Hypertrophic Cardiomyopathy
Challenging Questions

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• No disclosures
Hypertrophic Cardiomyopathy

- ? Genetics ?
- ? Myectomy ?
- ? Natural history ?
- ? ICD ?
Hypertrophic Cardiomyopathy

• Question 1: what about genetics and HCM?
Hypertrophic Cardiomyopathy

Old Definition: Severe hypertrophy of the myocardium in the absence of a known etiology.
"Essentially, HCM is always a genetic disorder..."

Marian and Roberts
Circ 1995: 92: 1336-1347
Over 16 defective sarcomeric genes with over 500 different mutations

2 nonsarcomeric genes (PRKAG2 and LAMP2)
Mutation

Poison polypeptide

Altered sarcomere structure and function

Compensatory Hypertrophy
Diagnosis of HCM - Year 2020

- Diagnosis
  - Genotype
- Clinical Severity
  - Echo
Early age
Spontaneous LVH

Massive LVH
Reverse curve septum
Sudden death

Later age
Triggered: HTN, AS

Mild LVH
Sigmoid septum
CHF, AF

Age related
Hypertrophic Cardiomyopathy

• Genetics - Clinical implications
  – Screen all first degree relatives
    • Adults every 5 years
    • Adolescents every year (especially athletes)
  – Obtain family history of HCM or sudden death
Hypertrophic Cardiomyopathy

• Question 1 : what about genetics and HCM?
• Question 2 : what about different types of myectomy?
LVO obstruction
Caused by SAM
Present in 2/3 pts with HCM
Major cause of symptoms
Ischemia
Diastolic dysfunction
Mitral regurgitation

Gradient 100 mmHg

LAP 45 mmHg
Relieve obstruction
Septal myectomy
Transaortic approach
Operative risk < 1%
Results
Gradient < 10 mmHg
No residual MR
Complete relief sx

Gradient 10 mmHg

LAP 12 mmHg
Septal Myectomy – 10 yr F/U
Symptomatic Improvement

Mayo Data
350 patients

Before
Postop

% Pts

I-II
III-IV
Hypertrophic Cardiomyopathy

It is clear that surgical myectomy will result in marked long-lasting symptomatic improvement in over 90% of patients with severe symptoms and obstruction.
Septal Myectomy – Long term outcome

Survival

Years

Myectomy
Non-Obstructive HCM
Obstructive HCM

Ommen et al: JACC 2005
Maron et al: NEJM 2003
Apical Variant
HCM
Apical HCM

Diffuse symmetric T wave inversions

Disease of diastole

Abnormal effective operative compliance
Novel “apical myectomy”
Remove massive myocardium
Through apical approach
Increase LV volume
Improve effective operative compliance
Improve symptoms of dyspnea
Adverse natural history apical HCM

Apical ischemia

Apical aneurysm + mid obstruction
Once development of apical aneurysm

- More symptoms
- Higher risk VT
- Increased risk embolic events
May need contrast enhancement to identify apical aneurysms
Another type of “myectomy”

- Transapical approach
- Resect midventricular obstruction
- Resect apical aneurysm
Hypertrophic Cardiomyopathy

• Question 1: what about genetics and HCM?
• Question 2: what about different types of myectomy?
• Question 3 and 4: what about natural history and sudden death?
Sudden death in HCM pts
Sudden death in HCM pts

Initial studies at referral centers
High annual mortality (4-7%/yr)
Majority due to sudden death
Sudden death in HCM pts

Unselected populations
Much lower mortality
Comparable to general population
Sudden death in HCM pts

Low Annual Mortality Overall

- Koffland: 0.8%
- Maron: 1.3%
- Spirito: 0.6%
- Cecchi: 0.7%
- Mayo: 1%

Most Common Cause Sudden Death In Young

1. Coronary HD
2. Ruptured aorta
3. Coronary anomalies
4. Idiopathic LVH
5. HCM 48%
Sudden death in HCM pts

✅ Unpredictable - years go by

✅ AICD can be lifesaving

✅ Can we predict who is at risk?
2011 ACCF/AHA Guideline for the Diagnosis and Treatment of Hypertrophic Cardiomyopathy

A Report of the American College of Cardiology Foundation/American Heart Association Task Force on Practice Guidelines

Developed in Collaboration With the American Association for Thoracic Surgery, American Society of Echocardiography, American Society of Nuclear Cardiology, Heart Failure Society of America, Heart Rhythm Society, Society for Cardiovascular Angiography and Interventions, and Society of Thoracic Surgeons

