Anatomy of the ventricular septal defect in outflow tract defects: Similarities and differences

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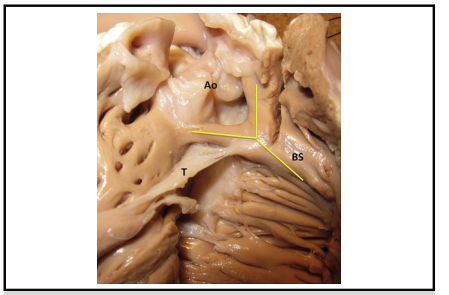
ABSTRACT

Objective: The study objective was to analyze the anatomy of the ventricular septal defect found in various phenotypes of outflow tract defects.

Methods: We reviewed 277 heart specimens with isolated outlet ventricular septal defect without subpulmonary stenosis (isolated outlet ventricular septal defect, 19); tetralogy of Fallot (71); tetralogy of Fallot with pulmonary atresia (51); common arterial trunk (54); double outlet right ventricle (65) with subaortic, doubly committed, or subpulmonary ventricular septal defect; and interrupted aortic arch type B (17). Special attention was paid to the rims of the ventricular septal defect viewed from the right ventricular side and the relationships between the tricuspid and aortic valves.

Results: The ventricular septal defect was always located in the outlet of the right ventricle, between the 2 limbs of the septal band. There was a fibrous continuity between the tricuspid and aortic valves in 74% of specimens with isolated outlet ventricular septal defect, 66% of specimens with tetralogy of Fallot, 39% of specimens with tetralogy of Fallot with pulmonary atresia, 4.6% of specimens with double outlet right ventricle, 1.8% of specimens with common arterial trunk, and zero of specimens with interrupted aortic arch type B ($P < .005$). When present, this continuity always involved the anterior tricuspid leaflet.

Conclusions: The ventricular septal defect in outflow tract defects is always an outlet ventricular septal defect, cradled between the 2 limbs of the septal band. However, there are some differences regarding the posteroinferior and superior rims of the ventricular septal defect. These differences suggest an anatomic continuum from the isolated outlet ventricular septal defect to the interrupted aortic arch type B rather than distinct physiologic phenotypes, related to various degrees of abnormal rotation of the outflow tract during heart development: minimal in isolated outlet ventricular septal defect; incomplete in tetralogy of Fallot, tetralogy of Fallot with pulmonary atresia, and double outlet right ventricle; absent in common arterial trunk; and excessive in interrupted aortic arch type B. (J Thorac Cardiovasc Surg 2015;149:682-8)



An outlet VSD viewed from the right ventricle, located between the 2 limbs of the septal band.

Central Message

The VSD in outflow tract defects is always an outlet VSD, cradled between the 2 limbs of the septal band. Differences between these defects regarding the presence of aortotricuspid continuity suggest an anatomic continuum, related to various degrees of abnormal rotation of the outflow tract during the heart development.

Author Perspective

This anatomic study offers a new perspective on the group of outflow tract defects. The cornerstone of these defects is the outlet VSD, which always opens in the outflow tract of the right ventricle between the 2 limbs of the septal band, irrespective of the presence or absence of a fibrous continuity between the aortic and the tricuspid valves. This anatomic approach underlines the anatomic difference between outlet VSD and isolated central perimembranous VSD. By reinforcing the hypothesis of an embryologic origin common to all outflow tract defects, it might help us to clarify the classification and terminology of VSD.

Supplemental material is available online.

Outflow tract defects including cardiac neural crest defects (so-called conotruncal defects) and transposition of the great arteries (TGA) are due to an abnormal formation of the outflow tract during cardiac development.[1] Cardiac neural crest defects are due to a defect of migration of cardiac neural crest cells, leading to a lack of elongation of the outflow tract and eventually to an abnormal development of the outlet part of the ventricular septum.[2] This common embryologic origin expresses itself within different anatomic phenotypes, including common arterial

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Funded by a grant from the French Society of Cardiology.
Received for publication May 27, 2014; revisions received Nov 17, 2014; accepted for publication Nov 29, 2014; available ahead of print Feb 20, 2015.
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0022-5223/$36.00

http://dx.doi.org/10.1016/j.jtcvs.2014.11.087

Abbreviations and Acronyms
CAT = common arterial trunk
DORV = double outlet right ventricle
IAA = interrupted aortic arch
PA = pulmonary atresia
TGA = transposition of the great arteries
TOF = tetralogy of Fallot
VSD = ventricular septal defect

trunk (CAT); tetralogy of Fallot (TOF) with or without pulmonary atresia (PA) or absent pulmonary valve; double outlet right ventricle (DORV) with subaortic, doubly committed, or subpulmonary ventricular septal defect (VSD); some forms of interruption of the aortic arch; and isolated VSD opening in the outlet of the right ventricle, with outlet septal malalignment.[3] From a purely anatomic standpoint, these congenital heart defects are described as different, but all share the presence of a VSD, as described by several authors.[4-8] Embryologically, this VSD is secondary to a lack of fusion between the outlet septum and the septal band or septomarginal trabeculation because of an incomplete rotation of the outflow tract.[3,9] The aim of this study was to analyze the anatomy of the VSD found in the various phenotypes of outflow tract defects on heart specimens and to determine its localization, its anatomic characteristics, and the rims and relationships to adjacent structures to search for similarities or differences among these various phenotypes.

