Coarctation of the Aorta
Strategies for Improving Outcomes

Lan Nguyen, MD[a], Stephen C. Cook, MD[b,*]

KEYWORDS

• Congenital heart disease • Aortic coarctation • Bicuspid aortic valve • Cardiac surgery • Stent • Aneurysm • Aorta • Treatment outcome

KEY POINTS

- Coarctation of the aorta is defined by a discrete narrowing of the aorta.
- Transcatheter systolic coarctation gradient ≥20 mm Hg is an indication for intervention with treatment choice guided by patient age and anatomy of obstruction.
- Follow-up imaging should be tailored to early identification of recoarctation/aneurysm with directed intervention.
- Hypertension is common in the aging patient with coarctation despite successful repair.
- Lifelong routine evaluation by cardiology specialists with expertise in adult congenital heart disease is required to identify late-onset complications.

INTRODUCTION

Coarctation of the aorta (CoA) is a common congenital heart defect (CHD) found in approximately 1 per 2900 live births[1–3] and is the seventh most common type of CHD.[4] Still, this is likely an underestimate, because the diagnosis may be delayed, even in the pediatric population.[4,5] In simple terms, coarctation is characterized by discrete narrowing of the thoracic aorta adjacent to the ligamentum arteriosum. Importantly, discrete coarctation is an aortopathy that lies within a spectrum of arch abnormalities ranging from discrete narrowing to a long segment of arch hypoplasia. The prognosis of untreated coarctation was extremely poor during the presurgical era with median survival age of 31 years and a quarter of patients dying before the age of 20 years.[6] Since the first surgical repair of aortic coarctation performed in the 1940s, treatment of coarctation has dramatically changed. Overall, survival into adulthood is now expected. However, these patients continue to require lifelong follow-up for management of associated problems including arterial hypertension, atherosclerotic disease, recoarctation, and aneurysm formation.

CAUSE AND PATHOGENESIS OF COARCTATION

Histologic examination of localized aortic coarctation lesions has demonstrated the presence of a tissue ridge extending from the posterior aortic wall and protruding into aortic lumen. This ridge consists of ductal tissue with in-folding of the aortic media.[6] In older patients, aortic intimal proliferation also contributes to the narrowing at the site of coarctation.[7] The cause of discrete aortic coarctation remains unclear, but is likely multifactorial. Prenatal environmental exposures have been associated with CoA and other left-sided lesions.[8] However, there is a growing body of literature that suggests a genetic basis for development of these lesions. Case series have described

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[a] Department of Cardiovascular Medicine, Heart and Vascular Institute, University of Pittsburgh, Scaife Hall S560.1, 200 Lothrop Street, Pittsburgh, PA 15213, USA; [b] Department of Pediatrics, The Adult Congenital Heart Disease Center, Heart Institute Children's Hospital of Pittsburgh of UPMC, 4401 Penn Avenue, Pittsburgh, PA 15224, USA

* Corresponding author.

E-mail address: stephen.cook@chp.edu

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clustering of coarctation cases in families. Evaluation of families with an index case of left ventricular outflow tract abnormalities of aortic valve stenosis, CoA, or hypoplastic left heart syndrome suggest a strong genetic influence, with an estimated sibling recurrence risk of greater than 30-fold.[9] Recently, mutations in the NOTCH1 gene have been identified in individuals with left ventricular outflow tract malformation, including coarctation.[10] In particular, the NOTCH1 variant R1279H seems to be more common in individuals with aortic coarctation.[11] NOTCH1 mutations have also been shown to contribute to abnormal epithelial-to-mesenchymal transition in endothelial cells, which is an important step in the development of the left ventricular outflow tract. Mechanical models have suggested that abnormalities of blood flow, defective endothelial cell migration, and excessive deposition of aortic duct tissue at the aortic isthmus can result in coarctation. Furthermore, embryonic studies in zebrafish have highlighted the importance of intracardiac hemodynamics in epigenetic control of distal chamber development.[12]

ASSOCIATED CONGENITAL HEART LESIONS

Although CoA can be an isolated CHD, it is also commonly found in other congenital syndromes and cardiovascular anomalies. Thus, deliberate investigation for the presence of coarctation should be made in these patients. The most common cardiovascular malformation associated with CoA is bicuspid aortic valve (BAV). Prior autopsy examination showed 46% of patients with CoA have congenital BAV.[13] More modern studies in patients with repaired coarctation found similar results with up to 45% to 62% prevalence of BAV.[14–16] The coincidence of BAV and CoA is difficult to determine, because BAV is very common and not everyone is screened for the presence of coarctation. In a study of 102 patients with BAV diagnosed by computed tomography (CT) imaging, 22% of patients either had prior coarctation repair, or were found to have CoA.[17] The coexistence of BAV and coarctation is important to consider, because it places the patient at a higher risk of aortic complications.[18] In a study following 341 patients with BAV over a median of 7 years, patients with bicuspid valve in the presence of coarctation had 7.5 times increased risk of ascending aortic complications, most commonly dilation of the ascending aorta.[19] The same group also found that among patients with aortic coarctation, the presence of a BAV was an independent risk factor for the development of aortic wall complications.[16]

Turner syndrome has a strong association with CoA. In a study of 132 girls diagnosed with aortic coarctation who subsequently underwent karyotyping, Turner syndrome was diagnosed in 5.3%.[20] CoA is found in 18% of patients with Turner syndrome.[21] Williams syndrome, a congenital and multisystem genetic disorder, has been associated with supravalvular aortic stenosis. Aortic arch abnormalities, including coarctation, are present in 10% of patients with Williams syndrome.[22] Coarctation can also be present in congenital cardiovascular anomalies involving multiple left-sided lesions, including Shone syndrome and hypoplastic left heart syndrome.

NONCARDIAC ASSOCIATIONS

The link between intracranial aneurysms and CoA was described well before the surgical era, accounting for 5% deaths in patients with aortic coarctation on autopsy review.[23] In the modern era, with the availability of brain MRI the reported prevalence of intracranial aneurysms in patients with CoA is approximately 10%,[24] which is five times more common than the average population (Fig. 1). In one study, hypertension was more common in the population of coarctation patients with

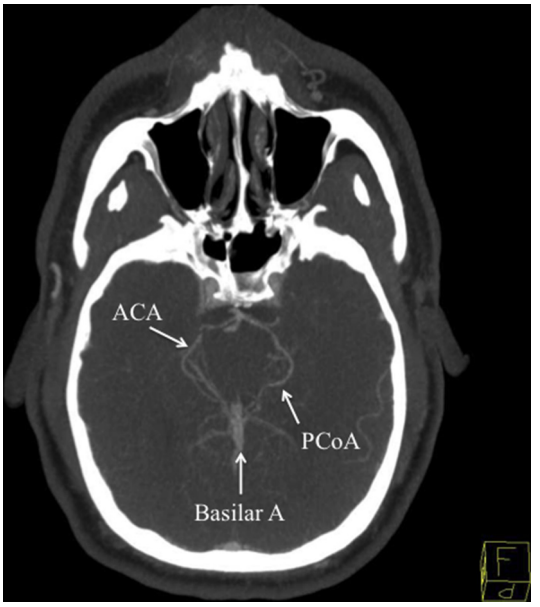

Fig. 1. Computed tomography angiography of the head showing normal anatomy of the circle of Willis without cerebral artery aneurysm in a 36 year old with coarctation of the aorta. Given that hypertension may play a role in the growth of intracranial aneurysm, these patients should be monitored and treated if indicated. ACA, anterior cerebral artery; basilar A, basilar artery; PCoA, posterior communicating artery.