2023 ESC Guidelines pro diagnostiku a léčbu kardiomyopatií: *Echokardiografie*

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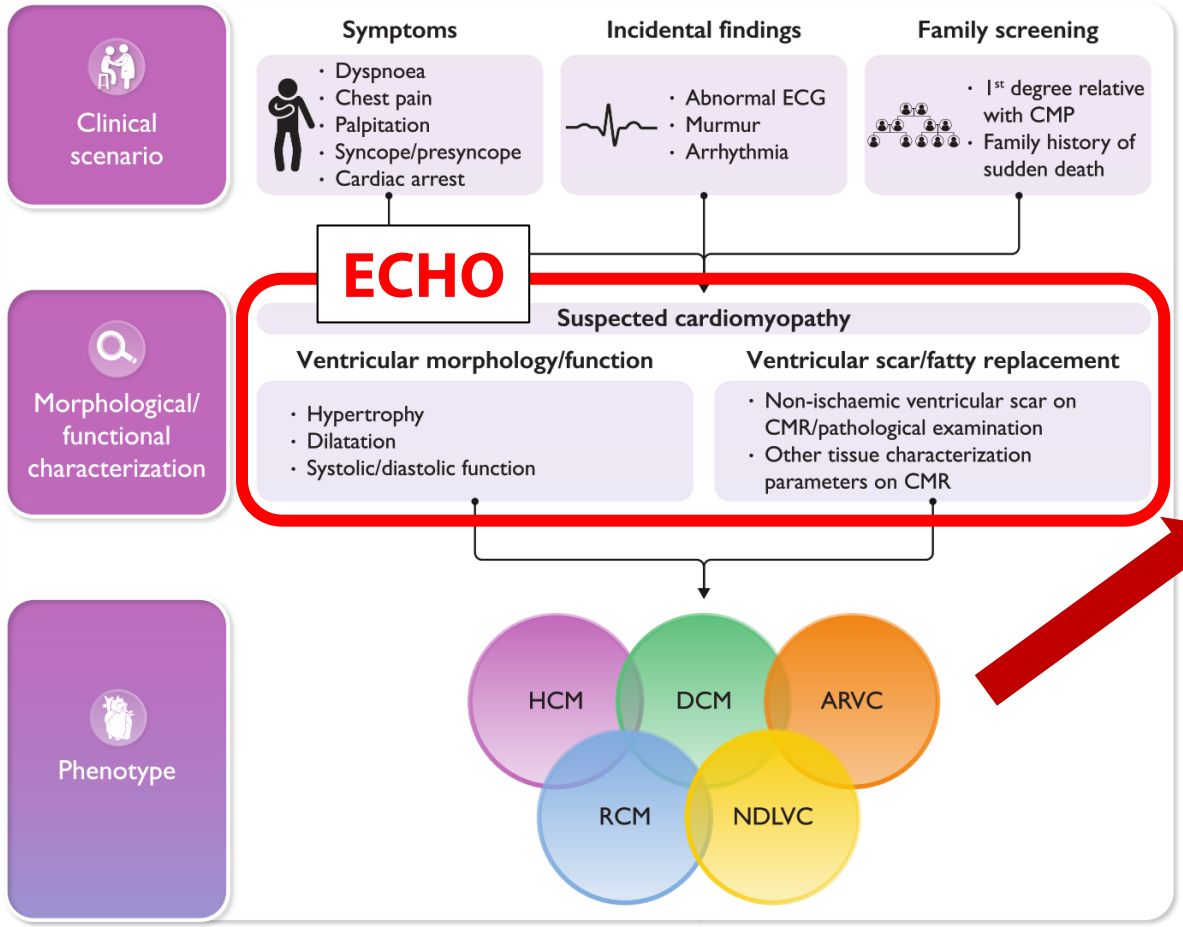
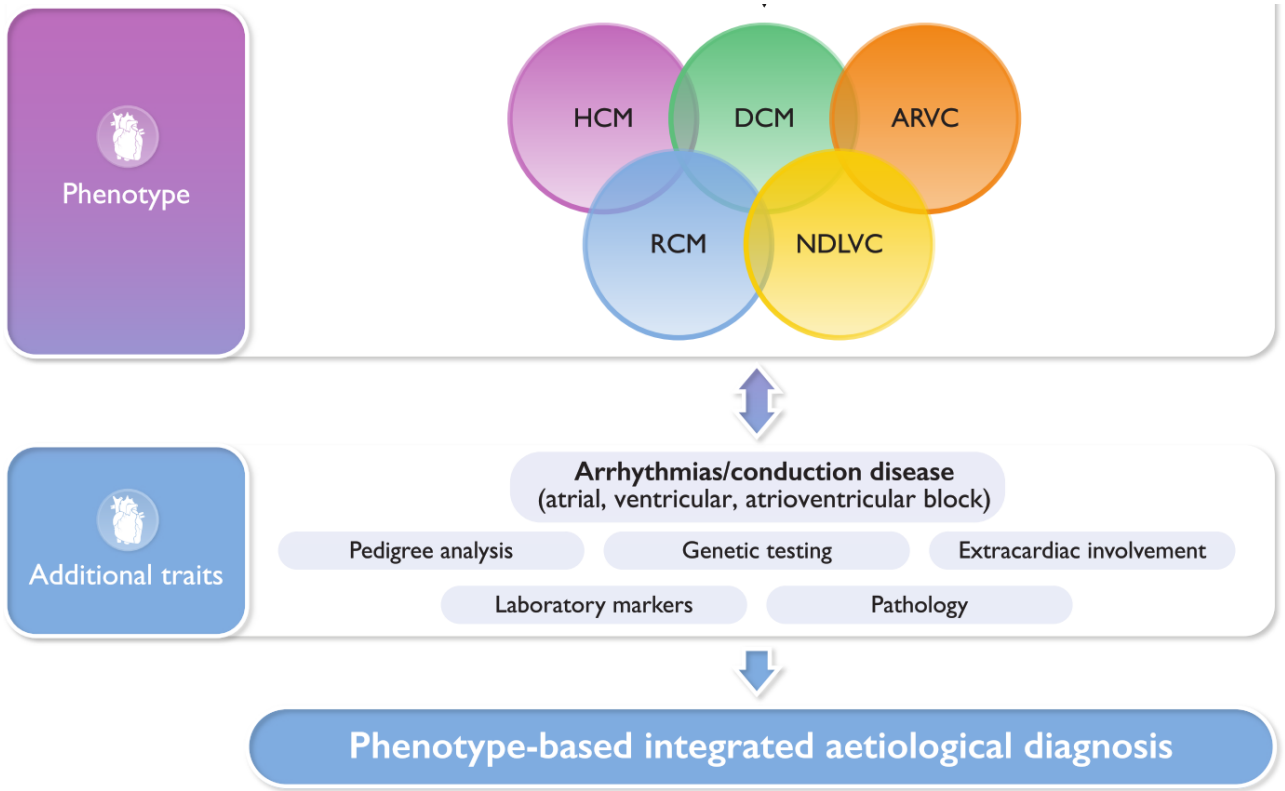
2023 ESC Guidelines: Fenotypický přístup



„Cardiomyopathy Mind-Set“



2023 ESC Guidelines: Echokardiografie

Recommendation Table 2 — Recommendations for diagnostic work-up in cardiomyopathies

Recommendations	Class[a]	Level[b]
It is recommended that all patients with suspected or established cardiomyopathy undergo systematic evaluation using a multiparametric approach that includes clinical evaluation, pedigree analysis, ECG, Holter monitoring, laboratory tests, and multimodality imaging.[63]	I	C
It is recommended that all patients with suspected cardiomyopathy undergo evaluation of family history and that a three- to four-generation family tree is created to aid in diagnosis, provide clues to underlying aetiology, determine inheritance pattern, and identify at-risk relatives.[64–66]	I	C

Recommendation Table 4 — Recommendation for echocardiographic evaluation in patients with cardiomyopathy

Recommendation	Class[a]	Level[b]
A comprehensive evaluation of cardiac dimensions and LV and RV systolic (global and regional) and LV diastolic function is recommended in all patients with cardiomyopathy at initial evaluation, and during follow-up, to monitor disease progression and aid risk stratification and management.[78,83–102]	I	B

???

konkomitantní chlopenní vada

tlakové poměry malého oběhu

perikardiální výpotek

…

Echokg & kardiomyopatie

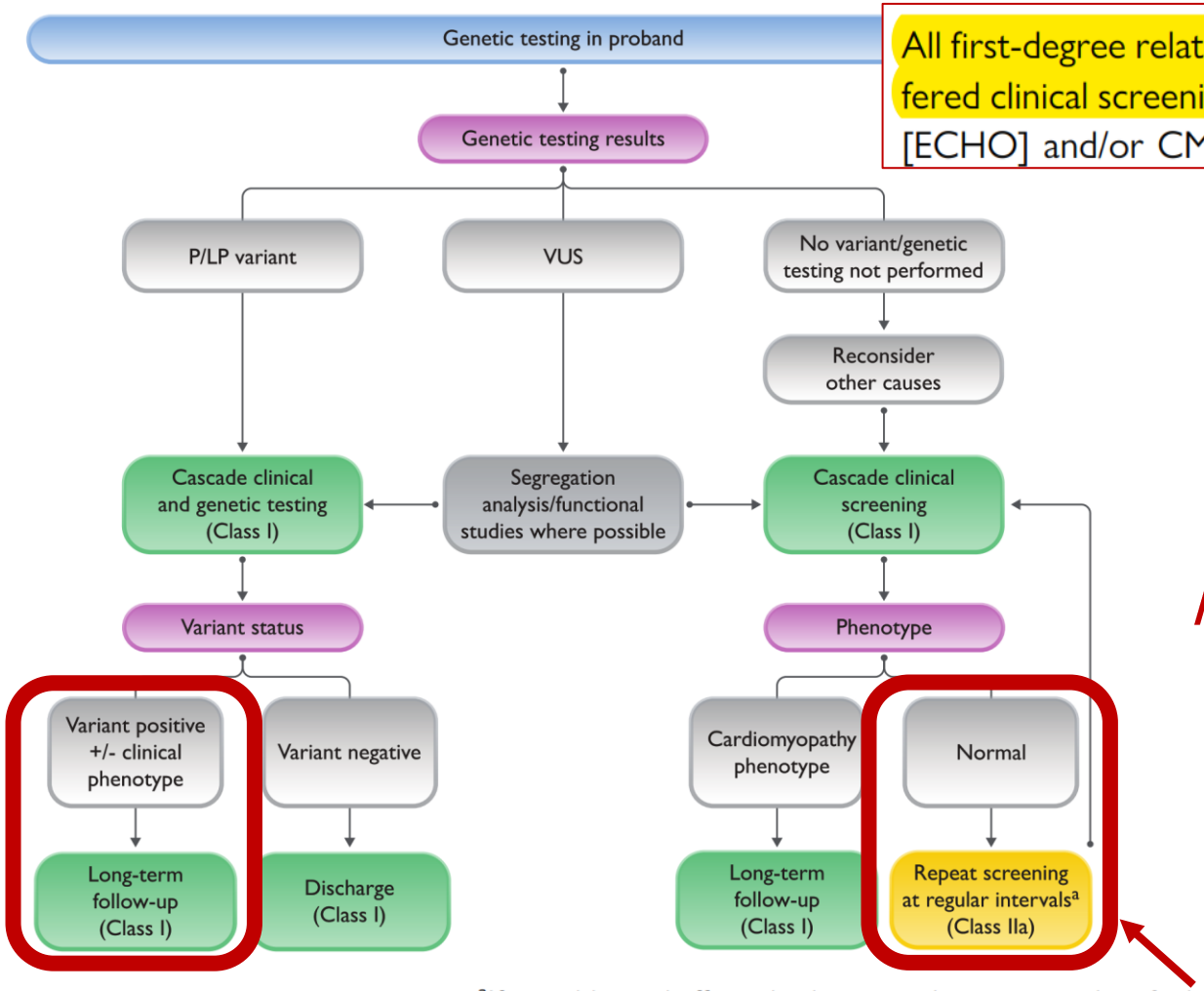
- **diagnostika kardiomyopatie**, její komplexní morfologicko-funkční charakteristika
- **riziková stratifikace a prognostické informace**
- **vedení a monitorace léčby** (farmako- i nefarmakologické; obecné, specifické)
- **sledování progrese**, event. regrese onemocnění
- **screening** prvostupňových příbuzných

Recommendations	Class[a]	Level[b]
It is recommended that all clinically stable patients with cardiomyopathy undergo routine follow-up using a multiparametric approach that includes ECG and echocardiography every 1 to 2 years.	I	C
Clinical evaluation with ECG and multimodality imaging is recommended in patients with cardiomyopathy whenever there is a substantial or unexpected change in symptoms.	I	C

2023 ESC Guidelines for the management of cardiomyopathies

Echokg & rodinný screening

All first-degree relatives of patients with cardiomyopathy should be offered clinical screening with ECG and cardiac imaging (echocardiogram [ECHO] and/or CMR). In families in whom a disease-causing genetic



ECHO + EKG á 1-3 roky do 60-ti let věku /event. další vyšetření/, poté á 3-5 let

[a]If no additional affected relatives and no variant identified on genetic testing, consider earlier termination of clinical screening.

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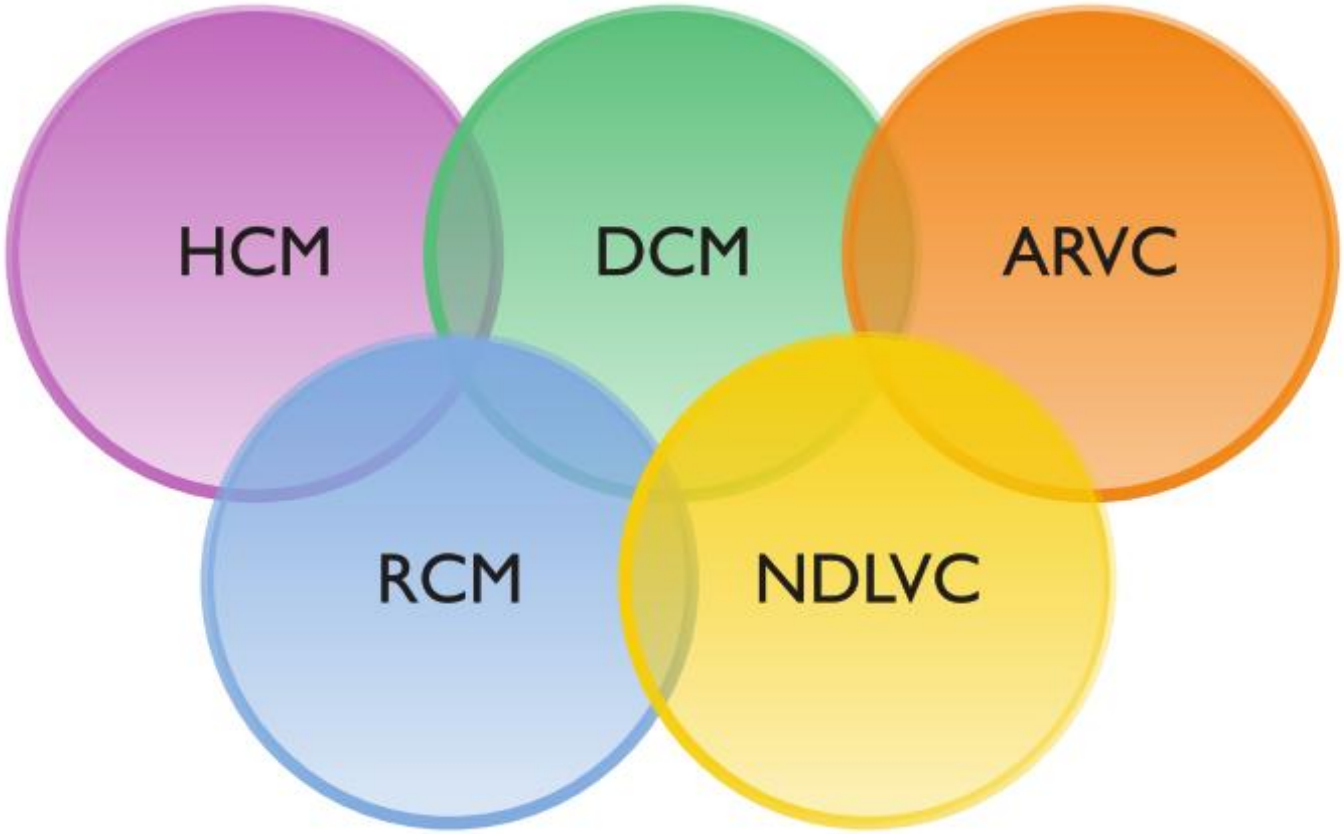
Fenotypy kardiomyopatií



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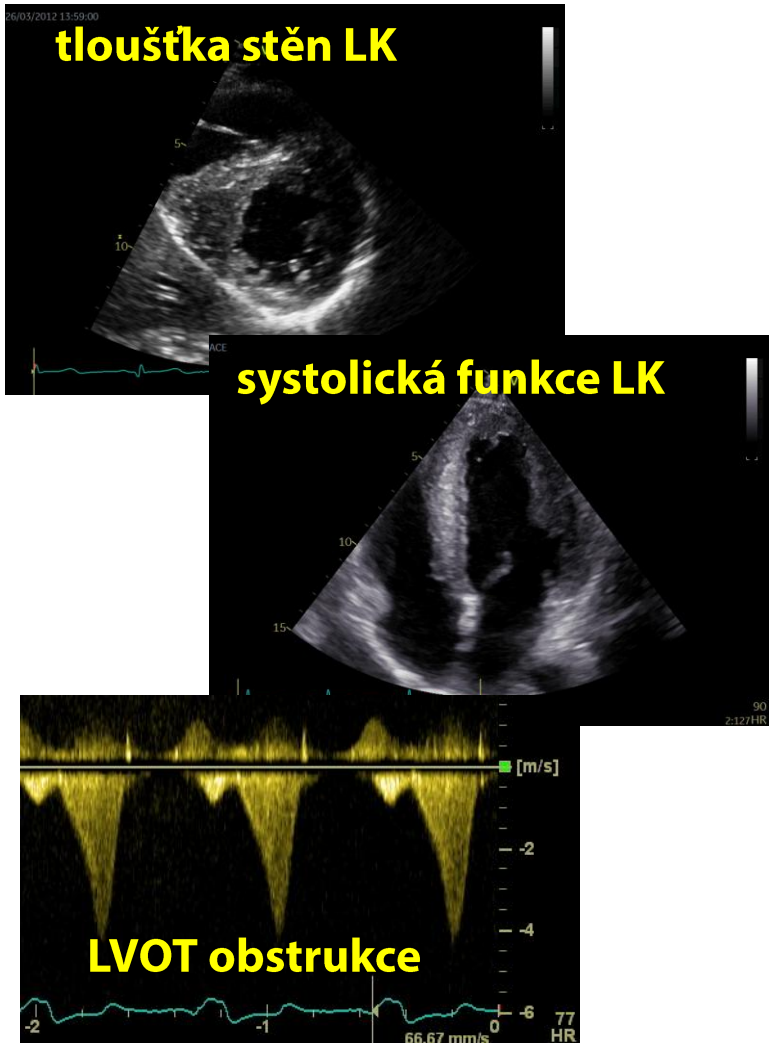
Hypertrofická kardiomyopatie & echokg

