Left Ventricular Dysfunction in a Young Man

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Learning Objectives

- Isolated ventricular noncompaction (IVNC) is a rare genetic cardiomyopathy
- Familial occurrence requires screening of first degree relatives
- Prognosis is poor given limited treatment options

Case Information

History of Present Illness:
A 20-year-old Mexican man presented with dyspnea on exertion for one month. He denied any chest pain, orthopnea, PND, or lower extremity edema. He was experiencing progressively worsening palpitations over that last month. He had a 21-year-old brother who died of sudden cardiac death last year with autopsy-proven dilated cardiomyopathy.

Physical Exam:
- T 37 BP 103/65 P 73 R 18 SaO2 98%
- Thin man in no acute distress
- Regular rate, rhythm, 3/6 holosystolic murmur at apex radiates to axilla. Laterally displaced PML, no S3, I VPD
- Soft, abdomen with diffuse discomfort, no hepatomegaly
- No lower extremity edema

Pertinent Lab/Maging Findings:
- BNP 1726  AST 94 and ALT 100
- Troponin 0.15 → 0.13 → 0.17
- Echo → Ejection fraction 10%, 4 chamber dilated cardiomyopathy, 4+ mitral & tricuspid regurgitation, multiple trabeculations in left ventricle (LV)
- Cardiac MRI → hyper-trabeculated LV with multiple endocardial recesses and noncompacted myocardium
- Cardiac Catheterization → normal coronaries, cardiac output 3.8L/min, pulmonary capillary wedge pressure 18mmHg, elevated pulmonary artery pressures

Pathogenesis:

- Arrest of normal endomyocardial morphogenesis in embryonic development
  - Normal embryonic development
    - Myocardium initially a meshwork of loosely interwoven muscle fibers
    - Trabeculae alternate with recesses that communicate with the ventricular cavity
    - During weeks 5-8 ventricular myocardium is compacted and recesses turn into capillaries

Diagnosis:

Jenni Criteria (by Echo)
- Absence of coexisting cardiac problems
- 2 layer left ventricle wall structure: thin/compacted outer layer and thick/noncompacted internal layer
- Apical and mid-ventricular segments of inferior and lateral wall are affected
- Blood flow from LV cavity into trabecular recesses by Doppler

Investigative Studies

- Cardiovascular MRI: Enlarged left ventricle with deep trabeculations from noncompaction (arrow)
- Left Ventricular angiogram: Marked trabeculations (arrow) with severe mitral regurgitation
- Autopsy example: Compacted epicardial layer and noncompacted endocardial layer

Hospital Course

- Medical management for heart failure  →  Diuresed 8 liters and symptomatically improved
- Multiple runs of non-sustained ventricular tachycardia  →  ICD placed
- Close clinical follow up arranged/cardiac transplant assessment
- Siblings in Mexico contacted and screening ECHOs arranged

Discussion

- Isolated ventricular noncompaction is a rare genetic cardiomyopathy.
- Male predominance (56-82%)
- Familial and sporadic cases
- Autosomal dominant and x-linked inheritance
- Pathogenesis → Embryonic arrest of compaction. (Fig. 4)
- Diagnosis  →  ECHO gold standard (Jenni Criteria) (Fig. 5)
- Clinical manifestations
  - Heart failure (systolic) from subendocardial hypoperfusion
  - Artrial thromboembolism from thrombi in trabecular recesses
  - Arrhythmias
- Treatment
  - Heart failure management, +/- ICD, +/- anticoagulation
  - Cardiac transplantation
- Prognosis
  - Sudden cardiac death 13-18%, thromboembolic events 24%, ventricular tachycardia 41%, heart failure >75%
- Limited treatment options result in poor prognosis

References