

## Ectopia Cordis and Cardiac Anomalies

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Ectopia cordis is a rare disease that occurs in 5.5 to 7.9 per million live births. Only 267 cases had been reported as of 2001, most (95%) associated with other cardiac anomalies. We studied the cardiac malformations associated in 6 patients with ectopia cordis. Depending on where the defect was located, the cases of ectopia were classified into four groups: cervical, thoracic, thoraco-abdominal, and abdominal. All 6 patients died before the third day of life, 4 during delivery. Three of the patients were included in the thoracic group, whereas the other 3 belonged to the thoraco-abdominal group. All the patients had associated ventricular septal defects, 3 double-outlet right ventricle (50%) and the rest (50%) tetralogy of Fallot-pulmonary atresia. Two patients with double-outlet right ventricle presented mitral-valve pathology, a parachute valve and an atresic mitral valve. None of these cardiac anomalies have been reported to date.

**Key words:** *Ectopia cordis. Congenital heart defects. Tetralogy of Fallot. Ventricular septal defect.*

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### Anomalías cardíacas en la ectopia cordis

La ectopia cordis es una enfermedad rara, con una incidencia de 5,5-7,9 casos por millón de nacimientos. Hasta el año 2001 se han descrito 267 casos, la mayoría de ellos (95%) con otra cardiopatía asociada. Estudiamos las cardiopatías asociadas a 6 casos de ectopia cordis. En función de la zona afectada, los pacientes con ectopia cordis se clasifican en 4 grupos: cervical, torácica, toraco-abdominal y abdominal. Los 6 pacientes descritos fallecieron antes de los 3 días de vida, cuatro de ellos en el momento del nacimiento. Tres pertenecían al grupo de ectopia torácica y otros tres al de toracoabdominal. Todos tenían comunicación interventricular, asociada a doble salida del ventrículo derecho en 3 (50%) y a tetralogía de Fallot-atresia pulmonar en otros 3 (50%). En 2 corazones con doble salida existía afección de la válvula mitral, estenosis por válvula en paracaídas y válvula mitral atrésica. Este tipo de anomalías no ha sido descrito anteriormente.

**Palabras clave:** *Ectopia cordis. Defectos congénitos del corazón. Tetralogía de Fallot. Comunicación interventricular.*

## INTRODUCTION

Ectopia cordis is a rare disease that it is defined by the abnormal position of the heart outside the thorax associated with defects in the parietal pericardium, diaphragm, sternum, and, in most cases, cardiac malformations. The designation ectopia cordis was first proposed by Abbott in 1898,<sup>1</sup> although cases of patients with similar defects had been described for decades with other designations. Byron<sup>2</sup> classified ectopia cordis into four types: cervical, thoracic, thoracoabdominal, and abdominal. The abdominal group included patients with an anomaly of the abdominal midline

(omphalocele) who met the defining characteristics of the disease. In 1958, Cantrell<sup>3</sup> reported a syndrome with five defects: anomalies of the chest wall, abdomen, diaphragm, pericardium, and heart.

We report the results of an anatomic study of the type of congenital cardiac malformations present in a series of 6 patients with ectopia cordis, 3 of the thoracoabdominal type and 3 of the thoracic type.

