Early Neonatal Cyanosis as a Presentation of a Rare Cardiac Anomaly: Truncus Arteriosus Type IV

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Keywords

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Abstract

Truncus arteriosus (TA) is a rare congenital heart anomaly presenting with mild cyanosis and congestive heart failure. It occurs when the blood vessels coming out of the heart in the developing baby fail to separate completely during development, leaving a common trunk responsible for the pulmonary and systemic perfusion. There are several variants of TA, depending on the specific anatomy and arterial connection. We report a case of a full-term newborn who developed cyanosis and desaturation during the first day of life. Investigations and echocardiography were consistent with TA type IV. Prostaglandin infusion was immediately started, and then, a successful palliative right modified Blalock Taussig shunt was performed.

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Introduction

Truncus arteriosus (TA) is characterized by a single arterial trunk arising from the normally formed ventricles by means of a single truncal valve and ventricular septal defect (VSD). Systemic venous blood and pulmonary venous blood mix at the VSD level, and the resulting desaturated blood is ejected into the single outlet [1]. Mixed venous blood that exits through unified outlet leads to increased pulmonary blood flow and desaturated systemic flow. Hence, TA manifests as congestive heart failure and mild cyanosis early in the neonatal period or later depending on the specific anatomy and the severity of increased pulmonary blood flow [2]. Additionally, valvular insufficiency and stenosis could contribute to the clinical picture. Valvular regurgitation, as in 50% of patients, can worsen congestive heart failure, whereas critical coarctation, seen in 10% of patients, can lead to cardiovascular collapse and shock or even death [3]. We describe a case of a neonate with a rare type of TA who presented with early severe cyanosis and murmur rather than congestive heart failure due to associated pulmonary atresia and restricted pulmonary blood flow.

Fig. 1. CXR showing decreased pulmonary blood flow with an enlarged cardiac silhouette. CXR, chest X-ray.

Case Report

A full-term male newborn was delivered by normal vaginal delivery, with no significant perinatal incidence. Parents were non-consanguineous, and they had no family history of congenital heart disease (CHD). The baby cried immediately after birth. He was pink and active with an Apgar score of 9 and 9 at 1 and 5 min, respectively. Vital signs and clinical examination were unremarkable with oxygen saturation reaching up to 96% in room air. However, gradually within the first 8 h after birth, the infant developed cyanosis with oxygen saturation dropped to 80% at 8 h of age. He had severe cyanosis with minimal respiratory distress and a grade 3 systolic murmur on the left sternal border. Cyanosis did not respond to oxygen well and oxygen saturation stayed at 85–90% even with high oxygen nasal prong. Chest X-ray showed decreased pulmonary blood flow with a mildly enlarged cardiac silhouette (shown in Fig. 1). On suspension of CHD, the patient was admitted to the neonatal intensive care unit, and prostaglandin was started.

Urgent echocardiography showed TA with a confluent origin of left and right pulmonary arteries supplied by a small patent ductus arteriosus (DA). Hence, prostaglandin infusion was carried on to preserve the DA flow.

Repeated detailed echocardiography after 2 days showed large bidirectional perimembranous subaortic VSD (8 mm), persistent foramen ovale (3.5 mm) with a left-to-right shunt, over-riding aorta (trunk) (50%) with a nonstenotic truncal valve (shown in Fig. 2), pulmonary atresia, persistent DA (4 mm) supplying confluent right and left pulmonary arteries, and significant collaterals (shown in Fig. 3). Findings were consistent with TA type IV.

Meanwhile, the patient was referred to a cardiac center where a right modified Blalock Taussig (RMBT) shunt was performed. Right pulmonary and innominate arteries were connected. Post-operative echocardiography showed a good flow through the shunt to the right pulmonary artery. Hence, oxygen saturation improved, and the patient maintained >85% saturation on room air. Consequently, the patient was discharged on anticoagulants and diuretics. However, the patient was scheduled for a Rastelli operation when he reaches >6 months old for a better outcome.

Fig. 2. Echocardiography showing a large VSD with over-riding aorta (trunk). VSD, ventricular septal defect.

Fig. 3. Doppler ultrasound showing collateral flow from the aorta.

Discussion

Early neonatal cyanosis and distress may herald critical pulmonary, cardiac, neurological, or metabolic disorders. Hence, prompt and early diagnosis of underlying pathology is imperative for appropriate and timely management of this condition [4]. Being one of the crucial causes of early cyanosis, CHDs are diagnosed in about 6 per 1,000 live births [5]. TA is a rare cardiac disorder representing just 0.7% of all CHDs and about 4% of all critical CHDs [6].

