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Brief Report

Exteriorisation of the heart in two siblings

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Abstract We report two siblings with isolated ectopic hearts. Neither child had associated congenital diseases. To the best of our knowledge, this is the first reported familial occurrence of ectopic hearts.

Keywords: Ectopia cordis; familial disease

EXTERIORISATION OF THE HEART, SO-CALLED "ectopia cordis", is a rare congenital malformation, with an incidence ranging from five to eight per million live births. Ectopic hearts are found either completely or partially outside the thoracic cavity, and usually lack pericardial coverage.[1] Such ectopic hearts can be otherwise normal, or can be complicated by associated intracardiac congenital malformations.[2] Non-cardiac malformations are also common.[2] A familial association has been reported in one case associated with Cantrell's pentalogy,[3] but otherwise all reported cases of ectopic hearts have been sporadic.[1,2,4] We report here the occurrence of this anomaly in 2 siblings.

Case report

A baby girl was transferred to our institution at the age of 2 days with an ectopic heart for further evaluation and possible surgical intervention. She was born at 36 weeks gestation as the sixth child of a healthy 33-year-old lady, one pregnancy having been incomplete. This pregnancy had been complicated by polyhydramnios, although there was no maternal history of infection or undue intake of medications. Physical examination revealed a well-developed pink neonate. Saturation of oxygen was 100% on partial pressure of oxygen of 0.3 and minimal ventilatory support. She weighted 2.6 kilograms. The heart was located completely outside the chest cavity, lacking pericardium and with the apex pointing anteriorly (Fig. 1). The peripheral pulses were normal. There was decreased air entry in both lungs, but abdominal examination showed no abnolmalities. The X-ray of the chest showed endotracheal and nasogastric tubes to be in position, and the stomach was on the left side. The rib cage was normal. The cardiac silhouette was sharp, with continuous inferior cardiac margin. There were moderate bilateral pleural effusions (Fig. 2, Panel A). Cross-sectional echocardiography was performed with the probe placed directly over the heart, and revealed normal intra-cardiac anatomy and normal ventricular function. Ultrasound studies of abdomen and head were normal. The karyotype was 46XX with normal chromosomes.

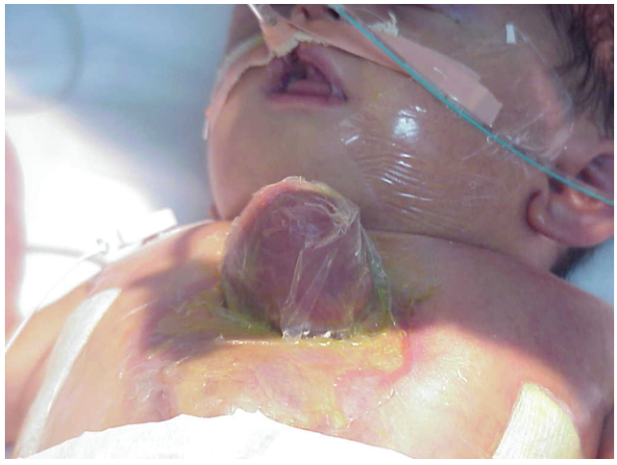

Figure 1.
The ectopic heart, covered with sterile plastic, is located outside the chest cavity, with the apex pointing anteriorly.

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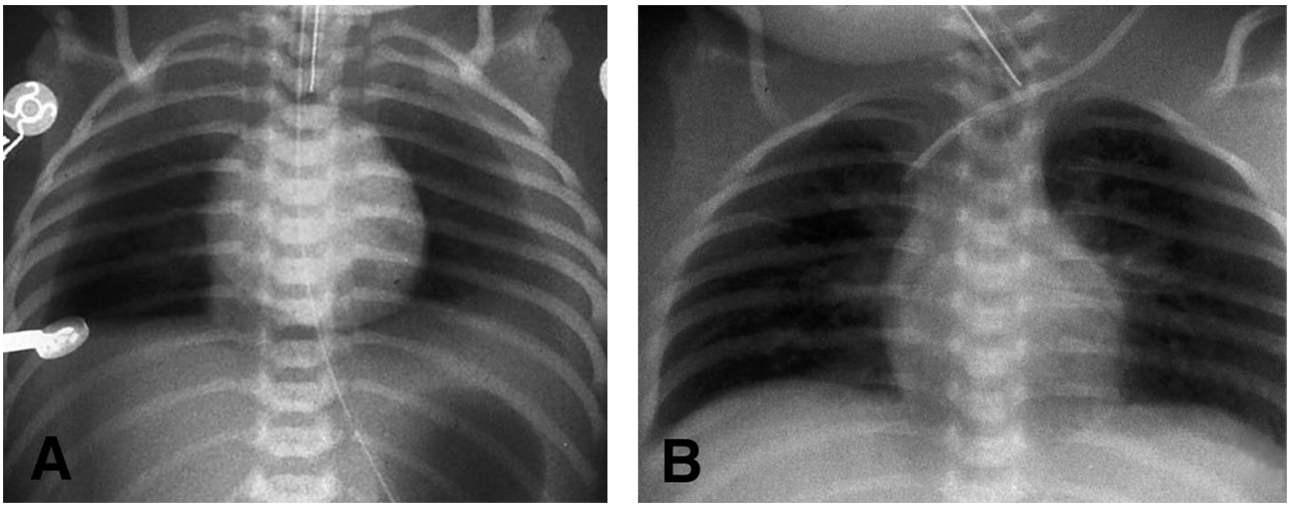

