Infiltrative and Restrictive Cardiomyopathy: Recognition by Echo

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Restrictive Cardiomyopathy

• Least common of the cardiomyopathies
• The cardiac chambers cannot stretch normally = stiff/noncompliant
• Filling is restricted
• Normal LV and RV size,
• Atrial enlargement – reflects increased ventricular filling pressures/atrial pressure
Pathophysiology

- Rigid Myocardium
- Diastolic ventricular pressure $\uparrow$
  $\rightarrow$ Venous congestion
  $\rightarrow$ 
  - Jugular vein distention
  - Hepatomegaly and ascites
  - Peripheral edema

- Ventricular filling $\downarrow$
  $\rightarrow$ CO $\downarrow$
  $\rightarrow$ 
  - Weakness
  - Fatigue
90% x small EDV = small SV
Case 1: Coach

78 year old man

- Progressive exertional dyspnea
- Peripheral edema & S3
- Echo – LVH, normal ejection fraction
Small LV
High RWT
Low
normal EF
Small LV
High RWT
Low normal EF
Tall E
Short DT
Small a wave
D dominant PV
Very low e’
What is amyloid

• Any misfolded protein that aggregates and stains with congo red (birefringence)
• Implication in pathogenesis of alzheimers disease (β amyloid)
• Systemic amyloidoses
The Systemic Amyloidoses

• Primary (AL) or light chain disease
  – Plasma cell dyscrasia
  – Immunoglobulin light chains
  – 12 month survival without treatment
  – 6 month survival with cardiac disease

• Familial (AF)
  – Mutations in transthyretin (TTR)
  – Ile 122 of particular interest
The Systemic Amyloidoses

• Senile systemic amyloid (SSA)
  – TTR-based non-genetic (ie, TTR normal)
  – Cardiac predilection
  – Male gender, onset after age 60

• Secondary amyloidosis (AA)
  – Chronic inflammatory states
Cardiac Amyloid: Summary

- Commonly mis-identified on echo
- Think of amyloid when CHF but normal LV systolic function - and low voltage ECG
- Sparkling myocardial pattern now simulated by harmonic imaging
- Clinical heart failure
- Strain evaluation useful
Amyloid heart disease: echo

• Normal ejection fraction until late
  – may look like HCM early on

Diastolic dysfunction predominates
  ✦ large atria, high grade DD

• LV walls thickened
  – but ECG: *low voltage*

• Sparkling, granular myocardial pattern – not specific

• Mild valvular thickening & regurgitation

• Peric. effusion: no tamponade
Restrictive inflow, Absent A
Depressed E prime
GS=-16.2%
Cardiac Amyloidosis

Hypertensive Heart Disease

Hypertrophic Cardiomyopathy

14mm  14mm  13mm

Mean Wall Left Ventricular Thickness
# Left Ventricular Structure and Function in Transthyretin-Related Versus Light-Chain Cardiac Amyloidosis

Candida Cristina Quarta, Scott D. Solomon, Imran Uraizee, Jenna Kruger, Simone Longhi, Marinella Ferlito, Christian Gagliardi, Agnese Milandri, Claudio Rapezzi and Rodney H. Falk

<table>
<thead>
<tr>
<th></th>
<th>Overall (n=172)</th>
<th>AL (n=80)</th>
<th>ATTRm (n=36)</th>
<th>ATTRwt (n=56)</th>
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<tbody>
<tr>
<td>E/A</td>
<td>1.6 [1–2.5]</td>
<td>1.6 [1–2.5]</td>
<td>1.3 [0.9–1.8]</td>
<td>2 [1.2–2.8]</td>
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GLS correlates with WT
- Apical cherry
- TTRwt had worst GLS but best
Corrado S., 79 year old retired ‘executive’ in waste management

From the data shown, what is the most likely diagnosis?

1. Cardiomyopathy
2. Ischemic heart disease
3. Constrictive pericarditis
4. Something else
<table>
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<th>Pre</th>
<th>Post</th>
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<tr>
<td>Heart rate (bpm)</td>
<td>62</td>
<td>76</td>
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<tr>
<td>E (cm/s)</td>
<td>82</td>
<td>100</td>
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<tr>
<td>TD e (cm/s)</td>
<td>6</td>
<td>6</td>
</tr>
<tr>
<td>E/TD e</td>
<td>13</td>
<td>16</td>
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</tbody>
</table>

Corrado S
Effect of HR on DFP and LVP
Rise in PCWP

Flat stroke volume response to exercise

Kitzman Circulation 1991
74 year old man
s/p TAVR
Still has HF
Case 2: The Attorney

58 year old with dizziness and presyncopal symptoms

left leg pain, numbness and swelling in both lower extremities.
Fabry’s Disease

- Fabry Disease
- Anderson-Fabry Disease
- α-galactosidase A deficiency
- Leukocyte levels
- Angiokeratosis
- X-linked recessive
- Affects predominantly males
- Affects homozygous females
Fabry Disease Cardiac Manifestations

- Left ventricular hypertrophy
- Hypertrophic cardiomyopathy, generally symmetric.
- Conduction defects: Short PR interval, Qt prolongation often with RBBB, tachyarrythmias, SCD.
- Aortic root dilatation
- Aortic and mitral valve insufficiency
- Ischemic disease, either due to endothelial dysfunction, microvasculature dysfunction or secondary to severe LV hypertrophy.
Clinical manifestations

Whorl-like corneal opacifications

Angiokeratomas

ESRD

CNS (CVA and TIA)
Recombinant α galactosidase Rx

- IV infusion enzyme replacement therapy reduces glycosphingolipid tissue deposition
- Can reverse wall thickness and mass

*NEJM 2001; Vol345#1: Eur J Clin Investig 2004; 34 (12):838*