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Risk Factors – Literature

- Out-of-hospital arrest
- Sustained VT
- Family history sudden death – HCM
- Massive hypertrophy
- Syncope
- NSVT on Holter
- TMET – VT or drop BP
Sudden death in HCM pts

<table>
<thead>
<tr>
<th>Maximal wall thickness (mm)</th>
<th>Incidence of sudden death/1,000 person-yr</th>
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<tbody>
<tr>
<td>&lt;15</td>
<td>0</td>
</tr>
<tr>
<td>16-19</td>
<td>2.6</td>
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<tr>
<td>20-24</td>
<td>7.4</td>
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<tr>
<td>25-29</td>
<td>11.0</td>
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<tr>
<td>&gt;30</td>
<td>18.2</td>
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NEJM 342:24:1778, Sept 19, 2000
ACC/AHA Guidelines

Really Bad
- Arrest
- Sustained VT

Bad
- FH HCM and Sudden death
- LVH > 30 mm
- Unexplained syncope

Somewhat Bad
- NSVT
- BP drop TMET
- LVO
- CAD
This is the best that we had..

But positive predictive accuracy < 15%

20-25% incidence
Inappropriate shocks
(younger active population of HCM)
Sudden death in HCM pts

What’s New?

✓ Age is important

✓ MRI with gadolinium

✓ Risk scores
HCM Pts > 60 y/o

Sudden death
0.2%/yr

Maron BJ et al
Circulation 2013:127:585
Sudden death in HCM pts

Age is important

✓ 76 y/o man with syncope after getting up in the middle of the night

✓ 23 y/o college student who suddenly passes out in class
Myocyte Disarray
Gadolinium Enhancement Defects on MRI

1293 HCM pts – F/U 3.3 yrs

Incidence SCD events

<table>
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<th>% Late Gadolinium Enhancement</th>
<th>0</th>
<th>&lt;10%</th>
<th>11-19%</th>
<th>&gt;20%</th>
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<td></td>
<td>0</td>
<td>5</td>
<td>20</td>
<td>25</td>
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Chan et al: Circulation 2014:130:484-495
HCM Risk-SCD Calculator (ESC)

- Prognostic model from retrospective multi-center longitudinal cohort study
- 3675 consecutive patients from 6 centers
- Follow-up 5.7 years
- 198 (5%) died suddenly or AICD fired

O’Mahony et al European Heart Journal 2014:35:2010
HCM Risk Score

Low risk
5 yr < 4%
No ICD

Intermediate risk
5 yr 4-6%
ICD may be considered

High risk
5 yr > 6%
ICD should be considered

ESC guidelines
European Ht J 2014
Patients who had appropriate firings from ICD vs ESC risk score

Maron et al: AJC 2015

60% of patients who had appropriate ICD firings were “low risk”
Patients who had appropriate firings from ICD vs ESC risk score

“Missed”

- Young Severe LVH
- Young Strong FH

22% of patients who had appropriate ICD firings were “low risk”

Mayo Data
Putting it altogether
Consideration of AICD in HCM

FH sudden death
1° relative
Sudden death < 40 y/o
Sudden death with HCM

Massive LVH
> 30 mm
> 25 mm*
young age
MRI GE

Syncope
Unexplained
Young
Recent

All others: Calculate ESC risk score
Consider AICD if 5 yr risk > 6%
Putting it altogether

Consideration of AICD in HCM

This needs to be a shared decision making process involving the needs and preferences of the patient and resources available.
Hypertrophic Cardiomyopathy

- ? Genetics ?
- ? Myectomy ?
- ? Natural history ?
- ? ICD ?
48 y/o man with HCM – Asymptomatic
No FH HCM or sudden death
On lopressor 200 mg per day
Septal thickness 3.2 cm
LVO gradient 90 mmHg
Normal coronaries – 2 mm septal
What would you do now?

1. Implant ICD
2. Add disopyramide
3. Septal ablation
4. Septal myectomy
HCM Related Death or Adverse Clinical Events in 70 Patients with LV Apical Aneurysms

- Survival free from HCM related mortality and adverse events

- Log-rank test p<0.001

- 1.7%/year

- 8.1%/year

- HCM patients without LV apical aneurysms
- HCM patients with LV apical aneurysm

Years from First Evaluation