

CASE REPORT

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Prenatal diagnosis of Pentalogy of Cantrell: a case report

Diagnóstico prenatal de pentalogía de Cantrell: reporte de caso

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ABSTRACT

The Pentalogy of Cantrell is the conjunction of five congenital defects (defects of the heart, pericardium, diaphragm, sternum and anterior abdominal wall) which represents a unique challenge for the obstetrician gynecologist. Although its incidence is small, it is essential to identify it early in the first trimester. A case of Pentalogy of Cantrell is reported, diagnosed in the first prenatal control in the second trimester of pregnancy.

Key words: Ectopia cordis, Pentalogy of Cantrell, Congenital abnormalities.

RESUMEN

La pentalogía de Cantrell es la conjunción de cinco defectos congénitos (defectos del corazón, pericardio, diafragma, esternón y la pared abdominal anterior), que representa un desafío único para los gineco-obstetras. Aunque la incidencia es pequeña, es fundamental identificarlo en el primer trimestre. En este reporte se comunica un caso de pentalogía de Cantrell diagnosticado en su primer control prenatal en el segundo trimestre de gestación.

Palabras clave. Ectopia cordis, Pentalogía de Cantrell, Anomalías congénitas.

INTRODUCTION

Cantrell's syndrome (CS) or Pentalogy of Cantrell was described for the first time in 1958. It involves multiple congenital anomalies, such as a medial anterior abdominal wall defect, distal sternal cleft, anterior diaphragm defect, apical pericardium defect with pericardial-peritoneal communication and intracardiac abnormalities⁽¹⁾.

The prevalence of this syndrome varies between 1:65 000-200 000 live births; in developed countries, 5,5 per million live newborns are reported⁽²⁾. Most of the cases are sporadic, predominantly in males, although an association has been found with dominant inheritance linked to the X chromosome, aneuploidies such as trisomy 18 and 21, viral infection, exposure to toxic substances such as aminopropionitrile and teratogens such as quinidine, warfarin, thalidomide, and even vitamin A deficiency⁽³⁾. It is postulated that the transverse septum defect is responsible for this malformation, which occurs between days 14 and 18 of embryonic development. The defect occurs in the process of migration or fusion of the anterior midline of the body^(1,4,5). The wide range of this syndrome could reflect the moment of the defect's development.

One of the few cases of the pentalogy of Cantrell diagnosed prenatally in our country is reported, with a review of the main literature on prenatal diagnosis.

CASE REPORT

The 39-year-old patient, G3P1011, began her prenatal control at 17 weeks of gestational age. Ultrasound was performed, observing a single active gestation of 17 weeks and 6 days with a fetal weight of 172 g. The fetus showed a supraumbilical abdominal wall defect, which compromised the lower portion of the sternum and where the liver, intestines and heart



protruded (the heart and the lower portion of both lungs were observed). Umbilical cord insertion was normal (Figures 1 to 4). The Doppler study evidenced the alteration of the vasculature and specified the extent of the defect (Figures 3 and 4). Likewise, a bilateral lip-palatal defect was observed (Figure 1A), abnormal position in perma-

