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Echocardiography

The Windsock Syndrome: Subpulmonic Obstruction by Membranous Ventricular Septal Aneurysm in Congenitally Corrected Transposition of Great Arteries

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Anomalies of the membranous portion of the interventricular septum include perimembranous ventricular septal defect and/or membranous septal aneurysm (MSA). In congenitally corrected transposition of the great arteries (L-TGA in sinus solitus), the combination of ventricular inversion and arterial transposition creates a unique anatomic substrate that fosters subpulmonic left ventricular outflow tract obstruction by an MSA. The combination of an L-TGA with subpulmonic obstruction by an MSA is referred to as the windsock syndrome. We report a case of windsock syndrome in a 25-year-old man which is to our knowledge the first three-dimensional echocardiographic description of this congenital entity. (Echocardiography 2013;30:E243-E248)

Key words: congenitally corrected transposition of great arteries, L-TGA, obstruction, aneurysm, windsock

Anomalies of the membranous portion of the interventricular septum include perimembranous ventricular septal defect and/or membranous septal aneurysm (MSA). Anatomically, MSA frequently resembles a windsock, a conical cloth tube used to show wind direction.

In otherwise normal hearts, the protrusion of the MSA from the higher pressure left ventricle (LV) into the lower pressure right ventricle (RV) rarely causes significant right ventricular outflow tract (RVOT) obstruction. This is because the MSA is located infracristal and distant from the pulmonic valve (PV).[1]

In congenitally corrected transposition of the great arteries (TGA) (also referred to as L-TGA in situs solitus) the combination of ventricular

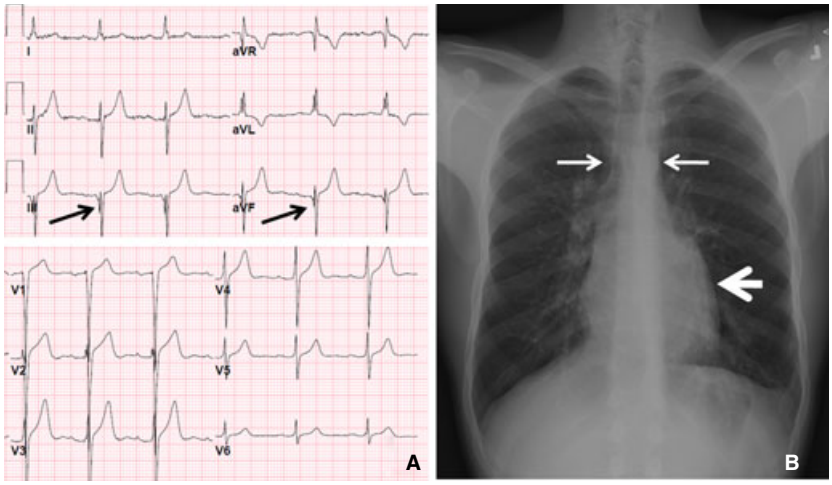

Figure 1. A. Electrocardiogram demonstrates normal sinus rhythm with a pseudo-infarct pattern in the inferior leads (black arrows) and absence of normal Q-waves in left-sided leads (aVL, V5, V6), consistent with the diagnosis of corrected transposition of the great arteries (L-TGA). **B.** Chest radiograph in postero-anterior projection. Vascular pedicle is abnormally straight (thin arrows) because the normal arterial relationship in this patient with L-TGA is lost (thick arrow).

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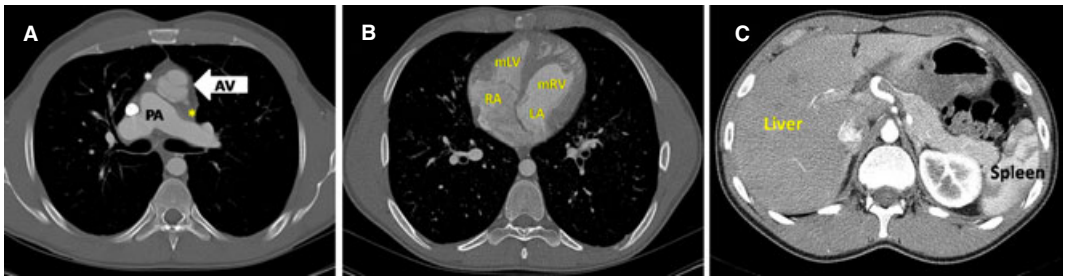

