Biventricular Non-Compaction with Right Ventricular Predominance in a Hispanic Male

Jose D. Burgos, M.D., FACP
Arianna Nieto-Meraz, MSIV
Juan Arenas, M.D., PGY1
Sarmad Said, M.D., PGY1
Omar Sosa, M.D., PGY1

INTRODUCTION
Non-compaction of the ventricular myocardium is a recently recognized genetic cardiomyopathy characterized by distinctive features including prominent trabeculations of the myocardium and deep endocardial recesses.1 It is assumed to occur as an arrest in normal endomyocardial morphogenesis and is mostly associated with other congenital malformations. The left ventricle is usually involved but both ventricles can be affected. A predominately right ventricular involvement is present in fewer than half of the cases of biventricular non-compaction and a predominant right ventricular non-compaction is extremely uncommon and has been reported in only once.2 We report a case of biventricular non-compaction in a 52 year old male that predominantly involved the right ventricle.

CASE REPORT
A 52 year-old Hispanic male with history of 20 pack-year smoking, presented with chest pressure like sensation for 2 months prior to the admission. Initially he went to his primary care physician who presumptively treated him as gastritis and also found him to be hypertensive. The month before hospital admission, he had noticed progressive decrease in his exercise tolerance. There was no significant cardiac history in his family. Physical examination revealed normal vital signs. Cardiovascular examination revealed an aortic early diastolic murmur of grade 3 in the Erbs area and a pansystolic murmur of grade 4/6 at the apex with radiation to the mid-axillary line, neck examination revealed a jugular venous distention of about 8 cm and the chest examination demonstrated reduced air entry over both lung bases with mild basal crackles. His extremities had grade 1 edema over the ankles. Laboratory investigations showed an elevated BNP of 1768 pg/ml.

His EKG revealed sinus tachycardia with occasional premature ventricular complexes, possible left atrial enlargement and non specific ST-T wave changes. Chest X-ray had features of cardiomegaly and pulmonary edema.

Transthoracic echocardiogram demonstrated moderately dilated left ventricle, prominent trabeculae of the left ventricle suggestive of left ventricular (LV) non-compaction, with severely reduced systolic function estimated ejection fraction of 20-25%, with global hypokinesia of the LV. Right ventricle also had prominent trabeculation suggestive of RV non-compaction. In addition there was right atrial enlargement, moderate to severe mitral regurgitation, moderate aortic regurgitation and severe pulmonary hypertension.

He underwent left heart catheterization which demonstrated normal coronaries and elevated left ventricular end diastolic pressure. Cardiac MRI was performed to confirm the echocardiography findings and it demonstrated left ventricular dilatation with global hypokinesia, excessive trabeculation of the left ventricular apex and mid ventricular segments, the thickness of the trabecular portion of the myocardium was twice that of the adjacent compacted myocardium. The right ventricle was also dilated with mildly depressed function, exhibited hypertrabeculation of the apex and the lateral wall. The left ventricular ejection fraction was estimated to be 22%. The left atrium was dilated with mild mitral regurgitation (Figure 1-2).

DISCUSSION
Myocardial non-compaction is a rare congenital cardiomyopathy secondary to interruption of the normal development of the myocardium during embryogenesis.1 It occurs more frequently in males, and its prevalence varies from 0.06 to 0.24%.5-7 It usually involves the left ventricle alone; however, a biventricular involvement can occur.2,4 According to Chin and coworkers, approximately one half of the cases are familial.1 A genetic linkage to a mutation in the G4.5 gene and maps to the chromosome

Figure 1 and 2. Cardiac MRI shows trabeculation of the left ventricular apex and mid ventricular segments. The right ventricle also appears dilated with mildly depressed function, exhibiting hyper trabeculation of the apex and lateral wall.

Continued on page 9
Xq28 was described in familial occurrence.\(^8\)

Isolated non-compaction of the ventricular myocardium is an uncommon disease that has been reported in only a few clinical studies and case reports. It was first described in a pediatric population in 1990,\(^4\) and later characterized in adults. In 1995, it was categorized as unclassified cardiomyopathy in a report by the World Health Organization.\(^9\)

Clinical features on ventricular non-compaction are non-specific; however these include heart failure, arrhythmias, and thromboembolic events.\(^1,4,5,11\) Echocardiogram is the key method of diagnosis.

The diagnostic criteria are summarized as:\(^12,13\)
- Absence of coexisting cardiac anomalies
- Existence of at least four prominent trabeculations and deep intertrabecular recesses.
- Presence of blood flow in the deep intertrabecular recesses, perfused from the ventricular cavity as detected by color Doppler imaging.
- Having a ratio greater than two between the noncompacted subendocardial layer and compacted subepicardial layer at the end of systole.

A left ventricular apical and inferior wall involvement is present in all adult cases, and a right ventricular apical involvement is present in 41% of patients.\(^4\)

Although the echocardiography has an important role in the diagnosis, contrast ventriculography, computed tomography or MRI can also be used for assessment, especially in patients with poor image quality on echocardiography.\(^13,14\) Cardiac MRI demonstrates the extent of involvement and localization.\(^15\) The two different myocardial layers can be seen on T2 images after an intravenous injection of gadolinium-diethylenetriaminepentaacetic acid (DTPA) injection.\(^16\)

The management comprises treatment for heart failure, arrhythmias and thromboembolic events. However there is no distinct therapy based on large-scale studies. Long-term anticoagulation is useful, especially in patients with left ventricular dysfunction, atrial fibrillation or thrombosis.\(^17\)

**CONCLUSION**

Biventricular non-compaction is a unique disorder accompanied by life threatening complications. In left ventricular non-compaction cases, right ventricle must also be examined carefully for right ventricular involvement. Early diagnosis with improvements in echocardiographic imaging and appropriate medical therapy may increase the probability of event free survival.

**REFERENCES**


Jose D. Burgos, M.D., FACP, Assistant Professor of Medicine, Department of Internal Medicine, Texas Tech University Health Sciences Center - Paul L. Foster School of Medicine.

Arianna Nieto-Meraz, MSIV, Department of Internal Medicine, Texas Tech University Health Sciences Center - Paul L. Foster School of Medicine.

Juan Arenas, M.D., PGY2, Department of Internal Medicine, Texas Tech University Health Sciences Center - Paul L. Foster School of Medicine.

Sarmad Said, M.D., PGY1, Department of Internal Medicine, Texas Tech University Health Sciences Center - Paul L. Foster School of Medicine.

Omar Sosa, M.D., PGY1, Department of Internal Medicine, Texas Tech University Health Sciences Center - Paul L. Foster School of Medicine.