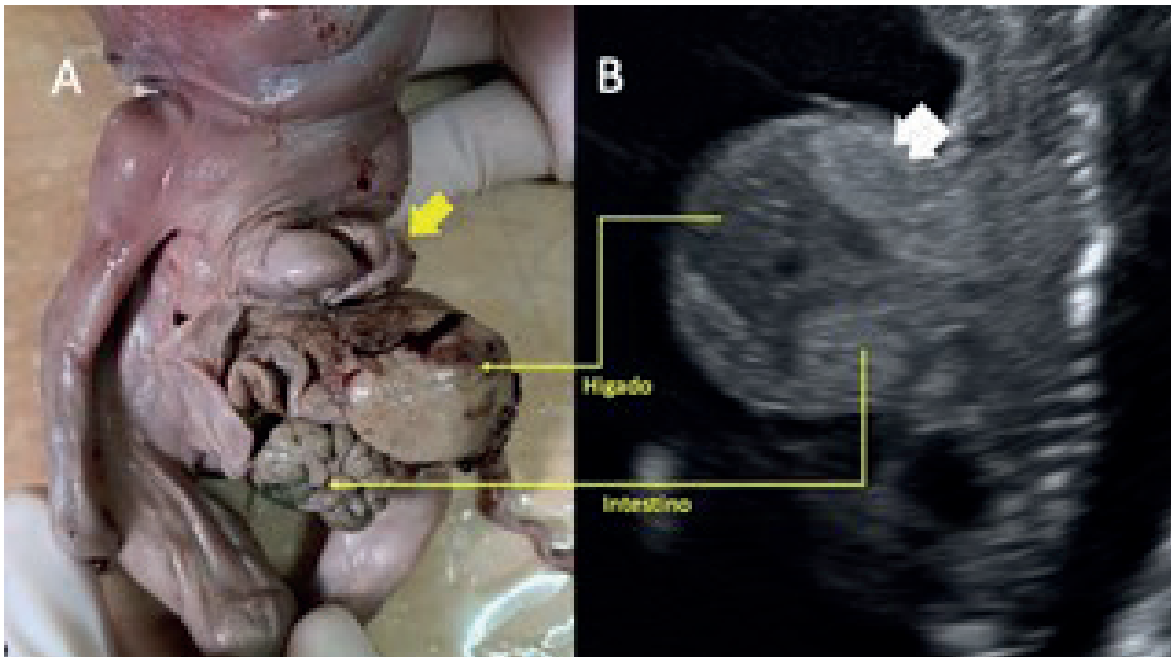
nent flexion of the hands, and preaxial polydactyly (Figure 4B). The diagnosis of Cantrell syndrome (CS) was made, to rule out probable aneuploidy.

A complete evaluation of the heart was not achieved due to fetal position. Therefore, an examination was planned in 4 weeks. The patient

FIGURE 1. IMAGE SHOWING THE CORRESPONDENCE BETWEEN THE 3D IMAGE (RIGHT) AND THE ANATOMY OF THE PRODUCT. NOTE THE EXTENSIVE ANTERIOR WALL DEFECT CONTAINING THE LIVER, INTESTINES AND HEART. YELLOW ARROW: BILATERAL LABIOPALATINE DEFECT.



FIGURE 2. A: PRODUCT IMAGE WITH ECTOPIA CORDIS. YELLOW ARROW: RIGHT VENTRICLE THROUGH INFERIOR STERNAL DEFECT. B: SAGITTAL SECTION OF THE ULTRASOUND, WHERE THE ABDOMINAL MEMBRANE-CONTAINED DEFECT IS OBSERVED. WHITE ARROW: FETAL LUNG WITHIN THE DEFECT. THE YELLOW ARROWS SHOW THE CORRESPONDENCE BETWEEN THE STRUCTURES VISIBLE ON ULTRASOUND AND THE FINAL PATHOLOGY.





continued her prenatal control, but the requested genetic tests were not performed. Three weeks later, she was admitted due to ruptured membranes, with 20 weeks 6 days of gestation, producing a stillbirth on the same day. Prenatal findings were confirmed. At the time of delivery,

the membrane covering the defect was ruptured, showing syndromic facies with low ear implantation, hypertelorism and a bilateral lip-palatal defect (Figures 1, 2 and 4), which increased the suspicion of aneuploidy. Family members did not authorize the autopsy.

FIGURE 3. A: CORONAL SECTION OF THE DEFECT. THE ARROW INDICATES THE VISION OF FOUR CHAMBERS OF THE HEART. ADJACENT HYPERECHOIC AREAS CORRESPOND TO EXTRA-THORACIC FETAL LUNGS. INSET SHOWS THE ECTOPIA CORDIS OF THE PRODUCT. **B:** SAGITTAL SECTION WITH USE OF POWER DOPPLER SHOWS THE SILHOUETTE OF THE VASCULARITY, IN WHICH THE AORTIC ARCH UP TO THE EXTRA-THORACIC HEART IS HIGHLIGHTED (DOTTED LINE).

