

# A Rare Case of Vascular Ring and Coarctation of the Aorta

in Association with CHARGE Syndrome

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A male neonate presented with CHARGE syndrome, a multiorgan genetic disorder involving the Coloboma of the eyes, congenital Heart defects, nasal choanal Atresia, growth and development Retardation, Genitourinary disorders, and Ear anomalies and deafness. Moreover, he had a rare case of vascular ring—consisting of a right aortic arch with retroesophageal brachiocephalic artery—combined with coarctation of the mid-aortic arch. He underwent both vascular ring and aortic arch repair at our institution.

To our knowledge, this is the 4th documented case of this exceedingly rare type of aortic arch anomaly combined with aortic arch obstruction. Moreover, it is the first confirmed case of these combined disorders occurring in CHARGE syndrome.

This report describes a truly rare case and reveals the limitations of echocardiography in detecting complex aortic arch anomalies while illustrating the benefits of advanced imaging prior to surgical intervention. (*Tex Heart Inst J* 2017;44(2):138-40)

**Key words:** Abnormalities, multiple/diagnosis/genetics; aorta, thoracic/abnormalities/diagnostic imaging; aortic coarctation/complications/diagnostic imaging/surgery; cardiac surgical procedures; CHARGE syndrome/genetics/pathology; heart defects, congenital/genetics; infant; tomography, x-ray computed; treatment outcome

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**C**HARGE syndrome is a multiorgan genetic disorder involving the Coloboma of the eyes, congenital Heart defects, nasal choanal Atresia, growth and development Retardation, Genitourinary disorders, and Ear anomalies and deafness, and it is highly associated with congenital heart disease and aortic arch (AA) anomalies.<sup>1</sup> Although many different types of congenital heart disease have been described, conotruncal and AA anomalies are the most prevalent.<sup>1</sup> The variety of AA anomalies associated with CHARGE syndrome is immense, and complete vascular rings have been reported. However, a right AA with a retroesophageal brachiocephalic artery and AA obstruction has rarely been reported in the literature.<sup>2-4</sup> To our knowledge, this is the only case—confirmed via genetic testing—of this lesion's occurring within the context of CHARGE syndrome.

## Case Report

A term (39-wk) male infant with a prenatal history of cleft lip and palate was delivered at another institution. Shortly postpartum, the infant had difficulty with secretions, dyspnea in the supine position, and dusky undertones. The infant was responsive to supplemental oxygen and had a negative hyperoxia test. A 4-extremity blood pressure evaluation yielded normal results. The infant's inability to handle secretions and his continued need for respiratory support led to intubation at 6 hours of life. He subsequently was transferred to our facility.

Our initial evaluation revealed multiple congenital anomalies (right cleft lip, bilateral cleft palate, inferior left-iris coloboma, hypertelorism, and bilateral ear dysplasia). CHARGE syndrome, suspected by our genetics colleagues on the patient's initial presentation, was confirmed via *CHD7* gene mutation testing. The infant was successfully weaned from mechanical ventilation to supplemental flow from a nasal cannula, but he continued to manifest audible stridor. The initial evaluation at our facility was significant for a right AA on a chest radiograph. A 2-dimensional echocardiogram revealed a possible right-dominant double AA with atresia of the left arch distal to the left subclavian artery. There was additional concern for distal right-AA narrowing before the origin of the right subclavian artery. On Doppler, the peak systolic gradient in the distal right AA was 50 mmHg. The cardiac situs was normal, and no major intracardiac defects were identified, except for a small-to-moderate fenestrated atrial septal

defect. A computed tomographic (CT) angiogram of the chest established the diagnosis of a right-sided AA with a retroesophageal brachiocephalic artery arising from a Kommerell diverticulum, as well as coarctation of the distal right AA near the origin of the right subclavian artery (Fig. 1). Cardiac catheterization revealed a 50-mmHg pressure gradient from the proximal right AA to the thoracic aorta. The aortic root angiogram was consistent with the CT angiogram in showing the area of coarctation in the right AA (Fig. 2).