Figure 2. Computed tomography (CT) of the chest demonstrates findings typical of corrected transposition of the great arteries (L-TGA) in situs solitus. **A.** The aortic valve (AV) is positioned anterior and to the left of the pulmonic valve consistent with L-TGA in situs solitus. Visualization of the coronary ostia (asterisks) confirms that the anterior semilunar valve is the AV. The immediate branching of the posterior great vessel confirms its identity as the pulmonary artery (PA). White asterisk, right coronary artery; yellow asterisk, left main coronary artery. **B.** The four-chamber view demonstrates ventricular inversion characteristic of L-TGA. The right atrium (RA) is connected to the rightward positioned morphologic left ventricle (mLV; venous ventricle). The left atrium (LA) is connected to the trabeculated and leftward positioned morphologic right ventricle (mRV; systemic ventricle). **C.** Normally positioned liver on the right and spleen on the left indicative of visceral situs solitus.

inversion, chronic pressure elevation in the morphologic RV and arterial transposition creates a unique anatomic substrate that fosters subpulmonic obstruction of the morphologic left ventricular outflow tract (mLVOT). The lack of infundibulum in the morphologic LV—which places the PV in direct continuity with the mitral valve—leads to protrusion of the MSA directly underneath the PV. This occasionally results in marked subpulmonic mLVOT obstruction.

This windsock syndrome pathophysiology of subpulmonic obstruction by the MSA in congenitally corrected TGA[2] is exemplified by multimodality imaging in this case report. To our knowledge, this is the first description of the windsock syndrome visualized by three-dimensional transesophageal echocardiography (3DTEE).

Case Report:

A 25-year-old man who had been diagnosed with a congenitally corrected transposition of great arteries as a child and bronchiectasis as an adult, presented to our institution with a chief complaint of hemoptysis for the prior 2 days. He was unable to provide any further details of his cardiac history except that he had not undergone any surgical repair. He reported being an active young man who played soccer twice weekly in an amateur league.

On physical examination, patient was afebrile and appeared in no apparent distress. Blood pressure was 105/70 mmHg; pulse was regular at 70 beats/min. He was breathing comfortably at 14 breaths/min. Skin and oral mucosa revealed no signs of cyanosis. There was no evidence of elevated jugular venous pressure or peripheral edema. Lung exam on auscultation revealed diffuse wheezing, decreased breath sounds at the base of left chest. On heart exam, there was a

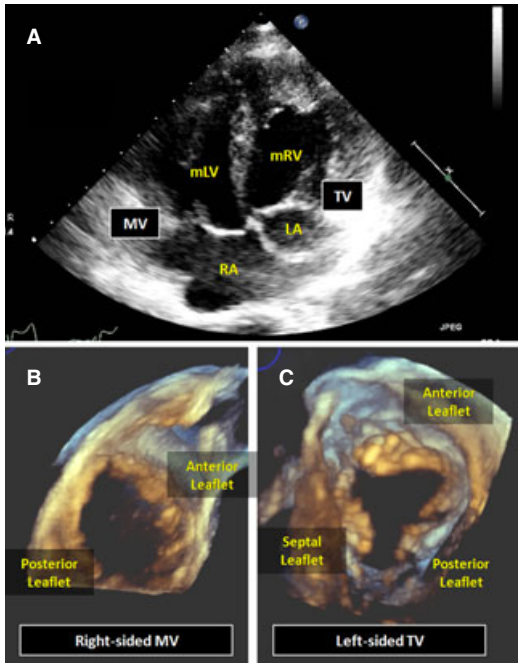

Figure 3. Echocardiograms demonstrate abnormal position of atrioventricular valves. **A.** 2D transthoracic echocardiogram in apical four-chamber view shows ventricular inversion. The tricuspid valve (TV) is identified as the more apically positioned atrioventricular valve and is abnormally positioned on the patient's left side. It separates the left atrium (LA) from the morphologic right ventricle (mRV). The mitral valve (MV) is identified as the more basally located atrioventricular valve and is abnormally positioned on the patient's right side. It separates the right atrium (RA) from the morphologic left ventricle (mLV). Movie clip S1 corresponds to this panel. **B.** 3D transesophageal echocardiograms confirm that the right-sided bileaflet atrioventricular valve is the MV. Movie clip S2 corresponds to this panel and shows the mitral valve from the left ventricular perspective. **C.** 3D transesophageal echocardiograms confirm that the left-sided trileaflet atrioventricular valve is the TV. Movie clip S3 corresponds to this panel and shows the tricuspid valve from the right ventricular perspective.

harsh mid-systolic murmur best heard at the upper sternal borders with wide radiation across the precordium. The remainder of the physical exam was normal.

