Case Report

Noncompaction Cardiomyopathy and Stroke: Case Report and Literature Review

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Background: To describe a rare case of isolated noncompaction cardiomyopathy and stroke and to review the medical literature on noncompaction cardiomyopathy. *Methods:* Retrospective chart review of the case was performed. Extensive literature review on etiology, clinical presentation, diagnosis, and management of noncompaction cardiomyopathy was also performed. *Results:* Our patient is a healthy 20-year-old woman who presented with sudden onset left face and arm weakness and hypesthesia. Magnetic resonance imaging (MRI) brain showed right middle cerebral artery (MCA) infarct. Magnetic resonance angiography head showed right MCA artery (M2) cutoff. MRI neck was nonsignificant. Echocardiogram was suggestive of noncompaction of left ventricle. Cardiac MRI confirmed the noncompaction of the left ventricle myocardium, which was thought to be the etiology of stroke. Patient was started on anticoagulation for secondary stroke prevention. *Conclusions:* Isolated left ventricular noncompaction cardiomyopathy (LVNC) is a rare form of primary genetic cardiomyopathy, which occurs because of the arrest of the process of compaction of ventricular myocardium during embryogenesis. Noncompaction cardiomyopathy is usually associated with other primary cardiac structural abnormalities like dysfunctional cardiac valves. In isolated noncompaction cardiomyopathy, there are no other primary cardiac structural abnormalities. The most common clinical features seen in LVNC include left ventricular systolic dysfunction, congestive heart failure, arrhythmias, and cardiac embolic events theorized to result from thrombus formation within the intertrabecular recesses. As it is a rare disease, evidence-based recommendations for preventing thromboembolic events in isolated left ventricular noncompaction have not been established. **Key Words:** Noncompaction—cardiomyopathy—stroke—atrial fibrillation—anticoagulation.

Case Summary

A 20-year-old right-handed Caucasian female with history of migraines with aura, taking oral contraceptive pills, presented to emergency room with sudden onset of weakness and sensory loss on left face and arm accompanied with right frontal headache. Headache was different from her typical migraine attacks and was not associated with aura. Noncontrast computed tomography (CT) head was normal. Because of minor neurologic symptoms and possibility of complex migraine, intravenous tissue plasminogen activator was not given. Magnetic resonance imaging (MRI) of brain showed moderate-sized acute

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infarct in right middle cerebral artery territory (Fig 1, A). Magnetic resonance angiography head showed cutoff of the superior division of right middle cerebral artery (MCA) artery (Fig 1, B). Magnetic resonance angiography neck was nonsignificant. Fat sat MRI brain did not show any dissection. A transthoracic echocardiogram showed hypertrabeculated left ventricular apex with deep crypts suggestive of left ventricular noncompaction cardiomyopathy, ejection fraction of 55%, normal cardiac valves, normal-sized left atrium, no thrombus, and no patent foramen ovale (Fig 2, A). Patient had no signs or symptoms of heart failure. Given ejection fraction of 55% and lack of heart failure clinical features, she was classified as stage B heart failure. Cardiac MRI was done to confirm findings of echocardiogram. Cardiac MRI showed prominent trabeculations in the left ventricular mid and apical regions, which confirmed the diagnosis of isolated left ventricular compaction cardiomyopathy (Fig 2, B-D). Telemetry did not reveal any arrhythmia. Lipid panel, Hemoglobin A1c, Anti-nuclear antibody, Erythrocyte sedimentation rate, C-reactive protein were all within normal limits. Hypercoagulable work-up was negative. Given all the above findings and cortical infarct, final diagnosis was cardioembolic right MCA infarct due to left ventricular noncompaction cardiomyopathy. After discussion with cardiology, patient was started on lifelong anticoagulation with coumadin with goal international normalized ratio of 2-3. Genetic testing revealed a heterozygous novel variant in the *lamin A/C* gene. She followed up in cardiology and stroke clinic and was continued on anticoagulation for indefinite period.

Discussion

Introduction

Isolated left ventricle noncompaction (LVNC) is a rare primary genetic cardiomyopathy. During embryogenesis, early myocardium consists of the interconnecting fibers separated by sinusoids that connect to the left ventricular cavity. This spongy myocardium has a trabecular appearance. The "spongy" myocardium begins to "compact" and develop into mature musculature during weeks 5-8 of embryogenesis. The sinusoids shrink and eventually become capillaries in the coronary circulation. In LVNC, this process of "compaction" is arrested and there is persistence of these large trabeculations that are continuous with left ventricular cavity without any communication with epicardial circulation.[1,2] The exact cause of the arrest of compaction is not known. Rare in any case, noncompaction is almost invariably associated with other congenial cardiac malformations but even rarer it can present as isolated LVNC.[3] Although LVNC is primarily the disease of left ventricle, associated involvement of right ventricle has been described in past.[2]

Epidemiology

Noncompaction cardiomyopathy, which was first found as an interesting autopsy finding in 1932, then became an echocardiographic curiosity 3 decades ago, has now become a rather common finding in living subjects with the introduction of various newer cardiovascular imaging modalities in addition to echocardiography, including cardiac MRI, cardiac CT, and contrast ventriculography.[2] Prevalence of noncompaction cardiomyopathy in those adults who are referred for echocardiography for some clinical reason has been estimated between .01% and .27%.[4-7] Prevalence in the entire population is estimated to be between .014% and 1.3%.[8-10] The incidence is estimated between .05% and .25% per year.[4] Males are more commonly affected than females. Prevalence in males has been reported around 56%-82%.[3,4,11,12]

Genetics

Noncompaction cardiomyopathy can be inherited or develop sporadically as well. The most common pattern

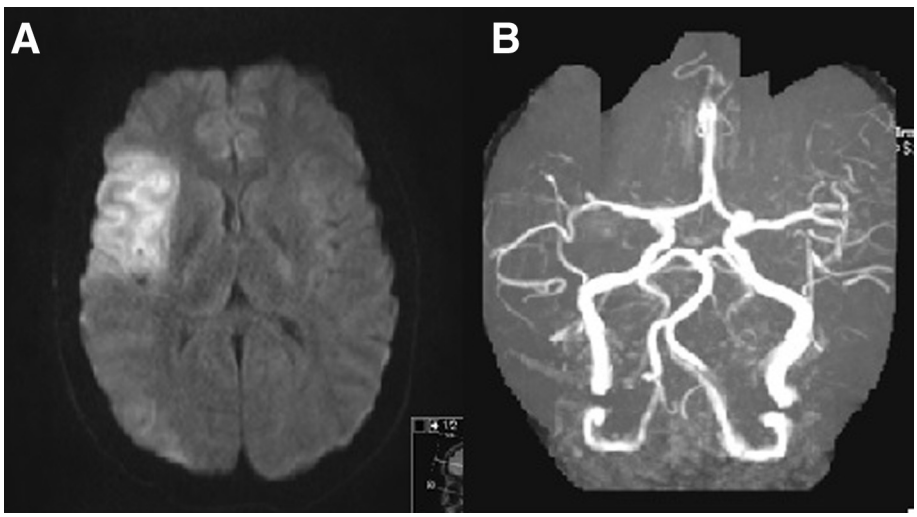

Figure 1. *(A) DWI MRI brain showing acute right MCA infarct. (B) Intracranial MRA showing cutoff at the superior division of right MCA artery. Abbreviations: DWI, diffusion-weighted imaging; MRA, magnetic resonance angiography; MRI, magnetic resonance imaging.*