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Case Report

Cardiocentro ERNESTO CHE GUEVARA SANTA CLARA CUBA

Thoracic *ectopia cordis* in a human embryo of eight weeks

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ABSTRACT
Defects of the ventral body wall occur in the thorax, abdomen and pelvis, and when they affect the thoracic region, with total or partial displacement of the heart outside the cavity, they give rise to thoracic *ectopia cordis*. The case of a human embryo of 22 mm skull-spine, week 8, stage 21 of embryonic development according to Carnegie, from voluntary abortion with misoprostol, is presented. As abnormal feature, in the external morphological examination the presence of an exposed cardiac apex in the ventral region of the chest was noted, which led to the diagnosis of thoracic *ectopia cordis*. The morphological study of aborted embryonic specimens may reveal developmental abnormalities that are not usually diagnosed due to the smallness of the product and the precocity of the loss.
Key words: Human embryo, Thoracic *ectopia cordis*, Diagnosis

Ectopia cordis torácica en embrión humano de 8 semanas

RESUMEN
Los defectos de la pared corporal ventral se producen en el tórax, el abdomen y la pelvis; cuando afectan la región torácica, con desplazamiento total o parcial del corazón fuera de la cavidad, dan origen a la *ectopia cordis* torácica. Se presenta el caso de un embrión humano de 22 mm de longitud cráneo-raquis, semana 8, estadio 21 del desarrollo embrionario según Carnegie; proveniente de aborto voluntario por misoprostol. En el examen morfológico externo se constató como detalle anormal la presencia de un ápex cardíaco expuesto en la región ventral del tórax, lo que llevó al planteamiento diagnóstico de *ectopia cordis* torácica. El estudio morfológico de especímenes embrionarios abortados puede poner en evidencia anomalías del desarrollo que usualmente no son diagnosticadas por la pequeñez del producto y la precocidad de la pérdida.
Palabras clave: Embrión humano, *Ectopia cordis* torácica, Diagnóstico

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INTRODUCTION

Abnormalities or defects of the ventral body wall occur in the thorax, abdomen and pelvis, and depending on the location and size of the abnormality, the abdominal viscera (gastroschisis), the urogenital organs (bladder or cloa-

cal exstrophy), and the heart are affected. The latter case is called *ectopia cordis* or cardiac, exocardia or ectocardia [1].

Variants of cervical, thoracic and abdominal *ectopia cordis* are known, showing total or partial displacement of the heart outside the thoracic cavity, with an incidence between 5 and 7.5 cases per 1,000,000 live births and it is most frequently described in males[2]. This defect appears to be due to insufficient progress of cephalic and lateral folding and to incomplete development of the body wall structures, including muscles, bone and skin[3]. Published cases suggest family inheritance problems[4,5], and evidence show a mutation in a gene mapped in Xq25 - q26, which is in correspondence with the ventral midline as an important field of development[2].

Thoracic shape appears as a result of lack of complete fusion of the lateral folds in the formation of the thoracic wall in the fourth week[6]. There may be a sternal defect and heart protrudes through the anterior wall; in the most frequent variants the sternum may be absent, or present, with a large defect; it may be incomplete, when the heart is covered by skin or pericardial sac; or complete, if the heart is exclusively coated with visceral pericardium. Although the normal position of the heart is affected, its embryogenesis is not compromised; however, it may be associated with defects such as ventricular septal anomalies and tetralogy of Fallot[3,5].

CASE REPORT

The case of a human embryo of 22 mm skull-spine, obtained from voluntary abortion with drug method (misoprostol), is presented with prior informed consent of the woman. Now it is part of the *Embrioteca* of the Faculty of Medicine in Villa Clara.

