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case report of pentalogy of cantrell

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Pentalogy of Cantrell (PC) is an extremely rare multiple congenital malformation syndrome
It was first described in 1958 by James R. Cantrell et al.1
It comprises of the following five characteristics:
(1) midline, upper abdominalwall disorder (e.g. omphalocele, gastroschisis)
(2) defects of the lower sternum (i.e. cleft sternum or absent sternum)
(3) anterior diaphragmatic defect (i.e. hypoplastic diaphragm, anterior diaphragmatic hernia)
(4) pericardial abnormality (e.g. ectopia cordis)
(5) congenital abnormalities ofthe heart (e.g. tetralogy of Fallot, ventricular septal defect, atrial septal defect).1
They first described the syndrome, attributed these anomalies to
developmental failure between 14 and 18 days of embryonic life
involving an inappropriate differentiation of a segment of the lateral
mesoderm however the exact pathogenesis of this condition remains unclear. 2
The incidence of the syndrome is about 1 in 65,000 live births.3

In these cases, the prognosis depends on the severity of the anomaliessuch as cardiac and extracardiacdefects and other associated anomalies. There are a few reported survivors after corrective surgery.4

I. Case Report:

A 24 Year old Primigravida with 13 Weeks 2 Days Period of gestation came for routine antenatal checkup & NT scan was done which showed features of Pentalogy of Cantrell & Genetic Testing was suggested but due to Patient non-affordability routine serum marker tests were not done &patient was advised for Termination of Pregnancy.

Patient was induced with Cerviprime(Dinoprostone) gel 6 hours apart & Patient aborted a Fetus weighing 40 g at 1:15 AM on 17/01/22 & Check scan was done &showed Retained Products of Conception for which USG guided Dilatation & Curettage done under Short General Anesthesia & Pre-conceptional Counselling for next Pregnancy was advised at the time of discharge.

