Hypertrophic Cardiomyopathy and Beyond- Echo Hawaii 2018

Lawrence Rudski MD FRCPC FACC FASE
Professor of Medicine
Director, Division of Cardiology and Azrieli Heart Center
Jewish General Hospital, McGill University
President, Canadian Society of Echocardiography

Disclosure: Small holding of GE Stock outside managed portfolio

Utility of Echocardiography

• Diagnosis – What is the disease
• Severity & Prognostication – Is it relevant
• Guiding Therapy – treatment and procedures
• Screening – For some conditions
HCM PHENOCOPIES

- HCM/HOCM
- Amyloidosis
- Storage Diseases
- Non-Compaction
- Athlete’s Heart
- Hypertensive Heart Disease +/- CAD
- Normal Variant
- Other causes of SAM without LVH

Echo Patterns

- Dimensions and Thickness and Function
- Myocardial Appearance
- Strain Pattern
- It’s all about the MITRAL VALVE
- Non-echo correlates...History, P/E, EKG, bloods
- Complementary Imaging Modalities
- Genetics
Primary HCM

- Disorder of myocardium affecting 1:500 adults

- 30-60% genetically transmitted (mostly AD transmission)

- Phenotypic, genotypic, intragenic heterogeneity
  - More than 150 mutations affecting 10 genes encoding sarcomeric proteins identified so far.

ESC 2014 Guidelines on Diagnosis and Management of HCM
HCM Diagnosis
Hallmark of Diagnosis is:

ASH + SAM

But...
Can have HCM with:

NO SAM and NO ASH

Definition (Cont’d)

• In general, > 15 mm wall thickness
• genotype-phenotype correlations have shown that virtually any wall thickness (incl. normal range) are compatible with the presence of HCM mutant gene
• mildly ↑ LV thickness should be distinguished from certain extreme expressions of physiologically based athlete’s heart
Asymmetric Septal Hypertrophy

- Septal:Posterior wall thickness of 1.3-1.5:1
- 90% specificity for HCM but not diagnostic
- Degree and location can vary

Extent and Distribution of Hypertrophy

- B. Maron – 4 types of ASH
  - 10% anterior septum alone
  - 20% anterior and posterior septum
  - 52% septum and anterolateral wall
  - 18% ONLY posteroseptal, apical-septal, or anterolateral wall. (may miss my m-mode)
Morphologic variants of hypertrophic cardiomyopathy: A, normal or mildly hypertrophied LV (the electrocardiogram is often abnormal); B, idiopathic subaortic stenosis (IHSS) or LV outflow tract (LVOT) obstructive HCM; C, asymmetrical septal hypertrophy (ASH); D, elderly HCM; E, midcavity obstructive HCM; F, reversed ASH; G, LV wall thinning, low LVEF, and left and right atrial enlargement; H, mixed LVOT and midcavity obstructive HCM; I, apical HCM; J, cavity obliteration; K, biventricular hypertrophy and obstruction; and L, symmetric hypertrophy.

Rakowski and Wigle - TGH

Echocardiography-Guided Genetic Testing in Hypertrophic Cardiomyopathy: Septal Morphological Features Predict the Presence of Myofilament Mutations

Josepha Binder, MD; Steve R. Ommen, MD; Bernard J. Gersh, MBChB, DPhil; Sara L. Van Driest, MD, PhD; A. Jamil Taik, MD, Rick A. Nishimura, MD; and Michael J. Ackerman, MD, PhD

Mayo Clinic Proceedings; Apr 2006;

8% Gene+ 79% Gene+ 30% Gene+ 41% Gene+

FIGURE 2: Genotype status based on age at diagnosis of hypertrophic cardiomyopathy (HCM) and echocardiographic septal contour.
Does Size Matter?
LVH and Sudden Death

How do you measure the septum?

