PENTALOGY OF CANTRELL: A CASE REPORT

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ABSTRACT
The pentalogy of Cantrell is a rare syndrome characterized by five anomalies that are associated: epigastric defect of the over umbilical abdominal mean line, defect of inferior breastbone third, defect of the anterior segment of the diaphragm, pericardial defects and congenital cardiac malformations.

Describes a case reported in Hospital “Isidro Ayora” in Quito, it was diagnosed with a ultrasonography and the defects was visualized with a magnetic resonance and macroscopic examination in the newborn.

PALABRAS CLAVE
Cantrel syndrome, Ectopia cordis, defects of the de la pared toraco-abdominal.

ABSTRACT
The pentalogy of Cantrell is a rare disease, with five characteristics: epigastric defect over umbilical abdominal mean line, defect of inferior breastbone third, defect of the anterior segment of the diaphragm, pericardial defects and congenital cardiac malformations.

This article describe a case from Hospital Isidro Ayora, the diagnosis was made by fetal ultrasonography in the second trimester of pregnancy and confirm with magnetic resonance and macroscopic characteristics in delivery moment.

OBJETIVES
Know the radiology characteristics of the disease
Identify all the disorders in the sick patients and its association with other malformations
INTRODUCTION

A wide range of birth defects that are diagnosed daily with the use of ultrasound as part of antenatal care in pregnant women and more in patients with risk factors favoring the presentation of this pathology in which if required other imaging techniques are employed as in the case of MRI which is widely used in the case of more complex pathologies.

It is important to know the contribution made by ultrasonographic study as first-line study in obstetric high-risk patients, which is aimed at assessing in uterus anatomical defects for proper clinical and genetic counseling decision according to the case ensuring the welfare bio-psychosocial child and their parents.

In 1958, Cantrell et al described this syndrome as an association of entities found in patients who are suffering from it, which stand are: defects of the anterior and the midline epigastric, the lower portion of the sternum, the above diaphragm, pericardium diaphragmatic and congenital cardiac defects of these the major are atrial septal defects, ventricular, pulmonary stenosis, tetralogy of Fallot and atrioventricular defects. (4) The commitment of the anterior diaphragm and pericardium determines thoraco-abdominal evisceration heart (ectopia cordis). Moreover, the defect closure of the anterior abdominal wall, in the midline, resulting in a omphalocele or gastroschisis, causing evisceration of abdominal organs (liver, intestine, etc.). (1, 4), diseases that may be mild or very complex, and carry fatal outcomes for the product.

MATERIALS Y METHODS

Imaging studies and case analysis were conducted in the Obstetric and Gynecology Hospital “Isidro Ayora” and Hospital “Eugenio Espejo” from Quito.

Ultrasound equipment brand GE Voluson 730 ProV with 3.5 MHz convex transducer. Using harmonics, Color Doppler, Power Doppler, spectral Doppler, image optimization with use of low frequency, high densities, adequate focus number was used according to the needs and use of 3D and 4D.

Magnetic resonance imaging (MRI) Philips 1.5 Tesla (T). Axial, coronal and sagittal images in relation to fetal were performed. Ultrafast sequences were used in T1 THRIVE and T2w_TSE_, TR user defined 80ms, Flip angle (deg) 90ms, TR 1000ms. Body coil and an additional placed on the pelvis.

CLINIC CASE

Pregnant patient of 42 years old of Colombian origin, Nanegalito resident for 10 years, occupation merchant, without personal, family or gynecological pathological data of importance.

At 8 weeks gestation, obstetric ultrasound reported without pathology and normal pregnancy according to checks gynecological performed. (Figure 1)

The patient presented to the ultrasound department with 18 weeks of her second pregnancy which has a 16-year period between births, we performed obstetric ultrasound where you can observe the presence of a single fetus with active fetal movements and the following findings:

Head: absence of skull bones allowing deformation of the fetal head, so measuring DBP and CC not considered for estimation of gestational age, also we can see schizencephaly with protrusion of the frontal brain exceeded to the previous region, palate
and cleft lip with a separation of about 5 mm, the nasal defect formation. (Figure 2.9)

**Chest:** 4-chamber heart, heart rate rhythm with defect of the anterior wall of the chest so the presence of ectopia cordis associated with ventricular septal defect of about 2.2 mm in diameter stands. (Figure 3)

**Abdomen:** closure defect of anterior abdominal wall so it fails to identify intestine and liver out of the abdominal cavity, without coating them, regarding gastrochisis, without observing extrusion abdomino-pelvic organs other. (Figure 4)

**Spine:** it was properly identified in all segments without evidence of melting defects or meningocele. (Figure 5)

**Extremities:** 4 extremities with appropriate aspect were identified.

**Umbilical cord:** thick and normal characteristics.

**Placenta:** posterior, homogeneous, of adequate thickness, without alterations. (Figure 5)

**Amniotic fluid:** characteristics and normal amount.

Obstetric MRI confirmed the ultrasound findings with greater precision of anatomical structures, also ruling out possible alterations at the level of the spine (Figure 6), also can observe the presence of low brain tissue at the skull base and the frontal region with significant liquid content occupying the cephalic cavity, showed brain membranes them impressed continued or be in contact with the amnion (Figure. 7, 8)
Figure 6. Obstetric IRM. Ectopia cordis and gastroschisis (arrows), normal spine.

