Hypertrophic Cardiomyopathy

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Disclosures

No Real or Potential Conflict of Interests for this Talk

Off-label use
Gadolinium MR contrast for the Heart
Outline

HCM intro / background
LV morphology / function
Focus on Echo, but **multimodal imaging** valuable
Role of UCA – don’t miss HCMap
Seeing SPIRAALS
Importance of MV assessment
Case Example

HCM background

Genetic disorder, sarcomeric mutation

Results in LVH – (exclude other etiologies)

All ages are at risk

- interstitial fibrosis, fiber disarray, impaired function

Preserved function with conventional measures

- impaired myocardial mechanics
Value of Echo

LVH description
LV sys / dias fnx
RVH / RVOT
RVSP
LVOT – *provocation*
MV anatomy / physio
Myectomy guide
Screening relatives

Table 1: Echocardiographic evaluation of patients with HCM

1. Presence of hypertrophy and its distribution; report should include measurements of LV dimensions and wall thickness (septal, posterior, and maximum)
2. LV EF
3. RV hypertrophy and whether RV dynamic obstruction is present
4. LA volume indexed to body surface area
5. LV diastolic function (comments on LV relaxation and filling pressures)
6. Pulmonary artery systolic pressure
7. Dynamic obstruction at rest and with Valsalva maneuver; report should identify the site of obstruction and the gradient
8. Mitral valve and papillary muscle evaluation, including the direction, mechanism, and severity of mitral regurgitation; if needed, TEE should be performed to satisfactorily answer these questions
9. TEE is recommended to guide surgical myectomy, and TTE or TEE for alcohol septal ablation
10. Screening

LV Wall Thickness

- Preferentially involves the [basal] IVS
  - AL, IL, and apex may be involved
- Unexplained WT >15mm (any segment)
  - measure all 16 segs *(less from base to apex)*
- IVS/PW ratio >1.3 (normotensive) or >1.5 (HBP)
  - interstitial fibrosis, fiber disarray, impaired function

*Carefully assess apical and anterolateral segments*
Differential

LVH
HCM
ESRD
Cardiac Amyloid
Glycogen-Storage Diseases
Anderson-Fabry’s Disease
Freidreich’s Ataxia
Thick LV walls
Septal Hypertrophy

Diffuse Hypertrophy

Apical Hypertrophy

Spiral Hypertrophy

Amyloidosis
Anterolateral regional HT

CMR sole dx of HCM phenotype in a minority of patients (5%)
* particularly the AL LV free wall

Typical EKG (echo to exclude CAD)
? Apical Dyskinesis?
Apical-variant HCM

2DE:
LV apical aneurysm
SAM

Not specific for HCM

May occur with or without LVOT obstruction

Due to hydrodynamic ‘drag’ and LV ‘pushing’ force

Exclude: Small hyperdynamic LV, MV repair, TGA, anomalous pap muscle, anterior infarct
Mitral Regurgitation

SAM creates impaired MV leaflet coaptation

- Eccentric *(away from the LVOT)*
  - If central (or anterior) MR jet
    - Suspect intrinsic MV pathology
Mitral Valve Pathology

172 HCM & 172 controls
- AMVL 26 vs 19mm
- PMVL 14 vs 10mm
AMVL/LVOT ratio >2.0
- Greater LVOTO

Abnormal pap muscles
- Accessory muscles
- Aberrant insertion

Normal Apical displacement
Abn chordal attach
Bifid PM
$PM_{LVH}$ with Mid-cav oblit
Elongated AMVL
LVOT obstruction

Rest > 30mmHg (outcome = HF, death); <30 *NOCM

Typical ‘dagger-shaper’ Doppler spectrum
- Lobster Claw: midsystolic decrease in LV ejection velocity from truncation of longitudinal shortening

Most ‘rest’ <30mmHg (>50 = therapeutic target)
- *provocation: ROUTINE Valsalva

LVOT provocation

Recommended for EVERY HCM echo study

- Doppler echo @ rest only **is not adequate**
- semi-supine, sitting, standing (if needed)

**TM exercise Doppler (if LVOTO < 50mmHg)**

- Dobutamine **NOT recommended**
- Amyl Nitrite – simple, quick, readily available

