Hypertrophic cardiomyopathy and family screening

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The consultation....
"My son here is 17, and I am quite worried about him. He got very breathless last week after his rugby game, and he just didn’t feel right. The coach was worried that he might have that SADS condition. You know my husband’s nephew died when he was 26, and it was very sudden"

- Investigate the dyspnoea
- Consider a cardiac cause
- Look for more information on the family history

Evaluate the patient with symptoms

- History of syncope, seizures, febrile convulsions
- Symptoms limiting exercise? or afterwards?
- Medication use / performance drugs
- Physical examination – vital signs including BP, murmur, effect of exercise on murmur, wheeze, stigmata of systemic conditions (Marfan’s, neuromuscular diseases, rashes eg erythema nodosum)
- ECG
- Consider echo and chest x ray
- Three-generation family tree

Post mortem analysis

- Looking for red flags for a cardiomyopathy
- Increased heart weight
- Asymmetrical left ventricular thickening
- Histology – fibrosis, myocyte disarray

Left ventricular hypertrophy does not always mean HCM
Consider HBP, look for presence of atheroma...

Family screening approach for cardiomyopathy in adults

3.3. Female age 59. "Heart problem"
2.2. Male SCD age 29
2.4. Female, died age 76 with heart failure and AF
4.7. Our proband
Clinical screening

• History, physical examination
• ECG and echo
• ECG changes can precede echo changes!
• On Echo: HCM is present if there is LV wall thickening in diastole ≥15mm, or ≥13mm in first degree family members of a HCM patient.
• Cardiac MRI can help with borderline cases
• Screen first degree relatives – and second degree, if athletic or symptomatic
• Clinical screening every 3-5 years in at-risk persons.

Confirmed HCM

• Lifestyle changes - "Sports for fun"
• Risk stratification – with personal history of syncope, family history of SCD, left ventricular wall measurements, and presence or absence of arrhythmia
• Implantable cardiac defibrillator if ≥1 risk factor
• B blockers commonly used (for Left Ventricular Outflow Tract gradient and arrhythmia prevention)
• Calcium channel blockers / Disopyramide / Advanced interventions
• Yearly specialist assessment.

Genetic testing in HCM

• 11-gene HCM panel typically offered to HCM probands.
• Quoted mutation identification rate between 60-70%
• Challenges – “private mutations”
• Challenges – need for segregation studies, and variable penetrance and expressivity
• Challenges – informed consent
• Genetics is an important adjunct to, but does not replace, clinical assessment.

Summary

• Identifying and verifying the presence of HCM is key
• Post mortem report – or information from the treating cardiologists
• Clinical screening is fast, though a negative screen does not rule out future development of HCM
• Screen first degree relatives and some second degree.
• Repeat screening is offered every 3-5 years in the adult population, and more frequently to age of ~22
Patient referral

• Family members of patients with confirmed HCM
• Any details available on the affected family member
• Family history
• Send to: Family Heart Screening Clinic, Mater Heart House, 54, Eccles St, Dublin 7.
• 01-803-4431
• Email familyheartscreening@mater.ie