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Case Report

The Berry Syndrome: A Cardiovascular Malformation: A Case Report

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Abstract

Although echocardiography remains the primary imaging tool when suspecting a congenital cardiac anomaly, it comes short in terms of accurately specifying the type of abnormality in the domain of cardiovascular malformations in which a well-studied dimensional topography is the basis of a successful surgical approach assuming the latter is possible. Ultrasound limitation is principally due to a limited acoustic window and the inexperience of imagers. In this article, we report a unique combination of 2 malformations discovered in the same patient group in the so-called 'Berry syndrome', its US and CT feature with contemporary surgical methods as well as the outcome of the patient in hand.

Keywords: *Berry Syndrome; Interrupted Aortic Arch; Aortopulmonary Window; CT Angiography; 3D Reconstruction*

Abbreviations

US: Ultrasound; CT: Computed Tomography; ER: Emergency Room; APW: Aortopulmonary Window; IAA: Interrupted Aortic Arch; CBP: Cardiopulmonary Bypass

Introduction

Berry syndrome is a unique neonatal cardiac association of anomalies: a distal aortopulmonary window (APW), the aortic origin of the right pulmonary artery (RPA), an intact ventricular septum, and an interrupted aortic arch (IIA) with a patent ductus arteriosus (PDA) [1,2]. In our case, the aorta is directly connected to the pulmonary trunk as they subdivide into left and right (Figure 1), therefore we classified the case as an incomplete berry syndrome or a pure association between an APW and an IIA type A.

Case Report

A 3-month-old male presented to the ER with fatigue, rapid breathing, and a pale skin tone. The patient had previously consulted with a cardiologist at the demand of the family doctor suspecting a heart issue due to the unclear cause of prolonged pallor. After performing a Doppler ultrasound, the cardiologist suspected an aortic interruption in addition to serious pulmonary arterial hypertension and referred it to our radiological department to further specify the anomaly with a CT angiography (Figure 2). A rare association of APW and IIA type

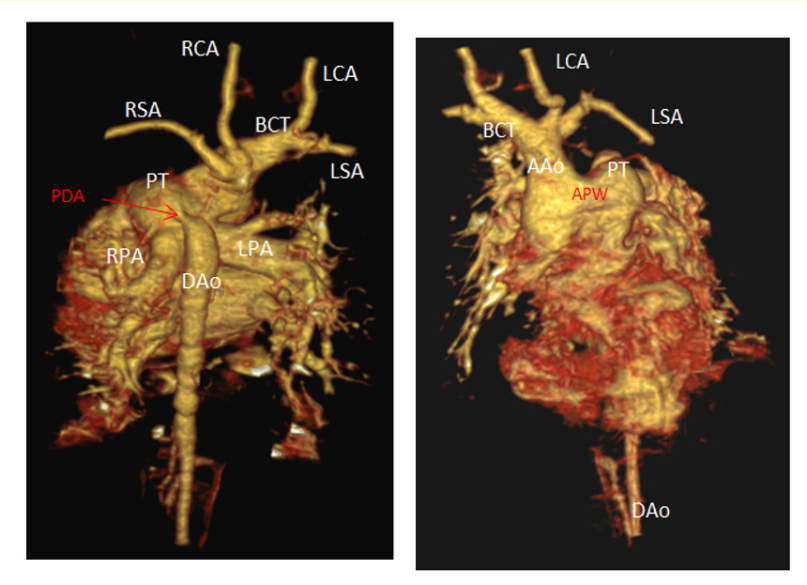

Figure 1: *Three dimensional reconstruction showing the topography of the PDA and visualize the APW type III and IIA type A with normal emergence of the pulmonary arteries.*

AAo: Ascending Aorta; DAo: Descending Aorta; APW: Aortopulmunary Window; PT: Pulmonary Trunk; BCT: Brachiocephalic Trunk; LCA: Left Carotid Artery; RCA: Right Carotid Artery; RSA: Right Subclavian Artery; LSA: Left Subclavian Artery; PDA: Patent Ductus Arteriosus.

A was diagnosed which may be a variant from the Berry syndrome described in the late 20s since the ventricular septum was intact, the ductus arteriosus was present and the aorta was connected to the main pulmonary artery.