Electrocardiogram demonstrated normal sinus rhythm with a pseudo-infarct pattern in the inferior leads (black arrows) and absence of normal Q-waves in left-sided leads (Fig. 1A), consistent with the diagnosis of L-TGA. Chest x-ray (CXR) imaging in postero-anterior projection demonstrated an abnormally straight vascular pedicle and the ventricular border on the left appeared more vertical than usual (Fig. 1B). These CXR findings were also consistent with the diagnosis of L-TGA.

On computed tomography (CT) of the chest the aortic valve (AV) was positioned anterior and to the left of the PV consistent with L-TGA in situs solitus (Fig. 2A). The four-chamber view of the heart showed ventricular inversion characteristic of L-TGA. (Fig. 2B). Normally positioned liver on the right and spleen on the left were indicative of visceral situs solitus (Fig. 2C). In addition, there was evidence of diffuse airway disease with bronchiectasis identified especially throughout the right middle lobe and both lower lobes.

Patient then underwent two-dimensional (2D) and 3D transthoracic and TEE for the evaluation of the murmur using a Philips iE33 ultrasound system (Philips Healthcare, Andover, MA, USA). Transthoracic echocardiogram in apical four-chamber view revealed ventricular inversion. (Fig. 3A; and movie clip S1) consistent with L-TGA. 3D transesophageal echocardiograms

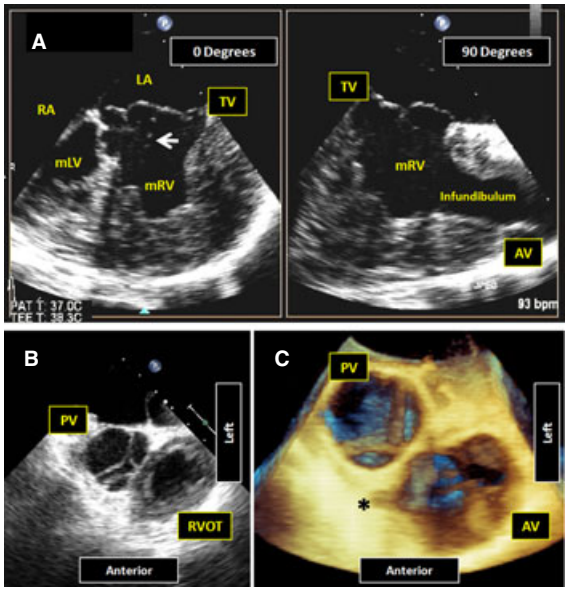

Figure 4. A. Biplane transesophageal echocardiogram at 0 and 90 degrees demonstrates a left-sided hypertrophied morphologic right ventricle (mRV). The following 2 findings help identify this ventricle as the mRV: (1) presence of a septal chorda consistent with the anatomy of the tricuspid valve (TV); and (2) presence of an infundibulum in continuity with a semilunar valve (which in this patient with corrected transposition of the great arteries = L-TGA is the aortic valve = AV). LA = left atrium; mLV = morphologic left ventricle; RA = right atrium. Movie clip S4 corresponds to this panel. **B.** 2D transesophageal echocardiogram demonstrates abnormal relationship of the pulmonic valve (PV) which is side-by-side with and not in continuity with the right ventricular outflow tract (RVOT). The findings are consistent with transposition of great arteries. **C.** A cranial view on 3D transesophageal echocardiogram shows abnormal position of the semilunar valve consistent with L-TGA. The semilunar valve located anterior and to the left is identified as the AV by the presence of the ostium of the right coronary artery (asterisk). The other semilunar valve is the PV.

