Spectrum of the congenital anomalies of the thoracic aorta

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Learning objectives

For an accurate interpretation of the radiological findings of the congenital anomalies, it is necessary to understand the anatomy and the embryology. Therefore, we intend to approach these objectives:

- Describe the radiological findings of the congenital anomalies of the thoracic aorta using MDCT and MRI.
- Explain the embryological development of the thoracic aorta and its major branches and identify the errors in the embryological development that causes the congenital anomalies.
Background

Development of the Aortic Arch

In normal embryonic morphogenesis, the primitive circulatory system is initially functional by the end of the third week of fetal development [10]. Therefore, most aortic arch anomalies occur in between the third and eighth week of gestation.

The normal development of the human aortic arch and great vessels involves the formation and selective regression of six paired pharyngeal vascular arches that connects the two embryonic arterial vessels (ventral and dorsal aortas) through the pharyngeal pouches [11] (Table 1 and Fig. 2). During this period, the vascular arches arise in a marked cranio-caudal sequence that conditions their formation and asymmetric regression. At the end of the fifth week of the fetal development, the third, fourth and sixth arches are well developed, and the first and second pairs of pharyngeal arches have already regressed. The fifth arch never develops or appears briefly and then regresses [10] (Fig. 3).

Aortic Arch Anomalies

These are congenital vascular abnormalities that affect the development of the primitive vascular arches and their derivatives, accounting for 0.5% to 1.6% of all congenital heart diseases [10]. The normal development of the aortic arch involves the regression of the right ductus arteriosum, the right dorsal aorta and right aortic arch, with an aorta that descends on the left and has a normal branching pattern [6]. After birth, the ductus arteriosum is left sided and extends from the left pulmonary artery to the aortic isthmus. Subsequently, the ductus constricts and form the ligamentum arteriosum. The most common symptomatic aortic arch anomalies are a double aortic arch, right aortic arch with left ligamentum/ductus arteriosum, followed by a left aortic arch with an aberrant right subclavian artery [10].

Vascular Rings

These are congenital vascular anomalies in which the thoracic vessels and ligamentous structures completely or partially encircle and compress the trachea and the esophagus. Usually, vascular rings result from abnormal persistence and/or regression of the primitive pharyngeal aortic arches, most commonly the third, fourth, and sixth arches.

Some rings are completely vascular, such as the double arch with both arches patent, and other rings, more difficult to conceptualize and diagnose, are completed by a fibrotic
vascular remnant. They may be classified in incomplete or complete vascular rings [8 and 10] (Fig. 3). Some complete vascular rings may have a Kommerell diverticulum, a dilation of the aortic arch at the origin of aberrant subclavian artery (formed secondary to the persistence of a remnant of the regressed side of the embryologic forth pharyngeal arch). Incomplete vascular rings are frequent and most of them are nearly asymptomatic, even if indentation of the esophagus is evident on imaging. Therefore, symptoms depend on the mass effect and obstruction that causes the rings around the esophagus and trachea.

At CT and MR fibrotic vascular remnants are non-enhanced structures, this characteristic makes them difficult to visualize during imaging. However, when part of a symptomatic ring, ligamentous structures lead to traction and compression on the trachea, therefore evaluation of the trachea is essential to identify a vascular ring [8].

**Double aortic arch**

Is the most common symptomatic complete vascular ring and results from the persistence of both the right and left fourth pharyngeal aortic arches and the dorsal aorta. Both arches may be patent, or a segment of one arch may be atretic. The left arch is usually patent rather than atretic and rarely, the right arch is atretic. Right dominance occurs in 66%, left dominance in 16%, and codominance in 17% of the cases [10].

The right aortic arch is usually larger, higher, and more posterior than the left. The proximal descending aorta can be on the right or midline and the rest of the descending aorta is often on the left. The ligament is usually left sided and less commonly, it can be right sided or bilateral and pulmonary arteries are typically normal. Each arch gives rise to the ipsilateral carotid and subclavian arteries, which ascend in a typical symmetrical conformation [8 and 10]. At chest film there is a posterior and bilateral lateral impressions on esophagus and bilateral lateral impressions on the trachea.

In symptomatic patients with double aortic arch the surgical planning, if both arches are patent, is directed toward to transection of the nondominant, the smallest or the atretic arch segment.

