

# DIAGNOSIS OF PENTALOGY OF CANTRELL IN FIRST TRIMESTER

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## INTRODUCTION/BACKGROUND :

**Pentalogy of Cantrell** (or **thoraco-abdominal syndrome**) is a rare syndrome that causes defects involving the diaphragm, abdominal wall, pericardium, heart and lower sternum.

Its prevalence is less than 1 in 1,000,000. Intrauterine diagnosis of this pentalogy is impossible before 12<sup>th</sup> week of gestation, because of herniation of bowel out of abdomen is a normal event in fetal development at this time, but after that ultrasonography is a useful method even in the first trimester. Differential diagnosis of fetal abdominal wall defect after 12<sup>th</sup> week is Omphalocele, pentalogy of Cantrell and Gastroschisis. If midline abdominal wall defect is present together with other abnormalities specially ectopia cordis one should consider pentalogy of Cantrell.

**MATERIALS & METHODS :** It's a case study .High resolution ultrasound was done using volume probe of SAMSUNG A35 MACHINE

**FINDINGS :** A 29 years primigravida with no positive history of diabetes, hypertension , smoking and with spontaneous conception came for first trimester screening. The findings were:

- 1) A single live intrauterine pregnancy of 12 week 1 day with CRL of 55.8 mm seen with grossly malformed baby showing increased nuchal translucency of 10.8mm
- 2) There was present an anterior abdominal wall defect as well as lower thoracic/ sternal defect with ectopia cordis , along with pericardial defect and 4 chamber heart was lying outside the chest cavity .
- 3) Stomach bubble is also seen outside along with liver .
- 4) The limbs were grossly normal with normal length of umbilical cord and normal placental tissue.



## DIFFERENTIAL DIAGNOSIS :

According to the above findings two differentials were put :

- 1 ANASARCA WITH PENTALOGY OF CANTRELL.
2. AMNION RUPTURE SEQUENCE OR BODY STALK DEFORMITY

## DISCUSSION & CONCLUSION

The findings supporting Pentalogy of Cantrell were: ectopia cordis, pericardial and sternal defect and anterior abdominal wall defect. Diaphragmatic defect was not sure. In addition there were no visible limb abnormalities as all the four limbs were normally seen. Placenta appeared normal with normal cord length. No midline facial defect and encephalocele was noted .So ,the first possibility was PENTALOGY OF CANTRELL. No supportive evidence of amnion rupture sequence or body stalk deformity was found.

The patient refused karyotype. Since both the deformities have poor prognosis termination was offered and accepted by the patient..

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