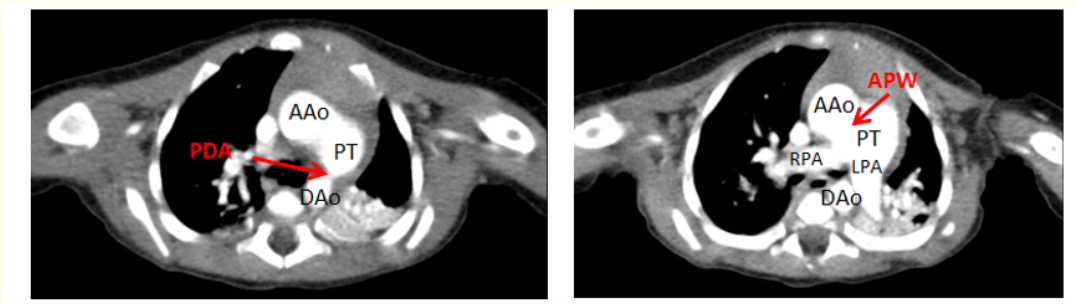

Figure 2: *Axial contrast enhanced CT demonstrating the associative anomalies in 2D: APW, PDA in addition to IIA diagnosed by the following of the aortic tract on the console.*

The infant installed breathing difficulties shortly after the CT was performed and was immediately rushed to another facility where a specialized surgical consult confirmed the urgency of immediate intervention. Vitals showed a heart rate of 150 bpm, oxygen saturation at 82% in room air, and respiratory rate at 42 breaths per minute before the surgery.

Two main reconstructions were made: first; a reconstruction of the aortic arch and second the closing of the aortopulmonary window using a pericardial patch. The subsequent hospitalization was uneventful, and the patient was discharged in good clinical condition on postoperative day 21. One year later, follow-up echocardiography showed satisfactory growth and the patient remained asymptomatic.

Discussion

Berry syndrome is an extremely rare disease, accounting for only 0.046% of all reported cases of congenital heart anomalies. The syndrome consists usually of a distal aortopulmonary (AP) window in 79% of cases, aortic origin of the right pulmonary artery (RPA) 96% of cases, an intact ventricular septum in 99% of cases, and an interrupted aortic arch in 85% with a patent ductus arteriosus in 34% of cases [1].

Since first described by Berry., *et al*. in 1982, among the nearly 100 patients that has been reported in the English literature so far [1], only 4 fall statistically in the case in which the primitive arterial trunk posteriorly did not disturb the normal flow of embryogenesis and consequently avoiding the 'straddling' of the pulmonary bifurcation to this undivided truncal segment rather than to the main pulmonary arterial trunk [2]. The right pulmonary artery (RPA), thus instead of relating to the aorta and the left pulmonary artery (LPA) to the pulmonary trunk, as we witness in the usual berry syndrome, the normal embryological course of events took place despite the persistence of the failure of the partitioning of the truncus arteriosus and the aortic sac due to conotruncal ridge mal-functioning [3]. As a result, the APW came through but with a normal origin of the right pulmonary artery (Figure 3).

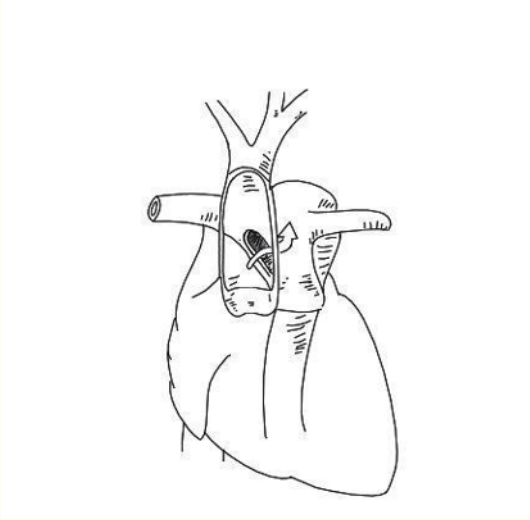

Figure 3: *Diagram simplifying the defects encountered in our case.*

According to a review of the literature between 1981 and 2011, combined APW/IAA was found in 8 cases with one case of a ventricular septum defect (VSD) in addition to the combination [4]. Moreover, a PubMed-based search was conducted in the literature for cases as-

sociating APW and IAA not under the berry syndrome diagnostic umbrella. The keywords included 'interruption of the aortic arch with aortopulmonary window 'interruption of the aortic', 'aortopulmonary window'. Only two cases were published with this same particular set associations: IIA with APW and that includes in 2013 and 2015 [5,6]. All things considered, cases similar to ours, setting aside the nomenclature, are estimated, to our best knowledge to be around 13 cases as a higher estimate.

