CANTRELL’S PENTALOGY: CASE REPORT

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ABSTRACT: Cantrell’s Pentalogy is a rare syndrome that features five characteristics that are the following: Inferior Sternal Defect, Inferior Diaphragmatic defect, diaphragmatic pericardia defect, and abdominal wall defects and cardiac alterations.

The case was reported on the Obstetrics And Ginecology Hospital Isidro Ayora, which was diagnosed by echo graphic means and other findings were described by MRI and macroscopic visualization of defects observed during labor.

KEY WORDS: Cantrell’s Sd, ectopic cordis, abdominal and thorax wall defects.

OBJECTIVES

- Learn about the radiologic findings of the pathology
- Identify all the disorders on patients with the disease and it’s association with other malformations.

INTRODUCTION:

There is a wide range of congenital defects that are diagnosed on daily basis by using ultrasonography as part of prenatal care in pregnant women and even more on patients with risk factors that favor the presentation of this type of pathology and in which if required, other radiologic tests as the MRI should be performed specially on complicated pathologies.

It’s important to know the contribution of ultrasonography as the first line study in high risk obstetric patients, which is guided for the valuation of anatomical intrauterine defects, being useful for taking the appropriate clinical decision and genetic counseling according to the case, thus ensuring the biological psychological and social welfare of the child and the parents.
In 1958 Cantrell and colleagues described this syndrome as an association of entities found on patients who were suffering mostly of defects on: the anterior wall on the medial epigastric line, sternal inferior section, anterior diaphragm, diaphragmatic pericardium and congenital cardiac alterations including intraauricular communications, interventricular communications, pulmonary stenosis, Fallot’s Tetralogy and atrio-ventricular defects. (4)

The commitment of the anterior diaphragm and pericardium determines the thoracoabdominal heart evisceration (ectopic cordis). Besides the closing defect of the anterior abdominal wall, in the midline, leads to an omphalocele or gastroschisis, resulting in evisceration of abdominal organs (liver, intestine) (1.4). Pathologies that present on mild or very complex manners can lead to fatal outcomes for the product.

CLINICAL CASE:

The reported case is about a 42 years old pregnant patient of Colombian origin, residing in Nanegalito for about 10 years until the present day, she is a merchant without any personal, familiar or gynecological pathological data of importance.

At 8 weeks of gestation an ultrasound is performed and in which it was reported an unchanged and normal pregnancy and also according to the past gynecological examinations. (Figure 1)

Figure 1: Supra public obstetric ultrasound performed at eight weeks of gestation without pathological data.

The patient attends to the ultrasound department for studying her second pregnancy currently 18 weeks of gestation, which has an intergenesic period of 16 years; the obstetrical ultrasound is performed and reveals the presence of a single fetus with active movements, alive and with the following findings:

Head: absence of skull bones allowing deformation of the fetal head, because of this the measurement of biparietal diameter is not considered for estimation of gestational age. It is also visualized the presence of schizencephaly with protrusion of frontal brain contents into the superior anterior zone, cleft lip and palate with a separation of about 5 mm, nasal defect formation detected. (Figure 2)
Figure 2: Supra public obstetric ultrasound performed at eighteen weeks of gestation, fluid content observed at head level with little parenchyma at the base of the skull, plus the absence of bones in the cranial vault. An enlarged head is displayed. Cleft lip and palate with a 5 millimeters separation.

Thorax: Four chambered heart with rhythmic heart rate, defect on anterior chest wall demonstrating the presence of ectopic cordis associated with VSD of approximately 2.2 millimeters of diameter. (Figure 3)  
Figure 3: Ectopic Cordis. Four Chambered Heart with VSD

Abdomen: Closing defect on anterior abdominal wall so it is possible to detect liver and intestine out of the abdominal cavity without coating elements, in relation with gastrochisis. No extrusion of abdominal-pelvic organs. (Figure 4)  
Figure 4: Gastrochisis, Intestinal loops and liver protruding outside of the abdominal cavity in direct contact with the amniotic fluid.

Vertebral Column: The column is properly identified in all segments with no evidence of fusion defects or presence of meningocele. (Figure 5)  
Figure 5: Vertebral Column and placenta without alterations

Extremities: Four limbs were identified with no abnormalities.  

Umbilical Cord: Normal thickness and features.

Placenta: Posterior, homogeneous, of adequate thickness, no alterations can be observed. (Figure 5)

Amniotic Fluid: Normal amount and normal features.
Obstetric MRI is performed which confirms the sonographic findings with greater precision of anatomical structures, besides discarding the possible alterations in the spine (Figure 6) the presence of small brain tissue at the base of the skull and frontal region is detected with significant liquid content occupying the cephalic cavity in which the brain membranes appeared to be in contact with the amnion.(Figure 7,8).

