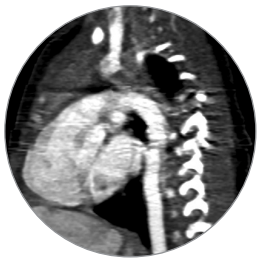

Interrupción de arco aórtico en síndrome de Down: presentación de un caso con revisión radiológica

Interrupted Aortic Arch with Down's Syndrome: A Case Report with Radiological Review

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Summary

Introduction: Interrupted Aortic Arch (IAA) is an obstructive anomaly, with a rare frequency of isolated presentation in the general population, being different when it presents concomitance with other cardiac anomalies and / or genetic syndromes such as DiGeorge Syndrome and Down Syndrome. **Case report:** The case of a newborn with facies characteristic of Down Syndrome, diagnosed with Interrupted Aortic Arch type A, is presented through computed tomography angiography. **Discussion:** Three types of Interrupted Aortic Arch (IAA) have been described so far, in which a persistent arterial ductus that allows continuity between the pulmonary artery and the descending aorta has been identified as a common feature. There are no reports of diagnostic algorithm for the detection of this pathology, but it has been studied using 2D echocardiography, computed tomography angiography (CT) and magnetic resonance imaging (MRI). Its definitive treatment is surgical. **Conclusion:** A case report of the interruption of the aortic arch is presented with a comprehensive review of the literature that correlates with radiological images, to cause an impact on the generation of knowledge of said pathology, which allows for a timely diagnosis, strengthens our medical behavior. and leads to better management of medical resources.

Resumen

Introducción: La interrupción del arco aórtico (IAA) es una anomalía obstructiva, poco frecuente en la población en general, siendo diferente al momento de presentar concomitancia con otras anomalías cardiacas o síndromes genéticos, como síndrome de DiGeorge y síndrome de Down. **Presentación de caso:** Se trata de una recién nacida con facies características del síndrome de Down, diagnosticada con interrupción de arco aórtico tipo A, mediante angiografía por tomografía computarizada. **Discusión:** En la literatura se han descrito hasta el momento tres tipos de interrupción del arco aórtico (IAA), en los cuales se ha identificado como característica común un conducto arterioso persistente que permite la continuidad entre la arteria pulmonar y la aorta descendente. No existen informes de algoritmo diagnóstico para la detección de esta patología, pero sí se ha estudiado mediante ecocardiograma 2D, la angiografía por tomografía computarizada (TAC) y resonancia magnética (RM). Su tratamiento definitivo es quirúrgico. **Conclusión:** Se presenta un caso de la interrupción del arco aórtico con una revisión amplia de la literatura que se correlaciona con imágenes radiológicas, con el fin de generar conocimiento sobre dicha patología lo que permite un diagnóstico oportuno, fortalece la conducta médica y conduce a un mejor manejo de los recursos médicos.

Introduction

Aortic arch disruption (AAD) is a malformation rare congenital, which represents the most severe form of obstructive abnormalities of the aortic arch (1), in which the continuity has been lost luminal between the proximal aorta and the descending aorta. It was first described in 1778 by Steidele (2), who reported that the flow into the descending aorta is the ductus, which remains permeable and provides continuity between the trunk of the pulmonary artery and distal aorta. It has an incidence of three cases per million of live newborns, and represents 1 % of all cardiovascular abnormalities (3). It has a strong association with chromosomal diseases such as 22q11 (syndrome of DiGeorge) with an incidence of approximately 25 (4), in trisomy 21 (Down syndrome) associated to cardiovascular malformations in 44-50 % of cases (5) and with obstructive arch abnormalities aortic in approximately 0.69 of 1000 births alive (6). Given its low incidence it becomes a little studied, which makes it necessary to generate clear knowledge of this pathology with the in order to make a timely diagnosis. It is presented the case of a newborn with aortic dissection diagnosed by CT angiography computerized (CT).