- DUNNO!
- If look at CT/MRI, no such thing as left or right septum
Mid-ventricular Form
Apical Variant

Apical Trapping and Apical Infarction
Systolic Anterior Motion (SAM)

- Anterior motion of Mitral leaflets in systole resulting in movement of leaflets into the LVOT and thus impediment to ejection of the stroke volume out the aortic valve.
- Varying degrees: mild, mod., severe (septal contact for >30% of systole)
- May result in echo-bright contact point on septum, which rarely can become nidus for IE.

SAM AND LVOTO

Late Peaking/Dagger Shaped

Latent = < 30 mmHg at rest and increasing To > 30 mmHg with Valsalva or standing Manifest = > 30 mmHg at rest
SAM - Venturi ??? Lift??? Or Drag..

If SAM is caused by the Venturi mechanism (LIFT), high flow velocity in the LVOT should be found at SAM onset.

If velocity is low at SAM onset, then lifting forces are decreased and drag forces are increased.
Septal hypertrophy $\rightarrow$ altered angle of attack $\rightarrow$ leaflet drag $\rightarrow$ LVOT obstruction

Sherrid et al. JACC 2000.

Elongated leaflets $\rightarrow$ more LVOT obstruction

Sherrid et al. JACC 2000
Hypertrophied papillary muscles obstruct LVOT

SAM – Beginning at low velocity
MR or LVOT Flow

LVOT is Late vs. Early Peaking
LVOT is Later onset
LVOT is Lower Velocity
TIPS: Get MR first & Ensure Good alignment

Compared with the previous study, the gradient is higher/lower/similar...??????
Gradients: Not always the same day in and day out!

Not All Dynamic LVOT Obstruction Is Due to HCM!

- If LV systolic function becomes hyperdynamic in patient with basal septal hypertrophy, LVOT becomes obstructed in dynamic fashion and behaves the same as HOCM
  - Elderly hypertensives – “Granny SAM”
  - Post-op intravascular depletion + inotropes (esp in patients post AVR for AS, or post MV repair with long anterior leaflet)
  - Initial presentation of amyloidosis
  - Acute LVOT obstruction: acute ant-apical MI (esp if preexisting basal hypertrophy) w/ compensatory hyperdynamic motion of inf-basal wall → SAM
  - TAKOTSUBO
MR in HCM

Schwammenthal E Circ 1998

Don’t Forget the RV
### Screening q12 months in adolescence and q5 years during adulthood

### MRI in HCM
Predicted 5-year event rates relative to LGE by % left ventricular mass for risk of end-stage HCM with systolic dysfunction, sudden cardiac death events, and total
- LV Wall Thickness
- LA size
- Maximum LVOT gradient

