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Early diagnosis and management of congenital heart diseases with a focus on transposition of the great arteries: case report

Diagnóstico e manejo precoces das cardiopatias congênitas com enfoque na transposição das grandes artérias: relato de caso

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Abstract

The transposition of the great arteries, characterized by exchange of the respective ventricles of origin between the aorta and pulmonary artery trunk, is responsible for approximately 5.7% of critical congenital heart diseases, with a high mortality rate in cases of late diagnosis and treatment. The objective of this study is to report a case of this heart disease, diagnosed by fetal echocardiography in the 27th week of gestation, to emphasize the importance of prenatal diagnosis for early management and reduction of the mortality rate of individuals with this heart disease.

Keywords: Transposition of the Great Arteries; Diagnosis; Case management.

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Resumo

A transposição das grandes artérias, caracterizada pela troca dos respectivos ventrículos de origem entre a artéria aorta e o tronco da artéria pulmonar, é responsável por aproximadamente 5,7% das cardiopatias congênitas críticas e apresenta uma alta taxa de mortalidade em caso de diagnóstico e tratamento tardios. Portanto, o objetivo deste estudo é relatar um caso dessa cardiopatia, diagnosticado por ecocardiografia fetal na 27ª semana de gestação, de forma a enfatizar a importância do diagnóstico pré-natal para o manejo precoce e redução da taxa de mortalidade de indivíduos com esta cardiopatia.

Palavras-chave: Transposição das grandes artérias; Diagnóstico; Manejo do caso

INTRODUCTION

Critical congenital heart diseases (CCHD) are the main trigger of acute heart failure in newborns, with a high mortality rate[1]. The transposition of the great arteries (TGA) is responsible for 5.7% of CCHD, and it is usually characterized by the exchange of the respective ventricles of origin between the aorta and the pulmonary artery trunk[1,2].

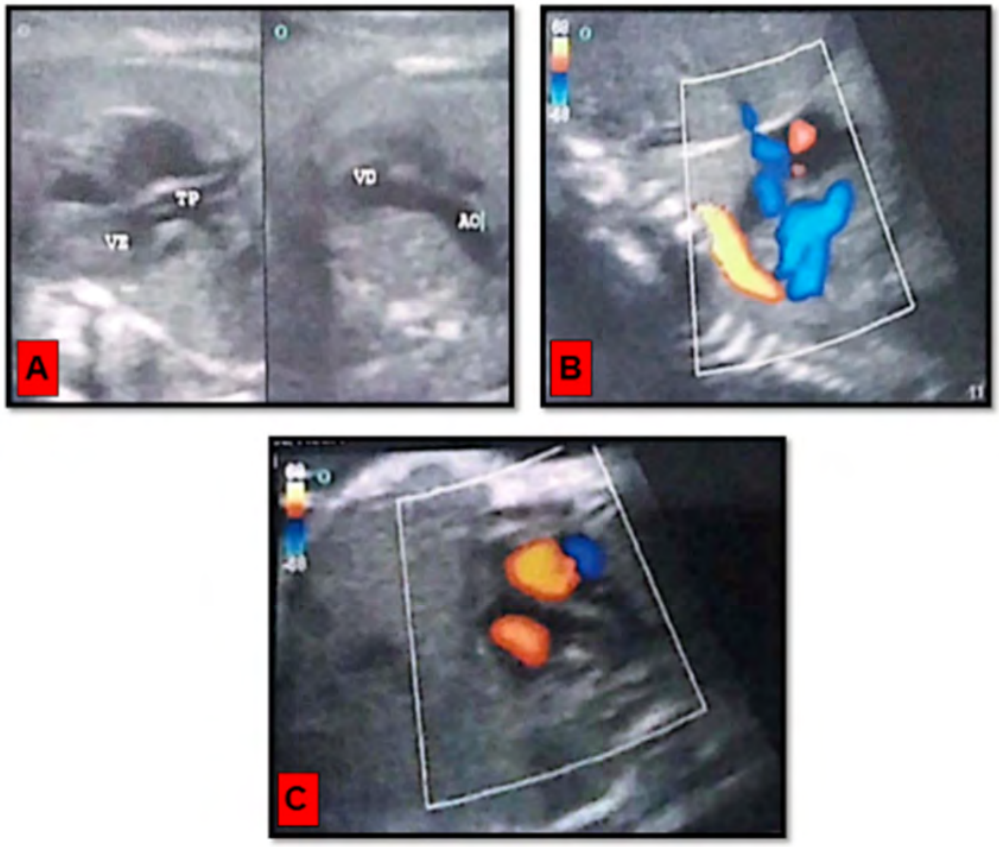
The management of these patients is complex and involves, among other procedures, monitoring, oxygenation, use of prostaglandins, septostomy, and surgery for arterial replacement[2-4]. Due to the severe clinical condition of TGA, early diagnosis (especially in the prenatal period) and management in the first year of life are essential for a better prognosis[2].

