Canadian Journal of Cardiology 35 (2019) 373–375

Editorial

Congenital Aortic Arch Anomalies: Lessons Learned and to Learn!

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See article by Lodeweges et al., pages 438–445 of this issue.

Development of the aortic arch is complex and can result in a heterogenous spectrum of aortic arch anomalies as a result of selective persistence or involution of the paired aortic arches and its branches during embryogenesis to form a definitive aortic arch. Aortic arch anomalies can occur in isolation, in association with congenital heart defects, or in genetic syndromes (eg, 22q11.2 microdeletion syndrome).

Robert Gross first used the term, "vascular ring," in his landmark article in 1945 when he described an earlier autopsy on a 5-month-old baby with respiratory issues and feeding difficulties.[1] At the time of autopsy in 1931, a ring of blood vessels was found that encircled the esophagus and trachea in a fashion that: "the esophagus was indented from behind, whereas the trachea was compressed on its anterior surface. The pathological findings at once suggested that a division of some part of the so-called "vascular ring" during life would probably have relieved the pressure on the constricted esophagus and trachea."[1] Later, in 1945, he applied this knowledge clinically to successfully divide a double aortic arch (DAA) in a 1-year-old child with persistent wheezing and multiple hospital admissions for respiratory tract infections, fever, and stridor.[1]

In this issue of the *Canadian Journal of Cardiology*, Lodeweges et al.[2] describe the different types of congenital aortic arch anomalies (CAAA) according to their anatomical heterogeneity, presenting symptoms, and differences in age at clinical onset and outcome. The authors included 69 affected individuals of 15,602 patients (0.4%) registered in the **Con**genital **Cor**vitia (CONCOR) database (large registry of adults with congenital heart disease in the Netherlands). Although this report inherits all limitations of a retrospective observational study, it provides very useful and helpful information for clinicians: (1) left aortic arch with an aberrant right subclavian artery is the most common anomaly (43.5%), followed by a DAA (30%), and right aortic arch (23.2%); (2) associated congenital cardiac anomalies are frequent (64%); (3) 54% of the population developed symptoms at a median age of 8.5 years (range, 0-53 years) independent of type of arch anomaly ($P = 0.738$); (4) there was an important delay in diagnosis of more than 1 year (median delay, 11 months; range, 1-639 months) in 35% of the patients; (5) respiratory, "asthma-like" symptoms were most frequent, followed by gastrointestinal symptoms (dysphagia for solid food), and were mainly observed in patients with a DAA; (6) patients with a DAA were significantly more frequently symptomatic than patients with other forms of CAAA (85% vs 50% vs 30%; $P < 0.001$); and (7) symptoms were the main indication for surgery in most patients (90%); surgery was performed, without perioperative mortality, mainly in patients with a DAA, followed by right aortic arch with aberrant left subclavian artery and left aortic arch (76% vs 44% vs 17%; $P < 0.001$). Surgery was not performed in 40 patients (58%) because they were either asymptomatic (72.5%), or they had only mild symptoms or a high mortality risk because of concomitant congenital cardiac anomalies (27.5%).

Definition and Classification

Morphological classification is critical, not only for imaging specialists, clinicians, and surgeons to describe CAAA, but also for researchers to compare study populations. CAAA result from abnormal development of the aortic arch and its branches and encompasses a wide, heterogenous spectrum, with or without a vascular ring. Since Gross' description,[1] the phrase, "vascular ring," has been used to refer to a group of CAAA that encircle and compress the esophagus and trachea. By definition, the term refers only to a vascular ring that surrounds the esophagus and trachea, and this term should not be used for aortic arch abnormalities that do not surround the trachea and esophagus. Hence, vascular rings encompass a wide and heterogenous spectrum of aortic arch anomalies (eg, DAA, other vascular structures), all of which compress the trachea or esophagus by forming a complete or partial ring.

There have been various classification schemes in attempts to categorize vascular rings. The classification proposed by

Received for publication January 8, 2019. Accepted January 23, 2019.

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See page 375 for disclosure information.

https://doi.org/10.1016/j.cjca.2019.01.011

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Table 1. Main types of vascular rings and their relative frequency

Type	Lodeweges et al.[2] (n = 69)	Literature	Reference/population
DAA	30.4%	49%	van Son et al.[10] Surgical, pediatric population (n = 37)
		54%	Backer et al.[11] Surgical population (n = 209)
Right	66.7%	75% of all DAA	Backer et al.[11]
Left	4.8%	18% of all DAA	Backer et al.[11]
Balanced	9.5%	7% of all DAA	Backer et al.[11]
Right aortic arch, left ligament, aberrant left subclavian artery	23.2%	46%	Backer et al.[11] Surgical population
		30%	van Son et al.[10] Surgical population
Left aortic arch, aberrant right subclavian artery	43.5%	0.5%-2% of general population	Turkvatan 2009[12]
		11% of vascular ring	van Son et al.[10] Surgical population
Innominate artery compression	1.4%		
Right cervical aortic arch	1.4%		

Note the different denominators (**Con**genital **Cor**vitia [CONCOR] registry vs the surgical population).
DAA, double aortic arch.

