Int J Pharm Bio Sci 2014 Jan; 5(1): (B) 744 - 751

Case Report **Radiodiagnosis**

International Journal of Pharma and Bio Sciences

ISSN 0975-6299

A CASE OF MULTISYSTEM ANOMALIES ASSOCIATED WITH GASTROSCHISIS IN THE UNBORN FETUS – CASE REPORT WITH POST-DELIVERY CORRELATION

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ABSTRACT

Gastroschisis is a congenital anomaly which shows extra abdominal herniation of fetal bowel loops, occasionally with portions of liver/ stomach, into the amniotic cavity through a paraumbilical defect in the anterior abdominal wall. The defect is usually right sided. Gastroschisis is decreasing in incidence owing to early detection by the advanced techniques in imaging and more efficient and thorough antenatal assessment. Omphalocoele is mostly associated with other chromosomal anomalies. On the contrary, gastroschisis is not alwaysaccompanied by a fixed set of anomalies except in 7 – 30% cases wherein anencephaly, cleft lip and palate, ectopia cordis, ASD, diaphragmatic hernia scoliosis, syndactyly, amniotic band and intestinal atresias. However this 25 year old primi gravida at 28 weeks, showed elevated maternal sAFP, short femur, cardiomegaly with severe pulmonary hypoplasia, median cleft lip, bilateral echogenic kidneys with renal cysts, diaphragmatic hernia, umbilical cord cyst and polydactyly with bilateral equinus deformity may occur. The unusual permutation of anomalies seen in this case is what makes it a rare entity worth discussion.Ultrasound was indicated by a clinical IUGR and done for anomaly screening. One week later, she returned following intrauterine fetal demise after which imaging modalities were employed to confirm the antenatal findings. This study emphasizes the need for strict antenatal screening and suspicious eye to rule out even unrelated anomalies in each case.

KEY WORDS :Gastroschisis, Omphalocoele, paraumbilical, cardiomegaly, antenatal

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INTRODUCTION

Antenatal ultrasound done for routine screening or for obstetric indications such as evaluating an elevated maternal serum alpha fetoprotein (AFP) is often sensitive to diagnose congenital anterior abdominal wall defects, namely, gastroschisis and omphalocoele. Routine antenatal screening is pivotal to detect and to distinguish between the different types of anterior abdominal wall defects. Gastroschisis is a congenital malformation defined by the freely floating bowel loops herniating through a full-thickness paraumbilical abdominal wall defect. Absence of a sac around the bowel loops is an important distinguishing feature. It is a rare anomaly with reported incidence rates of 1 to 6 per 10,000 live births [1,2] occurring in young mothers. The incidence is 16- fold higher in women younger than 20 years than in women of 30 years or more. Other risk factors associated with gastroschisis include tobacco and illicit drug use, low socioeconomic status, and vasoconstrictive drugs [3–6]. This anomaly is seen as isolated, rarely associated with other chromosomal anomalies (0.8%–3%). [1, 7, 8]Associated extra intestinal structural anomalies may be present, occurring in 5% to 28.6% of cases, and include cardiovascular and central nervous system anomalies most frequently. [9-14] The purpose of this study is to emphasize on the significance of impeccable routine anomaly screening and to depict a pictorial representation of the antenatal findings and to correlate them with the post-IUD imaging findings. Gastroschisis is believed to occur 5 to 8 weeks after conception, and the current hypothesis regarding the pathogenesis is an early disruption of the right omphalomesenteric artery [3, 4]. Most cases are diagnosed antenatally, and affected newborns require surgical repair of the defect soon after delivery.

CASE REPORT

25 year old primi gravida married for a year presented at around 27 weeks and was clinically detected with intrauterine growth retardation. This doubtful finding led to the need for an ultrasound to rule out any fetal anomalies incompatible with life and toe confirm the dates. The moment the curvilinear probe (3.5 – 5 MHz) was placed transabdominally, one anomaly after the other unfolded in a serial manner. The real time B mode USG was done and a single intrauterine fetus in cephalic presentation was detected with adequate liqor (AFI – 13.7 cm). A three-vessel umbilical cord was noted which showed 2 cysts arising from the cord. Good fetal cardiac activity was seen at first. The biometry revealed accurate biparietal diameter, head circumference, abdominal circumference as per the gestational age expected. The anatomy was then studied in detail. Head – Midline falx seen with normal bilateral lateral ventricles and no identifiable intracranial lesion was seen. Cerebellum visualized as normal. Face – median cleft lip was seen. Spine- entire spine visualized in long and transverse axes and appeared normal. In the Thorax, the left lung was visualized but right lung was not visualized and right sided cardiac deviation was noted. The cardiac ratio revealed a global cardiomegaly. Heart – deviated to the right with the left superior vena cava draining abnormally into the coronary sinus, with concordant ventro-sinus connection. The great vessels seen arising from the respective ventricles in normal configuration. In addition, multiple cystic spaces seen entering the thoracic cavity suggestive of a diaphragmatic hernia. Abdomen- situs appears normal. The cavity shows multiple hyperechoic specks of calcification and small cystic spaces in the periphery along with multiple specks of calcification on the peritoneal surface of the liver. A hyperechoic lesion seen protruding through the anterior abdominal wall, arising by the side of the umbilical cord through a defect with small bowel loops as its content. No sac identified. Both kidneys appear enlarged and echogenic with few renal cysts seen on both sides. Bladder appears normal with estimated fetal weight of 107.3 grams. Table 1 depicts the antenatal findings pertaining to this case report

