Marvin Louis Roy Lu et al., J Clin Exp Cardiolog 2015, 6:4
http://dx.doi.org/10.4172/2155-9880.S1.024

OMICS International
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5th International Conference on
Clinical & Experimental Cardiology
April 27-29, 2015 Philadelphia, USA

Gerbode defect: A rare form of atrioventricular septal defect in a 23 year old woman presenting with progressive dyspnea

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Introduction: Gerbode defect is a rare type of ventricular septal defect (VSD) defined as the presence of an anatomic shunt between the left ventricle (LV) and the right atrium (RA). Its incidence reportedly ranges from 0.05-0.08% of all catheterized congenital heart defects.

Case: A 23-year-old woman with systemic lupus erythematosus presented to Einstein Cardiology clinic with progressive dyspnea on exertion and fatigue over the last one month accompanied by intermittent pressure-like left-sided chest discomfort. She denies any other symptoms. Vital signs were within normal limits. Physical exam revealed a high-pitched systolic ejection murmur at the left sternal border radiating to the back and bilateral carotids. Laboratory exam was normal. Electrocardiogram showed normal sinus rhythm. Echocardiogram showed normal valves and biventricular function with ejection fraction of >55%, and a small perimembranous LV to RA communication. The jet is eccentric and cursing along the septal leaflet of the tricuspid valve. The vena contracta of the jet is 0.38 cm with a rough estimate of 0.34cm2 by 3D. She underwent right heart catheterization with saturation run which revealed a significant step-up in oxygen saturation from the inferior vena cava (68.6%) to the RA (78%). A diagnosis of Gerbode defect is made and she is currently undergoing evaluation for surgical versus percutaneous closure of the shunt.

Discussion: There are 3 types of Gerbode defect depending on the location of the shunt. In type I, the defect is in the atrioventricular membranous septum located above the tricuspid valve. Type II describes a defect below the tricuspid valve where shunting initially goes through a VSD with atrial shunting occurring through a deficiency in the septal leaflet of the tricuspid valve. Type III is a combination of both types. Patients with this defect usually present with progressive dyspnea and exercise intolerance similar to decompensated heart failure. Diagnosis can be made through echocardiogram with the presence of a dilated RA in the setting of a high Doppler gradient between the LV and RA. Spontaneous closure is very rare. Hence, surgical closure is recommended in all patients who are symptomatic with no other obvious cause, largely with excellent results.

Biography

Marvin Louis Roy Lu has completed his MD at the age of 25 at the University of Santo Tomas in the Philippines and is currently a 2nd year resident in Internal Medicine at Einstein Medical Center, Philadelphia.

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