Item to assess	Primary imaging modality	Comments
LV wall thickness	ECHO/CMR	• All LV segments from base to apex examined in end-diastole, preferably in the 2D short-axis view, ensuring that the wall thickness is recorded at mitral, mid-LV, and apical levels. • CMR is superior in the detection of LV apical and anterolateral hypertrophy, aneurysms,[580] and thrombi,[581] and is more sensitive in the detection of subtle markers of disease in patients with sarcomeric protein gene variants (e.g. myocardial crypts, papillary muscle abnormalities).[159,582,583]
Systolic function (global and regional)	ECHO/CMR	• Ejection fraction is a suboptimal measure of LV systolic performance when hypertrophy is present. • Doppler myocardial velocities and deformation parameters (strain and strain rate) are typically reduced at the site of hypertrophy despite a normal EF and may be abnormal before the development of increased wall thickness in genetically affected patients.
Diastolic function	ECHO	• Routine examination should include mitral inflow assessment, tissue Doppler imaging, pulmonary vein flow velocities, pulmonary artery systolic pressure, and LA size/volume.
Mitral valve	ECHO	• Assess presence and degree of SAM and mitral regurgitation. The presence of a central- or anteriorly directed jet of mitral regurgitation should raise suspicion of an intrinsic/primary mitral valve abnormality and prompt further assessment.
LVOT	ECHO	• See *Figure 12*.
LA dimensions	ECHO/CMR	• Provides important prognostic information.[365,525,584] • Most common mechanisms of LA enlargement are SAM-related mitral regurgitation and elevated LV filling pressures.
Myocardial fibrosis/LGE	CMR	• The distribution and severity of interstitial expansion can suggest specific diagnoses. Anderson–Fabry disease is characterized by a reduction in non-contrast T1 signal and the presence of posterolateral LGE.[134,155] In cardiac amyloidosis, there is often global, subendocardial or segmental LGE and a highly specific pattern of myocardial and blood-pool gadolinium kinetics caused by similar myocardial and blood T1 signals.[585,586]



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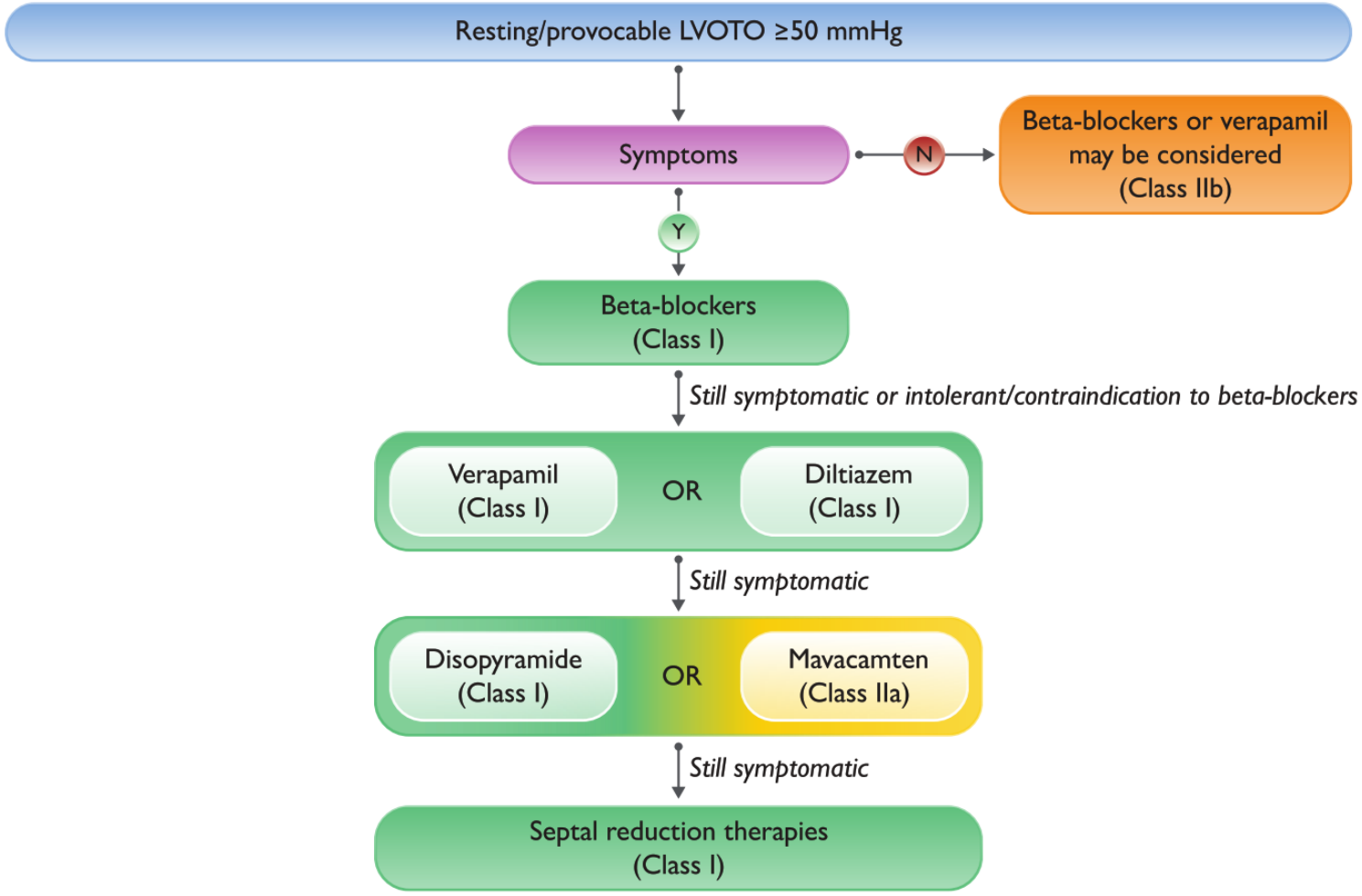
Hypertrofická kardiomyopatie: LVOT obstrukce



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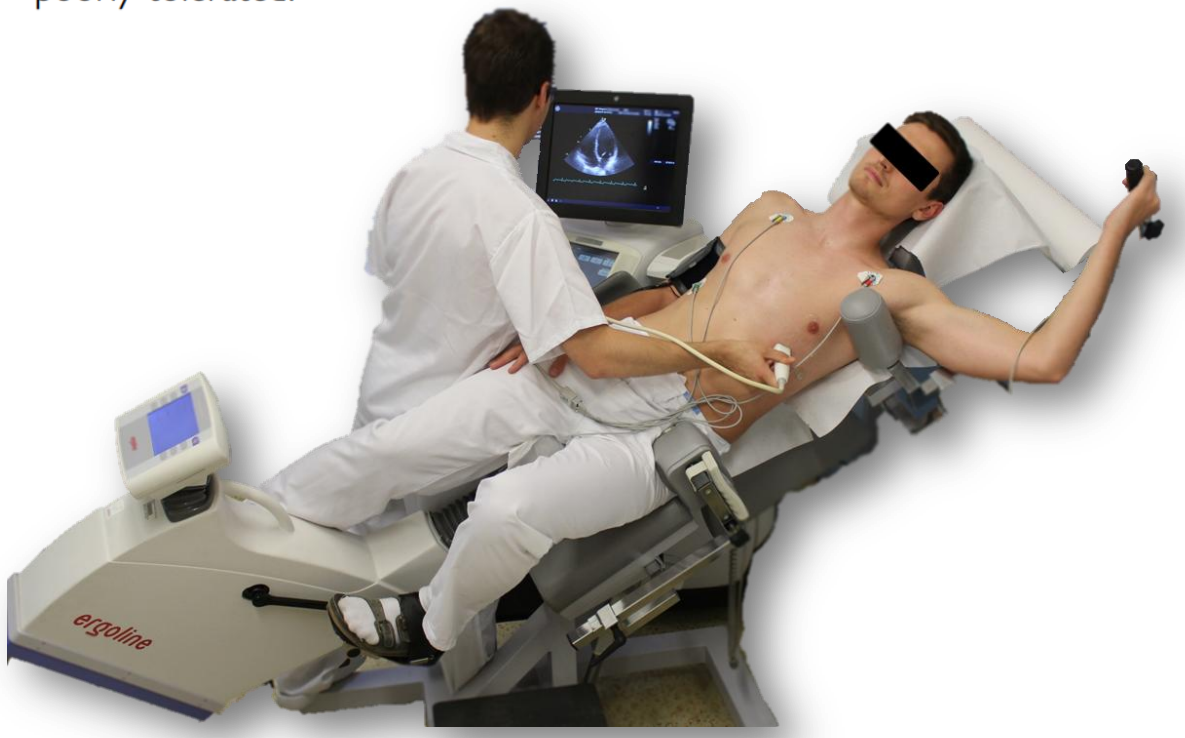
HKMP: echokg & obstrukce levé komory

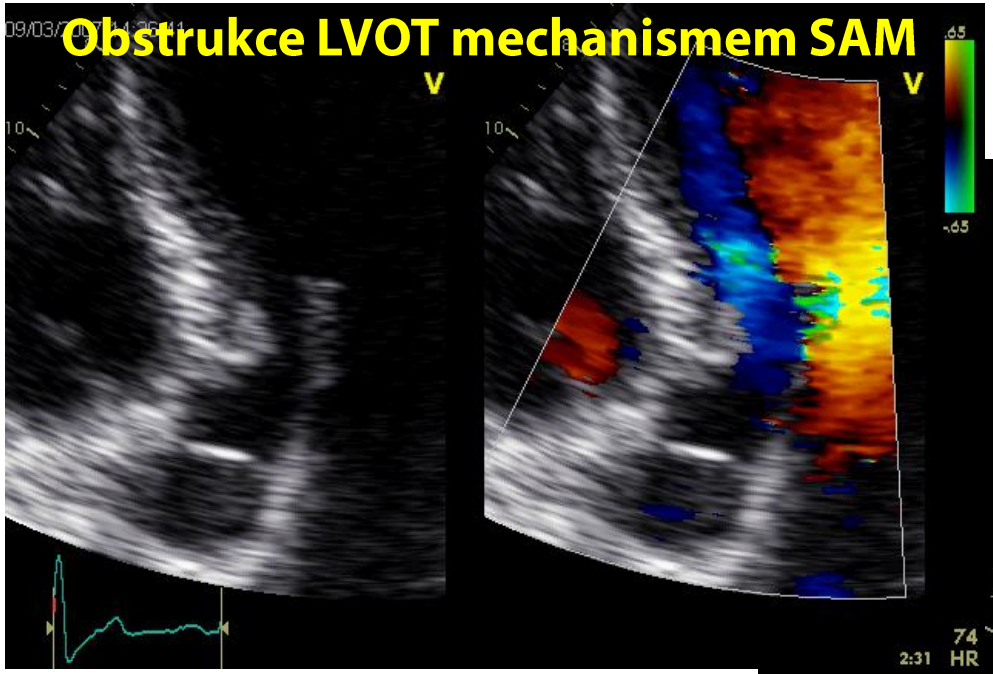
Recommendations	Class[a]	Level[b]
In all patients with HCM, at initial evaluation, transthoracic 2D and Doppler echocardiography are recommended, at rest and during Valsalva manoeuvre in the sitting and semi-supine positions—and then on standing if no gradient is provoked—to detect LVOTO.[84,86,365,525,584,587,589–594]	I	B
In symptomatic patients with HCM and a resting or provoked[c] peak instantaneous LV outflow tract gradient <50 mmHg, 2D and Doppler echocardiography during exercise in the standing, sitting (when possible), or semi-supine position are recommended to detect provocable LVOTO and exercise-induced mitral regurgitation.[588,595–598]	I	B

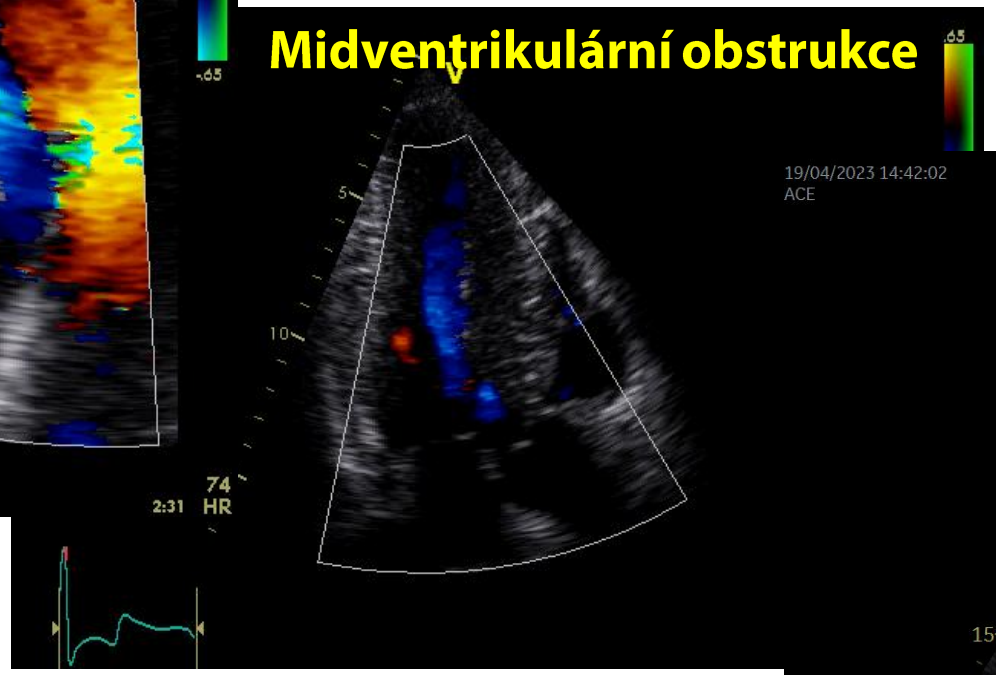
(*Figure 12*).[587,588] Exercise stress echocardiography is recommended in *symptomatic* patients if bedside manoeuvres fail to induce LVOTO ≥50 mmHg. Pharmacological provocation with dobutamine is not advised, as it is not physiological and can be poorly tolerated.

