**Unrepaired partial atrioventricular septal defect in a 64-year-old woman with Down syndrome**

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Background: Congenital heart defects (CHD) are often repaired in the newborn period as early surgical repair results in optimal long-term outcomes. CHD is frequently described in patients with Down syndrome (DS) including ventricular septal defect (VSD), atrioventricular septal defect (AVSD), and other complex CHD. As a result of advances in surgical and medical therapies, patients with DS and CHD that have undergone early surgical repair are now living well into the 6th decade of life.

Methods: We present a 64-year-old woman with past medical history significant for DS and unrepaired CHD.

Results: Diagnostic evaluation included a 12-lead electrocardiogram that demonstrated a low-right atrial rhythm and non-specific intraventricular conduction delay. Transthoracic echocardiogram (Figure 1) demonstrated a large ostium primum atrial septal defect with a bidirectional shunt (primarily left-to-right), mild-moderate tricuspid valve regurgitation, moderate right ventricular (RV) enlargement, normal RV systolic function, and mildly elevated RV systolic pressure. There was aneurysmal dilation of the main and branch pulmonary arteries. The left atrioventricular (AV) valve was trileaflet with multiple clefts and moderate regurgitation. There was normal left ventricular size and systolic function. Lastly, an inlet VSD was identified, but occluded by tricuspid aneurysmal tissue. This constellation of anatomical findings was consistent with partial AVSD.

Conclusions: Given the continued increase in life expectancy, patients with DS and CHD should receive appropriate counseling throughout their lifetime including all available medical and surgical therapies. In clinical practice, multiple medical co-morbidities is the rule rather than the exception in the adult DS patient of advanced age (>45 years). A thorough investigation of all comorbidities accompanied by appropriate levels of care, risk assessment, and benefits of diagnostic or therapeutic interventions must be discussed openly with the patient and/or decision-makers. This will ultimately lead to the highest quality of care for patients with DS from birth to adulthood.

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| **Figure 1.** Echocardiography findings demonstrate anatomy consistent with unrepaired partial atrioventricular septal defect. Subcostal coronal image (A) demonstrating primum atrial septal defect (yellow star) with left-to-right shunt (B) seen on color Doppler. Apical 4-chamber view (C) demonstrating mild right atrial and ventricular enlargement. From this view, the inlet ventricular septal defect (red arrow) is occluded by tricuspid aneurysmal tissue. The basilar short-axis view (D) demonstrates a large cleft (red arrow) in the left atrioventricular (AV) valve with trileaflet appearance (E) and multiple jets (orange arrows) of AV valve insufficiency (F). Lastly, the main and branch pulmonary arteries are severely dilated (F). AV = atrioventricular; LA = left atrium; LPA = left pulmonary artery; LV = left ventricle; MPA – main pulmonary artery; RA = right atrium; RPA – right pulmonary artery; RV = right ventricle. |