Lying to standing; *squatting to standing*: simple
Systolic Function

Normal LVEF / FS%
- Abnormal global strain, regional mechanics

Once LVEF falls, mortality >10% / yr
- sudden death rate also increased

“Burned-out” HCM (small V: isch / infarction – fibrosis)
- LV thinning / dilation, low EF

TDI: e’ & s’

Pulsed-wave / color mode
- high-amplitude, low-velocity signals (tissue V)

Systolic V (longitudinal function) is reduced

Classic Clinical Research trial (Nagueh et al)
- Early mitral annular V < controls (LVH +/-)

E’ <13cm/s ➔ Se 100% / Sp 93% for gene + HCM

S’ <9cm/s ➔ pathologic LVH vs physio LVH

S’ <4cm/s ➔ prognostic (death, HF admission)
3DE

LV volumes, function, mass
LA volumes, function
Mechanistic insights into SAM
  * Deformational geometry of LVOT
    * Medial aspect of MV = SAM
    * Resultant lateral narrow LVOT

Post ETOH / repair
It’s HCM, not Athletic

~1% Olympic level athletes have WT >15mm (and diffuse LVH pattern)

LVEDD <45mm (not >55mm) … and concentric vs eccentric

Poor EX (vs supernormal / superhuman)

Low Em’ < 9cm/s

Diastolic dysfunction

No regression after termination of training (good luck….)

Provocable LVOTO

Reduced strain (longitudinal, circum, radial)… & dyssynchrony

CMR fibrosis / ischemia

Eur J Appl Physiol 2004;92:592-7
CCT
CMR

- Provides opportunity for growth in ECHO skills
  - Recognition of **apical HCM** appearance
  - Improved understanding of MV pathology, **spirals**
- Particularly useful for presence, location, extent
- ~10% very focal / localized and missed by echo
- ~30% have RVH (RVOTO ~ septal-marginal bundle)
Assess LV & RV volume and function
- Simpson’s rule (preferred > biplane)
- LVOTO +/- SAM (MR +/- MV disease)

Determine maximal WT and LV mass
- Regional vs global (spiral)
- Septal vs apical
- RV involvement

Location / extent LGE
* Differential Diagnosis *
AS; athlete; LVCN; Sarcoid; Amyloid; Fabry; LVH + CAD
Assessment of Fibrosis

The presence of replacement fibrosis can be detected using the inversion recovery late enhancement technique following gadolinium-DTPA administration (LGE).

Fibrosis is the main component of chronic MI which is visible with the LGE.
Fibrosis imaging in HCM

LGE in INFARCT

LGE in HCM
HCM – a wide variety of scar

Normal LGE    Extensive Scar

Many patients have no detectable scar
LGE Prognosis

- N = 217 HCM
- LGE + vs LGE -
- 1° endpoint:
  - Death
  - VT or VF
  - ICD implant
  - ICD firing
  - Hosp CV admission

Various potential mechanisms for LGE in HCM:
- plexiform fibrosis (myocyte disarray)
- expanded interstitial fibrosis
- macroscopic replacement fibrosis (ischemia / infarct)

Regardless: LGE+ is a poor prognostic marker

Seeing Spirals

LH-spiral common; clinical significance unknown
86%; Belgian and Italian population
Older, more HBP, LV mass, LVOTO
? Less CMR LGE

Modified Centerline 180 Chord Technique
N = 132; all with ASH-CM; CMR performed

HT (hypertrophy) defined as:
>15mm; >1.3 ratio (HT to normal)
Radial, circumferential, and longitudinal extent

Results

Mean WT 22±5mm
Circumferential extent 131°; long 64%
HT basal AS, counter-clockwise (LH) [viewed from apex]

Florian A, et al. JACCi 2012; 5 (7): 702-711 (Reichek N. Editorial 712-714)
CMR “spirals”

CMR rat:
- 25 X 25 X 39µm
- 3D fiber sheet continuity
- counter-directional subendo & subepi architecture

UK mouse CMR:
- 7T
- black blood functional
45M, dyspnea, syncope, obesity
Peak gradient <20mmHg
Exercise (Doppler)

Peak gradient >100mmHg
Peak gradient >120mmHg and severe eccentric MR
Why Contrast guided?

