Spongy Heart, A Rare Congenital Heart Disorder.

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Background

Left ventricular non-compaction (LVNC), previously called spongy myocardium, is a rare congenital myocardial disorder characterized by prominent LV trabeculae, a thin compacted layer and deep inter-trabecular recesses. The prevalence among patients undergoing echocardiography (ECHO) is approximately 0.014 to 1.3 % [2]. LVNC can be either sporadic or familial. Various studies have linked LVNC to autosomal dominant inherited disorders.

The clinical manifestations of LVNC are variable. Some patients may not experience cardiac symptoms. Clinical features may include dyspnea, chest pain, palpitations or syncope. The major complications of LVNC include heart failure, atrial or ventricular arrhythmias, sudden cardiac arrest, and thromboembolic events [5].

We are presenting a case of an African American male patient who presented to the emergency department (ED) complaining of the sudden onset of slurred speech and blurry vision. Further evaluation revealed LVNC. We are discussing this rare cause of congenital heart disease and the importance of understanding the different presentations and complications associated with it.

Case Presentation

The patient was an African American male in his late 40s who arrived to ED complaining of the sudden onset of slurred speech and blurry vision. At the time of evaluation, his slurred speech had resolved, but he complained of persistent bilateral blurry vision. There were no other associated symptoms.

His physical examination was unremarkable, except for an elevated blood pressure of 160/100 mmHg. He did have a family history significant for a twin brother who died at age 46 due to an unknown cardiac reason. He had a past medical history of a transient ischemic attack (TIA) 7 years prior.

The patient was placed on the stroke protocol, an EKG was performed, blood work was obtained and imaging studies were ordered.

RESULTS:

- EKG: Normal sinus rhythm with no evidence of ischemia.
- CT of the head: Negative for acute intracranial findings.
- CTA of Head/Neck: No evidence of stenosis or occlusion.
- Chest X-ray: Moderate cardiomegaly.

LABORATORY VALUES	
Pro-BNP	389
Total cholesterol	228
LDL	155
HDL	46
Triglycerides	73

VITAL SIGNS

BP: 160/100 mmHg
HR: 73 bpm
Temp: 98.0 degrees
O2: 98% on room air

Hospital Course

Transesophageal Echocardiography revealed an ejection fraction of 30-35%, a left to right shunt in the patent foramen ovale, left ventricular non-compaction, left ventricle with abnormal septal wall motion, global hypo-kinesis and significant trabeculation of all the walls, except the septum. An MRI showed acute infarction of the right occipital and right parietal lobe. Repeat Chest X-ray showed stable cardiomegaly.

For his newly diagnosed LVNC, the patient was started on apixaban, low dose carvedilol, a high intensity statin, an ACE-inhibitor and Aspirin. Cardiology, Neurology, physical therapy and the primary care team monitored the patient during his hospitalization. Once his medications where optimized he was discharged and advised to follow up with his primary care physician and cardiology as an out patient.

Imaging:

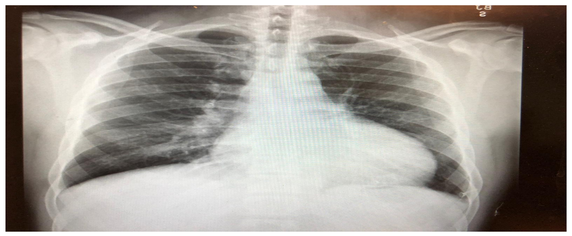

Figure 1. Chest X-Ray showing enlarged cardiac silhouette.

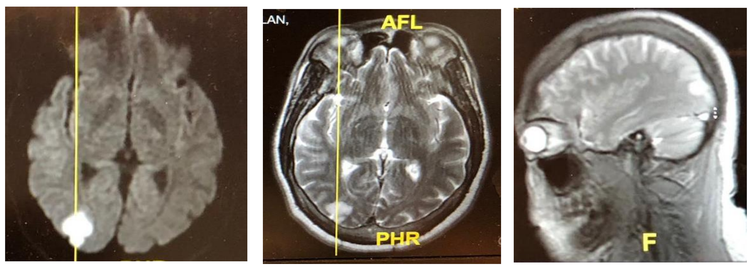

Figure 2. MRI showing an area of acute ischemia in the cortex of the right occipital lobe and white matter of the right parietal lobe.

Literature Review

The ventricular wall of a normal heart consists of a compacted layer of myocardial fibers set in the matrix of connective tissue. Pronounced hyper-trabeculation may be the result of altered regulation in cell proliferation, differentiation, and maturation during ventricular wall formation [8].

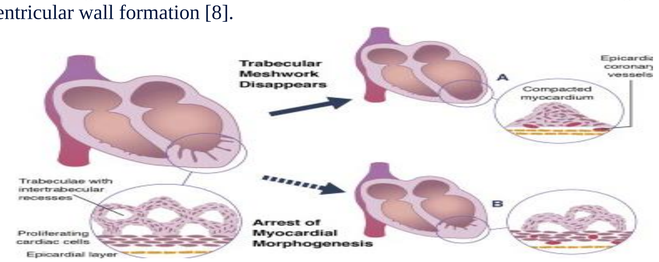

Figure 1.Embryonic Development of LVNC. The myocardium starts off as a meshwork of fibers that regress at weeks 5 to 8 to form compacted outer and inner smooth muscle layers (A). In LVNC, there is retarded myocardial morphogenesis and persistence of the trabecular meshwork (B). [9]

Discussion

- Echocardiography is the preferred initial test. The diagnosis is established by identifying specific morphologic criteria. The most widely accepted criteria is the Jenni Criteria [4].
- This patient presented with classical TIA symptoms. He had a positive family history for sudden cardiac death. Congenital heart diseases should be suspected when there is an unexplained family history of death at a young age, heart failure or stroke. In this setting, it is wise to pursue further evaluation of possible cardiac and neurologic etiologies. In our case, workup for a source of stroke yielded the diagnosis of LVNC. The hyper-trabeculation predisposed our patient to clot formations.
- Medical optimization of different comorbidities is key to enhancing patient outcomes, as trabeculations may develop during adult life in response to remodeling. Understanding the pathophysiology and the initial work up is key for a proper diagnosis. The timely detection of LVNC and its severity, would allow for interventions such as lifestyle modification, medication optimization, oral anticoagulation, defibrillator implantation and/or even cardiac transplantation.

Conclusion

In summary, ventricular non-compaction varies in presentation. Thromboembolic events, acute decompensated heart failure, arrhythmias or even sudden cardiac death might be the initial presentation. Given that prognosis for these patients is generally poor, we recommend management with a multidisciplinary team. Our goal is to provide evidence based data regarding the evaluation and proper management of these patients to improve their outcomes

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