Hypertrophic Cardiomyopathy

"Diagnostic Work-up of concomitant mitral valve pathology"

10 min

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Centro Cardiologico Monzino, IRCCS
Milan
ITALY

Palermo,
October 24 and 25

Hypertrophic CMP

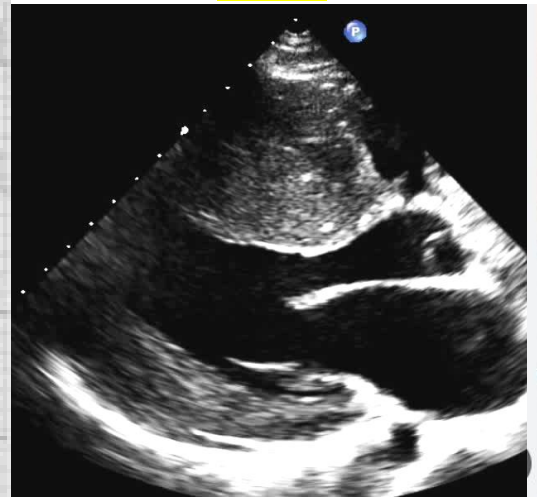
Diagnostic work-up

Clinical data

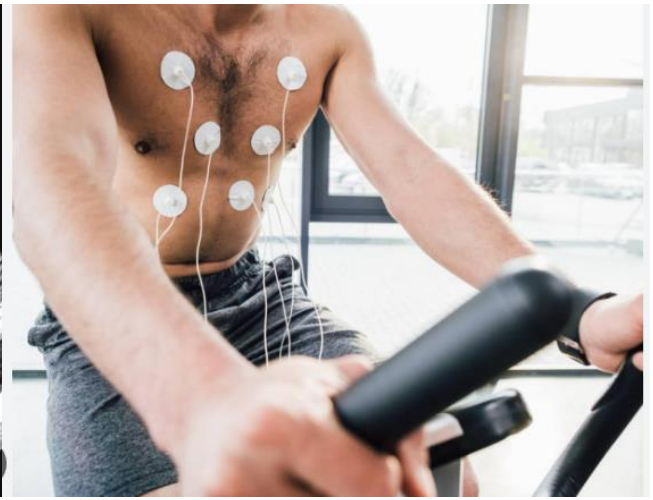
Genetics

ECG

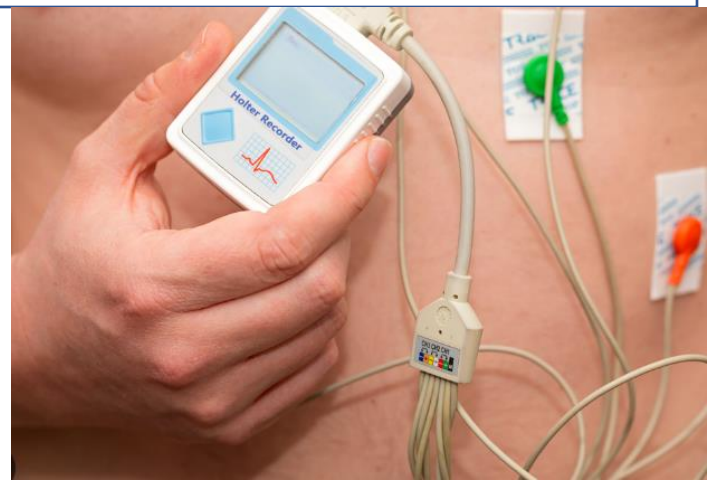
ECHO



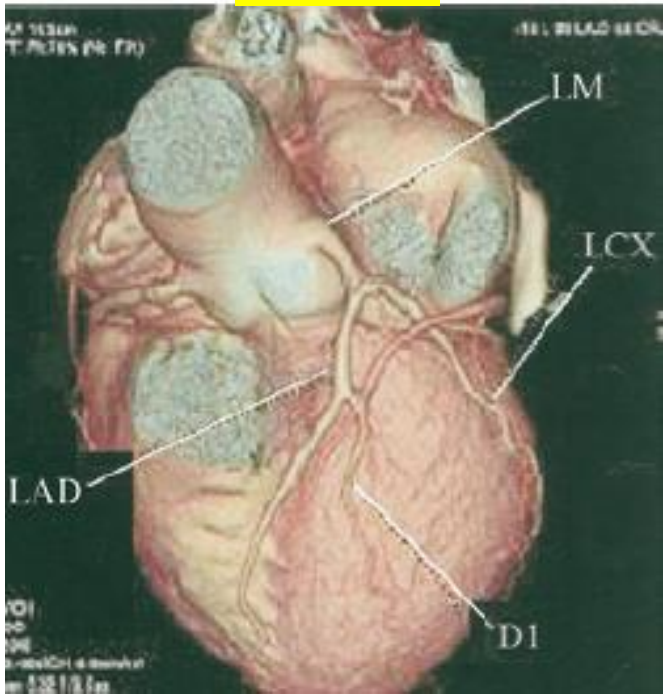
EXERCISE (ECG /ECHO)



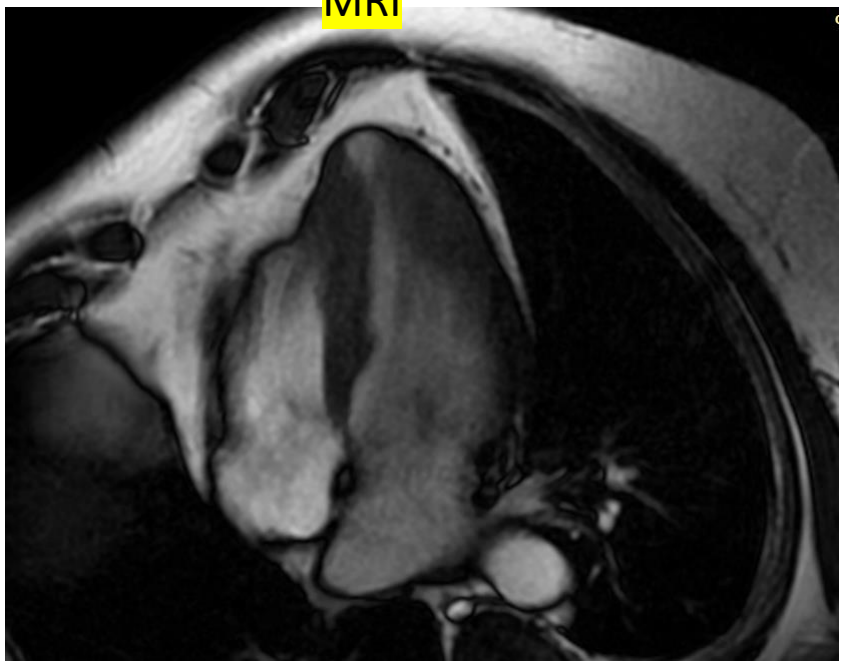
HOLTER

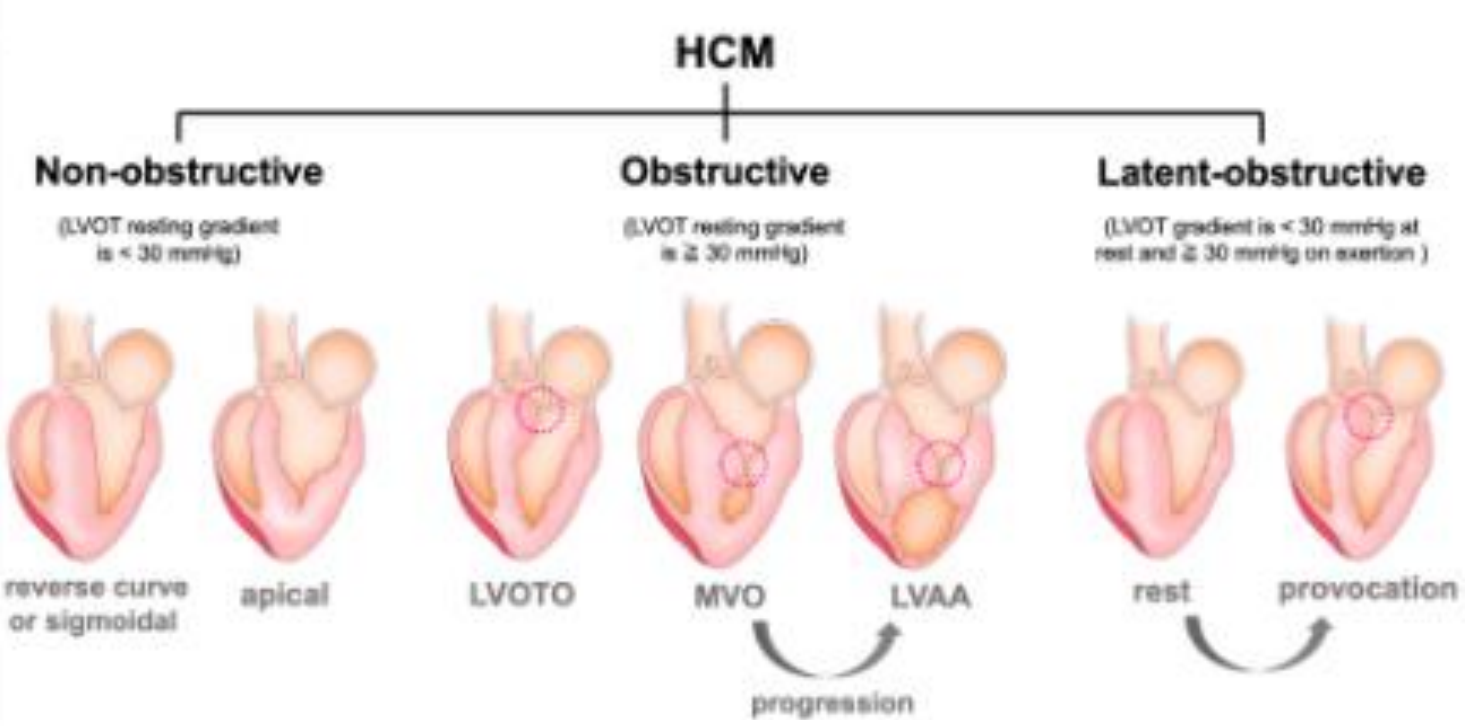


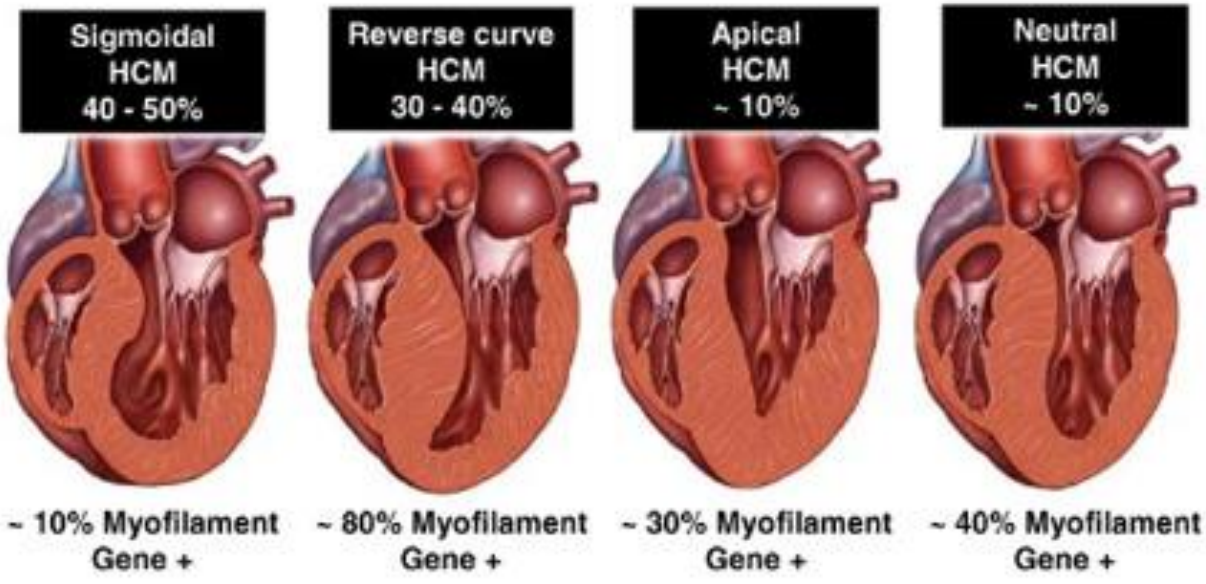
Cardiac CT



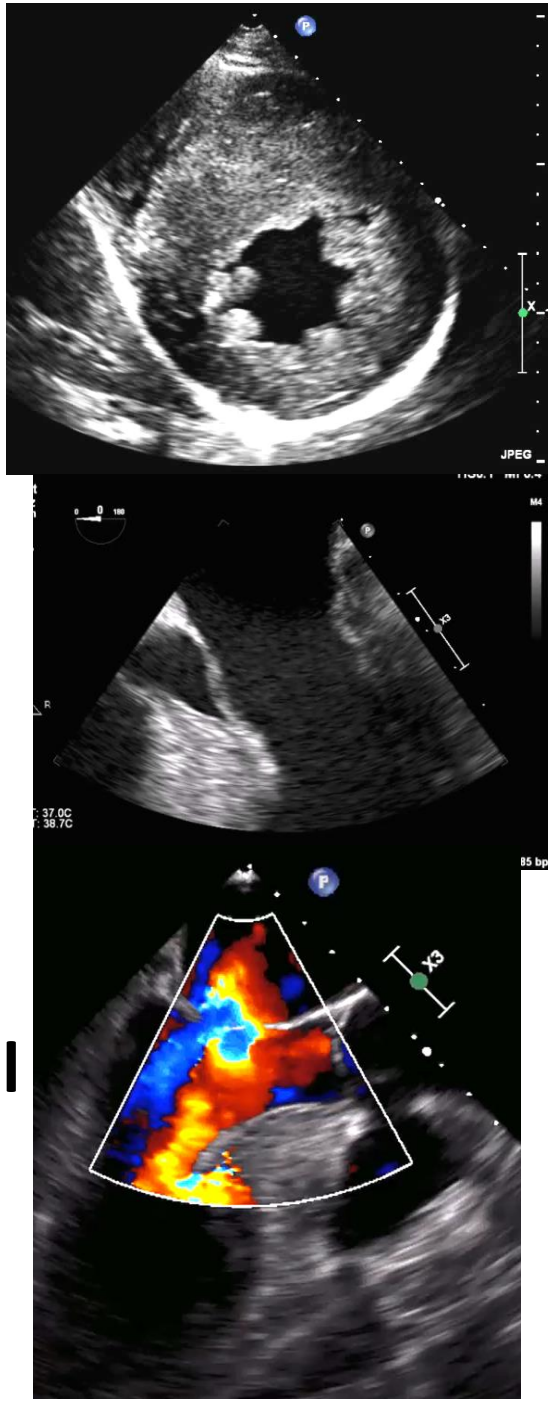
MRI



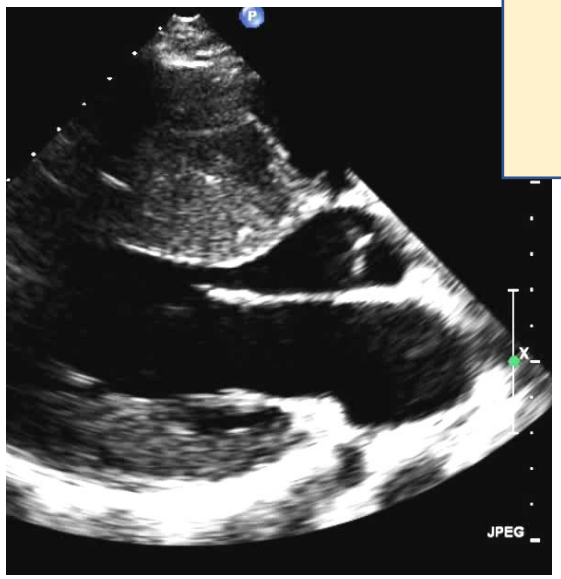

HCM
Non-obstructive
(LVOT resting gradient is < 30 mmHg)
Obstructive
(LVOT resting gradient is ≥ 30 mmHg)
Latent-obstructive
(LVOT gradient is < 30 mmHg at rest and ≥ 30 mmHg on exertion)
reverse curve or sigmoidal
apical
LVOTO
MVO
LVAA
rest
provocation
progression

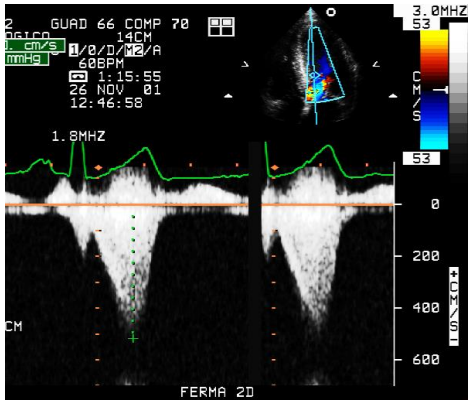

Sigmoidal HCM 40 - 50%
Reverse curve HCM 30 - 40%
Apical HCM ~ 10%
Neutral HCM ~ 10%
~ 10% Myofilament Gene +
~ 80% Myofilament Gene +
~ 30% Myofilament Gene +
~ 40% Myofilament Gene +

