Giant Ascending Aortic Aneurysm: Prenatal Echocardiographic Diagnosis: A Case Report and Review of the Literature

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Abstract: Congenital aneurysms of ascending aorta are unusual. We present a case report of a 32-year-old pregnant woman, 33 weeks of gestation, referred to perform a fetal echocardiogram with cardiomegaly and fetal right atrium enlargement, observed in a routine obstetric ultrasound. Fetal echocardiogram revealed a giant ascending aortic aneurysm. A female neonate spontaneous delivered at 36 weeks of gestation required mechanical respiratory assistance for respiratory distress and paradoxical respiratory movements. Partial sternal cleft and anomalous umbilical cord implantation at supraumbilical abdominal midline was founded. The diagnosis of ascending aortic aneurysm was confirmed by postnatal transthoracic echocardiography (TTE) and cardiovascular multislice computed tomography (MSCT). The use of echocardiogram and Doppler color flow mapping as well as the multislice computed tomography to establish a detailed morphologic analysis are discussed.

Keywords: Congenital aneurysms of ascending aorta, Fetal echocardiography, Transthoracic echocardiography, Doppler color flow mapping, Multislice computed tomography, PHACE syndrome, Sternal cleft, Anomalous umbilical cord implantation.

INTRODUCTION

32-year-old pregnant woman, 33 weeks of gestation was referred to perform a fetal echocardiogram. In a routine obstetric ultrasound, a fetal cardiomegaly and right atrium enlargement was observed. His maternal history showed five pregnancies, two spontaneous abortions and diabetes mellitus type I. The fetal 2-dimensional (2D) Doppler echocardiogram reveled a giant ascending aorta aneurysm measuring 28,6 mm in diameter originated 4 mm above the sinotubular junction (Figure **1**) and extended to the origin of the brachiocephalic trunk (BcT) which was included in the aneurysm; the remaining portions of the aortic arch and descending aorta were normal. There was no family history of Marfan syndrome or other connective tissue diseases.

At 36 weeks of gestation, a female was born by spontaneous delivery weighting 3090 g. and Apgar scores were 7 and 8 at 5 and 10 minutes, respectively. Due to respiratory distress and paradoxical respiratory movements, a mechanical respiratory assistance was required. At middle anterior thoracic wall, partial sternal cleft was found, and a depressed erythematous "V" shape zone on the manubrium sterni region with ectopic beat was observed. No bone or cartilage was palpable in this area (Figure **2**). An anomalous umbilical cord implantation was seen at supraumbilical abdominal midline (Figure **2**). The brain and abdominal ultrasonography were normal.

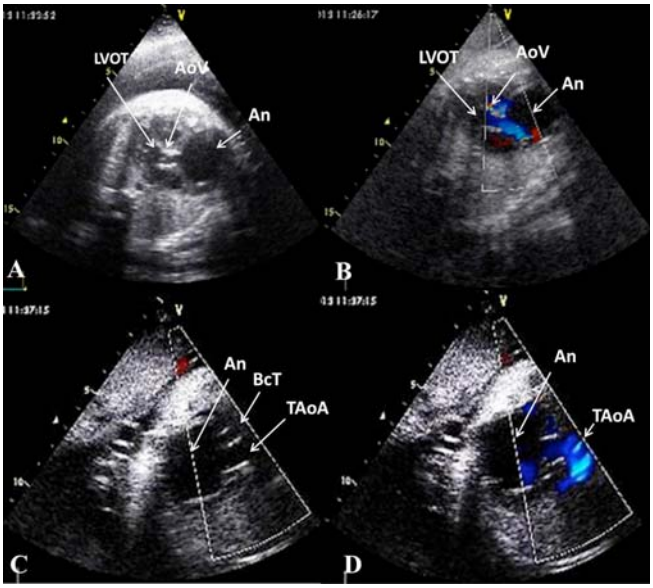

Figure 1: Fetal echocardiographic features 2D and Doppler color flow mapping. **A** and **B**: Transversal views showing left ventricle outflow tract (LVOT), aortic valve (AoV) and aortic ascending aneurism (An). **C**: Transversal echocardiographic view showing aortic ascending aneurysm (An), brachiocephalic trunk (BcT) and transversal aortic arch (TAoA). **D**: Transversal echocardiographic view and Doppler color flow mapping showing the ascending aortic aneurysm and transversal aortic arch.