Despite advances, early TGA diagnosis remains suboptimal[5]. This study aimed to present the case of a patient with TGA diagnosed in the prenatal period, emphasizing the relevance of early diagnosis and management for a favorable prognosis.

CASE REPORT

A 27-year-old pregnant woman presented at the 27th week of pregnancy for fetal echocardiography (ECHO), and TGA and right-to-left shunt through the foramen ovale and ductus arteriosus were diagnosed (Figure 1). After two months of fetal ECHO, a preterm male was born (weight 3,062 g) with an Apgar score of 8 and 9 in the first and fifth minutes, respectively.

Figure 1. Echocardiographic images of the transposition of the great arteries and the right-to-left shunt through the foramen ovale and the ductus arteriosus.



A. exchange of the ventricular origin between the aorta artery (AO) and the pulmonary artery trunk (TP), respectively, for the right (VD) and left (VE) ventricles; **B and C.** Right-to-left shunt through the patent foramen ovale and patent ductus arteriosus. Images obtained from the exams provided by the mother of the patient.

At six hours of life, the newborn presented with a pulse oximetry of 60% and central cyanosis, and proceeded to intubation in the neonatal intensive care unit. The ECHO showed increased valvular heart disease and dilation of the right cavities. In the hemodynamics room, balloon atrial septostomy (Rashkind procedure) guided by ultrasound demonstrated enlargement of the interatrial communication and moderate tricuspid regurgitation.

At nine hours of life, during the Rashkind procedure, the newborn developed hypotension refractory to two intravenous infusions of 0.9% saline solution (10 ml/kg) and dopamine (5 mg/kg/min). Returning from the hemodynamics room, the newborn was still intubated, on positive pressure ventilation, hypotensive, under prostaglandins, and presented with a skin rash in the chest and neck and edema in the lower limbs that progressed to the facial and cervical regions.

At 12 hours of age, the newborn remained with refractory hypotension (blood pressure of 44 x 26 mmHg), persistent hypoxia, and tachypnea, and antibiotic therapy with ampicillin and

gentamicin was started. On the fourth day of life, the patient developed eyelid edema, lactic acidosis, and coagulopathy, with the occurrence of bleeding and anemia; thus, blood products were prescribed. Despite improvements in coagulation and acid-base disorders, an arterial switch operation (ASO)[3], associated with tricuspid plasty and closure of pre-existing communications (cardiopulmonary bypass), was performed on the fifth day of life due to the severity of the condition; no intercurrences were observed.

In the immediate postoperative period, a Tenckhoff catheter was installed to start peritoneal dialysis due to progressive elevation of urea and creatinine and diagnosis of acute kidney injury. On the first postoperative day (PD), the newborn was extubated but developed hemodynamic instability. On the second PD, purulent secretion was released through the Tenckhoff catheter, given the refractoriness to clinical treatment, and antibiotic therapy was adjusted for piperacillin and tazobactam. On the third PD, an abdominal wall edema with progressive worsening appeared, while on the eighth PD, the patient developed difficult-to-control anasarca associated with hyponatremia.

