Abnormalities of the Aortic Arch

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Abstract

Congenital abnormalities of the aortic arch, which take many forms, result from aberrant development of one or more components of the embryonic pharyngeal arch system and represent less than 1% of all congenital cardiac defects. They occur with about equal frequency in both sexes with no geographical or racial predominance. There are five primary groups of anatomic arch anomalies: double aortic arch, right aortic arch with mirror-image (i.e., normal) branching, right aortic arch with abnormal branching, left aortic arch with abnormal branching, and cervical aortic arch. Aortic arch anomalies are associated with a chromosome 22q11 deletion in approximately 20% of patients.
Clinically, aortic arch anomalies can be divided into those that cause (or are likely to cause) physiologic abnormalities, and those that do not. Physiologic abnormalities resulting from aortic arch anomalies include tracheobronchial compression, esophageal compression, and abnormal blood flow patterns. The location and severity of compression varies with the configuration of the lesion. Prompt diagnosis and treatment of these congenital abnormalities can be lifesaving. Surgical division of the vascular ring is indicated in any patient with symptoms of airway or esophageal compression.

Keywords

aortic arch anomalies; double aortic arch, vascular ring, congenital heart defect, congenital aortic defect, 22q11 chromosome deletion

Definition and Embryology

Congenital abnormalities of the aortic arch, which take many forms, result from aberrant development of one or more components of the embryonic pharyngeal arch system. Early in the course of embryonic morphogenesis, six symmetric pairs of pharyngeal arch arteries develop in conjunction with the branchial pouches, forming the primitive vascular supply to the brachiocephalic structures. As cardiovascular morphogenesis proceeds, a patterned process of regression and growth of different pharyngeal arch components leads to the normal mature configuration of the thoracic aorta and its branches. The mature aortic arch is left-sided and gives rise, in order, to the right-sided innominate artery (which then branches into right common carotid and subclavian arteries), the left common carotid artery, and the left subclavian artery. Any segment of the embryonic aortic arch system can regress or persist abnormally, resulting in an extensive array of aortic arch anomalies.

Etiology

The various forms of this anomaly occur very early in embryologic development. They result from the abnormal or incomplete regression of 1 of the 6 embryonic branchial arches. Evidence

from human genetic studies and animal models suggest that chromosome 22q11 deletions may be an important cause of anomalous development of the aortic arch. Familial recurrence of double aortic arch has been reported, supporting a genetic etiology for this anomaly.

Classification

Aortic arch anomalies can be classified by morphogenetic abnormality or clinical implications. Anatomically, aortic arch anomalies consist of abnormal sidedness or position of the aortic arch (which concerns the course of the aortic arch itself) and abnormal branching of the arch vessels (which concerns the order and pattern of origin of the major branches from the aortic arch). Abnormalities of sidedness/position and branching frequently occur together. There are five primary groups of anatomic arch anomalies: double aortic arch, right aortic arch with mirror-image (*i.e.*, normal) branching, right aortic arch with abnormal branching, left aortic arch with abnormal branching, and cervical aortic arch.

Clinical description

Aortic arch anomalies are developmental abnormalities that are present in the fetus. The postnatal age at which the anomaly is identified may vary, although in most patients, double aortic arch is diagnosed in early infancy. Clinically, aortic arch anomalies can be divided into those that cause (or are likely to cause) physiologic abnormalities, and those that do not. Physiologic abnormalities resulting from aortic arch anomalies include tracheobronchial compression, esophageal compression, and abnormal blood flow patterns. Tracheobronchial and esophageal compression are most often caused by anomalies constituting a vascular ring, which is formed when the abnormally patterned arch vessels completely encircle the trachea and esophagus. The most frequently encountered forms of vascular ring are double aortic arch (which constitutes a spectrum of anomalies, with variable dominance and patency of the right and left arches, and occasional abnormalities of branching), right aortic arch with aberrant origin of the left subclavian artery from a retroesophageal diverticulum, and right aortic arch with mirror-image branching and a left-sided ductus arteriosus passing between the descending aorta and the proximal left pulmonary artery. However, anomalies that do not form a true ring can also cause clinically significant tracheobronchial and/or esophageal compression. The other potential physiologic implication of aortic arch anomalies is anomalous blood flow, which is relatively uncommon and occurs when there is isolation of a subclavian, carotid, or innominate artery (i.e.,

origin of these vessels from the proximal pulmonary artery by means of a ductus arteriosus). Typically, the isolated artery becomes occluded due to closure of the ductus supplying it, and there may be "steal" from the cerebral circulation through the circle of Willis in order to provide arterial flow to the occluded arch branch.

Symptoms and physical findings produced by vascular rings are primarily those of airway or esophageal compression. Individuals with a narrow or tight ring have a significant degree of constriction of one or both of these structures and present very early in life. However, a small number of patients do not manifest symptoms until later in life, and others remain entirely asymptomatic. Common symptoms include stridor, respiratory distress, apnea, and/or a characteristic high-pitched, brassy cough. Additional findings include a history of asthma, recurrent pneumonia, or evidence of dysphagia or difficulty with feedings. In some cases, airway symptoms are worsened or aggravated by feedings. Intercostal retractions during respiration are observed in some infants with severe obstruction. Others may try to maintain a position in which the head is hyperextended to improve breathing and minimize the obstruction. Air-trapping and evidence of pulmonary hyperinflation may also be present in one or both lungs in severe cases.

Symptoms of airway obstruction predominate in patients who present in infancy or the first few years of life. Dysphagia and symptoms related to the esophagus are the more likely presenting findings in older children and adults with vascular rings.

Diagnostic methods

Imaging studies: chest radiography, echocardiography, MRI (the best single imaging study for the diagnosis and characterization of vascular rings). Cardiac catheterization usually is not indicated. Frequently, the diagnosis of a vascular ring is made initially with barium esophagography.

Epidemiology

Vascular rings are uncommon anomalies and make up less than 1% of all congenital cardiac defects. They occur with about equal frequency in both sexes. No geographical or racial predominance exists. The 2 most common types of complete vascular rings are double aortic arch and right aortic arch with left ligamentum arteriosum. These make up 85-95% of the cases.

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Management

In summary, the most common aortic arch anomalies of clinical importance are vascular rings, which frequently become symptomatic in the neonatal or early infant period and require surgical division in order to relieve the tracheobronchial/esophageal compression. A variety of aortic arch anomalies occur in patients with other forms of complex congenital heart disease, such as tetralogy of Fallot, but vascular rings are frequently found alone or in conjunction with ventricular septal defects. Testing for a chromosome 22q11 deletion should be considered in all patients with aortic arch anomalies given the frequent association of this syndrome with aortic arch anomalies and the clinical implications for the patient and family.

Surgical division of a vascular ring is indicated in all symptomatic patients. To avoid serious complications such as or significant tracheal or bronchial damage, surgery should not be delayed, especially in patients with symptoms of airway compression. Prompt diagnosis and treatment of these congenital abnormalities can be lifesaving. Long-term prognosis after surgery is excellent; lifestyle implications are minimal and, most likely, related to residual symptoms or associated anomalies.

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