Table 1
Antenatal parameters at 27-28 weeks of gestation

PARAMETER	(IN cm)	WEEKS	DAYS
BPD	7.1	28	5
HC	25.94	28	0
AC	21.2	27	6
FL	4.75	25	4

DISCUSSION

The biometry revealed short femur with femoral length falling below 5th percentile and bilateral equinus deformity. A week after the prenatal diagnosis, patient returned and intrauterine death was diagnosed. Following the evacuation, post IUD ultrasound and radiographs were taken to correlate the prenatal findings and added findings like hypertelorism and polydactyly were confirmed. Reports from several countries have documented an increase in the incidence of cases over the past three decades. There are two possible explanations for this increase. First, birth defects such as gastroschisis may be ascertained more accurately by central registries. Second, there may be a new environmental factor. The highest rate of the defect is found in infants of mothers under 20 years of age. The first trimester use of some medications have been attributed as the causative agents of gastroschisis such as aspirin, ibuprofen, pseudoephedrine, and phenylpropanolamine. Moreover, early maternal exposure to x-rays and/or organic solvents raised risk levels. Other causes include alcohol, cocaine, marijuana, and smoking were associated with increased risk of gastroschisis. Many congenital conditions need to be ruled out antenatally, but some are diagnosed only clinically for eg: Adult-onset Still disease in pregnancy can be confused with many other diseases, but its diagnosis, after exclusion of other infectious, malignant, and rheumatic conditions, can portend good maternal and fetal outcomes. [15]

IMAGES

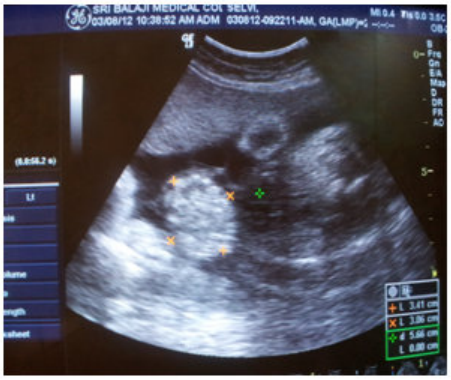

Figure 1
an antenatal ultrasonogram of the live fetus which showsa hyperechoic mass protruding through a defect the anterior abdominal wall lying to the right of the umbilical cord resembling a bunch of bowel loops adherent to each other but with no sac around the content.

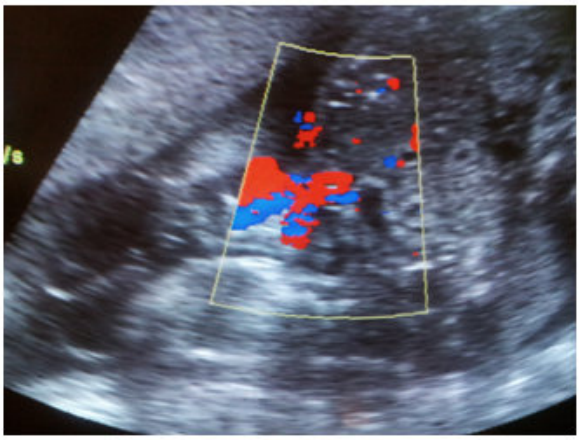

Figure 2
an antenatal ultrasonogram of the live fetus which shows a three- vessel umblical cord lying adjacent to the defect in the anterior abdominal wall

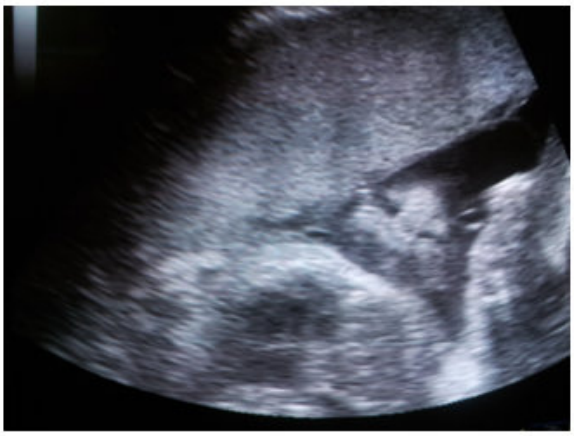

Figure 3
an antenatal ultrasonogram of the live fetus which shows the median cleft lip in antenatal ultrasound scan