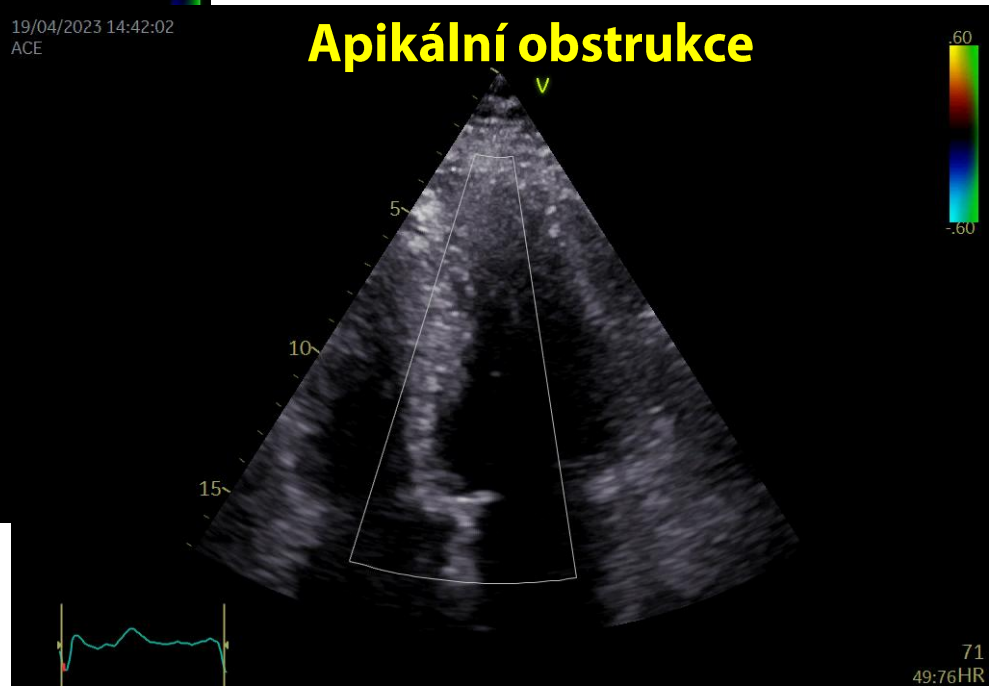


2023 ESC Guidelines for the management of cardiomyopathies

Hypertrofická kardiomyopatie: echokg & dynamická obstrukce není jen LVOTO

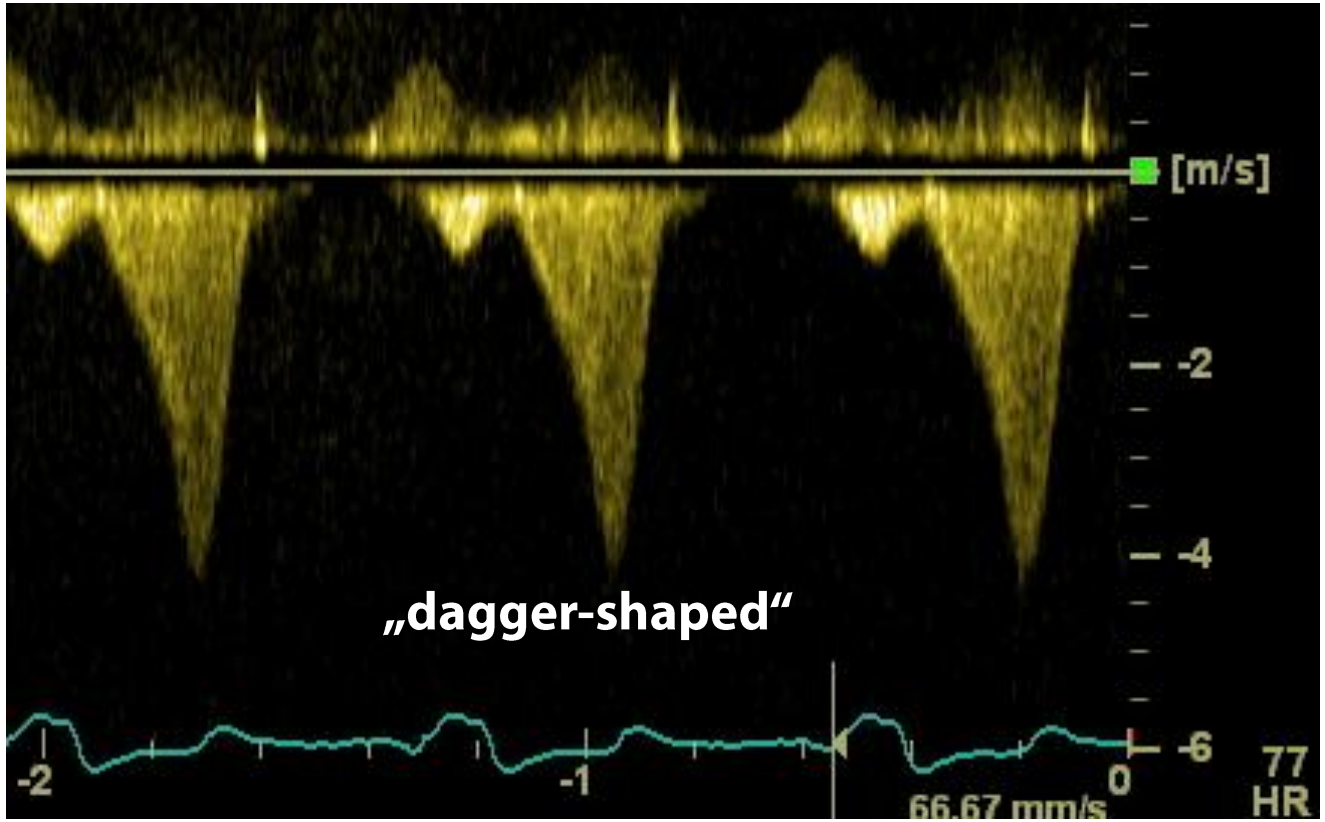




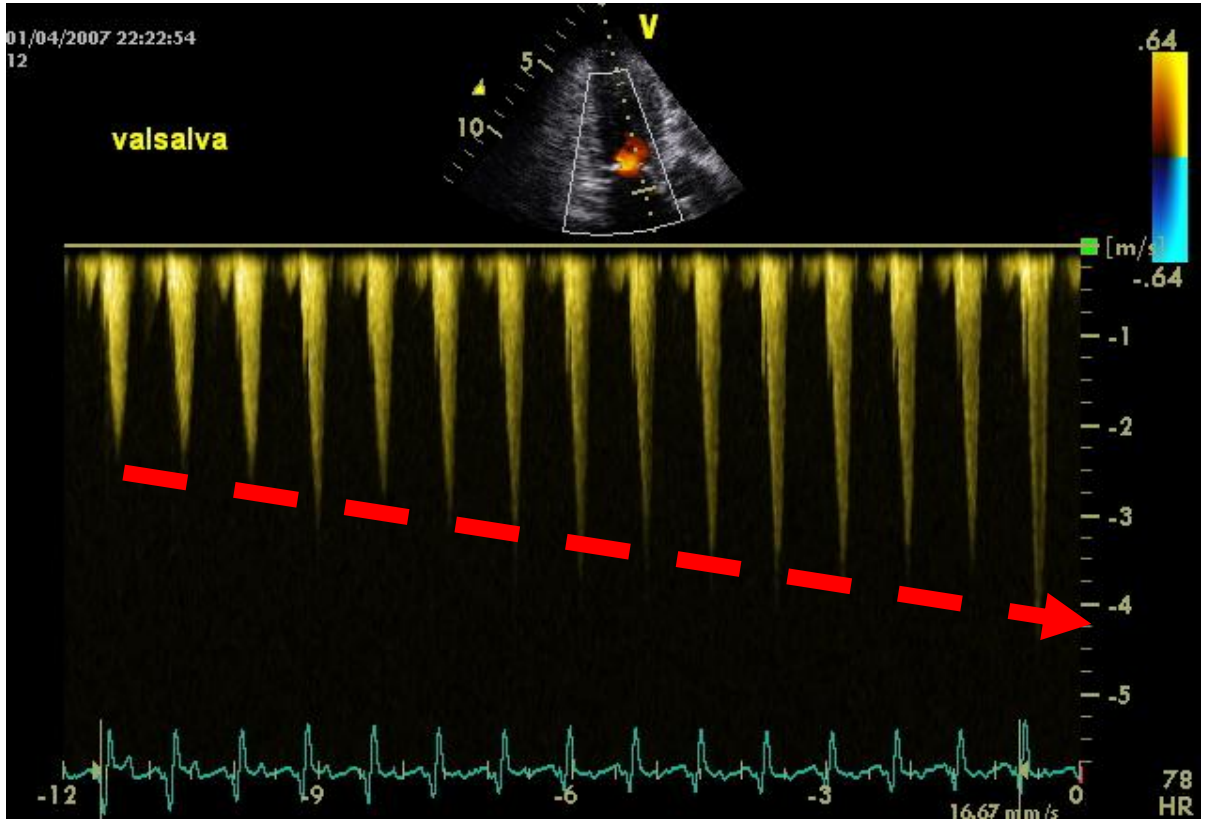


HKMP: echokg & dynamická obstrukce

CW Doppler



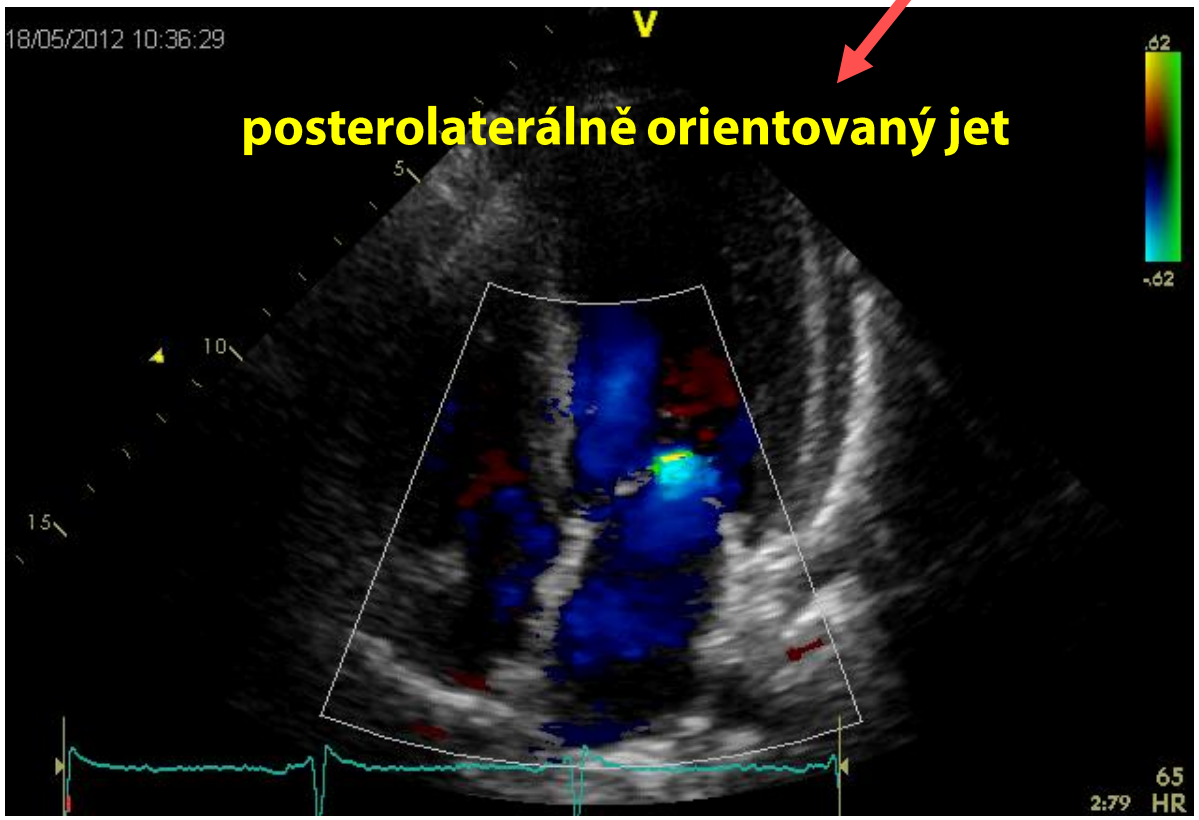
Provokace latentní obstrukce

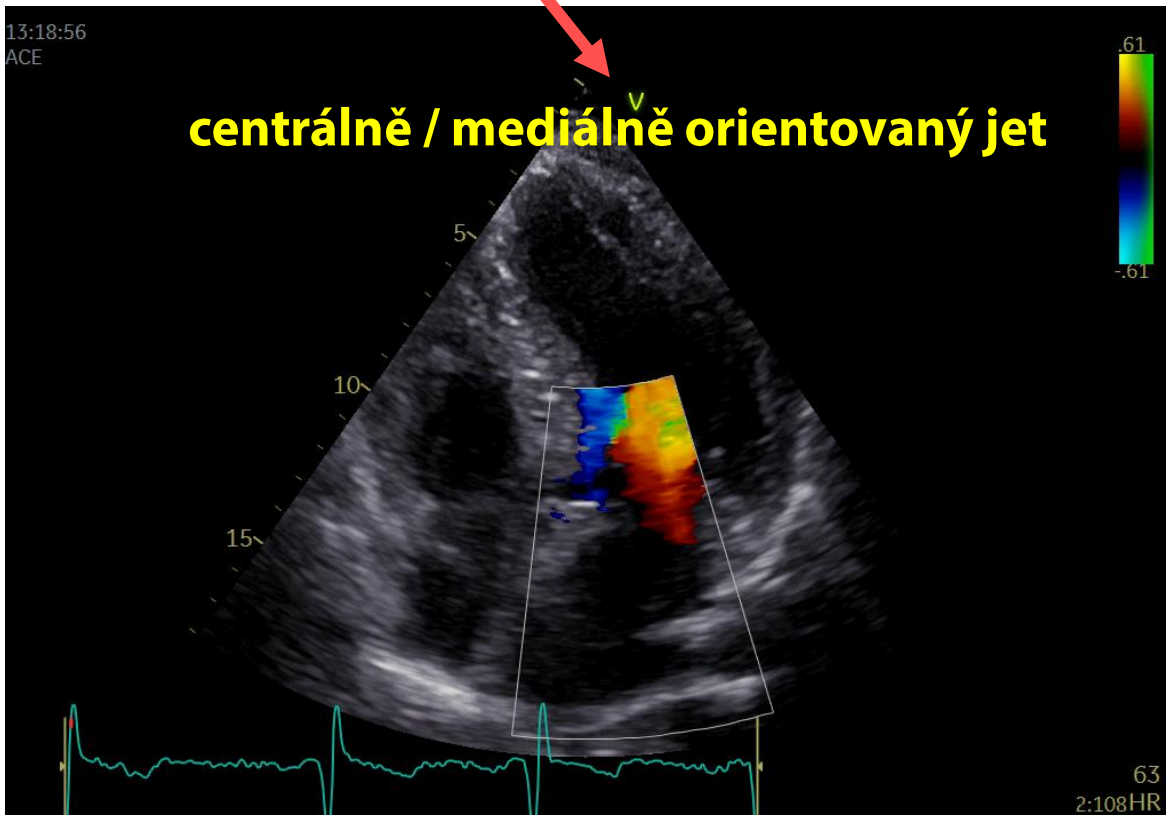


HKMP: echokg & mitrální regurgitace

v důsledku LVOTO mechanismem SAM vs. strukturální změny chlopně

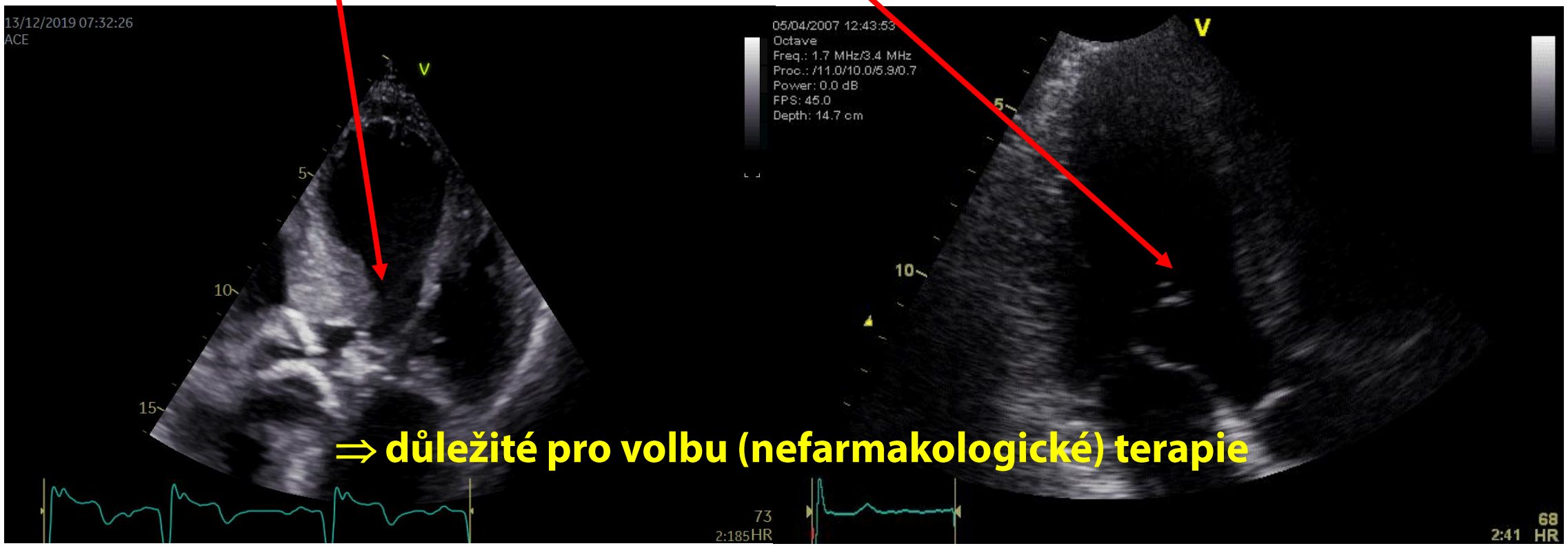
⇒ důležité pro volbu (nefarmakologické) terapie





HKMP: echokg & abnormality mitrálního aparátu

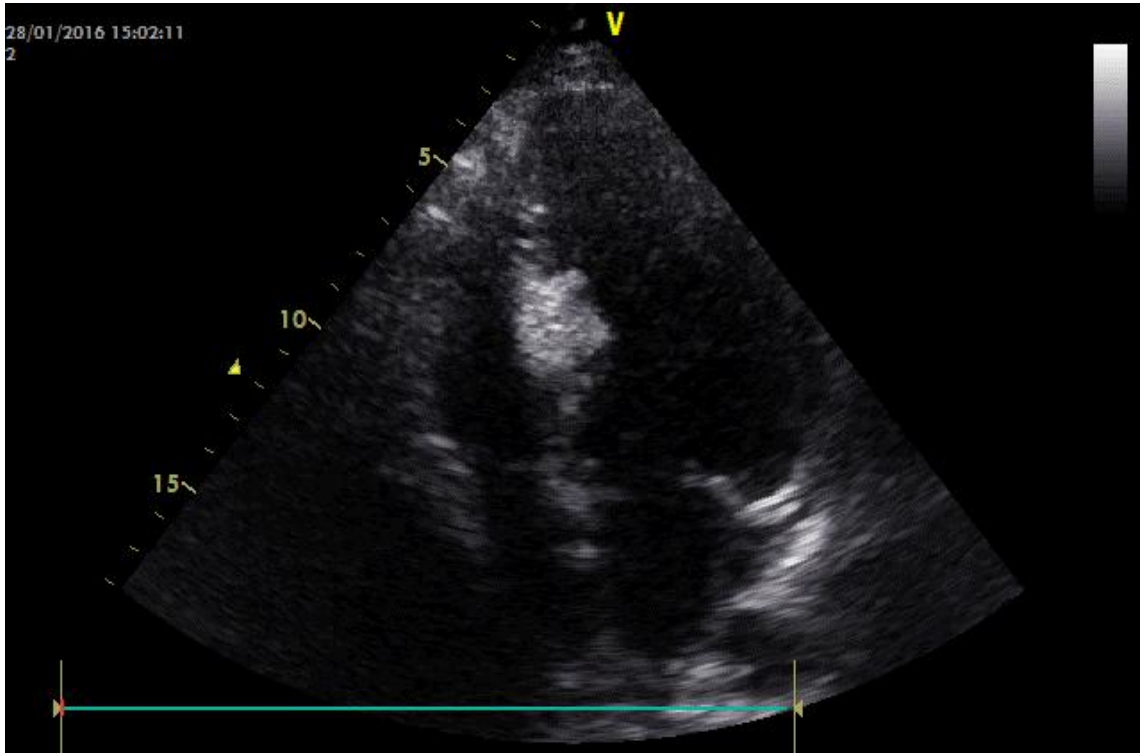
anteriorní / apikální posun papilárních svalů, bifidní papilární sval,
anomální úpon papilárního svalu přímo na přední cíp, hypermobilní papilární svaly,
akcesorní šlašinky

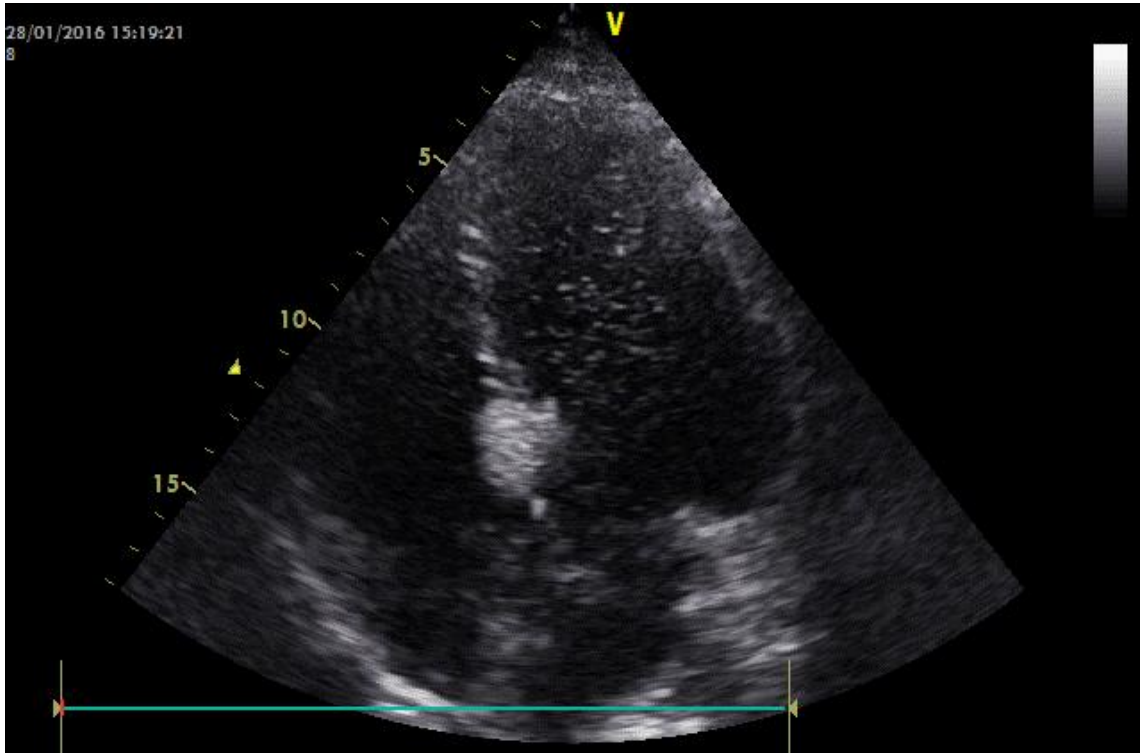


⇒ **důležité pro volbu (nefarmakologické) terapie**

HKMP: echokg & septální redukční terapie

echo-kontrastní látka intrakoronárně:
- lokalizace a velikost oblasti IVS určené k infarzaci
- prevence komplikací: bez opacifikace papilárních svalů, stěny pravé komory