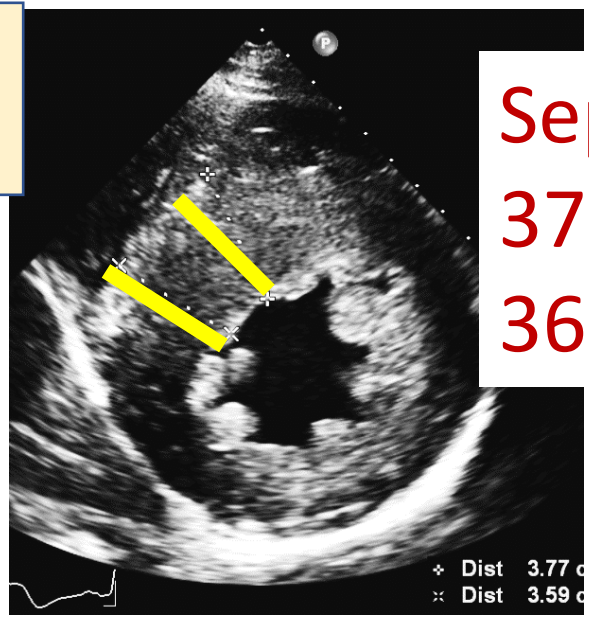
Maron et al


JPEG
X3

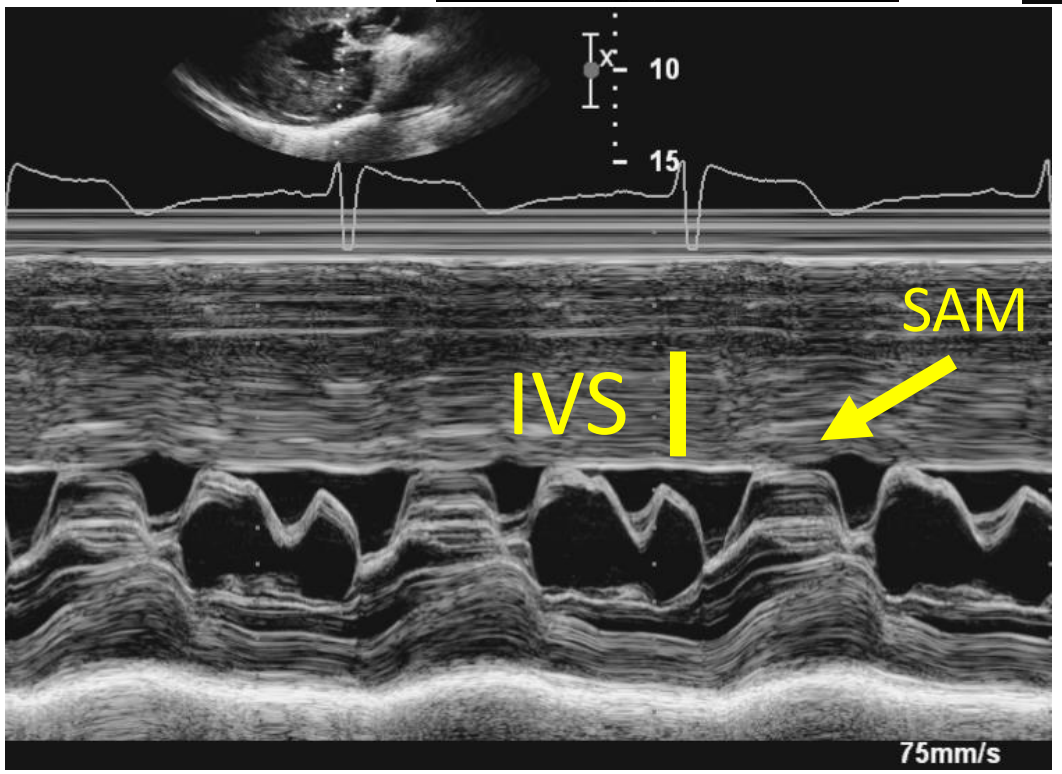
Hypertrophic CMP

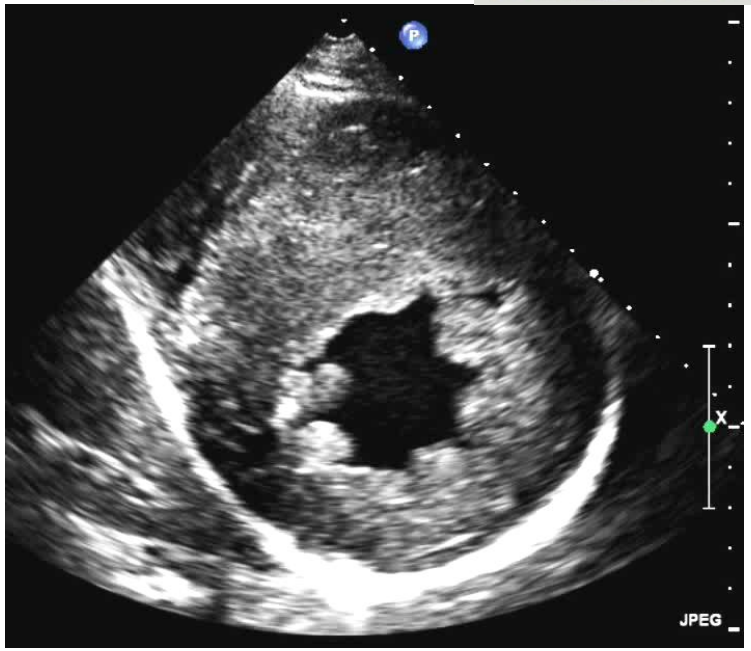






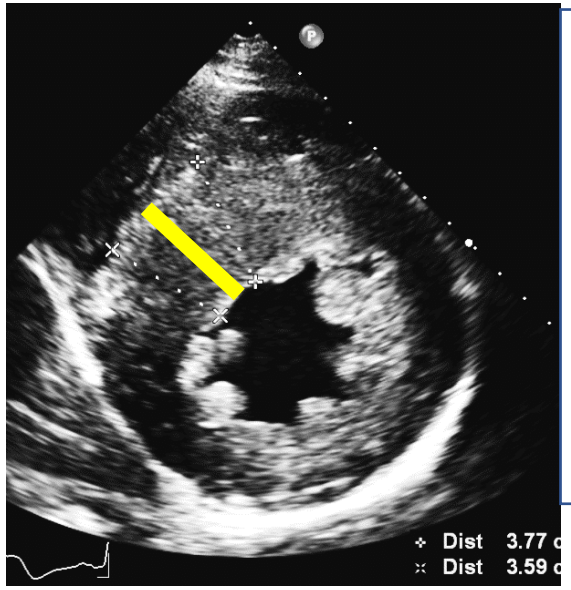
Septal thickness 37 and 36 mm



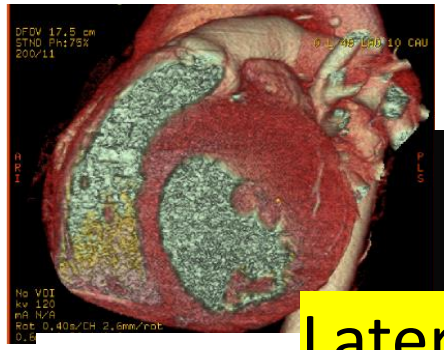


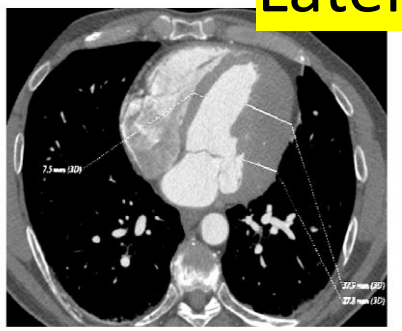
DIAGNOSIS by IMAGING

- **maximal end-diastolic wall thickness of ≥15 mm anywhere in the LV, in the absence of another cause of hypertrophy in adults. (ECHO or CMR**
- **ECHO: generally IV septum is involved**
- **More limited hypertrophy (13–14 mm) can be diagnostic when present in family members of a patient with HCM or in conjunction with a positive genetic test.**
- Inherited in an autosomal dominant pattern, **the distribution of HCM is equal by sex, although women are diagnosed less commonly than men.**

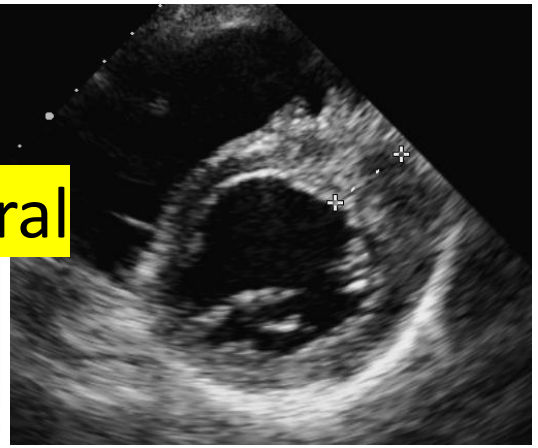


Less Common cases

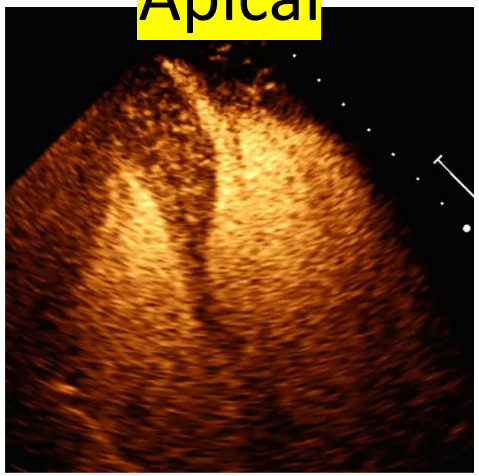




Lateral



Apical



Exclude Phenocopies

Athlete'sHeart

Long standing hypertension

Restrictive CMP

Left side obstruction

Amyloidosis

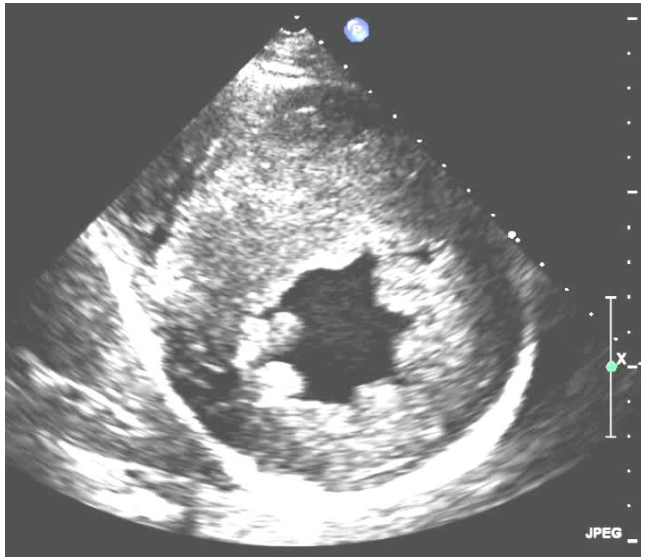
Hypertrophic CMP

FENOCOPIE:

mitochondrial myopathies, glycogen/ lysosomal storage diseases in children, and Fabry, amyloid, sarcoid, hemochromatosis, Danon cardiomyopathy

Amyloidosis

Hypertrophic CMP



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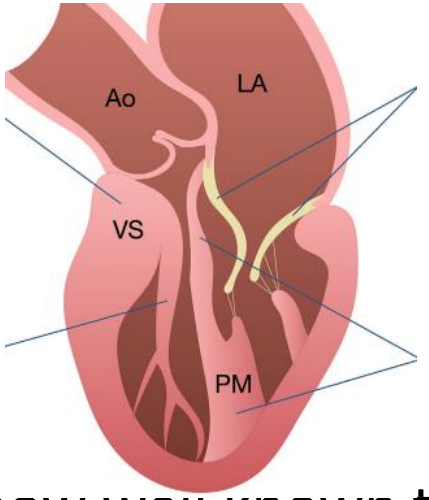
https://doi.org/10.1007/s12574-020-00467-9

REVIEW ARTICLE

The role of echocardiography for diagnosis and prognostic stratification in hypertrophic cardiomyopathy

Leonard Mandeş[1,2] · Monica Roşca[1,2] · Daniela Ciupercă[1] · Bogdan A. Popescu[1,2]

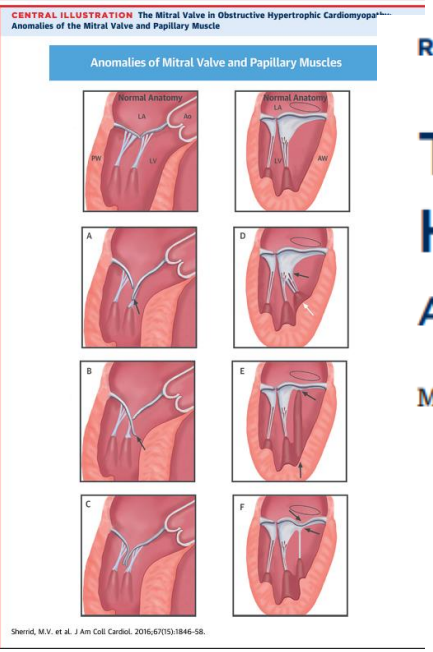
MV apparatus in Hypertrophic CMP



While initially thought to be a disease limited to the myocardium, it is now well known that up to **59% of pts diagnosed with HCM have at least one abnormality of the MV apparatus** (MVA) as a **direct effect of genetic mutations .**

More commonly, **leaflet elongation and excessive leaflet tissue are present in about 50% of pts**, while other anomalies **like chordal elongation, prolapse and direct insertion of the papillary muscle into the anterior leaflet are present in about 25% of cases** .

Other anomalies: **myocardial crypts**, **myocardial bridging, and right ventricular (RV) hypertrophy.**



REVIEW TOPIC OF THE WEEK

JACC 2016

The Mitral Valve in Obstructive Hypertrophic Cardiomyopathy

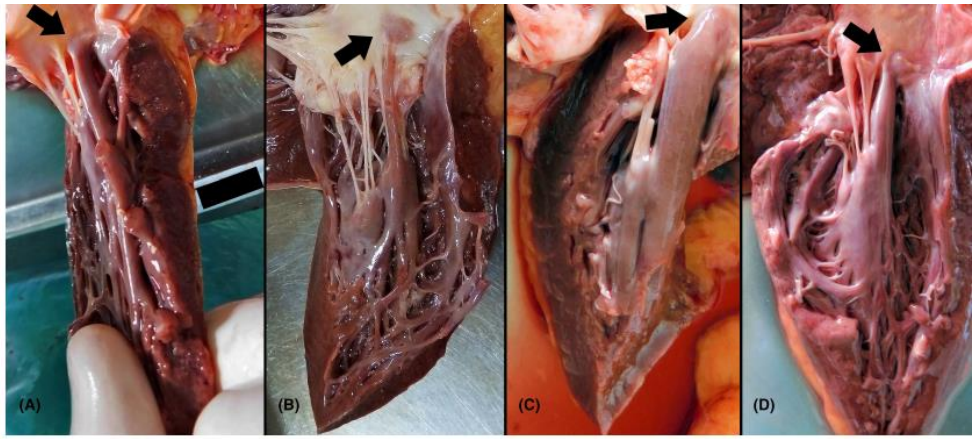
A Test in Context

Mark V. Sherrid, MD,[a] Sandhya Balaram, MD,[b] Bette Kim, MD,[c] Leon Axel, MD, PHD,[d] Daniel G. Swistel, MD[e]

MV anomalies

a wide array of malformations of the papillary muscles and chordae, that can be detected by ECHO and by CMR

Lekovic Forensic Science 2022



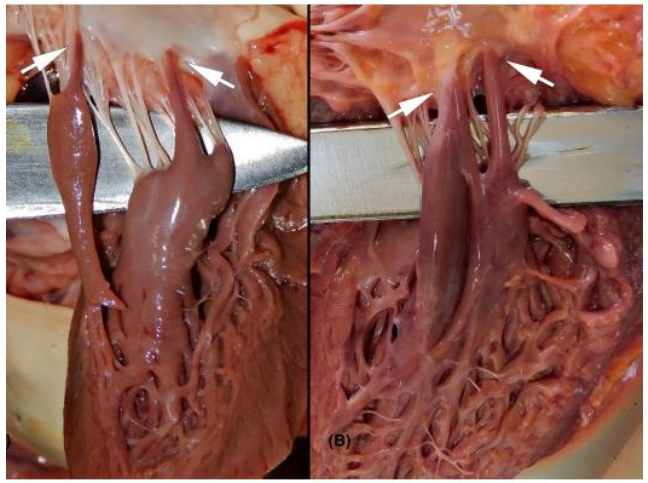
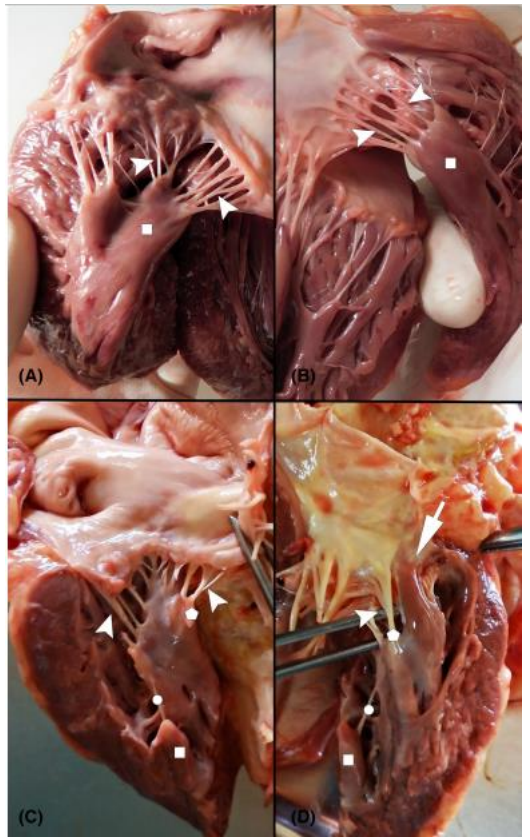
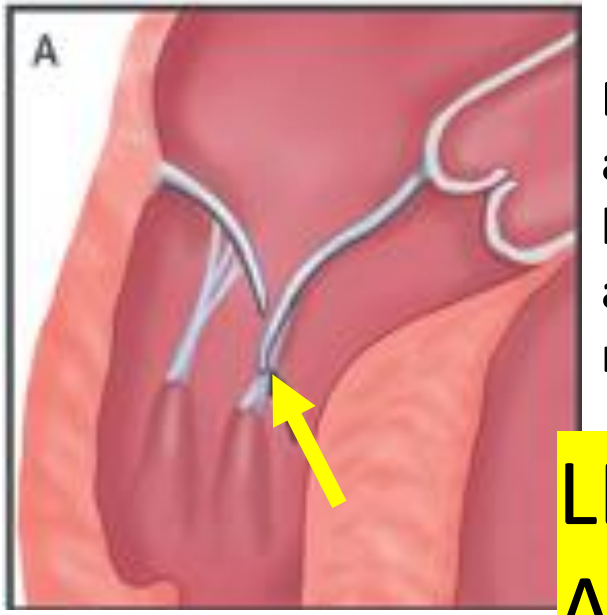


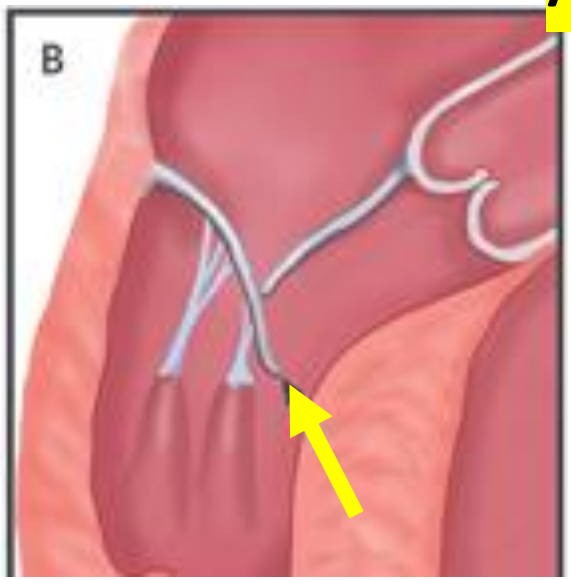
FIGURE 1 Anatomy variations of anomalous anterior papillary muscles. They varied in size and number of bellies, ranging from (A) discrete ones (case 16) to those with (B) moderate adjacent bellies (case 2) to a prominent single one (C—Case 18 and D—Case 5). Note the difference in the site of origin from the free ventricle wall and the place of insertion into the leaflet (A on the free edge vs. B on the base of a leaflet). [Color figure can be viewed at wileyonlinelibrary.com]



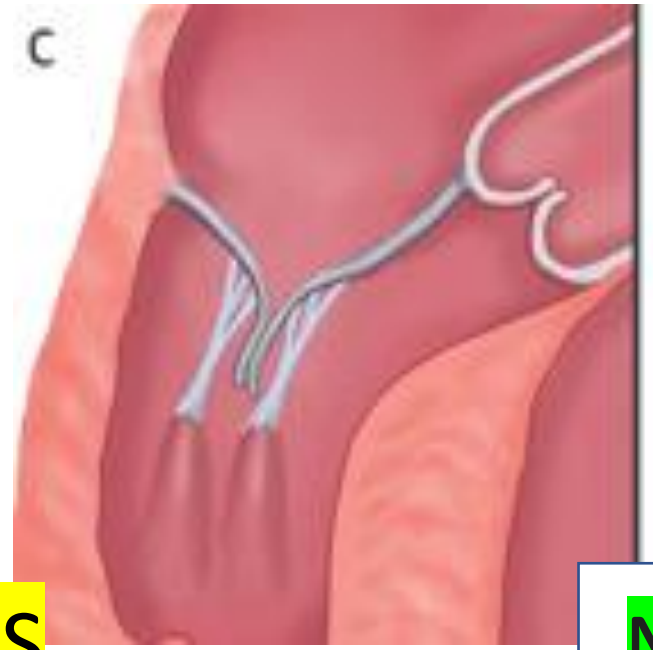


Elongation of anterior mitral leaflet;
arrow points to residual leaflet

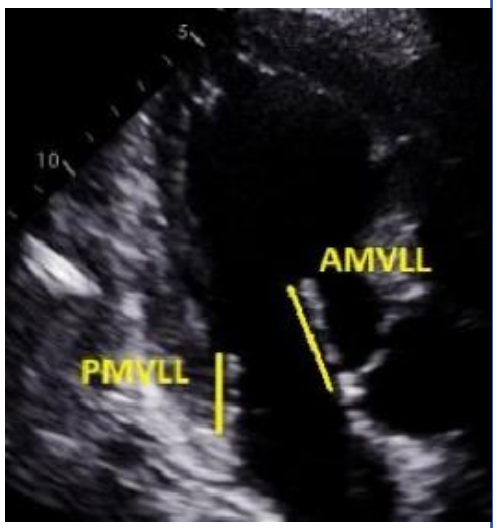
LEAFLET ANOMALIES



Elongation of posterior mitral leaflet;
arrow points to residual leaflet

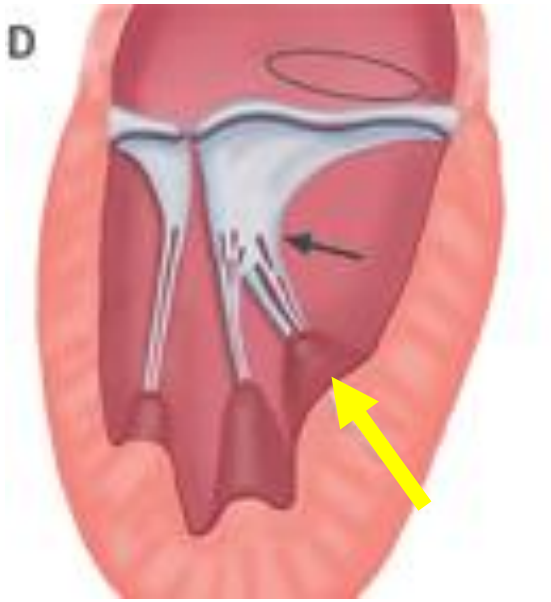


Elongation of both mitral leaflets; the anterior leaflet is most often the longer of the 2.

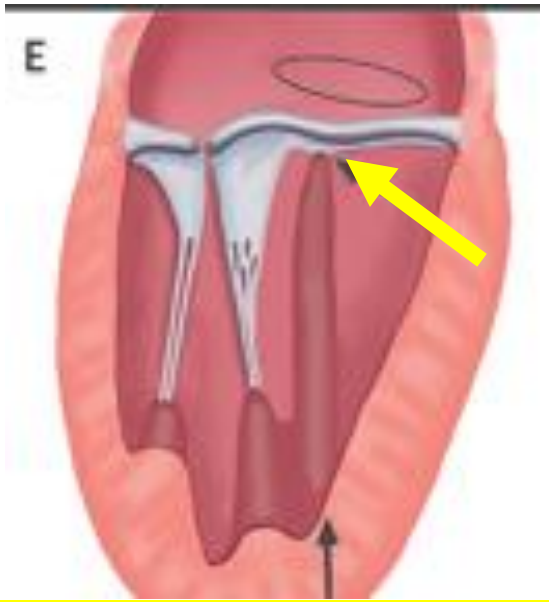


Normal leaflet length averages are 22–24 mm for the anterior and 12–13 mm for the posterior leaflet;

In HCM, the anterior mitral leaflet (AML) averages 34 mm compared with healthy individuals with an average of 24 mm.

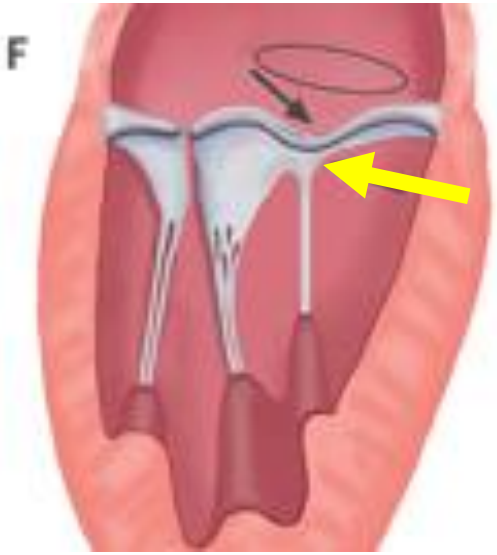


Additional thickened, anteriorly-displaced anterolateral papillary muscle-bifid papillary muscle (white arrow).



