Atresia of the aortic arch in 4-year-old child: a clinical case study

NIGRO STIMATO, Vittoria, et al.

Abstract
Atresia of the aortic arch is a rare congenital heart defect with a high mortality when associated with other intracardiac defects. Cardiac magnetic resonance (CMR) provides the exact anatomy of the aortic arch and collateral circulation and is useful to diagnose-associated aortic arch anomalies. This report describes the case of a 4-year-old child with atresia of the aortic arch, referred to our institution with the diagnosis of aortic coarctation and bicuspid aortic valve. On clinical exam, the femoral pulses were not palpable and there was a significant differential blood pressure between the upper and lower limbs. The echocardiography showed a severely stenotic bicuspid aortic valve but was limited for the exact description of the aortic arch. CMR showed absence of lumen continuity between the ascending and descending aorta distal to the left subclavian artery, extending over 5 mm, with the presence of a bend in the arch and diverticulum on either side of the zone of discontinuity, suggesting the diagnosis atresia of the aortic arch rather than coarctation or interruption. The patient benefited from a successful [...]

Reference

DOI: 10.3389/fped.2015.00019
PMID: 25853109
Atresia of the aortic arch in 4-year-old child: a clinical case study

Vittoria Nigro Stimato¹* , Dominique Didier² , Maurice Beghetti¹ and Cécile Tissot¹

¹ Cardiology Unit, Child and Adolescent Department, Geneva University Hospital, Geneva, Switzerland
² Magnetic Resonance Unit, Department of Radiology, Geneva University Hospital, Geneva, Switzerland

INTRODUCTION

Aortic arch obstructive lesions can have a large spectrum of presentation, going from coarctation through atresia or interruption of the aortic arch. Echocardiography is the exam of choice for assessment of aortic arch anomalies in children, but is sometimes not sufficient in older patients with poor echocographic windows. Cardiac magnetic resonance (CMR) provides the exact anatomy of the aortic arch and collateral circulation and is useful to establish the surgical strategy.

CASE REPORT

We report the case of a 4-year-old boy referred to our institution for surgical treatment of aortic coarctation and bicuspid aortic valve. The patient had no complaint. On clinical exam, the child was in excellent condition. The cardiac exam revealed a harsh 4/6 systolic murmur at the aortic area. The femoral pulses were not palpable, the child was hypertensive on the upper extremities with a significant differential blood pressure between the upper and lower limbs (Table 1).

The electrocardiogram showed sinus rhythm with signs of left auricular hypertrophy, but no ventricular hypertrophy. The chest radiography (Figure 1) showed mild cardiomegaly (ICT 3.3), preserved systolic and diastolic function (mitral E/A ratio 1.6; mitral A deceleration time 0.10 s, E/E’ 22 secondary to increased left heart filling pressure), and tortuous aortic arch with narrowing and acceleration of flow at the aortic isthmus (Figure 3). CMR with angiography showed absence of continuity between the ascending aorta distal to the left subclavian artery (Type 1) and the descending aorta, extending over 5 mm. There was a bend in the arch and diverticulum on either side of the zone of discontinuity, suggesting the diagnosis atresia of the aortic arch rather than coarctation or interruption. The patient benefited from a successful surgical commissurotomy of the aortic valve and reconstruction of the aortic arch with a homograft. The post-operative CMR confirmed the good surgical result. This case emphasizes the utility of CMR to provide good anatomical information to establish the exact diagnosis and the operative strategy.

DISCUSSION

Atresia of the aortic arch is a rare congenital anomaly. It is often associated with other cardiac defects, particularly persistent patency of the arterial duct, ventricular septal defect, and sub-aortic or aortic valvar stenosis. Although the distinction between coarctation and interruption is easily made, the difference between interruption and atresia of the aortic arch is not so evident. The two lesions, although producing identical hemodynamic...
Table 1 | Arterial blood pressure at four limbs in our patient.

<table>
<thead>
<tr>
<th>Limb</th>
<th>Systolic (mmHg)</th>
<th>Diastolic (mmHg)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Right upper</td>
<td>128</td>
<td>82</td>
</tr>
<tr>
<td>Left upper</td>
<td>114</td>
<td>80</td>
</tr>
<tr>
<td>Right lower</td>
<td>93</td>
<td>71</td>
</tr>
<tr>
<td>Left lower</td>
<td>99</td>
<td>61</td>
</tr>
</tbody>
</table>

The differential of blood pressure is maximum 15 mmHg for systolic blood pressure (SBP) and 10 mmHg for diastolic blood pressure (DBP). Normal values at 4 years SBP 102–115 mmHg DBP 62–71 mmHg.

consequences, both produce discontinuity between the transverse and descending components of the aorta. When the aortic arch is interrupted, however, part of its transverse component is completely absent, with varying distance then possible between the ends of the patent components of the arch. When the involved segment is atretic, it remains as an anatomic entity, although being fibrous and lacking any luminal patency. The persisting presence of the fibrous connection between the patent components of the arch also limits the length of the gap between them. In our patient, the presence of a diverticulum on either side of the zone of discontinuity, along with the identification of a fibrous strand by the cardiac surgeon, confirmed the presence of atresia rather than interruption of the aortic arch. The classification into types A, B, and C is used to distinguish the site of both interruption and atresia of the aortic arch. In a large series of patients with interruption, 42% of patients were of type A, with the arch interrupted at the aortic isthmus beyond the origin of the left subclavian artery, 53% of type B, with the interrupted segment between the origin of the left common carotid and left subclavian artery, and 4% of type C, when the interruption was between the innominate and the left common carotid artery (1).

