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Pentalogy of Cantrell: About a case report at the General Reference Hospital of Panzi

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ABSTRACT: Pentalogy of Cantrell is an extremely rare birth defect. It associates 5 malformations including: a supraumbilical hernia of the abdominal wall, a defect of the anterior part of the diaphragm and the diaphragmatic pericardium, an anomaly of the lower part of the sternum and cardiac malformations.
We most often find a notion of consanguinity of the parents. The long-term prognosis for children with this anomaly depends mainly on the complexity of the associated heart defect.
We report a case of pentalogy of Cantrell in a 2-day-old male newborn with extra thoracic cardiac ectopia.

KEYWORDS: pentalogy, Cantrell, cardiac ectopia, Panzi, hospital.

1 INTRODUCTION

Pentalogy of Cantrell is an extremely rare congenital anomaly at 5.5 to 7.9 per million live births (1). It corresponds to a defect in the closure of the umbilical ring concerning the supra-umbilical portion of the abdominal wall secondary to an anomaly in the development of the mesoderm during the first stage of embryonic development. This pathology associates five major malformations: a supraumbilical hernia of the abdominal wall, a defect of the anterior part of the diaphragm and the diaphragmatic pericardium, an anomaly of the lower part of the sternum and cardiac malformations (2). The appearance of the Pentalogy is considered sporadic. We report a case of Pentalogy of Cantrell in a newborn male, referred to us by the general referral hospital of Kaziba, a hospital located 60 km from the general referral hospital of Panzi.

2 CLINICAL OBSERVATION

It was a newborn male, the last of three siblings, with no family history of congenital malformations or consanguinity. The mother was 25 years old. The pregnancy was not followed medically and therefore, no antenatal ultrasound was performed by the mother during the period of evolution of the pregnancy. He was born vaginally after 38 weeks of amenorrhea and sent to the neonatology department of Panzi General Hospital on the 2nd day of life.

On physical examination, the newborn weighed 3100 g, he had a head circumference of 35 cm, a height of 49 cm. He was feverish, temperature 38°C. He had the heart completely out of the thoracic cavity and beating, an Omphalocele as well as the absence of the lower part of the sternum (Fig. 1).

The patient was put under medical treatment for neonatal infection. Surgical management was rejected because of the lack of an adequate technical platform. He had died at 17 days of life in a row due to the absence of a cardiovascular surgeon in our region.

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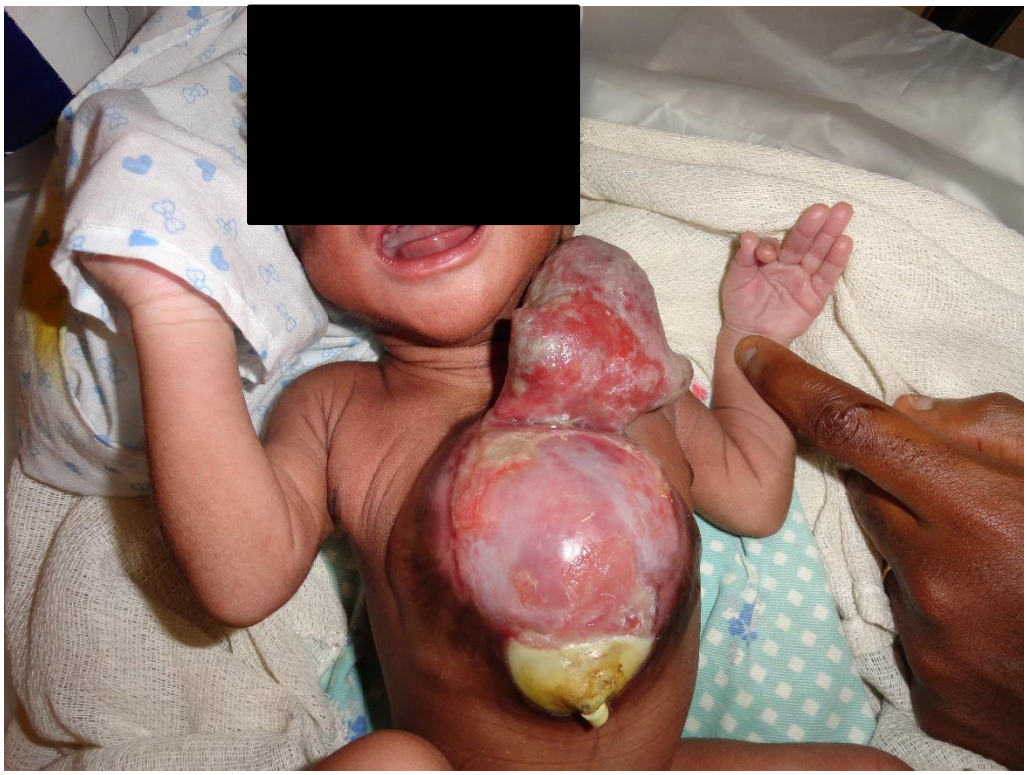

Fig. 1. Extra thoracic cardiac ectopia

3 DISCUSSION

Cantrell's pentalogy is an extremely rare birth defect, first described by Cantrell in 1958 (2). The estimated incidence of this rare syndrome is approximately 5.5 per million live births (1). This pathology is said to be complete if it associates the five major abnormalities: a supraumbilical hernia of the abdominal wall, a defect in the anterior part of the diaphragm and the diaphragmatic pericardium, an anomaly in the lower part of the sternum and cardiac malformations (3). Incomplete forms of the syndrome with a combination of at least two defects have been reported in the literature (2). The cause of Pentalogy of Cantrell is not clearly understood and it is unclear whether it represents an extreme spectrum of midline malformations, as it shares some characteristics with a midline defect and coexists with other malformations from the midline (4). In addition to the classic anomalies described by Cantrell et al, a few cases have been reported with the coexistence of other syndromes such as Edwards and Goltz-Gorlin syndrome. Structural abnormalities including craniofacial (eg cleft palate, supernumerary nostrils), central nervous system (hydrocephalus and neural tube defects), skeletal and abdominal abnormalities have been reported (2). There is a male predominance with a male/female ratio of 2.7/1. The most common form is the thoracic or thoracoabdominal type. Abdominal wall abnormalities include omphalocele, diastasis recti, umbilical hernia, or a combination of these abnormalities. The most common form is that with omphalocele (3). Diagnosis can be made antenatally with prenatal ultrasound during the first trimester of pregnancy (5,6). Prenatal magnetic resonance imaging (MRI) allows visualization of fetal abnormalities and helps in planning surgical management (7). The survival rate of the full Pentalogy of Cantrell form is less than 20% (8).

4 CONCLUSION

Pentalogy of Cantrell is a rare pathology. The diagnosis is possible thanks to the ultrasound of the first trimester of pregnancy. Its prognosis is linked to the severity of the heart defect. The treatment is multidisciplinary.

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