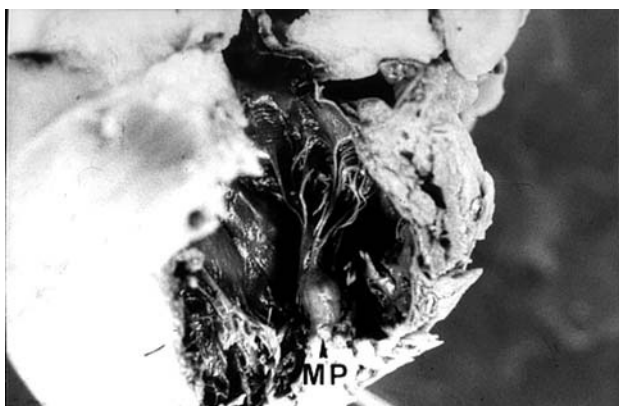
## CLINICAL CASES

### Case 1

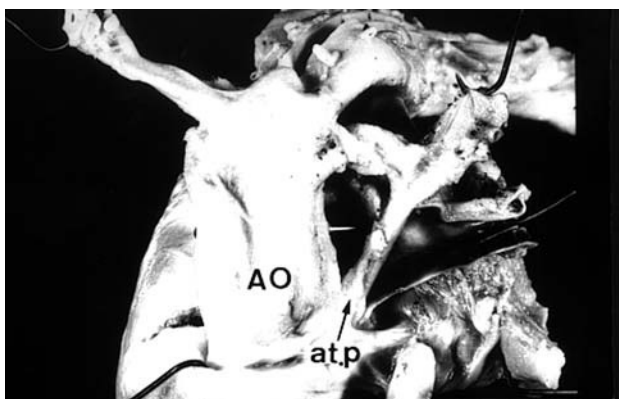
A newborn girl, gestational age 38 weeks, birth weight 2685 g. Maternal history of miscarriage. At birth she was cyanotic, had ears with a low implantation, omphalocele of 6.6 cm covered by a membrane, and an open sternum (incomplete thoracoabdominal

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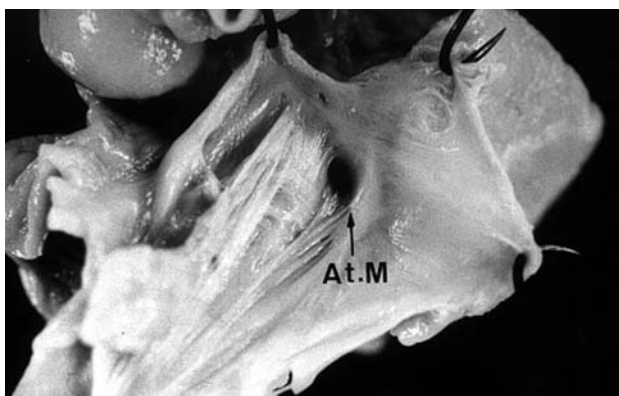
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**Fig. 1.** Left ventricle with parachute mitral valve in case 1. All the cords of the mitral valve are inserted in a single medial papillary muscle (PM).



**Fig. 2.** Pulmonary atresia with perimembranous ventricular septal defect in case 2. The aorta is very dilated and displaced forward, atretic right ventricular outflow tract with hypoplastic pulmonary trunk.



**Fig. 3.** Left atrium from case 3, showing mitral atresia and a central zone with an unperforated hollow corresponding to the atretic mitral valve (At.M).

ectopia cordis). The abdominal organs were visible. The physical examination disclosed cervicoaxial hypotonia, the heart beat was auscultated through the sternum, and the vesicular murmur was reduced in both hemithoraces. Surgery was performed to close

the opening with artificial tissue, but the infant presented progressive general deterioration, polypnea, and tachycardia. Ventilation was begun due to atelectasis of both pulmonary lower lobes, but she died within 24 h. In the cardiac post mortem study, the heart was medial, the superior vena cava opened onto the coronary sinus, and there was a ventricular septal defect due to poor alignment of the infundibular septum (0.6 0.5 cm). In addition, a parachute mitral valve (Figure 1), right double-outflow ventricle, pulmonary stenosis, and coronary arteries opening on the anterior and left coronary sinuses were present.

### Case 2

A newborn boy, gestational age 38 weeks, birth weight 2980 g. The mother had a history of metrorrhagia in the first trimester of pregnancy. At birth, the infant had generalized cyanosis that intensified with crying, and omphalocele with a split sternum (incomplete thoracoabdominal ectopia cordis). The physical examination revealed normal pulmonary auscultation and reinforcement of the second heart sound, without murmurs. Surgery was performed on the day of admission to resect the omphalocele sac and reinforce the anterior thoracoabdominal wall with synthetic tissue. The patient developed anuria and died 3 days after birth. In the cardiac post mortem study, there was a perimembranous ventricular septal defect, pulmonary atresia (Figure 2), and coronary arteries opening on the posterior and left sinus.

### Case 3

A newborn boy, gestational age 42 weeks, birth weight 2900 g. At birth he had Apgar score 7 and 9 (5 min), cyanosis of the skin and mucous membranes, polypnea with flail chest, omphalocele with an open sternum, and medial heart. In the chest radiograph the diaphragm was absent and the abdominal organs were in the chest (incomplete thoracoabdominal ectopia cordis). Surgery was performed but the patient died within 24 h. The cardiac post mortem study disclosed drainage of the superior and inferior cava veins into the right atrium, drainage of the pulmonary veins into the left atrium, mitral atresia (Figure 3), perimembranous ventricular outflow septal defect, and emergence of the pulmonary artery and aorta from the right ventricle. In addition, pulmonary valve stenosis and mild hypoplasia of the left ventricle were observed. The coronary arteries arose from the anterior and left coronary sinuses.

