IJHS
Interamerican Journal of Heath Sciences 5 (2025) - ISSN 2953-3724
DOI: 10.59471/ijhsc2025238

CASE REPORT

Pentalogy of Cantrell: A challenge in prenatal diagnosis. A case report in Bolivia

Pentalogía de Cantrell: Un desafío en el diagnóstico prenatal. A Propósito de un caso en Bolivia

Benito Aguirre-Cruz[1], Jhossmar Cristians Auza-Santivañez[2], Elizeth Mendoza-Contreras[1], Ritdber Alfredo Quispe Sarmiento[3], Pestañas-León Alexin Raúl[1], Delly Espejo-Alanoca[4], Blas Apaza-Huanca[2], Dilma Villca-Villca[5]

[1]Hospital San Antonio de los Sauces, Chuquisaca, Bolivia.
[2]Ministerio de Salud y Deportes. Instituto Académico Científico Quispe-Cornejo. La Paz, Bolivia.
[3]Caja de Salud de Caminos y RA, Cochabamba, Bolivia.
[4]Ministerio de Salud y Deportes. La Paz, Bolivia.
[5]Hospital Obrero N° 4. Caja Nacional de Salud - Regional Oruro. Oruro, Bolivia.

Received: 15-11-2023 **Revised:** 19-02-2024 **Accepted:** 23-05-2024 **Published:** 24-05-2024
How to Cite: Aguirre-Cruz B, Auza-Santivañez JC, Mendoza-Contreras E, Quispe Sarmiento RA, Pestañas-León AR, Espejo-Alanoca D, et al. Pentalogy of Cantrell: A challenge in prenatal diagnosis. A case report in Bolivia. Interamerican Journal of Health Sciences. 2025; 5:238. https://doi.org/10.59471/ijhsc2025238

Corresponing author: Jhossmar Cristians Auza-Santivañez

ABSTRACT

Introduction: pentalogy of Cantrell is a rare congenital anomaly with an incidence of less than one in 100 000 pregnancies. It is characterized by a defect in the abdominal wall at the level of the supraumbilical midline, the lower segment of the sternum, the anterior diaphragm, the diaphragmatic pericardium, and cardiac abnormalities such as ectopia cordis. Few cases of this entity have been reported in the literature in Bolivia. It is a constellation of congenital defects that pose a unique challenge, hence the importance of diagnosis in the prenatal stage.
Clinical Case and Discussion: an 18-year-old pregnant woman came to our hospital, was consulted by Gynecology and Obstetrics, and an ultrasound was performed which revealed a defect in the anterior abdominal wall with viscera outside the fetal cavity (liver, heart, stomach and intestinal loops, 24,3 weeks of pregnancy and multiple fetal malformations. A discussion of the case was held with specialists in Gynecology and Obstetrics, Pediatrics, Genetics, Family Physician, Imaging and Psychology. Taking into account the poor prognosis and the complexity of the anomalies, the patient was told that this could represent a threat and risk for her. At the request of the parents and with prior informed consent, the patient was admitted to the gynecology service, prepared and admitted to the operating room for cesarean section. A female product was obtained, with multiple malformations, a large defect in the anterior thoracoabdominal wall from the navel to the upper third of the body of the sternum, omphalocele. Part of the heart outside the thoracic cavity, presence of a uterine cavity, and a large thoracic cavity. of defects in the diaphragm, liver and intestinal loops outside the abdominal cavity.
Conclusions: the case highlights the importance of early prenatal diagnosis, even in settings with limited resources. Despite the lack of access to more complex studies, the appropriate use of ultrasound, combined with the expertise of the operator, allowed this complex congenital malformation to be identified at an early stage. The timely identification of these conditions not only facilitates multidisciplinary counseling for the pregnant woman and her family, but also opens the possibility of making informed decisions, including the option of terminating the pregnancy before the period of viability, if the parents so consider. In severe cases, such as ours, where the prognosis is generally unfavorable, an early diagnosis can better prepare families emotionally and psychologically, as well as allowing them to consider all possible options with due ethical and medical guidance.

