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Aberrant right subclavian artery - a marker for chromosomal abnormalities?

- review -

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ABSTRACT.
The aberrant right subclavian artery or the aberrant right subclavian artery syndrome is a rare anatomical variant of the right subclavian artery origin but it is the most common form of congenital anomaly of the aortic arch. The normal or abnormal appearance of the aortic arch can be explained by the regression or the persistence of one or other of the two embryonic aortic arches. The regression of the right aortic arch after the origin of the right subclavian artery will result in a normal aspect of left aortic arch with normal vascular anatomy. While the right ductus arteriosus is regressing, the left one persists. This anomaly is apparently found in the general population in approximately 0.5-1.4% of the cases and in approximately 3% of cases with congenital heart malformations.
The present review is focus on the prevalence of aberrant right subclavian artery in Down syndrome. Until present it is known that ultrasound evaluation in the screening programs for Down syndrome is of uncertain value and studies on larger number of cases are reconsidered. In addition we suggest that the echocardiographic examination in the first and second trimesters should be included as a first step of the diagnosis in such congenital anomalies.

Keywords: subclavian artery, congenital anomaly, Down syndrome, ultrasound, echocardiography.

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Introduction

Typically, from the transverse portion of the aortic arch three major arterial trunks will rise: the brachiocephalic trunk (from which the right subclavian artery and the right common carotid artery arise), the left common carotid artery and the left subclavian artery [1]. In case of developmental abnormalities, four trunks will rise from the aortic arch: the right common carotid artery, the left common carotid artery, the left subclavian artery and the right subclavian artery, the latter having its origin in the proximal portion of the descending aorta at the junction with the ductus arteriosus. Its origin is usually in a more dilated portion of the aorta, called the 'Kommerell diverticulum' [2]. The aberrant artery, which has its origin in the left half of the chest, will cross the midline in the posterior part of the mediastinum on its way to the proximal part of the right upper limb, usually behind the esophagus, producing a vascular ring around the trachea and esophagus that can clinically manifest by dysphagia (also known as dysphagia lusoria or Bayford-Autenrieth dysphagia) [3]. Recurrent laryngeal nerve paralysis given by the abnormal position of the artery is called the Ortners syndrome. It was not observed any clinical implication of the abnormal origin and path of the aberrant right subclavian artery [2].

The embryological theory of the aortic arch abnormalities

The physiopathological understanding of this aortic arch abnormalities development is facilitated by understanding the embryological development of the aortic arch. Edwards developed a theory that in early embryonic life the ascending aorta is split in two aortic arches (right and left) that will further cross together to give rise to the descending aorta. These two arches are located anterior to the vertebral column [2]. These two arches form a vascular ring around the trachea and esophagus. Each of them will give rise to two vessels: the right and left common carotid arteries and the right and left subclavian arteries, respectively. The right and left pulmonary arteries are connected by two ductus arteriosus to the corresponding right and left aortic arches near the origin of the right and left subclavian arteries [4].

The aberrant right subclavian artery is resulting from the partial regression of the right aortic arch between the origin of the right common carotid artery and the right subclavian artery. Thus, the left aortic arch gives rise to the right common carotid artery, the left common carotid artery, the left subclavian artery and the aberrant right subclavian artery (Figure 1). While the right ductus arteriosus is regressing, the left one persists.

The implication of aberrant right subclavian artery in Down syndrome

Pathological and clinical studies show that in cases of Down syndrome, the incidence of the anomaly is greatly increased, ranging between 2.9 and 100% in those with other associated cardiac malformations and from 0 to 5.4% in cases where this is the single cardiac abnormality [5]. The first observation about this association was made by Chaoui and contributors [1]. Recent prenatal ultrasound studies indicate an incidence of aberrant right subclavian artery diagnosis in the second trimester of pregnancy of around 35% in fetuses having 21st trisomy and of only 1-4% among fetuses with no chromosomal abnormalities [6]. First trimester screening for 21st trisomy by the combination of nuchal translucency with maternal serum levels of free-β human chorionic gonadotropin and pregnancy-associated plasma protein A has a detection rate of 90% with 5% false-positive results [7]. Associating additional ultrasound markers such as nasal bone presence or absence, the frontomaxilar angle value, the aspect of tricuspid valve or ductus venosus flow, the detection rate can increase to over 95% with less than 3% false-positive results. The first trimester ultrasound scan between 11-13 weeks can only identify the presence and position of the right subclavian artery in 80% of cases [2].