Anomalous anterolateral papillary muscle head (lower arrow) inserts directly into the middle of the anterior mitral leaflet (top arrow). This may contact the septum (not shown) and obstruct there, or may tent anterior leaflet into the LVOT.

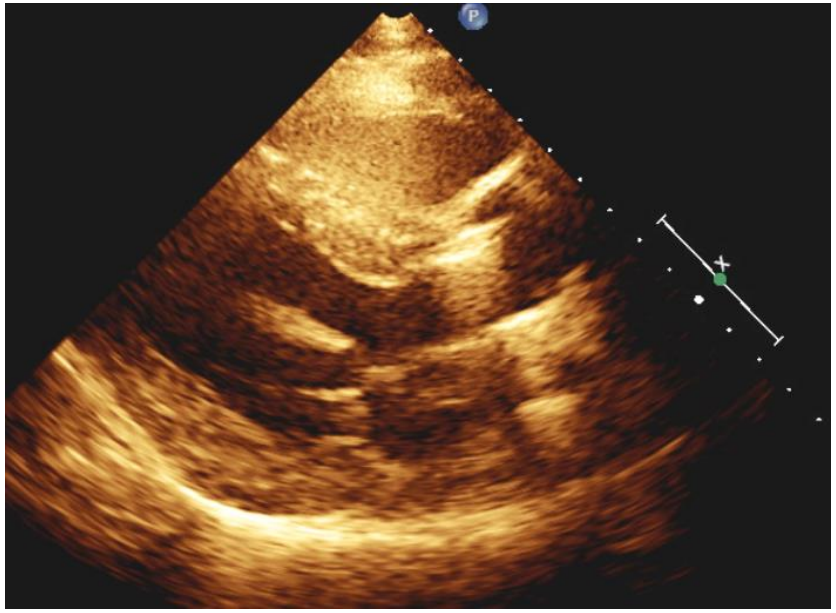
PAPILLARY MUSCLEs (and or CHORDAE) ANOMALIES

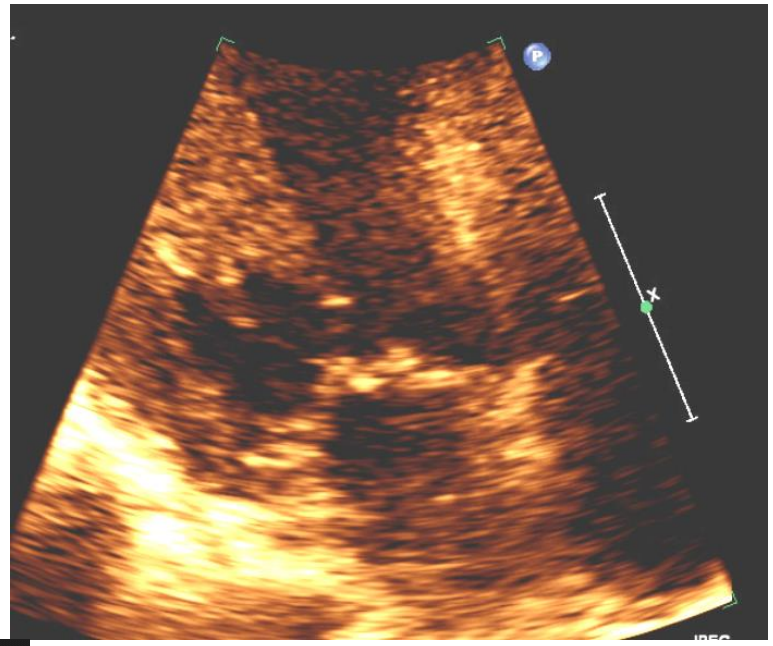


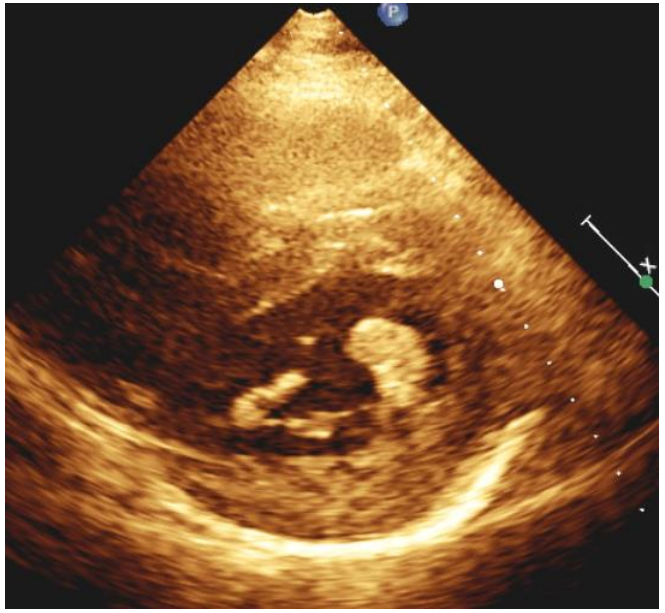
Abnormal chord, under tension, inserts into the midportion of the anterior leaflet and tents it into the LVOT (arrows), predisposing to systolic anterior motion

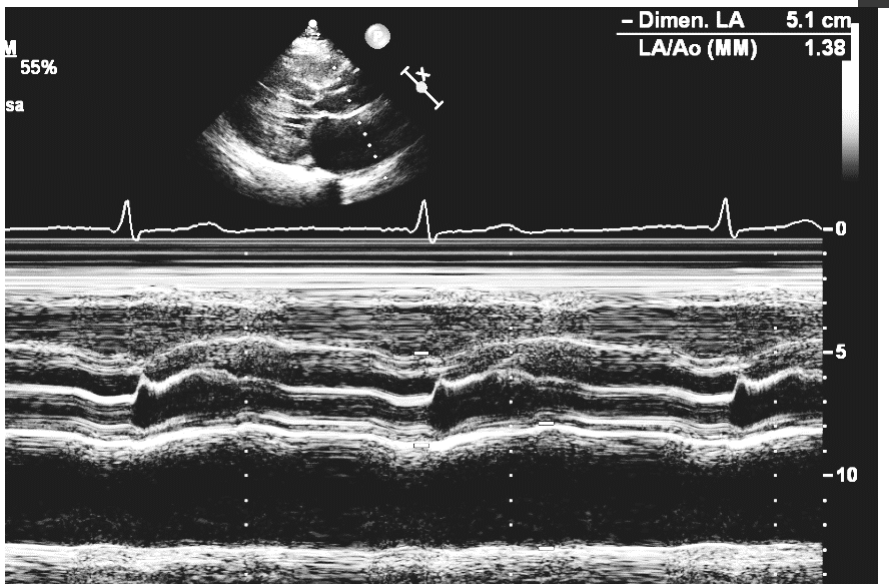
Cases with LVOT Obtruction And MV apparatus anomalies

TTE 2017

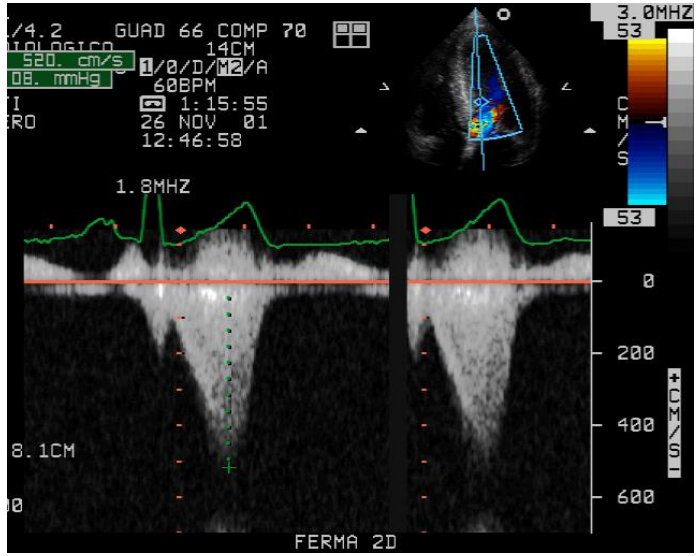




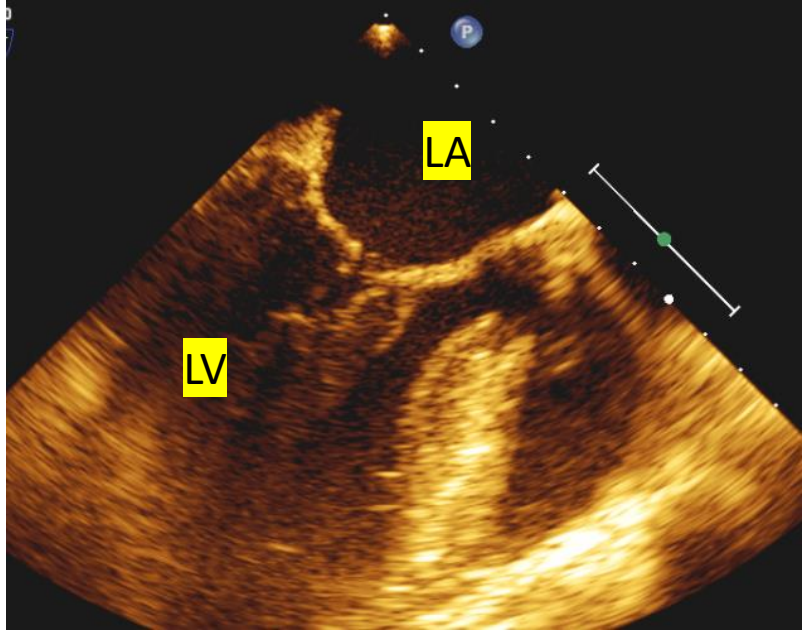




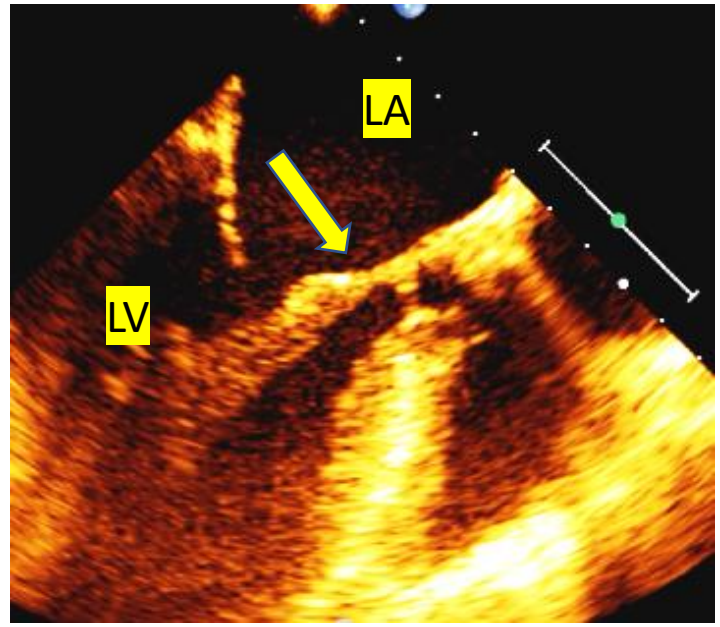
anteriorly-displaced anterolateral papillary

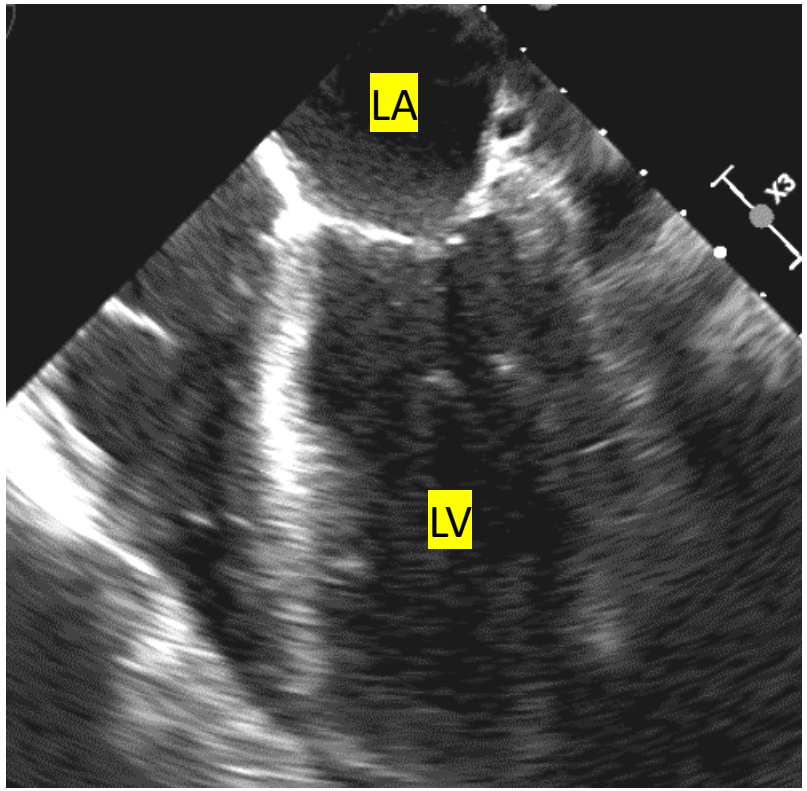


TEE

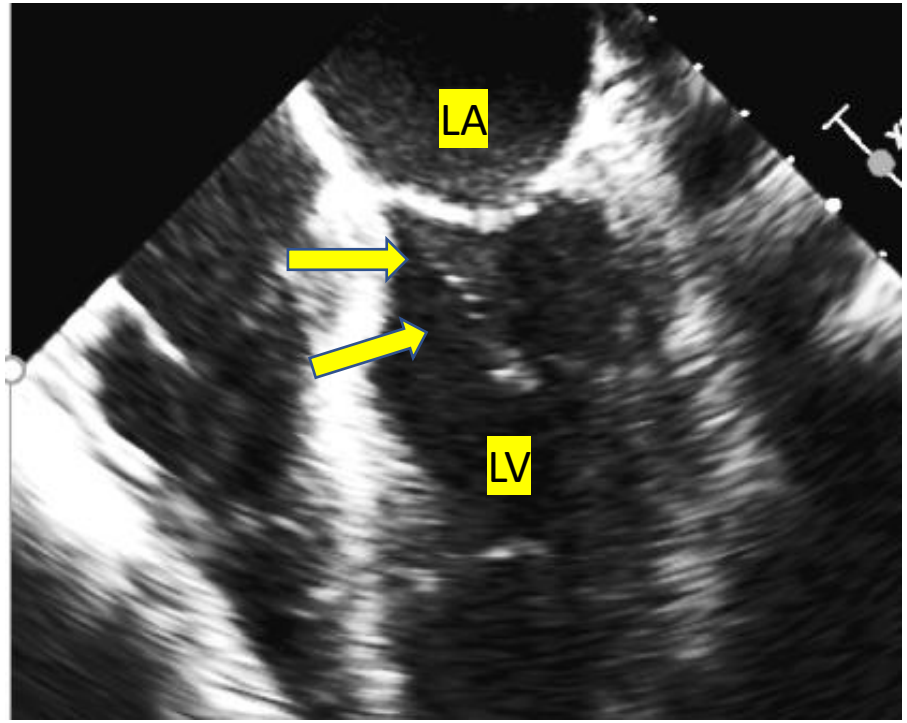


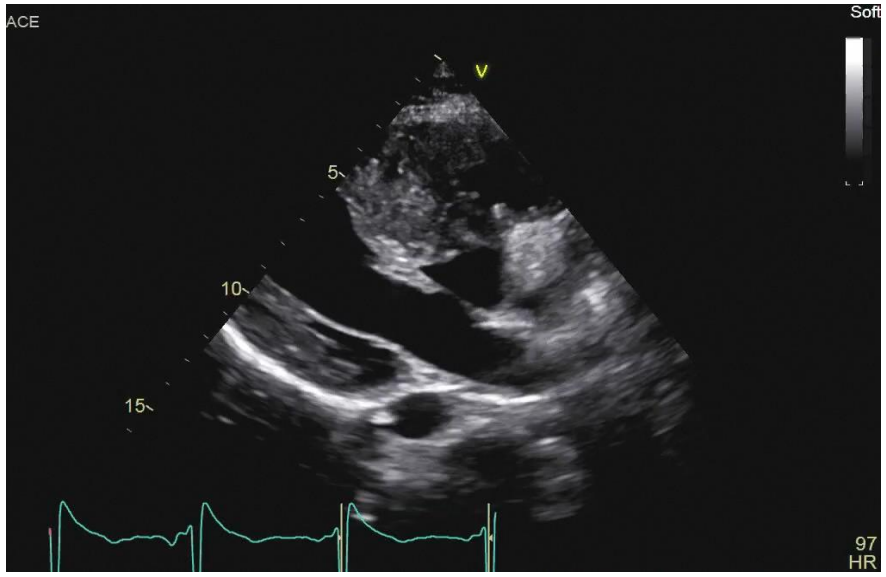
Anomalous anterolateral papillary muscle head inserts directly into the middle of the anterior mitral leaflet

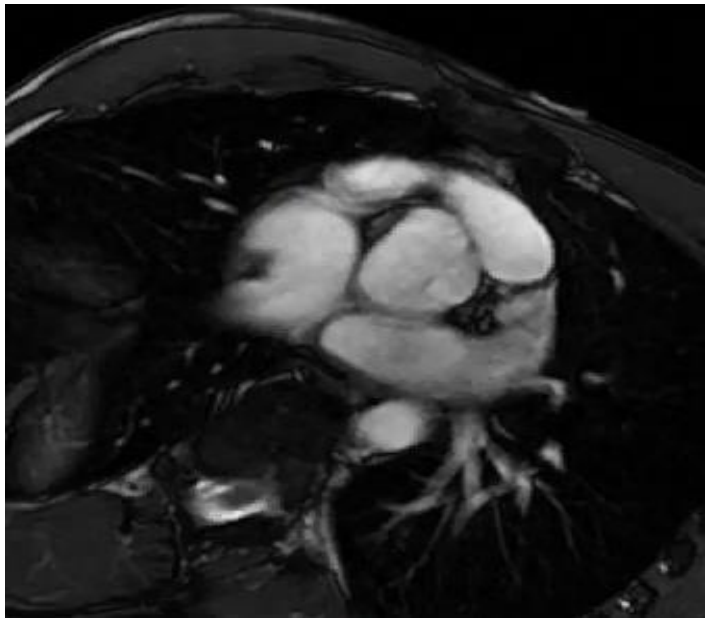


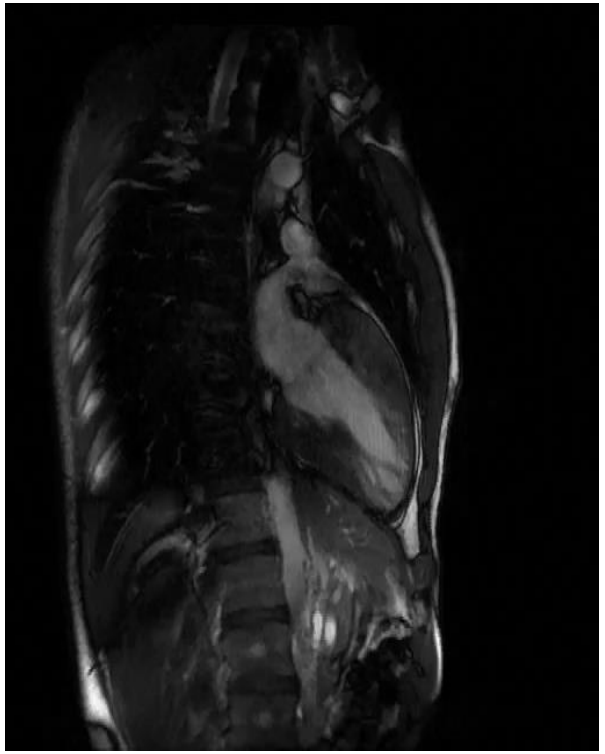


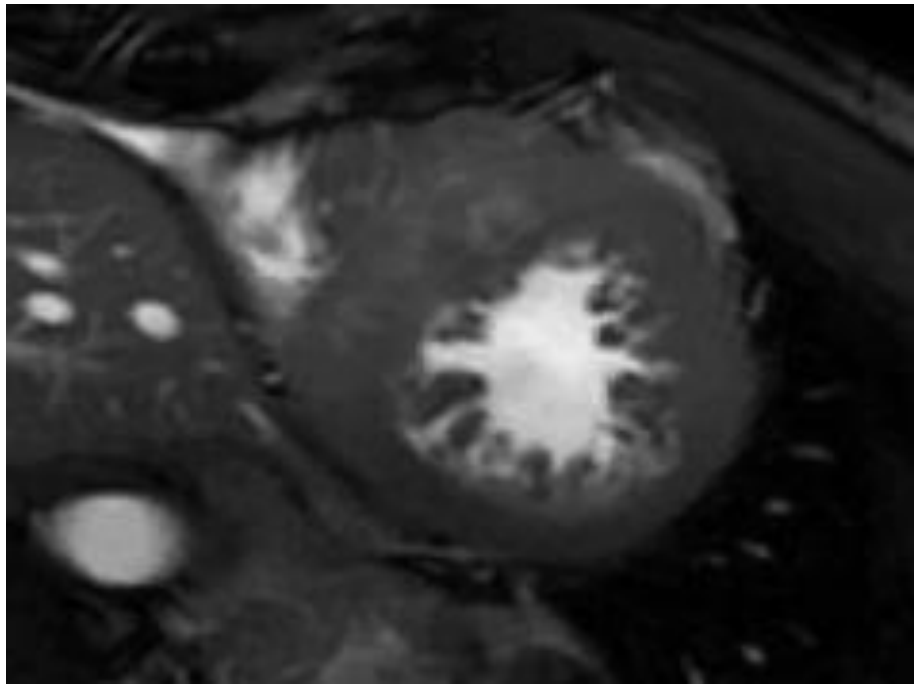
Abnormal chord, under tension, inserts into the midportion of the anterior leaflet and tents it into the LVOT











