MEGACYSTIS MICROCOLON INTESTINAL HYPOPERISTALSIS SYNDROME- A LETHAL RARE ANOMALY



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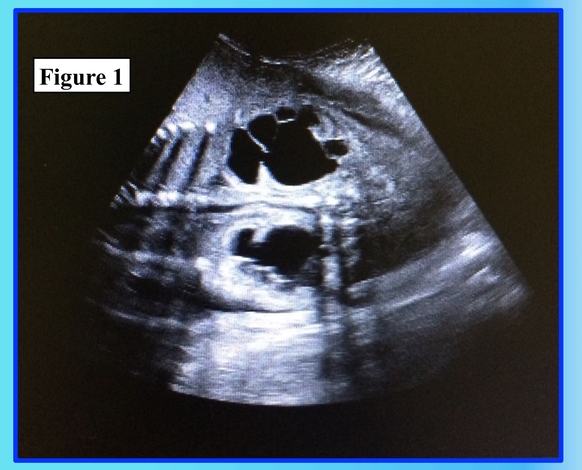
Figure 3: Massive distended abdomen secondary to distended urinary bladder
Figure 4: After drainage of urine abdominal distension reduced

Background:

Megacystis microcolon intestinal hypoperistalsis syndrome (MMIHS) is an unusual disorder characterized by a distended non-obstructed bladder, microcolon, and decreased intestinal motility. MMIHS is a rare condition and, to our knowledge, not more than 150 cases have been reported over the past 10 years[1].
Here we present a 34-year-old gravida3 para2 woman with fetal, bilateral hydronephrosis (Figure:1) and greatly distended bladder and mild polyhydramnios, detected during a prenatal ultrasound. Based on the USG findings, MMIHS was suspected. Presence of ganglionic cells on rectal biopsy confirmed the diagnosis of MMIHS postnatally.

Case:

We present a 34-year-old gravida3 para2 woman with fetal bilateral hydronephrosis, greatly distended bladder and mild polyhydramnios, detected during a prenatal ultrasound. Based on the USG findings, MMIHS was suspected. A female infant was born at 34 weeks of gestation by an emergency caesarean section because of breech during labour. The neonate weighed 4.2 kg with an Apgar score of 4 in 1 min and 6 in 5 min. Physical examination showed a distended abdomen (Figure:3) and bladder, as well as bilateral palpated enlarged kidneys. Abdominal x-ray showed a dilated stomach and minimal gas in the distal bowel segments (Figure:2). Abdominal ultrasound imaging showed dilated stomach with distal duodenal atresia. Fluoroscopy gastrografin revealed narrow bowel loops and jejunal malrotation at the right side. USG KUB showed distended urinary bladder, bilaterally enlarged kidneys, with tortuous and dilated ureter. A vesicostomy was performed to facilitate the drainage of urine (Figure:4). Ganglion cells were detected following rectal biopsy and female karyotype (46,XX) was confirmed by chromosomal analysis. The diagnosis of MMIHS was confirmed. The parents refused to give consent for any surgical correction. At the time of this report, the child was 2-months-old and had normal physical and neurological development, but total parenteral nutrition has been required.



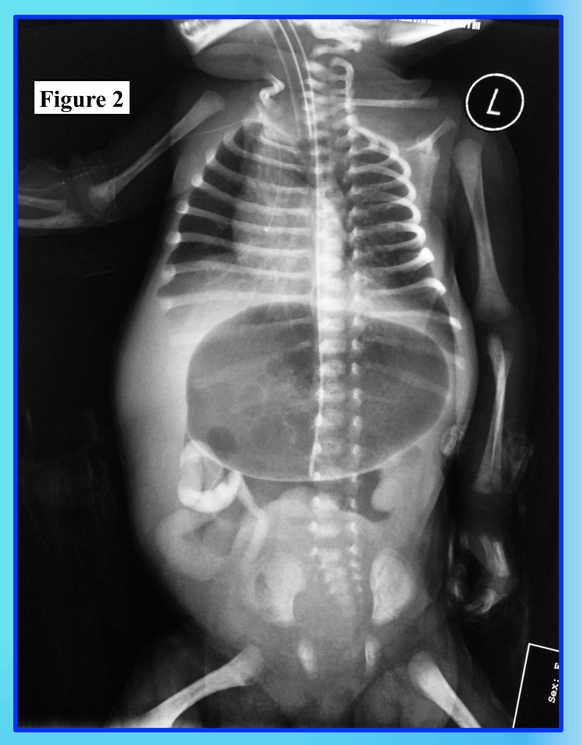


Figure 1: Antenatal scan shows fetal bilateral hydronephrosis
Figure 2: Abdominal X Ray shows dilated stomach

Discussion:

MMIHS is characterized by a massively enlarged fetal urinary bladder, microcolon, hypoperistalsis throughout the intestinal tract and incomplete intestinal rotation. Hydro-uretero-nephrosis and a hugely distended urinary bladder. The frequency of the disease is observed 3-4 times more in female than in male patients. In previous studies reported that 80% of infants died within the first year and 90% died within the first 2 years after birth. As a treatment of choice, no effective surgical method has been defined yet. Several intestinal diversions have failed to be successful[2-3].

In our case, antenatal USG revealed Huge enlarged bladder and bilateral hydronephrosis at 29 weeks gestation and resulted with a live birth at 34 weeks gestation. We suggest that an antenatal USG finding of a fetus with massive enlarged urinary bladder or intra abdominal mass with hydronephrosis should alert the physicians for MMIHS. Prenatal diagnosis of this rare disease is important for optimal counseling and postnatal appropriate management. Also genetic counseling is indicated for future pregnancies[4].

References:

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Acknowledgement: We would like to thank,
Dr. Nasser Shaikhan Taaeeb Al Shafouri
(Department of Paediatrics, Ibri Regional Hospital)
for providing us with the neonatal photographs (Figure 3 & 4).