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Case Report

Encephalocole

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Introduction

Omphalocele is rare type of Malformation. It is estimated to occur in 1/5000 of pregnancies (1-4). This mainly occurs due to closing defect of abdominal wall and sometimes its also associated with chromosomal abnormalities. This midline defect that occurs at umbilical ring often contains midgut and other abdominal organs such as liver, spleen and gonads. The etiology of Omphalocoele is not known , but there are various theories that have been proposed. The first theory proposed is persistence of primitive body stalk, second is failure of bowel to return to abdomen and the last theory includes failure of complete lateral body fold migration and body wall closure.

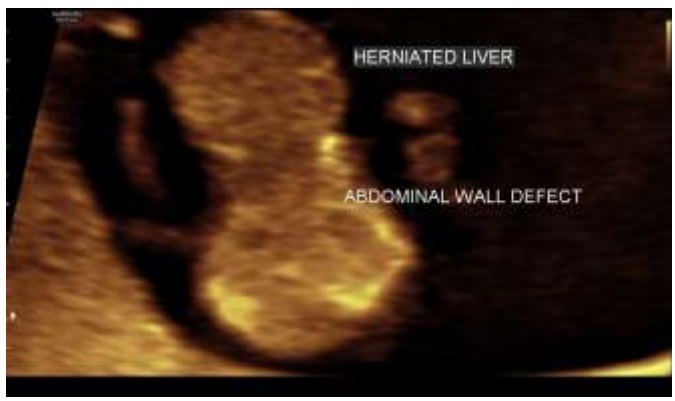
The Hernia contents are covered by outer amniotic layer, middle whartons jelly and inner peritoneal layer. Early screening allows prenatal counselling and safe delivery at tertiary care centre. Omphalocele is associated with other anomalies like Beckwith Wiedemann syndrome, pentalogy of cantrell, spinal defects syndrome. (5-8).

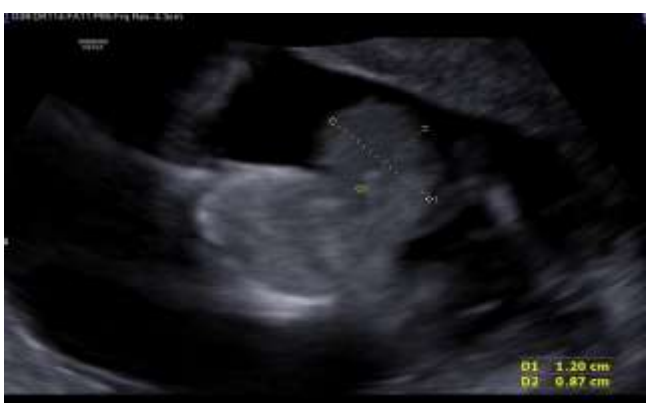
With availability of high-resolution prenatal Ultrasound accurate diagnosis of omphalocele can be made. Early screening and prenatal counselling can help in safe delivery at tertiary care centre.

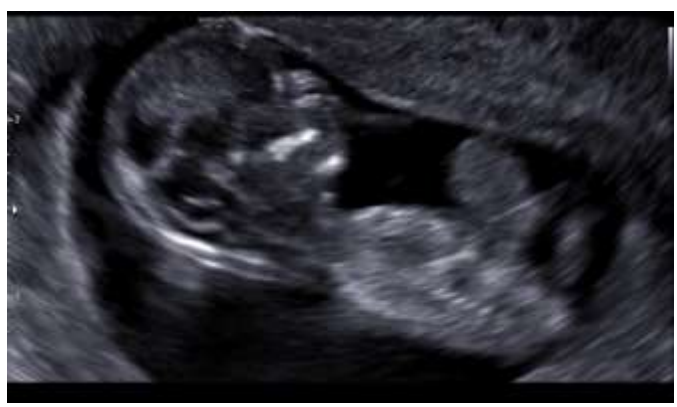
Case Report

40 yrs old primigravidae conceived by IVF in first attempt reported for routine Antenatal USG examination. There was no family history of any congenital anomaly or any previous spontaneous abortion. Ultrasound of 12 weeks gestation showed defect in anterior abdominal wall in the midline with a mass in the midline protruding through defect. Abdominal wall defect with sac containing liver covered with peritoneum. Herniated sac measures 12 mm x 8.7 mm x 13.6 mm. There is exomphalos. No foetal ascites or polyhydroamnios is seen. No other anomaly seen in fetus.