A case without apparent MV lesions (a part elongated leaflets)

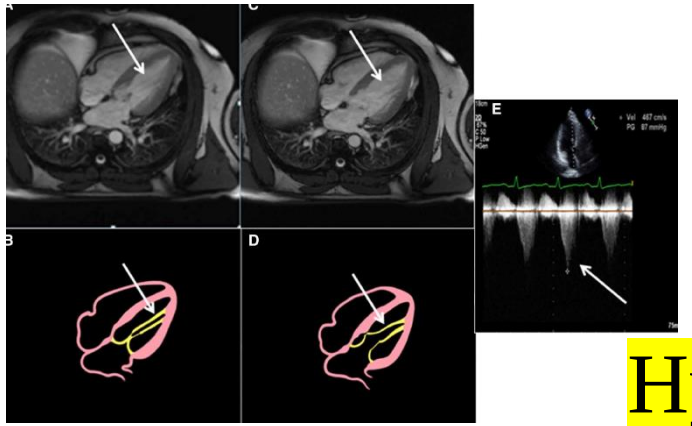
Note the presence of apparently normal papillary muscles (??) and a **very trabeculated LV**

Anterior MV lenght, papilarry and chordal abnormalities may contribute to SAM EVEN in cases without Severe Septal Hypertrpphy

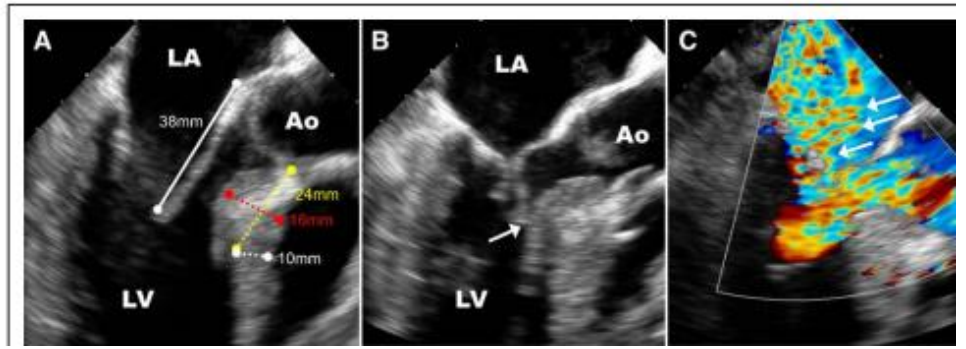
Left Ventricular Outflow Tract Obstruction in Hypertrophic Cardiomyopathy Patients Without Severe Septal Hypertrophy

Implications of Mitral Valve and Papillary Muscle Abnormalities Assessed Using Cardiac Magnetic Resonance and Echocardiography

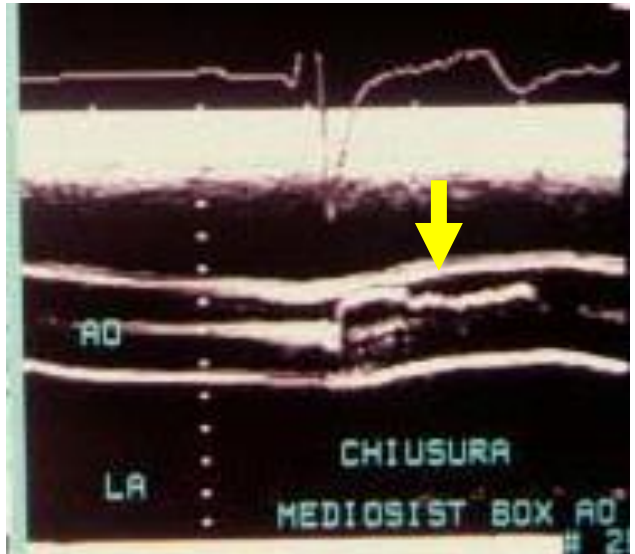
Parag Patel, MD; Ashwat Dhillon, MD; Zoran B. Popovic, MD, PhD; Nicholas G. Smedira, MD; Jessica Rizzo, RDCS; Maran Thamilarasan, MD; Deborah Agler, RDCS; Bruce W. Lytle, MD; Harry M. Lever, MD; Milind Y. Desai, MD

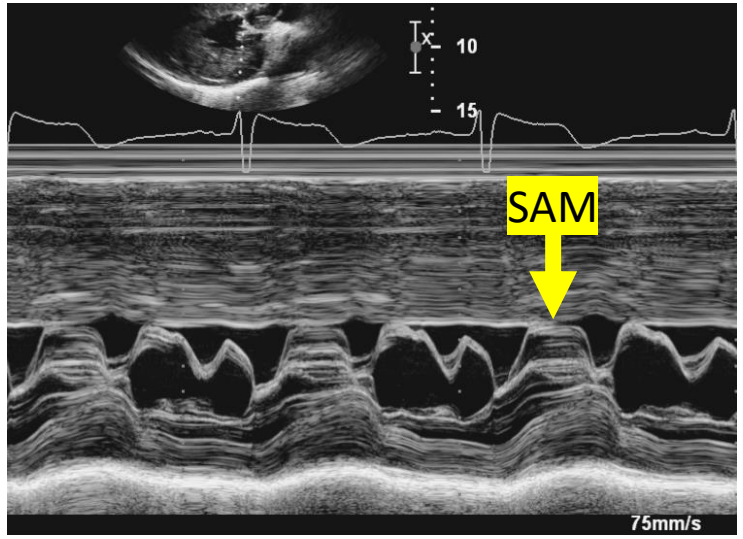


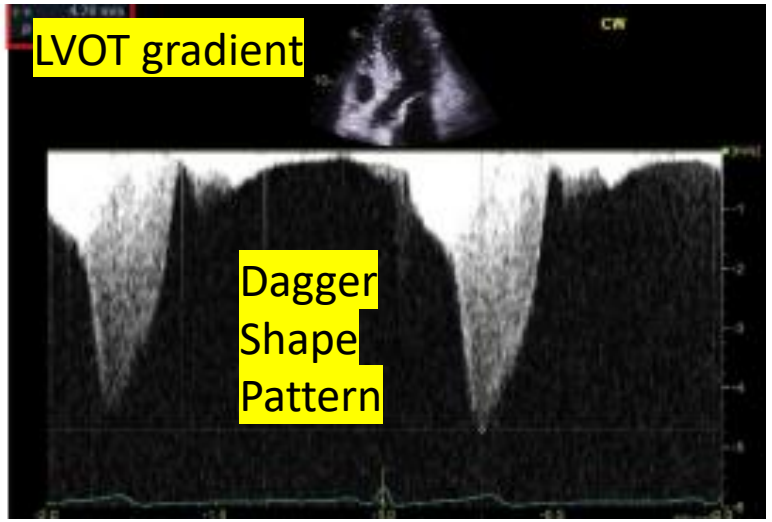
Maron et al Hypertrophic Cardiomyopathy

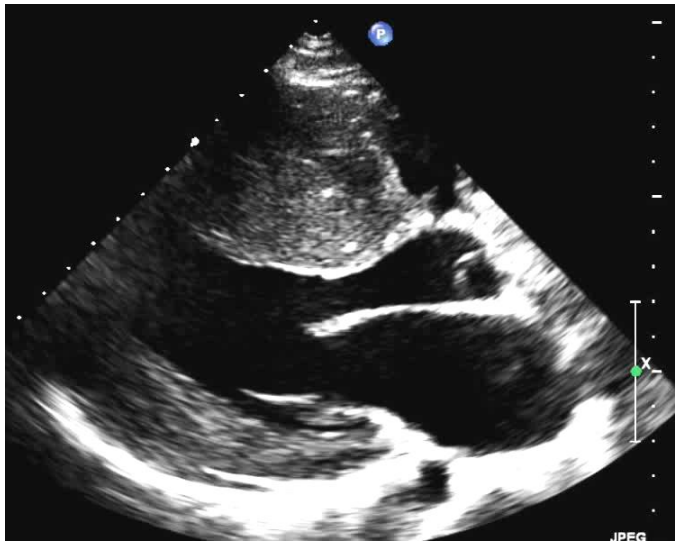


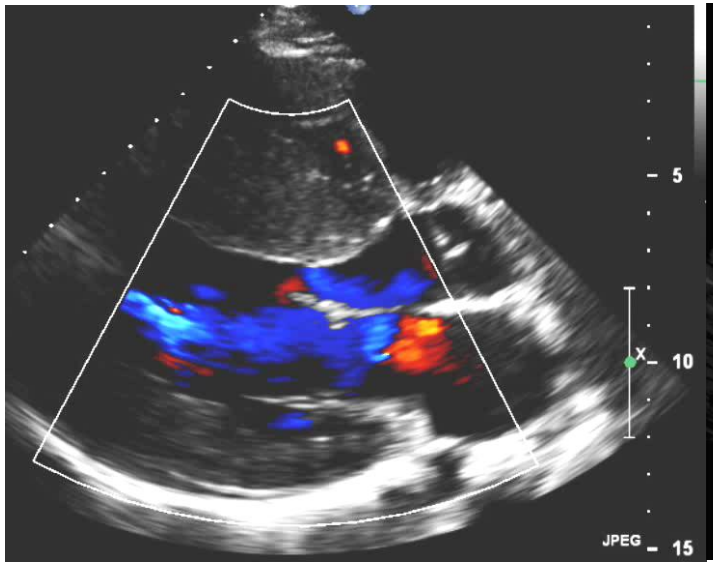
Hypertrophic Obstructive Cardiomyopathy : SIGNS

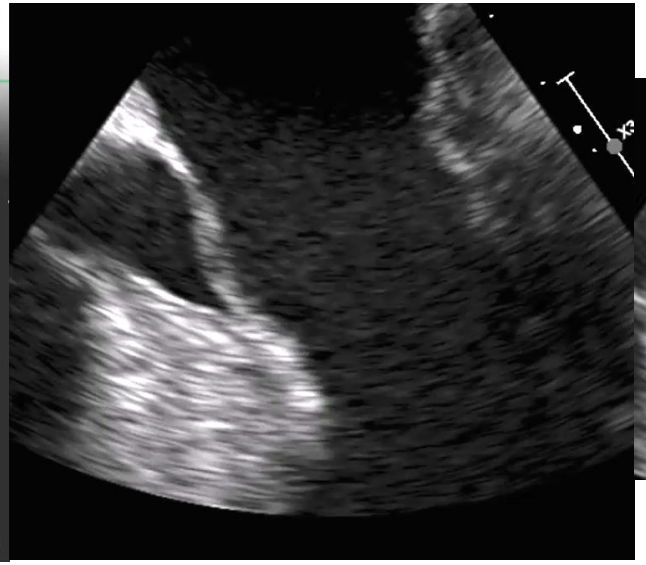


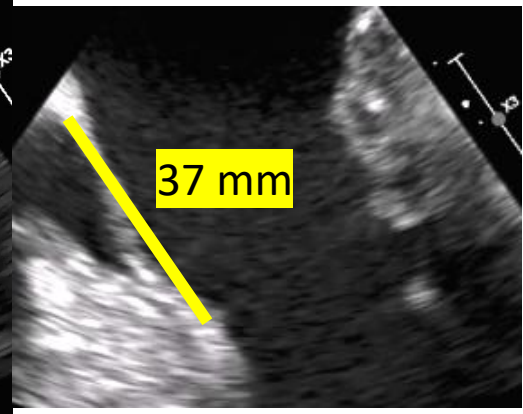




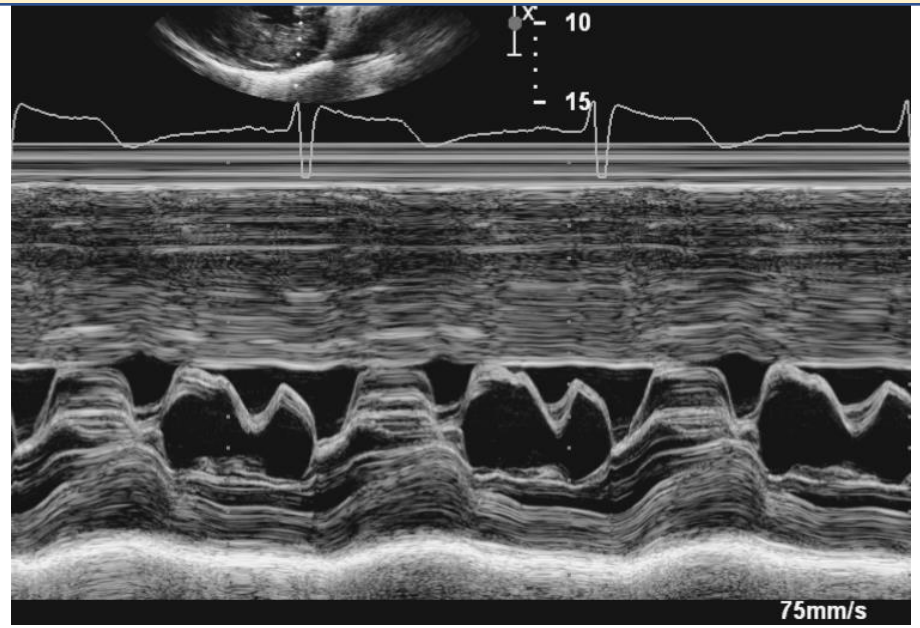


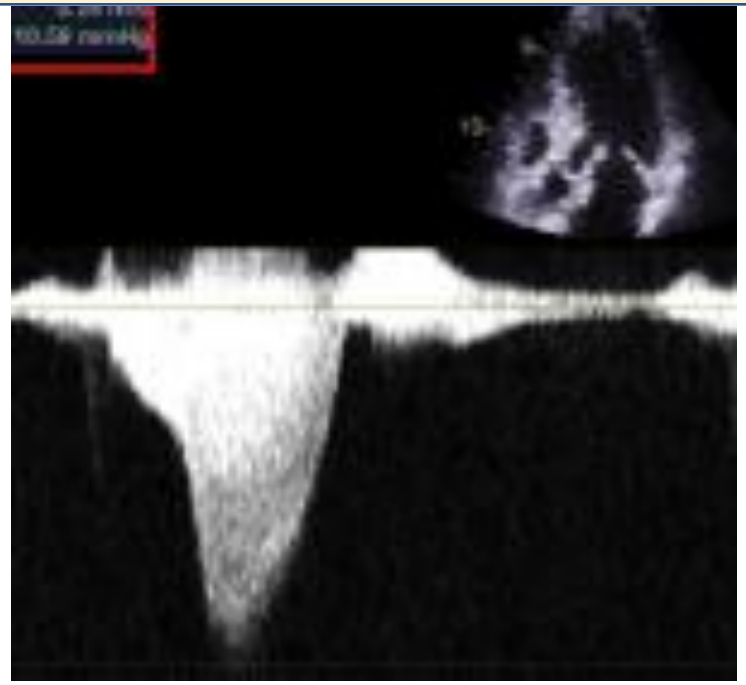




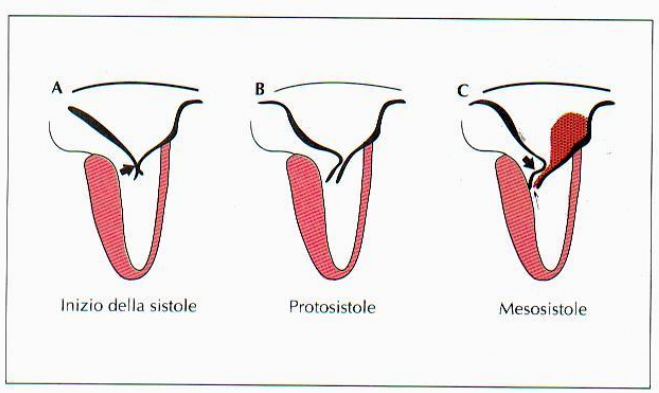


DURATION of SAM contact correlates with the severity of obstruction



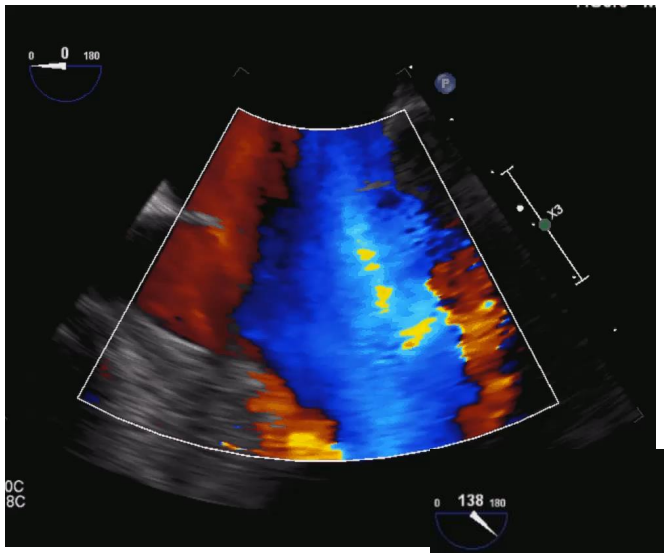


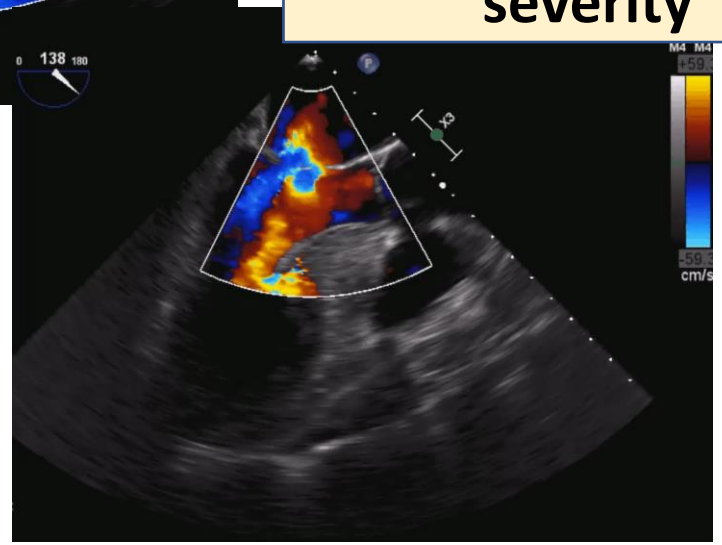
LVOT Gradient 100 mmHG



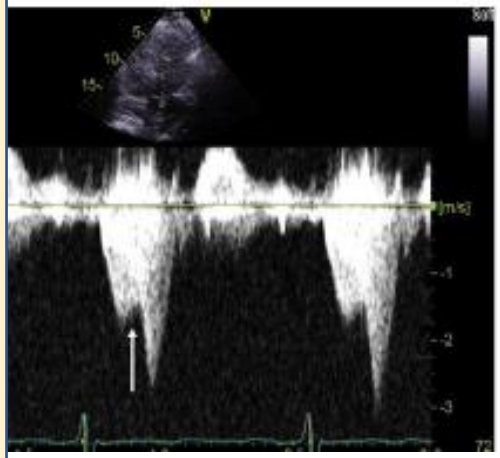
SAM

MR and obstruction are related to the systolic timing of SAM; the **site of the Doppler sample interrogation** is variable; rapid **changes in pre load and afterload influence gradient severity**

