

# Cardiac abnormality ectopia cordis

## Abstract

Ectopia cordis is a rare disease, with an incidence of 5.5-7.9 cases per million births. Until 2010, 267 cases have been described, most of them (95%) with other associated heart disease. We study the heart diseases associated with 6 cases of ectopia cordis. Depending on the affected area, patients with ectopia cordis are classified into 4 groups: cervical, thoracic, thoracoabdominal and abdominal. The 2 patients described died before 3 days of life, four of them at birth. Three belonged to the thoracic ectopia group and another three to the thoraco abdominal group. All had interventricular communication, associated with double exit of the right ventricle in 3 (50%) and tetralogy of pulmonary fallot-atresia in another 3 (50%). In 2 hearts with double outlet there was mitral valve involvement, parachute valve stenosis and atrial mitral valve. This type of anomalies has not been described previously.

For this ectopia cordis can be complete, due to the absence of skin and parietal pericardium, or partial if it exists under the pericardium sternum or on the skin sternum. Most cases are diagnosed in the second trimester, but can be visualized from 10 weeks gestation by means of Doppler ultrasound. With established prenatal diagnosis, cesarean birth is recommended, followed by corrective or palliative surgery.

Also vaginal delivery is associated with an increased risk of cardiac compression, damage to herniated viceras or rupture of cardiac chambers. At the time of birth, a fetal echocardiogram is necessary since 80% of cases are associated with intracardiac abnormalities such as tetralogy of fallot. Thus surgical treatment is the only therapeutic option associated with most cardiac protection. Multiple surgical techniques have been described, therefore, this disease can be addressed in one or two surgical times. And the efficacy and possibility of surgery depend on the class of ectopia cordis and the congenital cardiac defects present or absence of extracardiac malformation.

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## Introduction

Ectopia cordis is a rare disease that is defined by an abnormal position of the heart outside the chest associated with defects of the parietal pericardium, diaphragm, sternum and, in most cases, heart disease. The ectopia cordis nomination was first proposed by Abott in 1898, although patients with similar defects had been described in previous decades with other nominations. Byron classified ectopia cordis into 4 groups: cervical, thoracic, thoracoabdominal and abdominal. This last group includes patients with abdominal midline (omphalocele) disease that meet the defining characteristics of the disease. In 1958, Cantrell published a syndrome with 5 defects: thoracic, abdominal wall, diaphragm, pericardium and heart abnormalities.

## Clinical cases

### Case 1

Newborn, 38 weeks gestation, with a birth weight of 2,685g. Maternal history of previous abortion. At birth, cyanosis, atrial pavilion of low implantation and an 6x6 cm omphalocele, covered by a membrane and with the open sternum (incomplete thoracoabdominal cord ectopia) were observed. The abdominal viscera were observed. On physical examination, cervicoaxial hypotonia stood out, the heart beat was auscultated through the sternum, and the vesicular murmur was diminished in both hemithorax. She was operated with artificial tissue closure and presented progressive general deterioration, with polypnea and tachycardia. Breathing assisted by atelectasis of both lower pulmonary lobes was established, despite which the patient died at 2 h. In the cardiac necropsy, there was a medial heart, a left superior

vena cava that led to the coronary sinus, and an interventricular communication due to poor alignment of the infundibular septum (0.6x0.5 cm). In addition, a parachute mitral valve, a double-outlet right ventricle, pulmonary stenosis and coronary arteries in the anterior and left sinus were associated.

### Case 2

Newborn of 38 weeks gestation and birth weight of 2,980g. Maternal history of metrorrhagia in the first trimester of pregnancy. At the time of birth, generalized cyanosis was observed that intensified with crying, omphalocele with sternal cleft (incomplete thoracoabdominal cord ectopia), and the examination highlighted a normal pulmonary auscultation and a second single reinforced noise, without murmurs. The day of admission was intervened by resection of the omphalocele sac and strengthening with synthetic tissue of the thoracoabdominal anterior face.

## Discussion

Ectopia cordis is produced by segmental defects of mesodermal development during the third week of intrauterine life and by alterations of the amniotic bands, which simultaneously produce cerebral and thoracoabdominal malformations. When there is a multiple pregnancy, only one of the fetuses is affected. Rarely, ectopia cordis is associated with chromosomal abnormalities. The existence of ectopia cordis with severe heart disease has been confirmed in perinatal age by vaginal echocardiography at 10-12 weeks gestation or abdominal at 20-22 weeks.

For this until 2010, 267 patients have been described, 102 (39.2%) of the thoracic group and 99 (38%) of the thoracoabdominal. 90%

died in the first year of life. Cases of the cervical group rarely survive one day. The majority (95%) have an associated heart disease. The type of heart disease that occurs most frequently is interventricular communication, present in 59% of cases, followed by interatrial communication in 35%, stenosis or pulmonary atresia in 36%, tetralogy of Fallot in 22%, right ventricle diverticulum in 13%, upper left vena cava in 12% and double outlet of the right ventricle in 13%. Other heart diseases, such as the single ventricle, transposition of large vessels and complete atrioventricular defect rarely occur. In our experience, the 6 cases belonged to the thoracic groups (3 cases) and thoracoabdominal groups (3 cases). The majority presented some of the common heart diseases, such as interventricular communication (6 cases) and pulmonary duct obstruction (5 cases) associated with more complex defects: the right ventricle with double outlet (3 cases; 50%) and atresia or Parachute stenosis of the mitral valve, abnormality not previously described.<sup>1-3</sup>

It can also be said ectopia cordis can be complete, due to absence of skin and parietal pericardium, or partial if it exists under the pericardium sternum or over the skin sternum. The existence of a partial inferior or superior sternum defect without total opening with the presence of parietal pericardium and skin favors surgical treatment and prevents chest compression when the viscera is introduced into the cavity. In recent years, surgical correction has been attempted in one or two phases with variable results, fundamentally dependent on the type of associated heart disease.<sup>4-13</sup>

## Conclusion

Patients with ectopia cordis have severe heart disease, mostly trunk-shaped abnormalities. Two of our patients had defects not previously described. Despite having initiated surgical treatment attempts, the survival of patients with these heart diseases is very low, and most of them die within the first week of life. Therefore, it can be concluded that the EC is a lethal anomaly that requires rapid medical and surgical interventions. Surgery in these patients with complex life-threatening intracardiac anomalies has the only chance of survival, which should still be attempted despite the poor results so far. The success of the repair is dictated by the presence and severity of the intrinsic heart defects and associated congenital anomalies, rather than by the type of surgical approach. In this way, various investigations continue to be carried out to respond and treat pathologies such as these.

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## Conflicts of interest

No conflicts to declare.

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