J. Fetal Med. (March 2016) 3:31–34
DOI 10.1007/s40556-016-0076-0

BRIEF COMMUNICATION

Ventricular Septal Defect with Overriding Great Artery, Differential Diagnosis, Case Presentation, and Review of Literature

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Received: 29 December 2015 / Accepted: 2 February 2016 / Published online: 27 February 2016

Abstract Embryologically, conotruncal abnormalities result from abnormal development of the conotruncal septum expressing variably as double outlet right ventricle, tetralogy of Fallot, absent pulmonary valve syndrome, complete transposition, corrected transposition, and pulmonary atresia with ventricular septal defect (PA-VSD). It is not very uncommon to see a VSD with one single large artery overriding the defect, along with nonvisualization of the right ventricular outflow tract and ventricular origin of the Pulmonary artery. In situations with above findings and a near normal four chamber heart view the differential diagnosis include common arterial trunk (CAT), and pulmonary atresia with VSD and rare disorders like aortic atresia. CAT is characterized by a single great artery supplying the coronary, cranial and pulmonary circulation. Similar findings in PA-VSD may create confusion in prenatal diagnosis of these two conditions, more so in CAT type II/III. The diagnosis of CAT type I is comparatively easy as the pulmonary artery arises immediately beyond the origin of the trunk. This article discusses a case, the approach to and differential diagnosis of a single great artery overriding a VSD with a near normal four chamber view. The role of cardiac screening during the early morphology scan and the role of 4D imaging with color and power Doppler angiography are also highlighted for the correct diagnosis and description of spatial relationships of the vessels and chambers. Correct prenatal diagnosis of these conditions is important for patient counseling and prognostication.

Keywords VSD · Override · Pulmonary atresia · Common arterial trunk · Bilateral SVC · Pulmonary atresia with VSD · MAPCA

Introduction

It is not very uncommon to see a ventricular septal defect (VSD) with one single large artery overriding the defect, along with non visualization of the right ventricular outflow tract and ventricular origin of the pulmonary artery. In situations with the above findings and a near normal four chamber heart view, the differential diagnosis includes common arterial trunk (CAT), and pulmonary atresia with VSD (PA-VSD) and rare disorders like aortic atresia. CAT is characterized by a single great artery supplying the coronary, cranial, and pulmonary circulation. Similar findings in PA-VSD may create confusion in prenatal diagnosis of these two conditions, more so in CAT type II/III. The diagnosis of CAT type I is comparatively easier as the pulmonary artery arises immediately beyond the origin of the trunk. Aortic atresia with VSD may be evident by demonstration of a very small calibre aorta receiving reverse flow from the ductus. This article discusses a case, the approach to, and differential diagnosis of a single great artery overriding a VSD with an apparently normal four chamber view.

Electronic supplementary material The online version of this article (doi:10.1007/s40556-016-0076-0) contains supplementary material, which is available to authorized users.

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Table 1 Features favoring the diagnosis of PA-VSD in cases of single great artery overriding a VSD

Normal semilunar valve morphology
Absence of dysplastic valve with stenosis and/or reflux
Overriding vessel committed equally to both ventricles
MAPCA
Demonstration of a very small PA from RV with reverse flow from the ductus