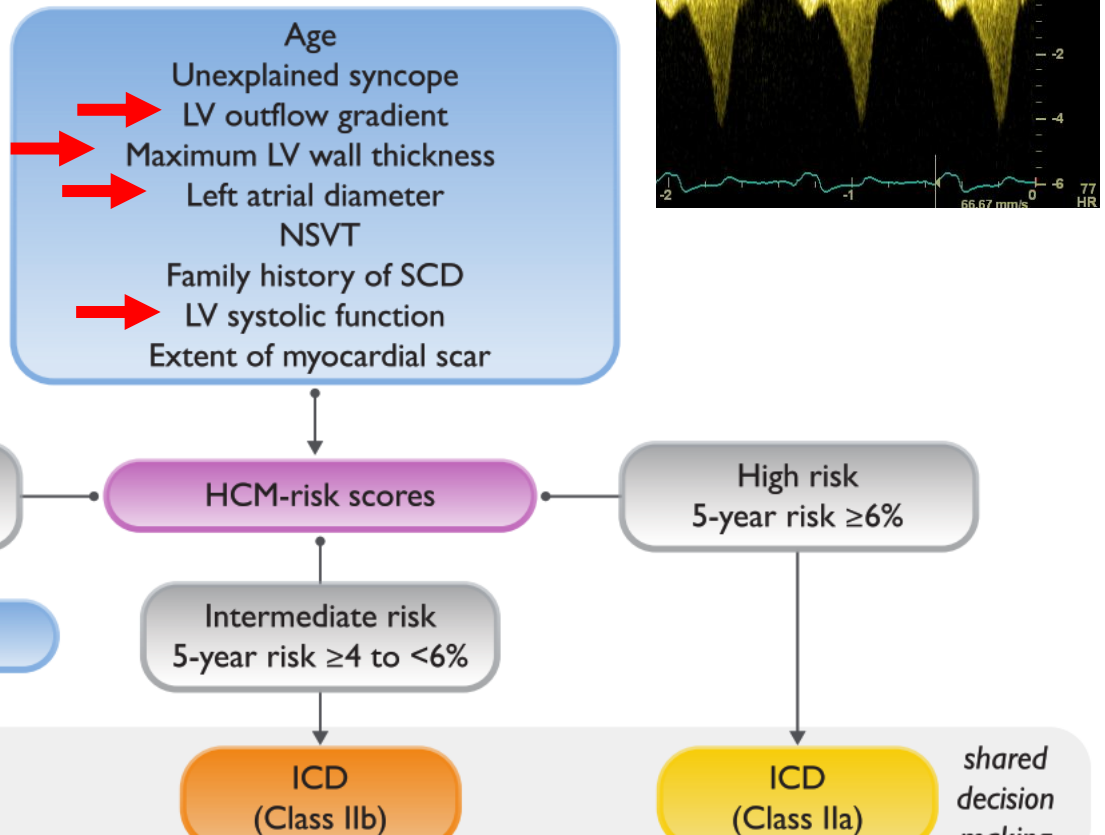


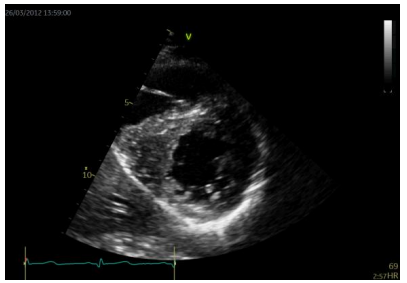


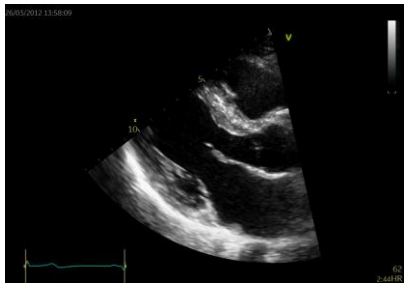
2023 ESC Guidelines for the management of cardiomyopathies

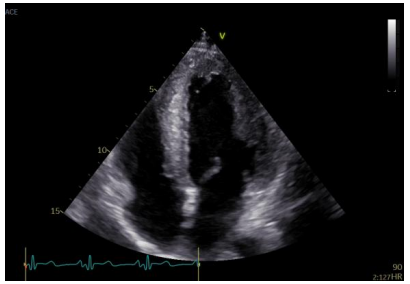
HKMP: echokg & primární prevence SCD

For patients who are in the low-risk category (<4% estimated 5-year risk of SCD), the presence of LVEF <50% may be considered in shared decision-making with patients about prophylactic ICD implantation, acknowledging the lack of robust data on the impact of systolic dysfunction on the personalized risk estimates generated by HCM Risk-SCD or validated paediatric model (e.g. HCM Risk-Kids).[89,315,841–844]	IIb	B









2023 ESC Guidelines for the management of cardiomyopathies

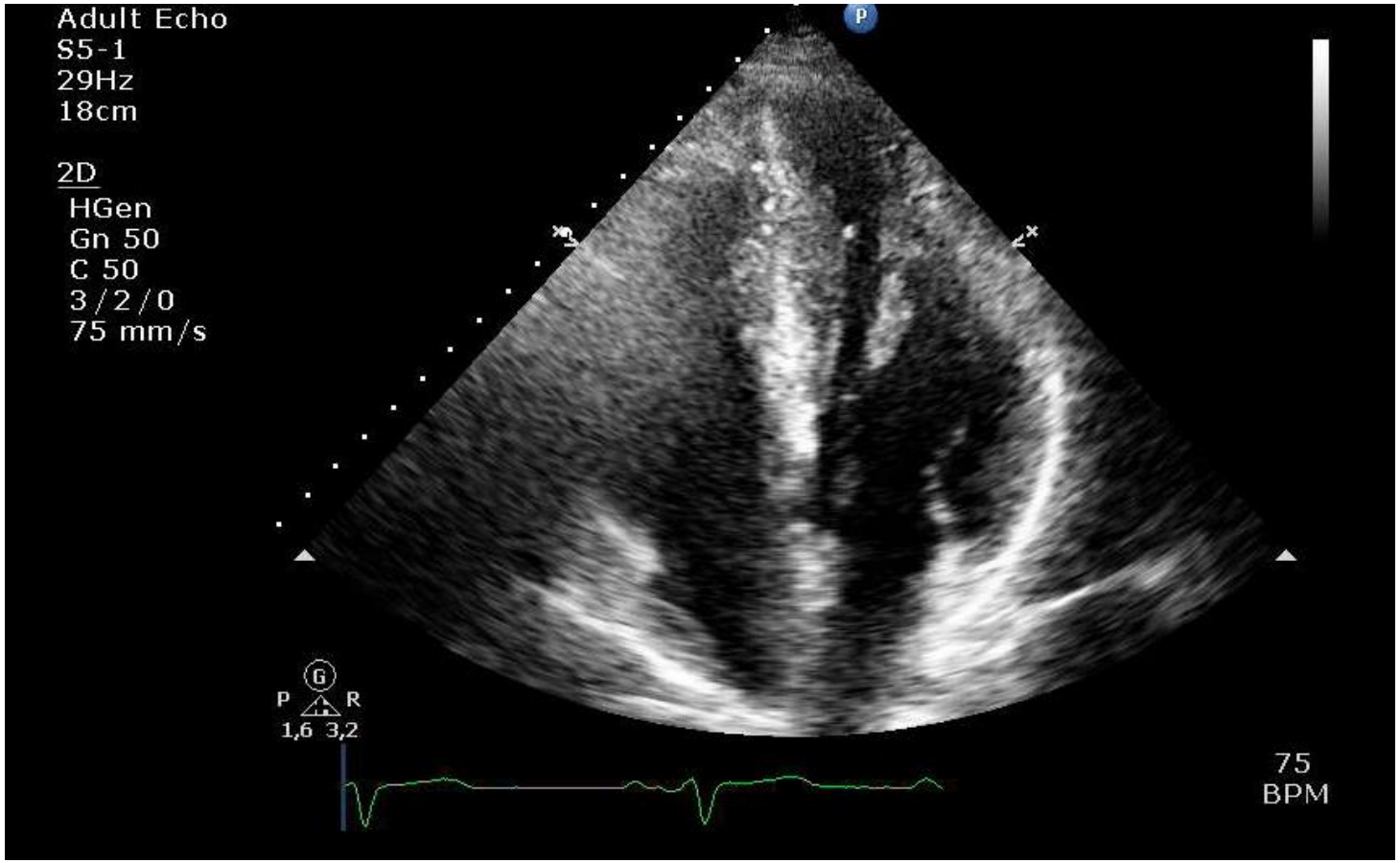
HKMP: echokg & primární prevence SCD

Prognostický význam EF LK < 50%

Events at Any Time	Total (n=6793), n (%)	HCM Without LVSD (n=6240), n (%)	HCM-LVSD (n=553), n (%)	Adjusted HR (95% CI)*	*P* Value
NYHA class III/IV	1121 (16.5)	922 (14.8)	199 (37.3)	2.0 (1.7–2.4)	<0.001
All-cause death	554 (8.2)	416 (6.7)	138 (25.0)	1.8 (1.5–2.2)	<0.001
Cardiac transplantation	110 (1.6)	46 (0.7)	64 (11.6)	11.0 (7.5–16.2)	<0.001
LVAD implantation	15 (0.3)	4 (0.1)	11 (2.0)	26.5 (8.2–85.7)	<0.001
Sudden cardiac death	90 (1.4)	70 (1.2)	20 (3.9)	3.9 (2.4–6.3)	<0.001
ICD implantation	1646 (24.2)	1345 (21.6)	301 (54.4)	2.2 (1.9–2.5)	<0.001
Appropriate ICD therapy	239 (14.5)	163 (12.1)	76 (25.2)	1.6 (1.4–1.8)	<0.001
Atrial fibrillation	1494 (23.2)	1229 (20.9)	265 (49.3)	2.1 (1.8–2.4)	<0.001
Stroke	116 (2.8)	88 (2.3)	28 (8.4)	2.5 (1.9–3.3)	<0.001

SHaRe Registry, Marstrand P et al., Circulation 2020;121: 1371

HKMP: echokg & aneuryzma hrotu levé komory



HKMP: aneuryzma hrotu levé komory

Prognostický význam aneuryzmatu hrotu LK pro primární prevenci SCD shledán nedostatečným

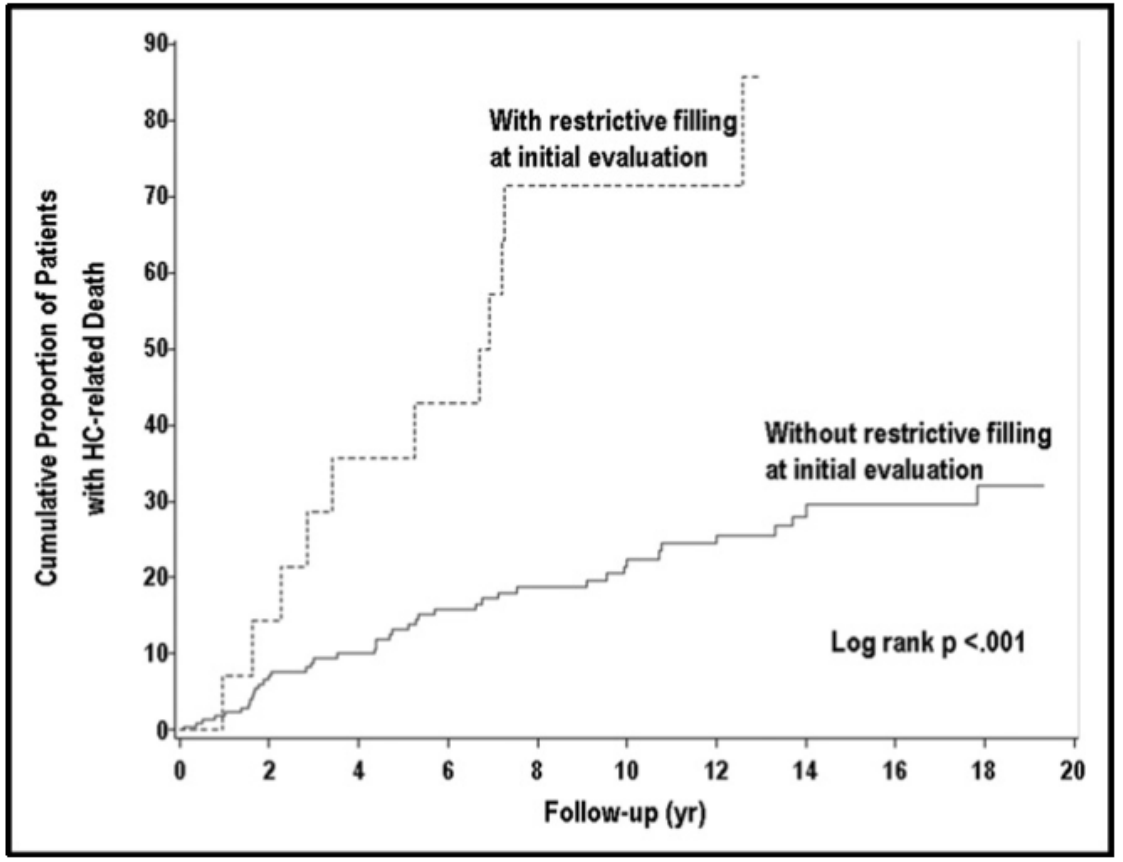
Study name or first author	Details and quality of evidence			Summary of key findings		
	Study type	Number of patients	Key inclusion and	Relevant outcome(s)	Key findings	Conclusion(s)
Rowin *et al.*, 2017[80]					vent rate in t. int 21/93 /93 pts) 2/93 pts) erapy hythm T in 89%	**Conclusion:** HCM patients with LV apical aneurysms are at high risk of arrhythmic sudden death **Limitations:** selection bias, high prevalence of confounders in patients with events (previous VT/VF and LV systolic function)
Lee *et al.*, 2022 [81]					vent rate int 14/ l: 7/160 rapy 4.4% (7/160 pts). Rhythm not specified	**Conclusion:** LV apical aneurysms in HCM are a high-risk phenotype, associated with increased risk of adverse cardiovascular events, including malignant ventricular arrhythmias **Limitations:** selection bias, high prevalence of confounders in patients with events (previous VT/VF and LV systolic function)

Table S2). All these studies were retrospective and the absolute number of events is too small to assess the independent predictive value of apical aneurysms. In two small series that described a selected subgroup of HCM patients with mid-ventricular obstruction, there was no increase in incidence of SCD events. In the only series that provides a detailed analysis of SCD events, the majority were appropriate ICD interventions for monomorphic VT, suggesting significant inclusion bias.[737] Finally, a large proportion of individuals with events had other important risk markers including prior sustained ventricular arrhythmia. Based on the current data, the Task Force recommends that individualized ICD decisions should be based using well-established risk factors and not solely on the presence of an LV apical aneurysm.

2023 ESC Guidelines for the management of cardiomyopathies

HKMP: echokg & diastolická dysfunkce LK

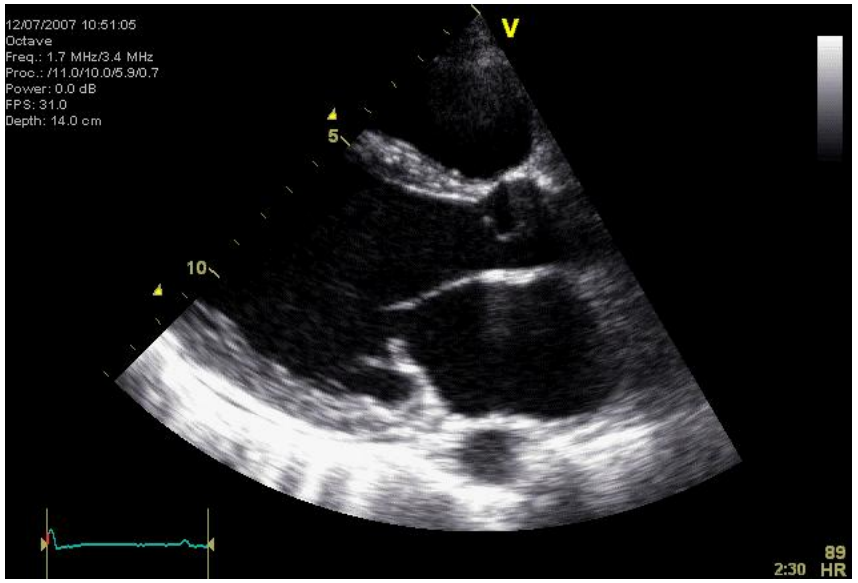
Prognostický význam restriktivního plnění LK

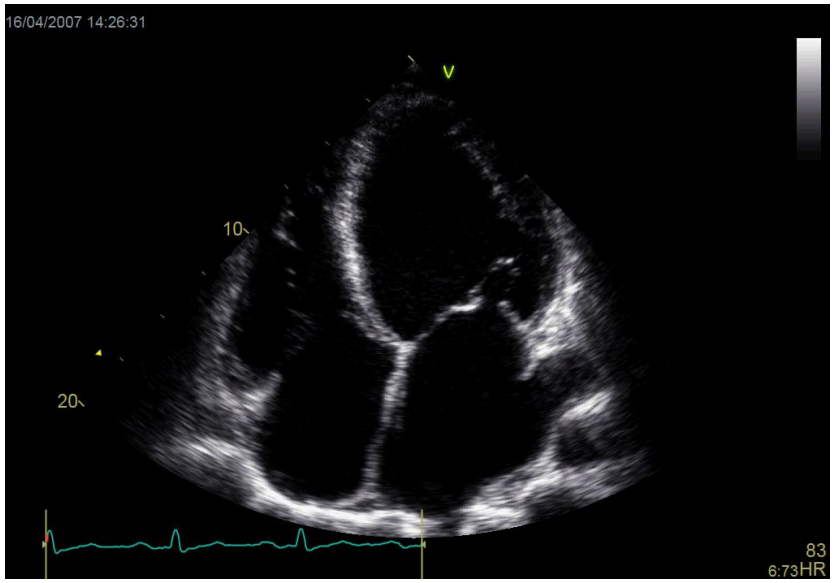


Biagini E et al., Am J Card 2009;104:1727

Dilatační kardiomyopatie & echokg

Dilated cardiomyopathy is defined by the presence of LV dilatation and systolic dysfunction unexplained solely by abnormal loading conditions or CAD. Left ventricular dilatation is defined by LV end-diastolic dimensions or volumes >2 z-scores above population mean values corrected for body size, sex, and/or age. For adults this represents an LV end-diastolic diameter >58 mm in males and >52 in females and an LVEDV index of ≥75 mL/m^2 in males and ≥62 mL/m^2 in females by ECHO.[9,845,846] Left ventricular global systolic dysfunction is defined by LVEF <50%.[9]

