

Case Report: Truncus Arteriosus Late Diagnosis and Atypical Presentation

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Abstract

A 6 months-old male, referred to the emergency of a quaternary hospital, for a suspicion of Tetralogy of Fallot because of an excessive head sweating and central cyanosis when intense crying presented since birth. In the neonatal unit, the pulse oximetry test was normal, being discharged in his third day of life. Echocardiogram showed a truncus arteriosus with an atypical anatomy which restricted the pulmonary flow and delayed the clinical signs. It is frequently associated to DiGeorge syndrome and the patient in question showed phenotypical signs that could have led to an earlier diagnosis when reminded this association. Patient was submitted to surgery 15 days after the admission.

Keywords: Truncus arteriosus; Tetralogy of Fallot; DiGeorge syndrome; Congenital cardiopathy.

Introduction

Truncus arteriosus (TA) is a rare congenital cardiopathy, responsible for less than 1% of the congenital heart diseases, with an annual incidence of 7:100.000 live births. Usually, presents itself isolated, without other associated malformations, with a slightly increased frequency in boys [1.] A disturbance in the development of the truncoconal septum results in a common arterial trunk, which collectively rises to the aorta and the pulmonary arteries. Is usually associated with

a sub arterial VSD and agenesis of the ductus arteriosus in most of the cases [2,3]. In TA, there is as single semilunar valve with variable anatomy and with variable degrees of disfunction regarding its morphology and diameter [3]. There's a frequent genetic association with DiGeorge Syndrome [3].

There are two classifications for TA, developed by Collet-Edwards and Van Praagh and both take into consideration the origin of the pulmonary arteries (PA) [4]. Generally, the diagnosis is suspected right after birth

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when there's alteration in the pulse oximetry test in the neonatal unit. In other cases, the diagnosis should be suspected in the presence of a cardiac murmur and/or signs of congestive heart failure, usually present in the first few weeks of life.

Patients with TA have high mortality rates in the first year when not submitted to surgery [2,6] and a high tendency of having pulmonary hypertension, which makes fundamental its early surgical correction. This case reports the case of a baby with a TA late diagnosis, with a unique anatomic presentation and signs of alteration in the pulmonary circulation.

Case report

H.S.C., 6 months-old male, referred to the emergency of a quaternary hospital, for a suspicion of Tetralogy of Fallot because of an excessive head sweating and central cyanosis when intense crying presented since birth. In the neonatal unit, the pulse oximetry test was normal, being discharged in his third day of life. In the physical examination, showed a good condition, a good weight (6.9 kg/15 lbs) and a length of 61cm/2ft. Pulse oximetry was 85% in both upper and lower limbs, arterial pressure of 100x62 mmHg (between p95 and p95+12), HR 138 bpm, RR 50 brpm, besides a syndromic face suggestive of DiGeorge Syndrome. The cardiac exam showed no cardiac impulses, a symmetric chest; a regular heartbeat with systolic murmur +5/+6 in the lower left sternal border radiating to back and a hyperphonestic and constant unfolding of the second murmur. Respiratory exam was normal, apart from a discrete tachypnea.

Normal abdominal exam. Rest of physical exam without changes.

Echocardiogram showed: a short common arterial trunk, dividing itself in ascending aorta and pulmonary trunk (PT), with the PT originating the RPA. Trileaflet truncal valve with discrete regurgitation. 13 mm sub truncal VSD. Aortic arch to the right, with a suggestive image of the LPA originating from the ductus arteriosus with signs of stenosis. A Angio CT was performed and showed a LPA with important stenosis in its origin, receiving influx from the ductus which originated from the LCA. An additional finding was described- a retroesophageal left subclavian artery.

Patient was submitted to surgery 15 days after admission with closure of the ductus, disconnection of the LPA (which emerged from the ductus). Separation of the common arterial trunk, with mending of the aorta with bovine pericardium. After the ventriculostomy, a ventriculoseptoplasty was performed with bovine pericardium as well to then connect the RPA (serving as a RVO posterior wall); in the anterior portion, a number 10 monocuspid was anastomosed and at last, the LPA was connected to the RPA. A residual 4mm VSD was kept. In the surgery, no thymus and a right vicarious lung were observed. ECC time was 156 minutes and 125 minutes of anoxia. After surgery, patient went to the CICU using epinephrine and milrinone. Vasoactive drugs were suspended in the 5th post-op and extubated in the 7th. Patient was discharged in the 29 post-OP with medications for congestive heart failure and a close follow-up in the clinic.