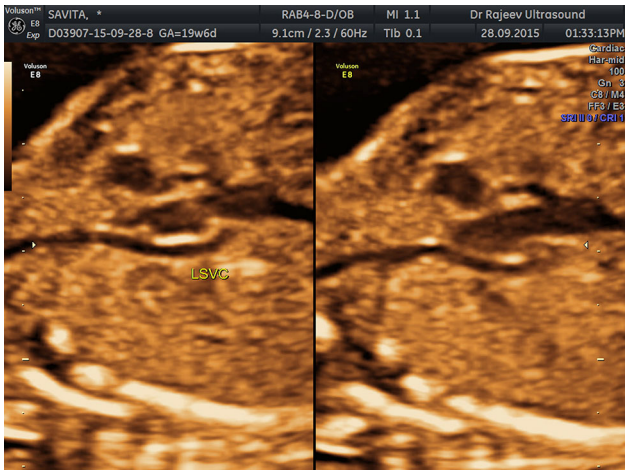

Fig. 1 Persistent Left SVC

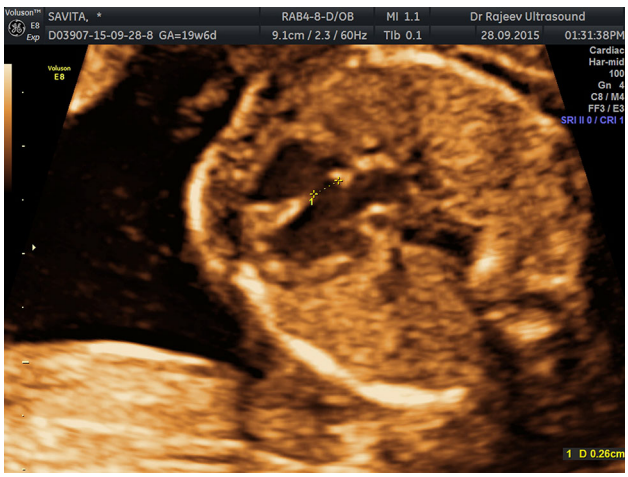

Fig. 2 Four chamber view demonstrating a large VSD

Report of Case

A 26-year-old primigravida with 4 months amenorrhea presented to us for an anomaly scan. This was a spontaneous conception with previous scans including the early morphology scan (nuchal translucency scan) was unremarkable. Her combined first trimester risk assessment done elsewhere, performed on a nonFetal Medicine foundation (FMF) accredited platform showed a low risk for trisomy 21 (1:3098), trisomy 18 (1:100,000) and trisomy 13 (1:100,000). Her nuchal translucency was reported as 2.0 mm at a CRL of 64 mm, the nasal bone had been reported present, there was no tricuspid regurgitation, and no reverse flow in ductus venosus 'a' wave.

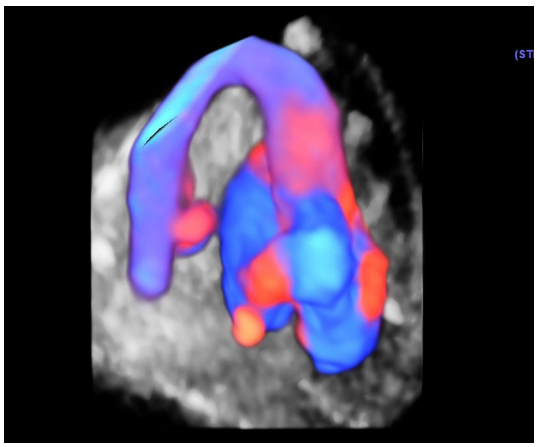

Fig. 3 3D power doppler angiogram showing a single large vessel, committed to both ventricles. MAPCA are seen arising from the descending aorta

The four chamber view and outflow tracts were not commented upon in this early morphology scan done elsewhere. The current scan demonstrated an 18 weeks 4 days single live gestation. Cardiac assessment revealed a bilateral superior vena cava (SVC) with the persistent left SVC draining the right atrium (RA) through a dilated coronary sinus (Fig. 1). The four chamber view was unremarkable otherwise.

A slightly angulated four chamber view demonstrated a 3 mm outlet VSD (Fig. 2) with a single large artery overriding the VSD and receiving inflow from both the ventricles. The pulmonary arteries could not be demonstrated arising from the right ventricle or proximal overriding large artery. The ductus arteriosus (DA) could not be identified. Careful retrospective analysis of the saved 4D STIC volumes with power Doppler (HD flow, Voluson E8) demonstrated two symmetric vessels arising from the mid descending thoracic aorta supplying either lung [major aortopulmonary collateral arteries (MAPCA)] (Fig. 3). Atrial and visceral situs were confirmed and no other cardiac or visceral abnormality was identified. The thymus appeared normal in size and echogenicity.

Genetic amniocenetsis was performed including FISH analysis for 22q11 microdeletion. The karyotype turned out to be normal. The patient however, opted for termination of pregnancy. A diagnosis of pulmonary atresia with VSD was suspected. Fetal autopsy though offered, was not accepted by the family.

Discussion

Embryologically, conotruncal abnormalities result from abnormal development of the conotruncal septum expressing variably as double outlet right ventricle, tetralogy of Fallot, absent pulmonary valve syndrome, complete transposition, corrected transposition, and pulmonary atresia with VSD and CAT [1, 2].