Postnatal 2D Doppler echocardiogram demonstrated a non-hemodynamically significant patent ductus arteriosus and 4 mm. ostium secundum interatrial septal defect with left to right shunt. A giant ascending aortic aneurysm measuring 30 mm. in diameter

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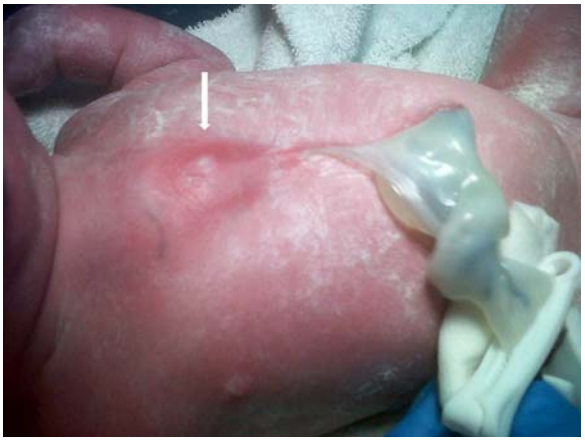

Figure 2: Picture of the patient chest walls showing superior sternal cleft, depressed erythematous "V" shape zone on the sternal manubrium region (arrow) and anomalous umbilical cord implantation.

originated 4 mm above the sinotubular junction and extended to the origin of the brachiocephalic trunk partially included in the aneurysm. The remaining aortic arch including the left common carotid artery, the left subclavian artery and the aortic isthmus appeared normal. The aortic valve, aortic annulus and the aortic root were normal without transvalvular pathologic gradients (Figure **3**).

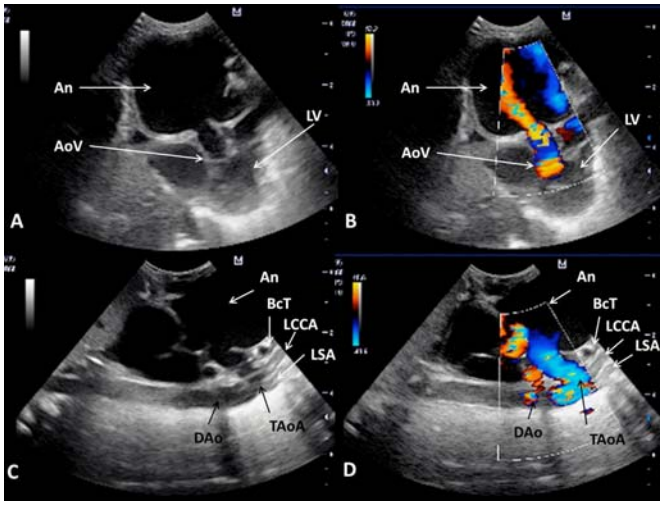

Figure 3: Postnatal echocardiographic features 2D and Doppler color flow mapping. **A** and **B**: Suprasternal short axis views showing left ventricle (LV), aortic valve (AoV) and aortic ascending aneurism (An). **C** and **D**: Suprasternal long axis views showing aortic ascending aneurysm, brachiocephalic trunk (BcT), left common carotid artery (LCCA), left subclavian artery (LSA), transversal aortic arch (TAoA) and descending aorta (DAo).

The third day of life the patient was referred to the regional cardiovascular reference hospital where a cardiovascular MSCT confirmed the prenatal and postnatal echocardiographic findings (Figure **4**).

An ascending aortic aneurysm 33 mm. long and 32 mm. large and including the proximal portion of the brachiocephalic trunk without involvement of the aortic root was observed as well as absent collarbone and sternal manubrium with superior sternal cleft. The sternal body was fused only at the bottom.