The mortality of interruption or atresia of the aortic arch is high during the first year of life, reaching about 76% without surgical repair (2). Some cases can survive through infancy and late diagnosis has been described in the literature (3–5), almost all late survivors have been shown to have no additional

FIGURE 1 | Chest X-ray with prominent right cardiac border secondary to dilatation of the ascending aorta (double arrow) and absent aortic knob (arrow).

FIGURE 2 | Long axis parasternal view showing doming of the aortic valve, aliasing of flow secondary to stenosis, and left ventricular hypertrophy. Short axis view showing bicuspid aortic valve. Abbreviations: AoV, aortic valve; LA, left atrium; LV, left ventricle; RV, right ventricle.

FIGURE 3 | Suprasternal view of the aortic arch with severe narrowing of the aortic isthmus and aliasing of flow. Abbreviations: Asc Ao, ascending aorta; Desc Ao, descending aorta.
intracardiac anomalies. In this setting, symptoms resemble those of long-standing systemic hypertension and coarctation. Central nervous system symptoms may be seen secondary to vertebral steal, particularly in type B or C. In our patient, the good clinical tolerance by possible slow progression of the aortic valve stenosis in a child in whom obstruction of the aortic arch may have been present soon after birth, in the setting of ductus arteriosus closure. Atresia of the aortic arch may have allowed for collateral vessels formation already during the fetal life, allowing sufficient blood flow supply to the lower part of the body. Some degree of aortic valve stenosis may have been present since fetal life and may have
allowed for hypertrophic adaptation of the left ventricle and better hemodynamic tolerance to an associated obstructive lesion of the aortic arch. Similar evolutions have already been described in the literature (6).

Obstructive lesions of the aortic arch are normally diagnosed in the neonatal period by physical examination, but the diagnosis is sometimes missed when the stenosis is mild with a significant number of asymptomatic patients being diagnosed during infancy (7, 8). Echocardiography is a good tool to visualize the aortic arch, particularly in small children, and is most often sufficient to have a good anatomical picture of the arch. CMR particularly 3D gadolinium-enhanced angiography provides the exact anatomy of the aortic arch and collateral circulation (9, 10), and is useful to diagnose-associated aortic arch anomalies like double aortic arch, right-sided aortic arch with mirror image branching and aberrant right subclavian artery (11), essential to establish the preoperative strategy (12). CMR is also important in detecting residual aortic arch hypoplasia, recurrent narrowing and aneurysmal formation in the post-operative period (13) and is recommended by the American Heart Association for every adult patient following aortic root surgery (14). We used CMR in our patient because of poor echographic windows to establish the exact diagnosis, the preoperative strategy, and also to assess the post-operative result.

The impact of bicuspid aortic valve and severe aortic valve stenosis in patients with obstructive lesions of the aortic arch on medium and long-term outcome was recently described by Sugimoto et al. (15), with bicuspid aortic valve representing a risk factor for later regurgitation or stenosis.

CONCLUDING REMARKS

Careful clinical exam is essential in pediatric patients with arterial hypertension and heart murmur to avoid the long-term morbidity associated with undiagnosed obstructive lesions of the aortic arch, the most common diagnosis being coarctation. Echocardiography is essential for the diagnosis but sometimes not sufficient to have a good anatomical description of the anatomy. CMR provides good anatomical information to establish the definitive anatomical diagnosis and the operative strategy.

ACKNOWLEDGMENTS

We wish to thank Dr. Robert H. Anderson, retired Professor of Pediatric Cardiac Morphology, Institute of Child Health, University College of London/UK, for his valuable expert opinion about the anatomical diagnosis of our patient.

REFERENCES


Conflict of Interest Statement: The authors declare that the research was conducted in the absence of any commercial or financial relationships that could be construed as a potential conflict of interest.

Received: 18 August 2014; accepted: 23 February 2015; published online: 20 March 2015.


This article was submitted to Pediatric Cardiology, a section of the journal Frontiers in Pediatrics.

Copyright © 2015 Nigro Stimato, Didier, Beghetti and Tissot. This is an open-access article distributed under the terms of the Creative Commons Attribution License (CC BY). The use, distribution or reproduction in other forums is permitted, provided the original author(s) or licensor are credited and that the original publication in this journal is cited, in accordance with accepted academic practice. No use, distribution or reproduction is permitted which does not comply with these terms.