In the Weeds: Identifying the Underlying Etiology in a Patient with Suspected Hypertrophic Obstructive Cardiomyopathy

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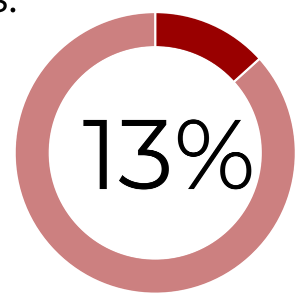
Introduction

Hypertrophic cardiomyopathy (HCM) is one of the most prevalent cardiomyopathies.[1] Hypertrophic Obstructive Cardiomyopathy (HOCM) is a subtype of HCM, in which the interventricular septal myocardium hypertrophies disproportionately and obstructs the outflow tract from the left ventricle to the aorta. Symptomatic patients are classically young athletes who experience decreased cardiac output, syncope with exercise, or sudden cardiac death due to ventricular arrythmias.



of estimated HCM cases go undiagnosed

Diagnosis of HOCM is made only with echocardiogram or genetic testing, which can at times result in missed diagnosis if patients do not have pronounced echo features or identified genetic mutations.

Case Description

- 57 y.o. female
- Presented with grade III/IV systolic murmur.
- Murmur documented for 4 years with no further workup.
- After 4 years, a referral was made to cardiology. Echocardiograms were obtained.
- After imaging, there was a high clinical suspicion for HOCM, but due to a history of uncontrolled hypertension and missing medical records, hypertrophy secondary to hypertension could not be ruled out.
- Genetic testing for HOCM was negative, despite patient having extensive family history of cardiac disease.
- Further referred for a cardiac MRI, and while hypertension could not be ruled out as a contributing factor, HOCM was placed as the leading differential.
- Ultimately, the pt went to the operating room for ventricular septal myectomy/cryoablation, aortic valve replacement, and maze of left atrium for atrial fibrillation

Timeline

2005 — First time medical records indicated a murmur

May 2013 — "Murmur sounded louder today."; III/VI systolic ejection murmur"

May 2014 — First transthoracic echocardiogram (TTE) ordered

Jan 2015 — Cardiology examined pt. and referred for transesophageal echocardiogram (TEE)

June 2015 — Pt. went for TEE but could not tolerate procedure. PCP referred her back to cardiology.

Sept 2015 — Pt. lost to follow up

Feb 2016 — Appt. with genetic counselor and repeat TTE ordered

May 2016 — 24 gene panel result is negative

May 2020 — Cardiology appt. "Need workup for HOCM." Ordered Cardiac MRI

June 2021 — Cardiology appt. "equivocal HOCM diagnosis, suspect hypertensive disease.

