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Case Report

A case report on acrania with omphalocele

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ABSTRACT

Congenital/fetal malformations are structural, functional, and/or biochemical molecular defects present at birth. Prevalence of congenital fetal anomalies in developing countries are 2 to 3% of newborns and about 94% of severe birth defects. Acrania is a lethal congenital malformation characterized by partial or complete absence of flat bones in cranial vault. Another anomaly, omphalocele, a ventral defect of the umbilical ring resulting in herniation of the abdominal viscera, is one of the most common congenital abdominal wall defects seen in the newborn. Omphalocele is associated with trisomy. Incidence of acrania in India 1.4 to 2 per 1000 births. Incidence of omphalocele 1 in 4000 births. a case of 29-year-old, G2P1L1 with 14 weeks 2 days of gestation attended OPD with ultrasonographic report showing acrania with omphalocele. The sonographic scan revealed fully developed fetal brain having no calvarium. The brain was covered by a thin membranous structure. Facial structures showing frog eye appearance, open spina bifida. Normal and well-coordinated movements were noted in in the fetal body parts. The fetal abdomen showed a central mass protruding out from anterior abdominal wall. The mass is covered by a membrane and contains small intestines. There was direct insertion of the umbilical cord into the midline of omphalocele. Patient was counselled regarding fetal condition and its outcome. Patient was willing for termination. Medical termination of pregnancy was done. A single dead female fetus of fetus and placental weighing of 250 gm.

Keywords: Ultrasonography; Omphalocele, Acrania, Open spina bifida

INTRODUCTION

A midline abdominal wall defect of variable size, called omphalocoele is covered by a membrane of amnion and peritoneum with Wharton's jelly between the two layers, and enclosing abdominal contents.[1] It forms when the lateral ectomesodermal folds fail to meet in the midline, leaving abdominal contents covered by amnion and peritoneum into which umbilical cord inserts.[2] Fetal acrania is in which fetal brain is not surrounded completely or partially by skull bones. Fetal acrania can be diagnosed from 11 weeks onward. At 11-14 weeks gestation, the majority of cranial ossification is in the lateral aspects of the frontal bones and lower parietal bones, and no vault ossification is visible in the midline on a perfect mid sagittal image on USG. The incidence of isolated fetal omphalocele was estimated as 1:2165 (0.046%).[3,4] The detection rate and accurate rate of fetal omphalocele were 100% and 100% respectively. The recurrence risk for an isolated omphalocele in a subsequent pregnancy is <1%. The acrania, anencephaly sequence begins with acrania, which affecting the central nervous system with an incidence of ~1:1000 pregnancies.[5]

CASE REPORT

In this report, we present, a case of 29-year-old female, G2P1L1 with 14 weeks 2 days of gestation. She had unremarkable medical and obstetric history. The parents were non-consanguineous, and there was no significant family history of congenital anomalies. On, ultrasound scan she was found to have an anencephalic fetus with a soft tissue mass resemble to brain tissue attached to the cranial end of the fetus (Figure 3) The mother had urban

background and prior antenatal visits no record of her 1st day of menstrual cycle and expected date of delivery. On ultrasound examination the presentation of fetus was cephalic and longitudinal lie. The sonographic scan revealed. There was a single live fetus with a fully developed fetal brain having no calvarium (acrania) and facial structures were normal. The fetus showed normal cardiac activity (137 beats /min) with normal rhythm. The fetal body parts showed normal and well-coordinated movements. Placenta was anterior with normal thickness. Both orbits were symmetrical in size and shape, due absent cranial vault they appear as frog eye (Figure 4), open spina bifida involving cervical and thoracic spine-suggestive of spinal dysraphism. The fetal abdomen showed a central mass protruding out from anterior abdominal wall. The mass is covered by a membrane and contains small intestines were noted. On general physical examination-patient is vitally stable.

On per abdomen examination-uterus corresponds to 12-14 weeks size. FHR present @ 138 bpm HB 11.4 g%, blood group-B positive, platelet count-2.5 L, urine routine-normal, RBS-60 mg/dl, TSH-1.10 mIU/L, coagulation profile-normal, serology-normal.

USG-A single live intrauterine fetus of 14 weeks 2 days period of gestation with acrania and omphalocele, FHR 148 bpm.

Herniation of the bowel loops out of abdominal cavity without membrane likely to be omphalocele.

Absent cranial vault, frog eye appearance, open spina bifida involving cervical and thoracic spine-suggestive of spinal dysraphism.