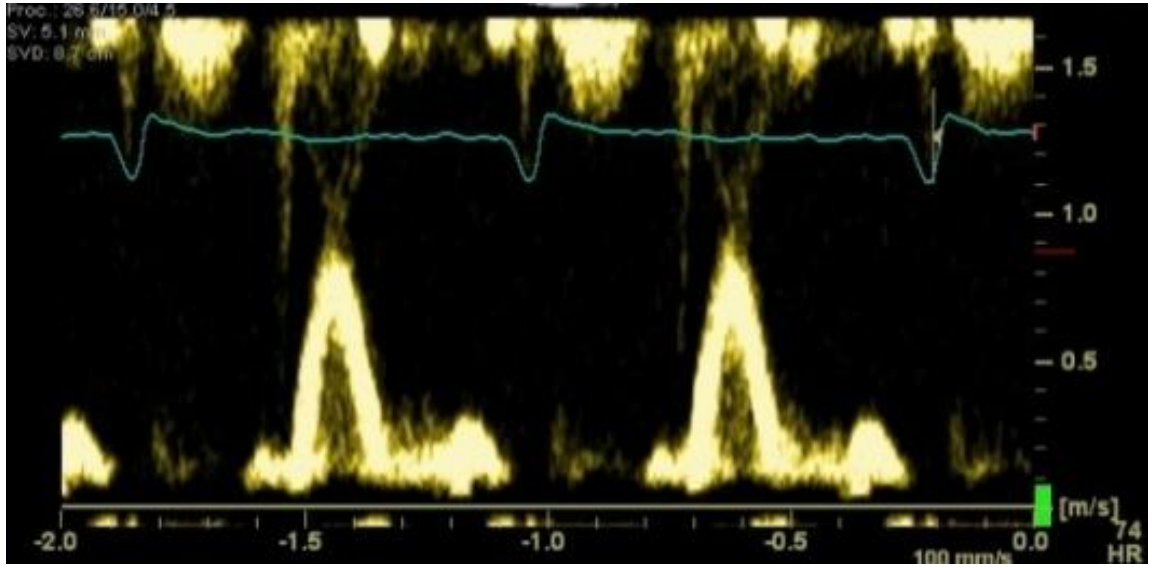


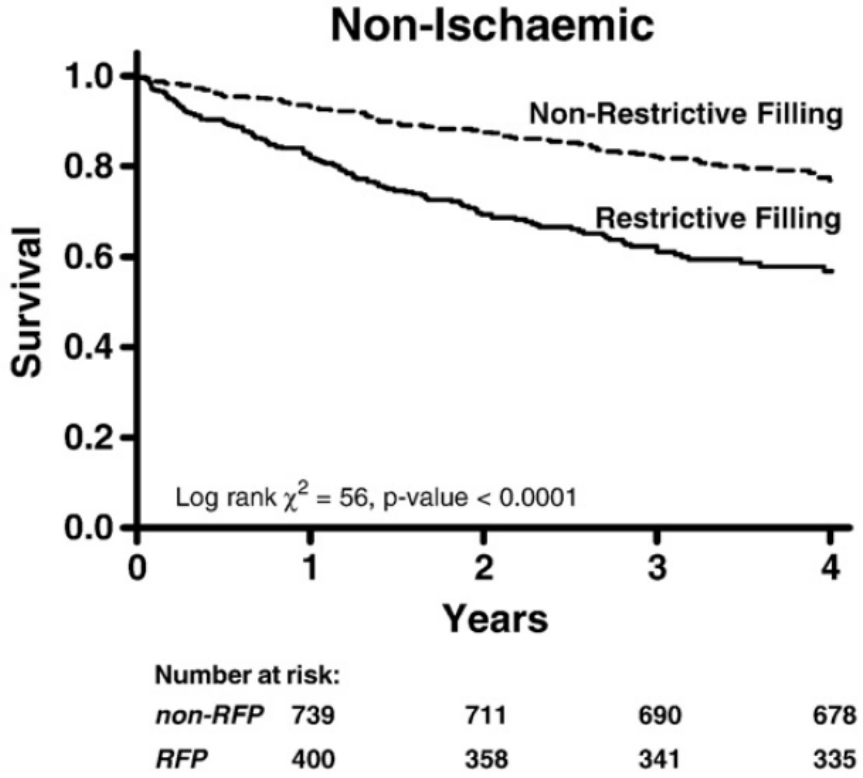


2023 ESC Guidelines for the management of cardiomyopathies

Dilatační kardiomyopatie & echokg

Prognostický význam restriktivního plnění LK





MeRGE collaborators, Eur J HF 2008;10:786

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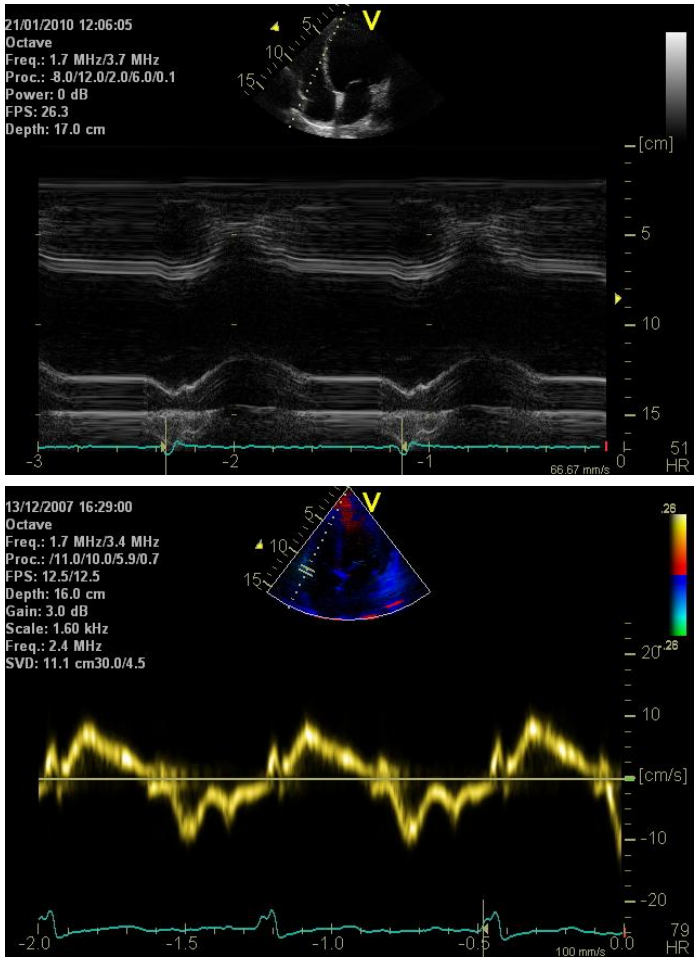
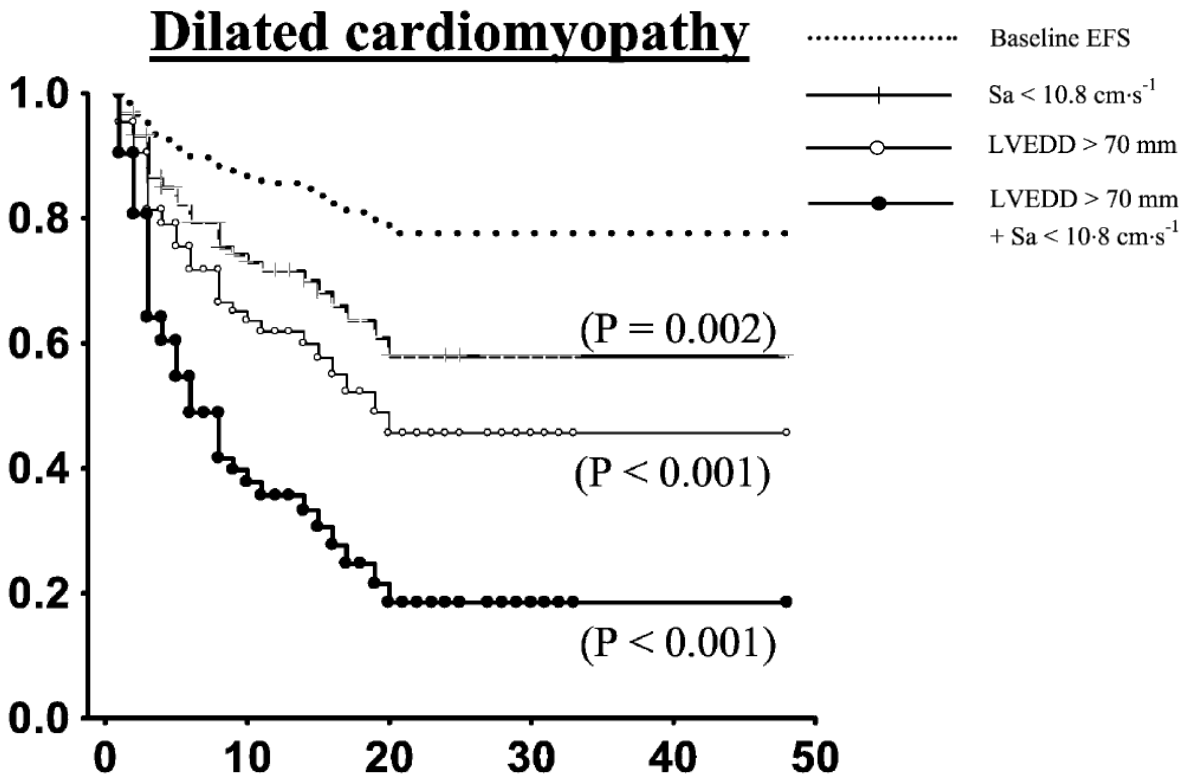
Dilatační kardiomyopatie & echokg

Prognostický význam systolické funkce PK





Meluzín J et al., EJE 2003;4:262

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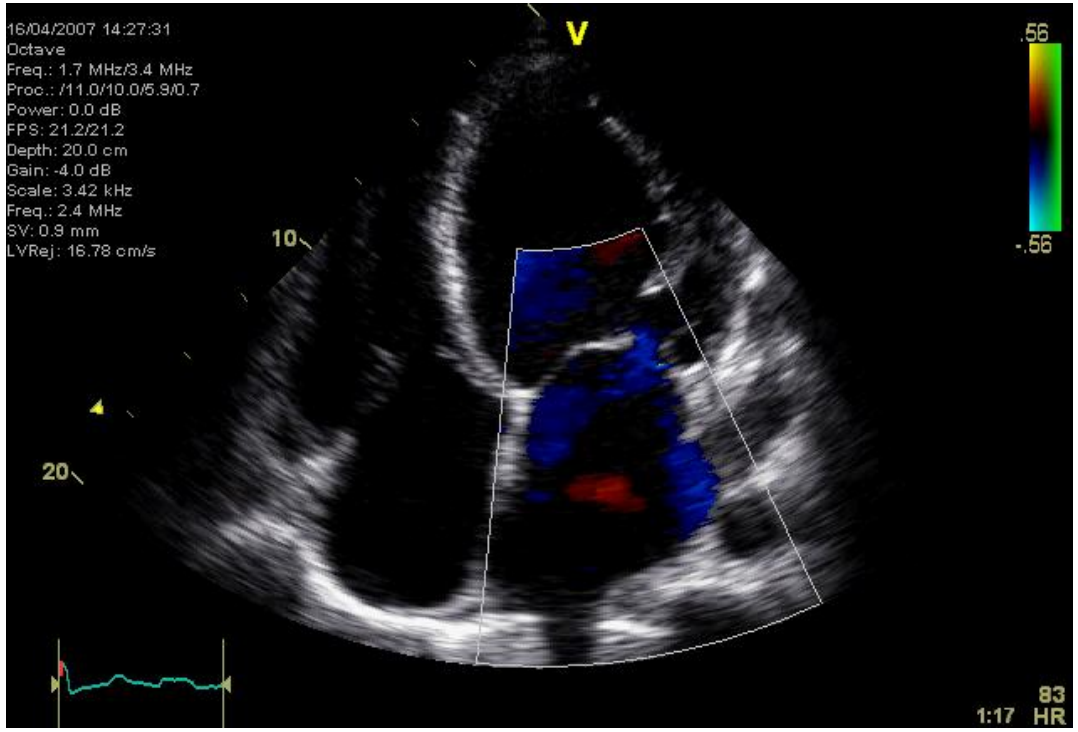
Dilatační kardiomyopatie & echokg

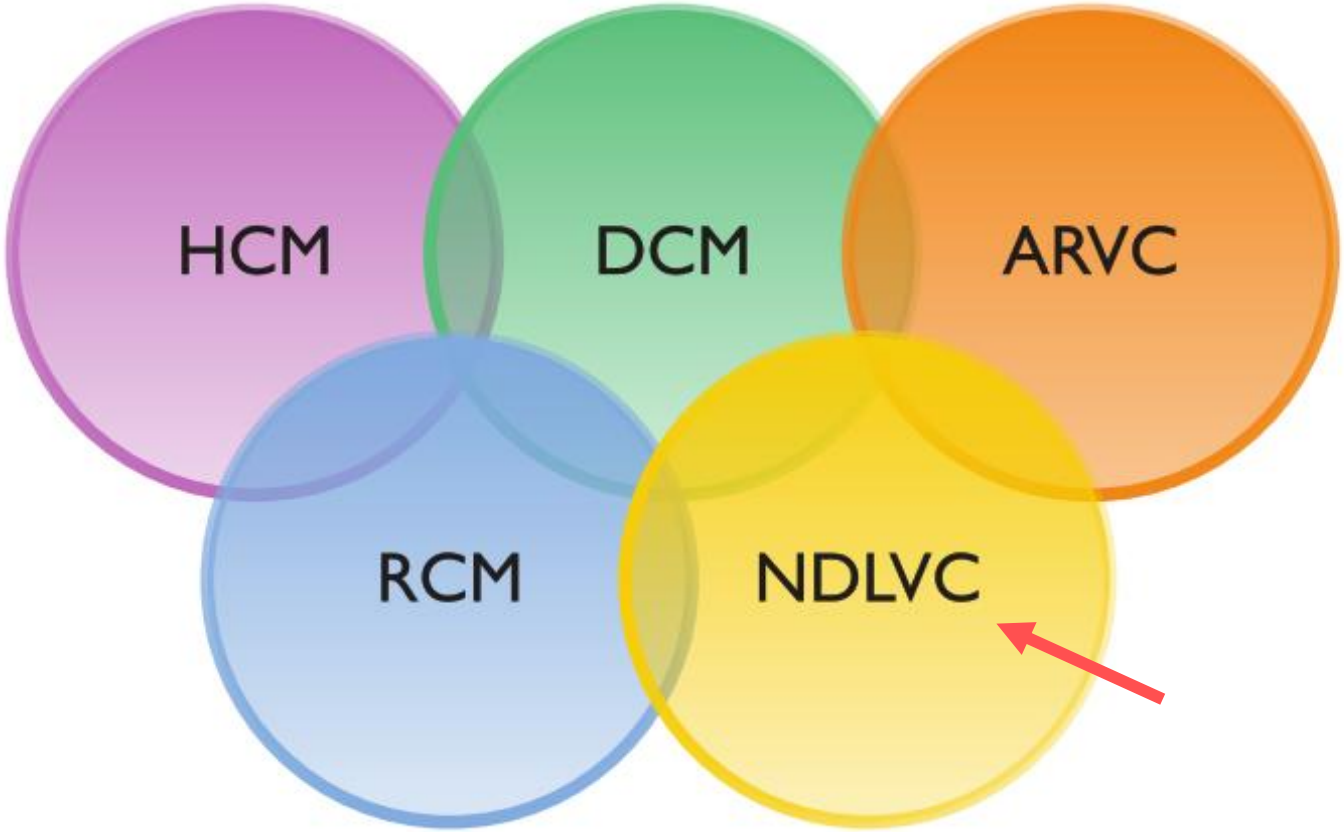
Funkční mitrální (a trikuspidální) regurgitace



- vyloučení primární MR jakožto příčiny dilatace/dysfunkce LK
  - vhodnost k perkutánní intervenční léčbě (M-TEE, T-TEER)

2023 ESC Guidelines for the management of cardiomyopathies

Nedilatovaná kardiomyopatie levé komory & echokg

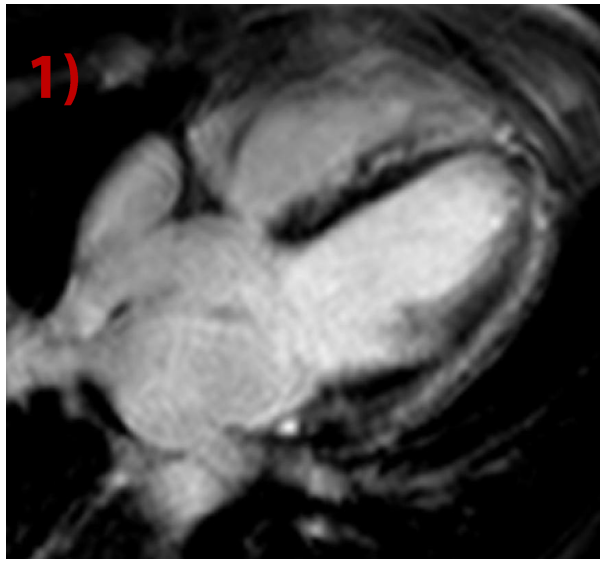


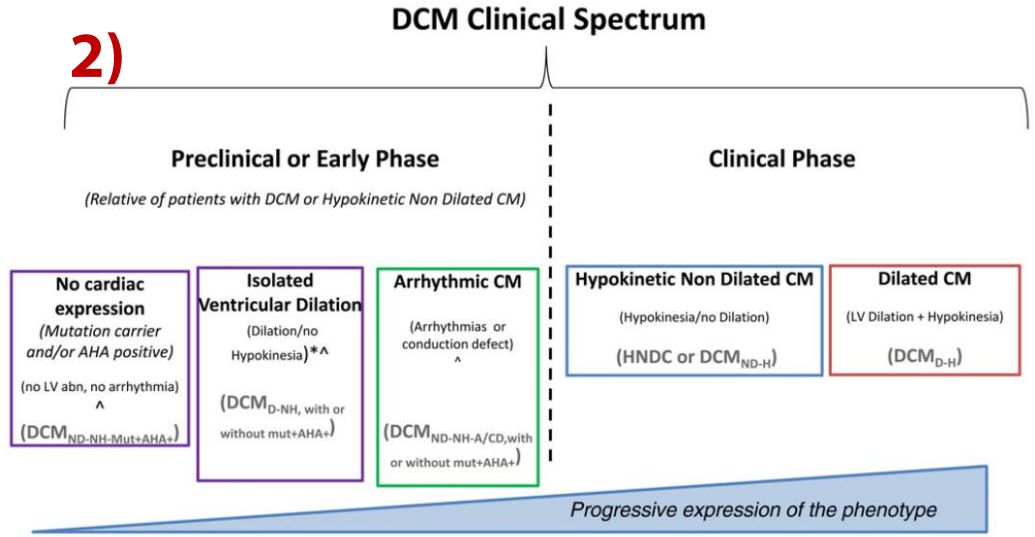
2023 ESC Guidelines for the management of cardiomyopathies

