

Pentalogy of Cantrell: A Report of One Case at the Yaounde Central Hospital in Cameroon

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ABSTRACT

Pentalogy of Cantrell is a rare multiple congenital malformation syndrome characterized by a combination of five features: A midline supraumbilical abdominal wall defect; a defect of the lower sternum; a defect of the diaphragmatic pericardium; deficiency of the anterior diaphragm and congenital cardiac anomalies. These defects can be diagnosed as early as the first trimester of pregnancy. The complexity of these anomalies, in particular the presence of any cardiac defects, determines the management as well as the prognosis. We report a case of pentalogy of Cantrell diagnosed by ultrasound at 32 weeks of gestational age, the fetus died 3 hours after delivery.

Keywords: Pentalogy of Cantrell, Ultrasound, Management.

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INTRODUCTION

Pentalogy of Cantrell is a rare and sporadic syndrome which associates five classic congenital malformations: Involving the abdominal wall, sternum, diaphragm, pericardium and heart.¹ This syndrome is very little described in African literature, particularly in Cameroon. The first case reported in Cameroon was in 2010 by Ngaha and collaborators² at the regional hospital of Bafoussam. We report a case of Cantrell's pentalogy detected by ultrasound at 32 weeks of gestation and confirmed at delivery.

CASE REPORT

A 20-year-old single primigravida was seen for her first antenatal visit in our outpatient clinic at 32 weeks gestation. The patient did not report ever being unwell or taking any medications early in the pregnancy. Despite the fact that the pregnancy was not desired, there was no attempt to terminate it. No other significant medical or family history was found.

On physical examination the general state was good. The patient was conscious. Her conjunctivae were well-colored with white sclera, and all the cardiorespiratory parameters were observed as normal (blood pressure was 100/58 mm Hg, the heart rate was 96 beats per minute and the respiratory rate was 20 breaths per minute). The



Fig. 1: Neonate at 32 weeks and 2 days with thoracolaparoschisis and exposition of thoracic and abdominal contents

cardiopulmonary examination was normal. The abdomen was distended with the fundal height at 27 cm, and fetal heart sounds were present at 138 beats per minute.

On vaginal examination, the cervix was posterior, soft, long and closed.

The routine ultrasound examination performed that day, reported a singleton pregnancy at 32 weeks, a live fetus with thoracic laparoschisis. The heart, lungs, liver, spleen and intestines were floating in the amniotic fluid. The β -hCG level and α -fetoprotein level requested were not done. At 32 weeks plus 2 days of gestation, labor spontaneously commenced and a live male infant weighing 1800 grams with thoracic laparoschisis (Fig. 1) as described in ultrasonography was delivered. The Apgar score was 3 over 10 at 1 minute and 1 over 10 at 5 minutes.

Resuscitation of the baby was not attempted because of financial restraints of the patient and his family. The baby died 3 hours after birth. Due to unavailability we did not conduct any genetic tests.

DISCUSSION

Pentalogy of Cantrell was first described in 1958 by Cantrell et al who named it so because of the presence of five major malformations. These malformations include a midline upper abdominal wall abnormality; lower sternal defect;

anterior diaphragmatic defect; diaphragmatic pericardial defect and congenital abnormalities of the heart.¹ The prevalence of pentalogy of Cantrell has been estimated at one in 65,000 to 5.5 in a million live births.³ This case was the first one since 5 years in our maternity corresponding to one in 18,000 deliveries.

The pathogenesis of pentalogy of Cantrell is unclear. The defects result from lack of fusion in the medial line of the mesoderm responsible for the formation of the thoracoabdominal wall. The heart protrudes through a sternal defect, which can be a cleft sternum or absent lower third of the sternum, producing ectopia cordis. The intestinal loops and liver protrude through the abdominal defect (involving the umbilical cord) and, create the spectrum of abnormalities found in the pentalogy of Cantrell.⁴ In 1993, a possible association was described in familial cases of a gene linked to the X chromosome in the Xq25-26.1 region.⁵ More recently, a case of pentalogy of Cantrell associated with administration of the labor-inducing drug, misoprostol was published, where the authors proposed that the failure of fusion may be secondary to vascular disruption.³ In our case, we did not find any family history of malformation.

The main abdominal wall malformation associated with this syndrome is omphalocele, found in 74.5% of patients, followed by deformed regions in the lower sternum (59.4%), diaphragm (56.8%) and pericardium (41.8%). Cardiac anomalies occur in 83% of cases, of which atrioventricular or huge ventricular septal defects are the most common malformation.⁶ In 1972, Toyama suggested a further classification of the syndrome: Class 1, definite diagnosis with all 5 defects present; class 2, probable diagnosis with 4 defects noted (including intracardiac and ventral abdominal wall abnormalities); and class 3, incomplete expression.⁷ In the case presented, we observed four evident anomalies of the pentalogy of Cantrell. Cardiac ultrasound was not done to detect abnormalities of the heart but upon physical examination the heart appeared abnormal. We think that our case was class 1. We did not find an association of a bilateral deep cleft lip/palate and left side clubfoot which were reported in others case reports.⁸

Ultrasound diagnosis is possible during the first trimester, when malformations such as ectopia cordis and ventral abdominal defects are visible. When small defects occur in the pericardium, diaphragm or in the inferior segment of the sternum without evident herniation of the heart, diagnosis is much more difficult.³

Three-dimensional imaging of cardiac, thoracic and abdominal malformations by ultrasound, magnetic resonance imaging and helical computed tomography angiography is useful for diagnosis and for guiding surgical decisions in patients with pentalogy of Cantrell.⁹ Our patient

started antenatal care late and the ultrasound diagnosis was made at 32 weeks. We did not have time to discuss with the neonatal unit about the management of the neonate before she spontaneously went into labor.

The management consists of corrective or palliative cardiovascular surgery, correction of the ventral hernia and diaphragmatic defects and correction of associated anomalies.³ We provided baby with comfort care without attempt of any active intensive care and surgical treatment because of unavailability of resources to treat that kind of severe congenital malformations, patient's financial difficulties and lack of social insurance policy. Besides it is ethically plausible not to spend limited financial resources on futile treatment of the neonate with poor prognosis while we have high mortality of near term and term babies without severe congenital malformations.

Concerning the prognosis, the pentalogy of Cantrell is associated with a high rate of mortality. In a 2008 study by Van Hoorn et al¹⁰ 37 of 58 patients (that's 63.8%) died within days after birth. The long-term prognosis for children with this anomaly depends to a great extent on the complexity of the associated congenital heart defect and the possibility of corrective surgery.^{11,12} The mean survival rate without any interventional surgery is about 36 hours.¹³ Studies show that even with careful monitoring in capable tertiary centers and multiple corrective surgeries, these patients have high morbidity and mortality rates and poor long-term prognosis.¹³ In our case, the severity of these anomalies resulted in neonatal death 3 hours after delivery.

CONCLUSION

Prenatal routine obstetric ultrasonography may help screen for pentalogy of Cantrell at an early stage. Thereafter, individual cases must be adequately evaluated antenatally for appropriate prenatal counseling and postnatal management when appropriate.

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