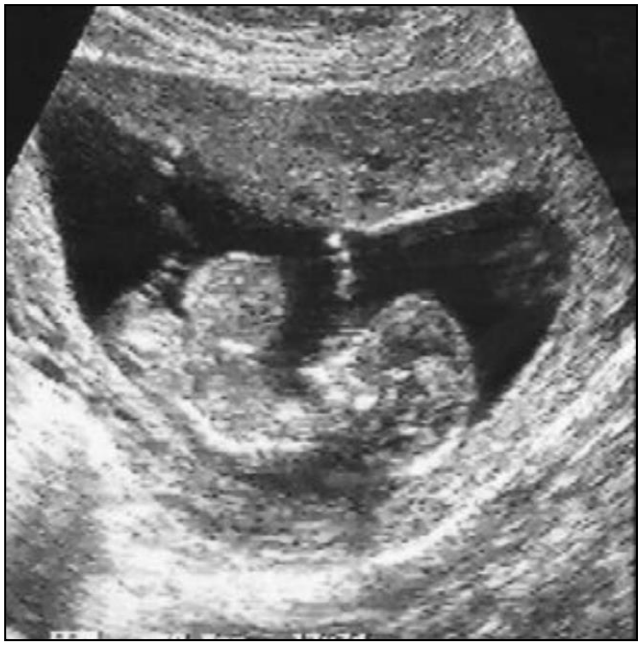

Figure 1: USG showing herniation of bowel loops and absent cranial vault.

Figure 2: Herniation of bowel loop (omphalocele).

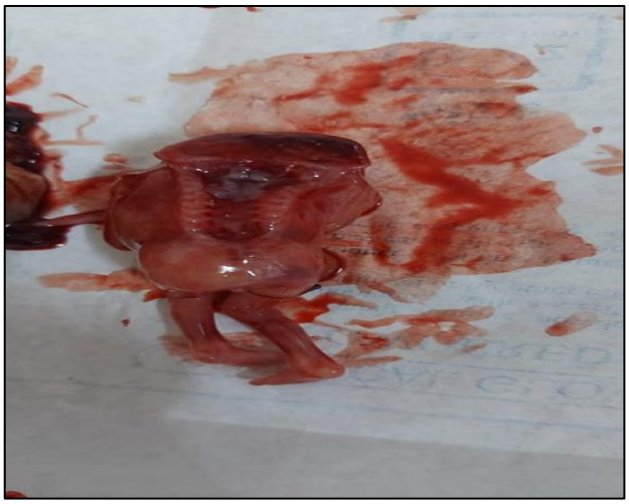

Figure 3: Absent cranial vault, spina bifid involving cervical and thoracic spine (spinal dysraphism).

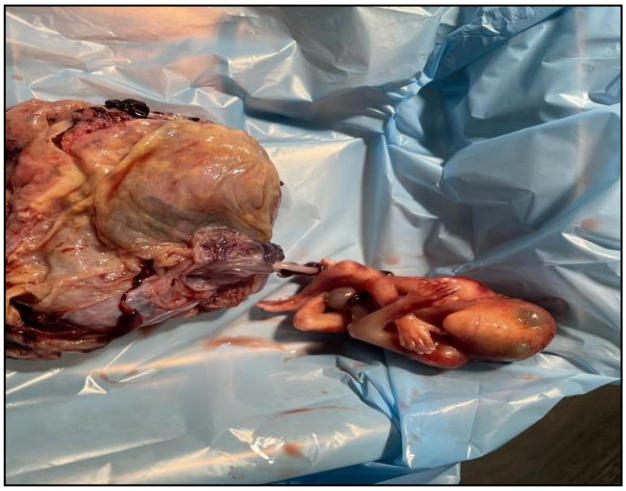

Figure 4: Absent cranial vault, frog eye appearance, with placenta.

Management

Patient was counselled regarding fetal condition and its outcome. Patient was willing for termination. After collecting required investigations, tab mifepristone 200 mg P/O tab misoprostol 400 mcg 4th hourly given inj. 10 IU oxytocin was started during expulsion procedure

After 3 hours expelled A single dead female fetus of fetus and placental weighing of 250 gm. Karyotyping of fetal specimen was denied by patient due to lack of affordability.

DISCUSSION

There is no inheritance pattern. Concomitant abdominal defect and neural tube defect are associated with trisomy 18. The cause of anencephaly is still a disputed entity but the defect is failure of closure of rostral neuropore.[6] Anencephaly is associated with anomalies of not only central nervous system but other system as well previous study has mentioned a wide range for percentage of fetus with associated anomalies like acrania, cleft lip, palate, amniotic band syndrome, oligo and polyhydramnios, VSD, omphalocele by Tan et al recorded 9.4% and David et al 84%.[7] Present study recorded a female fetus a study by Panduranga et al noticed preponderances in female fetuses than male in present study anomaly associated with anencephaly was spina bifida ,even though spina bifida was more common in lumbar region, craniospinal rachischisis of cervical region was found to be commoner according to Horowitz et al.[8]

CONCLUSION

Increasing awareness of maternal care, use of folic acid, early diagnosis, antenatal ultrasonography, proper counselling for this pregnancy and subsequent pregnancy can take care of the couple to face this dreaded complication of pregnancy. Pre-conceptional folic acid to be advised to all planned pregnancies. In high-risk pregnancy non-invasive test like triple test and quadruple test advised during early second trimester. Karyotyping should also be performed by amniocentesis or cord blood sampling or chorionic villus sampling. Early detection and termination of congenital anomaly will reduce the birth of babies with congenital anomalies. It will also ease the economic burden, psychological trauma to the parents and family.

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