KEYWORDS

Pregnancy; Pentalogy of Cantrell; Ectopia Cordis; Omphalocele; Ultrasonography; Prenatal Diagnosis.

RESUMEN

Introducción: la Pentalogía de Cantrell es una anomalía congénita, rara, con una incidencia menor a uno por cada 100 000 embarazos, se caracteriza por defecto de la pared abdominal a nivel de la línea media supraumbilical, segmento inferior del esternón, diafragma anterior, del pericardio diafragmático y alteraciones cardiacas como la ectopia cordis. En Bolivia se han reportado pocos casos de esta entidad en la literatura, es una constelación de defectos congénitos que plantean un desafío único, ahí la importancia en el diagnóstico en la etapa prenatal.
Caso Clínico y Discusión: gestante de 18 años, acude a nuestro hospital, se realiza interconsulta por Ginecoobstetricia, se realiza ecografía la cual reveló defecto de pared abdominal anterior con vísceras fuera de cavidad fetal (hígado, corazón, estómago y asas intestinales, embarazo de 24,3 semanas y múltiples malformaciones del feto. Se realiza una discusión del caso los especialistas de Ginecoobstetricia, pediatría, genética, médico de familia, imagenología y psicología. Teniendo en cuenta el mal pronóstico y la complejidad de las anomalías se explica a la paciente que esto puede representar una amenaza y riesgo para la misma. A solicitud de los padres y previo consentimiento informado, se ingresa a la paciente al servicio de ginecología se prepara e ingresa a quirófano para cesárea. Se obtiene producto sexo femenino, con múltiples malformaciones, un gran defecto de la pared toracoabdominal anterior desde el ombligo hasta un tercio superior del cuerpo del esternón, onfalocele. Una parte del corazón fuera de la cavidad torácica, presencia de defectos en el diafragma, hígado y asas intestinales fuera de la cavidad abdominal.
Conclusiones: el caso, pone en evidencia la importancia del diagnóstico prenatal temprano, incluso en entornos con recursos limitados. A pesar de la falta de acceso a estudios de mayor complejidad, el uso adecuado de la ecografía, combinado con la experticia del operador, permitió identificar esta malformación congénita compleja en una etapa temprana. La identificación oportuna de estas condiciones no solo facilita el asesoramiento multidisciplinario a la gestante y su familia, sino que también abre la posibilidad de tomar decisiones informadas, incluyendo la opción de interrupción del embarazo antes del período de viabilidad, si así lo consideran los padres. En casos graves, como el nuestro, donde el pronóstico es generalmente desfavorable, un diagnóstico temprano puede preparar mejor a las familias emocional y psicológicamente, además de permitirles considerar todas las opciones posibles con la debida orientación ética y médica.

PALABRAS CLAVE

Embarazo; Pentalogía de Cantrell; Ectopia Cordis; Onfalocele; Ultrasonografía; Diagnóstico Prenatal.

INTRODUCTION

Cantrell's Pentalogy is a rare congenital anomaly with an incidence of less than one in every 100 000 pregnancies. It is characterized by the following alterations: a defect in the abdominal wall at the supraumbilical midline, the lower segment of the sternum, the anterior diaphragm, the diaphragmatic pericardium, and cardiac abnormalities such as ectopia cordis.[1,2] Cantrell's pentalogy was first described in 1958 when Cantrell et al. published a series of five cases of this anomaly.[3] In Bolivia, few cases of this entity have been reported in the literature. It is a constellation of congenital disabilities that pose a unique challenge, hence the importance of prenatal diagnosis.[4,5] The heart, pericardium, diaphragm, sternum, and anterior abdominal wall defects are pathognomonic. Early diagnosis, supportive care, and planning with a multidisciplinary team are key components in managing patients with Cantrell's pentalogy.[5] This congenital disorder requires multidisciplinary care; a better understanding of this condition will improve early diagnosis. We report a case in a rural population where prenatal diagnosis was exceptional due to limitations from the imaging point of view. However, the clinical presentation was a pillar for said diagnosis. The challenge and complexity associated with Cantrell's pentalogy are evident.