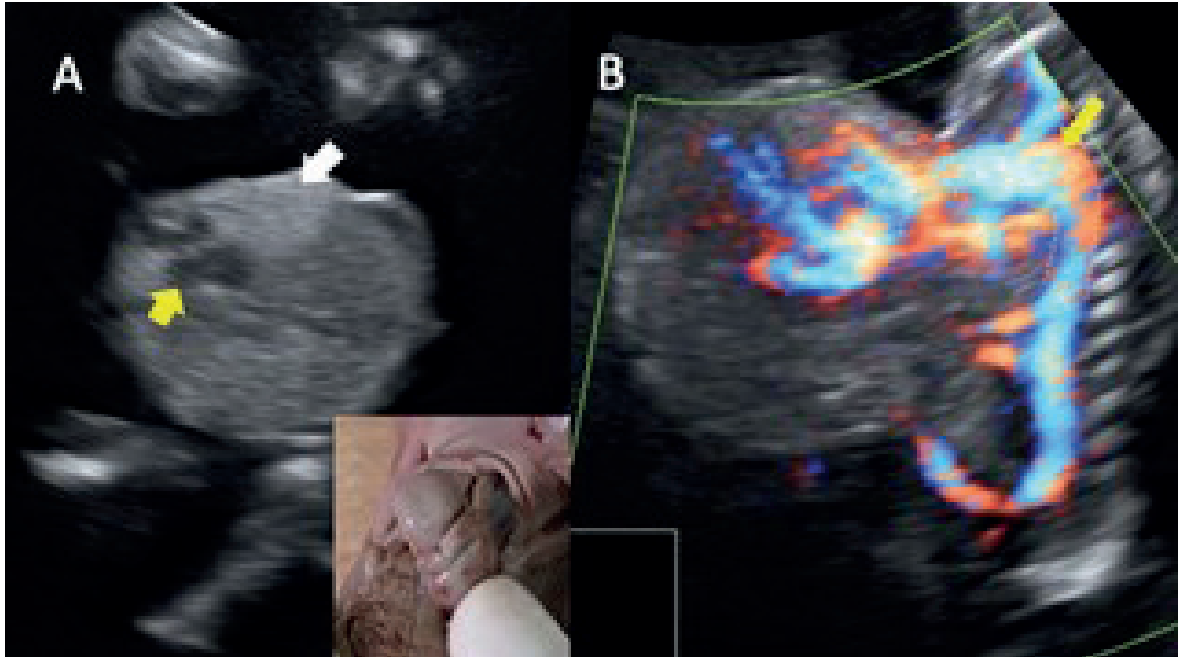
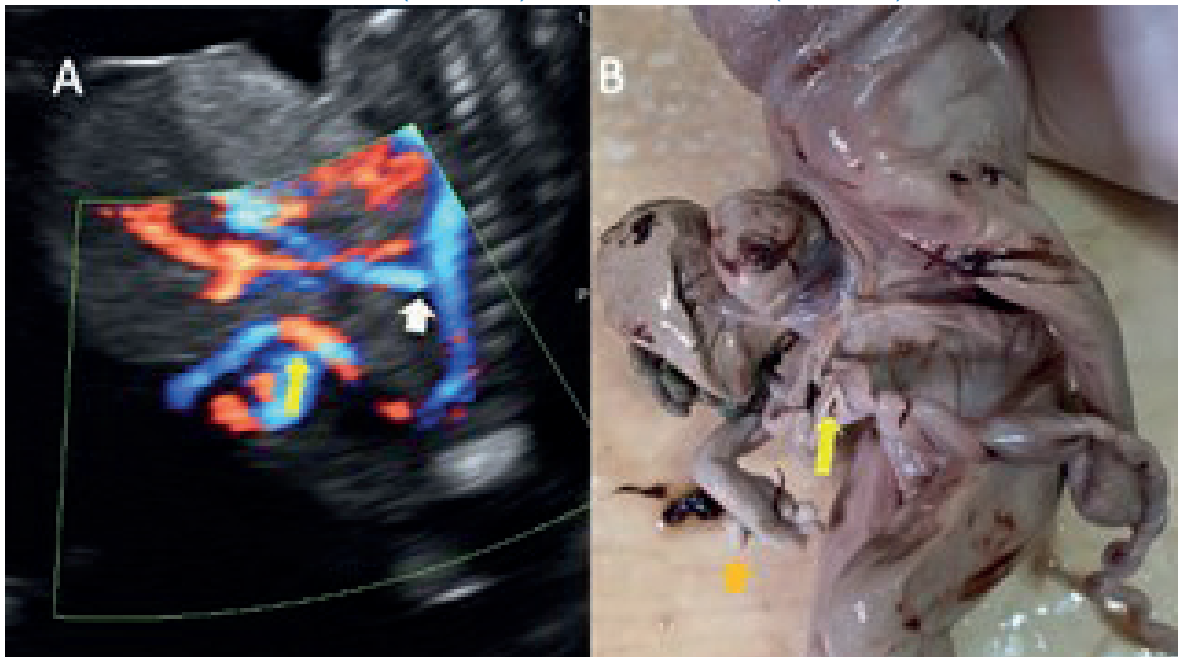