**Right aortic arch**

A right aortic arch is defined by the arch crossing over the right main bronchus, with the descending aorta running down by either side of the spine. It occurs from the persistence of the right and regression of the left fourth pharyngeal aortic arches and eighth dorsal aorta segment. A vascular ring may form depending on the level of the left fourth arch regression, the branching pattern of the vessels, the origins and course of the ligamentum
arteriosum and the location of the descending aorta. Three possible patterns of a right aortic arch may occur.

**Right aortic arch with mirror-image branching**

A right aortic arch with mirror-image branching is the second most common type of right arch anomaly and is usually associated with intracardiac defects. It is the result of an interruption of the double aortic arch just distal to the left ductus. Therefore, the aortic vessels originate in a mirror-image fashion, with the left brachiocephalic artery as the first branch, followed by the right common carotid and subclavian arteries.

If a right-sided ductus arteriosus persists, there is no concern for a complete vascular ring [6]. When a left sided ductus arteriosum is present, it usually originates from the innominate artery to the pulmonary artery, traveling anterior to the airway and esophagus and thus creating an incomplete vascular ring. When the left-sided ductus arteriosum originates from the descending aorta or from an aortic diverticulum, courses posterior to the esophagus, and enters the distal main pulmonary artery creating a complete vascular ring [6]. Associated congenital heart diseases with mirror-image right arch are the tetralogy of Fallot (25%), truncus arteriosus (35%) and DiGeorge syndrome [7].

**Right Aortic Arch with an Aberrant Left Subclavian Artery**

This type of anomaly is the most frequent pattern of a right aortic arch and appears in 12-25% of the symptomatic rings (7). Is form when during the embryological development there is a persistent right fourth pharyngeal arch and regression of the left fourth pharyngeal arch between the left carotid and subclavian arteries. This creates a branching configuration where the first artery is the left common carotid, followed by the right common carotid and the right subclavian arteries and at last the left aberrant subclavian artery. The left aberrant subclavian courses retroesophageal and usually originates from a Kommerell diverticulum. Symptoms may develop because of extrinsic compression of the esophagus and trachea by the retroesophageal trajectory of the aberrant left subclavian artery (dysphagia lusoria).

When the ligament is right sided (10% of these patients) or the ductus arteriosum is absent, an incomplete vascular ring is present. When the ligament or ductus arteriosum is left-sided, a complete vascular ring is form [7,8]. The association with congenital heart disease is around 5-12%, and usually occurs in the presence of conotruncal abnormalities such as tetralogy of Fallot or truncus arteriosus [6,7].

**Right Aortic Arch with an Aberrant Innominate Artery**
This rare anomaly is formed when there is a persistent right fourth pharyngeal arch and regression of the left fourth arch before the left common carotid artery, and results in an anomalous left innominate artery.

Contrary to the general rule that the first arch vessel courses contralateral to the side of the arch [6], the first arch vessel is the right carotid artery, followed by the right subclavian artery and retroesophageal innominate artery. When the ligament is right sided, an incomplete vascular ring is present and when the ligament is left sided, a complete vascular ring is formed. Usually, the innominate artery originates from an aortic diverticulum that also supplies a left-sided ductus or ligamentum arteriosum to the proximal left pulmonary artery, completing a vascular ring.

**Left aortic arch**

The normal left aortic arch is formed by the regression of the right fourth pharyngeal arch and persistence of the left fourth arch, eighth dorsal aorta segment and sixth pharyngeal arches. Has a left-sided descending aorta and left-sided ligamentum arteriosum. Two main anomalous patterns may occur.

**Left aortic arch with an aberrant right subclavian artery**

A left aortic arch with an aberrant retroesophageal right subclavian artery is the most common congenital aortic arch anomaly, occurring in approximately 1 in 200 individuals [6]. This anomaly develops when there is an early regression of the right fourth pharyngeal arch between the right common carotid and right subclavian arteries (rather than distal to the right subclavian artery). The right segment of the right dorsal aorta persists, so the right subclavian artery maintains its connection to the descending aorta.

This disposition originates four branches, rather than the normal three branches of the aortic arch. There will not be an innominate artery and the right subclavian artery will have its origin distal to the left subclavian artery, and the descending aorta will be on the left [7]. If there is a left-sided ligament, an incomplete vascular ring occurs. If there is a right-sided ligament or ductus a true vascular ring is formed, even thought, this condition is rare because the right ductus arteriosum almost always disappears [10].