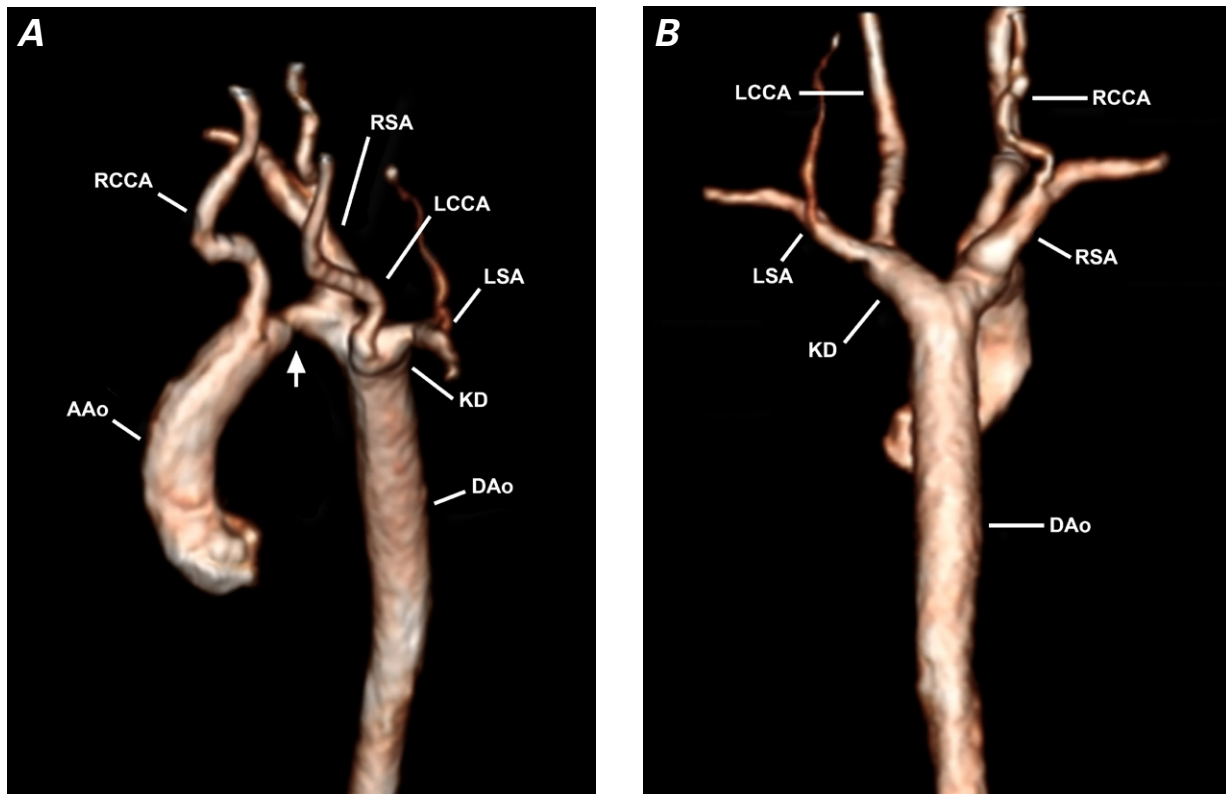
Having confirmed the patient's hemodynamic status by cardiac catheterization, we changed the surgical approach to a midline sternotomy. The infant underwent surgical division of the left ligamentum arteriosum, AA repair with homograft patch augmentation, and atrial septal defect repair. Homograft patch augmentation was performed because of the patient's small size and the inadequacy of native AA tissue to mobilize for an end-to-end anastomosis. There was no immediate postoperative sequela; however, there was a residual 30-mmHg blood pressure gradient from the aortic root to the left radial arterial line, established by direct measurement in the operating room.

Upon follow-up echocardiographic evaluation 6 weeks later, significant recurrent coarctation at the distal end

of the aortic patch produced a peak gradient of 65 mmHg. At the time of cardiac catheterization, the gradient was similar (-52 mmHg). The patient underwent successful balloon dilation of the aorta in the cardiac catheterization laboratory, with a resultant decrease in gradient across the arch to 20 mmHg. Since this procedure, the infant has not needed any further surgical or catheter-based AA intervention.