We prefer preserving the nomenclature 'berry syndrome' as we ought to think of it as a spectrum pf defects with some major ones and the association creates the variants. Furthermore, during the infant's follow-up visit, we recommended an aCGH analysis to the patient's family which later came back abnormal (aCGH abnormalities are linked to a deletion in the chromosome band 9p24.2). Consequently, this report accompanied with one previously made in 2013, showed a link has been made between Berry syndrome and deletion within chromosome band 9p24.2 through various hypotheses [7]. Although there is little evidence to support such claims, reports thus far show enough ground for future genetic research into the legitimacy of such links.

In order to diagnose such complex structural aberrances of the great cardiac vessels, an echography is typically the primary diagnostic imaging modality (Figure 4). However, its limitations may include an inadequate acoustic window, the inclusion of one or multiple associated malformations, PDA-induced distortion of the distal aortic arch, and insufficient expertise of the imager. In view of these difficulties, CT or MR imaging with reconstruction can be referred to an Echo in order to accurately recognize the type, and topography and simplify the conceptualization of complex and cumulative anomalies (Figure 1). A step further would be the recent prowess of 3D printing that have been developed to optimize surgical outcome considerably, especially in rare defects. 3D printing has been utilized to tangibly delineate the exact cardiac anatomy of a neonate with an aortopulmonary window associated with an interrupted aortic arch in order to familiarize the surgeon with the 3D real-time volumetric of the heart and its vascular malformations [8].

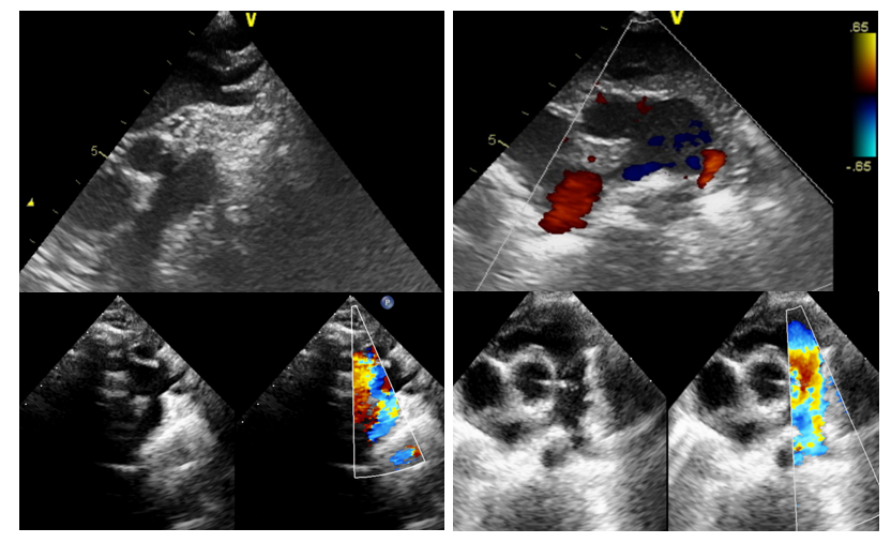

Figure 4: *Preoperative and postoperative cardiac doppler ultrasound demonstrating the repair in terms of anatomy and hemodynamically reflected by the elevated pulmonary hypertension previously caused by the APW.*

In terms of the surgical approach, Jacobson., *et al*. reported the first successful one-stage repair in infancy which later showed favorable results compared to staged approaches [9]. Similarly, in our case, we used a one-stage surgical repair via a median sternotomy with a CBP approach which was confirmed to carry successful outcomes for our patient in early and late follow-up visits. The surgical plan included: first the ligation of the arterial canal in its pulmonary part, subsequent resection of the insertion part of the arterial canal then finally the reconstruction of a latero-terminal anastomosis between the posterior surface of the aortic arch and the distal aorta. As for the large APW, a pericardium patch was used to seal it.

Conclusion

The complex surgical management of patients with such lesions results in substantial mortality and morbidity. In particular, the rate of re-interventions of either the pulmonary trunk, pulmonary artery branches, or aortic arch can be high. Therefore, prompt, and accurate diagnosis of cardiovascular anomalies and the full spectrum of the possible associations by noninvasive diagnostic methods is crucial to adequately reduce the overall prognosis and to establish the best surgical approach.

Conflict of Interest

The authors declare that there is no conflict of interest regarding the publication of this paper.

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