Dec 2023 — Myectomy

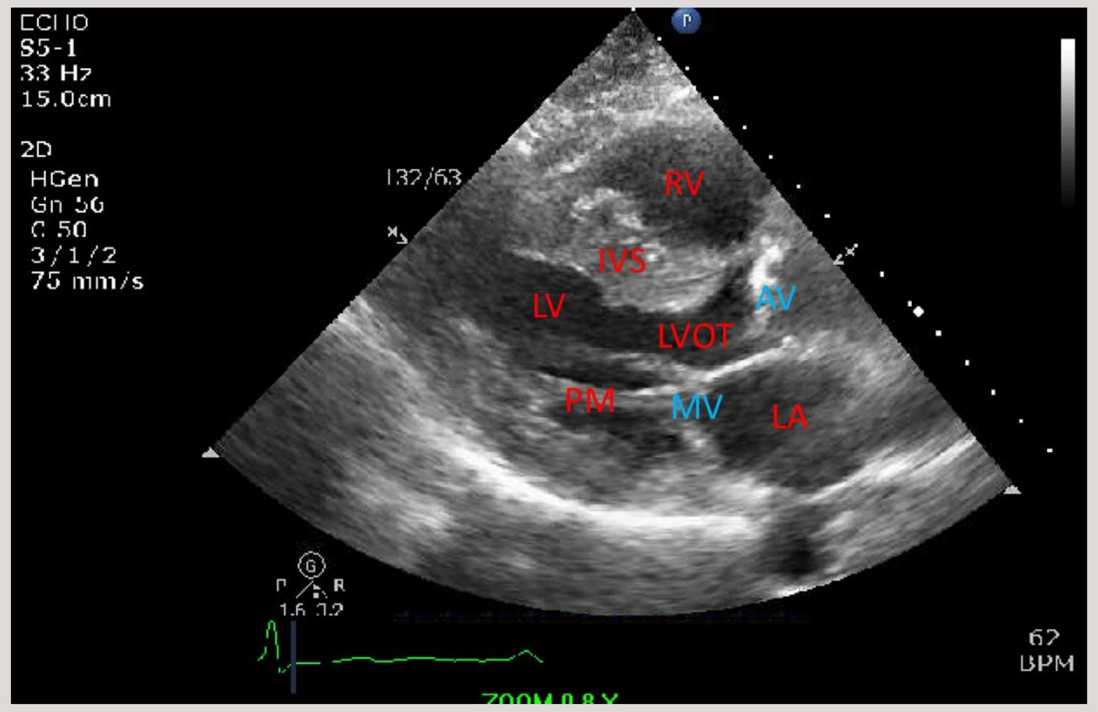


Figure 1: Parasternal long axis view of first TTE completed in 2014.

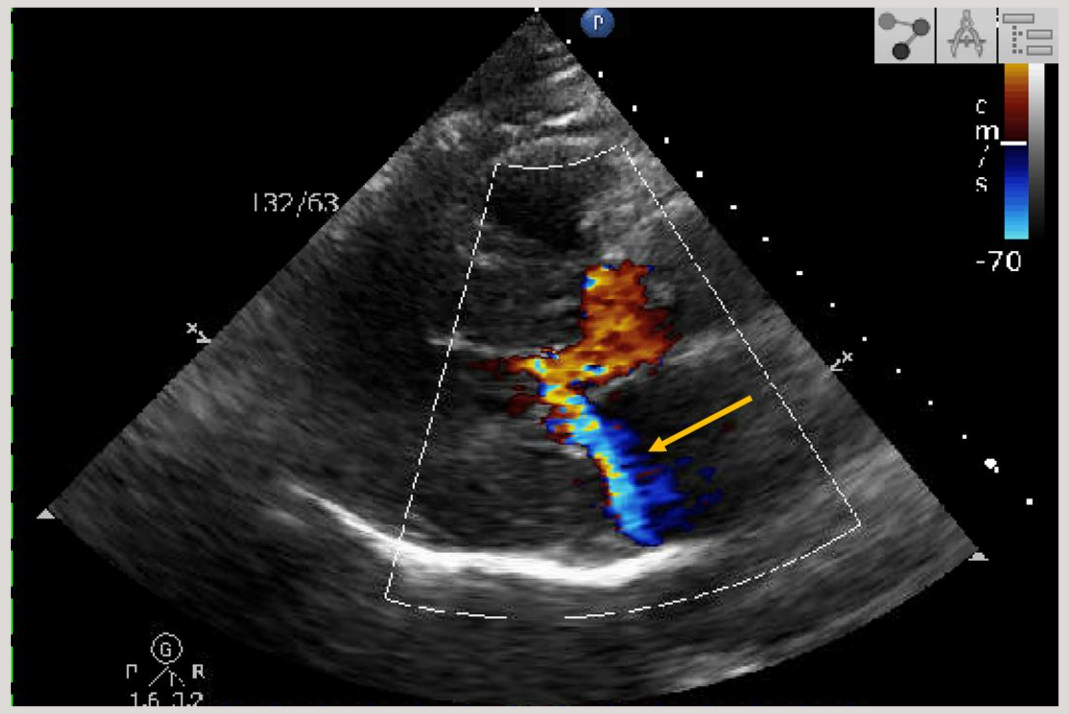


Figure 2. Parasternal long axis view of first TTE completed in 2014 with color Doppler jet, illustrating mitral regurgitation related to systolic anterior motion.