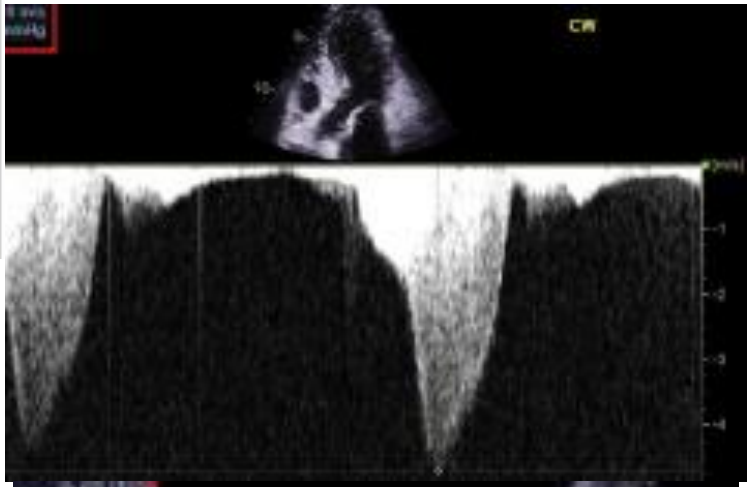




"Lobster claw" abnormality

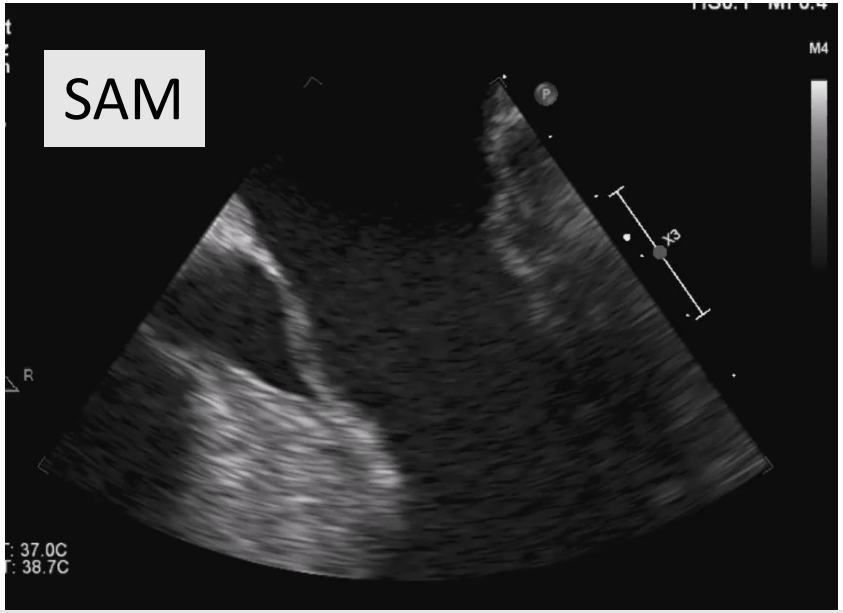


LV mid cavity

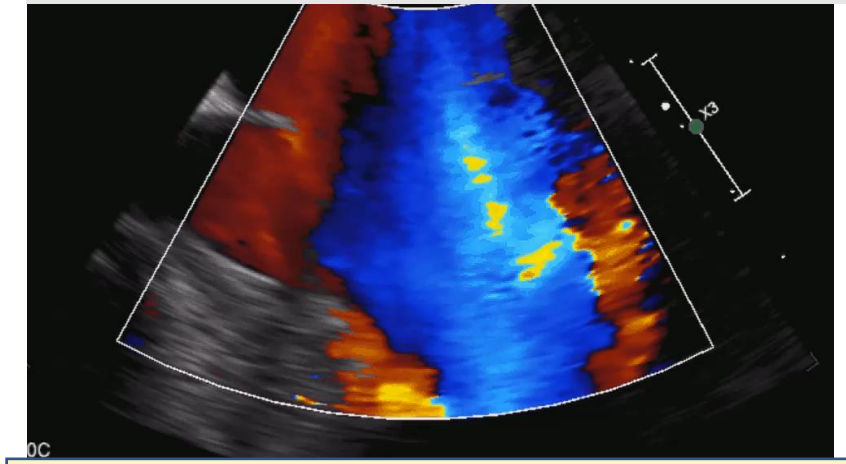


LVOT : 88 mmHG

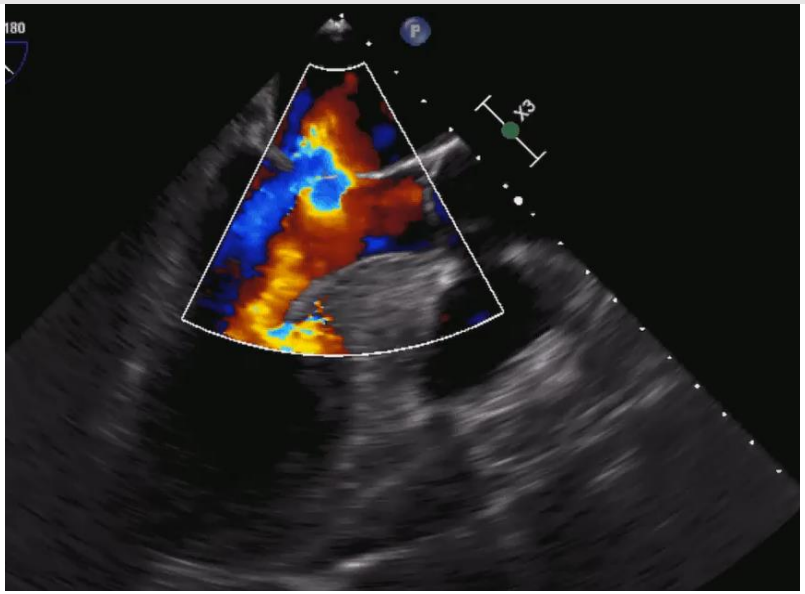
LVOT Valsalva : 110 mmHg



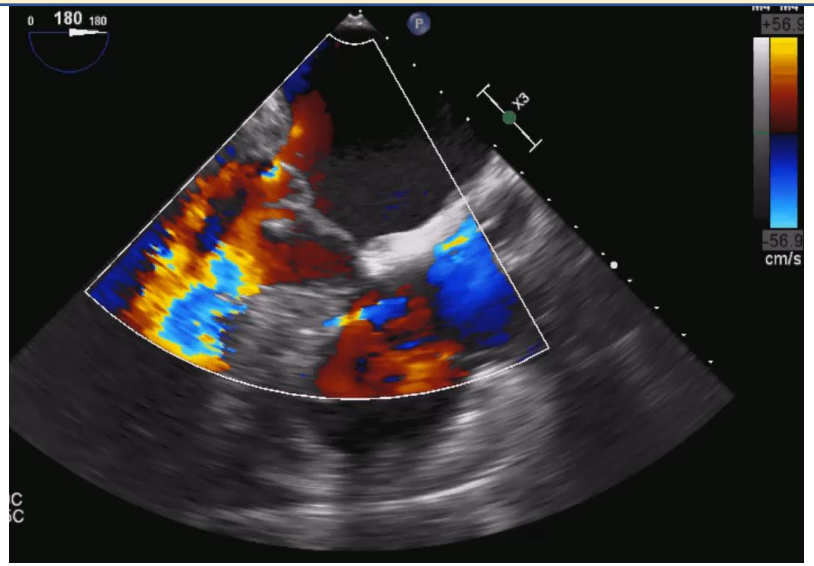
SAM, LVOT obstruction and MR jet (diverging toward post-lat LA)



SAM, LVOT obstruction and MR jet (diverging toward post-lat LA)



After myomectomy and MV repair: disappearance of SAM and MR



ESC European Society of Cardiology
European Heart Journal - Cardiovascular Imaging (2022) 00, 1–12
https://doi.org/10.1093/ehjci/jeac233
ORIGINAL PAPER
2022

Left ventricular anatomy in obstructive hypertrophic cardiomyopathy: beyond basal septal hypertrophy

Uxio Hermida [1], David Stojanovski[1], Betty Raman [2], Rina Ariga[2], Alistair A. Young[1], Valentina Carapella[1], Gerry Carr-White[3], Elena Lukaschuk [4], Stefan K. Piechnik[4], Christopher M. Kramer[5], Milind Y. Desai[6], William S. Weintraub[7], Stefan Neubauer [2,4], Hugh Watkins [4], and Pablo Lamata [1]* on behalf of the HCMR investigators

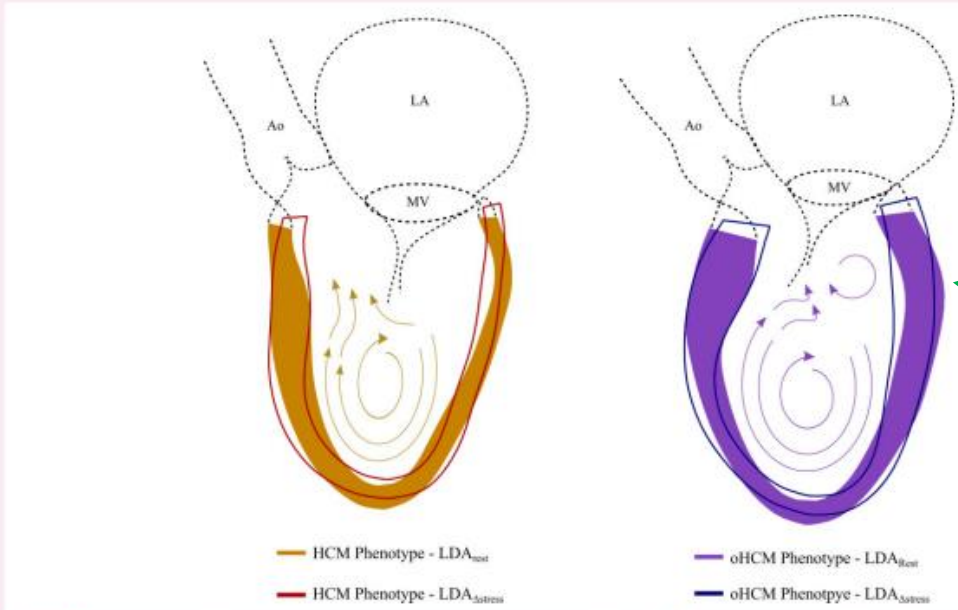


Figure 5 LV shape and LVOTO. Impact of LV shape on vortex formation during early systole, which changes the angle of attack of blood flow with respect to the mitral valve leaflets, increasing the systolic anterior motion of the mitral valve and consequently, LVOTO. Orange/left shape represents −3SD from the average shape along the LDA$_{rest}$ axis (extreme non-obstructive HCM phenotype at rest). Purple/right shape, +3SD (obstructive HCM phenotype at rest). Phenotypes resulting from the LDA$_{\Delta stress}$ are overlayed with lines. An estimation of the left atrium, mitral valve, aorta, and aortic

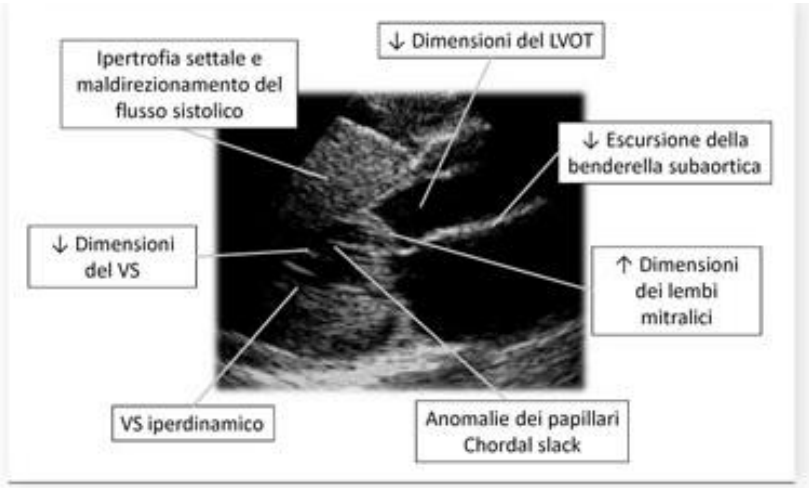
LV anatomy underpinning OHCM consists of basal septal hypertrophy, apical dilatation, LV lengthening, and LVOT inward remodelling.

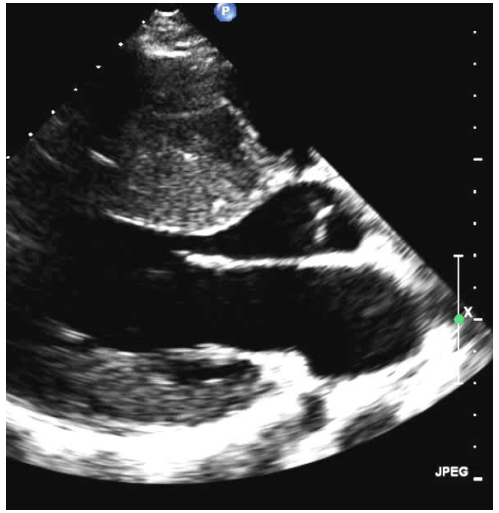
Genotype negative cases showed a tendency towards more obstructive phenotypes both at rest and stress.

Vortex in the protosystolic phase lead to a change of the angle between flow and leaflets facilitating SAM

SAM mulifactorial etiology
(structural and functional)

Olivotto et al



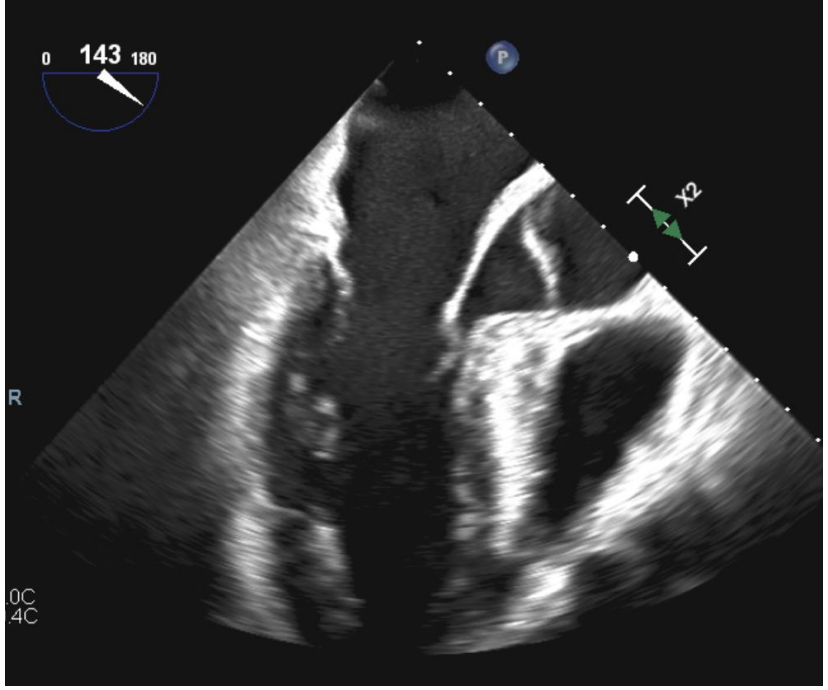


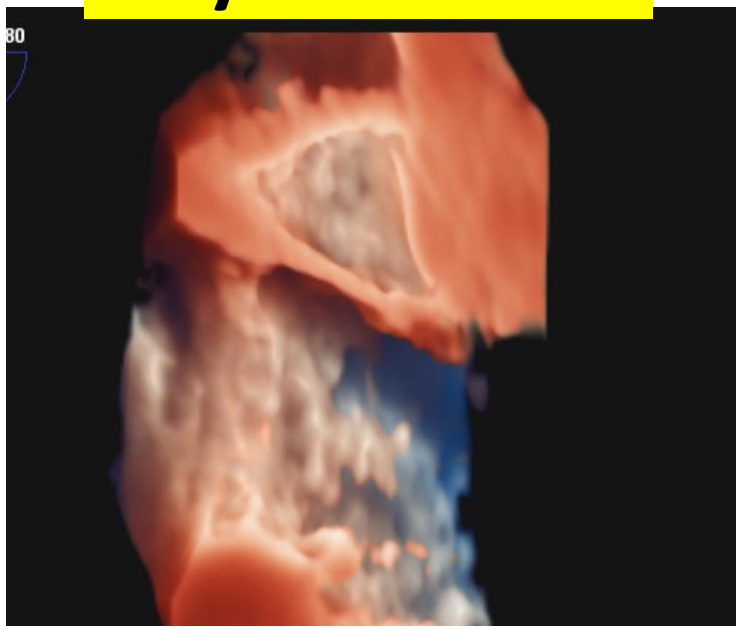
-
- Venturi Effect is a secondary determinant of SAM (and not primary as proposed in the past
- **Suggested Mechanisms of SAM**
- **Redondant Mitral leaflets inside a small and hypercontractile LV .**
- **Anomalies of the Papillary muscles that change the coaptation of the elongated MV leaflets**

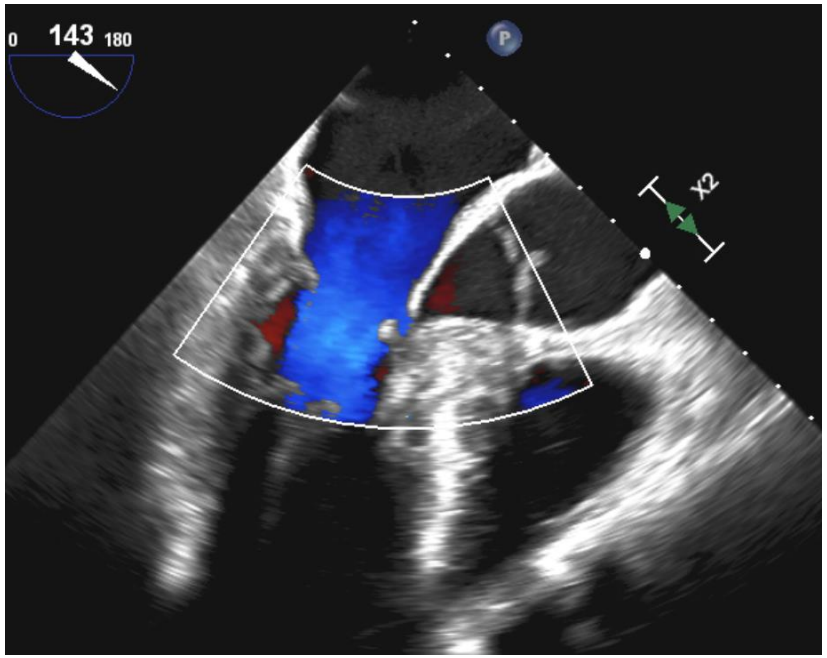
- **LVOT is reduced and it captures elongated MV leaflets and push leaflets towards the LVOT >>> causing obstruction**

