MEGACYSTIS– MICROCOLON– INTESTINAL HYPOPERISTALSIS SYNDROME- CASE REPORT AND REVIEW OF LITERATURE.

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HOW TO CITE THIS ARTICLE:
Gunjan Mehrotra, Rajesh Sharma, Manoj Kela, Avinash Vishwani, Sangram Singh. "Megacystis–microcolon–intestinal hypoperistalsis syndrome- case report and review of literature". Journal of Evolution of Medical and Dental Sciences 2013; Vol2, Issue 23, June 10; Page: 4152-4155.

ABSTRACT: Megacystis–microcolon–intestinal hypoperistalsis syndrome (MMIHS) is a rare, often fatal condition. Infants present with a functional obstruction of the gastrointestinal tract (GI), malrotation, micro colon, and a large unobstructed bladder.

MMIHS usually presents in female infants. We present a male infant diagnosed as having megacystis–microcolon–intestinal hypo peristalsis.

KEYWORDS- Megacystis–micro colon–intestinal hypoperistalsis syndrome

INTRODUCTION-Megacystis-microcolon-intestinal hypoperistalsis syndrome consists of distended unobstructed bladder, dilated small bowel, micro colon, and decreased or absent peristalsis with presence ganglion cells. Omphaloceles [1], cardiac malformations and multiple rhabdomyomata , [2, and 3] mild webbing of the neck and intra-abdominal testis has also been associated with this syndrome [1].

CASE REPORT-A 30-year-old G_2P_1 woman presented to our hospital with a routine ultrasound examination at 22 weeks. The ultrasonographic examination [fig. 1] revealed a single male fetus with a markedly distended bladder and normal to increased amniotic fluid volume. There was minimal distension of the renal pelvis, no other abnormalities were detected. The most likely diagnosis of these ultrasonographic findings was megacystis-microcolon-hypoperistalsis syndrome.

The patient was referred for genetic counseling and the examination was repeated in two weeks to assess the progression of the syndrome. The second examination performed at 24 weeks again demonstrated increased amniotic fluid. The bladder was further distended and extended into the left upper quadrant of the fetus filling the pelvis and maximum part of the abdomen.

Even after advice to terminate the pregnancy patient decided to continue it and delivered prematurely at 32 weeks. Baby was 1.5kg male presented with perforation peritonitis after 24 hours. Baby-gram showed gas under right dome of diaphragm [fig.2].Exploratory laparotomy was planned & on exploration a markedly distended bladder filling the abdominal cavity with no urethral stricture or gross evidence of mechanical outlet obstruction. Mild hydronephrosis was noted bilaterally without hydroureter along with small ileal perforation near ileo-ceacal junction and complete micro colon [fig.3]. Ileostomy was done & abdomen was closed. The baby died 24 hours after surgery due to cardiopulmonary dysfunction.

DISCUSSION- Megacystis-microcolon hypoperistalsis syndrome is rare and lethal in most cases. It was first described in 1974 by Berdon in 5 female infants, 2 of whom were sisters. The first reported ultrasound diagnosis was made by Vezina et al. in 1979. In all cases, the enlarged bladder and absence of oligohydramnios typical of this syndrome were easy to define by ultrasound. Polyhydramnios is present in approximately 25% of the cases [1]. The length of gestation and birth weight are within normal limits. Megacystis-microcolon-intestinal hypoperistalsis syndrome is more common in females perhaps due to under diagnosis in males who are labeled instead as "prune belly". Most cases are sporadic. An autosomal recessive inheritance has been supported by reports of affected siblings.