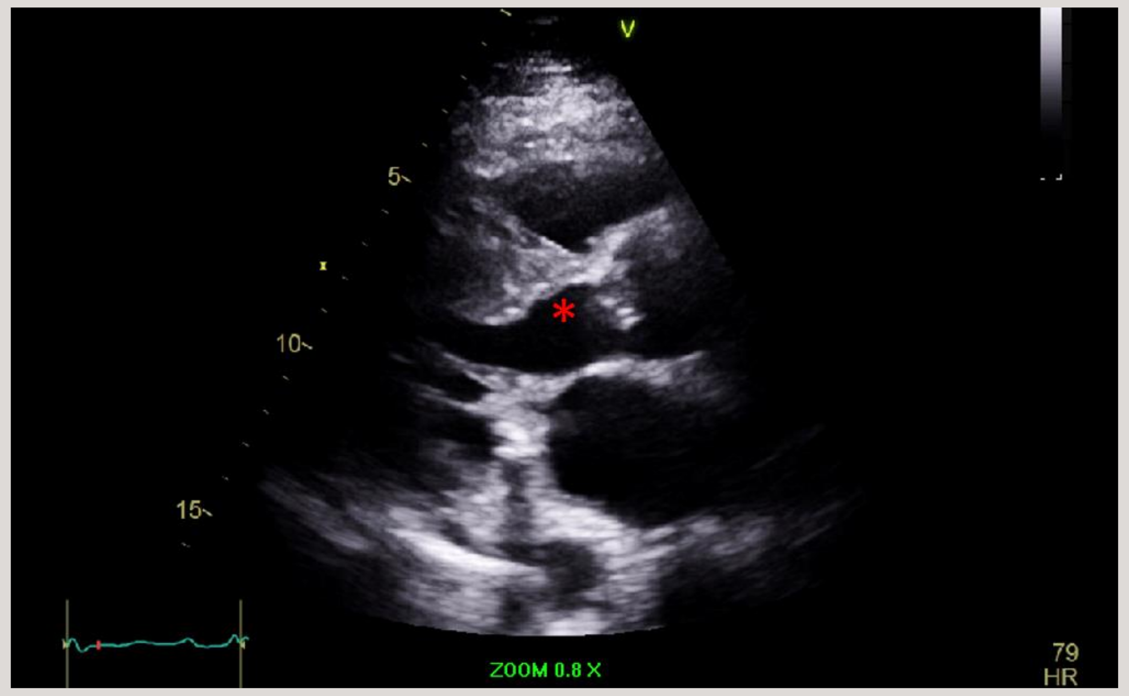


Figure 3: Parasternal long axis post 2023 myectomy. Asterisk denotes area of myectomy.

Clinical Significance

This case highlights several shortcomings of the medical system:

1. Fragmentation of EMR
- Absence of the pt's records prior to 2011 (pt obtained care at a different healthcare institution); thus, the duration and presence of both the initial murmur & HTN is unknown

2. Discontinuity in Patient-Provider Relationship
- At every cardiology clinic appt the pt was seen by a different provider who had varying interpretations of the radiologic imaging
- Inconsistencies in the diagnosis of HOCM vs HCM due to longstanding HTN

3. Provider Bias
- HOCM classically presents in young people in their second or third decade of life
- This pt was a black woman in her fifties. It is possible that the diagnosis of HOCM was not considered initially as a result
- By the time a full work-up had been conducted, her HTN had been poorly controlled for many years, making delineating between a genetic versus hypertensive cause of HOCM difficult

4. Lack of Scientific Knowledge
- The patient was tested for 24 gene mutations associated with HOCM and found to be negative for all of them
- Extensive family history of coronary artery disease and myocardial infarctions – high likelihood of genetic predisposition
- There are likely more genetic mutations that are linked to HOCM that have yet to be discovered

In all, this case sheds light on the inadequate diagnostic practices being relied on for the identification of HOCM and may explain why this cardiomyopathy is so grossly underdiagnosed.

References

1) Tuohy, C. V., Kaul, S., Song, H. K., Nazer, B., & Heitner, S. B. (2020). Hypertrophic cardiomyopathy: the future of treatment. European journal of heart failure, 22(2), 228–240.