UCA on LEFT side of IVS

UCA on RIGHT side of IVS
ETOH Ablation

Note: ICD placed (unexplained syncope)
ETOH Ablation
Guiding Myectomy

- Visual inspection limited = imprecise resection
  - Too large = VSD; too small = residual LVOTO
  - TEE monitoring is required
Echo Screening

- Family members of genetic + / SCD
  - Annually through adolescence (active phase)
  - Every 5 years for adults (exclude late onset)
  - At consideration for competitive athletics

- Echo should be comprehensive (pre-clinical)

Consider CMR for TDE echo or serial ECG change
Prognosis

- Massive LVH
  - $> 30 \text{mm (echo)}$; $? > 27 \text{mm CMR}$
- Resting LVOTO $> 30 \text{mmHg}$
- Burnt-out HCM (thin wall; EF $< 50\%$)
- LV aneurysm
- LAVI $> 34$
- LV scar burden (selected patients)
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<td>WT &gt;30mm</td>
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<td>Unexplained syncope</td>
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<td>Non-sustained VT</td>
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<td>* Other risk modifiers?</td>
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<td>BP fall with stress</td>
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* LVOTO; CMR LGE; 25-29mm; apical aneur; gene mutation

JACC 58: e212; 2011
Mr Google: HCM risk calculator

ICD – YES or NO?

25 years old
25mm max LV WT
25mmHg (Valsalva)
3 beat run NSVT
Conclusions

Echo – Excellent initial modality of choice
- Report the $WT_{\text{max}}$, regional distribution
- Evaluate MV apparatus carefully
- Recognize limitations (role for CMR or CCT)

Screen (imaging or genetic [when gene is known])
- Kids annually; Adults q 5 years (annually if athletes)

Provoke gradients routinely (lowest gradient = LLD position)
- Valsalva, position change, Amyl, exercise (not dobutamine)
South Rim, Grand Canyon

Photo: Vince Sorrell
History of Imaging

47 years ago Shah & Gramiak et al. Circ 1969
1st M-mode LVOTO from SAM

31 years ago Higgins et al. AJC 1985
1st CMR in HCM

4 yrs ago Florien et al. JACCi 2012
Spiral architecture of myocardial fiber array
Conventional Diagnostic Criteria for HCM

Non Apical HCM variants: IVS/PW > 1.3, IVS > 15 mm, SAM/LVOT obstruction
Apical HCM: exuberant apical LVH, spade-like LV cavity

HCM Diagnosis Unequivocal

Consider Risk Stratification

Echo-Doppler Predictors of Adverse Prognosis Non-Apical variants
- LVH (>30 mm)
- LVOT gradient (>30mm Hg)
- LV dysfunction (EF < 50 %)
- LA dilation (> 48 mm) or LAVI > 34 ml/m²
- Intraventricular dyssynchrony (> 45 ms) *

Differentiation from Pathologic LVH Criteria favoring HCM
- TDI longitudinal strain (> -10 %)
- Paradoxical longitudinal strain (linear mapping-TDI)
- Attenuated regional radial and circumferential strain (2D strain)
- Intraventricular dyssynchrony

HCM-Diagnosis Equivocal

Non-Apical HCM variants

Apical HCM
- 2D contrast-enhanced echo
- RT-3DE
- RT-3DE with contrast
- 2D-strain (paradoxic apical strain)

Differentiation from Physiologic LVH and Normals† Criteria favoring HCM
- Systolic mitral annular velocity(< 9 cm/s)
- Diastolic dysfunction (TDI)
- Lack of LVH regression post cessation of exercise
- Lack of LV dilatation
- Provocable LVOT gradient
- Attenuated of longitudinal, circumferential and radial strain (2D strain)†
- Intraventricular Dyssynchrony†
Extra Question

- Exercise in patients with known HOCM:
  1. Should be avoided if there is any resting gradient
  2. Contraindicated if exercise gradient >50mmHg
  3. No restrictions for any exercise
  4. Low to moderate intensity exercise is actually recommended
Challenges

Late presentation (burned out phase)
  - LV dilation and thin LV walls

Physiologic LVH – unable to stop training

Co-existent pathology (HBP and HCM)

Isolated basal septal hypertrophy
  - elderly Females (may have SAM)