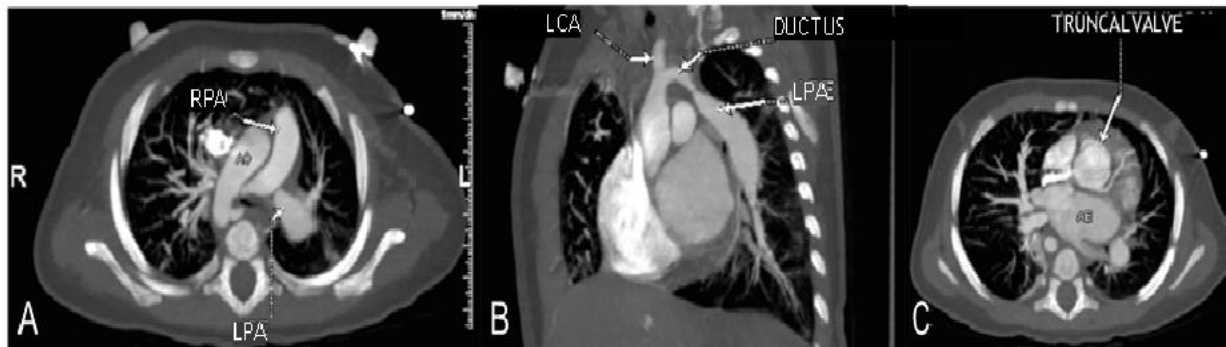


Figure 1: Angio-CT images. (a) RPA originated from the pulmonary trunk, disconnected from the LPA. (b) LCA emerging from the aortic arch, from which emerged the ductus arteriosus posteriorly the LPA (with important stenosis in its origin). (c) Truncal valve and dilated LA. APD=RPA: Right Pulmonary Artery; LCA: Left Carotid Artery; LPA: Left Pulmonary Artery.

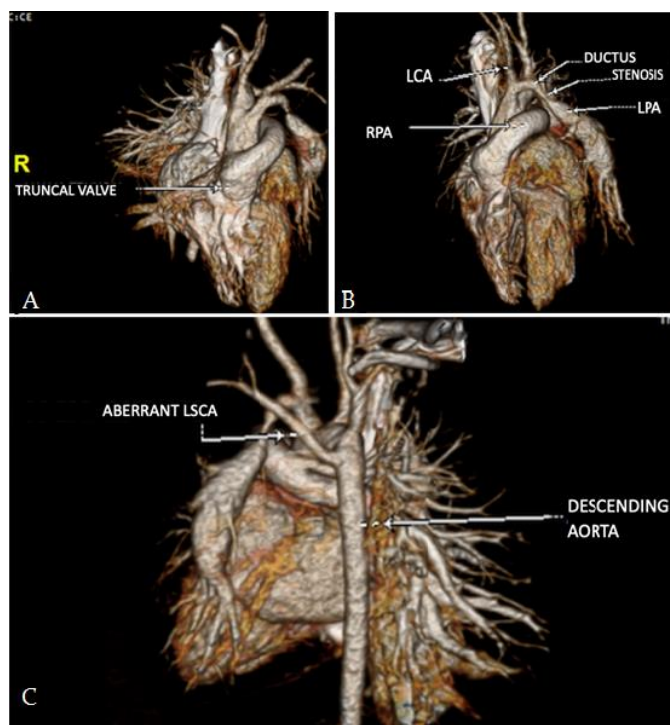


Figure 2: Tridimensional Angio-CT images. (a) Truncus bifurcating into ascending aorta and PT. (b) PT originating RPA. LCA emerging from the aortic arch and the ductus emerging from it and posteriorly the LPA (with important stenosis in its origin). (c) Posterior view of the aortic arch showing the anomalous origin of the LSCA. PT: Pulmonary Trunk; RPA: Right Pulmonary Artery; LCA: Left Carotid Artery; LPA: Left Pulmonary Artery; LSCA: Left Subclavian Artery.

Discussion

TA is a cyanotic congenital cardiopathy in which the pulmonary flow is determined by

the anatomy of the pulmonary arteries. In this case, the patient had a previous normal pulse oximetry test, delaying the cardiopathy

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diagnosis, which was only made at 6 months-old with evident signs of pulmonary hyper-flow.

The DiGeorge syndrome is diagnosed by the deletion of the 22q11.2 gene, described in 30% of the cases [3,7]. When suspected, a cardiac investigation should be performed. The patient described showed phenotypic characteristics from the syndrome, which might have contributed to an earlier diagnosis of the cardiopathy.

Other cardiac findings associated to TA include aortic arch to the right in 30% of the cases, interruption of the aortic arch in 13% and coronary anomalies [3,8]. Agenesis of the ductus arteriosus usually happens in 100% due to the communication of the trunk with the systemic and pulmonary circulations [3]. In this case, the aortic arch is to the right with the pulmonary arteries disconnected from one another and the LPA originated from the stenotic ductus-which is a very unusual

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presentation. It is possible that the anomalous origin and the stenosis of the LPA might have contributed to a lower pulmonary flow and less evident signs of congestive heart failure, which could have contributed to a late diagnosis and to advanced vascular changes in the pulmonary circulation.

Conclusion

TA is a rare cyanogenic congenital heart disease in which the pulmonary flow is determined by the anatomy of the pulmonary arteries. The surgical correction is indicated as soon as possible. The association with DiGeorge syndrome is frequent.

This case illustrated a patient with stigmas of the syndrome and a late diagnosis of TA, possibly due to the atypical anatomy and its degree of limitation to the pulmonary flow, therefore contributing to a less evident clinical.