Diagnosis of Omphalocoele was made. Patient was advised for invasive testing for karyotype and molecular testing for beck with Wiedemann syndrome was performed and it did not show any abnormalities. The patient delivered at term with baby weight was 2.1 kg. Baby was admitted in neonatal unit and was planned for surgery after 6 months.







Discussion

Physiological Herniation occurs at eight weeks of embryogenic development, when foetal midgut extends intro extra embryogenic celons, occupying proximal segment of umbilical cord.

Bowel normally undergoes 270-degree clockwise rotation first 90 degree occurs by 10th week extra coelomically at the base of umbilicus, 180 degree clockwise turns occurs at level of superior mesentry artery. Midgut returns to abdominal cavity at 12 weeks of age. There is theory of omphalocele that failure of bowel to returns to abdomen.

There are also many chromosomal abnormalities associated with it like trisomy's 18, 13, and 21. This can be cardiac genitourinary, gastrointestinal, Neural Tubes, Head & Neck defect with Beckwith Wiedemann syndrome.

Omphalocele containing only bowel have higher incidence of Karyotyping abnormalities. Mortalities rate is 80% when associated with anomalies and it increase to 100% when chromosomal or cardiovascular anomalies are present.

The size of omphalocoele does not affect prognosis.

USG can be done for prenatal assessment of foetus. Earliest detection of Omphalocoele can be at 12 weeks. Magnetic resonance imaging can help in better management and counselling.

Associated Malformation

Association of omphalocele with other malformation is reported in various cases. Most commonly found anomalies are cardiac anomalies(tetralogy of Fallot) ,skeletal (club foot)and nervous and renal malformation(anencephaly, hydrocephalus,spinal bifida).

Obstetric Management of omphalocele depends on whether they are isolated or associated with chromosomal or malformation abnormalities(8-9)). Those with polymalformative syndrome or chromosomal anomaly termination of pregnancy is indicated. In Isolated omphalocoeles pregnancy can be continued with regular prenatal monitoring and delivery in specialized centre.

Conclusion

Prenatal diagnosis of omphalocoele is significant as omphalocoele is associated with chromosomal anomalies, early non invasive diagnosis is essential to allow the termination of pregnancy to be preferred early.

Prognosis of omphalocele depends on the process of congenital malformation or the chromosomal abnormalities rather than size.

Reference

1. Weir E., Congenital abdominal wall defects, CMAJ., 2003, 169, 809-810

2. Sípek A., Gregor V., Horácek J., Masátová D., Abdominal wall defects from 1961 to 2000--inci- dence, prenatal diagnosis and prevalence by ma- ternal age, Ceska Gynekol., 2002, 67, 255-260 (in Czech)

3. Hsu C.C., Lin S.P., Chen C.H., Chi C.S., Lee H.C., Hung H.Y., et al., Omphalocele and gastroschisis in Taiwan, Eur. J. Pediatr., 2002, 161, 552-555

4. Tan K., Kilby M.D., Whittle M.J., Beattie R.B., Booth I.W., Botting B.J., Congenital anterior abdominal wall defects in England and Wales 1987–1993: retrospective analysis of OPCS data, BMJ, 1996, 313, 903-906

5. Nicolaides K.H., Snijders R.J., Cheng H.H., Gosden C., Fetal gastro-intestinal and abdominal wall defects: associated malformations and chromosomal abnormalities, Fetal. Diagn. Ther., 1992, 7, 102-115

6. Smigiel R., Jakubiak A., Lombardi M.P., Jaworski W., Slezak R., Patkowski D., et al., Co-occurrence of severe Goltz-Gorlin syndrome and pentalogy of Cantrell - Case report and review of the literature, Am. J. Med. Genet. A., 2011, 155, 1102-1105

7. Cohen M.M. Jr., Beckwith-Wiedemann syndrome: historical, clinicopathological, and etiopathogenetic perspectives, Pediatr. Dev. Pathol., 2005, 8, 287-304

8. Mohsni S, Chachia A, Attia L, Ben Temime R, Ghodbane I, Makhlouf T, et al. Omphalocèle récidivante. Maghreb Médical. 2008;28(389):143-5.

9. Fratelli N, Parageorghiou AT, Bhide A, Sharma A, Okoye B, Thilaganathan B. Outcome of antenatally diagnosed abdominal wall defect. Ultrasound Obstet Gynecol. 2007;30(3):266-70.