CLINICAL CASE

An 18-year-old pregnant woman from a rural area came to our hospital for evaluation. She had undergone a first-level prenatal check-up in her place of origin the previous month. On arrival, she reported that she had undergone neither laboratory tests nor an ultrasound scan. A consultation was requested with the obstetrics and gynecology department for evaluation. The patient did not report any relevant personal or family history, denies toxic habits,

IJHS
Interamerican Journal of Heath Sciences 5 (2025) - ISSN 2953-3724
DOI: 10.59471/ijhsc2025238

and did not report exposure to any harmful substances during pregnancy. On physical examination, the conscious, alert pregnant woman weighed 54,8 kg, was 154 cm tall, had blood pressure (BP) of 95/64 mm Hg, a heart rate (HR) of 59 bpm, uterine height (UH) 26 cm, Leopold maneuvers longitudinal situation, right dorsal position, indifferent presentation, fetal movements present, fetal heart rate (FHR) 143 bpm, gestational age 26 weeks, by date of last menstrual period (LMP).

Laboratory tests and an obstetric ultrasound were requested. The laboratory results were within normal parameters; the ultrasound report revealed a single product, female sex, particulate amniotic fluid, and a defect in the anterior abdominal wall with viscera outside the fetal cavity (liver, heart, stomach, and intestinal loops) (figure 1A-1B). The ultrasound report shows a pregnancy of 24,3 weeks and multiple fetal malformations, and from this conglomeration of findings, a possible diagnosis of "Cantrell's Pentalogy" is reached.

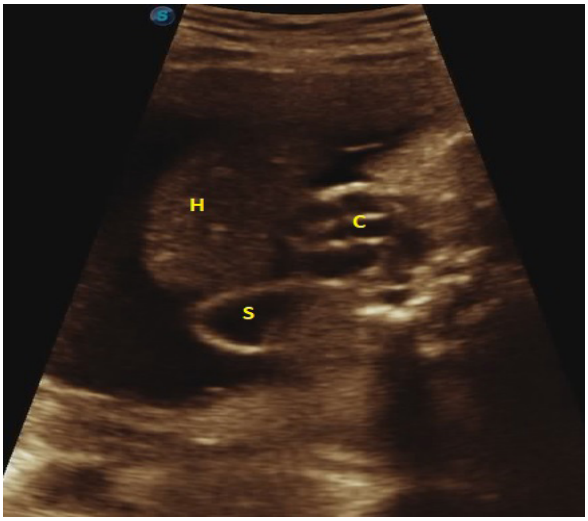

Figure 1A. Cross-section at the level of the lower thorax of the foetus, liver H can be seen outside the abdominal cavity, heart and stomach C and S partially outside the cavity

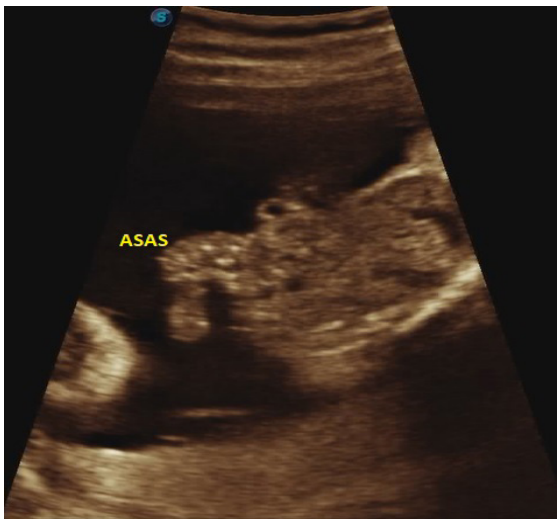

Figure 1B. A cross-section with a larger volume than the previous one at the level of the navel shows a defect in the abdominal wall with protrusion of hyperechoic intestinal loops outside the cavity