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Clinical case

24-hour female patient with a multiparous mother. At the time of delivery she weighed 3095 grams and was 49 cm tall. Physical examination showed facies characteristic of Down's syndrome. The mother denied any history of significance. A 2D echocardiogram was performed, from which a written report was received with results of hypoplasia of the aortic arch, aortic isthmus with aortic coarctation; posterior wide ventricular septal defect without hemodynamic repercussion, patent ductus arteriosus, large bidirectional short without hemodynamic repercussion; ostium secundum type interauricular septal defect without hemodynamic repercussion. After evaluating the outcome, cardio pediatrics decides to hospitalize the patient in the pediatric intensive care unit (ICU).

In the first hours of hospital stay, the patient develops respiratory distress and cyanosis, so medical management is initiated. A CT angiography (CT angiography) is taken, which, in its axial section, confirms a hypoplastic aortic arch (Figure 1A) and significant dilation of the pulmonary artery (Figure 1B) also visible in the sagittal and coronal sections (Figures 2 and 3). In addition, in the sagittal section, persistence of the ductus arteriosus and continuation of the descending aortic artery with the pulmonary artery are visualized (Figure 2A). The diagnosis of type A aortic aberration is confirmed due to the discontinuity between the ascending aorta continued by the right and left carotid artery and the pulmonary artery with significant dilatation of approximately 13 mm in diameter (Figure 2B). The image is reviewed in 3D reconstruction which confirms the above findings (figure 4). After the diagnosis of this pathology, the patient is treated with prostaglandin E1 during her stay in the pediatric ICU; however, the patient dies in her neonatal period.

Discussion

Disruption of the aortic arch is one of the most severe forms and the rarest congenital anomaly in the spectrum of obstructive lesions of the aortic arch, defined as loss of anatomical continuity between the ascending and descending aorta and considered an extreme form of coarctation (1-3). Flow into the descending aorta is provided by the ductus, which remains permeable and provides continuity between the trunk of the pulmonary artery and the distal aorta. Despite this, the blood that reaches the abdomen and lower limbs is partially oxygenated. Patients evolve with severe congestive heart failure, a tendency to shock and, in the absence of timely treatment, usually die in the neonatal period (3).

Today different forms of manifestation of IAA are recognized and different classifications have been described, for example, that of Celoria and Patton (1959), which classify it according to the site of interruption: Type A, the disruption is located distal to the left subclavian artery; type B, it is located distal to the left common carotid artery, thus the left subclavian artery arises distal to the disruption of the descending aorta; and type C, which is exceptionally rare, is between the trunk brachiocephalic artery (innominate artery) and the left common carotid artery (1).

Epidemiologically, IAA is found in about 3 cases per million live births and represents 1% of all cardiovascular abnormalities (3). But its incidence changes because of its frequent association with other cardiovascular abnormalities (patent ductus, ventricular septum defects, ventricular septal defects, etc.), as well as genetic diseases or syndromes, such as DiGeorge syndrome and that of Down, with a frequency of 25 % and 0,069 % respectively (4-6). Also, according to this classification, type B is presented in a 70 %, while type A is 28 % and type C is 1 % (1).

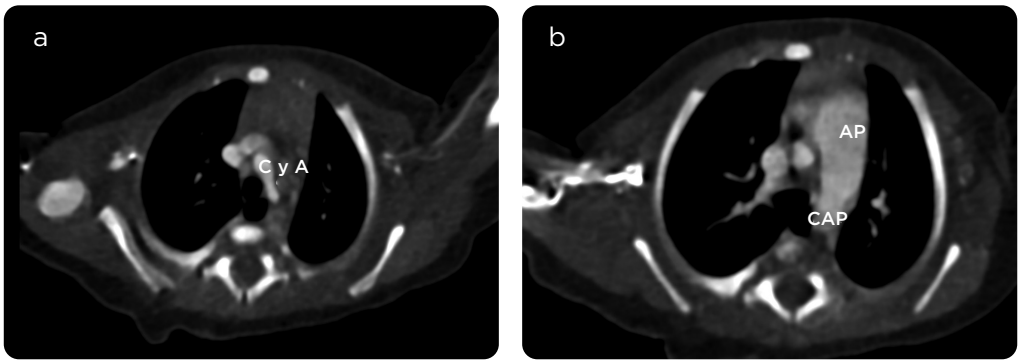

Figure 1. AngioCT, a) axial section: mild hypoplasia of the aortic arch (C&A). b) Significant dilation of the pulmonary artery (PA) and patent ductus arteriosus (PDA) without dilation.

