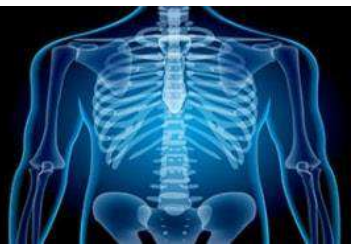
International Journal of Radiology and Diagnostic Imaging



E-ISSN: 2664-4444
P-ISSN: 2664-4436
www.radiologypaper.com
IJRDI 2020; 3(2): 80-82
Received: 16-02-2020
Accepted: 19-03-2020

Vijaykumar S. Mane
Associate Professor,
Department of Radiodiagnosis,
Bharati Vidyapeeth (Deemed to be) University, Sangli,
Maharashtra, India

Dr. Diksha Goyal
Junior Resident-2,
Department of Radiodiagnosis,
Bharati Vidyapeeth (Deemed to be) University, Sangli,
Maharashtra, India

Dr. Anil G Joshi
Professor & H.O.D,
Department of Radiodiagnosis,
Bharati Vidyapeeth (Deemed to be) University, Sangli,
Maharashtra, India

Corresponding Author:
Dr. Anil G Joshi
Professor & H.O.D,
Department of Radiodiagnosis,
Bharati Vidyapeeth (Deemed to be) University, Sangli,
Maharashtra, India

Early detection of pentalogy of cantrell by sonography

Vijaykumar S Mane, Dr. Diksha Goyal and Dr. Anil G Joshi

DOI: http://dx.doi.org/10.33545/26644436.2020.v3.i2b.100

Abstract
Pentalogy of Cantrell is a rare congenital condition with a thoraco-abdominal disruption showing five characteristics i.e. ectopia cordis and intracardiac anomalies; lower sternal defect; midline supraumbilical thoraco-abdominal wall defect; anterior diaphragmatic defect; defect of pericardium. Ultrasound can be used for its early and confirmatory diagnosis. This is a study of a rare case of Pentalogy of Cantrell based on sonography findings in a 23 year old primigravida presenting for routine antenatal check-up.

Keywords: Pentalogy of Cantrell, ultrasound, congenital anomalies

Introduction
A 23 year old pregnant woman came for a routine antenatal checkup at 12 weeks of pregnancy.
On performing sonography, various congenital anomalies were detected in the fetus.

Materials and Methods
A complete review of the patient record was obtained. A high end ultrasonography machine was used with convex and trans-vaginal probes.The findings were documented by performing trans-abdominal and trans-vaginal scans to aid in making an effective diagnosis.

Results
The Ultrasound findings were as follows:
- Single intrauterine live fetus with a corresponding gestational age of 12 weeks
- Placenta was normal, Amniotic fluid was adequate.
- An anterior abdominal wall defect with herniation of liver and bowel loops through it
- A defect in the lower anterior chest wall with heart lying outside the thoracic cavity
- The brain showed deficient skull vault with dilated ventricles suggestive of anencephaly

Since the patient had the following findings, it leads to the diagnosis of Pentalogy of cantrell- class 3.

Images

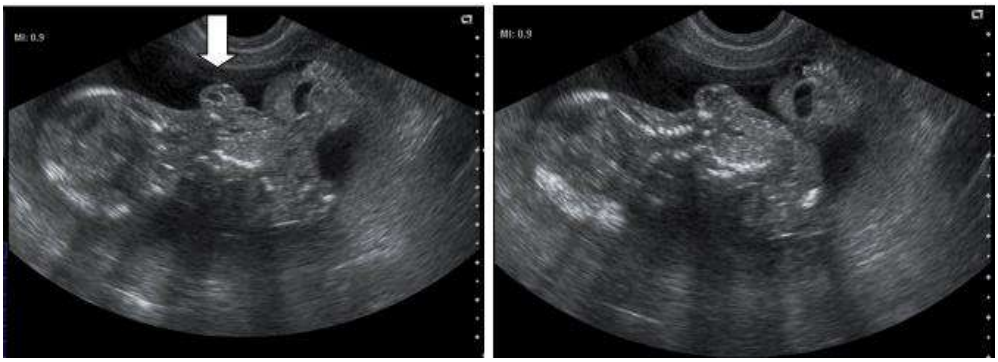


Fig 1 & 2: Showing lower anterior chest wall defect with heart lying outside the thoracic cavity