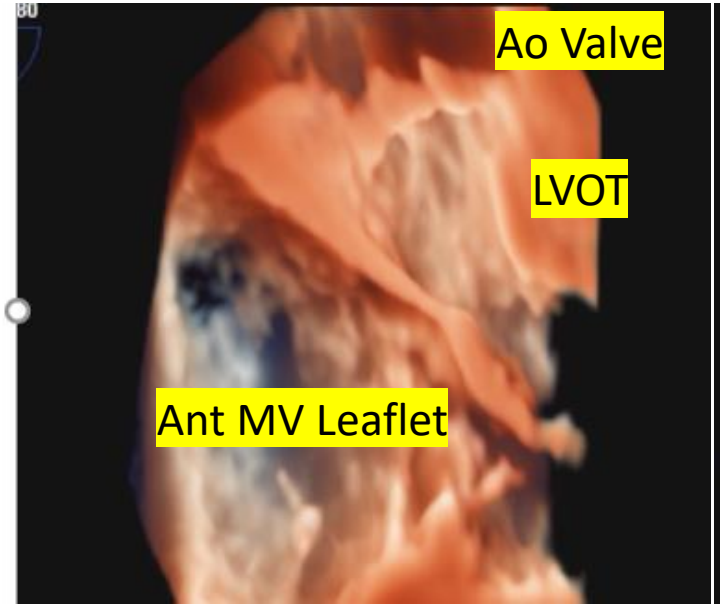
-

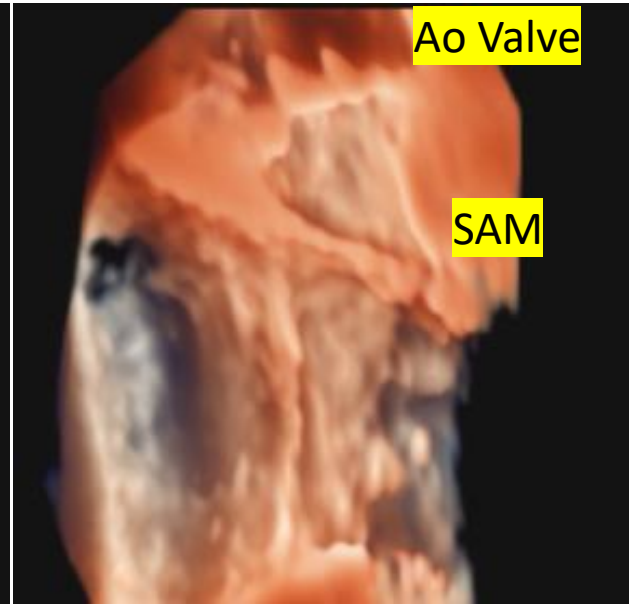
Very Small LVOT

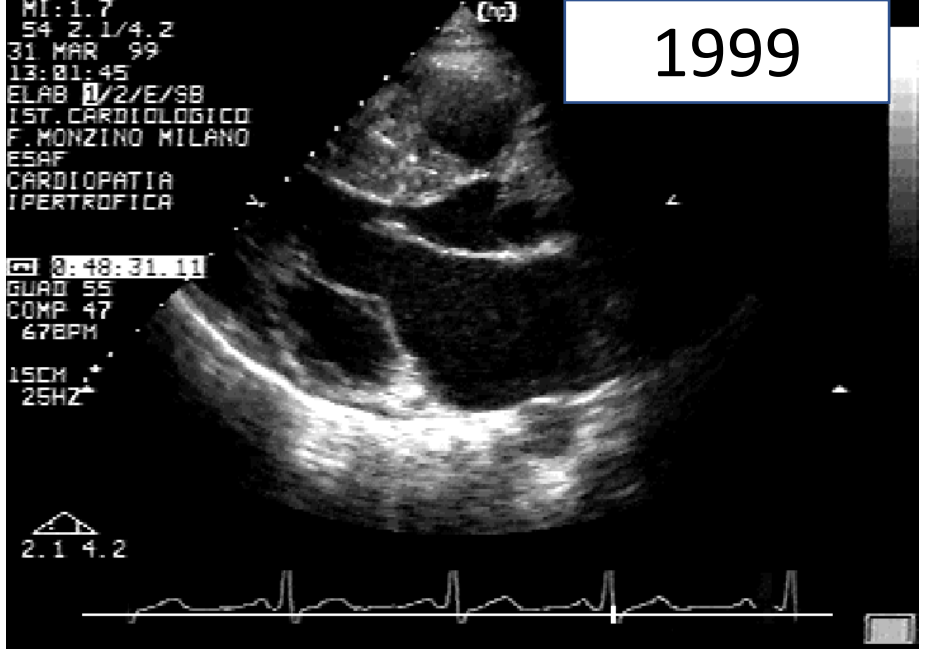


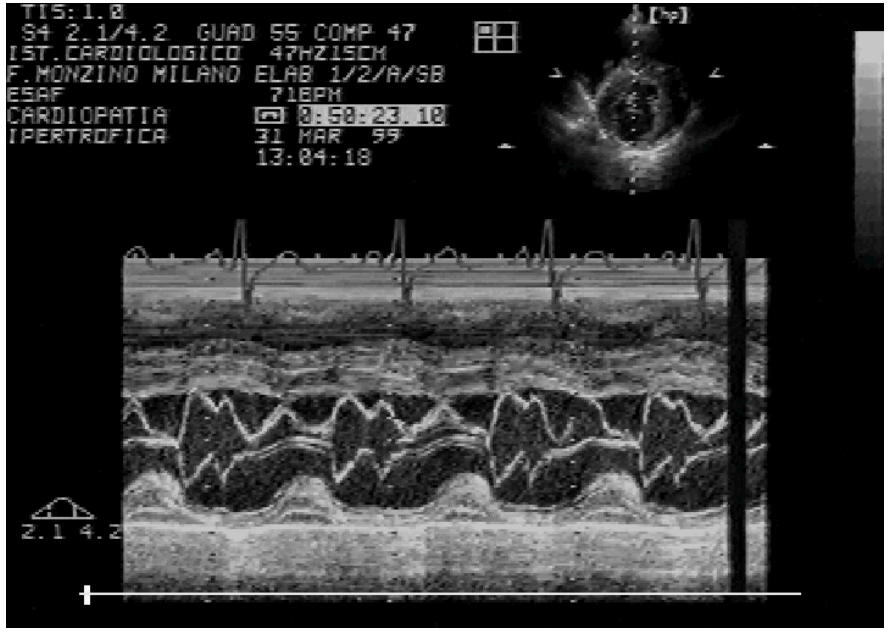


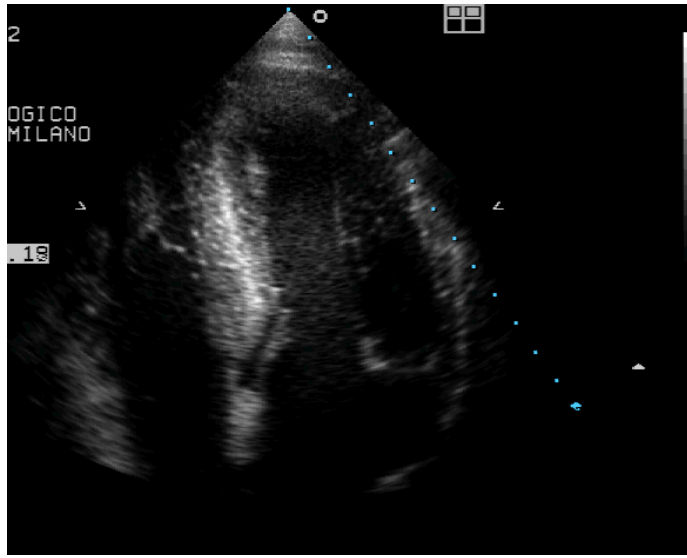




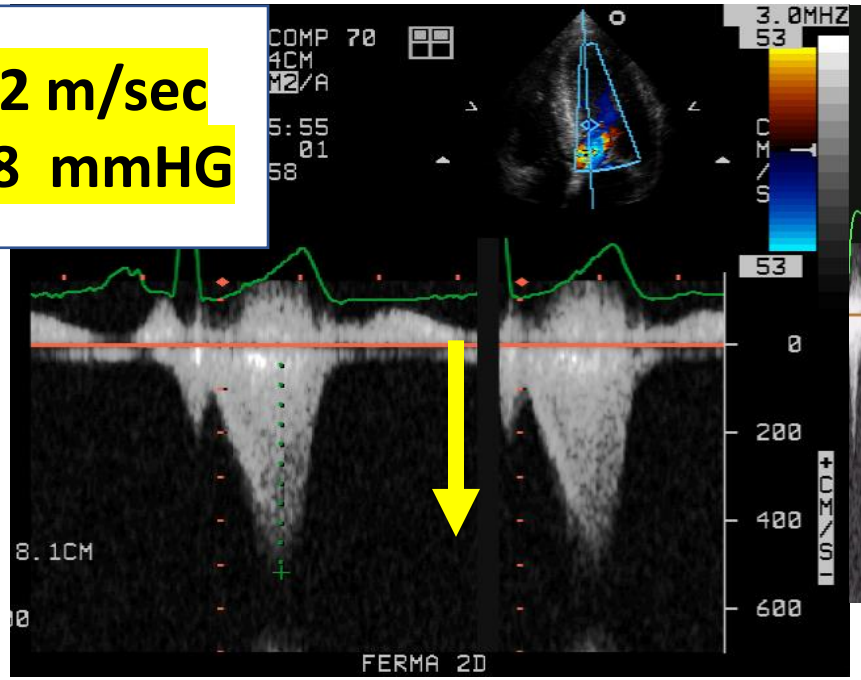




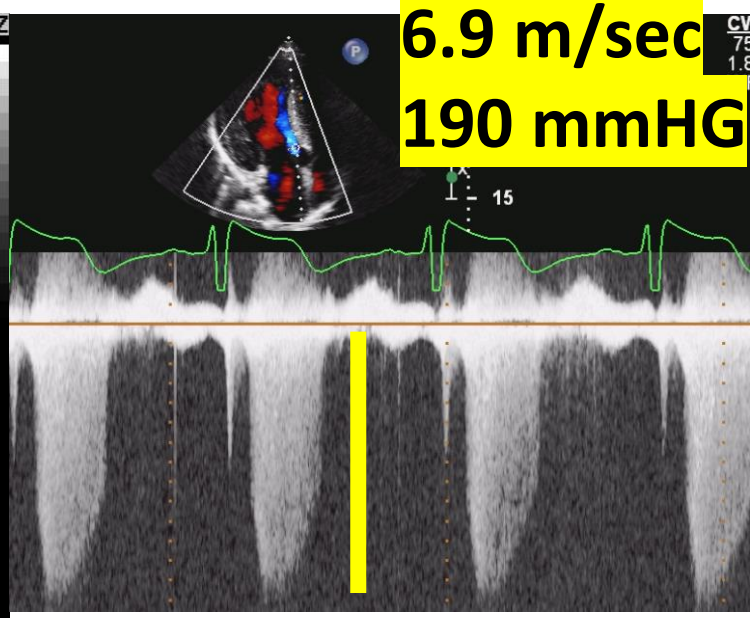




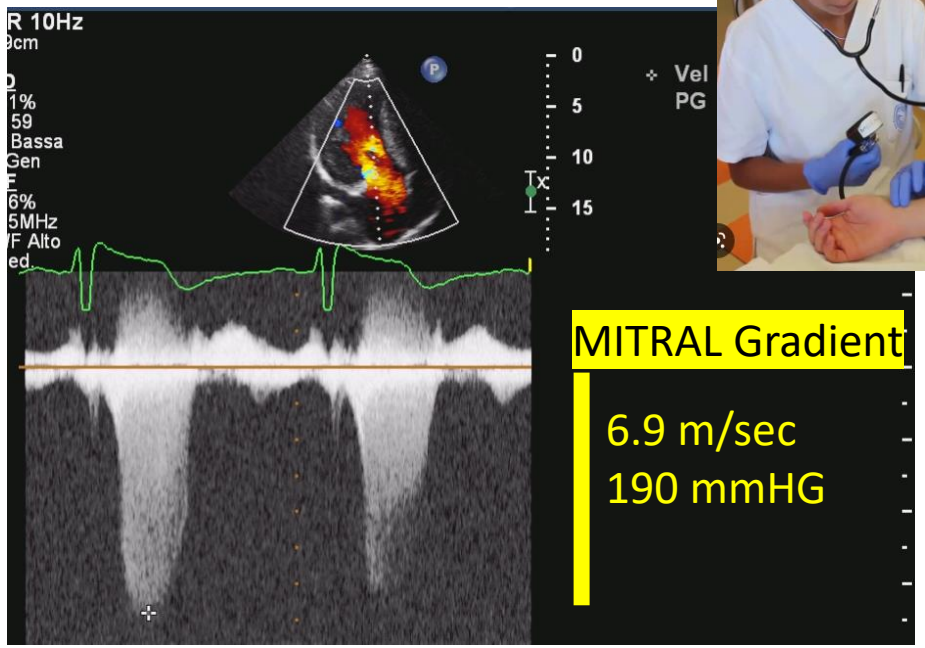
5.2 m/sec
108 mmHG

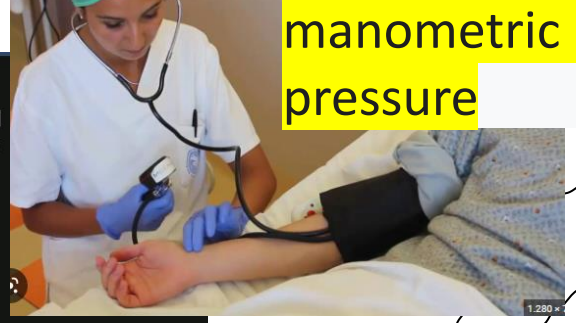


6.9 m/sec
190 mmHG



How to verify accuracy in gradient measurements ?

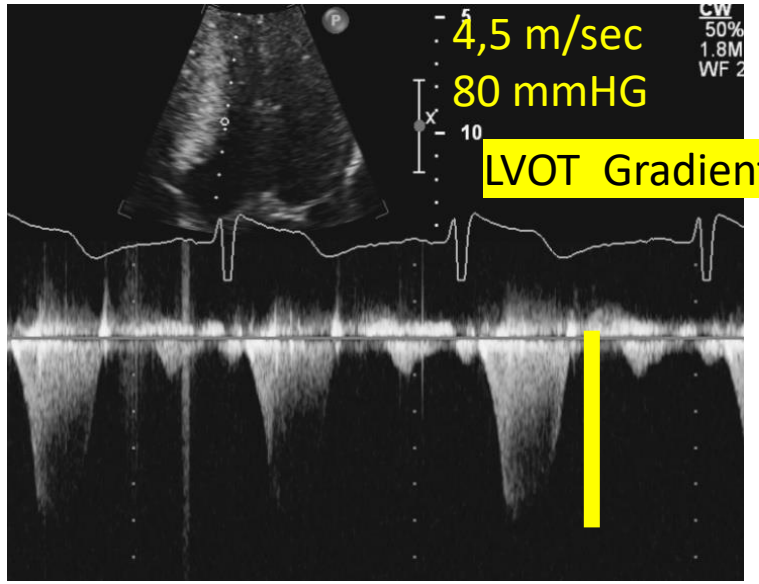


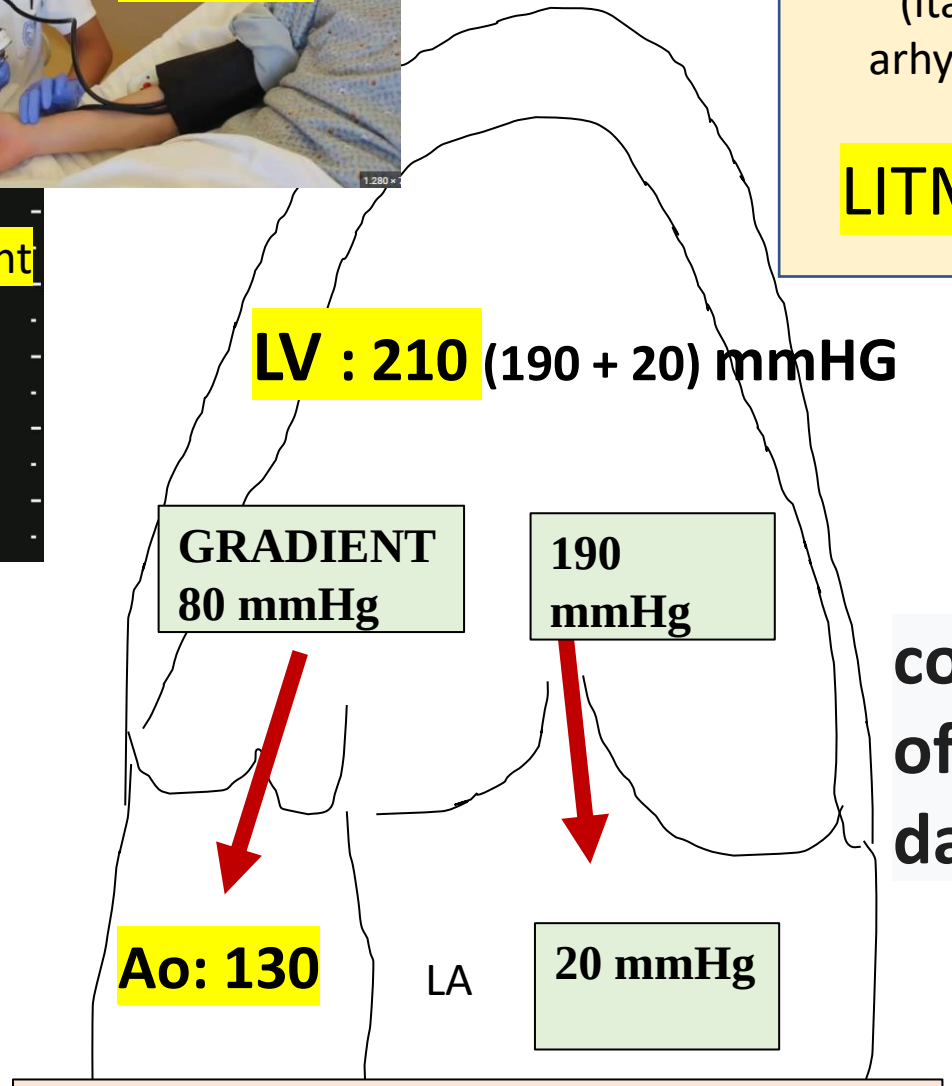


manometric blood pressure

«Prova del nove» (italian simple arhythmetic test)

LITMUS TEST

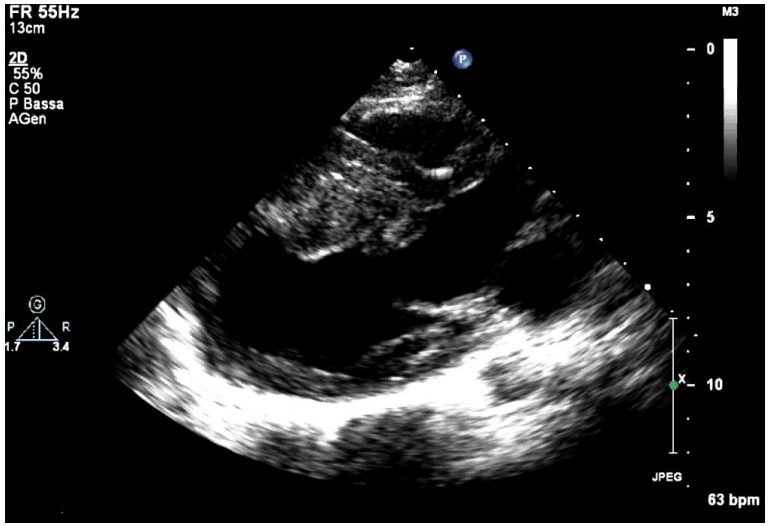


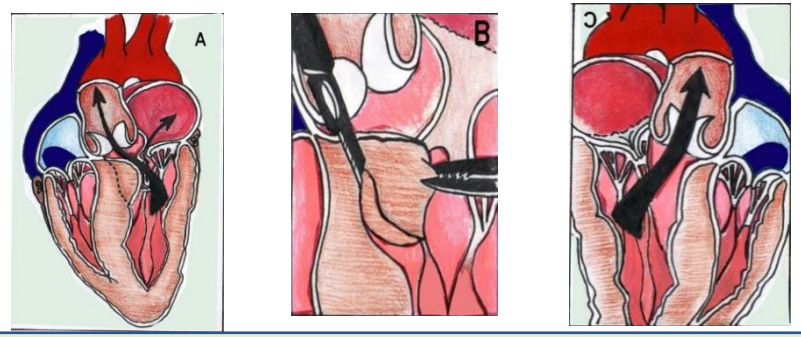


consistency of Doppler data

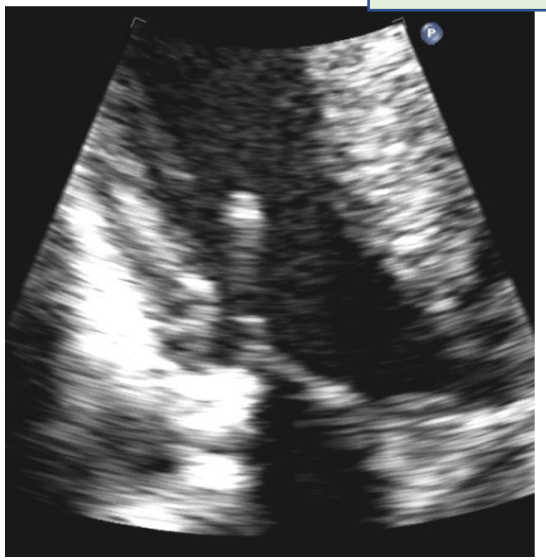
LV: 210 – 80 (grad LVOT) =
130 mm HG Ao pressure

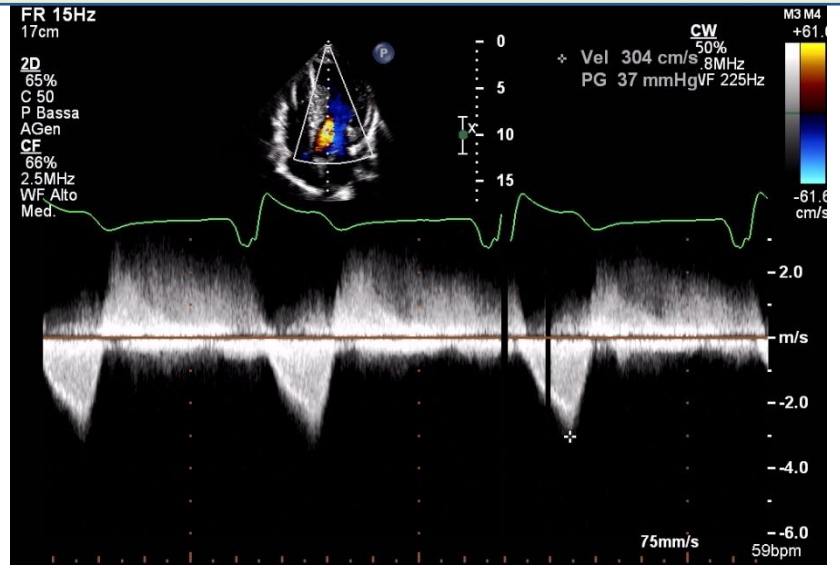
21 Year Lady: Morrow operation at 5 year; re-do at 10 year; ICD 2010;





NO SAM >>>> Ostruction due to the anterior motion of the posterior leaflet





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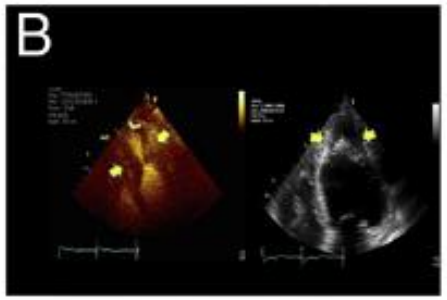
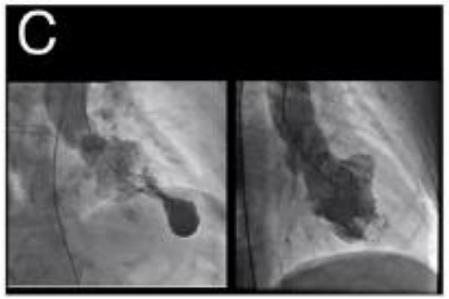
. 60, No. 17, 2012
0735-1097/$36.00
http://dx.doi.org/10.1016/j.jacc.2012.05.055

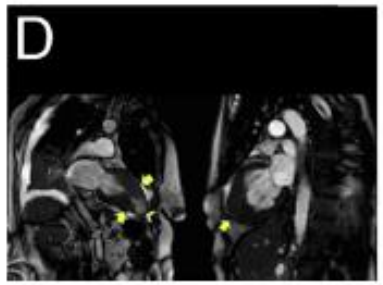
IMAGES IN CARDIOLOGY

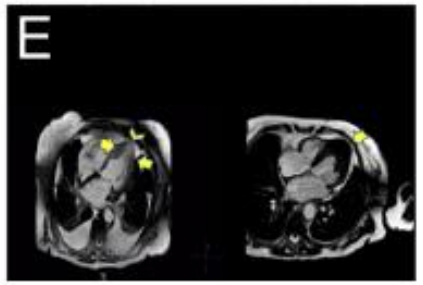
Apical Hypertrophic Cardiomyopathy With and Without Aneurysm

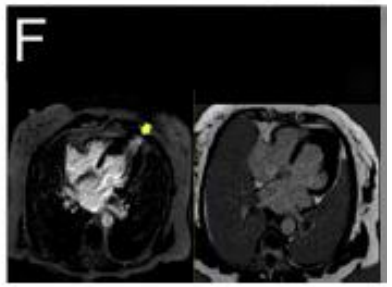
Arun Dahiya, MBBS,*† Paaladinesh Thavendiranathan, MD,* James D. Thomas, MD,*
Scott D. Flamm, MD*
Cleveland, Ohio; Brisbane, Queensland, Australia