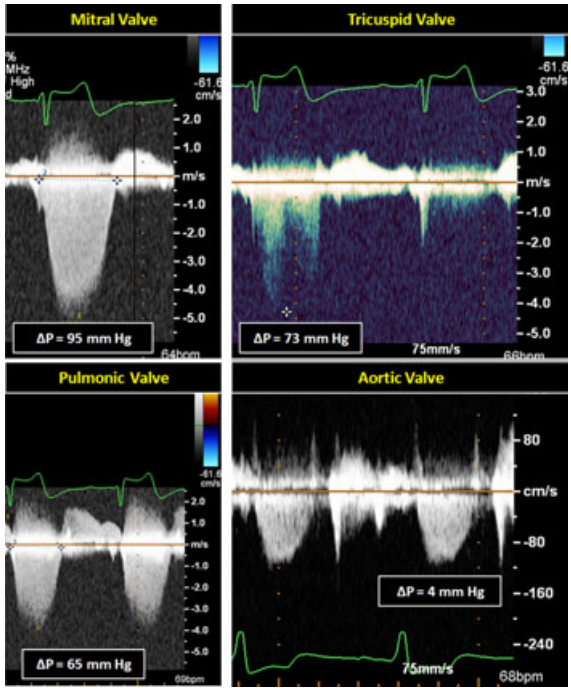

Figure 5. Top Panel: Continuous-wave Doppler tracings across the mitral valve and tricuspid valve. In this patient with L-TGA, the high peak pressure gradient of the tricuspid regurgitant jet (at least 73 mmHg) is expected given that the morphologic right ventricle is the systemic ventricle. However, the high peak pressure gradient of the mitral regurgitant jet (at least 95 mmHg) is abnormal given that the morphologic left ventricle serves as the venous ventricle in this patient. This abnormal mitral gradient is consistent with either pulmonary hypertension or left ventricular outflow tract (LVOT) obstruction. Bottom Panel: Continuous-wave Doppler tracings across the pulmonic valve (PV) and aortic valve (AV). There is no significant systolic gradient across the AV or the right ventricular outflow tract in this patient with L-TGA. In contrast, there is a markedly elevated systolic gradient (at least 65 mmHg) in the lower left panel indicative of either PV or LVOT stenosis. The presence of this outflow gradient in the venous ventricle rules out significant pulmonary hypertension as the etiology of the elevated mitral regurgitant jet gradient (seen in the upper left panel).

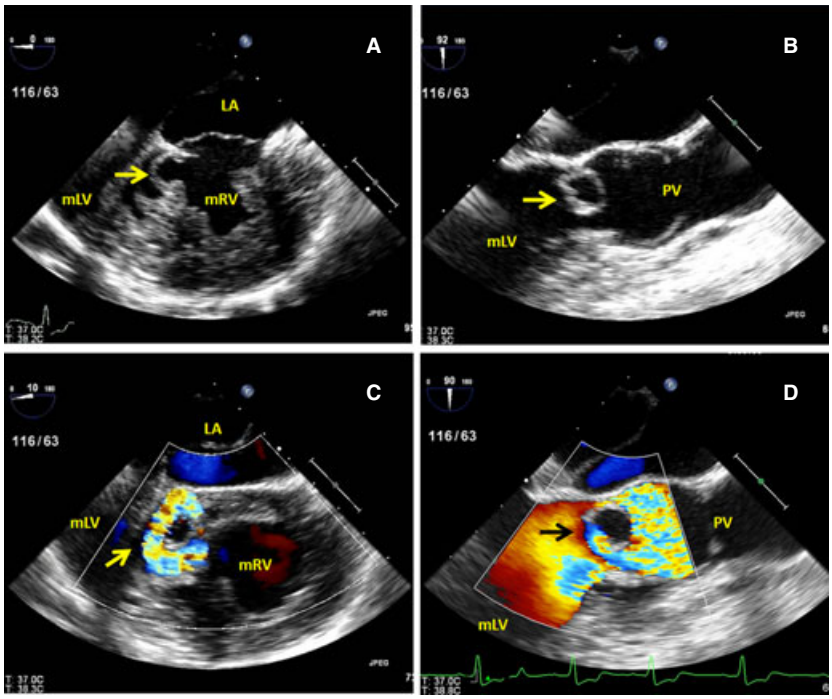

Figure 6. Two-dimensional and color Doppler transesophageal echocardiography demonstrates the left ventricular outflow tract (LVOT) obstruction (subpulmonic stenosis) by the membranous septal aneurysm (MSA) in this patient with L-TGA (windsock syndrome). **A.** Arrow points to MSA protruding into the mLV. Movie clip S5 corresponds to this panel. **B.** Arrow points to membranous septal aneurysm protruding into the LVOT just proximal to the PV. Movie clip S6 corresponds to this panel. **C.** and **D.** Arrows point to flow acceleration around the membranous septal aneurysm in the LVOT in short axis (**C.**) and long axis (**D.**) indicative of subpulmonic stenosis. LA = left atrium; mLV = morphologic left ventricle; mRV = morphologic right ventricle; PV = pulmonic valve. Movie clip S7 corresponds to **D.**

confirmed that the right-sided bileaflet atrioventricular valve is the mitral valve (Fig. 3B; and movie clip S2) and that the left-sided trileaflet atrioventricular valve is the tricuspid valve (Fig. 3B; and movie clip S3).