The patient is informed about the findings and receives medical counseling by the gynecologic and obstetrical committee of the hospital resulting in a recommendation for a therapeutic abortion because of the malformation process and lack of probabilities for chances of life of the product. The patient decides not to participate and continues on with pregnancy despite the probabilities. Five weeks later the patient returns for the therapeutic abortion as she and her partner decided to proceed with the intervention which is made by inducting labor successfully obtaining the expulsion of a 23 week product, with severe malformations that reflects on minimal myocardial sporadic contractions that ceased after a few minutes, findings from ultrasonographically and MRI are confirmed (Figure 7). Macroscopic and ultrasonographic correlation data
allowed for the presumptive diagnosis of Cantrell pentalogy's defect in association with midline defects and probably amniotic flanges affecting the skull. A pathologic analysis was not performed due to the mother not consenting for the examination.

Figure 7: Open lip schizencephaly (Tip of arrow) and the presence of cerebral membranes that appear to be in contact with amnios (white arrow)

Figure 7: Obstetric MRI, Axial image of the base of the skull shows normal structures, sagittal image shows increased CSF over the base of the skull with head deformities and contact between amnios and cephalic casing.

Figure 7. 23 week old fetus, Cranium-facial alterations are described, notice the insertion of ovular membranes at head level, probably related with amniotic brides that causes acrania without anencephaly, also it is shown the series of important defects of fusion between the medial facial line and thoracoabdominal area.

DISCUSSION:

This rare syndrome is presented in the population in about 5.5 / 1,000,000. It describes five malformation and were described by Cantrell, Haller, and Ravich in 1958 who identified: Supraumbilical abdominal midline defect, sternum defect at lower third level, lack of anterior segment of the diaphragm, pericardial defects and congenital cardiac malformations.

The frequency of presentation is higher in men in relation to women in a ratio of 2: 1. (6)

Most cases are sporadic but there has been found certain associations with family inheritance, dominant X-linked inheritance, viral infection, and exposure to toxic substances and teratogens such as quinidine, warfarin, thalidomide and even vitamin A deficiency (3), its etiology is still unknown though by knowing these associations it is described as multifactorial defect. (7)

The origin of the defects are believed to take place between days 14 and 18 of gestation due to disorders involving the mesoderm that determine the diaphragmatic, pericardial and intracardiac defects.
there is also a lack of fusion of the lateral folds in the body trunk that reflects with the presence of sternal defects and the defects of the abdominal wall show a relation with omphalocele (4).

It is known that the complete form of the disease carries five features mentioned previously but other forms have been described in the presence of atypical phenotypes. In 1972 Toyama suggested the following classification: (3)

Class 1: Accurate diagnosis if the five defects described by Cantrell are presented.

Class 2: Probable diagnosis with presence of four defects (intracardiac anomalies and abdominal wall defects must be present).

Class 3: Incomplete diagnosis, a combination of variations of the defects (always including sternal defects).

The clinical manifestations depend on the type and severity of the associated malformations, to the point of being so slight that are discovered after birth.

A frequent associated pathology with this Sd is the Ectopic Cordis presented in 80% of the cases and according to the Figueroa study, et al(2) in which it was found that VSD was present in all patients, as in the case of this patient this was detected by ultrasonography.

The third trimester ultrasonography is crucial in most cases of congenital malformations and in this case the malformation could be detected early.

This pathology can be diagnosed by ultrasonography if the following findings are found: epigastric omphalocele, lower sternal defect and cardiopathy, thus the possible diagnostic for Cantrell’s Pentalogy can be made. Both defects of the pericardium and diaphragm are difficult to see by this method.

Thoracic-Abdominal exposure of the viscera make incomplete cases and cases not involved with fatal pathologies like in the presented report candidates for a immediate surgical resolution for preventing sepsis (6). The thoracic defect should be corrected in first place on an ectopic cordis situation, later the abdominal defect should be corrected on a second intervention. However the corrections made on patients of reported cases feature subsequent complications and death in almost every patient.

When other malformations coexist, they can represent an anatomical or pathogenical relation however there are cases in which cranioccephalic and cleft lip palate alterations presence are not quite explained, for some authors the amniotic band constriction phenomenon is the reason for these alterations (3,7) because of the observable relation between ovular membranes and the subjects cranium.

CONCLUSION
Cantrell’s Pentalogy is a complex disease in which a multidisciplinary intervention from the medical team is needed to reach a precise diagnostic and an adequate intervention and prognostic.

Ultrasonography as an initial image method is of great value for the search and characterization of the founded malformations and is very useful tool in expert hands that allows precise diagnostics that correlate well with findings of other more complex studies like MRI, which has great value in obstetric pathology.

The surgical correction success and the prognostic of these patients depend on the type of presentation if it is partial or complete, the latter having unfavorable results specially when correlated with other malformations that in some cases are incompatible with life, leading to choose a radical solution like the one described on this article.

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Bibliography: