Case Studies: Thick Walls, Is this Hypertrophic Cardiomyopathy

Matt Umland, ACS, RDCS, FASE
Aurora Health Care
Milwaukee, WI
Case

Amyloid Heart Disease

Amyloidosis

• Amyloid
  A waxy translucent substance consisting primarily of abnormal protein that is deposited in various tissues
  Amyloid protein is almost insoluble and once it infiltrates between the tissues they become waxy and non-functioning
Amyloid

Left ventricle

Extensive Amyloid

Cardiac Amyloid

- Deposition of fibrils composed of low molecular weight units of a variety of serum proteins in the heart

- Most common types:
  - AL-primary amyloidosis-50%
  - AA- secondary amyloid (inflammation)- 10%
  - Familial syndromes- 5%
Cardiac Manifestation

- Cardiac Amyloid involvement is progressive
- Progression of diastolic dysfunction
- Arrhythmias
- Signs/symptoms of left and/or right heart failure in absence of CAD
- Dyspnea
  - Elevated JVP
  - Peripheral edema

2D Echo Features

- Granular appearance throughout the myocardium
- Thickened cardiac valves and papillary muscles
- Significant bi-atrial enlargement (due to elevated filling pressures)
  - Increased LV/RV wall thickness
  - Thickened inter-atrial septum
  - Pericardial/pleural effusions
Medial Annulus TDI

Lateral Annulus TDI
Amyloid Strain Bullseye

Case

Friedreich’s Ataxia
Friedreich’s Ataxia

- Symmetrically hypertrophied LV
- Prominent Papillary Muscle
- Absence of SAM

Clinical/Genetic Abnormalities in Friedrich’s Ataxia

NEJM 1996 335: 1169

- Autosomal recessive neurodegenerative disorder
- Ataxia
- Onset < 20 years; relentless course
Cardiac Manifestations

- Symmetrically hypertrophied LV
- Prominent Papillary muscle
- Absence of SAM
Case

Glycogen Storage Disease (PRKAG2)

PRKAG2

• Abnormal glycogen accumulation
• Prominent hypertrophy
• Electrophysiological abnormalities
4 Chamber global strain = -4%

Apical long axis global strain = -5%
2 Chamber global strain = -5%

Case
Glycogen Storage Disease (LAMP2)
LAMP2

• Abnormal glycogen accumulation
• Prominent hypertrophy
• Electrophysiological abnormalities
Global average strain = -9%

Case

Fabry’s Disease
Fabry’s Disease

• Deficiency of the enzyme alpha-galactosidase that causes a buildup of a type of fat called globotriaosylceramide in the body
• X-linked lysosomal disorder

Cardiac Manifestations

• Usually concentric wall thickening
• Outflow obstruction is uncommon
Global average strain = -12%
Last Case

HCM or Not
4 Chamber global strain = -5%

HCM or Not??