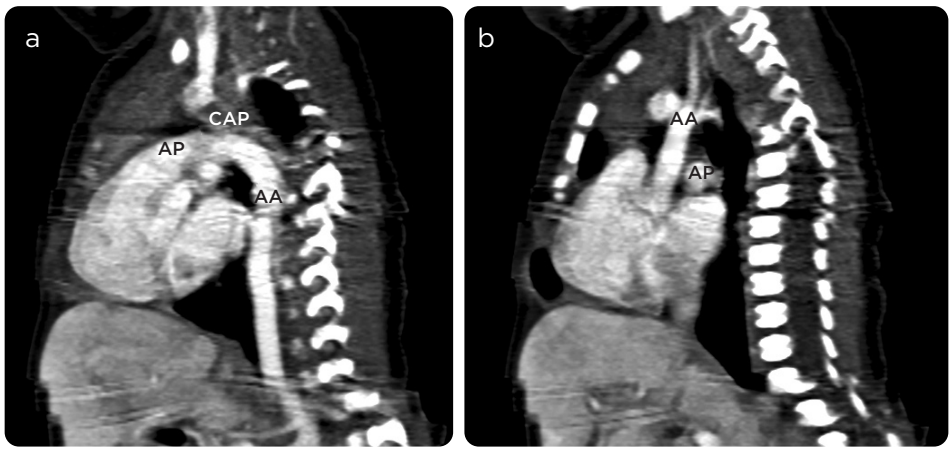

Figure 2. AngioCT, (a) sagittal split: pulmonary artery (PA) with significant dilation of 13 mm in diameter, patent ductus arteriosus (PDA) of 6 mm in diameter and the origin of the descending aorta from the ductus arteriosus, with obvious continuity b) Discontinuity and difference is observed of origin between the ascending aortic artery (AA) and the pulmonary artery.

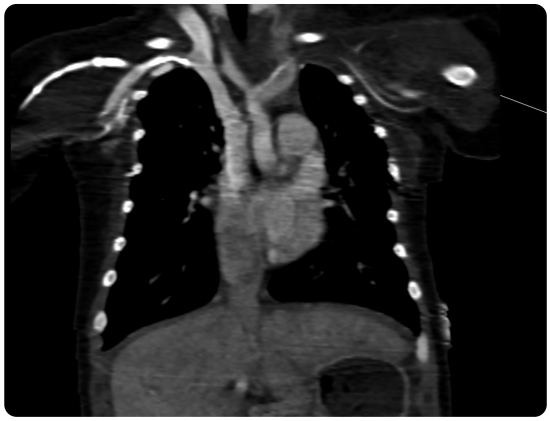

Figure 3. AngioCT, coronal section: non-congruent origin between the ascending aorta and the pulmonary artery, which is significantly dilated.