A transient anatomical and/or functional urinary tract obstruction has been considered. Srikanth [4] suggested that an intramural inflammatory process affecting the gastrointestinal and urinary tracts could lead to extensive fibrosis. The fibrosis would destroy the intestinal neural network, producing hypo peristalsis and causes neuromuscular in coordination of the bladder. The resulting bladder distension could interfere with the rotation of the intestine causing malrotation. Another theory suggests the urinary tract obstruction in this condition is secondary to the intestinal defect. It is not clear whether this is a primary neuropathy or myopathy [1]. At pathology, vacuolar degeneration of smooth muscle is observed. The ganglion cell distribution is normal but axonal dystrophy of the central, peripheral, and autonomic nervous systems [5] and dysplastic changes associated with increased laminin and fibronectin have been observed suggesting a primary myocellular defect of contractile fiber synthesis. This suggests a defect of glycogen-energy utilization but a deficiency of fiber synthesis could also be responsible for the disorder. In mice a deficiency in alpha3 subunit of the neuronal nicotinic acetylcholine receptor (a mediator of normal function of the autonomic nervous system) results in similar symptoms to those of megacystis-microcolon-intestinal hypoperistalsis[6].

On USG megacystis-microcolon-intestinal hypoperistalsis syndrome should be considered in the presence of an abnormally distended bladder with a normal or increased amount of amniotic fluid. Dilatation of the urinary bladder in conjunction with the absence of oligohydramnios should raise the suspicion of the diagnosis, particularly in a patient at risk. It should be noted though, that the dilatation of the bladder may not be visible in some case at the 18-20 weeks exam, and that the earliest sign maybe mild hydronephrosis [7].

The main differential diagnosis is obstructive uropathy. In a female fetus, a low urinary tract obstruction can be due to urethral agenesis, variants of caudal regression syndrome, or to the rare detrusor hypertrophy [8]. All of these cases are associated with severe oligohydramnios. Megacystis-microcolon-intestinal hypoperistalsis syndrome must be distinguished from isolated

"prune belly" caused by urethral obstruction. Prune belly occurs predominantly in males, is commonly due to posterior urethral valves and has a low recurrence risk.

Megacystis-microcolon-intestinal hypoperistalsis syndrome is lethal in most cases, with only about 6% survivors at one year [9]. An intrauterine death has been described. Intestinal dysfunction is the cause of death despite hyperalimentation. Septicemia has been reported as the cause of death in several infants [10]. There are no reported cases of long term survivors.

The prognosis is poor and treatment has been shown to be ineffective. Trans uterine- trans fetal bladder taps have been used to relieve bladder pressure in utero [11]. Recommendations for surgical management include palliative surgery. Hyper alimentation is required. Parasympathomimetics, synthetic gastrointestinal stimulants, adrenergic blockers, and multiple gastrointestinal hormones have not been effective in inducting adequate bowel function. Obstetrical management should include a careful search for associated anomalies. Considering the poor prognosis, termination of pregnancy may be offered to the patient.

REFERENCES:

1. Birth Defects Encyclopedia Ed. Buyse ML, Blackwell Scientific Publications, Cambridge, MA 02142 P. 978-9.
2. Winter RM, Knowles SAS: Megacystis-microcolon-intestinal hypoperistalsis syndrome: confirmation of autosomal recessive inheritance. J Med Genet 1986 23: 360-362.
3. Couper RT, Byard RW, Cutz E, Stringer DA, Durie PR: Cardiac rhabdomyomata and megacystis-microcolon-intestinal hypoperistalsis syndrome. J Med Genet 1991 28:274-276.
4. Srikanth MS, Ford EG, Isaacs H Jr, Mahour GH Megacystis microcolon intestinal hypoperistalsis syndrome: late sequelae and possible pathogenesis. J Pediatr Surg 1993 Jul;28(7):957-9
5. Al-Rayess M, Ambler MW Axonal dystrophy presenting as the megacystis-microcolon-intestinal hypoperistalsis syndrome. Pediatr Pathol 1992 Sep-Oct;12(5):743-50
6. Xu W, Gelber S, Orr-Urtreger A, Armstrong D, Lewis RA, Ou CN, Patrick J, Role L, De Biasi M, Beaudet AL Megacystis, mydriasis, and ion channel defect in mice lacking the alpha3 neuronal nicotinic acetylcholine receptor. Proc Natl Acad Sci U S A 1999 May 11;96(10):5746-51
7. Couper RT, Byard RW, Cutz E, Stringer DA, Durie PR: Cardiac rhabdomyomata and megacystis-microcolon-intestinal hypoperistalsis syndrome. J Med Genet 1991 28:274-276.
8. Prenatal Diagnosis of Congenital Anomalies, Eds. Romero R, Pilu G, Jeanty P, Ghidini A, Hobbins J Appleton & Lange1988 East Norwalk, CT 06855 p 291.
9. Chung MY, Huang CB, Chuang JH, ko SF, Chen L, Megacystis-microcolon-intestinal hypoperistalsis syndrome (MMIHS): a case report. Chang Keng I Hsueh 1998 Mar; 21(1):92- Chen CP, Wang TY, Chuang CY, Sonographic findings in a fetus with megacystis-microcolon-intestinal hypoperistalsis syndrome. J Clin Ultrasound 1998 May;26(4):217-20.
10. Stamm E, King G, Thickman D, Megacystis-microcolon-intestinal hypoperistalsis syndrome: prenatal identification in siblings and review of the literature. JUltrasound Med 1991 Oct; 10(10):599-602.

