

Case Report**Pentalogy of Cantrell: a case report**

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Abstract

Cantrell's pentalogy (CP), a rare congenital malformation, consists of the supraumbilical abdominal wall defect, the sterna lower part defect and agenesis of the anterior portion of the diaphragm, an absence of the diaphragmatic part of the pericardium, and a malformation of cardia. This case report presents a female neonate, who was born at 32 weeks of conception, weighing 1300 g and was admitted one hour after delivery. She had the five anatomical defects known for Cantrell's Pentalogy. Moreover, autopsy revealed a bilateral cleft lip and palate, a patent ductus arteriosus, and an atrial and ventricular septal defect.

KEYWORDS: Ectopia Cordis, Abdominal Wall, Defects, Cleft Lip and Palate Associated Transmembrane Protein 1, Human, Equinovarus.

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The first description of pentalogy of Cantrell was made by Cantrell et al in 1958, who reported 5 cases with this anomaly. A pentad of findings that included a defect of the lower sternum, a midline supraumbilical thoracoabdominal wall defect, a deficiency of the diaphragmatic pericardium, a deficiency of the anterior diaphragm, and congenital cardiac anomalies was reported in these cases.¹

Up to know, there are some case reports that have been classified as full spectrum, which have all of the five defects and incomplete forms with lesser defects of the pentalogy of Cantrell.²⁻⁴ In these cases, the prognosis depends on the severity of the anomalies, such as cardiac and extracardiac defects, and other associated anomalies. There are a few reported survivors after corrective surgery.²

Case Report

A female neonate, one hour after delivery, was admitted at Pathology ward of Ghaem Hospital for autopsy. She was born by a preterm de-

livery at 32 weeks of conception, weighing 1300 g, from a 39-year-old G1P0A0 woman with no significant finding in her medical history. His father has a past history of addiction to alcohol and opium. Immediately after delivery, the neonate transmitted to NICU, but she died one hour after delivery.

A prenatal sonogram at 20 weeks showed alive fetus with Gastroschisis. Further 3D sonogram showed a fetus with a defect in the body wall and sticking out liver (Figure 1 and 2).

In physical examination, there were a defect in thoracoabdominal wall with ectopia cordis, an absence of pericardium, an evisceration of the intestines and liver, and a bilateral cleft lip and palate.

Radiographic finding showed an absence of mid portion and the distal segment of the sternum.

In autopsy, a bilateral cleft palate and lip, an evisceration of the liver and intestines, and a thoracoabdominal schisis with ectopia cordis were seen (Figure 3 and 4). The extension

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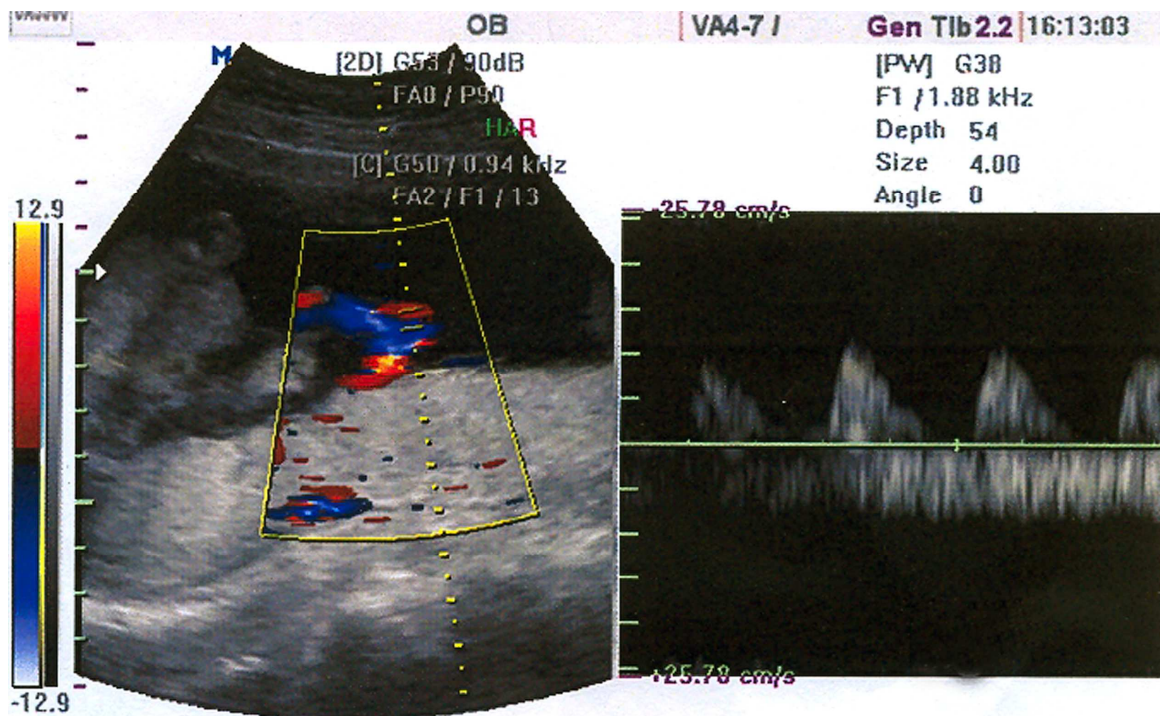


