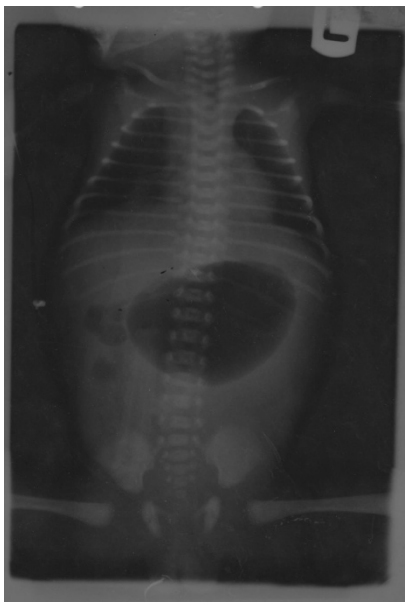

Figure 3. Plain x-ray shows large gastric bubble with scanty air in the proximal bowel

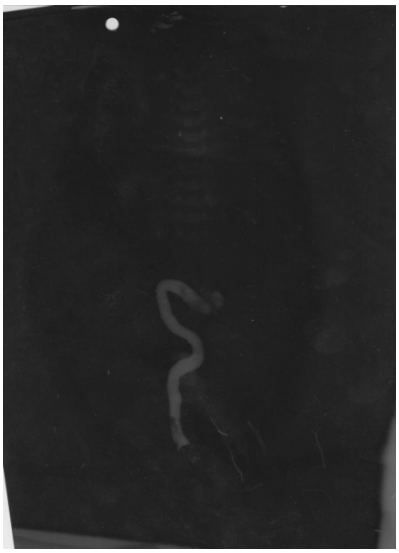

Figure 4. Barium enema shows microcolon

On the 6th day of life, the neonate had recurrent apnea episodes, and tracheal intubation was done. On the 7th day of life oliguria was seen and the level of serum BUN, creatinine and potassium increased. Voiding cysto-uretherography showed a massive urinary bladder with the irregular wall without intrinsic lesion, and vesicoureteral reflux was not noted (Figure 5). After proper management, the neonate had urinary flow again. During the next day's neonate suffered from severe sepsis and lab tests revealed leukopenia and thrombocytopenia. PTT were increased and frequently it

was treated with platelet, FFP, packed cell and GCSF. On the 13th day of life, the infant had reduced O2 saturation. She had bradycardia and massive tracheal bleeding and after all, the neonate had cardiac arrest and expired.

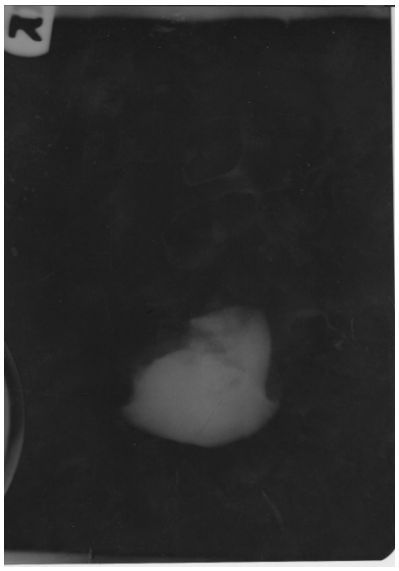

Figure 5. Voiding cysto-uretherography shows massive distention of bladder with irregular wall without vesicoureteral reflux