The newborn presented with septic shock, with episodes of hypotension responsive to volume expansion and hypoxia refractory to noninvasive ventilation, requiring intubation. The newborn also remained on peritoneal dialysis due to the persistence of anasarca.

In the first month of life, the patient developed cardiopulmonary arrest due to asystole and responded to cardiopulmonary resuscitation measures. Five days later, necrosis appeared in the right thumb and progressed to all limbs. In the following two days, the newborn evolved into a new cardiopulmonary arrest due to asystole, which was irreversible to resuscitation measures for 10 minutes. His death was confirmed after this period.

DISCUSSION

Despite recent advances in treatment, patients with CCHD have an estimated mortality rate between 15% and 25% in the first year of life[6], with TGA being one of the most lethal in the neonatal period[2]. These data corroborate the outcome of this case since the newborn died at one month and six days of life.

Although the prenatal detection rates of TGA are suboptimal (25% to 40%), an early diagnosis with fetal ECHO associated with Doppler contributes to early treatment and allows a better prognosis[1,7]. However, in this case, even with early detection of TGA, the patient evolved with a fatal outcome due to septic shock, a possible complication caused by the clinical severity and complexity of the procedures performed.

Domínguez-Manzano *et al.*[5] evaluated the impacts of prenatal diagnosis of TGA on early treatment and prognosis of 154 patients diagnosed with this CCHD prenatally (n = 88) and postnatally (n = 66) between 1998 and 2014. The authors demonstrated that the management should

be initiated within the first 48 hours postnatally, based on prenatal confirmation or early neonatal suspicion[5], to reduce the mortality rate in this group. For example, perioperative monitoring, cardiovascular examinations, and follow-up of serum lactate, tissue perfusion, and urine output stand out[3]. However, in cases of other heart diseases or association with clinical outcomes resulting from complex procedures, as observed in this case, early management may not be sufficient.

In cases of restrictive atrial septal defect associated with TGA, the Rashkind procedure should be performed within 24 hours postnatally and before the definitive treatment[7], as performed at nine hours of life in the present case. The treatment choice for TGA is ASO, whose ideal window for performance is up to three weeks of life[3,4,7], in line with its occurrence on the fifth day of life of the newborn in this case. Although ASO was performed within the period described in the literature, the timing of surgical correction did not influence the complications of the patient in this case report.

Cardiopulmonary bypass performed in ASO is considered the main cause of acute kidney injury in newborns, especially four days after surgery, and may increase mortality, making monitoring necessary and, in case of high risk, early initiation of peritoneal dialysis[7], as in the present case.

Even with advances in ASO, the perioperative mortality at 90 days from the time of the procedure remains higher than 5%[8]. The causes of death in the postoperative period of this procedure are multifactorial and commonly associated with cardiac, brain, respiratory, hepatic, splenic lesions, multiple organ failure, and sepsis[8]. Also, some of the risk factors for death in this period are low surgical weight (< 2.5 kg), aortic clamping time, episodes of acute heart failure, and use of cardiopulmonary bypass[8].

Further advances in the multidisciplinary management of patients with TGA, particularly in the postoperative period, are needed since the early detection of this congenital heart disease may not be sufficient. This will contribute to the prevention or attenuation of complications related to TGA and its definitive surgical treatment (i.e., ASO); therefore, improving the prognosis and reducing mortality.

CONFLICTS OF INTEREST

The authors declare no conflicts of interest.

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AUTHOR CONTRIBUTIONS

TJMBSV: Conceptualization, Data curation, Investigation, Methodology, Project administration, Resources, Writing – original draft, Supervision, Writing – review and editing. **IFGG:** Writing – original draft, Writing – review and editing. **EBC:** Investigation, Writing – original draft, Writing – review and editing. **AVF:** Investigation, Writing – original draft, Writing – review and editing. **FAP:** Conceptualization, Data curation, Investigation, Methodology, Project administration, Resources, Writing – original draft, Supervision, Writing – review and editing. **GPPMN:** Conceptualization, Data curation, Investigation, Methodology, Project administration, Resources, Writing – original draft, Supervision, Writing – review and editing. All authors read and agreed with the final version of the manuscript.

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