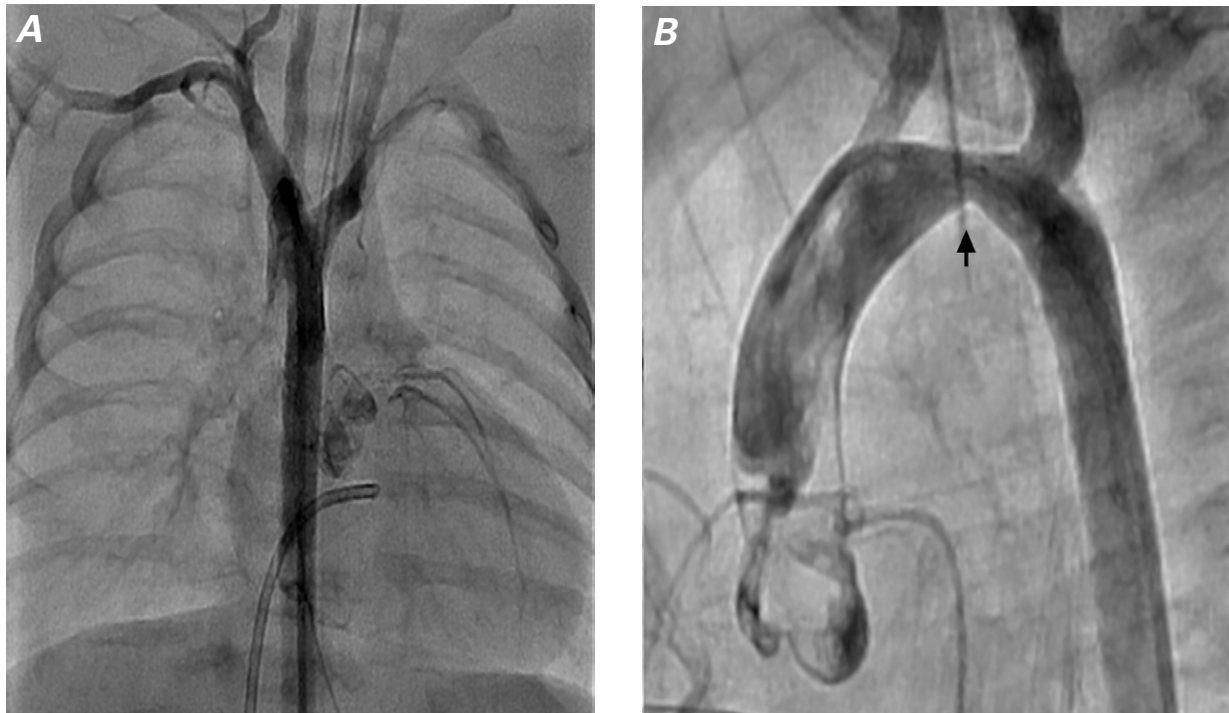
## Discussion

Right AAs have been well described; however, associated AA obstruction is highly unusual and rarely occurs in complex AA anomalies such as the one seen here. There is but a paucity of evidence to suggest a specific origin of causation. Alternative and altered in utero flow patterns could explain why this rare lesion is susceptible to AA obstruction, but the exact mechanism in this particular anomaly remains unidentified.<sup>5</sup> Two of the previous case reports of coarctation with this anomaly include a patient with 22q11 deletion and a patient with features that suggested CHARGE syndrome.<sup>2-4</sup> The association of 22q11 deletion with abnormal AA development is well documented.<sup>6</sup> Abnormal neural crest migration to develop the primitive pharyngeal arches,



**Fig. 1** Three-dimensional computed tomographic angiograms. **A)** Sagittal view shows the area of coarctation in the right aortic arch (arrow). **B)** Posterior view shows the Kommerell diverticulum.

AAo = ascending aorta; DAo = descending aorta; KD = Kommerell diverticulum; LCCA = left common carotid artery; LSA = left subclavian artery; RCCA = right common carotid artery; RSA = right subclavian artery



**Fig. 2** Angiograms of the aortic arch during cardiac catheterization. **A)** Straight anteroposterior view shows a right aortic arch. **B)** Lateral view shows the area of coarctation in the right aortic arch (arrow).

as observed in 22q11 deletion, causes AA embryologic anomalies.<sup>7</sup> Data suggest that this same principle could apply to CHARGE syndrome and lead to AA maldevelopment.<sup>7,8</sup> We recommend considering CHARGE syndrome as a part of the genetic diagnostic process in this type of AA anomaly. Overall, the question of whether coarctation in this anomaly follows an abnormal pattern of in utero circulation or an abnormal predetermined 4th AA development warrants further investigation.

The current report describes the importance of advanced imaging in clearly delineating the anatomy for purposes of surgical planning. In this particular situation, the only arch vessel that originated proximal to the obstruction was the right common carotid artery; thus, upon examination of the extremity, the clinician might be falsely assured of AA patency by the absence of a blood pressure gradient or the lack of a pulse discrepancy. In this case, a direct pressure gradient measured during cardiac catheterization confirmed the findings on CT angiography and the necessity for AA repair at the time of ligamentum division. Echocardiography remains the mainstay for the diagnosis of AA anomalies, and CT angiography and magnetic resonance imaging have complementary roles. Given the complexity of anomalies that are associated with a right AA and the association between right AA and retroesophageal brachiocephalic artery, we recommend advanced imaging before surgical intervention, to fully reveal the presence or absence of AA obstruction.

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