Nedilatovaná kardiomyopatie levé komory & echokg

1) přítomnost neischemické fibrózy/jizvy či tukové náhrady myokardu LK, bez ohledu na přítomnost globální či regionální systolické dysfunkce LK

2) izolovaná difuzní hypokineza LK bez přítomnosti fibrózy/jizvy





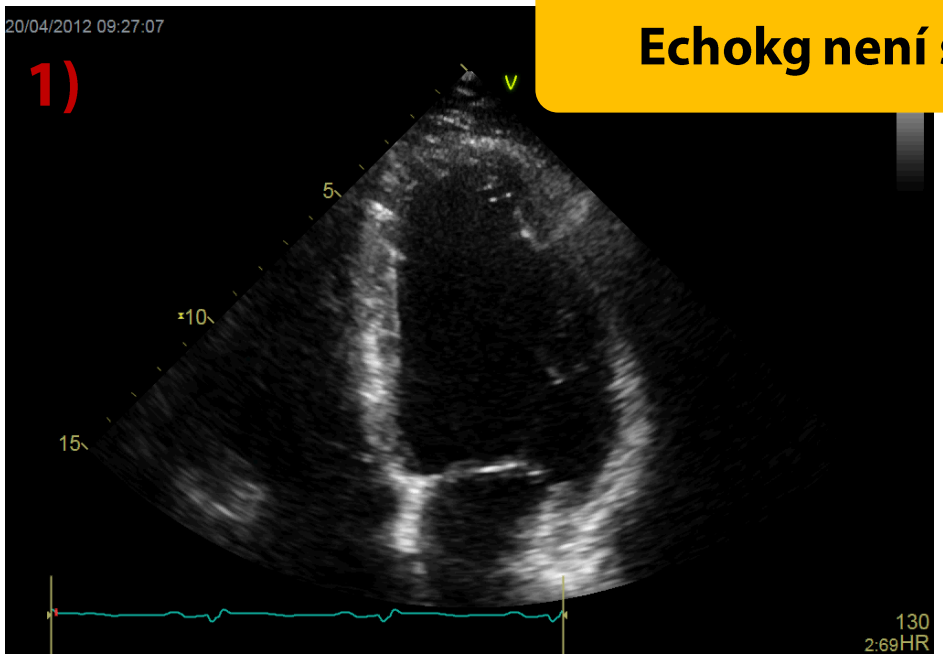
2023 ESC Guidelines for the management of cardiomyopathies

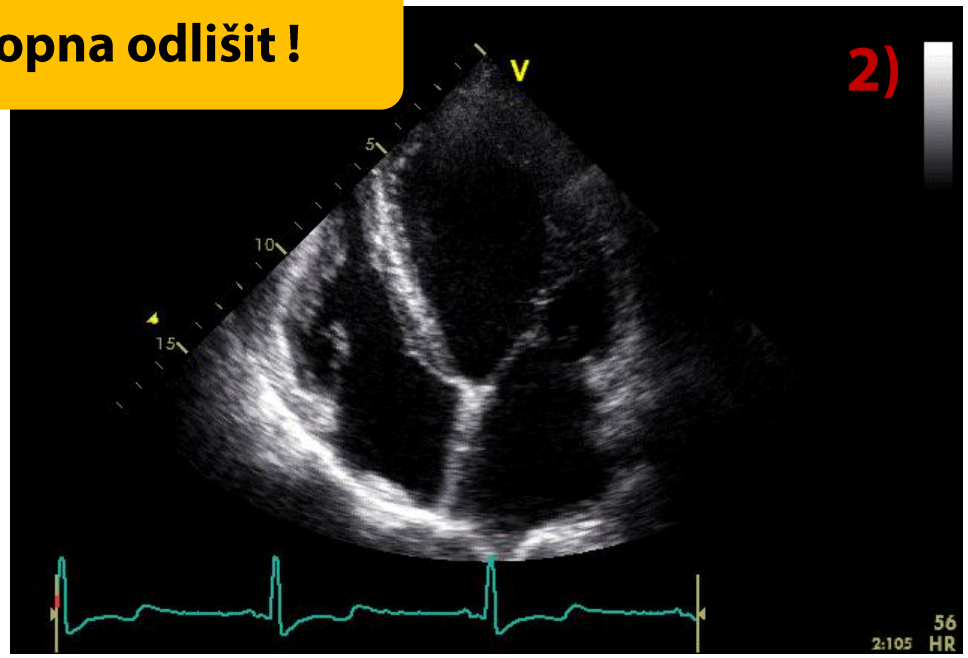
Nedilatovaná kardiomyopatie levé komory & echokg

1) přítomnost neischemické fibrózy/jizvy či tukové náhrady myokardu LK, bez ohledu na přítomnost globální či regionální systolické dysfunkce LK

2) izolovaná difuzní hypokineza LK bez přítomnosti fibrózy/jizvy

Echokg není schopna odlišit !





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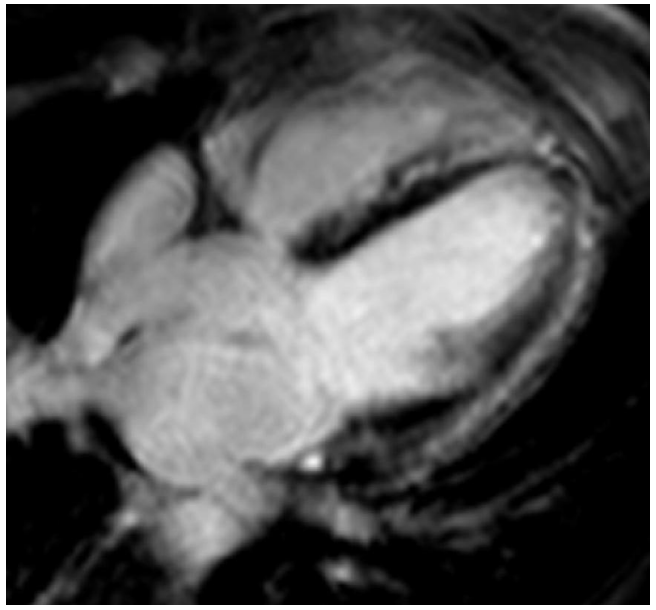
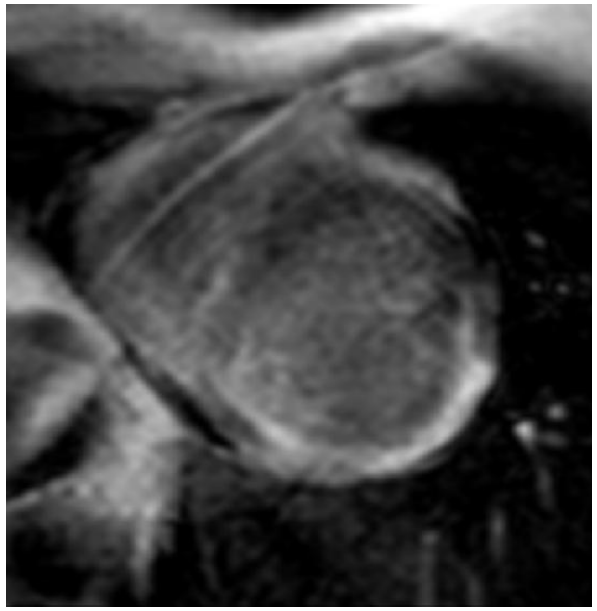
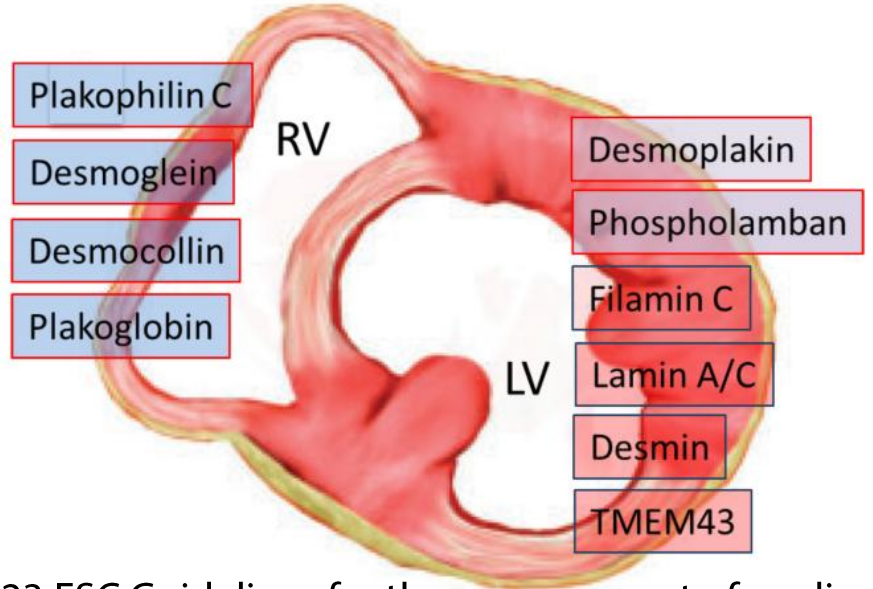
Arytmogenní kardiomyopatie LK ??

MRI-LGE





genetika

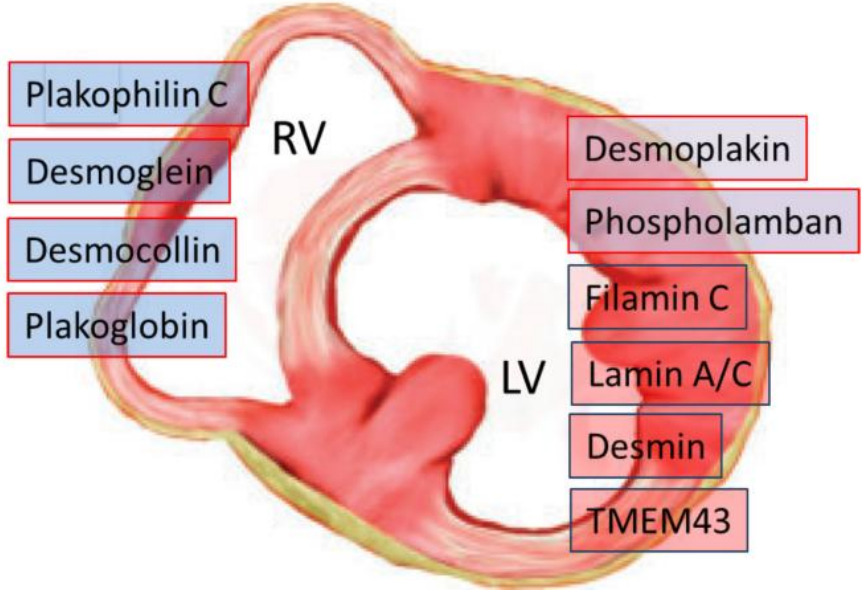


2023 ESC Guidelines for the management of cardiomyopathies

Arytmogenní kardiomyopatie: definice, Padovská kritéria 2020

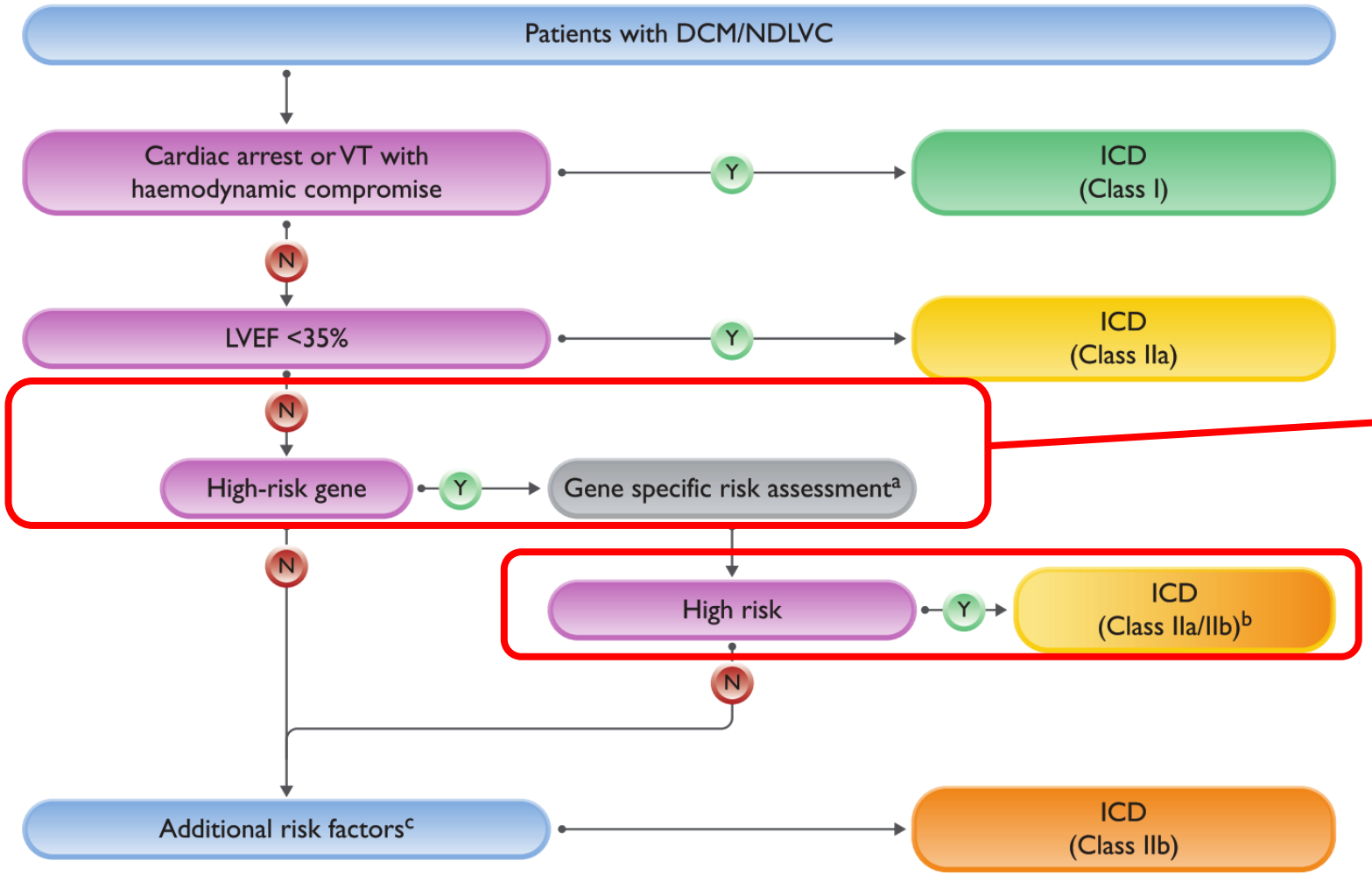
- **geneticky podmíněné** onemocnění srdečního svalu PK / LK / obou komor
- charakterizované **náhradou myokardu fibrózní / fibrolipomatózní tkání**
- **predisponující k potenciálně letálním komorovým arytmiím** bez ohledu na systolickou funkci komory

mutace v genech kódující desmosomální a non-desmosmální proteiny



Corrado D et al., Int J Card 2020; 319

Echokg & prevence SCD u NDLVC a DKMP



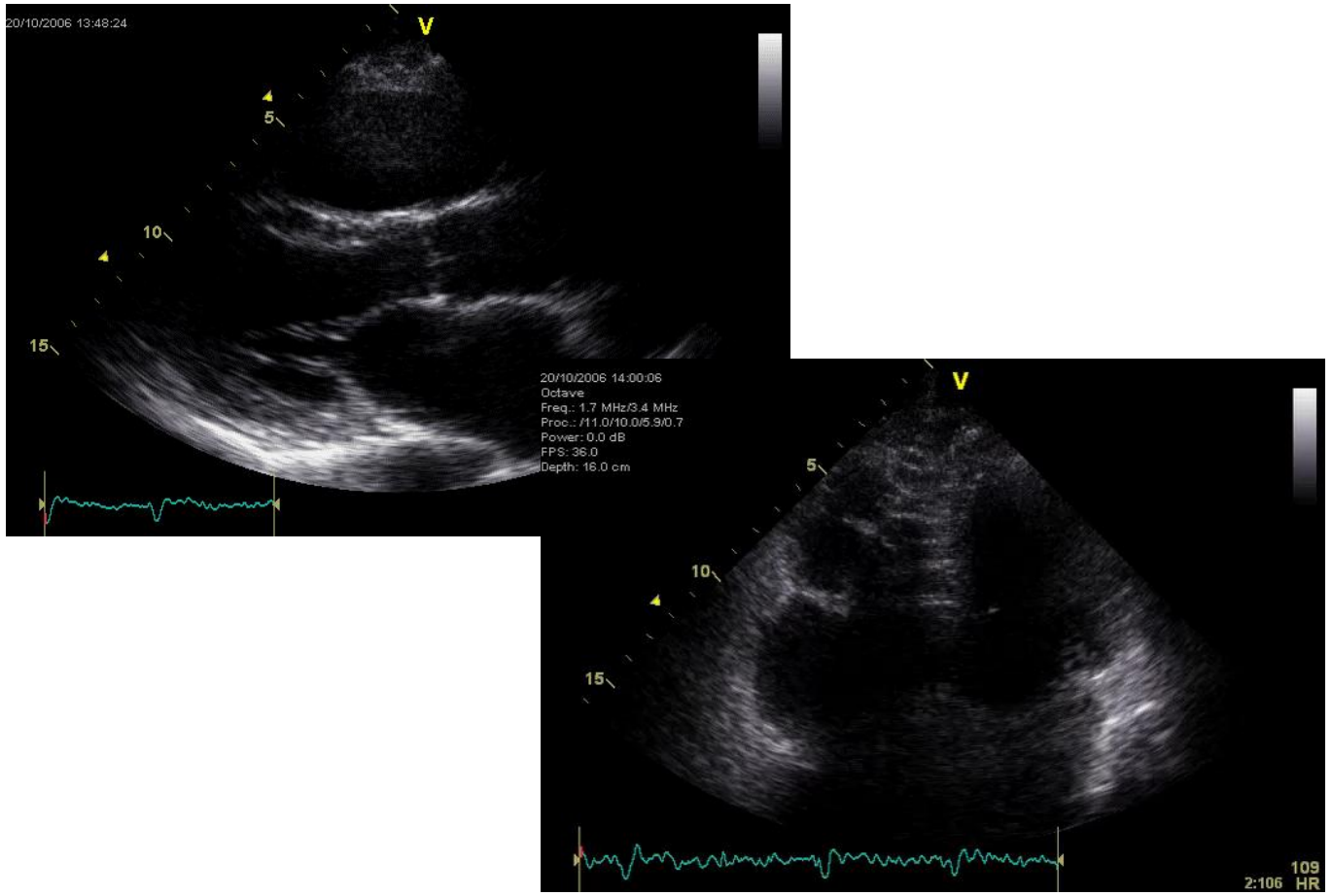
Gene	Annual SCD rate	Predictors of SCD
LMNA[185,186,438,541,865,878,879]	5–10%	Estimated 5-year risk of life-threatening arrhythmia using *LMNA* risk score (https://lmna-risk-vta.fr)
FLNC-truncating variants[866,867,880]	5–10%	LGE on CMR LVEF < 45%
TMEM43[868,881]	5–10%	Male Female and any of the following: LVEF <45%, NSVT, LGE on CMR, >200 VE on 24h Holter ECG
PLN[542,882,883]	3–5%	Estimated 5-year risk of life-threatening arrhythmia using *PLN* risk score (https://plnriskcalculator.shinyapps.io/final_shiny) LVEF < 45% LGE on CMR NSVT
DSP[185,186]	3–5%	LGE on CMR LVEF < 45%
RBM20[869]	3–5%	LGE on CMR LVEF < 45%

