Anatomy of the ventricular septal defect in congenital heart defects: a random association?

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Introduction: A ventricular septal defect (VSD) is part of most congenital heart defects (CHD).

Aim of the study: To determine the distribution of the anatomic types of VSD in CHD.

Material and methods: We analyzed morphologically 1178 heart specimens with CHD from the anatomic collection of the French Reference Center for Complex CHD. Special attention was paid to the localization of the VSD: muscular, membranous, outlet located between the two limbs of the septal band, inlet. The specimens were classified according to the anatomic and clinical classification of CHD (ACC-CHD).

Results: A VSD was present in 67% of all hearts and was:
- Constant, of a single type, in tetralogy of Fallot and variants and common arterial trunk: outlet, in complete atroventricular canal (CAVC): inlet, and in double-inlet left ventricle (DILV): muscular.
- Not constant with a predominant type, in 96% of double discordance (DD,inlet 82%), 62% of heterotaxy syndromes (Hetx, inlet 93%), 93% of interrupted aortic arch (outlet 80%), 87% of double outlet right ventricle (outlet 77%).
- Not constant, of variable type, in 68% of aortic coarctation (CoA: outlet 44%, membranous 35%, muscular 21%), 54% of transposition of the great arteries (TGA: outlet 40%, membranous 25%, muscular 25%, inlet 10%).
- Rare, in anomalies of pulmonary veins (5%), Ebstein anomaly (14%), double-inlet right ventricle (10%), coronary anomalies (25%).
- Isolated in 10% of all VSD: outlet 44%, membranous 36%, muscular 18%, inlet 2%.

Associations according to VSD type:
- outlet : 60% “conotruncal” defects (CTD), 10% TGA
- inlet : 57% CAVC, 13% DD, 10% Hetx
- muscular : 33% DILV, 26% TGA, 13% isolated
- membranous : 30% TGA, 28% isolated, 16% CoA.

Conclusion: The VSD is an integral part of the phenotype in some CHD. In CoA and TGA the VSD is not constant and its anatomic distribution is similar to that in isolated VSD, indicating a likely random association. This reinforces the hypothesis of different genetic mechanisms in TGA and CTD. This original approach, using the anatomic characteristics of one part of the phenotype, could provide new insights in the grouping and aetiology of CHD.