Figure 7. Schizencephaly open lip (arrowhead) and the presence of brain membranes impressing to take contact with the amnion (white arrow).

Figure 8. Obstetric MRI. Axial image of the skull base showing normal structures, sagittal section shows the increase of CSF over base of skull with head deformation and contact cephalic covered with the amnion.

We informed the patient of the findings, after which she received medical counseling by committee obstetric and gynecology of the hospital for a therapeutic abortion due to the fetus malformations and with absent prognostic for the baby, however the patient decides not enter and continue the pregnancy despite the null forecast, five weeks later the patient was readmitted for the decision she and her partner for the therapeutic abortion which was performed with induction of labor successfully, obtaining product of 23 weeks, with multiple and severe malformations, minimal and sporadic myocardial contractions, cease it within a few minutes.

The ultrasonography and MRI findings were confirmed. (Figure 6, 7, 8) With a good correlation between ultrasound and macroscopic findings for the Cantrell pentalogy in association with the facial midline cranial defect and possibly amniotic clamps condition that affected at the skull.

Could not perform pathological analysis because the mother did not authorize this procedure.

Figure 9. Fetus of 23 weeks. Facial abnormalities described the skull is displayed, note the inclusion of the egg membranes at the fetal head, probably related to amniotic flanges causing acrânea without absence of brain and important fusion defects in midline facial and thoracoabdominal.

DISCUSSION:

This rare syndrome present at about 5.5 / 1'000000 population and related five malformations described by Cantrell, Haller and Ravich in 1958 who identified: default epigastric midline abdominal supraumbilical, defect of the lower sternum, deficiency the anterior segment of diaphragm, pericardial defects and congenital cardiac malformations.

The occurrence is higher in men compared to women (2:1). (6) Most cases are sporadic, although found association with familial inheritance dominant X-linked inheritance, viral infection, toxic and teratogens exposure such as quinidine, warfarin, thalidomide and even to vitamin A deficiency (3), its etiology is unknown even, though these associations could be for multifactorial origin. (7)

The beginning of the defects are believed to take place between 14 and 18 weeks of gestation by disorders of the mesoderm (6) it determine the diaphragmatic, pericardial and intracardiac defects, there is also lack of fusion of the lateral folds in body stalk it determine sternal and abdominal wall defects for example omphalocele. (4) Is known that the presence of complete forms has five
characteristics but also have been described other incomplete forms with atypical phenotype of presentation (7) in Toyama in 1972 suggested the following classification: (3)

Class 1: accurate diagnosis; five defects described by Cantrell appear;
Class 2: probable diagnosis, with four defects (including intracardiac and abdominal wall anomalies)
Class 3: incomplete diagnosis, varying combinations of defects (always including sternal anomalies).

Clinical manifestations depend on the type and severity of associated malformations may be so mild that it is not discovered after birth.

One of the diseases often associated with this syndrome is ectopia cordis in 80% of cases, in Figueroa’s study, et al. (2) founded in all patients IVC such as in this patient who already sonographer could determine the presence of this disorder.

The first trimester ultrasound is essential in most cases of congenital anomalies and in this particular case we saw early yet.

You can sonographically diagnosed Cantrell if you find epigastric omphalocele, a defect in the lower portion of the sternum and heart defect. But the diaphragmatic and pericardial defect is very difficult to see, inferring their presence given the findings (4).

Exposure of thoraco-abdominal viscera in incomplete cases not associated with fatal diseases such as in this case the immediate surgical resolution to be possible to prevent sepsis (6); the priority is the thoracic defect in the case of ectopia cordis, in a second time to correct the abdominal defect, but in the corrections made in cases reported in the literature have further complications and death of almost all patients. When other malformations coexist, they may have a pathogenic or obvious anatomical localization but can find other types of malformations with unclear correlation as in this case: craniocerebral and cleft abnormalities that some authors attribute it to amniotic flanges (3, 7) which may be the case presented especially by observing the relationship of the ovular membranes with baby skull.

CONCLUSION:

The Pentalogy of Cantrell is a complex disease in which the multidisciplinary health team intervention is required to reach the accurate diagnosis and guide treatment and eventual prognosis.

Ultrasonography as an initial imaging method is very important for the search and characterization of defects found and is a very useful tool that in expertly hands allows us precise diagnoses and correlate well with findings from studies of greater complexity such as magnetic resonance with immense value in obstetric pathology.

Successful surgical correction and the prognosis for these patients is particularly focused on the type of partial or complete submission latter being especially with unfavorable results more if it is associated with other malformations that are life incompatible sometimes and requiring radical decisions like in this article.

CONFLICT OF INTEREST

none

BIBLIOGRAPHY: