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REVIEW

Hypoplasia, Pseudocoarctation and Coarctation of the Aorta – A Systematic Review

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Aortic arch abnormalities are uncommon and may be seen in association with other congenital cardiac anomalies. Coarctation, pseudocoarctation and hypoplastic aortic arch are known aortic arch abnormalities, with the former being well studied, whilst for the latter two, much less is known. There are similarities and differences that are important to distinguish among these three conditions in order to avoid errors in diagnosis that may result in unnecessary investigations, which may in turn result in physical or emotional harm to the patient. For this reason, we present a systematic review of the published literature providing an evidence-based overview that may be helpful to clinicians when faced with this diagnostic dilemma.

Keywords Aorta • Coarctation • Hypoplastic arch • Pseudocoarctation • Congenital heart disease

Introduction

The incidence of congenital heart disease (CHD) including even trivial lesions is approximately 75-81/1000 live births while the incidence of CHD requiring postnatal expert management is 2.5-3.0/1000 live births [1,2]. There have been important advances in the diagnosis and management of CHD in last few decades. The availability of sophisticated imaging technology has enabled accurate diagnosis of congenital cardiac anomalies in the prenatal period through to adulthood. Congenital anomalies of the aortic arch occur due to mal-development and involution of the six pairs of arches that arise from the paired dorsal aortae [3]. Development of the aorta is complex and occurs during the third week of gestation. The primitive aorta has ventral and dorsal segments. The ventral aorta fuses to form the aortic sac while the dorsal aorta joins to form the descending aorta. Subsequently, the six pairs of aortic arches develop further, connecting the aortic sac to the dorsal segment. During the fourth week of embryologic development of the heart, pharyngeal arches receive arteries from the heart. The cardiovascular system results from the development, fusion or regression of these six pairs of arteries coursing through the pharyngeal arches. Arches I, II and V regress, arch III forms the carotid system, and arch IV forms the aortic arch on the left and subclavian artery on the right. Arch VI forms both the pulmonary arteries and the ductus arteriosus on the left. These developmental milestones coupled with increased blood

Abbreviations: CHD, congenital heart disease; CT, computerised tomography; JVP, jugular venous pressure; MRA, magnetic resonance angiography; MRI, magnetic resonance imaging; TEE, transoesophageal echocardiography; TTE, transthoracic echocardiogram; VSD, ventricular septal defect

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flow through the aorta at six weeks of gestation are responsible for the development of a normal aortic arch. An anomalous development or insufficient increase in blood flow through the aorta can lead to a variety of aortic arch abnormalities that are frequently associated with other types of CHD [4]. Coarctation of the aorta is a well known and studied congenital condition, which is typified by a narrowing of the descending aorta typically located at the aortic isthmus or the insertion site of the ductus arteriosus distal to origin of left subclavian artery. Aortic coarctation comprises 6-8% of CHD with an incidence of 4/10,000 live births [1,2]. It has a male preponderance and is frequently associated with other cardiac anomalies such as a bicuspid aortic valve, transposition of the great arteries, ventricular septal defect and patent ductus arteriosus.

Hypoplastic aortic arch and pseudocoarctation of the aorta are two lesser known aortic anomalies that may be mistaken for the better known lesion of coarctation. This misdiagnosis may lead to unnecessary anxiety for the patient and their families, while adding cost and possible harm from unnecessary invasive investigations. Hypoplastic arch is mostly seen in the paediatric age group and has rarely been reported as an isolated finding. Hypoplastic aortic arch may be seen in as many as 81% of patients with coarctation of the aorta [5] or may occur in association with other congenital anomalies such as tetralogy of Fallot [6], ventricular septal defect [7], and pulmonary artery sling [8]. Both coarctation and hypoplastic aortic arch are thought to be part of the same disease spectrum with pathological association between the coarctation and tubular hypoplasia of the transverse arch. Both conditions may cause hypertension proximal to the obstruction; other symptoms and haemodynamic findings will depend on the presence of effective collateral circulation around the obstructed aortic segment. The third anomaly, pseudocoarctation of the aorta, consists of an elongated arch with a kink at the level of the isthmus. It does not cause obstruction to blood flow but can be associated with other congenital anomalies, including coarctation of aorta [9].

The diagnosis of these three anomalies can raise confusion due to their overlapping and similar clinical, radiological and pathophysiological features. This leads to unnecessary non-invasive or invasive investigations which may be costly, provoke anxiety, or pose risk to the patient. We present cases of hypoplastic arch and pseudo-coarctation of the aorta and compare and contrast these with true coarctation by reviewing the clinical presentation, imaging modalities and management of these three conditions.

Methods

We searched PubMed, Medline, EMBASE and Scopus using the terms ''pseudocoarctation of the aorta,'' ''hypoplastic arch,'' ''tubular aortic arch,'' and ''aorta.'' The search was repeated by a qualified librarian using the same search engines and terms. For pseudocoarctation, cases were selected for inclusion if they met the following criteria: English language, adult human patients, published in the last 20 years, true congenital form of pseudocoarctation (not pseudocoarctation ''like'' picture due to other causes such as aortic sarcoma and postoperative constriction), and included a description of clinical presentation and management of the patients. For hypoplastic arch we used the same inclusion criteria other than the age group. We included all cases, adult and paediatric, in the search strategy for hypoplastic arch cases due to the paucity of adult published data.

Findings

The search strategy resulted in 312 articles for pseudocoarctation and hypoplastic arch. After discarding studies and case reports that involved neonates and paediatric population (for pseudocoarctation) and articles addressing only surgical techniques, a total of 21 cases of pseudocoarctation and hypoplastic arch (18 pseudocoarctation including one case presented here and three hypoplastic arch cases) were included in this review. Articles were analysed for patient demographics, clinical symptoms and radiological studies. The findings are summarised in Table 1.

Hypoplastic Arch

Illustrative Case 1

A 22 year-old man presented to the outpatient cardiology clinic for a routine visit. The patient had a known history of a bicuspid aortic valve and coarctation of aorta diagnosed at four years of age. A peak gradient of 20 mmHg across the aortic valve and a 35-mm gradient across the aortic coarctation were measured at that time. The patient was under regular surveillance with no hypertension and was physically active without functional limitations. On examination, he had a normal carotid pulses and jugular venous pressure (JVP) and waveforms. Sitting blood pressures recorded were: left upper extremity 122/80, right upper extremity 158/70, left lower extremity 150/100 and right lower extremity 150/100 mmHg. The brachial pulses were asymmetrical (right stronger than the left) and a brachiofemoral delay was noted on the right side. The femoral, dorsalis pedis and tibial pulses were reduced on both lower limbs. Cardiac examination revealed a systolic click, a grade 2/6 ejection systolic murmur, and a high-pitched diastolic murmur at the left upper sternal border. The systolic murmur was loudest in the suprasternal area.

A chest radiograph was unremarkable. Transoesophageal echocardiography (TEE) demonstrated a bicuspid aortic valve with mild to moderate aortic regurgitation. The ascending aorta was hypoplastic with acute angulation of the aortic arch. Colour Doppler showed flow acceleration across the transverse arch. Mean Doppler gradient across aortic valve was 10 mmHg and in the descending aorta 16 mmHg. A nonobstructive web in the proximal descending thoracic aorta distal to dilated left subclavian artery was seen.