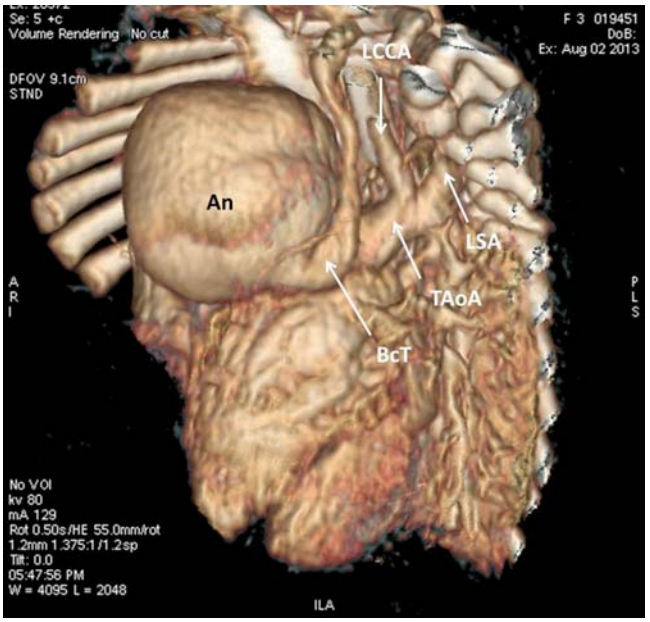

Figure 4: 3D volume rendering images of cardiovascular MSCT showing aortic ascending aneurysm (An), transversal aortic arch (TAoA), brachiocephalic trunk (BcT), left common carotid artery (LCCA) and left subclavian artery (LSA).

The reference hospital decided to admit the patient for surgical treatment. A resection of the aneurysm and replacement of the ascending aorta with autologous pericardial patch and re-implantation of the brachiocephalic trunk was performed. The patient died three hours later after surgery by multiorganic failure due to vasoplegic shock and no genetics or anatomophatological studies were performed.

DISCUSSION

The ascending aorta aneurysm in the fetus and the neonate is an uncommon disorder that presents a clear challenge for its treatment and can be observed isolated or associated to different genetic syndromes with neurological, cardiac, dermatologic or skeletal affectations [1-9].

The different genetic syndromes associated with aortic ascending aneurysm and reported in the literature are Marfan syndrome [2, 10, 11], Ehlers–Danlos syndrome [2], Loeys–Dietz syndrome [12], cutis laxa [13], PHACE syndrome [13-16], congenital–idiopathic aneurysm [1], familial thoracic aortic

aneurysm [12, 17] and Cantrell´s pentalogy [18] as well as sternal cleft [1, 6, 7, 14, 17], and midline abnormalities [6, 19].

PHACE is an acronym for a neurocutaneous syndrome encompassing the following features: posterior fossa anomalies, hemangiomas of the face, cardiac anomalies, arterial lesions, eye abnormalities and ventral developmental defects, such as sternal cleft or supraumbilical raphe [13-16].

Consensus Statement on Diagnostic Criteria for PHACE Syndrome stratified these criteria into two categories: 1) PHACE syndrome and 2) possible PHACE syndrome [20]. Major and minor criteria were determined for the following organ systems: cerebrovascular, structural brain, cardiovascular, ocular, and ventral/midline. PHACE syndrome requires the presence of a characteristic segmental hemangioma or hemangioma >5 cm on the face or scalp plus one major criterion or two minor criteria. Possible PHACE requires the presence of a hemangioma >5 cm on the face or scalp plus 1 minor criterion. The group recognized that it may be possible to have PHACE syndrome with a hemangioma affecting the neck, chest, or arm only or no cutaneous hemangiomas at all. In such cases, fulfillment of additional required criteria would also lead to a possible PHACE diagnosis [20].

In addition to the ascending aorta aneurysm, our patient had absence of collarbone and sternal manubrium and superior sternal cleft. Although she had two major criteria, it showed no changes in the cerebral ultrasound nor cutaneous hemangiomas affecting the face, neck, chest or arms to match criteria to possible PHACE diagnosis.

Three other associated syndromes were excluded: familial thoracic aorta aneurysm because we did not find familiar history of aortic aneurysm or dissection; Cantrell´s pentalogy because this condition is associated with partial lower sternal cleft and Marfan syndrome due to fact that no marked elongation of the long bones was observed in the fetal ultrasound [11] nor clinical postnatal examination.

The postnatal echocardiography and Doppler color flow mapping and cardiac multislice computed tomography (MSCT) play an important role to confirm the diagnosis, delineate the extent of the aortic aneurysm and rule out other anomalies. Hirata and coworkers [2], suggest that the surgery is indicated to avoid possible aortic rupture, correct the sternal cleft to protect the intrathoracic organs and improve respiratory mechanisms. We did not find in the literature to our scope, any reference to an intrauterine treatment [21].

CONCLUSSION

In summary, the prenatal echocardiographic and Doppler color flow mapping diagnosis permit to exclude other associated anomalies, follow the progression of the lesion and planning the perinatal management. About the management of these patients, there are few case reports in the literature, but all match that surgery [7, 8, 22] or endovascular treatments [4] are indicated only when the aneurysm presents signs of dissection, compresses other thoracic structures or enlarge.

CONFLICT OF INTEREST

The authors declare that there is no conflict of interest.

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