It was a 16 year-old primigravida mother, asthmatic, smoker, with no personal or family history of congenital malformations or known exposure to teratogens. The embryo showed some signs of maceration attributable to the abortion process, so it was not sent to histologic study. Little cefalocaudal curvature, exposed bowel loops unfused palatal crests, incomplete fusion of nasomedian processes, separation of the fingers and start of separation of toes were observed (**Figure 1**). Both the skull - spine length as the details provided by external morphological examination allowed to classify it as a specimen stage 21, according to Carnegie[7], corresponding to week 8. It was noticeable, when examined with magnification, the presence of an *apex cordis* exposed in the ventral region of the thorax (**Figure 2**), which was corroborated by analyzing in detail the anatomical part and the photos taken by magnification options of the image viewer, which led us to diagnosis approach of thoracic *ectopia cordis.*

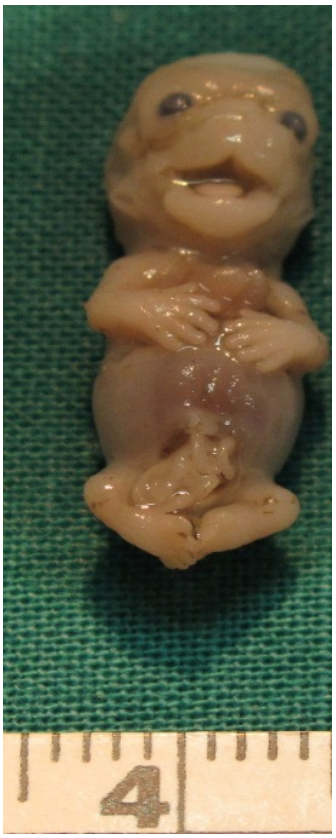

Figure 1. Front panoramic view of a human embryo of 8 weeks.

COMMENT

Isolated forms of *ectopia cordis* have been reported both in fetus[8-10] and in newborns[5,11], while variants associated with other anomalies trigger syndroms such as Cantrell's pentalogy[1,12], and thoracoabdominal syndrome[2,4], which can not be discarded in this case due to incipient signs of maceration which the specimen presented, its age and inability to do mi-

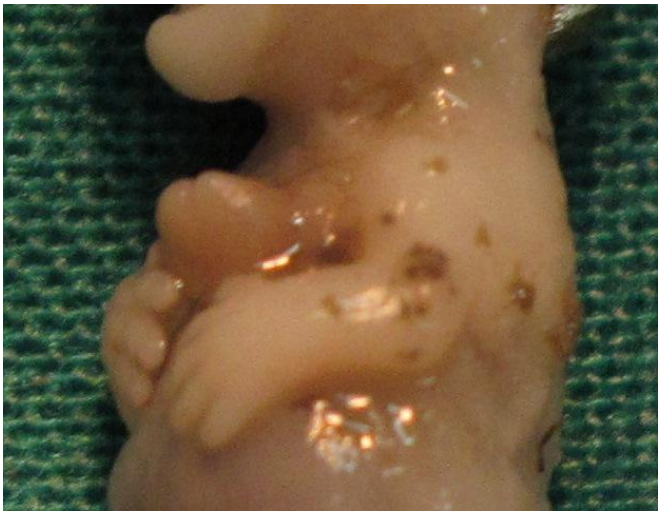

Figure 2. Selective side left view of the thorax.

croscopic and chromosome tests, which limited us to present it as an isolated form of thoracic *ectopia cordis*.

Although utero echocardiographic diagnosis has allowed, from 10 weeks of gestation, setting the prenatal diagnosis of cardiac ectopias, mainly of thoraco-abdominal forms accompanied by omphalocele[8] , identification of this malformation as early as the case presented (8 weeks) is novel. In this period , the presence of exposed bowel loops is attributed to the physiological umbilical hernia normal in this stage of development, so that diagnosis of omphalocele is not feasible as it is not until the tenth week that these loops return to the abdominal cavity. Labiopalatine defects cannot be stated either as the extent of development (morphologically speaking) that this important facial segment presents corresponds to the age of the specimen. The morphologic study of aborted embryo evidences developmental abnormalities that are not usually diagnosed due to the smallness of the product and the precocity of the loss. This would facilitate timely genetic counseling to women for future pregnancies.

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