TA anomaly involves failure of complete separation of the aorta and pulmonary artery during fetus development. Consequently, they originate jointly from the ventricles and supply systemic, pulmonary, and coronary arteries with mixed oxygenation blood [7].

TA is classified through different systems including Collett and Edwards [8] and a revised classification offered by Van Praagh and Van Praagh [9]. The Collett classification depends on the origin of pulmonary right and left ar-

teries. It could originate from the trunk with a short main pulmonary artery (type 1), arise separately from the posterolateral aspect of the common arterial trunk (type 2), arise oppositely from sides of the trunk (type 3), or as in our case, with atretic pulmonary artery and flow from the DA (type 4) [8]. On the other side, Van Praagh and Van Praagh [9] included accompanying aortic malformations in his classification.

Typically, TA presents with distress in the first few days of life or early infancy with mild cyanosis and symptoms of congestive heart failure from increased pulmonary blood flow [2]. However, TA type IV, as in our patient, usually presents with cyanosis more than congestive heart failure as the pulmonary flow is restricted with the accompanying pulmonary atresia. TA type IV is considered a variant of pulmonary atresia with VSD or a tetralogy of Fallot with pulmonary atresia rather than a TA. It behaves the same way as pulmonary atresia with decreased pulmonary blood flow and cyanosis as the first presentation [10, 11].

Chest X-ray in type IV showed decreased pulmonary blood flow as the main pulmonary artery is atretic, and the flow depends on the DA and collateral connections. Conversely, typical TA without pulmonary atresia shows pulmonary hyperperfusion depending on the anatomy and physiology. Nevertheless, echocardiography is the diagnostic investigation of choice.

Management initially entails correction of any metabolic abnormality, cardiac stabilization, and noninvasive ventilation. Furthermore, in type IV, prostaglandin is of paramount importance to keep the DA open and to reserve the pulmonary circulation. In our presented case early prostaglandin was crucial to maintain acceptable oxygen saturation until RMBT shunt was performed.

Primary surgical repair during the neonatal period is now possible by closing the VSD and inserting a conduit between the right ventricle and pulmonary artery. This has led to improved survival in patients with TA. One-year postsurgical survival of TA is >80%, whereas the observed 1-year survival in uncorrected TA is approximately 15% [12]. Another option is to do initial palliative pulmonary arterial banding allowing the infant to grow larger. Complete repair is then deferred until late infancy. However, no clear advantage of this approach is seen in morbidity or mortality [13].

TA type IV is approached differently. Similar to pulmonary atresia, an initial shunt followed by Rastelli operation is the surgical approach with the best outcome [14]. After an initial RMBT shunt, our reported case improved dramatically with saturation reached 85%, and echo confirmed good flow in the right pulmonary artery.

Conclusion

Early neonatal cyanosis could indicate several pathologic causes. CHDs are crucial entities to consider and manage immediately. However, considering the specific anatomy and physiology of the CHD is essential to manage the patient appropriately and to get the required outcome.

Although TA is a rare CHD, it should be diagnosed as early as possible to get a good outcome. Notably, TA is amenable to corrective surgery or at least a palliative operation with a good outcome depending on the specific anomaly. However, infants with TA type IV should be managed akin to pulmonary atresia with initial shunt to increase the pulmonary flow followed by the palliative procedure at later age. Subsequently, TA requires meticulous cardiology follow-up and probably medical and surgical further intervention according to the clinical situation.

Statement of Ethics

After full explanation, written informed consent for publication of this case was obtained from the father of the patient for publication of the case report and the accompanied images. However, ethical approval was not required in accordance with the Dubai Scientific Research Ethics Committee policies.

Conflict of Interest Statement

The authors declare no conflict of interest. The case was managed by our team. We have done the full research and writing of the manuscript.

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Author Contributions

Dr. A.M.H. and Dr. R.M.A. were responsible for the management of the case. They wrote the case report and reviewed the discussion. Dr. A.M.H.S. reviewed the case report and wrote the discussion. The case was formulated by Dr. A.M.H.S. and then reviewed by the full team.

Data Availability Statement

All data generated or analyzed during this study are included in this article. Further enquiries can be directed to the corresponding author.

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