Given these ultrasound findings, the case was discussed with specialists in obstetrics and gynecology, pediatrics, genetics, family medicine, imaging, and psychology. Subsequently, the pregnant woman and her family were informed about the risks, complications, and prognosis of the fetus. In addition, the genetics department was consulted, which determined the presence of a syndrome of amniotic band sequences and malformations incompatible with life. Taking into account the poor prognosis and the complexity of the anomalies, the patient is informed of the potential risks to her health during the continuation of the pregnancy; the team of specialists in the medical board and in agreement with genetics, recommend a legal termination of pregnancy (LTP), due to the condition of incompatibility with the life of the fetus. At the parents' request and with their prior informed consent, the hospital team agreed to perform the surgical intervention. The patient was admitted to the gynecology department, prepared, and admitted to the operating theatre for a cesarean section with regional anesthesia. A

female infant was born, weighing 715 grams, with multiple malformations.
Part of his heart was outside the thoracic cavity, and there were also defects in the diaphragm, edematous liver, and intestinal loops outside the abdominal cavity. (figure 2A-2B). The rest of the physical examination described a dysmorphic face, and the spine presented severe scoliosis. The diagnosis of "Cantrell's pentalogy" was reached based on all the findings.

Figure 2A. The image shows an edematous liver, stomach and intestinal loops outside the cavity, and the heart partially outside the cavity

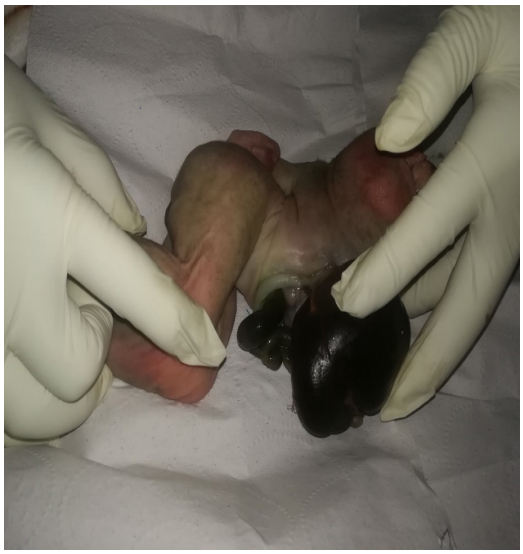

Figure 2B. The image shows the liver and intestinal loops outside the cavity, the heart outside, covered by the liver

The family was informed of the findings. The baby died a few minutes after being born. The parents did not give their consent for an autopsy, nor was a biopsy taken for further study.
On physical examination, the newborn presented a significant defect in the anterior thoracoabdominal wall, which extended from the navel to the upper third of the sternum, a distorted cord in the navel region due to an omphalocele.

DISCUSSION

Cantrell's pentalogy is a rare complex disorder of the ventral body wall, which refers to a complex combination of sternum defect, ectopia cordis, cardiac anomalies, defects of the anterior diaphragm and diaphragmatic pericardium and supraumbilical defect of the abdominal wall.[6,7] Other associated anomalies include midline anomalies such as facial cleft and encephalocele. Given the association with trisomies 18 and 13, genetic counseling is recommended. Most of these cases are sporadic, and the risk of recurrence is low. The prognosis for this disorder is poor.[8] Based on the 60 cases reviewed by Tayoma, the syndrome is classified as either complete or incomplete,

depending on the number of malformations present.[9] Definite diagnosis class 1 and all five defects are present. A probable diagnosis of class 2 is if four defects are present: intracardiac and ventral abdominal wall defects. Class 3: incomplete expression, with various combinations of defects present and includes a sternal anomaly.
In our clinical case, we classified it as class 2 of the syndrome due to the presence of a large defect in the anterior thoracoabdominal wall, a distorted cord in the navel region due to an omphalocele, the heart outside the thoracic cavity, and the presence of defects in the diaphragm, liver and intestinal loops outside the abdominal cavity. Our case had an unfavorable prognosis, in line with the literature, which suggests that mortality is higher in newborns with complete form and additional associated cardiac anomalies.[10,11] Prenatal diagnosis is essential, a skill in performing obstetric ultrasound and diagnosis even at 10 weeks gestation using traditional two-dimensional (2D) ultrasound imaging, at which stage omphalocele and ectopia cordis are common findings.[12,13,14] In Bolivia, not all health centers have high-resolution ultrasound equipment with new technologies such as 3D or 4D and specialists in obstetric sonography and ultrasound evaluation, which limits access to early diagnosis in rural areas. In our case, the patient did not receive an ultrasound during the first trimester of pregnancy, which delayed diagnosis until the 24th week of gestation. This delay highlights the importance of improving access to imaging technologies in regions with limited resources. The management and prognosis of the case depend on the size of the abdominal wall defect and associated anomalies. After prenatal diagnosis, termination of pregnancy can be offered in severe cases when amniocentesis shows an abnormal karyotype.[15,16]