Figure 1. Color Doppler showing ectopia cordis

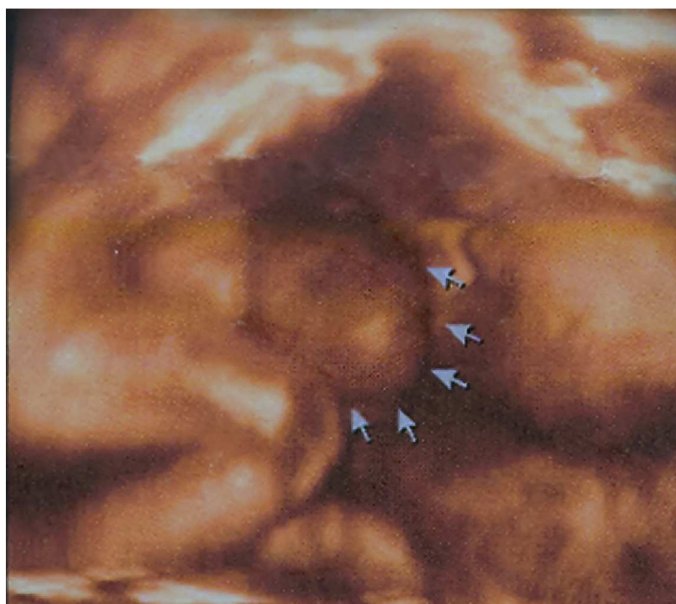


Figure 2. 3 dimation sonogram showing sticking out liver

of the thoracoabdominal wall defect was from the mid-sternum to the supraumbilical area. Additionally, left sided clubfoot was seen, and there was an anterior diaphragmatic defect.

There was a thoracic ectopia cordis of the heart with a rotation of 100 degrees infer superiorly, and its apex toward the midline of the

neck. An absence of pericardium was found. There were defects in the atrial and ventricular septum, and a patent ductus arteriosus. There was no remarkable finding in the rest of the internal examination.

Microscopic examination revealed chronic inflammation and congestion of the liver, epicardium, and bowel serosa.



Figure 3. Ectopia cordis, midline supraumbilical defect of thoracoabdominal wall and evisceration of liver and intestines

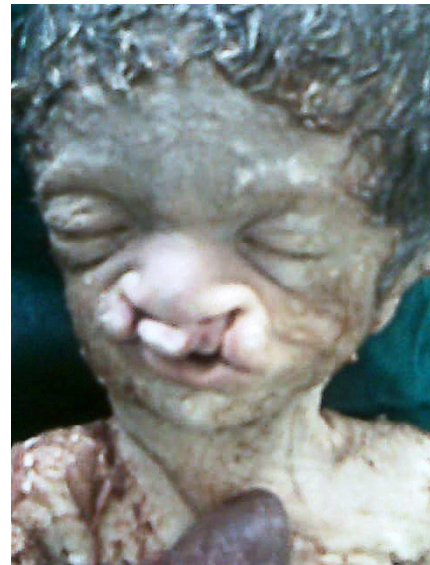


Figure 4. Bilateral cleft lip and palate

Discussion

Cantrell pentalogy is a rare congenital thoracoabdominal disruption, first described by Cantrell et al with five characteristic:

1) Ectopia cordis and intracardiac anomalies; 2) lower sternal defect; 3) midline supraumbilical thoracoabdominal wall defect; 4) anterior diaphragmatic defect; and 5) defect of diaphragmatic part of pericardium that results in relation between pericardial cavity and peritoneum.¹

Prevalence of pentalogy of Cantrell is about 1 per 65000 live births and classified as a developmental defect of midline anterior body wall. Full pentalogy of Cantrell is a severe and rare syndrome, but incomplete forms with combination of two or three defects were reported frequently.² Intracardiac anomalies that are constant portion of pentalogy of Cantrell are VSD (in 100% of cases), ASD (52%), pulmonary stenosis (33%) and Tetralogy of Fallot (20%).¹

Also sternal fusion defect is a rare malformation and an inferior type is seen in pentalogy of Cantrell.³

Cantrell offered a developmental failure in lateral mesoderm during day 14-18 as a reason for indecision of transverse septum of dia-

phragm, therefore migration of paired mesodermal fold does not occur.¹ Failure of the transverse septum to develop, as well as abnormal development of the myocardium, cause diaphragmatic and cardiac defect, respectively.⁴

Because of various phenotypes of abdominal wall defect in Cantrell's pentalogy, multiple factors is said to be responsible, including mechanical teratogens, major gene mutation, chromosomal abnormalities such as trisomy 13 and 18 and disrupted vessels defects.⁵

Mutation of TAS gene which mapped at Xq25-q26.1 area, is mentioned to have a role in fusion of sternum, multiple cardiac, diaphragmatic and anterior abdominal wall defects, and also additional abnormalities reported in some cases of Cantrell's pentalogy.^{6,7} Carmi et al reported some cases with encephalocele and cleft lip with or without cleft palate, in association with abdominal wall defects such as pentalogy of Cantrell.⁷

In some cases of Cantrell of pentalogy, aggregation of fluid in the chest and neck cavity was reported as a result of venous congestion because of cardiac failure, increased mediastinal pressure due to diaphragmatic herniation or Omphalocele.⁵

Abnormalities of the extremities are also reported in associated with few cases of pentalogy of Cantrell.⁸⁻¹⁰ One study reported arthrogyriposis, left thumb defect and shortening of left upper limb together with exencephaly.¹¹ Peixoto-Filho et al mentioned that clubfoot was seen in few cases.¹² Also in the presented case clubfoot was seen.

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.^{12,13} Differential diagnosis of fetal abdominal wall defect after 12th 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.¹⁴

Both 2D and 3D obstetric ultrasonography are recommended, but 3D ultrasonography is not necessary in first trimester.¹² Other diagnostic methods including CT-Scan and MRI can be used for confirmation.^{4,15,16}

Prognosis of pentalogy of Cantrell depends on severity of intra and extra cardiac defects,

pulmonary hypoplasia, extent of abdominal wall defect, cerebral anomalies and diaphragmatic herniation. The mean survival rate without any interventional surgery is about 36 hours.³ Studies showed that even with care monitoring in professional centers and multiple corrective surgeries, they had high morbidity and mortality rate and long time prognosis is poor.¹⁷

Conclusions

The presented case had all portions of pentalogy of Cantrell as well as midline supraumbilical wall defect and ectopia cordis, lower sternal, pericardial and diaphragmatic defects together with bilateral deep cleft lip/palate and left side clubfoot which were reported in other case reports. These severe anomalies resulted in her premature death.

Prenatal routine obstetric ultrasonography of the case revealed abdominal wall defect with evisceration of intestines and liver which confirmed by 3D ultrasonography and Gastroschisis was offered. But diagnosis of pentalogy of Cantrell was made after autopsy.

Conflict of Interests

Authors have no conflict of interests.

Authors' Contributions

AHJ and AAO performed autopsy and histological evaluation. AF approved the genetic diagnosis. HS wrote the manuscript. BJ edited the manuscript. All authors have read and approved the content of the manuscript.

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