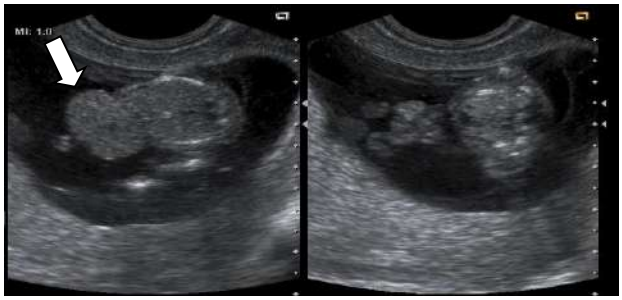


Fig 3: Showing herniation of liver and bowel loops is seen through an anterior abdominal wall defect

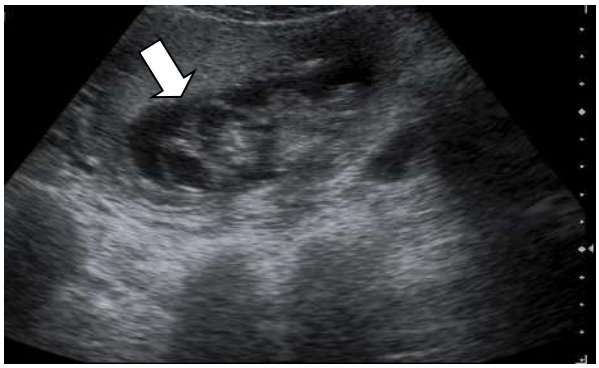


Fig 4: Head showing deficient skull vault with dilated ventricles s/o anencephaly

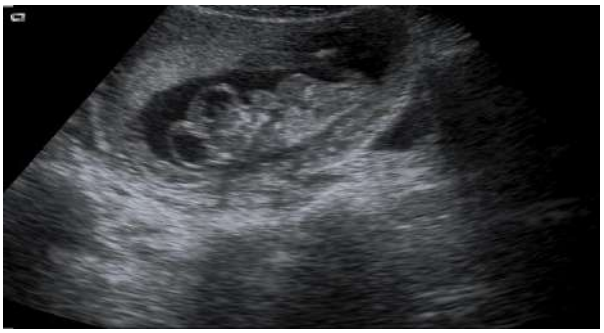


Fig 5: Showing a midline defect and herniation of the thoraco-abdominal organs is noted head showing deficient skull vault with dilated ventricles s/o anencephaly

Discussion

Pentalogy of Cantrell is a very rare syndrome (1) with an incidence of around 6 per million live births

It was first described in 1958 by Cantrell and consists of the following 5 anomalies:

1. A midline supraumbilical abdominal wall defect
2. A defect in the diaphragmatic pericardium
3. A defect in the lower sternum
4. A defect in the anterior diaphragm
5. Congenital intracardiac abnormality

The association of an omphalocele with ectopia cordis is the hallmark of this syndrome. Multiple variants have been reported, and few patients display the complete spectrum of disease.

The mortality rate is high and timely diagnosis on USG leads to appropriate management of the pregnancy.

In 1972, Toyama suggested an alternative classification system to describe the more common variants of this syndrome: (2)

Class 1: definite diagnosis with all 5 defects present

Class 2: probable diagnosis with presence of 4 defects (including ventral wall defect abnormalities and cardiac defects)

Class 3: incomplete expression (includes various combination of defects present, including a sternal abnormality)

On an embryologic basis, Cantrell et al proposed that the association of defects is mesodermal in origin, occurring in the third week of embryonic life.