---

Table 6 Summary of clinical applications

<table>
<thead>
<tr>
<th>No.</th>
<th>Clinical application</th>
<th>Echocardiography</th>
<th>Nuclear imaging</th>
<th>CMR</th>
<th>Cardiac CT</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>LV dimensions, wall thickness</td>
<td>Recommended as initial test</td>
<td>Not recommended</td>
<td>Recommended with inadequate echocardiography</td>
<td>Rarely needed if echocardiography and CMR are not feasible</td>
</tr>
<tr>
<td>2</td>
<td>LV EF and regional function</td>
<td>Recommended as initial test</td>
<td>Not needed if echocardiography and CMR are available</td>
<td>Recommended with inadequate echocardiography</td>
<td>Not needed if echocardiography and CMR are available</td>
</tr>
<tr>
<td>3</td>
<td>LV filling pressures</td>
<td>Recommended</td>
<td>Not recommended if it provides only indirect evidence</td>
<td>Not recommended</td>
<td>Cannot be used for this purpose</td>
</tr>
<tr>
<td>4</td>
<td>Pulmonary artery pressure</td>
<td>Recommended</td>
<td>Cannot be used for this purpose</td>
<td>Cannot be used for this purpose</td>
<td>Cannot be used for this purpose</td>
</tr>
<tr>
<td>5</td>
<td>LA volume and function</td>
<td>Recommended</td>
<td>Cannot be used for this purpose</td>
<td>Recommended with inadequate echocardiography</td>
<td>Rarely needed if echocardiography and CMR are not feasible</td>
</tr>
<tr>
<td>6</td>
<td>Dynamic obstruction</td>
<td>Recommended</td>
<td>Cannot be used for this purpose</td>
<td>Recommended with inadequate echocardiography</td>
<td>Cannot be used for this purpose</td>
</tr>
<tr>
<td>7</td>
<td>Mitral regurgitation</td>
<td>Recommended</td>
<td>Not recommended</td>
<td>Recommended with inadequate echocardiography</td>
<td>Not recommended</td>
</tr>
<tr>
<td>8</td>
<td>Ischemia/CAD (if clinically indicated)</td>
<td>Considered if nuclear and CT not feasible</td>
<td>Recommended</td>
<td>Research application</td>
<td>Not recommended</td>
</tr>
<tr>
<td>9</td>
<td>Cardiac metabolism and neurotransmission</td>
<td>Cannot be used for this purpose</td>
<td>Research application</td>
<td>Research application</td>
<td>Cannot be used for this purpose</td>
</tr>
<tr>
<td>10</td>
<td>Monitoring of invasive therapy</td>
<td>Recommended</td>
<td>Rarely needed if echocardiography and CMR are not feasible</td>
<td>Recommended with inadequate echocardiography</td>
<td>Rarely needed if echocardiography and CMR are not feasible</td>
</tr>
<tr>
<td>11</td>
<td>Image replacement fibrosis</td>
<td>Research application</td>
<td>Not recommended</td>
<td>Recommended test</td>
<td>Cannot be used for this purpose</td>
</tr>
<tr>
<td>12</td>
<td>Screening</td>
<td>Recommended</td>
<td>Not recommended</td>
<td>Recommended with inadequate echocardiography</td>
<td>Not recommended</td>
</tr>
</tbody>
</table>
Cardiac Amyloidosis

- LVH with speckled pattern
- Biatrial Enlargement
- Restrictive Filling/Low e’
- Pericardial Effusion
- Atrial Septal Thickening
- Thickened valves

Relative apical sparing of longitudinal strain using two-dimensional speckle-tracking echocardiography is both sensitive and specific for the diagnosis of cardiac amyloidosis. Heart 2012;98:1442–1449.

Remember that Amyloidosis and Aortic Stenosis are both diseases of the Elderly
LV Non-Compaction

- Left ventricular noncompaction (LVNC) is a cardiomyopathy characterized by prominent left ventricular trabeculae and deep intertrabecular recesses
- To be distinguished from (How?) LV hypertrabeculation – often seen in normal
- MRI required as echo insufficiently sensitive or specific...but remember, MRI ≠ TRUTH
LV Non-Compaction

Left ventricular noncompaction (LVNC) is a cardiomyopathy characterized by prominent left ventricular trabeculae and deep intertrabecular recesses. To be distinguished from (How?) LV hyper-trabeculation – often seen in normal... MRI required as echo insufficiently sensitive or specific...but remember, MRI ≠ TRUTH

First one I ever saw at my center.
Contrast is Key
How About This Guy?
64 year old with CVA and mild hypertension

Which of these has Fabry’s?

Courtesy Dr. F. Weidemann
LV Hypertrophy – Correlation with Enzyme and Screening in LVH/HCM Populations

"Prototypical" Fabry
Myocardial Fibrosis

*Grey zone* of LV wall thickness (13–15 mm)

Athlete’s Heart vs HCM

Maron, B J Heart 2005;91:1380-1382
Summary

- HCM presents in numerous forms
- Echo is primary imaging modality for diagnosis and prognosis but complementary imaging AND CLINICAL/SEROLOGIC/BIOCHEMICAL correlated
- Contrast and Strain Imaging Helpful
- Keep a broad differential diagnosis as many mimickers including NORMALS