### Case 4

A newborn girl, gestational age 40 weeks, with a birth weight of 2800 g. At birth she had central cyanosis

with respiratory distress. She died in spite of reanimation measures. The heart was covered by visceral pericardium and the sternum was open (complete thoracic ectopia cordis). In the cardiac post mortem study, the drainage of the cava veins and pulmonary veins in their respective atria was normal. There was a perimembranous subaortic ventricular septal defect and the aorta straddled the ventricular septum. The right outflow infundibulum, pulmonary valve, and pulmonary trunk (tetralogy of Fallot) were stenotic. A right arch existed. The coronary arteries had a normal origin.

### Case 5

A newborn boy, gestational age 36 weeks, birth weight 2700 g. The infant had an open sternum and extrathoracic heart covered only by visceral pericardium (complete thoracic ectopia cordis). At birth he presented generalized cyanosis and polypnea, and he died within a few hours. In the cardiac post mortem study, the right atrium was dilated and received drainage from two cava veins and two right pulmonary veins. The foramen ovale was permeable and the left pulmonary veins drained into the left atrium. A perimembranous infundibular septal defect was present. The aorta and pulmonary artery emerged from the right ventricle, both with a subvalvular infundibulum. The left ventricle was of normal size.

### Case 6

A male newborn, gestational age 38 weeks, birth weight 3000 g. He died a few minutes after birth. The sternum was open and the patient had an extrathoracic heart covered only by visceral pericardium (complete thoracic ectopia cordis). In the cardiac post mortem study, a bilateral superior vena cava was observed, with the right vena cava draining into the right atrium and the left vena cava into the coronary sinus and right atrium. The inferior vena cava was interrupted and continued with the azygos into the superior vena cava. The right pulmonary veins drained into the right atrium and the left pulmonary veins into the left atrium. The mitral valve was hypoplastic. A perimembranous septal defect of 0.6 0.6 cm was present. The pulmonary valve was atretic and the aortic valve saddled the interventricular septum. The left ventricle was well developed.

## DISCUSSION

Ectopia cordis is produced by segmental defects in mesodermal development in the third week of intrauterine life<sup>4,5</sup> and amniotic band anomalies that produce simultaneous cerebral and thoracoabdominal malformations.<sup>6</sup> When a multiple pregnancy exists, only one fetus is affected.<sup>3</sup> Ectopia cordis is rarely associated

with chromosomal abnormalities.<sup>7-12</sup> The existence of ectopia cordis with severe congenital heart disease has been confirmed in the prenatal period by vaginal echocardiography<sup>13</sup> at 10-12 weeks of gestation or by abdominal echocardiography at 20-22 weeks.<sup>13,14</sup>

As of 2001, 267 patients have been reported,<sup>9-20</sup> 102 (39.2%) of the thoracic type and 99 (38%) of the thoracoabdominal type. Ninety percent of the infants died in the first year of life. Cases of the cervical type rarely survive a single day. Most (95%) newborns have associated cardiac malformations. The most frequent cardiac malformation is ventricular septal defect, which is present in 59% of cases, followed by atrial septal defect in 35%, pulmonary stenosis or atresia in 36%, tetralogy of Fallot in 22%, right ventricular diverticulum in 13%, left superior vena cava in 12%, and double right ventricular outflow tract in 13%. Other cardiac malformations like single ventricle, complete transposition of the great arteries, and atrioventricular septal defect, occur only rarely. In our experience, the 6 cases were of the thoracic (3 cases) and thoracoabdominal types (3 cases). Most had some of the common cardiac malformations, like ventricular septal defect (6 cases) and obstruction of pulmonary outflow (5 cases), associated with more complex defects: double right ventricular outflow tract (3 cases; 50%), and parachute atresia or stenosis of the mitral valve, an anomaly that has not yet been reported.

Ectopia cordis can be complete, with the skin and parietal pericardium absent, or partial if there is pericardium under the sternum or skin over the sternum. The existence of an upper or lower partial sternal defect without a complete opening and covered by parietal pericardium and skin facilitates surgical treatment and alleviates the thoracic compression that results from introducing the heart in the cavity. In recent years, surgery has been attempted in one or two phases with variable results that depend mainly on the type of associated heart disease.<sup>15-20</sup>

## CONCLUSIONS

Patients with ectopia cordis present serious cardiac malformations, generally troncoconal anomalies. Two of our patients had malformations that have not been described previously. In spite of attempts at surgical treatment, few patients with these cardiac malformations survive and most of them die in the first week of life.

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