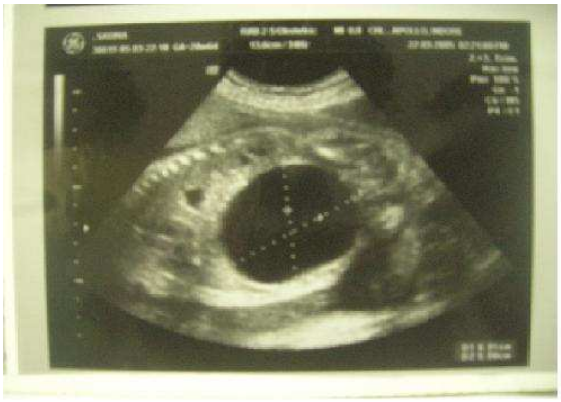

Fig 1] The ultrasonographic examination revealed a single male fetus with a markedly distended bladder and normal to increased amniotic fluid volume.

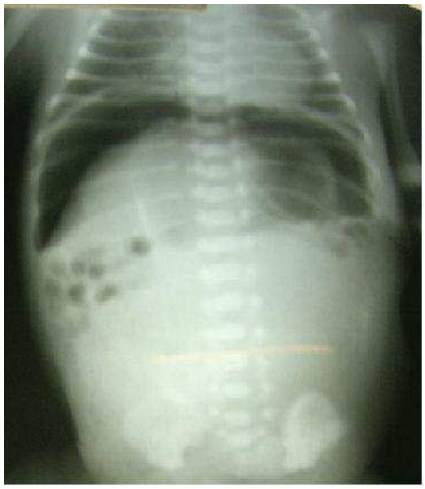

Fig 2] Baby-gram showed gas under right dome of diaphragm.

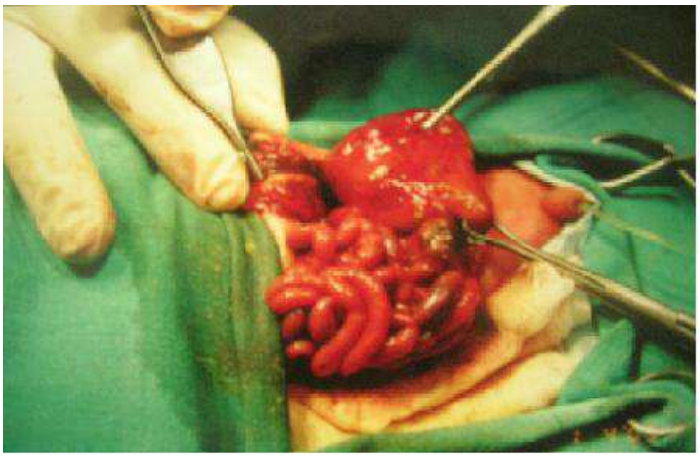

Fig 3] Mild hydronephrosis was noted bilaterally without hydroureter along with small ileal perforation near ileoceacal junction and complete micro colon.