



BMH Med. J. 2021;8(4):143-148. **Case Report**

## Pentalogy of Cantrell

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### Abstract:

It is a syndrome complex that consists of an upper anterior abdominal wall defect with an ectopia cordis. The cardinal features are upper anterior abdominal wall defect, lower anterior sternal defect, anterior defect of the diaphragm, diaphragmatic pericardial defect and associated congenital cardiac anomalies. The prognosis of the condition is poor, and if at all a favourable outcome always depends on the severity of the cardiac anomalies that coexisted in that condition. The exact pathology of the condition is not identified, however many associations have been described.

**Keywords:** Pentalogy of Cantrell, abdominal wall defect, sternal defect, diaphragm

### Introduction

The condition was first described by James R Cantrell in 1958 [1]. Five anomalies of this disease condition are the following:

1. Midline supraumbilical abdominal wall defect.
2. Defect in the diaphragmatic pericardium.
3. A defect in the lower anterior sternum.
4. A defect in the anterior diaphragm and finally.
5. Various intracardiac anomalies.

In most severe case heart herniates out through the diaphragmatic defect, resulting in ectopia cordis. Large omphalocele with ectopia cordis is the hallmark of the syndrome. Full display of the syndrome complex may happen only in fewer cases. Survival and prognosis always depend on the complexity of associated anomalies, especially the cardiac.

In addition to above mentioned classical features of this Syndrome complex, in the year 1972, Toyama suggested an alternative classification of the disease [2]:

Class 1: All the five cardinal features are evident, then it is a definite case of the Pentalogy of Cantrell.

Class 2: Four anomalies present including ventral wall defect and cardiac defects.

Class 3: Incomplete expression of the syndrome, various combinations of defects with external anomalies.

Various dermatological and skeletal manifestations also coexist with these conditions, so it may be corrected as hexalogy or hepatology of Cantrell.

### **Prevalence and Epidemiology**

Prevalence of this syndrome complex may vary from 1:65000 to 5.5: 1million live births [4]. This wide range happens due to the expressibility of the criteria made by the Toyama. Full-blown features of this syndrome complex, may not manifest in all the affected cases.

### **Aetiology, Embryology and Pathophysiology**

Cantrell et al postulated that the condition is due to the heterogeneous representation of various factors, and the pathology is of mesodermal origin [3]. One theory that came to its stand is the abnormal migration of splanchnic and somatic mesoderm [5] with breakage of the vitelline sac, in the third week of life.

Abnormal migration of transverse septum of the diaphragm occurs due to abnormal migration of myoblasts, premature atrophy of cardinal veins accounts for the pericardial defects. Mutations in the X-linked chromosome accounts for the midline defect. Most reported cases of Pentalogy of Cantrell are sporadic in occurrence, however, mutations in X linked chromosome have been associated with ventral midline defect.

A recent case report demonstrated, maternally inherited microduplication of chromosome 15q21.3 on the postnatal chromosomal microarray of a newborn with a prenatal diagnosis of Pentalogy. This region includes the gene ALDH1A2, which encodes for retinaldehyde dehydrogenase type 2, an enzyme that has a critical role in the development of the heart and diaphragm. Thereby demonstrating a biologically plausible explanation of the spectrum of anomalies in the Pentalogy of Cantrell.

Antenatal screening is needed [5,6], to detect the anomaly in the first trimester itself, more of it is successive scanning in the first trimester shows a progressive omphalocele of more than 1cm [7,8] should be considered as pathological or it's persistence after 14 weeks, both are ominous signs with fatal outcome.

### **Manifestations of Disease**

#### ***Clinical presentation***

Types of defects include:

1. Abdominal wall defects (giant omphalocele, diastasis of rectal muscles).
2. Anterior diaphragmatic hernia.
3. Anterior chest wall defect with extrusion of the heart outside the thoracic cavity.

4. Thoracoabdominal ectopic cordis.
5. Cardiac Anomalies, VSD, ASD, Tetralogy of Fallot, ventricular diverticula and ventricular aneurysm.
6. CNS Anomalies: Neural tube defects, encephalocele, hydrocephalus.
7. Craniofacial defects: Cleft lip/palate, exencephaly.
8. Limb defects: Club foot (15,16), absence of tibia/fibula, hypodactyly.
9. Skeletal anomalies: Scoliosis, lordosis, hemivertebra.
10. Abdominal organ defects: Intestinal malrotation, imperforate anus (15.16), polysplenia, gall bladder agenesis.

### **Case Report**

A young patient came for anomaly scanning. No history of consanguinity. The patient is not diabetic or hypertensive, she was on a regular antenatal check-up.

Following anomaly scan, found to have a large omphalocele with ectopia cordis. A large ventral thoracoabdominal defect was noted. Lower anterior sternal defect and anterior diaphragmatic defect. Cardiac defects included pericardial defect, large VSD and ventricular aneurysm.



**Figure 1: Omphalocele**



**Figure 2:** Left ventricular aneurysm



**Figure 3:** Sternal defect

### **Discussion**

Categorisation of Cantrell consists of all the five defects involving the fetus [1]. Later it was subdivided or classified by Toyoma [2], in three heads, where the first head with all five expressions, the second one with four expressions including ventral defect of the abdomen and lower sternal defects and class three with incomplete expressions with the presence of a sternal defect.

The case reported here consists of a classic representation of all five manifestations of what Cantrell had postulated. Defect in the anterior abdominal wall, defect in the lower sternum, diaphragmatic pericardial defect, defect in the anterior diaphragm and various intracardiac

anomalies, so one of the rarest presentation with all five categories. The time of presentation was also important at 20 weeks. In most cases, the age of presentation varies from 14 to 33 weeks. Nowadays, the implementation of anomaly scan [11] helps us to find the menace as a monstrous fetus. In suspected cases, for better delineation of anomalies prenatal MRI can also be carried out [12,13,14].

It is a herculean target to ascertain a treatment schedule for a fetus born out with defects of Pentalogy of Cantrell. A hectic task is needed to correct the abdominal, sternal and cardiac defects [17] and the probable outcome following such a procedure is also not that much worthwhile. Tahoma [2] stated 20% survival following such complex surgical procedures. Ghidini [18] stated 0% survival of all the 17 cases reported and Homberg in 1996 [19] stated 50% mortality in all the surgically embarked cases.

Pentalogy of Cantrell, a rare and complex disorder needs to be tackled prematurely, if it is carried out further its next outcome is always doom. Curbing the menace as early as possible is the most desirable one. Preventive radiology has a tremendous role in the field of anomaly scan.

A meticulously planned anomaly scan in the first trimester [11] would be able to pin point such major and most of the minor anomalies, what would have reflected at that period of gestation.

Familial recessive inheritance has been reported in certain cases [9]. Cases have also been reported in babies born out of consanguineous marriage [10]. It must be avoided by constant and repetitive self-realisation to all age groups and also in tribes where it is being practised widely.

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