A review of Ventricular Non-Compaction

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Abstract

Non-compacted myocardium has been categorized as unclassified congenital cardiomyopathy and is characterized by an altered structure of the left ventricular myocardium with extremely thickened, hypo kinetic segments consisting of two layers: thin, compacted myocardium on the epicardial side, and a thicker non-compacted endocardial layer. Non-compaction of the ventricular myocardium results from an arrested normal process of myocardial compaction (normally during the first month of fetal life). Right ventricular non-compaction may accompany left ventricular non-compaction in less than 50% of patients.

INTRODUCTION

Non compaction of the ventricles is a rare disorder of endomyocardial morphogenesis characterized by echocardiographic findings consisting of multiple, prominent myocardial trabeculations and deep intertrabecular recesses communicating with the ventricular cavity, in the absence of other structural heart disease.

COURSE

It affects the LV, and/or RV, and may result in usually systolic ventricular dysfunction with frequent progression to clinical heart failure, and potential complications including arrhythmias and thromboembolic events.

Non-compaction was initially described in children; however, recent studies have characterized this disease in adult populations, in whom this process may be underappreciated in prevalence. It can be familial. It is rarely associated with other congenital cardiac disorders (LCA from pulmonary trunk, LVOT/RVOT obstruction, Ebstein's anomaly Bicuspid AV, and isomerism of LA appendage).

EMBRYOLOGY

In the early embryo, the heart is a loose interwoven mesh of muscle fibers that forms trabeculae with deep intertrabecular recesses. During fetal ontogenesis->myocardium gradually condenses->large spaces within the trabecular meshwork flatten. These large intertrabecular spaces transform into capillaries as the process of compaction occurs from epicardium to endocardium and from base towards the apex, with trabecular compaction being more complete in the left ventricular than in the right ventricular myocardium. In the abnormal condition, the process of compaction arrests at an early stage, leaving numerous excessively prominent trabeculations and deep intertrabecular recesses in a segmental distribution usually from the apex.

DIAGNOSIS

The major echocardiographic criterion is a ratio of noncompacted to compacted (N/C) myocardium measured at a site of maximal thickness that is $>_2$ (at the end of systole).Color Doppler-> differentiates deep ventricular recesses that invaginate the non-compacted myocardium. It typically involves apical, lateral and inferior.

(Diffuse hypertrophy by HTN and VHDs where trabeculations are seen, coursing from the free wall to the septum). Other diagnostic studies can sometimes visualize trabeculations (CMR, CT, LVgraphy). Screening of family members is suggested.

GENETIC BASIS

Families with multiple individuals who have LVNC are likely to have a genetically inherited form. Not all affected individuals, however, must have an affected parent because, in all autosomal dominant diseases, a certain proportion of cases occur due to a new mutation (i.e., they are sporadic). The parent whose germ cells contain the new mutation will be clinically normal, since the mutation affects only a single germ cell, but can transmit the disease-causing allele to their offspring.

In isolated LVNC, familial recurrence is high and found in approximately 40 percent of patients. The inheritance

patterns of LVNC include X-linked inheritance in males with isolated LVNC, and an autosomal dominant pattern in some isolated and non-isolated cases. In the X-linked form, mutations in the gene G4.5 (TAZ), which encodes tafazzin, were initially identified by Bleyl and colleagues. This is the same gene previously identified by Bione and colleagues as causative of Barth syndrome; some patients with LVNC also have clinical features of Barth syndrome.

Figure 1



Echocardiogram of a LVNC Heart

PROGNOSIS

No major studies have been done. LVNC is associated with higher morbidity and mortality. It could be due to CHF, ventricular arrhythmias, or thromboembolic events. Prognosis is poor in symptomatic patients.

MANAGEMENT

No specific therapy for LVNC. Patients with reduced LVEF->standard medical therapy. Chronic warfarin-> a.fib and/ or an LVEF _<30-40%. Holter->? Detect asymptomatic arrhythmias. Heart transplantation-> only in pts with LVNC and end stage heart failure.

References

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