Figure 2.
Panel A shows the pre-operative chest X-ray in anterio-posterior projection, illustrating the left sided stomach and normal rib cage. Note the sharp cardiac silhouette, with a continuous inferior cardiac margin and moderate bilateral pleural effusions. Panel B shows the post-operative chest X-ray, also in anterio-posterior projection, showing a normal cardiac silhouette and resolved pleural effusions.

Family history

The parents are first-degree cousins, with 6 children. The fifth child, currently a 4-year-old boy, was also born with an isolated thoraco-abdominal ectopic heart. He underwent staged repair at the ages of one year, and eighteen months, at another institute. Currently, he is alive and well.

Surgery

In preparation for surgery, the heart was covered with plastic and sterile gauze. Frequent irrigation of the sterile gauze was performed to reduce evaporation and loss of fluids. Handling and examination were kept to a minimum to reduce the risk of infection. We used a combined surgical approach, with plastic and cardiothoracic surgeons joining their efforts to correct the defect. Initially, the skin on both sides of the ectopic heart was undermined all the way to the lateral mid axillary line. A space was created in the thoracic cavity by pushing the liver into the abdominal cavity, suturing it to the peritoneum and the abdominal wall. The ectopic heart was then reduced into the cavity overlying the middle of the diaphragm. The chest wall was reconstructed using Marlex mesh® (Bard Limited, Crawley, UK), which was sutured to the abdominal wall and to the remnant of the pericardium. This covered the heart relatively well. A bi-pedicled flap then was created on both sides of the chest and abdomen. Because of insufficiency of these skin flaps, we delayed suturing, covering the defect with Allo Derm® (Acellular dermal graft for human transplantation. Life Cell Corporation Branchburg, New Jersey). The Marlex mesh® covering the heart was itself covered with Allo Derm®. A Xeroform® dressing (Kimberly-Clark Corporation. Roswell, GA) was then applied, and this was also covered with abdominal pad and Kerlix® (Kendall Healthcare Products Company, MansField, MA).

The patient was brought back to the operating room after a few days to advance the bi-pedicled flaps and close the defect. The procedure was tolerated well, and the patient was transferred to the intensive care unit in stable condition.

Discussion

Exteriorisation of the heart, or "ectopia cordis", is a rare congenital malformation that results from failure of maturation of the midline mesodermal components of the chest and abdomen. Ectopia is classified, according to the degree of soft tissue coverage and the position of the heart, into cervical, cervicothoracic, thoraco-abdominal, and abdominal variants.[1] The exact cause and pathogenesis, however, are unknown.[2] Thus far, all anatomically normal ectopic hearts have been sporadic. Martin et al[3] indicated that the association of defects of sternal fusion with various cardiac, diaphragmatic, and anterior body wall malformations represents a developmental field complex that includes the pentalogy of Cantrell and ectopic heart. They presented a family in which three consecutively born brothers had extensive diaphragmatic defects. In addition, two had the pentalogy of Cantrell, and one of these two had an ectopic heart. This was the first reported case of familial pentalogy of Cantrell since its original description in 1958. An X-linked dominant inheritance has been suggested for the so-called

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thoraco-abdominal syndrome. This syndrome is composed of diaphragmatic and ventral hernias, hypoplasia of the lungs, and cardiac anomalies. Another syndrome that combines defects of the abdominal wall and the diaphragm, however, is the sporadic pentalogy of Cantrell. In previous series, and in case reports of ectopic hearts,[1,2,4–8] either anatomically normal or found in association with congenital cardiac diseases or other congenital malformations, the occurrence has been sporadic. To the best of our knowledge, our case report is the first case of familial incidence of isolated ectopic heart. The mode of inheritance in this family, and the genetic bases of its occurrence, are difficult to ascertain. Unlike the familial pattern of inheritance reported with the other syndromes of midline defects, the pattern of incidence in this family dose not suggests an X-linked dominant inheritance. We plan detailed analysis of the deoxyribonucleic acid from both the parents and all the siblings.

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