Collett and Edwards classified [3] CAT into different subtypes based on the anatomical origin of the pulmonary arteries. In type I, the main pulmonary trunk arises from the truncal artery just distal to the truncal valve. In type II and III, the pulmonary trunk is absent and the two pulmonary branches arise posteriorly and are separated from each other, but close to each other, type II or more laterally and widely separated (type III). Type IV now corresponds to PA-VSD.

PA-VSD and CAT, type II–III reveal some common features in the form of a large VSD overriding great artery and a near normal four chamber view. Diagnostic accuracy of PA-VSD and CAT II and III has been reported variably between 64.3 and 95.8 % [2, 4]. The following features have been found useful in differentiating these entities [5].

Abnormal semilunar valve morphology, dysplastic valves, and multiple, thickened, and echogenic leaflets are seen more commonly in CAT -II-III, rather than PA-VSD. Such dysplastic valves more often lead to stenosis or reflux [2, 5] in cases of CAT II–III.

The overriding vessel committed more to the right ventricle has been variably reported in CAT II and III by Vesel et al. [4] 22 %, and Volpe (13 %), and more than 90 % by Traisrisilp et al. [5]. The branch vessels supplying the lungs generally originate a lot more proximally in CAT as compared to PA-VSD. Demonstration of MAPCA from the descending aorta is diagnostic of PA-VSD though the demonstration of the same may not be technically feasible in all cases, especially with less experienced operators. A very small pulmonary artery arising from the right ventricle may be occasionally demonstrated and is diagnostic of PA-VSD. Retrograde filling of the pulmonary vessels may be seen through reverse flow from the ductus arteriosus.

The present case showed normal appearing semilunar valves with no regurgitation or high velocity flow to suggest stenosis. The great artery was committed equally to both the ventricles. The main pulmonary arteries could not be demonstrated arising from the right ventricle or the large proximal overriding artery. Symmetric vessels were seen arising from the descending aorta supplying the lungs suggesting the presence of MAPCA. The above features were clearly favoring the diagnosis of pulmonary atresia with VSD rather than CAT II and III in the present case.

Diagnosis of PA-VSD or CAT II and III requires diligent evaluation of the fetal heart either while screening or during a dedicated fetal echocardiography as the four chamber is usually completely or nearly normal. The evaluation of the outflow tracts leads to the diagnosis of the large VSD and absence of, or nonvisualization of the usual three vessel view or confluent main pulmonary artery. As discussed previously demonstration of a very small aorta with reversed flow through the ductus arteriosus or a very small confluent pulmonary artery from the RA with reversed flow through the ductus arteriosus, with or without the MAPCA will lead to a specific diagnosis of aortic atresia with VSD or PA-VSD, respectively (Table 1). In situations where a single artery is seen overriding the VSD, the evaluation should include defining whether the large artery is equally or preferentially committed more to the right or left ventricle.

The semilunar valve needs to be evaluated for number of leaflets, their thickness, and opening. Any stenosis or regurgitation across the valve should be documented. Origin of a single pulmonary artery branching from the overriding vessel or two separate pulmonary vessels and the level of their origins should be correctly ascertained using power Doppler. Origin of pulmonary vessels from the descending aorta literally confirms the diagnosis of PA-VSD and MAPCA. The size of the thymus gland, and other markers of 22q 11 deletion (DiGeorge) and other common aneuploidies must be positively looked for, in view of their common association with conotruncal abnormalities. Last but not the least, extracardiac abnormalities also need exclusion.

4D ultrasound and STIC volume acquisitions with and without power Doppler are extremely useful tools for diagnosing these abnormalities and demonstrating the spatial relationships of the vessels. Both ventricles draining one large overriding vessel and descending aortic origin of MAPCA were demonstrated in a single image using STIC with power Doppler acquisition, projecting in color power angiographic and glass body modes and then rotating in X, Y, and Z axis to best demonstrate the pathology. Diagnosis of VSD with large overriding vessel may be possible during the early morphology or nuchal translucency scan if the extended anomaly scan protocol including the four chamber view and three vessel view are made a part of the scan protocol using color or power Doppler [6].

Compliance with Ethical Standards

Conflict of interest None.

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