CONCLUSIONS

The case we present highlights the importance of early prenatal diagnosis, even in resource-limited settings. Despite the lack of access to more complex studies, the appropriate use of ultrasound and the operator's expertise made it possible to identify this complex congenital malformation at an early stage. This finding highlights the fundamental role played by accessible technology and the continuous training of healthcare professionals in the detection of fetal anomalies. The timely identification of conditions such as the Pentalogy of Cantrell not only facilitates multidisciplinary counseling for the pregnant woman and her family but also opens up the possibility of making informed decisions, including the option of terminating the pregnancy before the viability period if the parents so choose. Raising awareness of this rare condition among obstetricians, imaging specialists, and sonographers is essential to improve the capacity for early diagnosis. In severe cases, such as ours, where the prognosis is generally poor, early diagnosis can better prepare families emotionally and psychologically, as well as allow them to consider all possible options with the appropriate ethical and medical guidance.

REFERENCES

1. Cuesta-Guardiola, Tatiana, et al. "Diagnóstico prenatal de pentalogía de Cantrell." Progresos de Obstetricia y Ginecología [Internet]. 2015 [citado 2024 Oct. 26]; Disponible en: https://www.sciencedirect.com/science/article/abs/pii/S0304501315001958

2. Acosta JAR, Gutierrez EAO. 190 días de pentalogía de cantrell: reporte de caso y revisión de tema. S. F. J. of Dev. [Internet]. 2023 Aug. 17 [cited 2024 Dec. 14];4(5):1994-9. Available from: https://ojs.southfloridapublishing.com/ojs/index.php/jdev/article/view/2747

3. Fajardo Idrobo Blanca Viviana, Palencia Palacios Maribel, López Mosquera Valentina, Álvarez Soler Jaime Antonio. PENTALOGY OF CANTRELL. A STILLBIRTH CASE REPORT. Case reports [Internet]. 2022 June [cited 2024 Dec 13] ; 8(1): 73-84. Available from: http://www.scielo.org.co/scielo.php?script=sci_arttext&pid=S2462-85222022000100073&lng=en. Epub Feb 10, 2023. https://doi.org/10.15446/cr.v8n1.91323.

4. Pestañas León AR, Aguirre-Cruz B, Sosa-Palaviccini MO, Jhossmar Cristians Auza-Santivañez JCA-S, Apaza-Huanca B, Márquez Molina J, et al. Prenatal diagnosis of Fetal Cardiac Rhabdomyomaassociated with Familial Tuberous Sclerosis. Interamerican Journal of Health Sciences [Internet]. 2024 Feb. 23 [cited 2025 Jan. 3];4:183. Available from: https://ijhsc.uai.edu.ar/index.php/ijhsc/article/view/200

5. Williams, A. P., Marayati, R., & Beierle, E. A. Pentalogy of Cantrell. Seminars in Pediatric Surgery, 28(2), 106–110. [Internet]. 2019 [cited 2025 Jan. 3];4:183. Available from: https://doi.org/10.1053/j.sempedsurg.2019.04.006

6. Cantrell JR, Haller JA, Ravitch MM. A síndrome of congenital defects involving the abdominal Wall, sternum, diaphragm, pericardium, and heart. Surg Gynecol Obstet. 1958 Nov;107(5):602-14. Available from: PMID: 13592660