MATERIAL AND METHODS

We analyzed the anatomy of the VSD in human hearts with outflow tract defects using the heart specimens of the anatomic collection of the French Reference Center for Complex Congenital Heart Defects (M3C) in the Marie-Lannelongue Hospital. This collection includes 1366 heart specimens fixed in 10% formalin. We studied 277 heart specimens with outflow tract defects, including 19 isolated VSDs opening into the outlet of the right ventricle with overriding aorta but no subpulmonary stenosis (isolated outlet VSD, including malalignment and doubly committed phenotypes); 71 with TOF (8 of them with absent pulmonary valve); 51 with TOF-PA; 54 with CAT; 65 with DORV (including 36 with subaortic VSD, 4 with doubly committed VSD, and 25 with subpulmonary VSD); and 17 with interrupted aortic arch (IAA) type B.

We chose to exclude hearts with DORV with noncommitted VSD, because these defects are supposed to be related with distinct embryologic mechanisms.[10] We also excluded IAA type A because this defect is generally thought to be an extreme form of aortic coarctation.[11] There was no IAA type C in the collection. Although TGA is also an outflow tract defect, we chose to study 104 hearts with TGA and associated VSD separately because several epidemiologic, experimental, and genetic studies tend to demonstrate that the morphogenesis of TGA could involve in part the laterality genes, such as *ZIC3*, *CFC1*, and *NODAL*,[12-14] leading to a possible disturbance of the "rotational" aspect of wedging[15] and to the fusion of the outflow tract cushions in a parallel and not spiraling fashion.[16] In addition, we analyzed 15 hearts with dilated or restrictive cardiomyopathy without any associated congenital heart defects as controls.

The intracardiac anatomy of each heart specimen was studied with particular attention paid to the VSD. The exact localization of the VSD and its borders were carefully described viewed from the right ventricular side, as well as the relationship between the tricuspid and aortic valves, looking for a fibrous continuity between these 2 valves, and particularly which tricuspid leaflet was involved in this continuity.

We chose to define the curved surface of the VSD by the surface that will be closed by the surgeon.[9] This surface is different depending on whether each arterial trunk arises from its own ventricle or both arterial trunks arise from the same ventricle as in DORV. In DORV, the VSD, roofed by the outlet septum or its fibrous remnant, is different from the interventricular communication that lies in the plane of the ventricular septum and is roofed by the inner curvature of the heart.

The morphologically normal hearts were used as controls to analyze precisely the anatomy of the membranous septum and its relations with the limbs of the septal band or septomarginal trabeculation. We also looked for the presence or absence of a demonstrable outlet septum and its orientation when the outlet septum was deviated anteriorly (into the right ventricle) or posteriorly (into the left ventricle) relative to the axis of the remainder muscular ventricular septum.

Normal Anatomy of the Right Ventricle

To fully describe the morphologic features of the VSD, we have to clearly define the terminology used to characterize the different parts of the right ventricle (Figure 1).

The ventriculo-infundibular fold (grouping together parietal band and subpulmonary conus) is a muscular band that separates the anterior leaflet of the tricuspid valve from the leaflets of the pulmonary valve in the normal heart.[17-19] The left extremity of the ventriculo-infundibular fold represents the upper part of the ventricular septum: the outlet (or conal) septum, which is fused with the upper extremity of the septal band (or septomarginal trabeculation), between its 2 limbs, anterosuperior and posteroinferior. In the normal heart, the posteroinferior limb of the septal band, which carries the papillary muscle of the conus and its attachments, is not fused with the ventriculo-infundibular fold but is separated from it by the atrioventricular part of the membranous septum, connecting the anterior leaflet of the tricuspid valve with the leaflets of the aortic valve.[20] The interventricular part of the membranous septum connects the septal leaflet of the tricuspid valve with the right coronary and the noncoronary leaflets of the aortic valve (Figure 2). This will help us to analyze the localization of the VSD in the specimens and to describe precisely the rims of the VSD.

Statistical Analysis

StatView software (SAS Institute Inc, Cary, NC) was used for data analysis. The qualitative anatomic variables were presented with percentages. A chi-square test analysis was used to evaluate the possible differences between the parameters.

RESULTS

Localization of the Ventricular Septal Defect

All heart specimens, except 1 with an IAA type B, had a VSD. This VSD was, as expected, always located in the outlet of the right ventricle, between the 2 limbs of the septal band (outlet-type VSD) (Figure 3). However, we could distinguish some anatomic differences within these outlet VSDs, depending on the rims of the VSD and more precisely on the presence or absence of a fibrous continuity between the leaflets of the aortic (or truncal) and tricuspid valves, and on the presence or absence of an outlet septum.