- Abnormal migration of splanchnic and somatic mesoderm with premature breakage of the vitelline sac resulting in a midline defect.
- -Abnormal formation of the transverse septum of the diaphragm occurs because of abnormal migration of myoblasts.
- -Premature atrophy of the cardinal vein leads to associated pericardial defects

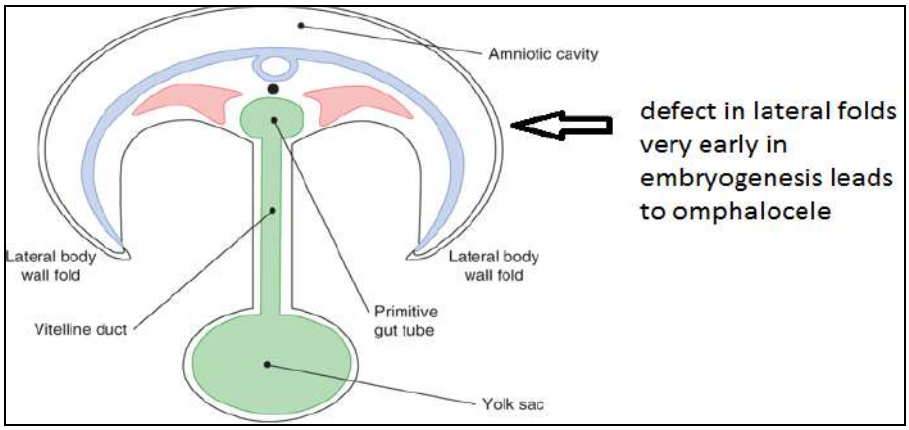


Fig 6: Abnormal migration of splanchnic and somatic mesoderm with premature breakage of the vitelline duct results in a midline defect

Pentalogy of Cantrell may be associated with various other congenital defects like:

- CNS malformations like neural tube defects, encephalocele, hydrocephalus
- Craniofacial defects like cleft lip and/or palate, exencephaly
- Limb defects like club foot, absence of tibia or radius, hypodactyly
- Abdominal organ defects like intestinal malrotation, polysplenia, gallbladder agenesis
- Skeletal manifestations like scoliosis, lordosis, and hemivertebrae

- A wide range of intracardiac anomalies have been associated with PC including septal defects, tetralogy/pentalogy of Fallot, total anomalous pulmonary venous drainage, and left ventricular diverticulum [3, 4, 5, 6, 7, 8]. Holt-Oram syndrome, which was seen in the family of Case 1, is associated with cardiac anomalies but has never been reported with PC.

A variety of mutations including deletions or duplications of the PORCN, thoraco-abdominal syndrome (TAS), ALDH1A2, and teneurin-4 (TENM4) genes have been identified [8, 9, 10, 11, 12] to be associated with Pentalogy of Cantrell. It can also be associated with rare anomalies, such as craniorachischisis, pulmonary extrophy, and Goltz-Gorlin syndrome [8, 13, 14, 15, 16].

Conclusion

Pentalogy of Cantrell is a complex syndrome of numerous fetal anomalies that should be borne in mind during the ultrasound evaluation of an omphalocele, sternal defects, ectopia cordis

Since the pathology is also chromosomal based, the counselling should be done for future pregnancies as it can be challenging to manage. Therefore, prenatal diagnosis and careful assessment is essential for appropriate postnatal management [17, 18].

Ultrasonography should always be the primary modality for diagnosis which gives an opportunity to parents to be adequately counseled regarding management options.