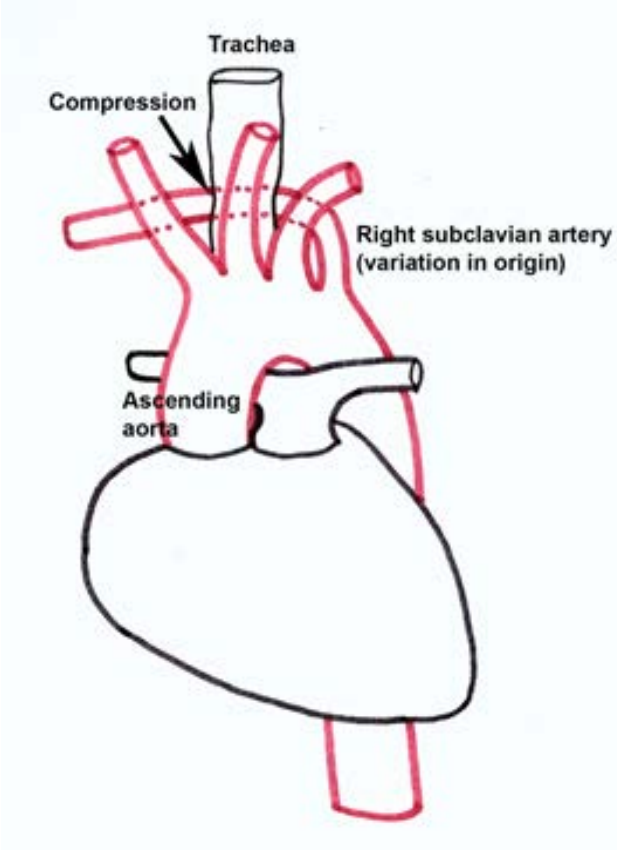

Figure 1 - The aberrant right subclavian artery

Regarding the ultrasound examination, the aberrant right subclavian artery can be considered a variant of the normal. Color Doppler examination is very useful when this diagnosis is suspected. The best examination section is obtained by the trachea and the three great vessels (pulmonary artery, superior vena cava and aorta) where the origin of the vessel at the aortic level may appear, along the ductus arteriosus as well as its route behind the trachea toward the right

clavicle and shoulder (Figure 2). The presence of the vessel is confirmed by demonstrating the presence of blood flow on pulsed Doppler [8]. Using transvaginal ultrasound between 11 and 14 weeks significantly increases the ability to make this diagnosis. While tridimensional ultrasound may be useful in a small number of cases, the color Doppler examination is sufficient only for the diagnosis in most of the cases.

Figure 2 – Transverse view of the upper chest. Images show the aberrant right subclavian artery (ARSA) which arises as a 4th branch of the aortic arch, behind the trachea, below the level of the aortic arch, at the level of the ductus arteriosus. The left subclavian artery (LSA) arises from the aortic arch as a 3rd branch.
Note that the Doppler velocity should be changed from the setting typically used in cardiac Doppler (max velocity usually around 50-55 cm/s) to slower velocities (around 18 cm/sec) in order to detect the flow in the aberrant right subclavian artery [9]

Ultrasound markers associated with this anomaly and 21st trisomy are the intracardiac hyperecogenic spots and the absence of the nasal bone. The aberrant right subclavian artery may also be associated with other chromosomal abnormalities. Its association with a trunk abnormality increases the risk for 22q11 deletion of the long arm of the chromosome [1].

The most important differential diagnosis is the azygos vein that passes posterior to the trachea before reaching the superior vena cava, which could complicate the distinction between the two vessels [5].

Conclusion remarks and future outlook

The prevalence of aberrant right subclavian artery in Down syndrome is around 37%. Including the ultrasound evaluation in the screening programs for Down syndrome is of uncertain value and studies on larger number of cases should be made, but it can be included in the echocardiographic examination in the first and second trimesters.

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