Most affected patients have minimal symptoms and rarely require surgical treatment. The symptoms develop when there is an extrinsic compression of the esophagus caused by the retroesophageal trajectory of the aberrant right subclavian artery, which is commonly referred to as "dysphagia lusoria" or by the subtle airway compression when the complete ring is formed.
Circumflex aorta

In this anomaly there is a left aortic arch with a right proximal descending aorta and a right-sided ductus arteriosus forming an incomplete vascular ring [8]. This anatomy is analogous to the right arch with left proximal descending aorta and left-sided ductus arteriosus, as both constitute a circumflex aorta. The left-sided distal aortic arch and the right proximal descending aorta run posterior to the esophagus and trachea, with a normal arch branching or with an aberrant right subclavian artery.

Other Congenital Anomalies of the Thoracic Aorta

Aortic coarctation

Is a common malformation characterized by a focal obstructive narrowing at the aortic isthmus. It accounts for 1.8-9.8% of congenital heart disease, with an incidence of 5-6% [10]. The narrowing is caused by abnormal fibromuscular ridge, arising from abnormal hyperplasia of the tunica media that protrudes and reduces the vessel lumen [1]. Most classifications are based according to the anatomic relationship of the ductus arteriosum in [1,10]:

Juxtaductal

· The left subclavian artery is the landmark for distinguishing between the more common distal and less common proximal locations.

Preductal region

· Occurs proximal to the ductus arteriosum and is associated with hypoplasia of the aortic isthmus.

· Usually presents in infancy in children less than 1 year of age and causes congestive heart failure.

Postductal region

· Distal to the insertion of the ductus and is likely the result of muscular ductal extension into the aorta.

· Is more common in children greater than 1 year and in adults.

An aortic coarctation is associated with multiple other abnormalities being the most frequent the bicuspid aortic valve (30-40%), Turner syndrome and ventricular septal...
defect [1,10]. It usually causes few symptoms or is asymptomatic, unless there is a critical stenosis or there are acquired complications. Hemodynamic compromise leads to the development of collateral vessels to maintain blood flow to the lower body, being usually via the internal mammary and intercostal arteries [3].

Chest radiography has a low to moderate sensitivity for detection of aortic coarctation, depending on the age of the patient, the degree of narrowing and the presence of associated cardiac defects [10]. Imaging findings include pulmonary venous congestion, prominent aortic arch, inferior rib scalloping, notching or sclerosis secondary to the intercostal collateral vessels that remodels its cortical, and normal or enlarged heart size. At CT, the coarctation is visible as an indentation of the aorta, with prestenotic and poststenotic dilatation.

The treatment depends on the age, clinical presentation and severity of the coarctation. Untreated critical coarctation is potentially lethal, with mortality up to 60% in the first year of life and about 25% of the remainder at age 20. The rest by age 40 have evident complications and survival past 50 years is unusual [5]. Post procedural surveillance is mandatory to monitor for residual coarctation, aortic arch hypoplasia, aneurysm formation at the site of repair, re-stenosis, aortic dissection, and pseudoaneurysm formation [1].

The differential diagnosis is pseudocoarctation. In this entity there will be a dilated and elongated aortic arch with a kinking at the ligamentum arteriosum, absence of collateral vessels and fibromuscular ridge, and there is no hemodynamic compromise [1,10].

**Interrupted aortic arch**

Is the most severe and extremely rare obstructive conotruncal anomaly that manifests neonatally, representing less than 1.5% of all congenital heart diseases [9]. In this anomaly there is a lack of luminal continuity between the ascending and descending thoracic aorta, being the systemic perfusion provided by the ductus arteriosus and/or aortic branch arteries. It is associated with others cardiovascular defects in up to 98% of the cases, such as a patent ductus arteriosum (97% of patients) and ventricular septal defects (90% of individuals) [9].