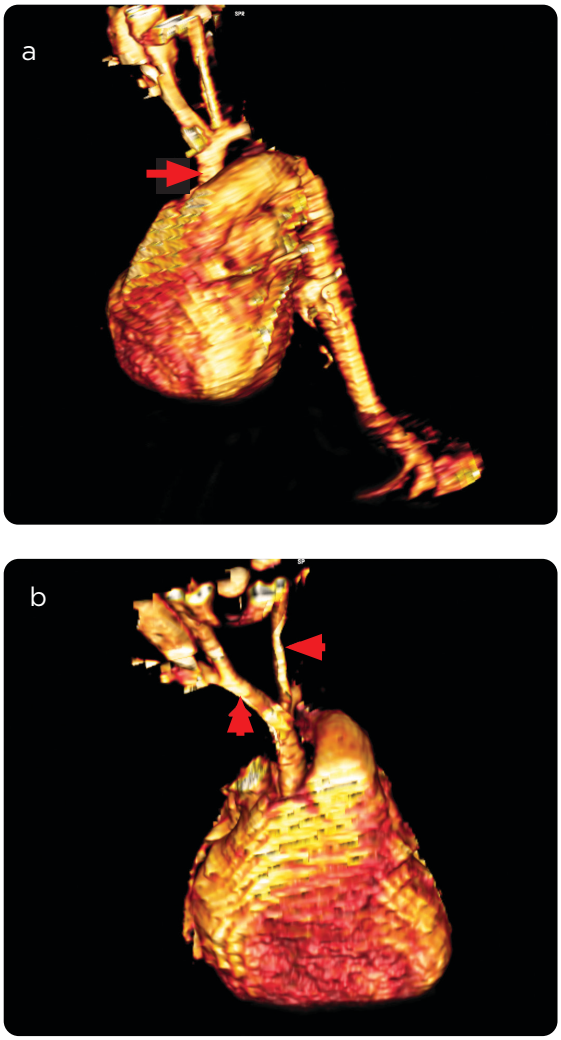

Figure 4. 3D angioCT reconstruction. Lateral and anterior view: type A aortic aberration accompanied by significant dilatation of the pulmonary artery of 13 mm in diameter (AP). The ascending aorta (large arrow) continues as the right (small arrowhead) and left (large arrowhead) common carotid arteries. A 6-mm diameter patent ductus arteriosus (PDA) gives rise to the descending aorta.

Embryologically, under normal conditions, the thoracic artery is formed by the creation of six bilateral aortic arches and the seventh intersegmental arteries, with the involution of several segments of these. Thus, according to the level of failure in the regression it can be classified as IAA type A, which is formed by the abnormal regression of the fourth left arch late in development, after the left subclavian artery has ascended to its normal position (7).

There are various mechanisms for diagnosing this pathology without the existence of a specific algorithm, including specific protocols or guidelines for each institution, adapted to the resources available. In the case described, a 2D echocardiogram was performed; however, this method is limited because it does not allow adequate observation of the continuation of the main pulmonary artery through the ductus arteriosus to the descending aorta (ductal arch), which may resemble a true aortic arch. However, it is possible to differentiate it by the absence of the origin of the brachiocephalic vessels. In addition, the cardiac anatomy in more than two thirds of the cases is characterized by a biventricular heart with normal connections and a large ventricular septal defect (1).

Echocardiography does not differentiate one case of IAA from another in which severe aortic coarctation and a hypoplastic aortic arch coexist, so complementary tests such as CT or MRI are necessary. These techniques demonstrate the morphological characteristics of AAI and the possible complex associated findings, due to their multiplanar capabilities, which facilitate the understanding of the abnormality and its anatomical relationships. However, CT has more advantages, such as shorter performance time with less sedation requirements and higher image resolution with simultaneous view of the airway (8).

Patients with IAA undergo cardiac surgery during the first year of life and have an overall survival rate at 16 years of about 59-70 %. The administration of prostaglandin E1 therapy in the neonatal period, to keep the ductus arteriosus open, has been shown to improve survival. The goal of treatment is to restore continuity of the aorta; however, the surgical approach is controversial. The most studied consists of reconstruction by end-to-end anastomosis, creation of a left subclavian flap, carotid reduction and placement of an interposition graft. Post-operative mortality is between 15 and 20 % (4).

Conclusion

The world literature describes AAI as a rare congenital heart disease, with few case reports and a low prevalence, which is diagnosed concomitantly with other pathologies, mainly associated with congenital syndromes. In reporting this case, the need to deepen the knowledge of this pathology is stressed, in order to make a diagnosis of type A aortic arch interruption, using the available tools such as 2D echocardiography and CT angiography. Radiological images are presented as a contribution to the development of a timely diagnosis in the medical institutions of the department, a region in which there are no reports of the pathology or data on confirmed cases.

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