References

1. Alagappan P, Chellathurai A, Swaminathan TS, Mudali S, Kulasekaran N. Pentalogy of Cantrell. Indian J Radiol Imaging. 2005; 15:81-4.
2. Toyama WM. Combined congenital defects of the anterior abdominal wall, sternum, diaphragm, pericardium, and heart: a case report and review of the syndrome. Pediatrics. 1972; 50:778-792.
3. Restrepo MS. Cerqua A, Turek JW. Pentalogy of Cantrell with ectopia cordis totalis, total anomalous pulmonary venous connection, and tetralogy of Fallot: A case report and review of the literature. Congenit. Heart Dis. 2014; 9:E129-E134. [Google Scholar] [CrossRef]
4. Singh N, Bera ML, Sachdev MS, Aggarwal N Joshi R, Kohli V. Pentalogy of Cantrell with left ventricular diverticulum: A case report and review of literature. Congenit. Heart Dis. 2010; 5:454-457. [Google Scholar] [CrossRef]
5. Balderrábano-Saucedo N, Vizcaíno-Alarcón A, Sandoval-Serrano E, Segura-Stanford B, Arévalo-Salas LA, de la Cruz, L.R.; Espinosa-Islas, G.; Puga-Muñuzuri, F.J. Pentalogy of Cantrell: Forty-two years of experience in the Hospital Infantil de Mexico Federico Gomez. *World J. Pediatr. Congenit. Heart Surg.* 2011; 2:211-218. [Google Scholar] [CrossRef]
6. Mallula KK, Sosnowski C, Awad, S. Spectrum of Cantrell's pentalogy: Case series from a single tertiary care center and review of the literature. *Pediatr. Cardiol.* 2013; 34:1703-1710. [Google Scholar] [CrossRef]
7. Ojha V, Sh C, Ganga KP, Saxena A, Gulati G. Congenital left ventricular diverticulum in Pentalogy of Cantrell: Puzzle solved on dual source CT. *Ann. Thorac. Surg.* 2019; 108:e205. [Google Scholar] [CrossRef] [PubMed]
8. Kylat RI. Complete and Incomplete Pentalogy of Cantrell. *Children (Basel)*. 2019; 6(10):109. Published 2019 Oct 6. doi:10.3390/children6100109
9. Steiner MB, Vengoechea J, Collins RT. 2nd. Duplication of the ALDH1A2 gene in association with pentalogy of Cantrell: A case report. *J. Med. Case Rep*. 2013; 7:287. [Google Scholar] [CrossRef] [PubMed]
10. Lombardi MP, Bulk S, Celli J, Lampe A, Gabbett MT Ousager LB, et al. Mutation update for the PORCN gene. *Hum. Mutat.* 2011; 32:723-728. [Google Scholar] [CrossRef] [PubMed]
11. Parvari R, Weinstein Y, Ehrlich S, Steinitz M, Carmi R. Linkage localization of the thoraco-abdominal syndrome (TAS) gene to Xq25-26. *Am. J. Med. Genet.* 1994; 49:431-434. [Google Scholar] [CrossRef]
12. MacTaggart B, Bowen C, Markowitz M, Chong J, Bamshad M, Ma X, Adelstein RS. Using whole-exome sequencing to identify genetic variants in patients diagnosed with pentalogy of Cantrell. In Proceedings of the 30th Annual Showcase of NIH Intramural Research, Bethesda, MD, USA, 14 September 2016. [Google Scholar]
13. Atis A, Demirayak G, Saglam B, Aksoy F, Sen C, Craniorachischisis with a variant of pentalogy of Cantrell, with lung extrophy. *Fetal Pediatr. Pathol.* 2011; 30:431-436. [Google Scholar] [CrossRef]
14. Smigiel R, Jakubiak A, Lombardi MP, Jaworski W, Slezak R, Patkowski D. Hennekam, R.C. 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. [Google Scholar] [CrossRef]
15. Madžarac V, Matijević R, Škrtić A, Duić Ž. Fistonić N, Partl JZ. Pentalogy of Cantrell with unilateral kidney evisceration: A case report and review of literature. *Fetal Pediatr. Pathol*. 2016; 35:43-49. [Google Scholar] [CrossRef]
16. Albu C, Staicu A, Popa-Stănilă RT, Milaș AC, Chiriac LB, Kovács TE. Multidisciplinary approach of assessing malformed fetuses exemplified in a rare case of pentalogy of Cantrell associated with craniorachischisis, pulmonary extrophy and right-sided aortic arch with aberrant brachiocephalic artery. *Rom. J. Morphol. Embryol.* 2018; 59:911-915. [Google Scholar]
17. Victoria T, Andronikou S, Bowen D, Laje P, Weiss DA, Johnson AM. Et al. anterior abdominal wall defects: Prenatal imaging by magnetic resonance imaging. *Pediatr. Radiol.* 2018; 48:499-512. [Google Scholar] [CrossRef]
18. Türkyilmaz G, Avcı S, Sıvrıkoz T, Erturk E, Altunoglu U, Turkyilmazlmaz SE. *et al*. Prenatal diagnosis and management of ectopia cordis: Varied presentation spectrum. *Fetal Pediatr. Pathol.* 2019; 38:127-137. [Google Scholar] [CrossRef]