Is classified into three types depending on the location of the aortic arch discontinuity and the origin of the subclavian artery. Type A is an interruption just distal to the left subclavian artery and makes up one third of cases. Its embryological development results from abnormal regression of the left fourth pharyngeal arch after ascension of the left subclavian artery to its expected position. The type B defect, the most common, occurs between the left common carotid and left subclavian arteries. It takes place when the left fourth pharyngeal arch regresses before normal ascension of the left subclavian artery.
Type C is the rarest type, appearing in 2-5% of the cases, and the interruption develops proximal to the left subclavian artery, between the innominate and left common carotid arteries. This type is the result from the regression of the ventral portion of the left third pharyngeal arch and left fourth arch, with persistence of the ductus caroticus, a structure that normally regresses [2,9,and 10].

CT and MRI findings that suggest this entity are nonvisualization of a portion of the aortic arch and the visualization of a single complete thoracic vascular arch on sagittal image (resembling a "normal" aortic arch). Consequently, sagittal imaging can be misleading if not reviewed carefully and may erroneously suggest the presence of an intact aortic arch in the setting of an interruption. Other findings are the presence of a patent ductus arteriosum and a "V" configuration of the great vessels on coronal imaging.

A neonate with an interrupted aortic arch may appear normal at birth. However, as the ductus closes, systemic blood flow decreases and pulmonary venous pressure rises. This causes congestive heart failure, respiratory distress, systemic hypoperfusion and decreased organ function. For these reason, this malformation needs to be repaired before the closure of ductus arteriosus. If left untreated, it has a mortality rate of more than 90% at the first year of age [10].

**Patent ductus arteriosum**

In the fetus, a patent ductus arteriosus connects the proximal descending aorta with the proximal left pulmonary artery and normally closes soon after birth. If the ductus does not close spontaneously, there is a continuous flow from the aorta to the pulmonary arteries. At CT, the ductus is visualized as a tubular structure extending from the underside of the aortic arch just below the origin of the left subclavian artery to the left pulmonary artery. It may be aneurismal or calcified, findings that may lead to rupture [3].

About 20% of the patients with unoperated large patent ductus arteriosus die by the third decade and the mortality increases to nearly two thirds by the age of 60, usually due to heart failure [5]. In surgically created systemic-to pulmonary circulation such as in a modified Blalock-Taussig shunt, CTA may have a role in evaluating its complication, for example perigraft seroma and pseudoaneurysm. It is also useful to evaluate systemic-to pulmonary connections by bronchial-to pulmonary or transpleural collaterals [2].

**Truncus arteriosus**

Is an uncommon conotruncal anomaly that accounts for 1% of cardiac lesions detected in fetal life [13]. Is the result from a septation failure during the development of the ventricular outlets and the proximal arterial segment of the heart tube. It consists in a single great
artery that originates from the base of the heart, overrides the ventricular septum and supplies the systemic, the pulmonary and coronary blood flow.

It is classified according to the branching pattern of the pulmonary artery (Collett and Edwards). In type I, the main pulmonary trunk arises from the truncal artery just distal to the truncal valve. In type II, there is a common origin of both pulmonary arteries with no main trunk. In type III, the pulmonary trunk is absent and the right and left pulmonary branches arise separately from the posterolateral aspect. Type IV corresponds to pulmonary atresia with ventricular septal defect and multiple major aorto-pulmonary collateral arteries [12,13].

In imaging, the morphologic features that allow the diagnosis of a truncus arteriosus are a single arterial trunk (larger in diameter than the normal aorta at a comparable age) positioned above the ventricular septum, abnormal truncal valve with two to five leaflets that are variably stenotic or incompetent, an atretic pulmonary valve, the conal septum is usually absent and there is a ventricular septal defect. An interrupted aortic arch is a common associated lesion, as are abnormalities of the mitral valve, coronary arteries, and pulmonary venous connections [12].

The prognosis is poor in untreated patients with more than 90% of mortality before the first year of age [13]. The main complications after truncal repair are right ventricle-pulmonary artery conduit stenosis or regurgitation, branch pulmonary artery stenosis, neo-aortic valve insufficiency or stenosis, ventricular septal defect patch leak and aortic arch obstruction [12].

Complete or D-transposition of the great arteries

Is a cyanotic condition that accounts for 5-7% of the cases of congenital heart disease [3]. This anomaly is characterized by ventriculoarterial discordance and atrioventricular concordance. The aorta arises from the morphologic right ventricle and is anterior and to the right of the pulmonary artery, which arise from the morphologic left ventricle. This malformation results in two separate circulations, one from the right ventricle through the aorta to the body and back to the right heart, and the other from the left heart to the lungs and back to the left heart.