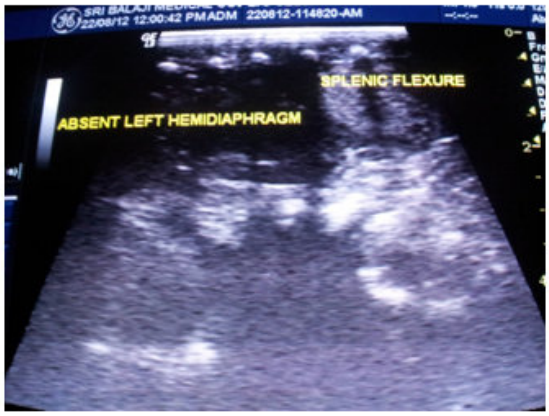

Figure 4
an image of post-delivery ultrasound image of the fetus following IUD which shows agenesis of the right lung and apparent cardiomegaly

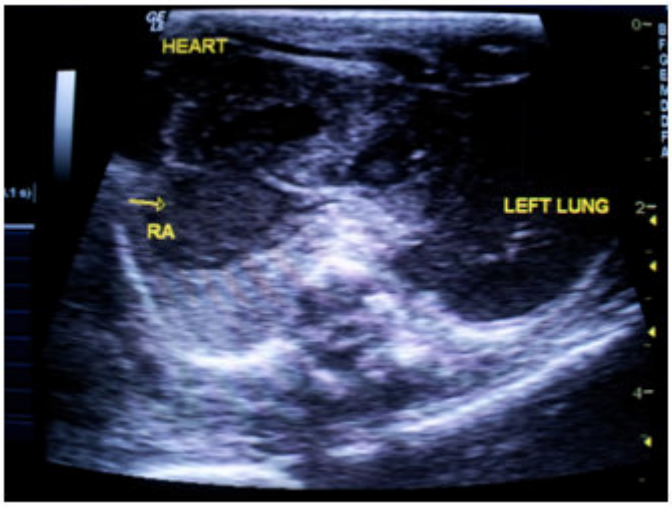

Figure 5
an image of post-delivery ultrasound image of the fetus following IUD which shows the absence of the left hemidiaphragm

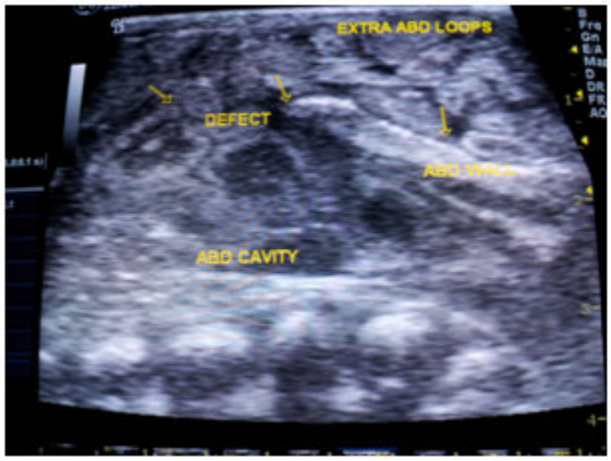

Figure 6
~~is~~ an image of post-delivery ultrasound image of the fetus following IUD which shows the paraumbilical anterior abdominal wall defect with the herniated and gangrenous bowel loops and no sac is seen around the loop of bowels.

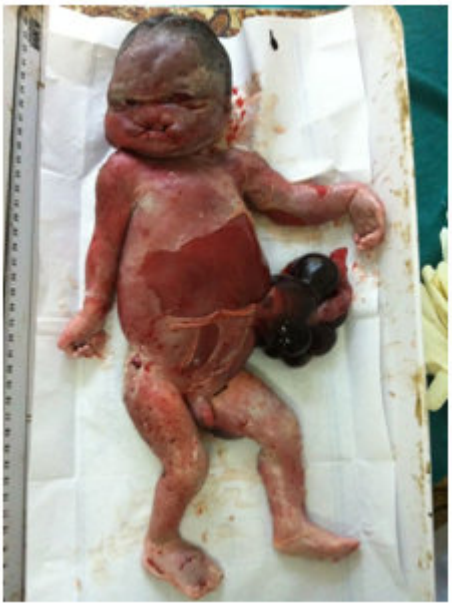

Figure 7

~~shows~~ the post-delivery image of the fetus with all the antenatally detected anomalies with loops of exposed gangrenous bowel loops protruding through the paraumbilical defect. Other visible findings include hypertelorism, median cleft lip, polydactyly, bilateral equinus deformity of the legs and narrowed chest wall.