7. Martin RA, Cunniff C, Erickson L, Jones KL. Pentalogy of Cantrell and ectopia cordis, a familial developmental field complex. Am J Med Genet. 1992;42 (6):839-841.Available from: https://doi.org/10.1002/ajmg.1320420619

8. Chandran S, Ari D. Pentalogy of Cantrell: an extremely rare congenital anomnaly. J Clin Neonatal. 2013;2(2):95-97. Available from: https://doi.org/10.4103/2249-4847.116410

9. Toyama WM. Combined congenital defects of the anterior abdominal wall, sternum, diaphragm, pericardium, and heart: a case report and review of the syndrome. Pediatrics. 1972 Nov;50(5):778-92. https://pubmed.ncbi.nlm.nih.gov/4263752/

10. Balderrábano-Saucedo N, Vizcaíno-Alarcón A, Sandoval-Serrano E, Segura-Stanford B, Arévalo-Salas LA, de la Cruz LR, Espinosa-Islas G, Puga-Muñuzuri FJ. Pentalogy of Cantrell: Forty-two Years of Experience in the Hospital Infantil de Mexico Federico Gomez. World J Pediatr Congenit Heart Surg. 2011 Apr;2(2):211-8. https://doi.org/10.1177/2150135110390528

11. Wen L, Jun-lin L, Jia H, Dong Z, Li-guang Z, Shu-hua D, Wei-jin L, Yun-hua G. Cantrell syndrome with complex cardiac malformations: a case report. J Pediatr Surg. 2011 Jul;46(7):1455-8. https://doi.org/10.1016/j.jpedsurg.2011.03.071

12. Suehiro K, Okutani R, Ogawa S, Nakada K, Shimaoka H, Ueda M et al (2009) Tratamiento perioperatorio de un neonato con síndrome de Cantrell. J Anesth 23(4):572–575. https://doi.org/10.1007/s00540-009-0785-9

13. Rossi A, Forzano L, Veronese P, Fachechi G, Marchesoni D (2011) Pentalogía incompleta de Cantrell durante el primer trimestre del embarazo. Minerva Ginecol 63(4):399–400.

14. Rodgers, Elizabeth B., et al. "Diagnosis of Pentalogy of Cantrell Using 2-and 3-Dimensional Sonography." Journal of ultrasound in medicine 29.12 (2010): 1825-1828.

15. Fernandez, M. S., et al. "Cantrell's pentalogy. Report of four cases and their management." Pediatric surgery international 12 (1997): 428-431.

16. Hornberger LK, Colan SD, Lock JE, Wessel DL, Mayer JE Jr.. Resultado de pacientes con ectopia cordis y defectos intracardíacos significativos. Circulation. 1996;94:32–7. PMID: 8901716

CONSENT

The patient's consent was obtained for the realization of this work.

FINANCING

The authors did not receive funding for the implementation of this study.

CONFLICT OF INTEREST

The authors declare that there is no conflict of interest.

CONTRIBUTION AUTHORSHIP

Conceptualization: Jhossmar Cristians Auza-Santivañez, Benito Aguirre-Cruz.
Research: Jhossmar Cristians Auza-Santivañez.
Methodology: Benito Aguirre-Cruz, Jhossmar Cristians Auza-Santivañez.
Visualization: Blas Apaza-Huanca, Dilma Villca-Villca.
Original draft: Benito Aguirre-Cruz, Jhossmar Cristians Auza-Santivañez, Elizeth Mendoza-Contreras, Ritdber Alfredo Quispe Sarmiento, Pestañas-León Alexin Raúl, Jhoel Vargas Apaza, Blas Apaza-Huanca, Dilma Villca-Villca.
Writing-revision and editing: Benito Aguirre-Cruz, Jhossmar Cristians Auza-Santivañez, Elizeth Mendoza-Contreras, Ritdber Alfredo Quispe Sarmiento, Pestañas-León Alexin Raúl, Jhoel Vargas Apaza, Blas Apaza-Huanca, Dilma Villca-Villca.