Biplane transesophageal echocardiogram at 0 and 90 degrees demonstrates a left-sided hypertrophied morphologic right ventricle (mRV) and a right-sided morphologic left ventricle (mLV). Both ventricles had normal systolic function (Fig. 4A; and movie clip S4). There was also an abnormal relationship of the PV which was side-by-side with the AV and not in continuity with the RVOT (Fig. 4B). A cranial view on 3D transesophageal echocardiogram showed an abnormal position of the semilunar valves (Fig. 4C). The findings were all consistent with TGA.

There was trace tricuspid and mild-to-moderate mitral regurgitation. Continuous-wave Doppler tracings demonstrated a high peak pressure gradient of the tricuspid regurgitant jet (at least 73 mmHg); this finding was expected given that the mRV is the systemic ventricle. However, spectral Doppler also revealed a high peak pressure gradient of the mitral regurgitant jet (at least 95 mmHg); this finding was abnormal and consistent with either pulmonary hypertension or left ventricular outflow tract (LVOT) obstruction (Fig. 5, top panels).

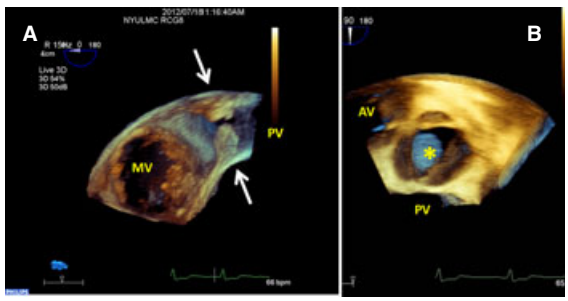

Figure 7. Three-dimensional transesophageal echocardiography demonstrates protrusion of the membranous septal aneurysm (MSA) into the left ventricular outflow tract (LVOT) consistent with windsock syndrome in this patient with L-TGA. **A.** Arrows point to the MSA protruding into the LVOT which is in continuity with both the mitral valve (MV) and the pulmonic valve (PV). The continuity between the mitral and the PVs (i.e. absence of an infundibulum) is characteristic of L-TGA. Movie clip S2 corresponds to this panel. **B.** A cranial view of the partly open PV demonstrates subpulmonic obstruction by the MSA (asterisk). The aortic valve (AV) is located anterior and to the left of the PV consistent with L-TGA. Movie clip S8 corresponds to this panel.

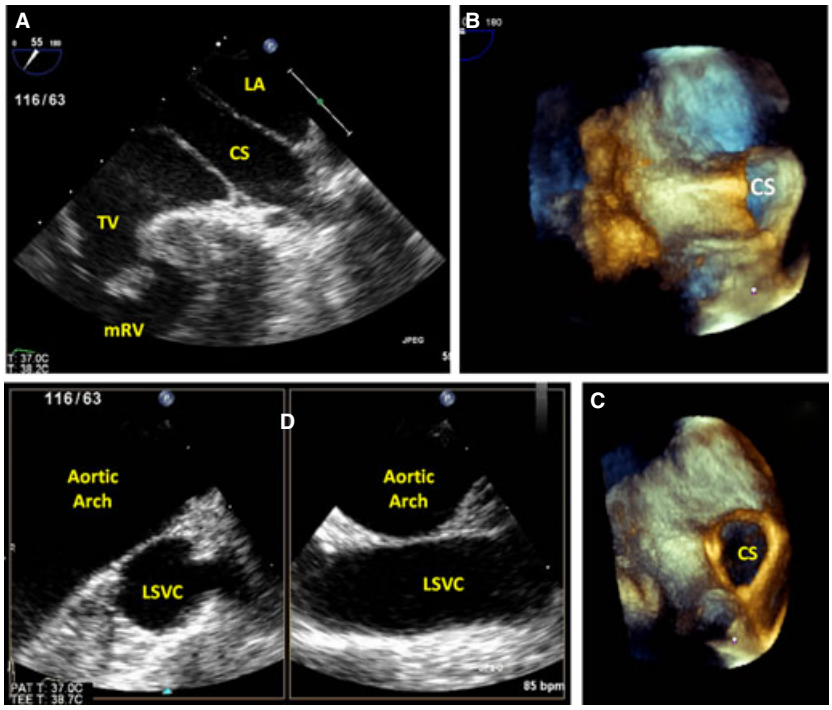

Figure 8. Two-dimensional (**A.** and **D.**) and 3D (**B.** and **C.**) transesophageal echocardiography demonstrates additional findings in this patient with corrected transposition of the great arteries (L-TGA), namely the persistence of the left superior vena cava (LSVC) resulting in a markedly dilated coronary sinus (CS). LA = left atrium; mRV = morphologic right ventricle; TV = tricuspid valve. Movie clip S9 corresponds to **B.** and **C**.

