Noncompaction Cardiomyopathy: Uncommon and Often Missed Cause of Congestive Heart Failure with Significant Clinical Implications

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Background

- Noncompaction cardiomyopathy, also known as noncompaction of the ventricular myocardium (NCVM), is an uncommon congenital cardiomyopathy
- Characterized by multiple prominent trabeculations in the ventricular wall and deep intra-trabecular recesses within the ventricular cavity
- Several clinical features that require special consideration in medical management
- Diagnosis and proper medical management is thought to be missed or delayed – likely due to lack of awareness and difficulty of diagnosis

Case Description

History of Present Illness

- 56 year old African American woman with a significant history of congestive heart failure (CHF) and hypertension
- Presented to the emergency department with angina-like chest pain starting 4 hours prior to admission
- Chest pain described as pressure-like that was worse with exertion and better but not relieved with rest

Significant Patient History

- Hypertension and CHF only known medical problems
- Past surgeries include hysterectomy and appendectomy
- Family history remarkable for diabetes, cancer, and heart attack
- Denies regular alcohol, tobacco, or drug use

Case Description

Initial Assessment

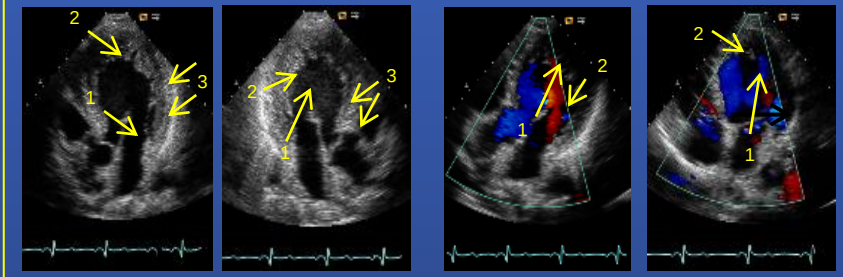
- Physical exam revealed S4 gallop and bilateral lower lobe crackles; otherwise unremarkable
- Troponin mildly elevated at 0.5 ng/mL
- Lab otherwise unremarkable
- EKG showed T-wave inversion in leads V3-V6 and mild anterior lead ST-elevation – unclear if acute myocardial infarction versus left ventricular strain

Hospital Course

- Urgent left heart catheterization performed: no underlying coronary artery disease, but severe global left ventricular dysfunction noted
- Follow-up 2D Doppler findings suggestive of congestive heart failure due to NCVM
  - Isolated left ventricular systolic dysfunction with an ejection fraction of 40%
  - Multiple trabeculations in left ventricle with blood flowing through them
  - Two distinct layers of myocardium – noncompacted and compacted layers
- Patient treated with conservative CHF management including optimization of cardiac medications and diuresis with oral furosemide
- Discharged with outpatient follow-up for further evaluation & management of NCVM including Holter monitoring and possible anticoagulation therapy

Echocardiographic Images

Figure 1 Figure 2 Figure 3 Figure 4



Figures 1 & 2: standard 2D echo demonstrating 1) trabeculations 2) intra-trabecular recesses 3) two distinct lavers of myocardium

Figures 3 & 4: 2D Doppler echo illustrating 1) blood flow through the intra-trabecular recesses 2) trabeculations

Echocardiographic Images

Etiology

- Rare congenital cardiomyopathy found in ~0.014% of echocardiograms
- Typically diagnosed in adulthood
- Cause thought to be due to arrest of embryonic myocardial development
- Both familial and sporadic forms identified

Diagnosis

- Diagnosis classically done with 2D echo, but cardiac MRI is an alternative modality
- No current universal diagnostic criteria

- Diagnosis suggested by: 1) prominent trabeculations 2) intra-trabecular recesses with blood flow 3) two distinct myocardial layers 4) no other possible cause present

Clinical Features & Management

- Clinical presentation can range from asymptomatic to life-threatening
- Three unique features: 1) progressive CHF 2) arrhythmias 3) intra-ventricular thrombi
- Management considerations: 1) optimize CHF treatment 2) Holter monitoring 3) aspirin vs. warfarin for anticoagulation therapy