CASE REPORT

Prenatal Diagnosis of Ectopia Cordis in Honduras: Case Report

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Abstract

Introduction. Ectopia cordis is an extremely rare congenital heart malformation occurring in 5.5 to 7.9 per million live births. It is characterized by the anomalous position of the heart outside of the thoracic cavity. We report a case of ectopia cordis with thoracic presentation due to its rare occurrence.

Clinical Case. A 24-year-old primigravid Hispanic woman was referred at 32 weeks of gestation to a routine ultrasonography. Detailed ultrasonography revealed a singleton fetus with heart situated outside the thoracic cavity. No other abnormalities were detected. The findings were strongly suggestive of ectopia cordis. At 38 weeks cesarean delivery was performed receiving a full term neonate girl with ectopic heart in the chest. The sternum was entirely deficient with an externally-visible beating heart over the chest wall. The abdominal wall was intact.

Discussion and Conclusions. Ectopia cordis is a rare congenital heart defect defined by anomalous position of the heart outside the thorax associated to defects in pericardium, diaphragm and sternum. Usually, it is also associated to other intracardiac congenital heart defects. First trimester prenatal diagnosis of ectopia cordis is feasible and can be performed at about the tenth week. Early surgical approach has provided the repositioning of the heart into the thoracic cavity. However, attempts at surgical correction have been largely unsuccessful. As this is considered a sporadic event, the recurrence risk is not increased over that of the general population.

Key Words

Ectopia cordis, congenital, heart defect

■ INTRODUCTION

Ectopia cordis is an extremely rare congenital heart malformation occurring in 5.5 to 7.9 per million live births. It is characterized by the anomalous position of the heart outside of the thoracic cavity. The thoracic presentation is the most common form (59%). The cause of ectopia cordis is currently unknown, and cases are sporadic. The prognosis is poor, and most infants are stillborn or die within the first few hours or days of life.(1,2)

We report one case of isolated thoracic ectopia cordis without associated congenital heart defect due to its rare form of occurrence.

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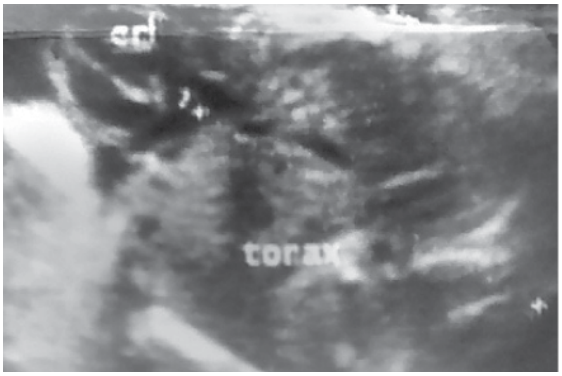
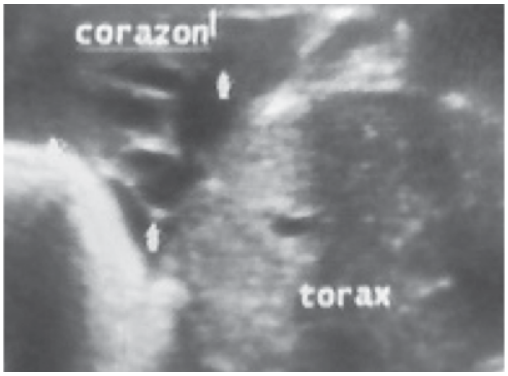
■ CLINICAL CASE

A 24-year-old primigravid Hispanic woman with uneventful previous medical history and pregnancy course was referred at 32 weeks of gestation to a routine detailed ultrasonography. The patient had received no regular prenatal care and had no family history of congenital anomalies. Ultrasonographic examination showed a singleton fetus with normal amniotic fluid volume. Fetal biometry was consistent with date (1670 gr). Detailed ultrasonography revealed a defect in the thoracic wall and the heart was located outside the thoracic cavity (Figures 1 and 2). No abdominal, craniofacial or intracardiac abnormalities were detected. These findings were strongly suggestive of ectopia cordis. At 37 weeks a new ultrasound confirmed a beating fetal heart protruding through the thoracic wall defect.





Figures 1 and 2: Fetal heart visibly outside thoracic cavity protruding through defect.