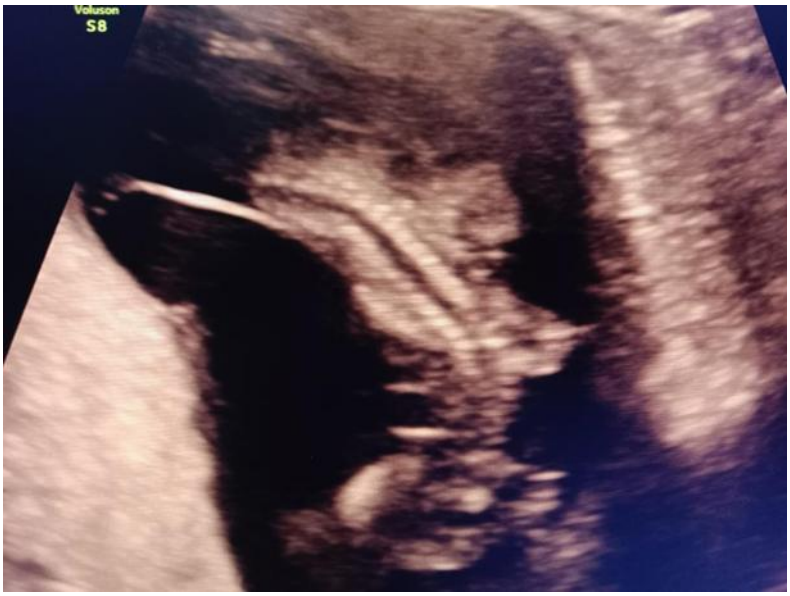


Image is showing SCOLIOSIS which is sideways curvature of the spine

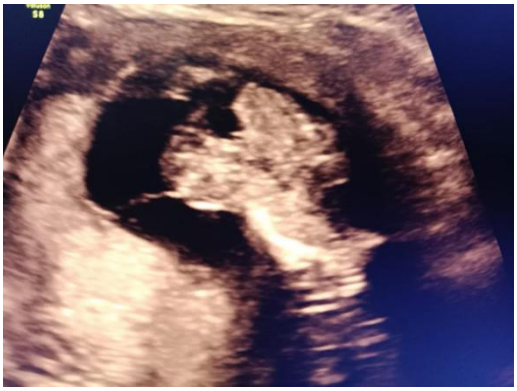
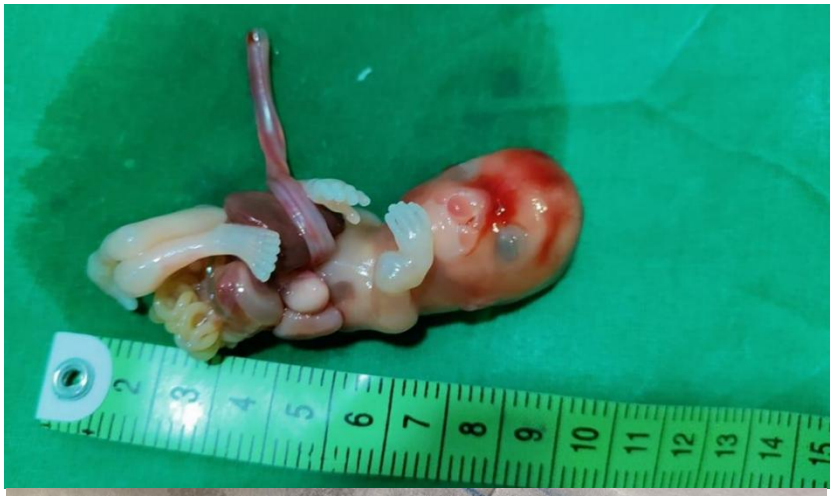
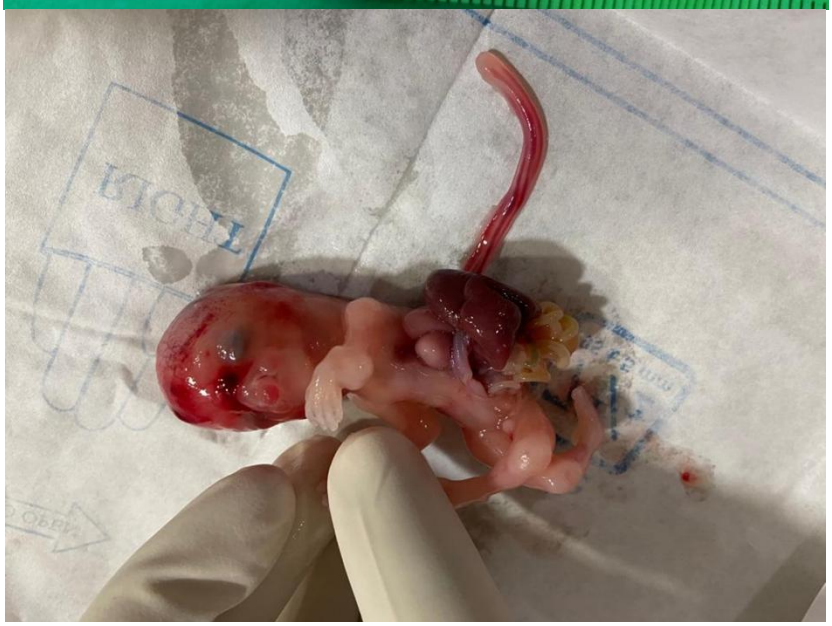


Image is showing OMPHALOCELE which is abdominal wall defect Where abdominal organs protrude through a hole into the base of the umblical cord.





On Exteranal Examination:
Scoliosis seen
Polydactyly seen –left foot-4 digits seen
Bilateral radius, ulna, tibia and fibula absent
Ruptured omphalocele showing heart (ectopia cordis), left lung. Liver, spleen, stomach,smalland large intestine and left kidney lying outside
Anal atresia seen

On Dissection:
Right lung and right kidney found in situ
Agenesis of the diaphragm seen
Congenital heart disease could not be detected

On Infantogram:
? Ruptured omphalocoele
Ectopia cordis
C-shaped scoliosis with widening of the cervical spinal canal
Multiple limb defects-Bilateral radius, ulna, tibia and fibula absent and plastic deformation of bilateral upper and lower limbs noted. Polydactylynoted.

II. Discussion:

The exact cause of Pentalogy of Cantrell remains still unknown, but thesyndrome is considered to be of heterogeneous origin,caused by multiple factors such as gene mutation, chromosomalabnormalities such as trisomy 13 and 18 and disrupted vessels defects and physical and chemical teratogens. Thewidely accepted hypothesis suggests that there is a failurein migration of the primordial mesodermic structures of thechest and abdomen during embryologic development atgestational age of 14–18 days.1

Mutation of TAS gene which mapped at Xq25-q26.1 area, is mentioned to has a roll in fusion of sternum, multiple cardiac, diaphragmatic and anterior abdominal wall defects, and also additional abnormalities reported in some cases of Cantrell's pentalogy.4

The cardiac abnormalitiesare the result of developmental disorders of epimyocardium.Pericardial and diaphragmatic defects are due to developmentalfailure of the transverse septum.

Intrauterine diagnosis of this pentalogy is impossible before 12th 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 12th week is Omphalocele, pentalogy of Cantrell and Gastroschisis.4

Most reported cases of Pentalogy of Cantrell are thought to be sporadic in occurrence; However, Mutations in Genes located on the X chromosome have been associated with ventral midline disruptions. An X-Linked dominant inheritance Pattern has been observed in some families.

III. Conclusion:

The present case had almost all the features of Pentalogy of Cantrell. infantogram showed ruptured omphalocele and ectopia cardis along with that it has scoliosis, polydactyly and bilateral radius, ulna,tibia and fibula were found be absent.

Pentalogy of Cantrell accompanied by ectopia cordis is a rare phenomenon with a broad range of clinical symptomatology and consequences, which demand a multidisciplinary team to be appropriately assessed for effective prenatal counseling and postnatal management.5

Prenatal imaging studies are essential not only for prenatal counseling but also for adequate postnatal therapeutic planning.

These can be easily diagnosed prenatally by two-dimensional (2D) or even three-dimensional (3D) ultrasonography combined with Doppler, karyotyping and genetic counselling for associated anomalies are critical since infant survival assessment and early diagnosis give the parents the option of terminating the pregnancy.

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