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Case Report

Unusual presentation of a rare congenital anomaly: interrupted aortic arch presenting in adulthood with myocardial infarction

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ABSTRACT

Interrupted aortic arch (IAA) is a very rare congenital cardiac anomaly in which there is complete interruption of a part of the aortic arch due to abnormal regression of a segment of the left fourth arch during development. The condition usually presents in the neonatal period with abrupt onset cardiac failure and shock after closure of the ductus arteriosus that maintains perfusion distal to the IAA in fetal life. In most cases, immediate surgery is necessary to save the life of the patient. In very rare cases, a massive collateral circulation can develop to compensate for the arch interruption and the condition may remain asymptomatic until adulthood, when it can present with early cardiovascular disease due to the chronic hemodynamic changes. Cross sectional imaging plays a major role in diagnosis and surgical planning in such adult patients. Surgery is usually recommended, even in asymptomatic cases, to prevent early onset cardiovascular diseases. Possibility of previously undiagnosed cardiovascular disease should always be a consideration when evaluating a young patient with myocardial infarction.

Keywords: IAA, Computed tomography angiography, Congenital heart disease

INTRODUCTION

Interrupted aortic arch (IAA) refers to a congenital cardiac anomaly in which there is complete discontinuity in any particular segment of the aortic arch so that the major systemic outflow from the heart is unable to follow its usual path. IAA is a rare congenital anomaly. The incidence is estimated to be approximately 2-3 in 1,000,000 live births. It is one of the least encountered congenital cardiovascular defects.[1] IAA is, in majority of the cases, associated with other congenital cardiac anomalies. Over 73% of cases associated with ventricular septal defect. Other complex cardiac anomalies may also be associated.[2] Such associated defects may further worsen the prognosis. It usually presents with severe heart failure in infancy. As it is a ductus dependent lesion and major systemic outflow to the distal segment of the interrupted aorta occurs through the ductus in fetal life, symptoms begin soon after closure of the ductus arteriosus in the neonatal period. Mortality rate is over 90% in untreated infants in the first year of life. However, there have been unusual cases reported in literature where IAA patients have survived into adulthood without surgery. Due to the rarity of such long-term survivors, there is a paucity of literature on the long-term prognosis and chronic complications in untreated IAA patients. We encountered a very unusual case of IAA in which the condition was present as an isolated anomaly and patient was undiagnosed and asymptomatic until the late fourth decade of life when he presented with myocardial infarction and was incidentally discovered to have IAA.

CASE REPORT

A 39 years old previously asymptomatic male patient presented to our hospital with sudden onset chest pain. ECG revealed left axis deviation with a lateral wall non-ST elevation myocardial infarction. Detailed clinical examination revealed asymmetric pulses in the limbs with weaker lower limb pulses as compared to upper limb pulses. Blood pressure measurements showed persistent hypertension in the upper limbs and normal to low

pressures in lower limbs. The patient was then referred to the radiology department for computed tomography coronary (CT) and aortic angiography for detailed vascular evaluation in view of young myocardial infarction and isolated upper limb hypertension.

CT angiography (CTA) was performed on a SIEMENS™ 256 slice dual energy CT scanner with retrospective ECG gating which did not require any heart rate control for a diagnostic coronary angiography scan. CTA revealed high grade stenosis of the proximal obtuse marginal artery arising from the left circumflex artery by a non-calcified plaque (Figure 1). The plaque was seen to cause over 90% occlusion of the vessel lumen. Rest of the coronary arteries were normal. Further, calcification was observed in the ascending aortic wall indicating chronic hypertension. The ascending aorta was also dilated. Focal complete interruption of the aorta was seen involving the distal aortic arch just distal to the origin of the left subclavian artery (Figure 2). The descending thoracic aorta was found to be reformed by a rich network of collateral arterial channels involving the bilateral intercostal arteries, internal mammary arteries, lateral thoracic arteries, mediastinal arteries, other branches of the subclavian arteries and even the anterior spinal artery, all of which were found to be dilated (Figure 3). Mild left ventricular hypertrophy and dilatation was seen, likely due to chronic upper body hypertension secondary to IAA. A hypoenhancing segment was seen in the anterolateral wall of the left ventricle consistent with infarcted myocardium. Functional imaging of the heart was performed by reformatting data generated from the retrospectively gated CTA dataset. Wall motion was found to be reduced in the infarcted segment (Figure 4). Ductus arteriosus was completely closed with no flow. No additional anomalies were found in either the heart or the rest of the visualized cardiovascular structures. Thymic remnant was also normally visualized. Rest of the lab tests including serum calcium and parathyroid hormone were within normal limits.

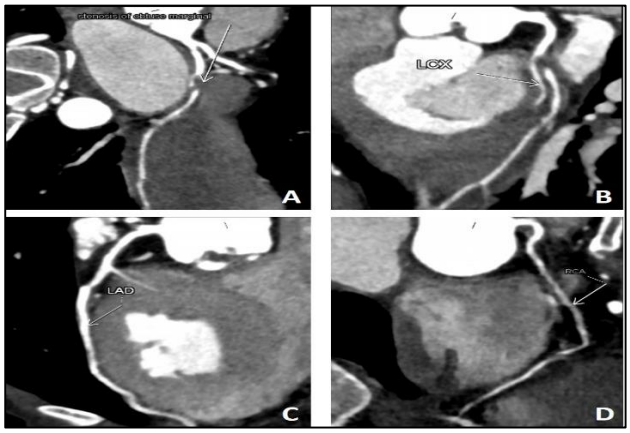

Figure 1 (A-D): CT coronary angiography showing high grade stenosis of proximal obtuse marginal artery (arrow), normal left circumflex, left anterior descending and right coronary artery.

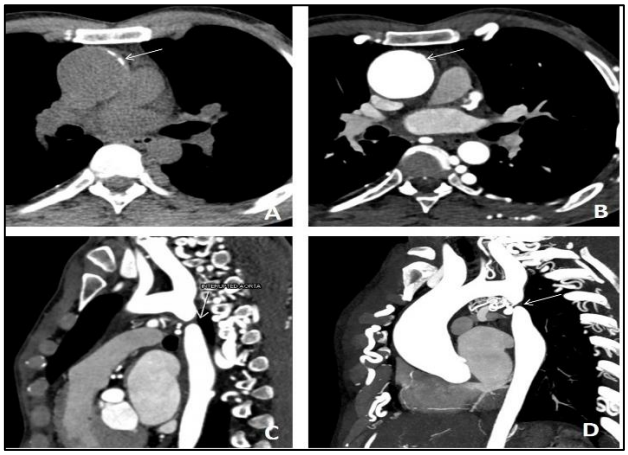

Figure 2 (A-D): CT aortography showing intimal calcification in ascending aorta (arrow), dilated ascending aorta (arrow), focal short segment interruption in the distal arch of aorta (arrows).

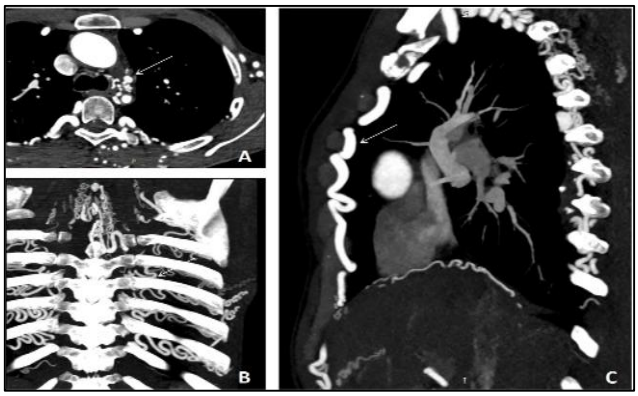

Figure 3 (A-C): CT aortography showing extensive collateral network formed by the mediastinal (arrow) and paravertebral arteries in intercostal arteries (arrow) and internal mammary arteries (arrow).

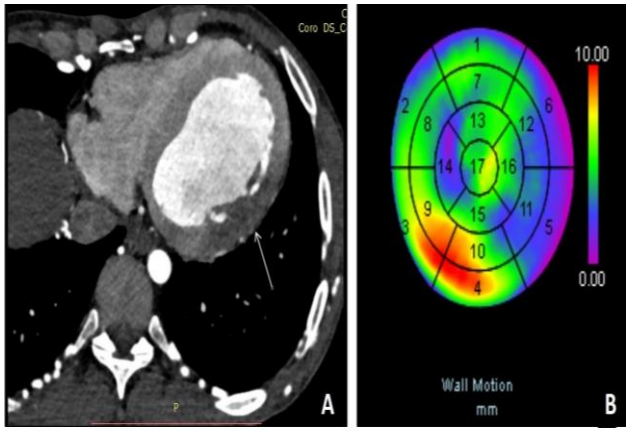

Figure 4 (A and B): Functional CT cardiac imaging showing mild dilatation and wall thickening of left ventricle with hypoenhancement of part of the lateral wall of left ventricle (arrow) and hypokinesia of segments 5, 6, 11 and 12 of left ventricular myocardium in left obtuse marginal artery territory.