This condition results in the death of about 90% of untreated infants in the first year of life [4]. But the prognosis improves, at least temporarily, if there is a second lesion such as a ventricular septal defect that allows a mixture of blood from the two circulations. This finding was the base for the atrial blood flow switch operations like the Senning (1959) and Mustard (1963). Either of these procedures was often performed in the first year of life and had a 10-year survival of 85-90% and about 80% at 20 years [4]. Then
in 1975, the Jatene operation demonstrated the feasibility of correcting completely the transposition by switching the aortic and main pulmonary artery trunks to redirect the ventricular outflow, reestablishing the normal circulatory pathway and each ventricle returned to its normal physiologic function.

**Congenitally corrected or L-transposition of the great arteries**

This anomaly accounts for 0.5% of congenital heart lesions and is characterized by atrioventricular and ventriculoarterial discordance (double discordance). This means that there is an inversion of ventricles and great arteries, with the morphologic right ventricle located on the left giving rise to the aorta (which is anterior and to the left of the main pulmonary artery). And the morphologic left ventricle is situated on the right and gives rise to the pulmonary trunk. As a result, blood flow through the pulmonary and systemic circuits occurs in normal series fashion.

L-transposition is associated with other congenital defects such as ventricular septal defect, pulmonary stenosis, tricuspid valve malformations and atrioventricular conduction abnormalities. Right heart failure is a late complication related to the high resistance systemic arterial circulation. [3]
Fig. 1: A. Embryonic pharyngeal aortic arches, cervical intersegmental arteries, ventral and dorsal primitive aorta. B. 3 to 4 weeks of fetal development. At this point the first and second pharyngeal arches have regressed and the third, fourth and sixth aortic arches have appeared in a marked craniocaudal sequence. The fifth arch never develops or appears briefly and then regresses. C. 5 weeks of fetal development. The dorsal aortic segments between the third and fourth pharyngeal arches begin to regress. The internal carotid arteries are formed by the ventral aorta and the third pharyngeal arches.

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Fig. 2: A. 6 weeks of fetal development. The dorsal aorta between the third and fourth arches has regressed. Connections between the first six cervical intersegmental arteries have regressed and the 7th intersegmental arteries enlarge and start to become the subclavian arteries. The pulmonary arteries arise from the sixth arches. B. 7 weeks of fetal development. The fourth and six arches undergo asymmetric remodeling. The left fourth pharyngeal arch and the left dorsal aorta, creates the definitive aortic arch and the cranial portion of the descending aorta. The right dorsal aorta (distal to the seventh cervical segmental artery) gradually loses its connection with the right fourth arch and the definitive descending aorta. The 7th cervical intersegmental artery and the remaining of the right dorsal aortic segment (distal to fourth arch) develop the right subclavian artery.

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**Embryology of the Normal Left Aortic Arch [7, 10]**

<table>
<thead>
<tr>
<th>Pharyngeal arches</th>
<th>Description</th>
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<tr>
<td>I and II arches</td>
<td>Creates the external carotid arteries.</td>
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<tr>
<td>III arches</td>
<td>Bilaterally: develops the common carotid arteries and the proximal segment of the internal carotid arteries. Right: proximal portion of the innominate artery.</td>
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<tr>
<td>IV arches</td>
<td>Right: forms the proximal portion of right subclavian and the innominate arteries. Left: persists as the definitive left aortic arch.</td>
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<tr>
<td>V arches</td>
<td>Rudimentary in humans.</td>
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| VI arches         | Right - suffers a partial regression in utero.  
|                   | - Ventral portion: right pulmonary artery.  
|                   | - Dorsal portion: regress.  
|                   | Left - has a dominant persistence.  
|                   | - Ventral portion: left pulmonary artery.  
|                   | - Dorsal portion: ductus arteriosum that later becomes ligamentum arteriosum. |
| Dorsal aorta      | Remnant of the right - part of right subclavian artery and distal segment of the right internal carotid artery.  
|                   | Left - becomes the descending aorta. |
| 7th intersegmental arteries | Bilateral persistence.  
|                   | - Right: distal portion of the right subclavian artery.  
|                   | - Left: forms the left subclavian artery. |

**Fig. 3: Table 1. Embryology of the Normal Left Aortic Arch.**

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**Complete Vascular Rings**

- Doubled aortic arch.