At 38 weeks cesarean delivery was performed receiving a girl with ectopic heart in the chest. (Figure 3) The placenta and umbilical cord were unremarkable. Physical examination showed a full-term neonate with a normal Apgar Score (8

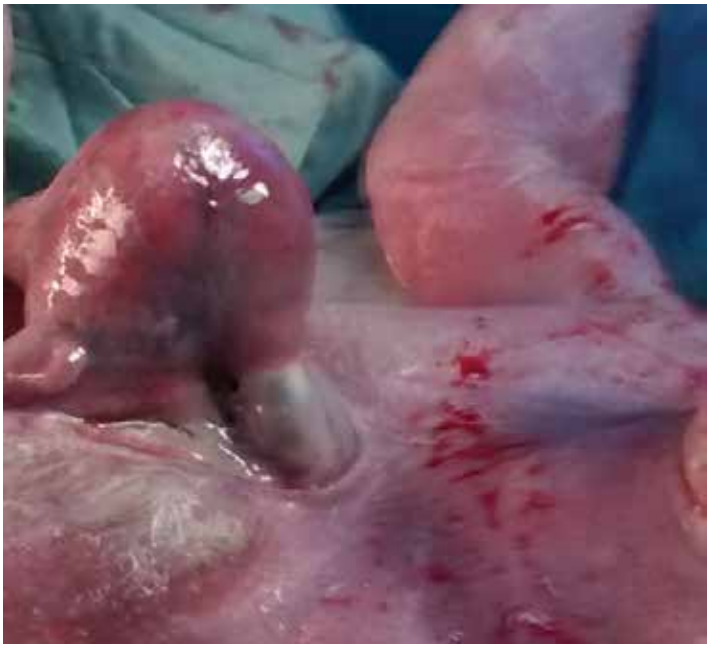


Figure 3: thoracic ectopia cordis

and 9 at one and five minutes respectively). The sternum was entirely deficient with an externally-visible beating heart over the chest wall. The abdominal wall was intact. No other visible defects were found. Postnatal echocardiography revealed no associated cardiac anomalies. Newborn died 24 hours after birth.

■ DISCUSSION

Ectopia cordis is a rare congenital heart defect defined by anomalous position of heart outside the thorax associated to defects in pericardium, diaphragm and sternum. It is a rare congenital abnormality occurring in 5.5–7.9 per million live births. It was first observed by Haller in 1706 and its name was first proposed by Abbott in 1898. It may occur as an isolated malformation, or it may be associated with a larger category of ventral body wall defects that affect the thorax, abdomen, or both. Usually, it is also associated to other intracardiac congenital heart defects, and troncoconal malformations are the most common ones. (3,4)

Cervical, cervicothoracic, thoracic, and thoracoabdominal types of ectopia cordis have been described. Cervical and cervicothoracic ectopia cordis are relatively rare, while the thoracic, abdominal and thoracoabdominal types are more common. Thoracic presentation is the most common form (59%) and has a better prognosis, making long-term survival possible. (5)

Most cases of ectopia cordis are diagnosed in the second trimester. First trimester prenatal diagnosis of ectopia cordis is feasible and can be performed at about the tenth week. The main ultrasonographic finding is a fetal heart beating outside the thoracic cavity. In the present case, no first trimester ultrasonography was performed and diagnosis was established during a third trimester ultrasonography.

When early antenatal ultrasonographic diagnosis of ectopia cordis is made other associated anomalies, such as omphalocele and craniofacial defects, should be looked for. With an established antenatal diagnosis, elective cesarean delivery followed by corrective or staged surgical palliation or repair of the ectopia cordis can be planned. Vaginal delivery could result in prolonged cardiac compression, damage to herniated viscera, or rupture of atrial diverticula or omphalocele sacs. Postnatal echocardiography is essential for the diagnosis of associated cardiac anomalies.(6,7)

The prognosis is poor, and most infants are stillborn or die within the first few hours or days of life. Surgery is the only therapeutic option. Early surgical approach has provided the repositioning of the heart into the thoracic cavity due to the greater elasticity of the thoracic wall. However, attempts at surgical correction have been largely unsuccessful because of the extent of the associated anomalies. Obstetric management should include a careful search for associated anomalies, especially cardiac anomalies, and assessment of fetal karyotype. As this is considered to be a sporadic event, the recurrence risk is not increased over that of the general population.(8,9)

■ CONCLUSIONS

First trimester prenatal diagnosis of ectopia cordis is feasible and can be performed at about the 10th week.
Careful search for associated anomalies, especially cardiac anomalies, and assessment of fetal karyotype
Thoracic presentation is the most common and has better prognosis, making long-term survival possible.
As this is considered to be a sporadic event, the recurrence

risk is not increased over that of the general population.

Diagnóstico prenatal de ectopia cordis en Honduras: Informe de un caso

Resumen

Introducción. La ectopia cordis es una malformación cardiaca congénita extremadamente infrecuente que ocurre en 5,5 a 7,9 nacidos vivos por millón. Se caracteriza por la posición anómala del corazón fuera de la cavidad torácica. Informamos un caso de ectopia cordis con presentación torácica debido a su rara ocurrencia.

