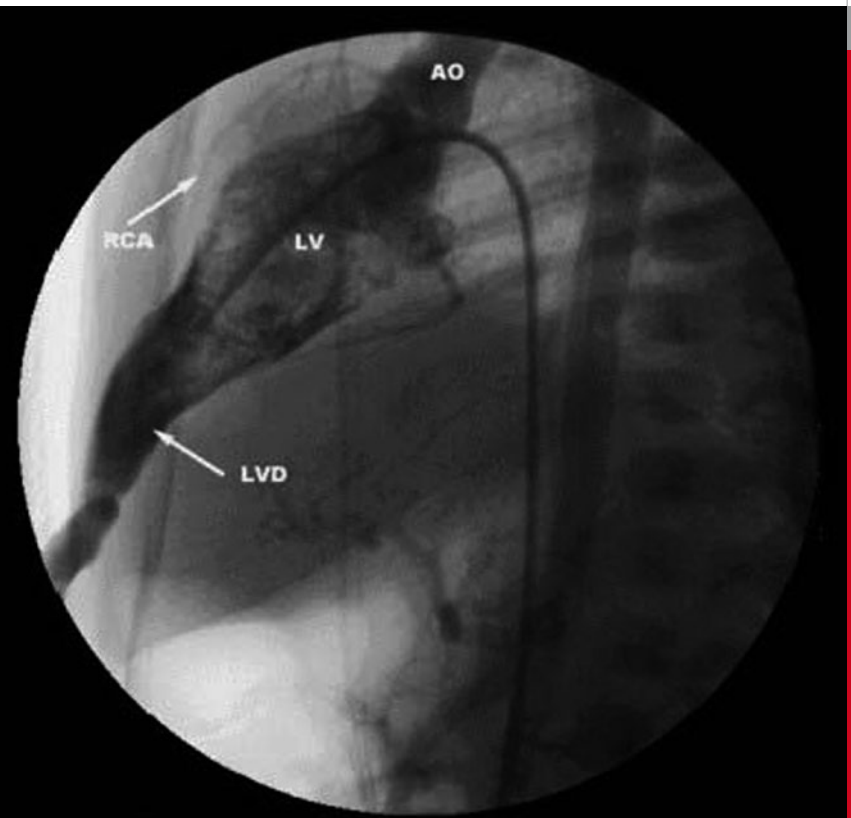
SWISS SOCIETY OF NEONATOLOGY

Cantrell's pentalogy: an unusual midline defect



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A full-term baby boy was transferred from a regional hospital with an abdominal tumor discovered at birth. Fetal ultrasounds had been normal and delivery unremarkable except for the presence of meconium stained amniotic fluid. Family history was negative except for an umbilical hernia of the two-year-old sister which had resolved spontaneously. The infant showed neither signs of respiratory distress nor hemodynamic instability.

Physical examination showed a pulsating purple tumor in the midline, just above the herniated umbilicus with dehiscence of the abdominal wall muscles. A sinuous pulsatile cord was also seen under that abdominal wall defect (see movie). Nipples were widely spaced, heart sounds seemed distant and located more on the right side, and there was no murmur.

The chest X-ray on admission showed cardiac dextroposition (Fig. 1). Echocardiography revealed partial abdominal herniation of the heart with a diverticulum of the left ventricle measuring almost 3 cm and contracting synchronously with the left ventricle. No other cardiac anomalies were found. The ECG was normal. A thoracoabdominal CT scan was performed and showed a bifid lower part of the sternum, a small omphalocele, a ventral diaphragmatic hernia with presence of intestines and the absence of the apical part of the pericardial membrane. The entire heart seemed to be in a dextroposition. No other abnor-

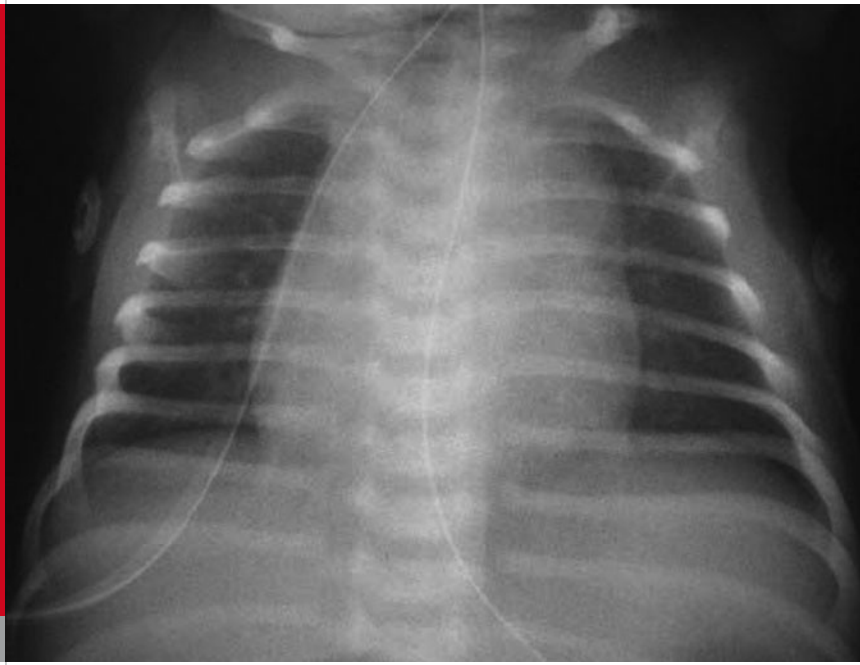

Fig. 1

Chest X-ray showing cardiac dextroposition.