- Right aortic arch with an aberrant left subclavian artery and left-sided ligamentum/ductus arteriosum.

- Right aortic arch with mirror-image branching and a left-sided ductus arteriosus that originates from the descending aorta.

- Left aortic arch with an aberrant right subclavian artery and a right sided ligamentum/ductus arteriosum (rare).

**Incomplete Vascular Rings**

- Right aortic arch with an aberrant left subclavian artery and a right-sided ductus/ligamentum.

- Right aortic arch with mirror-image branching and a left-sided ductus arteriosus that originates from the innominate artery.

- Left aortic arch with aberrant right subclavian artery and left ductus/ligamentum

- Innominate artery compressing the trachea.

- Circumflex aorta.
**Fig. 4:** Table 2. Vascular rings.

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**Fig. 5:** Double aortic arch. A. Embryological development: Persist both 4th pharyngeal aortic arches and the dorsal aorta. B and C. Both arches are patent with a proximal descending aorta on the midline. From each arch rises the ipsilateral carotid and subclavian arteries, which ascend in symmetrical conformation (red arrows).

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Fig. 6: Right aortic arch with mirror-image branching. A. Embryological development: There is an interruption of the double aortic arch distal to the left ductus. B, C and D. Mirror-image fashion of the aortic arch branching. First comes the left innominate artery (1), then the right common carotid (2) and the right subclavian arteries (3). The findings form an incomplete vascular ring.

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**Fig. 7:** Right aortic arch with an Aberrant Left Subclavian artery

A. Embryological development: The right 4th pharyngeal arch persist, with regression of the left 4th arch between the left carotid and subclavian arteries. B and C. The left aberrant subclavian (orange arrow) originates from a Kommerell diverticulum (green arrow) and courses retroesophageal forming an incomplete vascular ring. D. Extrinsic compression of the esophagus and trachea by the retroesophageal trajectory of the aberrant left subclavian artery (yellow arrow head).

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Fig. 8: Left aortic arch with an aberrant Right Subclavian artery A. Embryological development: The right 4th pharyngeal arch regress between the right common carotid and right subclavian arteries and a segment of the right dorsal aorta persists, connecting the right subclavian artery to the descending aorta. B and C. In this anomaly there are four arch branches: 1. Right carotid artery, 2. Left carotid artery, 3. Left subclavian artery, 4. Right aberrant subclavian artery. There is not an innominate artery.

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Fig. 9: Aortic Coarctation A and B. Focal obstructive narrowing at the aorta in the juxtaductal region with prestenotic and poststenotic dilatation. Yellow arrows: subclavian arteries. C. Chest film shows hiliar prominence and inferior rib scalloping/sclerosis (red arrows).

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**Fig. 10:** Interrupted aortic arch A and B. There is a lack of continuity between the ascending and descending thoracic aorta (yellow arrows) located distal to the left subclavian artery with a persistent 5th pharyngeal aortic arch (blue arrow head), corresponding in a type A arch interruption.

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Fig. 11: Patent ductus arteriosum A and B. Aneurismatic ductus (red *) connecting the left pulmonary artery (yellow arrow) and the proximal descending aorta just below the origin of the left subclavian artery. Black arrow head main pulmonary artery.

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**Fig. 12:** Truncus Arteriosus A, B and C. Single great artery that originates from the base of the heart with both pulmonary arteries arising separately from the posterolateral aspect of the great artery (type III).

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Fig. 13: D-transposition of the Great arteries A. CT in patient who underwent the arterial switch operation (Jatene). Notice the typical post-operative anatomy with the main pulmonary artery anterior to the ascending aorta. B. Pulmonary artery and ascending aorta runs in the same plane with lack of normal spiraling around each other.

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Imaging findings OR Procedure details

A Philips 1.5 tesla MRI was used to perform the angiographies and cardiac studies. Also a Philips Ingenuity CT was used, applying low-radiation-dose CTA when required and contrast media. Afterwards, isotropic 3D reconstructions were made.
Conclusion

Knowing the embryological development of the thoracic aorta helps to understand the anatomical findings that can be visualized by MRI and MDCT studies.

It is essential to recognize and diagnose these aortic features, because in some cases they may require surgical corrections.
References