Due the persistent risk of further cardiovascular events, decision was made to repair the IAA. Single stage open surgical repair of the IAA was done without cardiopulmonary bypass due to the distal location and short length of interruption in the aortic arch. Direct anastomosis of the ends of the interrupted arch with patch augmentation was performed. Post-surgical recovery was uneventful and patient was discharged home after five days. Patient is currently on follow up and is doing well.

DISCUSSION

IAA develops due to abnormal development of the embryonic aortic arches. The arch of aorta normally develops from the left fourth aortic arch, extending between the truncus arteriosus and the left dorsal aorta. Abnormal regression of any segment of the left fourth arch can lead to IAA.[3] The Celoria and Patton classification system is used to differentiate the various types of aortic arch interruption. It is based on developmental variations leading to IAA at different sites in the aortic arch and associated variations in the origin of the right subclavian artery. These lead to varied pathophysiological mechanisms and symptoms. The types of IAA according to this system are: type A: interruption distal to left subclavian artery, type B: interruption between the left common carotid and left subclavian artery, type C: interruption between the innominate and left common carotid artery and further sub-classification of each type according to the origin of right subclavian artery is: type 1: normal, from innominate artery, type 2: aberrant, from aorta distal to left subclavian artery and type 3: isolated, from right patent ductus arteriosus.[4]

The most common type of IAA is type B, which constitutes over 84% of cases. Type C is the least common. Our case was a type A1 IAA.

IAA is an uncommon congenital anomaly by itself. Isolated IAA is even rarer. In most cases, it is either associated with other congenital cardiac defects or seen as part of syndromic associations. Two major syndromes associated with IAA are DiGeorge syndrome and CHARGE syndrome. Almost 50% of IAA cases are found to have DiGeorge syndrome, which develops due to a deletion of 22q11.2 chromosome. These patients are almost always diagnosed in infancy with severe cardiac defects, thymic and parathyroid aplasia, facial dysmorphisms and associated immunodeficiency.

CHARGE is a less common association with IAA and consists of varying combinations of colobomas, congenital heart defects, choanal atresia, growth retardation, genitourinary anomalies and ear anomalies. Among association of IAA with other congenital heart defects, the most common is malalignment of the conotruncal septum leading to outflow obstruction and ventricular septal defects.[2] Our case was not associated with any other congenital cardiac anomalies, non-cardiac malformations or syndromes.

The pathophysiology of the condition depends on the time of closure of the ductus arteriosus since IAA is a ductus dependent lesion. In fetal life, flow through the patent ductus arteriosus perfuses the organs and limbs downstream from the obstruction. After birth, reduction in pulmonary vascular resistance leads to closure of the ductus arteriosus. This precipitates abrupt onset of hypoperfusion and cardiac failure. Positive feedback cycles further accentuate heart failure and death often results without early surgical intervention.[5] The features are more severe with more proximal lesions, with type C being the most severe due to interruption proximal to the left common carotid artery. In very rare cases with massive compensatory collateral circulation like in ours, it may remain asymptomatic until adulthood despite ductus closure, when it may present with chronic vascular changes leading to cerebrovascular or cardiovascular disease due to persistently increased pressure proximal to the interruption.

Management of the condition involves early diagnosis, preferably antenatal, using obstetric ultrasound. If undiagnosed, early neonatal echocardiography will usually reveal the diagnosis. Early management involves keeping the ductus arteriosus patent using prostaglandin E1. This can prevent abrupt onset cardiac collapse and death as it can maintain perfusion of the organs distal to the interruption.[6] This is, however, only a temporary measure and surgery should be performed as soon as possible. If shock develops, inotrope support may be needed to stabilize the patient prior to emergency surgery. Presurgical evaluation may be required with cross sectional imaging which may be performed with CT or magnetic resonance imaging (MRI). The definitive treatment is early surgery. Surgical approach depends on the associated cardiac defects and length of the interruption. Usually patch augmentation of the interrupted segment is required, which may be made out of native arterial tissue or synthetic graft material. Additional cardiac defects may necessitate a staged surgical procedure for repair.[7] Prognosis in neonates with multiple defects is often guarded, even with timely surgery.

CONCLUSION

IAA can, in very rare cases, present in adulthood in previously asymptomatic patients with early myocardial infarction. Cross sectional imaging plays a major role in diagnosis and surgical planning, especially in adults, due to development of an extensive collateral circulation. Surgical correction is recommended even in asymptomatic adult patients in order to correct hemodynamic abnormalities and prevent early cardiovascular disease. The possibility of undiagnosed congenital heart diseases should always be kept in mind

while investigating young patients with cardiovascular diseases.

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