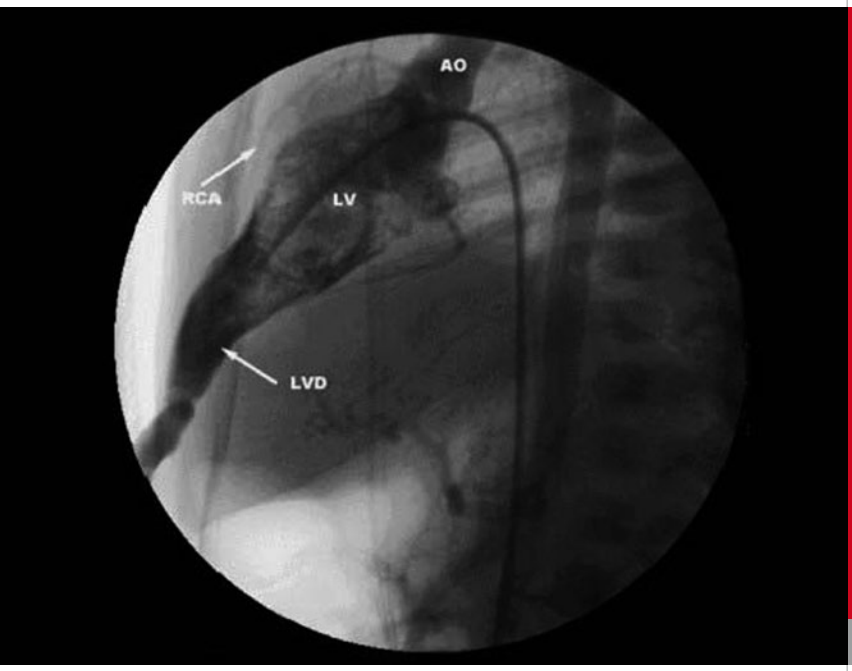

Fig. 2

Cardiac catheterization: left ventricular diverticulum (LVD); left ventricle (LV); aorta (AO); right coronary artery (RCA).

malities were encountered. These findings suggested the diagnosis of Cantrell's pentalogy. Abdominal and cerebral ultrasounds were normal and caryotyping revealed no chromosomal anomaly.

Continuous enteral alimentation was introduced to prevent intestinal incarceration into the diaphragmatic hernia and surgical resection of the left ventricular diverticulum was considered. Preoperative cardiac catheterization and angiography of the left ventricle showed the diverticulum and the coronary perfusion (Fig. 2).To prevent incarceration of the herniated bowel resection of the diverticulum and correction of the abdominal wall with closure of the diaphragm was performed.

The patient had an uneventful postoperative course and was discharged at four weeks of age on diuretics, aspirin and endocarditis prophylaxis. On follow-up at 3 and 6 months of age the patient was thriving with no signs of heart failure or arrhythmia. ECG was normal. Diuretics and aspirin were stopped at 6 months of age.

DISCUSSION

Ectopia cordis is a rare congenital malformation, occurring in 5.5 to 7.9 per million live births (1). It is characterized by complete or partial displacement of the heart outside the thoracic cavity. The association of anterior diaphragmatic hernia, omphalocele, congenital heart disease, sternal and pericardial defects was

first described by Cantrell and colleagues in 1958 (2). The type of ectopia cordis described in Cantrell's pentalogy syndrome is thoracoabdominal and is of better prognosis than other forms of ectopia cordis (cervical, cervicothoracic).

The etiology is unknown but seems to be caused by the failure of maturation of the midline mesodermal components of the chest and abdomen, occurring during the third week of gestation. This causes a defect of the anterior part of the diaphragm that leads to a displacement of the heart outside the thoracic cavity and prevents proper midline fusion of the developing chest wall (bifid sternum, widely spaced nipples) (3,4).

The survival rate of 50% or better is influenced by the presence or absence of other associated congenital anomalies, the type of heart defect and the age at surgery (mortality rate increases when surgery is done on the first day of life) (5). Common congenital cardiac anomalies associated with ectopia cordis are: VSD (almost 100%, but not in our case), ASD (53%), TOF (20%) and left ventricular diverticulum (20%). Severe forms of Cantrell's pentalogy include pulmonary hypoplasia, a large omphalocele, herniation of the liver or the bowel into the chest and hydrocephalus (5). Abdominal wall defect can be diagnosed prenatally as early as 14 weeks of gestation (6,7).

Severe cases of Cantrell's pentalogy need two-stage repair. Mild cases (as our patient) undergo a single-stage surgical intervention during the neonatal period and have a good prognosis. Cardiologic follow-up consists of echocardiography and ECG at 1, 3 and 6 months of life to exclude postoperative ventricular arrhythmia and heart failure. Thereafter, patients are seen at yearly intervals.

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