Caso clínico. Una mujer hispana primigesta de 24 años de edad, se refiere a las 32 semanas de gestación a una ecografía de rutina. La ecografía detallada reveló un feto único con el corazón situado fuera de la cavidad torácica. No se detectaron otras anomalías. Los resultados fueron muy sugestivos de ectopia cordis. A las 38 semanas se realizó la cesárea y nació una niña a término con ectopia cordis torácica. El esternón era defectuoso con un corazón, visible desde el exterior, que latía sobre la pared torácica. La pared abdominal estaba intacta.

Discusión. La ectopia cordis es una cardiopatía congénita rara definida por la posición anómala del corazón fuera del tórax asociada a defectos en el pericardio, el diafragma y el esternón. Por lo general, también se asocia con otros defectos congénitos intracardíacos. El diagnóstico prenatal de ectopia cordis en el primer trimestre es posible y se puede realizar aproximadamente en la décima semana. El abordaje quirúrgico precoz realiza el reposicionamiento del corazón en la cavidad torácica. Sin embargo los intentos de corrección quirúrgica en gran medida no han resultado exitosos.

Palabras clave

Ectopia cordis, defecto congénito del corazón

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Pancreatic Islet Cell Transplantation Markedly Improves Glycemic Control and Hypoglycemic Awareness

Written by Kristin Della Volpe

• May 6, 2016 SOURCE http://www.endocrineweb.com/professional/type-1-diabetes/pancreatic-islet-cell-transplantation-markedly-improves-glycemic-control

Transplantation of purified human pancreatic islet cells into patients with type 1 diabetes significantly reduces the risk of severe hypoglycemic events, and increases glycemic control and hypoglycemic awareness, according to findings from a phase 3 trial recently published online in *Diabetes Care*.

The trial, funded by the National Institutes of Allergy and Infectious Diseases (NIAID) and of Diabetes and Digestive and Kidney Diseases (NIDDK), was conducted by the Clinical Islet Transplantation Consortium (CIT), and designed in consultation with the U.S. Food and Drug Administration to enable potential future licensure of purified human pancreatic islet manufacture.

"The findings suggest that for people with life-altering severe hypoglycemia despite optimal medical management, islet transplantation offers a potentially lifesaving treatment that in the majority of cases eliminates severe hypoglycemic events, while conferring excellent control of blood sugar," Anthony S. Fauci, MD, Director of the NIAID, said to the press.

"… the promise of islet transplantation is undeniable and encouraging," said NIDDK Director Griffin P. Rodgers, MD. "Even with the best care, about 30% of people with type 1 diabetes aren't aware of dangerous drops in blood glucose levels."

Study Design

The study included 48 patients with type 1 diabetes for more than 5 years who had persistent impaired awareness of hypoglycemia and severe hypoglycemic events, despite expert management for at least 1 year prior to study enrollment.

All study participants received at least one transplant of islet cells injected into the portal vein. Subjects who still needed insulin 75 days after transplant underwent a second transplant (25), while one received a third transplant.

Nearly 90% of Patients Achieved Freedom from Severe Hypoglycemic Events

One year after the first transplant of islet cells, 88% of study participants were free of severe hypoglycemic events, had established near-normal control of glucose levels, and restored hypoglycemic awareness. These results persisted for 71% of the participants 2 years after transplantation. Freedom from insulin was achieved in 52% of patients at 1 year and 43% at 2 years

Risks and Benefits of Islet Transplantation

Although some side effects were serious, none led to death or disability.

"... as the immunosuppression drugs can have significant adverse side effects (infections and lowered kidney function), the treatment only makes sense for people who have frequent severe hypoglycemia despite optimal diabetes management or for those already on immunosuppressive drugs for a kidney transplant," said study coauthor Tom Eggerman, MD, PhD, NIDDK scientific officer for the CIT.

The researchers continue to follow participants to determine the longer-term benefits of islet transplantation and whether these outweigh the risks associated with immunosuppressive drug use.

First License-Enabling Trial

"Licensure is critical because it will ensure the quality, consistency, and safety of the islet product; provide greater patient access to islet cell transplant; and accelerate continued research that we hope will make this procedure suitable for a broader population of people with type 1 diabetes," said coauthor Nancy D. Bridges, MD, Chief of the NIAID Transplantation Branch.

Last month Science Daily reported on a study (see page 19) in which researchers discovered how to encourage lab-grown beta cells to mature into functional insulin-producing cells, which would allow autotransplants that may eliminate the need for immunosuppressive drug use.