Discussion

Megacystis microcolon intestinal hypoperistalsis syndrome (MMIHS) is a rare and lethal disease. This syndrome firstly reported by Berdon and his colleagues in 1976 (1). After that time, limited cases have been reported. This syndrome characterizes with abdominal distension due to the large nonobstructive urinary bladder, microcolon, aperistalsis or hypoperistalsis of the gastrointestinal system. It represents the most severe form of functional intestinal obstruction in the newborn and is generally associated with a fatal outcome (1,3,4). Pathogenesis of syndrome is not recognized. The presumable role of genetic, neurogenic, myogenic and hormonal mechanism have been suggested (3). The disease is common in a female infant. This ratio is 3-4/1(8). Gosemann *et al*., reported a clear preponderance for female to male (70.6 vs. 29.4%) (4). The present case was female. Prenatal ultrasonoghraphy is a very good diagnostic tool for diagnosis of this syndrome (3, 9-10). The performance of sonography in the 16th week of gestation can identify large bladder in 88% cases and fetal hydronephrosis in 57% cases. At this time, polyhydramniosis and oligohydramniosis are seen in 33% and 7%, respectively (3). In our case first sign of large bladder was visible at the 16th week of gestation and all sonographies that performed during follow up showed the normal volume of amniotic fluid. Most of these patients are born term with normal weight. Puri *et al*., reported that of the 182 cases that reported until 2005, 59% were born at term, 25.5% at 36 to 39 weeks of gestation, 12% at 32 to 35 weeks and 3% at 31 weeks and less. Also, they mentioned that the mean birth weight was normal (3 kg.) for gestational age (3). Our case was a near term with normal weight. The clinical symptoms of MMIHS are similar to other neonatal intestinal obstructions. Clinical symptoms include abdominal distention, bile-stained vomiting, and absent or decreased bowel sounds. Abdominal distention will resolve after inserting a catheter into the bladder (3). In our case, there was severe abdominal distension after birth and after inserting a urinary catheter, obvious reduction in bladder volume was seen. In MMIHS, radiologic studies are usually diagnostic. Radiologic findings include large gastric bubble with no abdominal gas in the distal segments in the plain x-ray of the abdomen, huge bladder with irregularities in bladder wall in voiding cysto-ureterogram and microcolon in barium enema. Less than 5% of cases have vesicoureteral reflux.Sometimes other abnormal findings such as bilaterally duplicated systems may be seen (3,11). Abdominal x-ray of our patient showed similar findings and VCUG revealed dilated bladder with the irregular wall without vesicoureteral reflux. Barium enema was done, and micro colon component of MMIHS syndrome was observed. Other anomalies may be accompanied by this syndrome including omphalocele, cardiac malformations, rhabdomyoma and intestinal malformation (12). Anomaly accompanied with our case was an atrial septal defect, and there was no other abnormal finding. Pathologic findings in this syndrome are variable. Histologic studies of the myenteric and submucous plexuses in most patients showed normal or even increased ganglion cells. Degeneration of smooth muscle was seen in some patients. This finding can be used for distinction between this syndrome and the other aganglionosis diseases like hirshprung;s disease (3). Mantan and *et al.,* believe that the presence of vacuolar degenerative changes in the smooth muscle cells of bowel and bladder suggest that MMIHS may be due to a visceral myopathy. Immunohistochemistry staining of a rectal and urinary bladder biopsy specimen have showed a deficiency of cytoskeletal proteins and increased collagen (12). Boman *et al.,* consider that the microscopic abnormalities in MMIHS are variable, inconstant and nonspecific. They involve the smooth muscle more than the intrinsic nervations of the gut and

the bladder (14). The biopsy study of our patient showed normal ganglion cells. This finding was according to MMIHS syndrome. In differential diagnosis, we excluded prune-blley syndrome. Ninety-five percent of Prune-belly syndrome occurs in the male. The cause of bladder dilatation during fetal life in this syndrome is obstruction of urinary tract system. Meanwhile this disease is accompanied by oligo hydramniosis, vesicoureteral reflux and lung hypopolasia (14-15).

Treatment of MMIHS is supportive and total parenteral nutrition (TPN) is necessary in all of the cases. Prokinetic drugs have no effect on intestinal or bladder peristalsism. Surgical manipulation has generally been unsuccessful (3,4) Raofi *et al*., believe that multi organ transplantation is the only way to rescue these patients (16). MMIHS is a fatal disease. The majority of patients died within one year after birth due to severe infection. Only a few cases have been reported to stay alive until 11 years of age (17). Although most cases of MMIHS syndrome occurs sporadic, the risk of recurrence in next pregnancies and sibling is about 25%. This issue has declared the probability of autosomal recessive transmission of disease (17-18).

Therefore, parents of such neonates should consider repeated sonographies during the next pregnancies especially if the fetus is female. In conclusion physician must considered Megacystis microcolon intestinal hypoperistalsis syndrome (MMIHS) in 1-each prenatal ultrasonoghraphy that show bladder dilatation with or without hydronephrosis especially if the fetus is female, also after birth each abdominal distention that resolve promptly after inserting a catheter into the bladder.

The goal of introducing this disease is to attract physicians to the probability of MMIHS so that they avoid from unnecessary surgical interventions such as diversion in this cases.

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