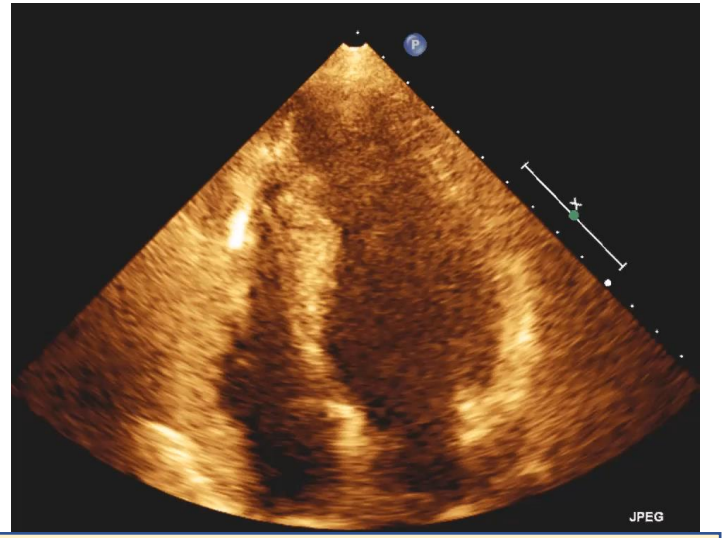




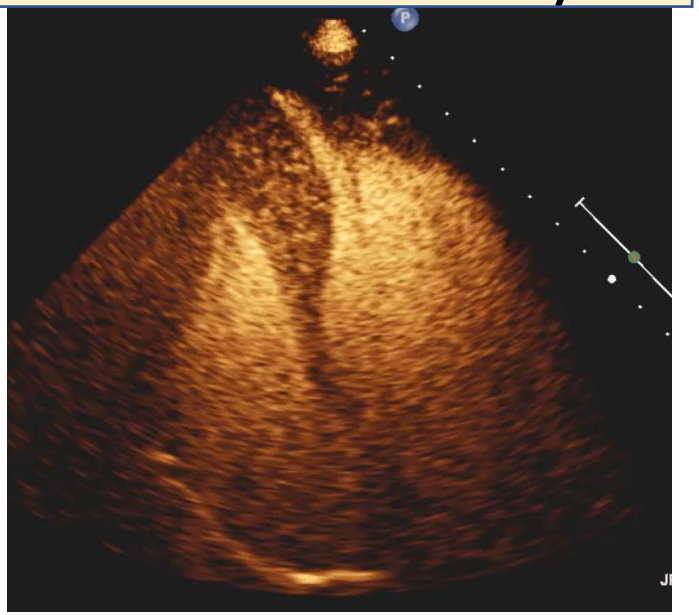




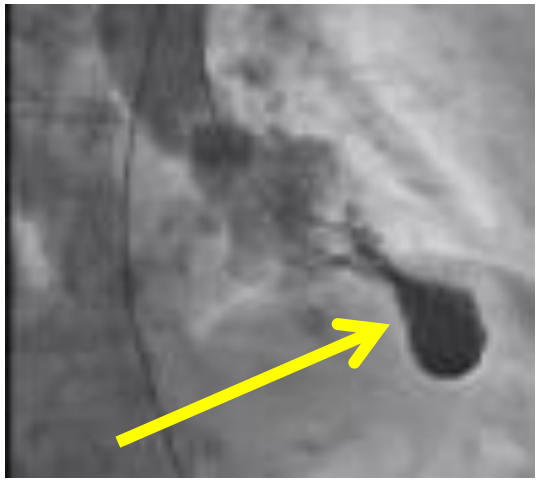
NO only HCM with LVOT obstruction but…



Apical Hypertrophic CMP without Aneurysm



1991 First Case with aneurysm in our series (21 year man)
VT hemodynamic instability (British Heart 1991)



Doppler evidence of abnormal intracavitary systolic and diastolic flow in hypertrophic cardiomyopathy with midventricular obstruction

Paolo Barbier, MD, and Antonio L. Bartorelli, MD
Milan, Italy

Am Heart 1993

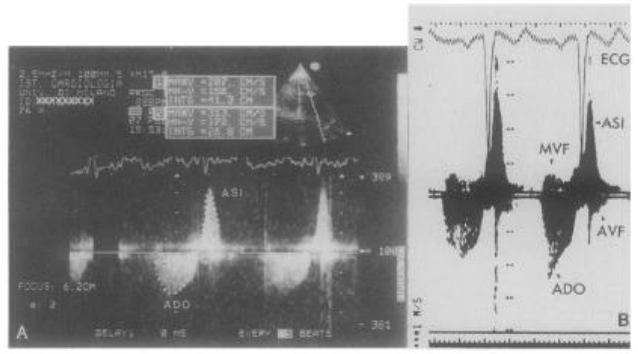
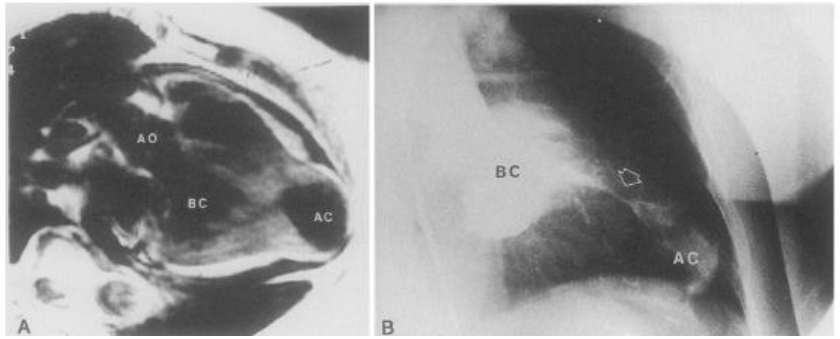
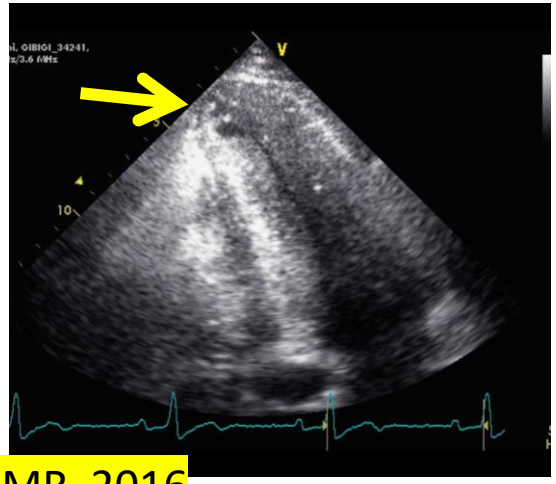


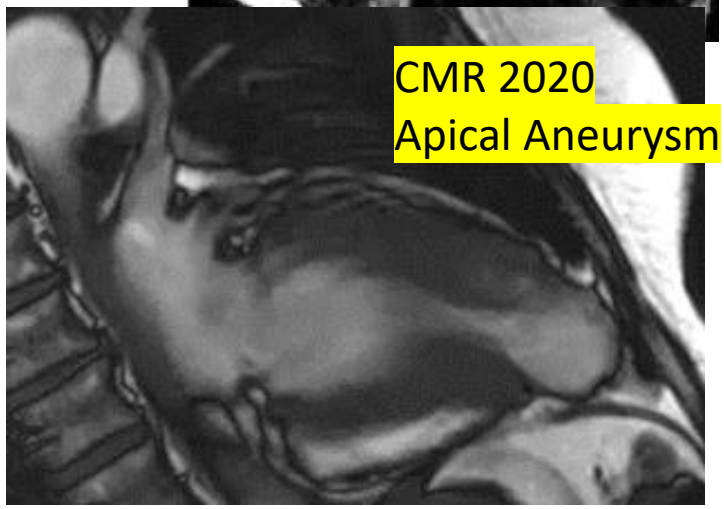
Fig. 3. Multigated continuous wave **(A)**, and nonimaging continuous wave **(B)** Doppler tracings of abnormal systolic and diastolic velocity patterns. *ADO*, Apical diastolic outflow; *ASI*, apical systolic inflow; *AVF*, aortic valve flow; *ECG*, electrocardiogram; *MVF*, mitral valve flow.



Doppler Pattern

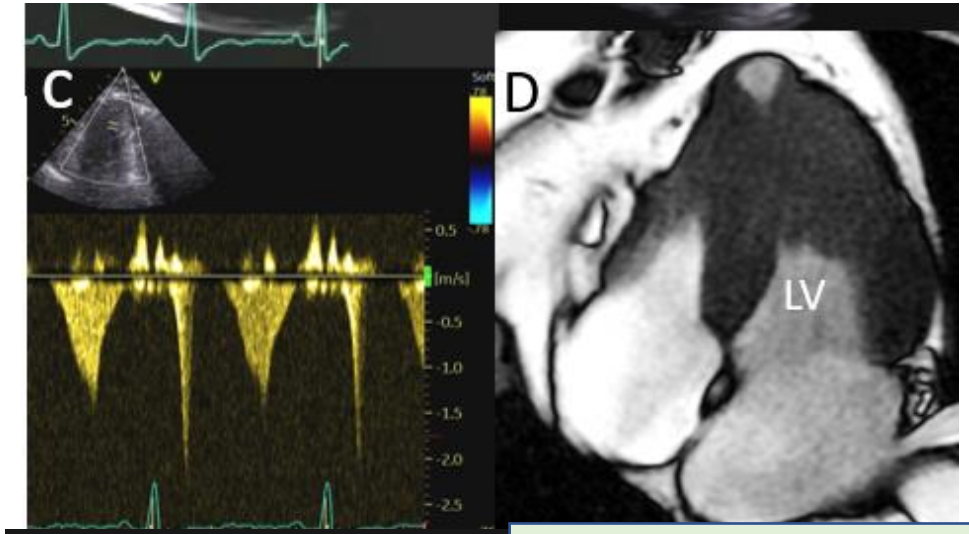


CMR 2016
Apical scar

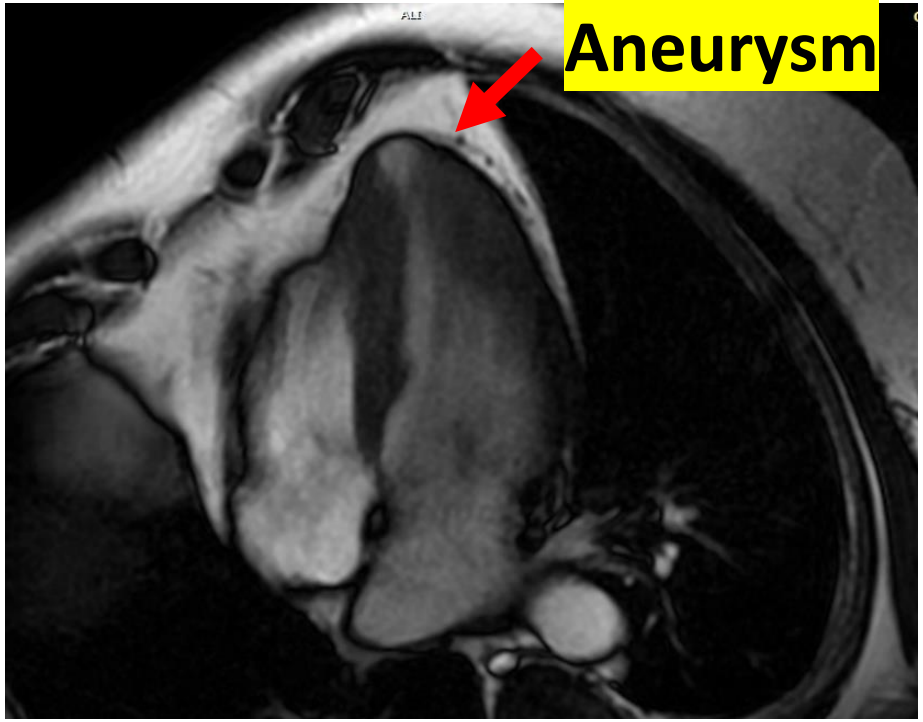


CMR 2020
Apical Aneurysm

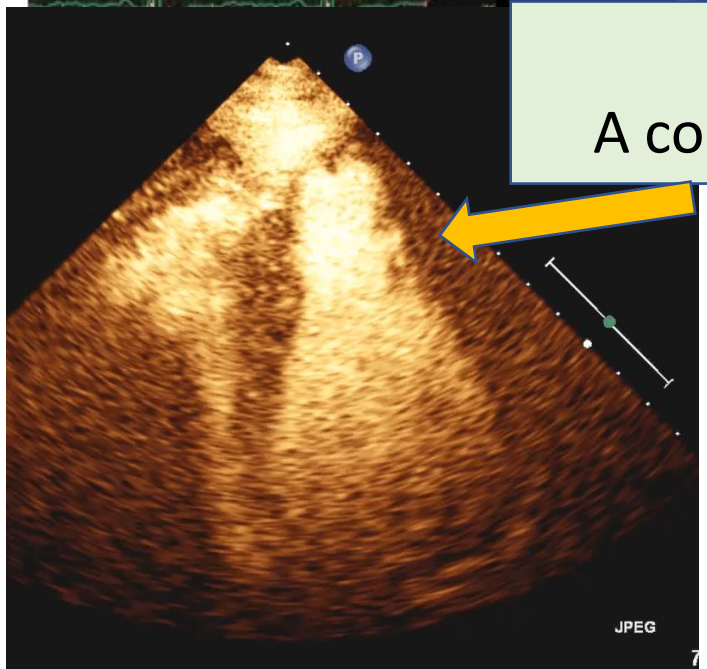
Relately Easy ECHO diagnosis even in small Aneurysms (with an accuracy similar to CMR thanks to contrast agents)

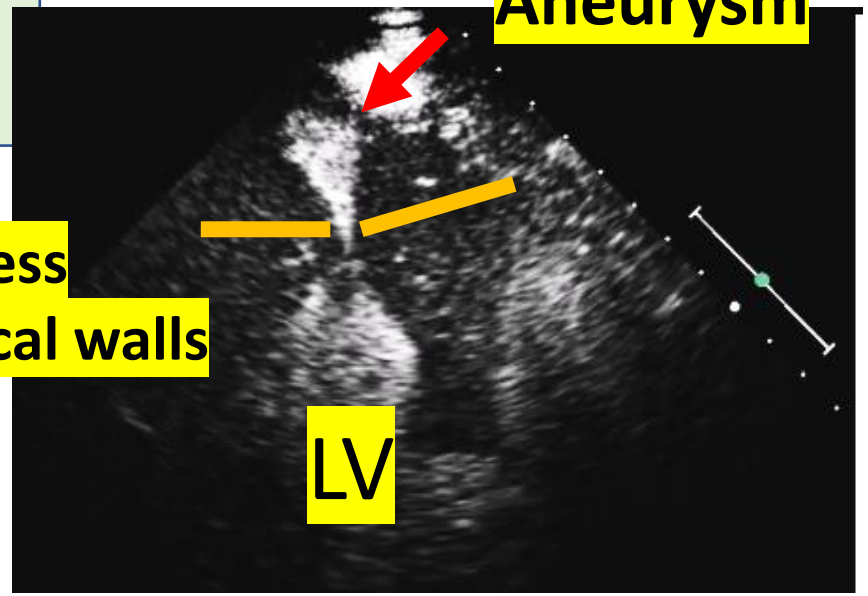


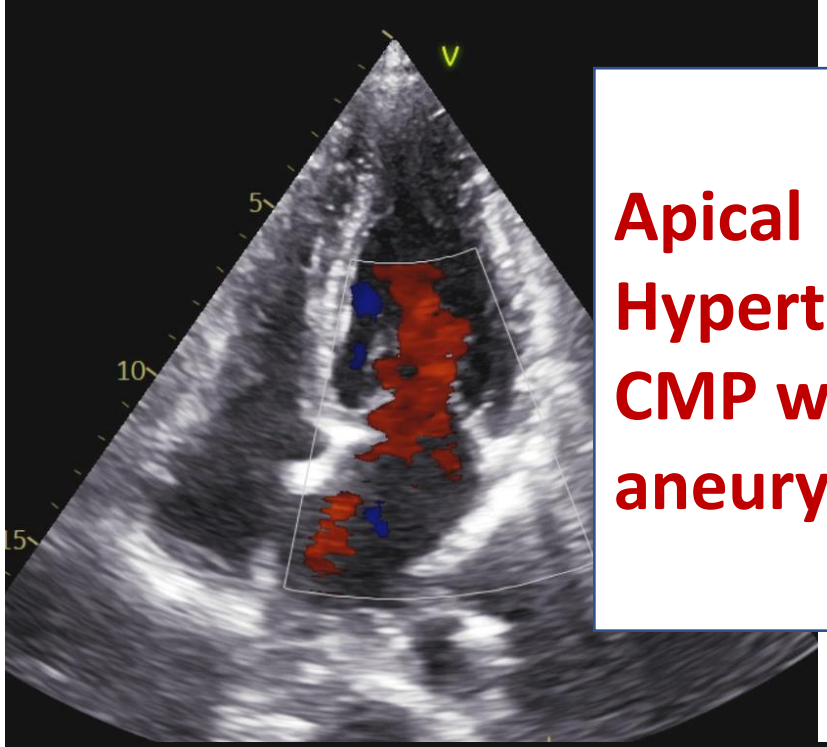
2023



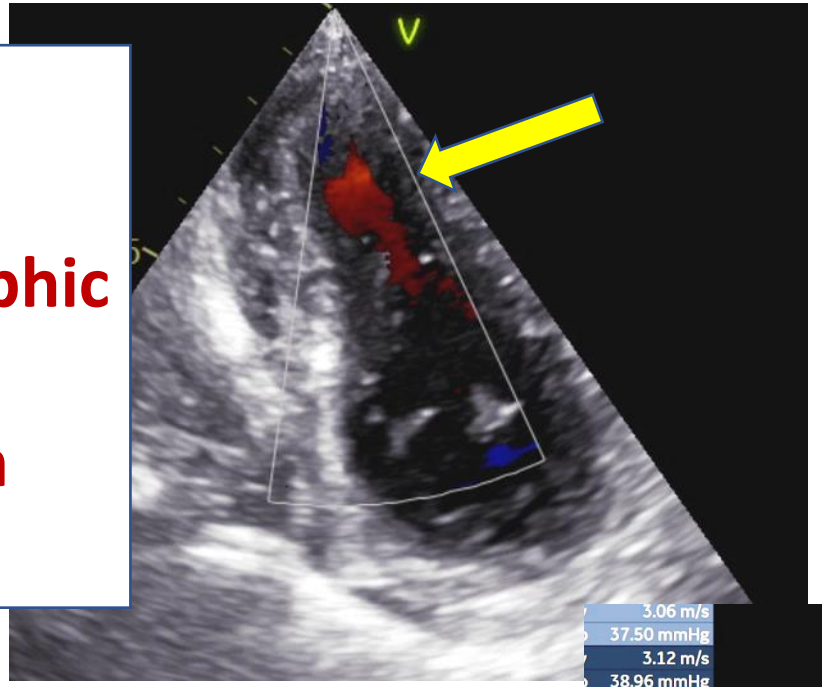
Subpoptimal ECHO
A contrast ECHO was performed



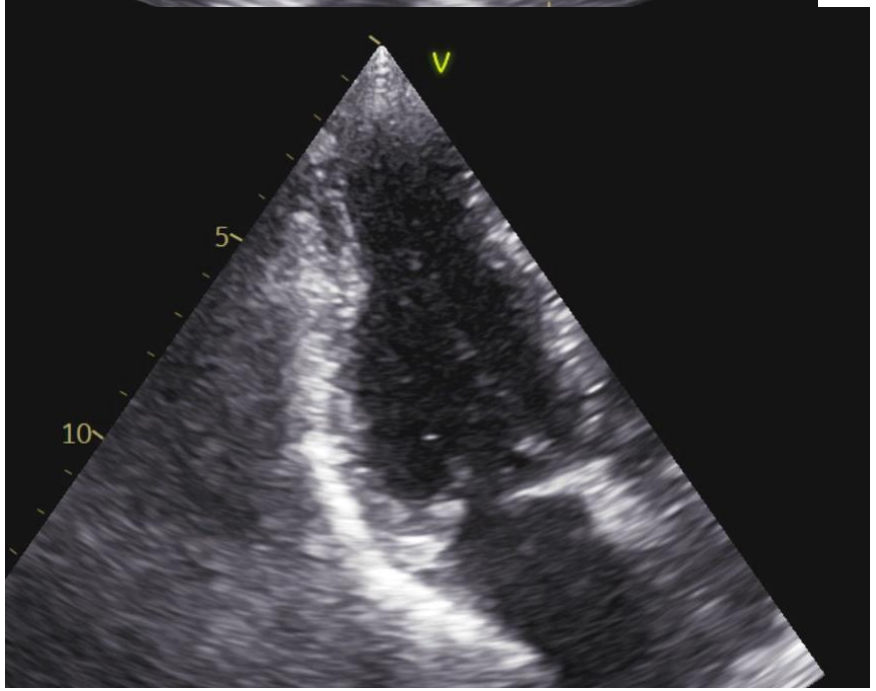


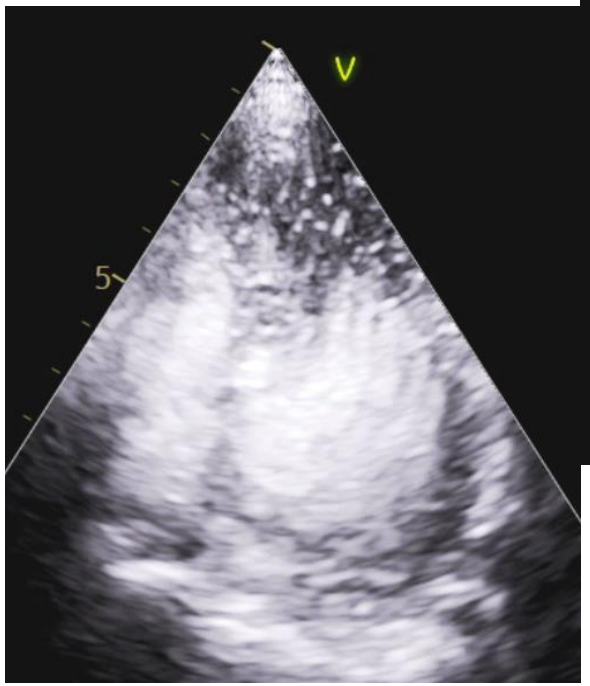


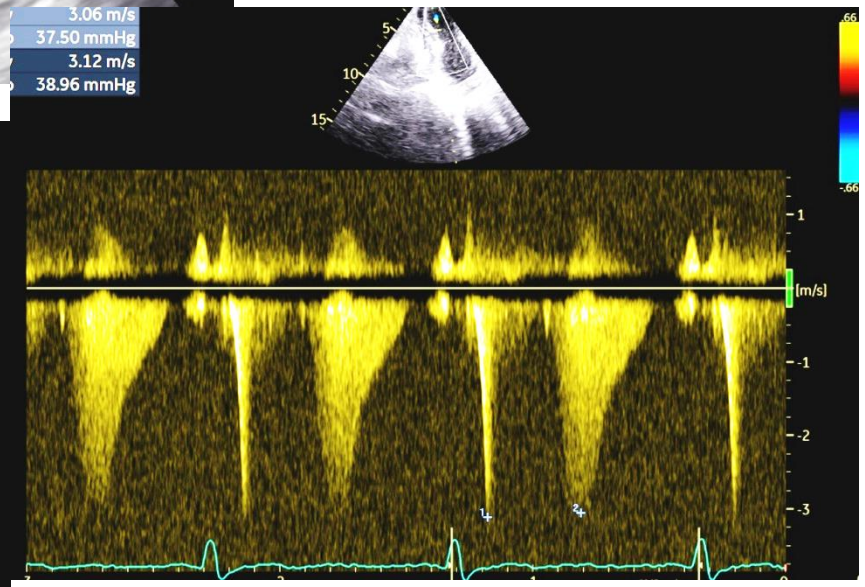
Apical Hypertrophic CMP with aneurysm



Void Pattern
(pulsed Doppler)in the apical Region at the level of the obstruction