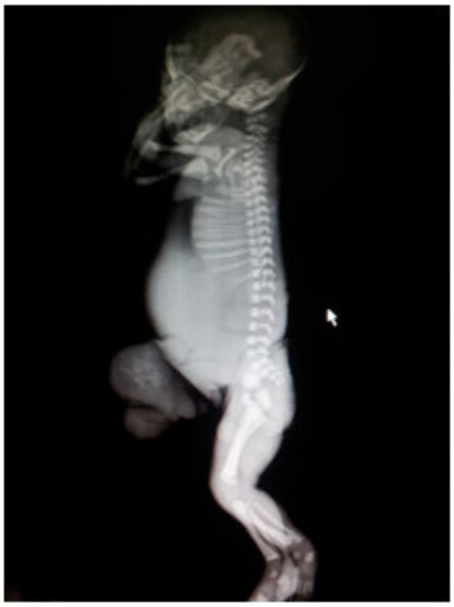

Figure 8

~~shows~~ the radiograph of the dead fetus in lateral view shows the protuberant abdomen with the extra abdominal mass of adherent bowel loops with no other bony deformity in the spine or limbs visualized

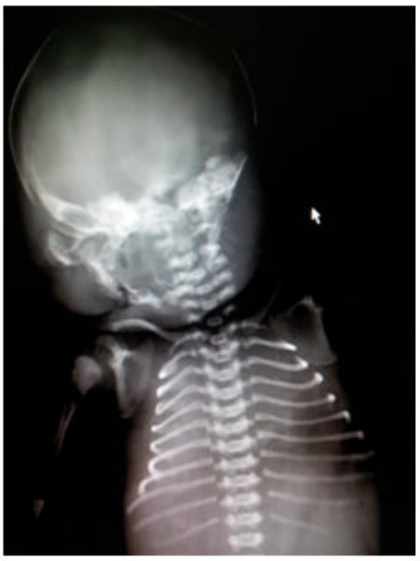

Figure 9

~~shows~~ the radiograph of the head and chest of the dead fetus post-delivery showing crowding of the ribs more on the right owing to the hypoplastic right lung with presence of few scattered lucencies in the left lung field probably due to the diaphragmatic hernia on the same side.

Table 1

Differences between omphalocoele and gastroschisis

Feature	Omphalocoele	Gastroschisis
Incidence	1:6000 to 4000	1:20000 to 30000
Etiology	Omphalomesenteric artery occlusion	Failure of gut migration from yolk sac to abdominal cavity
Surgical emergency	Yes	No
Covering Sac	Present (may be ruptured	Absent
Abdominal defect	Small to large	Small (2-4 cm)
Cord attachment	Cord onto sac	Cord onto abdominal wall
Herniated bowel	Protected	Edematous and matted
IUGR	Less likely	More common
Associated anomalies	55-80%	10-15%
Other chromosomal anomalies	Beckwidth Weidmann, pentalogy of Cantrell, bladder extrophy, CHD	Rare; intestinal atresia, Gall bladder agenesis, renal agenesis
Location of defect	Umbilicus	Right of umbilicus
Maternal age	Average	Younger
Mode of delivery	Cesarean/vaginal	Vaginal
Prognostic factors	Associated anomalies	Condition of bowel

CONCLUSION

Gastroschisis and omphalocoele are the two most common congenital abdominal wall defects. Described in the literature as early as the first century AD, today these anomalies are mostly detected antenatally due to routine maternal serum screening and fetal ultrasound. Differences between gastroschisis and omphalocoele are summarized in Table 1. However, the most important distinguishing feature in prognosis of the two conditions comes from not only the defect itself but from the differential rate of associated anomalies; the

risk of an associated structural or chromosomal abnormality in an infant with omphalocoele exceeds 50%, whereas infants with gastroschisis rarely have associated abnormalities, except for an increased incidence of intestinal atresia. Therefore, the long term outcome for neonates with omphalocoele is often determined by its associated anomalies, whereas infants with gastroschisis tend to achieve normal growth and developmental milestones as they progress through childhood. However this case report brings to light that association of gastroschisis with other anomalies should not be undermined and a keen suspicious look out for other anomalies is mandatory in gastroschisis cases as well. Moreover, the correlation of the intrauterine and post IUD imaging helps to identify the life threatening congenital anomalies and the importance of their early detection.

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