Carl Backer and Constantine Mavroudis has described the nomenclature most elegantly and is likely the most widely used system of classification.[3] In anatomic terms, there are 2 types of vascular rings: those that are complete and have vascular structures completely encircling the trachea and esophagus, and rings that are incomplete and only compress a portion of the esophagus or trachea without encircling it fully.[3]

Table 1 includes a summary of the main types of vascular rings and their relative frequencies. However, the true frequencies are difficult to compare because of the different denominators (CONCOR registry, adults with congenital heart disease vs surgical databases). The 2 classic true anatomic vascular rings are DAA, with 3 subcategories, and right aortic arch with a retroesophageal left subclavian artery and left ligamentum. Right aortic arches can be divided into 2 groups: the first is the right aortic arch with left ligamentum and retroesophageal left subclavian artery (true vascular ring). In this group there has been noted to be aneurysmal dilatation at the base of the left subclavian artery, called the Kommerell diverticulum. In many patients the Kommerell diverticulum might enlarge to the point of becoming an independent cause of posterior compression of the trachea and esophagus. The second group of right aortic arch is mirror-image branching where there is a left innominate artery as the first brachiocephalic branch and gives rise to the left carotid and left subclavian arteries (this is not a true vascular ring).[3,4]

The other 2 tracheoesophageal compression syndromes that have been included under the heading "vascular ring," but are an incomplete ring,[2] are innominate artery compression syndrome (the innominate artery compresses the trachea anteriorly by more than 75% of the original tracheal lumen) and the pulmonary artery sling (the left pulmonary artery originates from the right pulmonary artery and courses posteriorly between the trachea and esophagus in its route to the left lung hilum).

One of the most common CAAA (and found to be the most common in this study),[2] is a left aortic arch with aberrant right subclavian artery (arising from the descending thoracic aorta). This is not a vascular ring and is often picked up incidentally on imaging in asymptomatic patients. However, approximately 10% of adults with this anomaly have symptoms of dysphagia resulting from extrinsic compression of the esophagus because of the retroesophageal course of the aberrant right subclavian artery, termed dysphagia lusoria.[4]

Diagnosis Missed and Delayed

The study under discussion confirms our impression as clinicians: the correct diagnosis is often missed and frequently delayed because symptoms vary in degree. One of the important findings of this study is that the median age of clinical onset was 8.5 years whereas the median age at diagnosis was 16 years, which identifies how symptoms can be present for quite some time before correct diagnosis. Symptoms secondary to esophageal and tracheal compression often consist of recurrent respiratory infections, cough, dyspnea, difficulty feeding as an infant, or dysphagia as an adult. Patients who are not diagnosed until adulthood might describe a long history of "asthma" necessitating inhalers and recurrent hospital admissions. This underlines the fact that in patients with suspected asthma who do not respond to guideline therapy, or in whom the diagnosis is uncertain, tracheal pathology (such as subglottic stenosis or a vascular ring) must be considered.[2,4]

With regard to diagnosis, careful review of a patient's chest radiograph might provide a clue if the clinician is astute, because it can be useful in identifying the location of the aortic arch. In patients with a DAA, it might be difficult to discern which side the arch is located; in patients with a right aortic arch, the trachea will be pushed toward the left. A barium study was historically used to diagnose a vascular ring. Today, however, computed tomography or magnetic resonance are the reference methods for advanced imaging.[5-7]

Association With Congenital Heart Defects

CAAA can occur in isolation but might also be associated with structural congenital heart disease. In this study, 64% of CAAA patients had associated congenital cardiac anomalies.[2]

Some aortic anomalies are more strongly associated with cardiac anomalies (eg, right aortic arch with mirror image branching is associated with congenital heart disease in greater than 98% of cases, including tetralogy of Fallot, truncus arteriosus, tricuspid atresia, and transposition of the great arteries).[6,8] However, other arch variants, such as right aortic arch with a retroesophageal left subclavian artery, are more often associated with normal intracardiac anatomy. As well, it has been suggested that some CAAA have a genetic component. Various anomalies, including right aortic arch, cervical aortic arch, aberrant and isolated subclavian artery, and interrupted aortic arch, have been associated with microdeletions of locus 22q11.2.[8] For these reasons, echocardiography should be performed in all patients with an aortic arch anomaly, and genetic testing is recommended in syndromic patients and/or patients with learning difficulties. And last, but not least, patients with aortic arch anomalies should be assessed by a congenital heart disease specialist.

Surgical Treatment

Symptoms are an indication for surgery.[4] Patients with vascular rings due to DAA frequently become symptomatic. Therefore, it is understandable why published series, including this study,[2] described DAA as the most common type of vascular ring requiring surgery. In contrast, vascular rings due to right aortic arch with anomalous left subclavian artery cannot cause symptoms or only mild symptoms because the ring is relatively loose. In the subset of patients who are asymptomatic, watchful waiting is a reasonable approach. In this study, of the patients who underwent surgery, one-third had complete resolution of their symptoms. It must be explained to the patient that in some cases, relief of extrinsic compression might not resolve respiratory symptoms completely, if underlying tracheomalacia is present after a delay of the diagnosis. Similarly, relief of long-standing esophageal compression might not completely resolve dysphagia symptoms if the esophagus does not fully recoil.[4]

Strengths and Weaknesses

The strength of this retrospective observational study is that it is the largest of its type in an adult population. Although the study population is small, an inevitable consequence of the nature of this rare congenital anomaly, the review provides important insight and useful information to the clinician. The study, however, displays the weaknesses of a retrospective study and registry (eg, inclusion bias/report bias, data from many centres and clinicians).

Future Perspective

As in many other rare diseases, international registries and collaboration and/or merging of existing databases to increase the study population might shed further light into aortic arch anomalies and the different clinical practices around the globe. This also emphasizes the importance of an internationally accepted nomenclature and classification.[9]

Acknowledgements

Dr Erwin Oechslin currently holds the Bitove Family Professorship of Adult Congenital Heart Disease. Dr Nita Guron is the recipient of the Gordon B. Fellowship in Adult Congenital Heart Disease, Toronto, Ontario, Canada.

Disclosures

The authors have no conflicts of interest to disclose.

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