FIGURE 4. A: SAGITTAL SECTION WITH POWER DOPPLER, SHOWING THE AORTIC VESSELS (WHITE ARROW: MESENTERIC ARTERY) SUPPLYING THE ORGANS OF THE DEFECT. YELLOW ARROW: ENTRY OF THE UMBILICAL CORD IN THE LOWER PART OF THE DEFECT. **B:** IMAGE OF THE PRODUCT, WHERE THE ORIGIN OF THE UMBILICAL CORD IS OBSERVED IN THE LOWER PART OF THE DEFECT (YELLOW ARROW). POLYDACTYLY IS ALSO OBSERVED (ORANGE ARROW).





DISCUSSION

CS is composed of a supraumbilical abdominal wall midline defect, distal sternal cleft, apical pericardial defect with pericardial-peritoneal communication (the last two explain ectopic cordis), anterior diaphragm defect, and intracardiac abnormalities, such as defects of the ventricular septum, the atrio-ventricular septum, tetralogy of Fallot, pulmonary stenosis, among others^(6,7).

In the present case, all the characteristics were demonstrated, except for heart disease (Figures 1 to 4), since the optimal evaluation of the heart could not be performed. Not all cases express the five characteristics, so Toyama (1972) suggested classifying CS by the clinical findings of the pentalogy, which influence the prognosis and treatment of the disease.

Type 1: Exact diagnosis, the five defects described by Cantrell are appreciated.

Type 2: Probable diagnosis, with four defects stated by Cantrell, including intracardiac and abdominal wall abnormalities.

Type 3: Incomplete diagnosis: variable combination of defects, always including sternal anomalies⁽³⁾.

Ectopia cordis, which is defined as the extra-thoracic location of the heart, is one of the cardiac malformations associated with CS (Figure 3A), but it is not necessary for diagnosis nor is it always present.

Prenatal diagnosis of CS should ideally be made during the first trimester, and confirm the extent of involvement in the second, during routine examinations. 3D volumetric ultrasonography and power Doppler allow us to more accurately assess the extent of the disease and associated malformations (Figures 3 and 4). It should always be expanded with echocardiography, as heart disease is a determinant of prognosis. Fetal MRI can be performed in cases not associated with genetic defects, for postnatal preoperative planning^(2,8-12). During routine ultrasound, the suspicion of ectopia cordis and omphalocele are two ultrasound findings that should lead us to rule out CS⁽¹³⁾. When the defect is small, the presence of pericardial effusion associated with omphalocele in the first trimester requires confirmation in the second trimester^(2,7).

The variety of presentation of the phenotype is wide, so much so that Sepúlveda⁽¹⁴⁾ published a series of 7 cases with a diagnosis of ectopia cordis in the first trimester, in a period of 15 years in three medical centers; 4 were classified as CS, all characterized by the abdominal wall defect, which contained liver and heart. On the other hand, Zidere⁽⁷⁾ reports 3 cases with early prenatal diagnosis that, by the second trimester, the ectopia cordis had resolved, but a pericardial effusion persisted, revealing the communication between the pericardium and peritoneum. All cases were associated with severe heart disease.

The differential diagnosis includes isolated thoracic ectopia cordis (ectopia cordis can be thoracic, thoraco-abdominal, abdominal and cervical), amniotic band syndrome and anomaly of the body stem⁽¹³⁾. Genetic studies are necessary due to the high association with aneuploidies (mainly trisomy 18), which is the main suspicion in the present case. In addition, chromosome analysis also provides better information and advice to families.

CS with ectopia cordis has been associated with a high perinatal mortality rate, requiring multiple surgical procedures per stages⁽¹⁵⁻¹⁷⁾. Pośpiech-Gąsio, on the occasion of publication of a case, brings together published cases with prenatal diagnosis of CS, finding 57, the majority evaluated in the first trimester. Of these, only 10% survived, 10% had perinatal death and 10% were stillbirths⁽¹⁸⁾. We could say that the prognosis, in general, is based on the presence of ectopia cordis, the complexity of the intracardiac malformation and the malformations of other associated systems⁽¹⁹⁾.

Finally, the diagnosis of this pathology should be made in the first trimester. The combination of color Doppler and 3D volumetry further improves the detection of the defect, which should lead to timely advice to parents.

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