Continuous-wave Doppler tracings revealed no significant systolic gradient across the AV or the RVOT. In contrast, there was a markedly elevated systolic gradient (at least 65 mmHg) across the LVOT with a normally opening PV. This was indicative of either PV or LVOT stenosis (Fig. 5, bottom panels). The presence of this elevated mLVOT gradient ruled out significant pulmonary hypertension as the etiology of the previously noted elevated mitral regurgitant jet gradient.

Further echocardiographic evaluation demonstrated that the LVOT obstruction was due to subpulmonic stenosis by the MSA (Fig. 6; and movie clips S5–S7). This constellation of findings is indicative of the so-called windsock syndrome in this patient with L-TGA (Fig. 7; and movie clip S8).

Incidentally, the patient also had markedly dilated coronary sinus due to persistence of the left superior vena cava (Fig. 8; and movie clip S9).

Discussion:

Congenitally corrected TGA is a rare anomaly that occurs in <1% of all forms of congenital heart disease. Defects of the ventricular septum are described in as many as 70% of patients with congenitally corrected TGA; these include MSA and ventricular septal defects (VSD), typically perimembranous VSD.[3] The first description of an aneurysm of the membranous septum was in 1826 by René Laennec.[4]

Aneurysms of the ventricular septum are often asymptomatic in patients with otherwise normal cardiac anatomy. In normal cardiac anatomy, the infundibulum (also known as the conus arteriosus) separates the atrioventricular valve from the pulmonary artery and its valve. Given the parallel alignment of the great arteries in L-TGA, the membranous septum is in close proximity to the morphologic mLVOT in patients with congenitally corrected TGA. This unique relationship between the mLVOT and the ventricular septum makes it more likely that a MSA will cause subpulmonic obstruction. Bulging of the MSA from the mRV into the mLVOT occurs because the pressure is higher in the mRV than in the mLV.[5] The combination of an L-TGA with subpulmonic obstruction by a MSA is referred to as the windsock syndrome.[2]

The echocardiographic findings including an elevated Doppler velocity of the mitral regurgitation jet and a significant antegrade Doppler velocity along the mLVOT can mimic pulmonary stenosis. Three-dimensional transthoracic echocardiography (3DTTE) and TEE imaging as in this case is very helpful to understand the dynamic nature of this outflow tract obstruction. This differentiation is important as fixed pulmonary stenosis can also occur frequently in patients with L-TGA.

This is to our knowledge the first 3D echocardiographic description of the windsock syndrome, a combination of an L-TGA with subpulmonic obstruction by a MSA.

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Supporting Information

Additional Supporting Information may be found in the online version of this article:

Movie clip S1. Apical four-chamber view on 2DTTE reveals ventricular inversion consistent with L-TGA. This clip corresponds to Figure 3A.

Movie clip S2. 3DTEE en face view of the right-sided mitral valve seen from the left ventricular perspective. This clip corresponds to Figure 3B.

Movie clip S3. 3DTEE en face view of the left-sided tricuspid valve seen from the right ventricular perspective. This clip corresponds to Figure 3B.

Movie clip S4. 3DTEE biplane image demonstrates ventricular inversion and normal biventricular function. This clip corresponds to Figure 4A.

Movie clip S5. 2DTEE image in the mid-esophageal four-chamber view demonstrates the aneurysm of the membranous interventricular septum (arrows). This clip corresponds to Figure 6.

Movie clip S6. 2DTEE in the long-axis view of the morphologic left ventricle demonstrates the aneurysm of the membranous interventricular septum (arrows). This clip corresponds to Figure 6.

Movie clip S7. 2D color Doppler TEE in the long-axis view of the morphologic left ventricle demonstrates subpulmonic obstruction by the aneurysm of the membranous interventricular septum (arrows). This clip corresponds to Figure 6.

Movie clip S8. 3DTEE en face view of the pulmonic valve from the pulmonic artery side demonstrates the globular appearance of the membranous septal aneurysm in the subpulmonic region. This clip corresponds to Figure 7.

Movie clip S9. 3DTEE demonstrates markedly dilated coronary sinus consistent with persistent left superior vena cava. This clip corresponds to Figure 8.