



BMH Med. J. 2019;6(3):91-93. **Case Report**

Diagnosis of Pentalogy of Cantrell in First Trimester Ultrasound

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Abstract

Reporting first trimester diagnosis of a case of Pentalogy of Cantrell by Ultrasonogram. Routine antenatal ultrasound at 12 weeks and 4 days demonstrated a single live intrauterine fetus with anterior wall abdominal wall defect including herniated liver, bowel loops, ectopia cordis and cystic hygroma.

Introduction

Pentalogy of Cantrell is a rare congenital abnormality involving a midline anterior abdominal wall defect, defect of anterior diaphragm, defect of the apical pericardium, cleft distal sternum and intracardiac defect. The syndrome was first described in 1958 [1]. Affected individuals may or may not show the entire spectrum of structural abnormalities. The disorder was classified into 3 categories by Toyama [2]:

Class 1: Including all 5 components – confirmed diagnosis of Pentalogy of Cantrell

Class 2: Four defects including intracardiac and abdominal defect- probable diagnosis of Pentalogy of Cantrell

Class 3: Various combination of defects, always including sternal defect - incomplete expression of Pentalogy of Cantrell

Case presentation

Routine first trimester Nuchal Translucency scan at 12 weeks and 4 days was a combined trans-abdominal and trans-vaginal scan performed using Voluson 730Pro scanner. Scan showed a single live intrauterine fetus with generalized oedema and cystic hygroma (**Figure 1**). The nasal bone was absent. Ventral aspect of fetal abdomen show a globular supra umbilical mass containing fetal liver, bowel loops and herniated fetal heart through a sternal defect (**Figure 2**). Grey scale and colour Doppler evaluation confirmed the presence of fetal heart in the herniated mass (**Figure 3**). No evidence of intracardiac defect was made out.

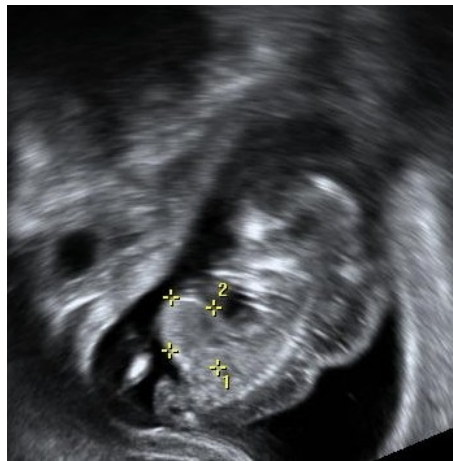


Figure 1: Generalised edema cystic hygroma and ventral defect

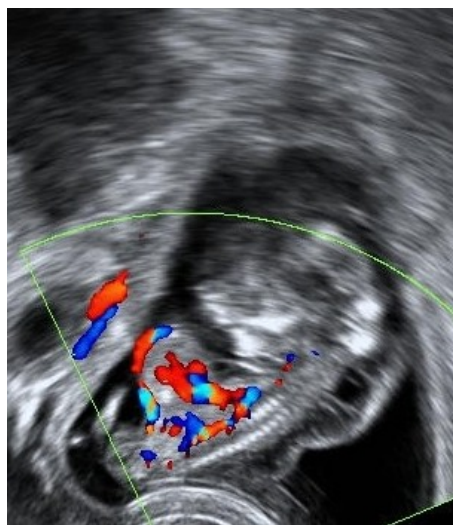


Figure 2: Omphalocele containing fetal heart



Figure 3: Pulsed Doppler showing cardiac activity in Omphalocele

Discussion

Pentalogy of Cantrell is a rare disorder with a prevalence of 1/65,000 - 1/200,000 [3]. The syndrome is considered multi factorial. Cantrell et al [1] proposed the theory of abnormal development of septum transversum, due to failure in differentiation of lateral mesoderm around 14-18 days of embryologic life. Most cases are sporadic, some case are associated with Trisomy 18. An X linked inheritance was also described. Very few cases are diagnosed in the first trimester and the routine use of trans-vaginal ultrasonography has improved the detection rate of such anomalies.

The differential diagnosis are amniotic band syndrome, body stalk syndrome and isolated ectopia cordis. The first trimester ultrasound diagnosis was first described Bennet et al [4] using grey scale and colour Doppler. Role of 3D ultrasonography and fetal MRI have also been reported [3,5]. These are more important in cases diagnosed in late gestation and deciding to continue pregnancy especially for pre operative planning. The diagnosis of ventral abdominal defect can only be confirmed after 12 weeks of gestation as the embryologic process of midgut herniation in to the base of umbilical cord may still be normal at the gestation.

Conclusion

Routine use of trans-vaginal ultrasonography has improved the visualization of normal and abnormal anatomy of a small first trimester fetus. Combination of colour Doppler further improves the detection of major defect like Pentalogy of Cantrell and hence timely counseling and intervention can be implemented.

References

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