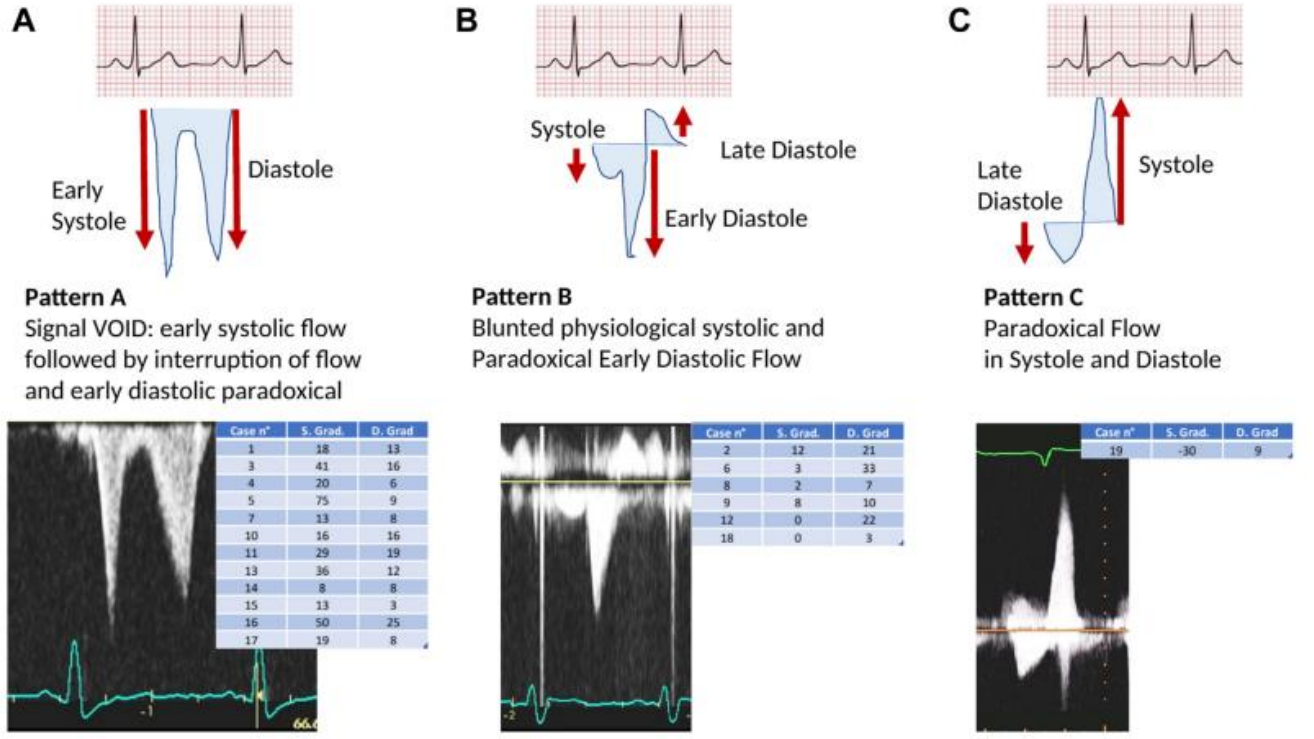
CLINICAL CASE SERIES

JACC Cases 2023

Is There a Typical Doppler Pattern in Patients With Apical Hypertrophic Cardiomyopathy With Aneurysm?

Gerardo Vito Lo Russo, MD,[a,b] Mauro Pepi, MD,[b] Saima Mushtaq, MD,[b] Valentina Mantegazza, MD,[b] Fabrizio Celeste, MD[b]

FIGURE 1 Schematic Representation of the Doppler Pattern



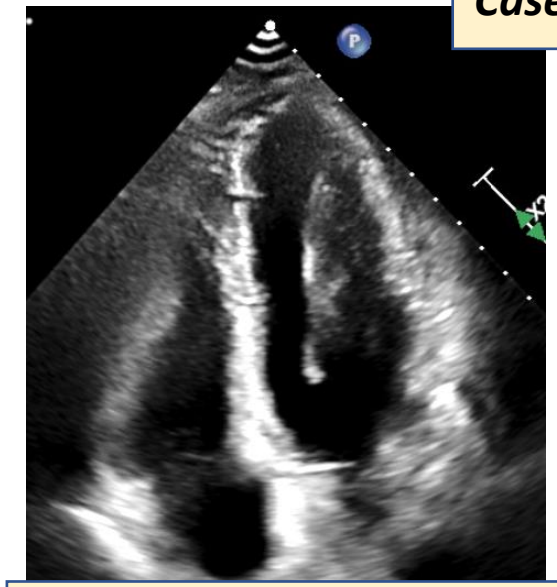
(A) Pattern A (more frequent). **(B)** Pattern B. **(C)** Pattern C. S. Grad = systolic gradient (mm Hg); D. G. = diastolic gradient (mm Hg).

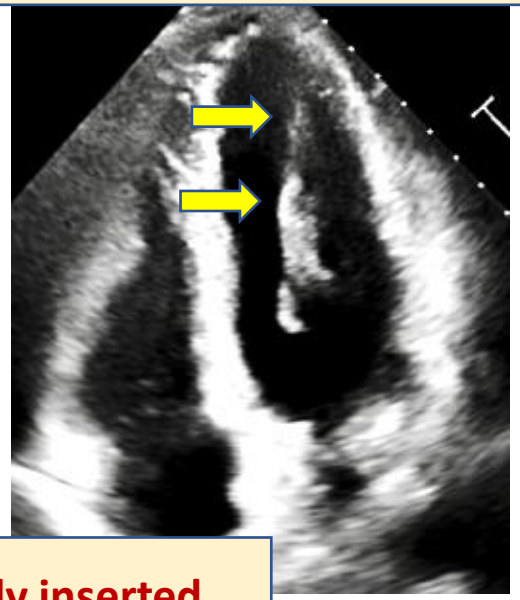
We indentified 3 different flow patterns at the midventricular level using CW Doppler.

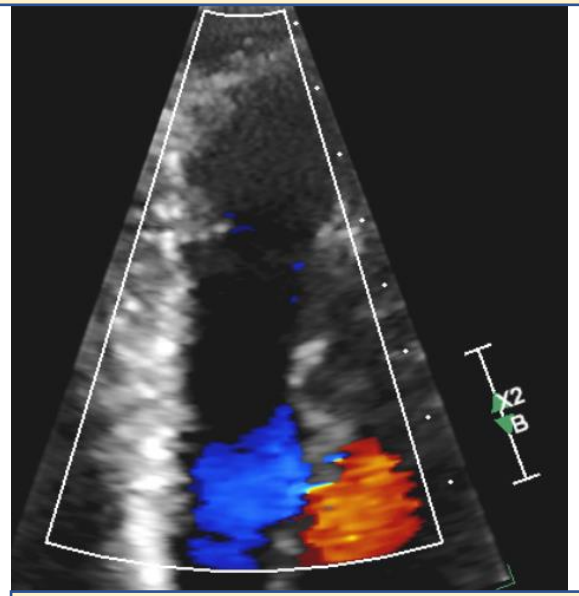
The most frequent was pattern A, followed by pattern B and pattern C.

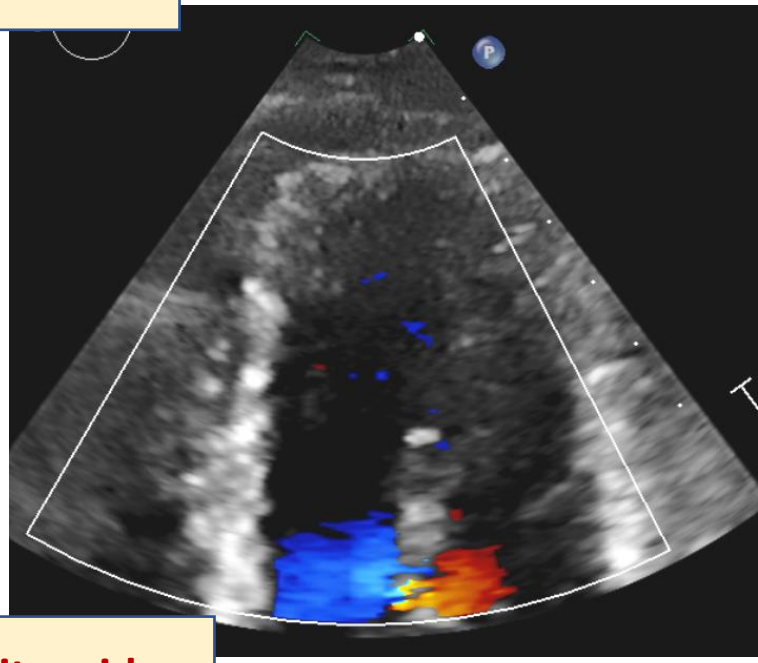
All are an expression of increased intracavitary pressure and consequently decreased coronary pressure that may contribute to the pathophysiology of aneurysm formation.

Case 1 : Apical HCP with apical bulging and anomalous papillary muscles

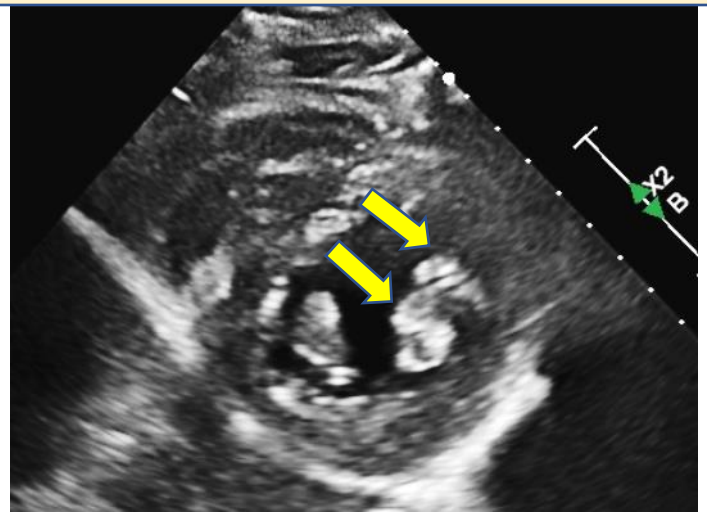




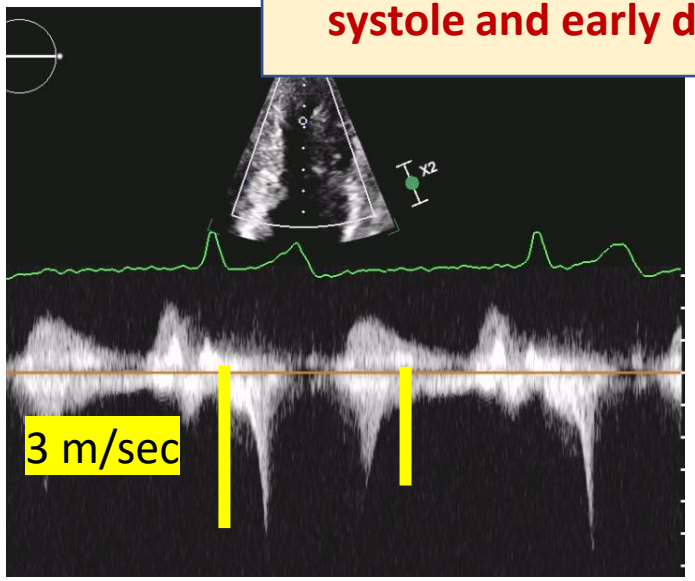


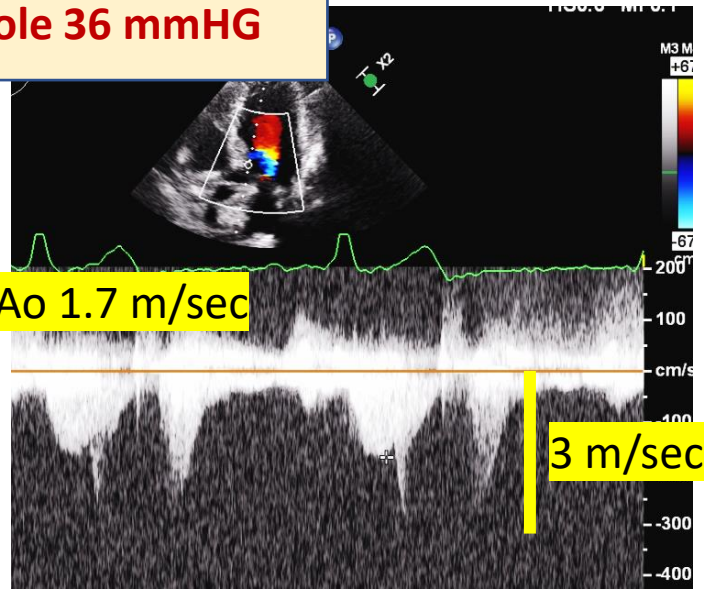


BIFID anterior and Apically inserted Papillary muscle



VOID PATTERN : max velocity mid systole and early diastole 36 mmHG

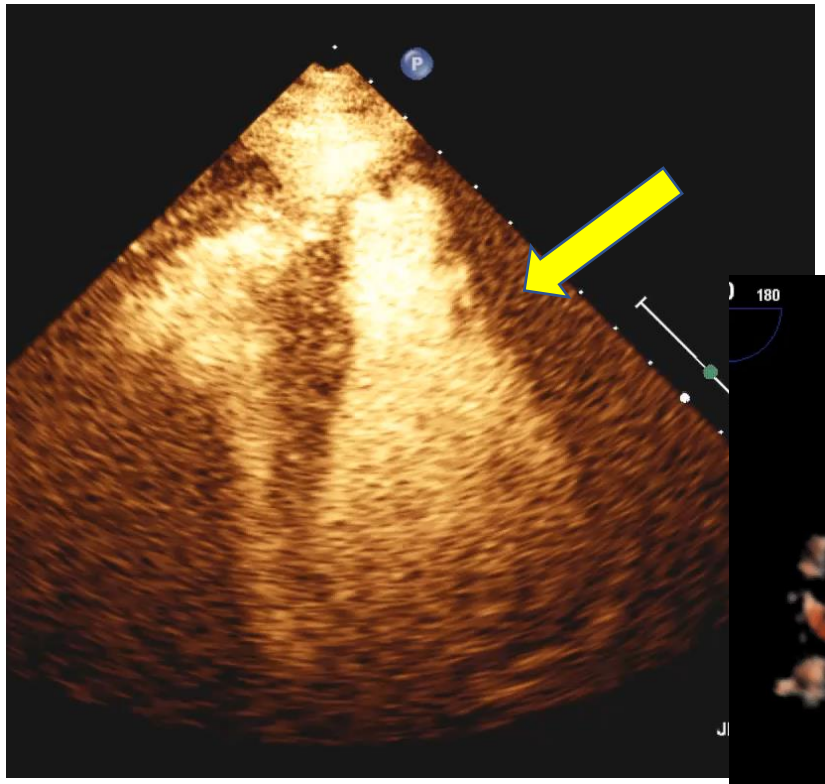


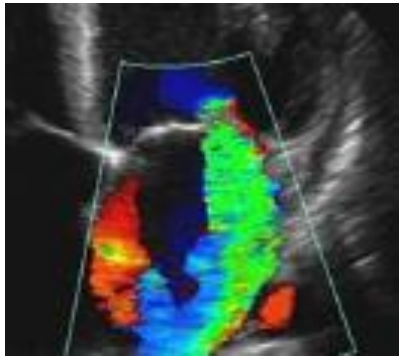


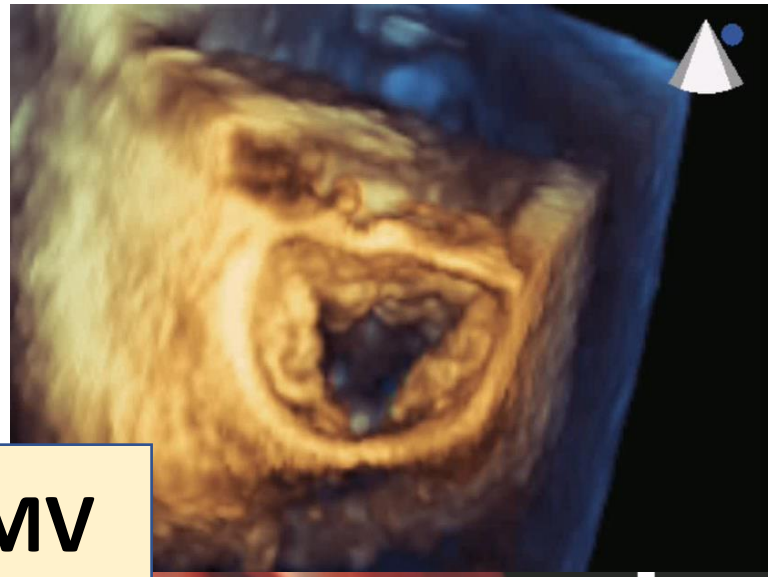
Case 2: Apical HCP with apical bulging and MV anomaly

Referred for severe MR >>>> very Uncommon Anomaly of the MV

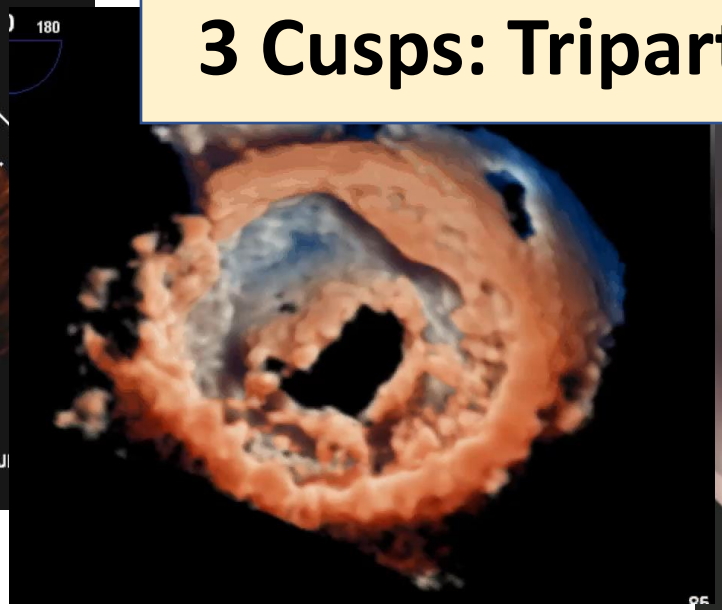
HOCM with a midventricular gradient and a complete cleft of the posterior MV leaflet

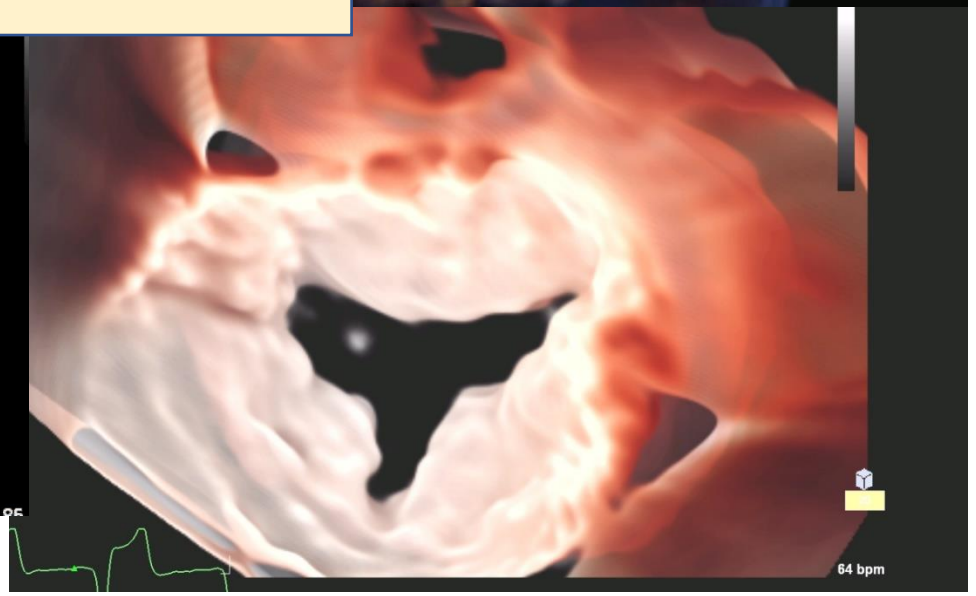






3 Cusps: Tripartite MV





Hypertrophic Cardiomyopathy

"Diagnostic Work-up of concomitant mitral valve pathology"

- **Anomalies in the MV apparatus** , such as the mitral leaflets, papillary muscles, and chordae tendineae, as well as apical-basal muscle bundle , that extend from the LV apex to the basal septum, **are often observed in patients with HCM.**
- **These anomalies** can be **detected by transthoracic and transesophageal echocardiography and by cardiac magnetic resonance.**
- A thorough **understanding of the complex anatomy in HCM is essential for determining the most appropriate treatment strategy for each patient.**

Centro Cardiologico
Monzino