2023 ESC Guidelines for the management of cardiomyopathies

Arytmogenní kardiomyopatie PK & echokg

regionální akineze / dyskineze / aneuryzma PK (! ne hypokineza)

dilatace PK, snížená FAC PK



Major

By 2D echo:

- Regional RV akinesia, dyskinesia, or aneurysm
- *and* 1 of the following (end diastole):
  - PLAX RVOT ≥32 mm (corrected for body size [PLAX/BSA] ≥19 mm/m^2)
  - PSAX RVOT ≥36 mm (corrected for body size [PSAX/BSA] ≥21 mm/m^2)
  - *or* fractional area change ≤33%

Minor

- Regional RV akinesia or dyskinesia
- *and* 1 of the following (end diastole):
  - PLAX RVOT ≥29 to <32 mm (corrected for body size [PLAX/BSA] ≥16 to <19 mm/m^2)
  - PSAX RVOT ≥32 to <36 mm (corrected for body size [PSAX/BSA] ≥18 to <21 mm/m^2)
  - *or* fractional area change >33% to ≤40%

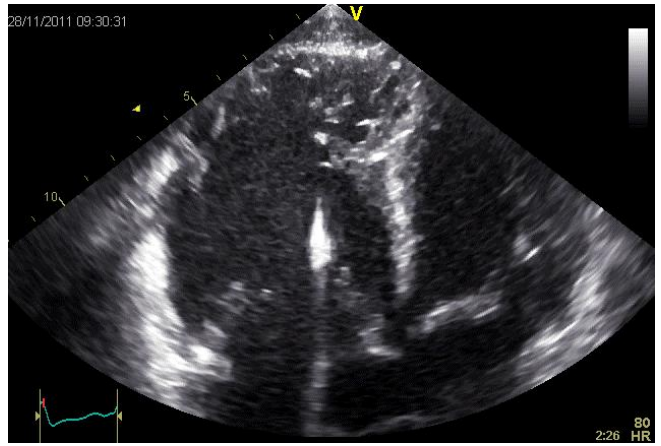
Marcus FI et al., Circulation 2010;121

Echokg & prevence SCD u arytmogenní kardiomyopatie PK

Recommendations	Class[a]	Level[b]
Secondary prevention		
An ICD is recommended to reduce the risk of sudden death and all-cause mortality in patients with ARVC who have survived a cardiac arrest or have recovered from a ventricular arrhythmia causing haemodynamic instability.[939,943,944,948,949]	I	A
An ICD should be considered in ARVC patients who have suffered a haemodynamically tolerated VT.[522,939,943–945,948–950]	IIa	B
Primary prevention		
High-risk features[c] should be considered to aid individualized decision-making for ICD implantation in patients with ARVC.[538,939]	IIa	B
The updated 2019 ARVC risk calculator should be considered to aid individualized decision-making for ICD implantation in patients with ARVC.[d,524,526,536–539]	IIa	B

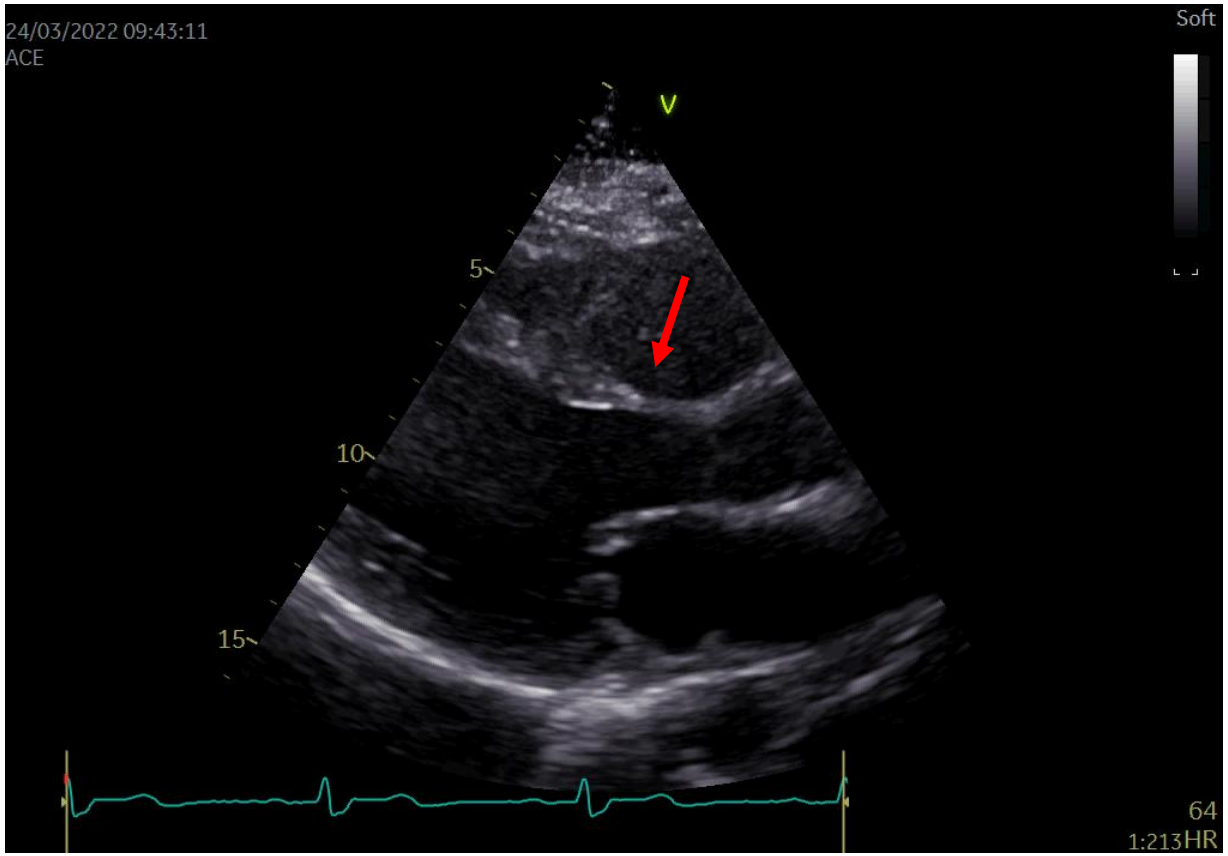
Známky vyššího rizika SCD:

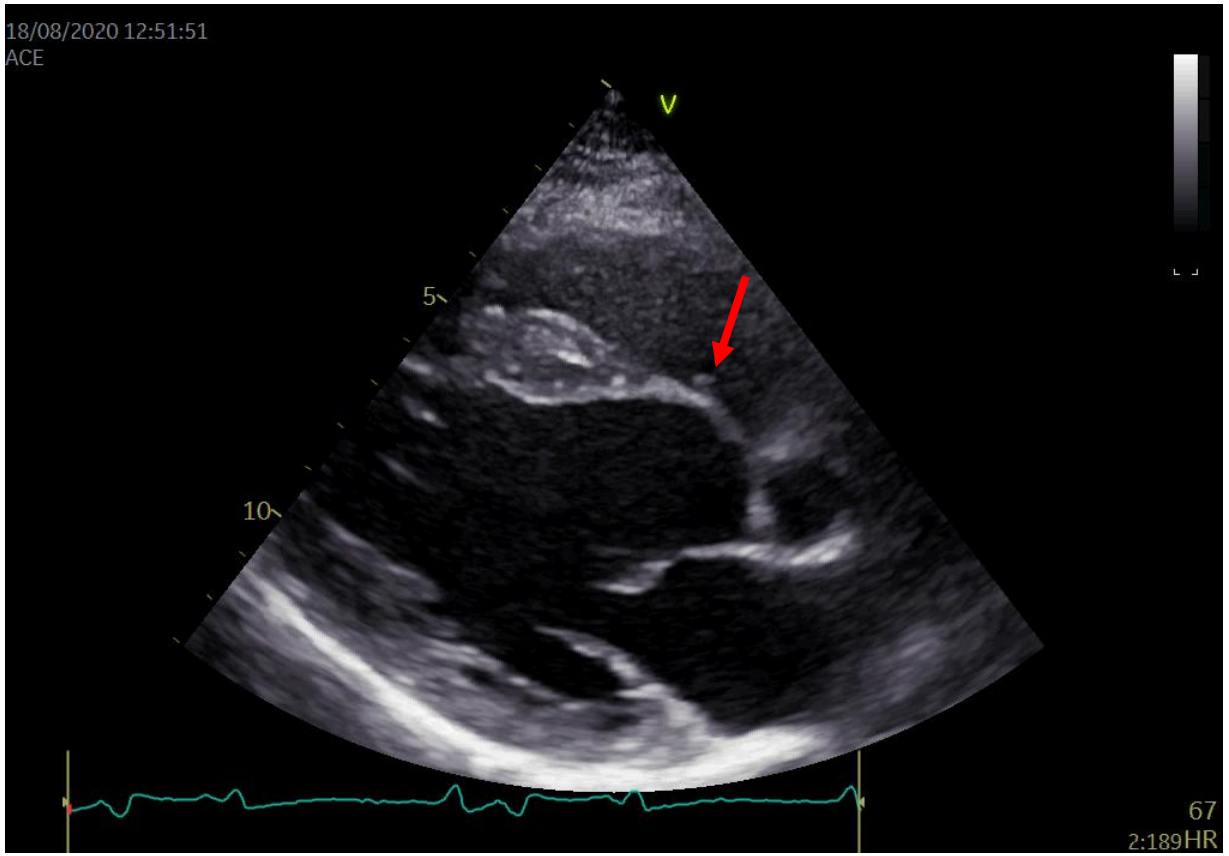
- arytmická synkopa
- nesetrvalé KT
- **EF pravé komory < 40%**
- **EF levé komory < 45%**
- setrvalá mKT vyvolaná při PSK



2023 ESC Guidelines for the management of cardiomyopathies

Echokg & sarkoidóza srdce





Restriktivní KMP & echokg

Intrinsic myocyte dysfunction

Genetic

- Primary RCM
- Variants in sarcomeric, cytoskeletal, nuclear envelope, filamin, titin genes

Storage

- Desmin
- AFD
- Danon
- Glycogenoses
- *PRKAG2* variants
- Iron overload/storage disorders

Non-genetic

- Drugs (e.g. chloroquine)

Endomyocardial disorders

- Endomyocardial fibrosis
- Hypereosinophilia
- Carcinoid
- Endocardial fibroelastosis
- Endocardial neoplasms
- Iatrogenic/drug toxicity

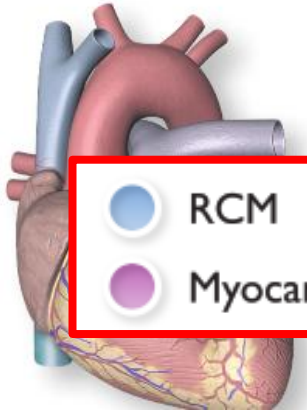
Myocardial extracellular matrix disorders

Infiltrative

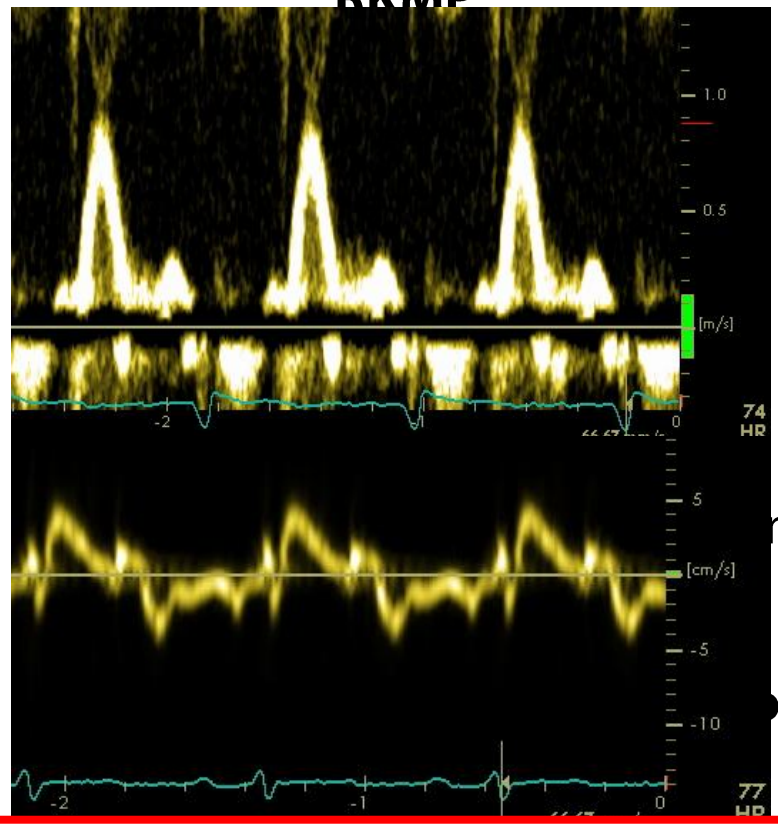
- Hyperoxaluria
- Amyloidosis
- Sarcoidosis

Fibrosis

- Radiation
- Chemotherapy
- Systemic sclerosis
- Inflammatory/granulomatous
- Diabetic heart disease



RKMP



ně)

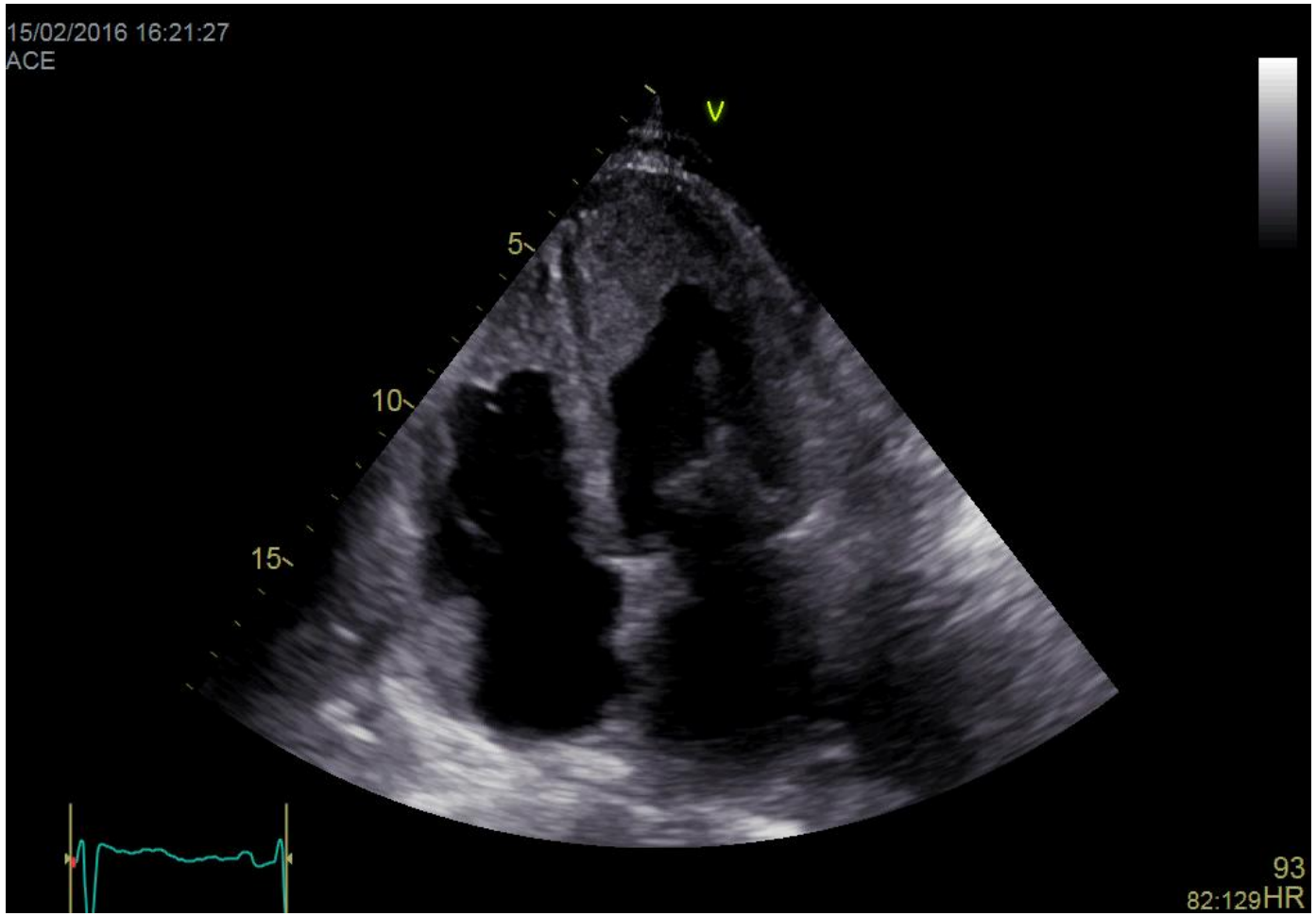
ru:

- RCM
- Myocardial diseases with occasional restrictive physiology, often in the context of LVH

2023 ESC Guidelines for the management of cardiomyopathies

Echokg & RKMP v důsledku patologie endokardu

endomyokardiální fibróza



Echokg & RKMP v důsledku patologie myokardu

myocytární dysfunkce – geneticky podmíněná či získaná (toxicita)
/ extracelulární infiltrace či fibróza/ intracelulární střádání

Tloušťka stěny levé komory

zesílená stěna

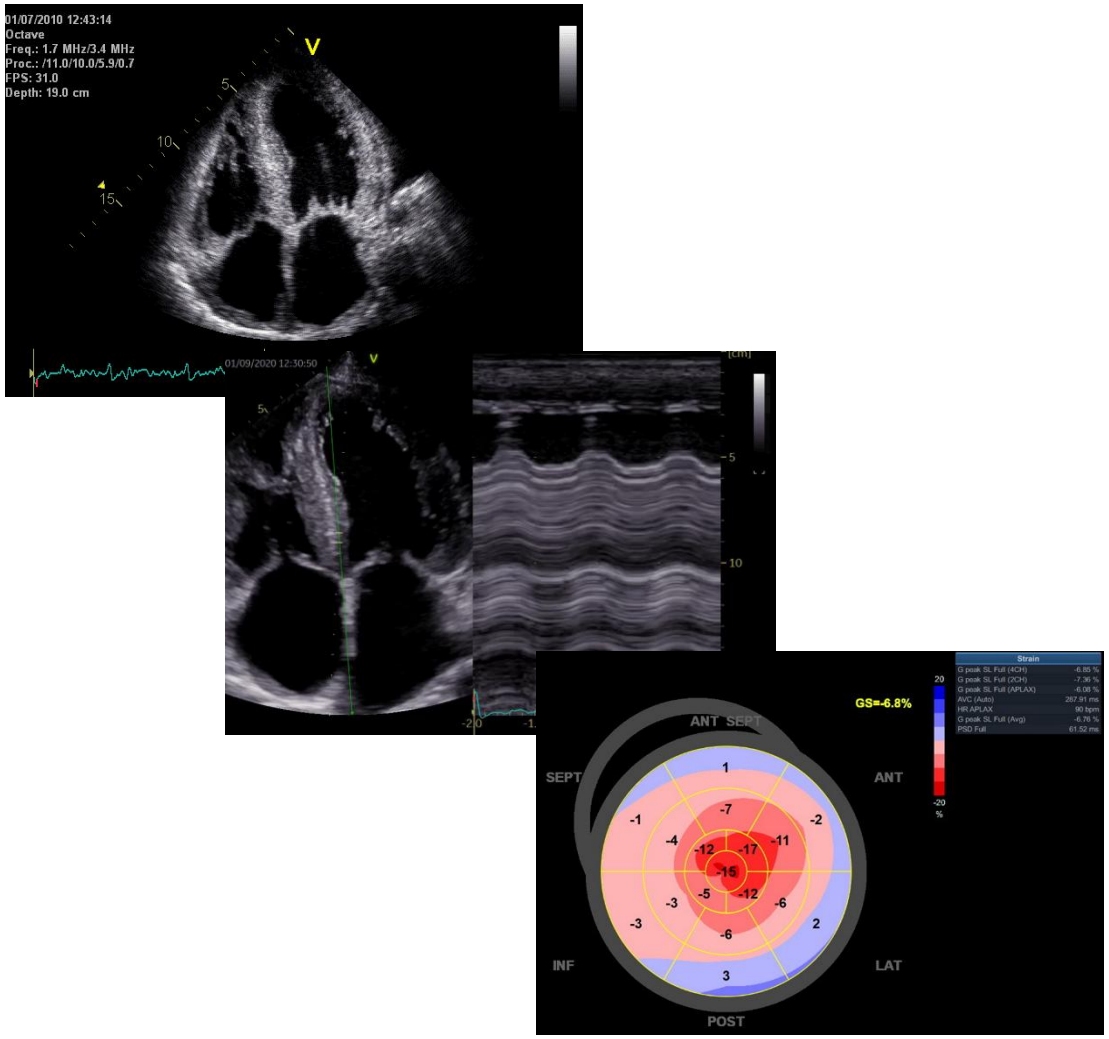
- amyloidóza
- Fabryho choroba
Danonova choroba
- PRKAG2 kardiomyopatie
- glykogenózy
- některé sarkomerické mutace

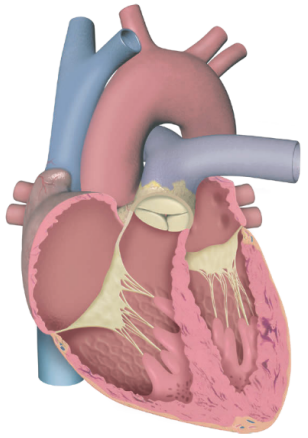
bez zesílení stěny

- idiopatická, resp. geneticky podmíněná RKMP
- kardiotoxická (radiace, chemoterapie)
- přetížení železem (hemochromatóza)
- sklerodermie
- diabetická

Rapezzi C et al., Eur Heart J 2022;43

Echokg & srdeční amyloidóza





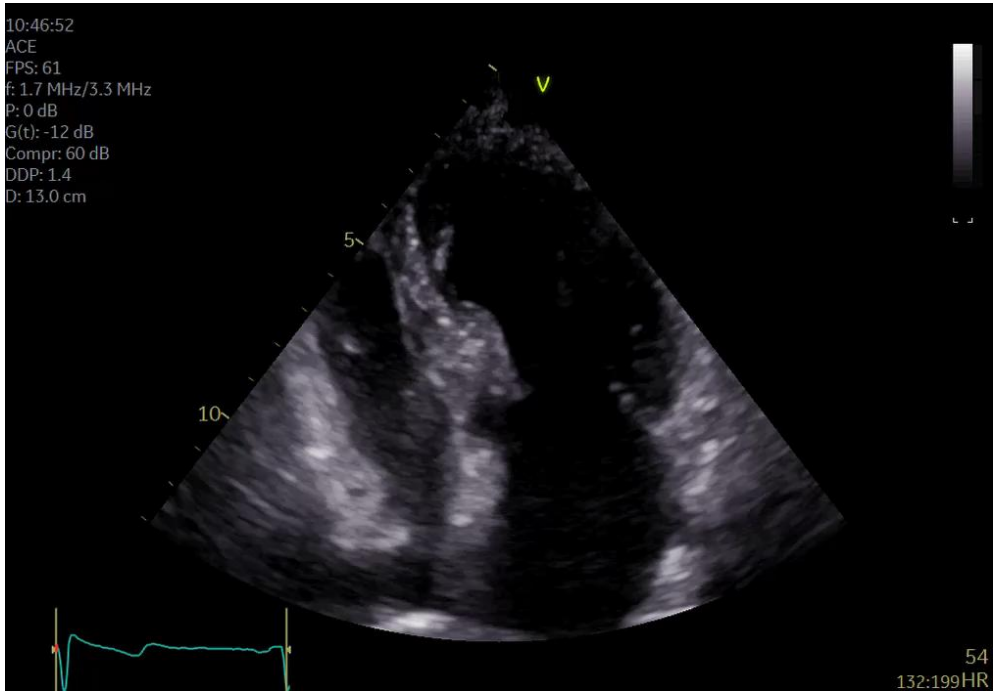
Left ventricular wall thickness ≥12 mm + ≥1 of

- Heart failure in ≥65 years
- Aortic stenosis in ≥65 years
- Hypotension or normotensive if previously hypertensive
- Sensory involvement, autonomic dysfunction
- Peripheral polyneuropathy
- Proteinuria
- Skin bruising
- Ruptured biceps tendon
- Bilateral carpal tunnel syndrome
- Subendocardial/transmural LGE or increased ECV
- Reduced longitudinal strain with apical sparing
- Decreased QRS voltage to mass ratio
- Pseudo Q waves on ECG
- AV conduction disease
- Possible family history of ATTR
- Chronically increased troponin levels
- Known multiple myeloma or MGUS

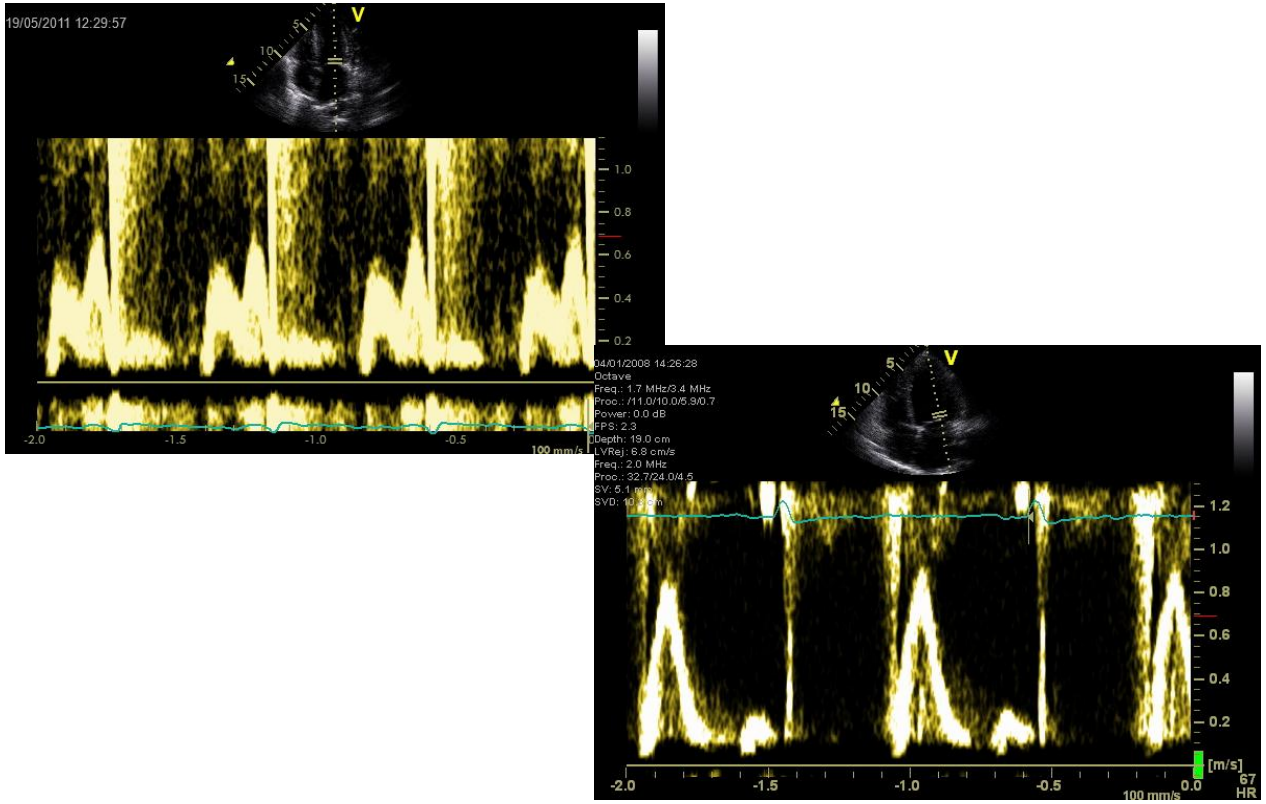
2023 ESC Guidelines for the management of cardiomyopathies

Echokg & srdeční amyloidóza

symetrické i asymetrické (IVS)
zesílení stěn LK



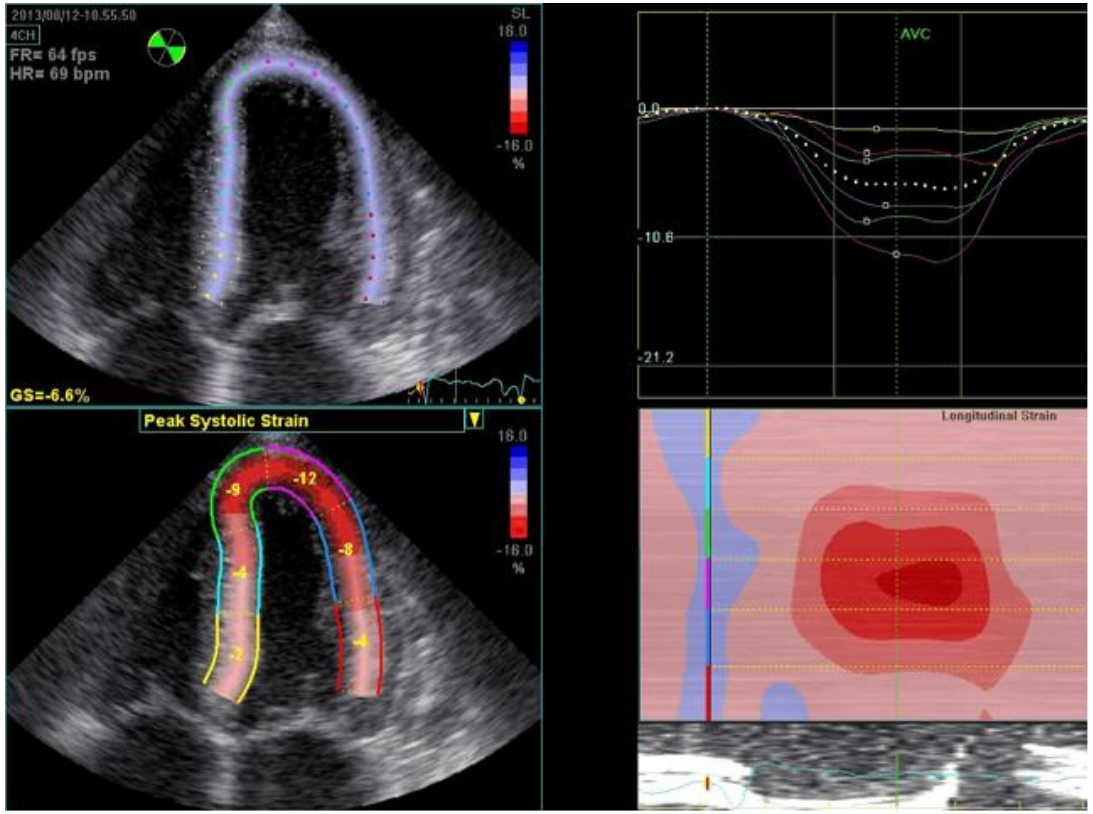
absence restriktivního plnění LK
nevylučuje srdeční amyloidózu



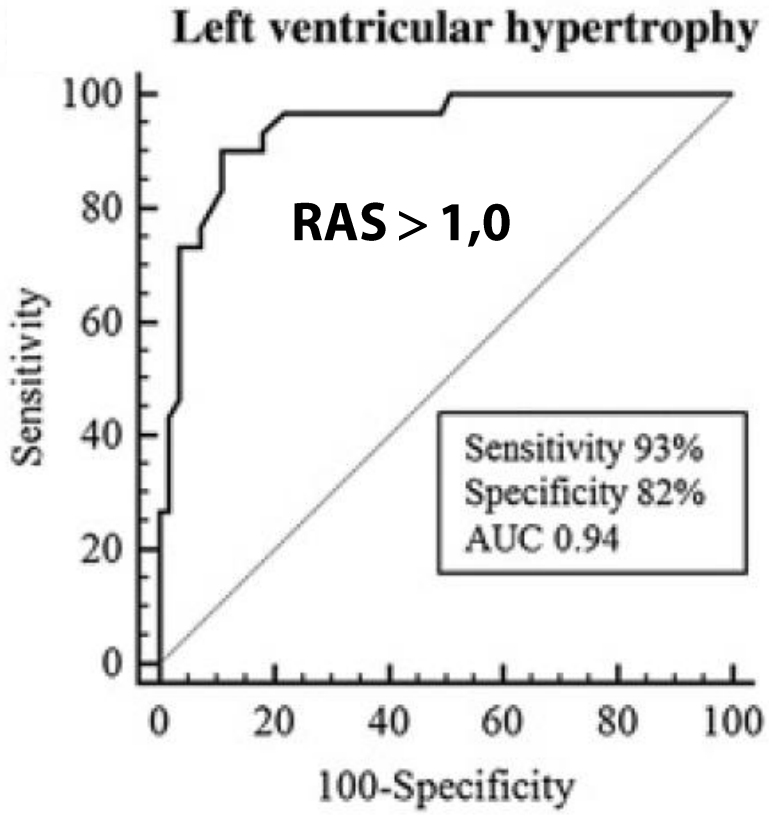
2023 ESC Guidelines for the management of cardiomyopathies

Amyloidóza a fenomén „Relative Apical Sparing“

Relativně zachovalá deformace hrotu vs. bazálních a středních segmentů LK



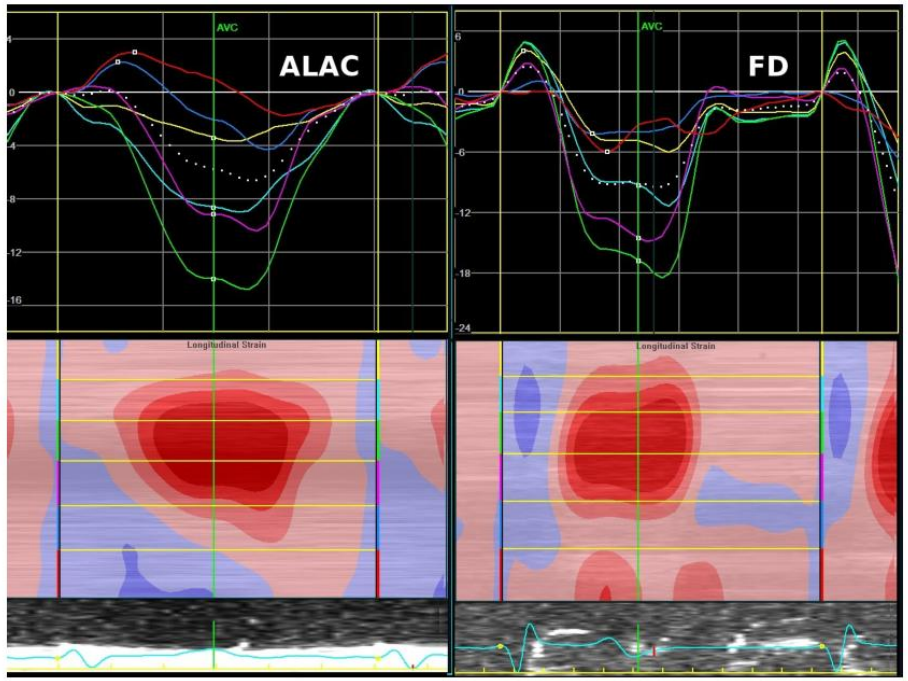
$$\text{Relative apical LS} = \frac{\text{Average apical LS}}{\text{Average basal LS} + \text{Average mid LS}}$$

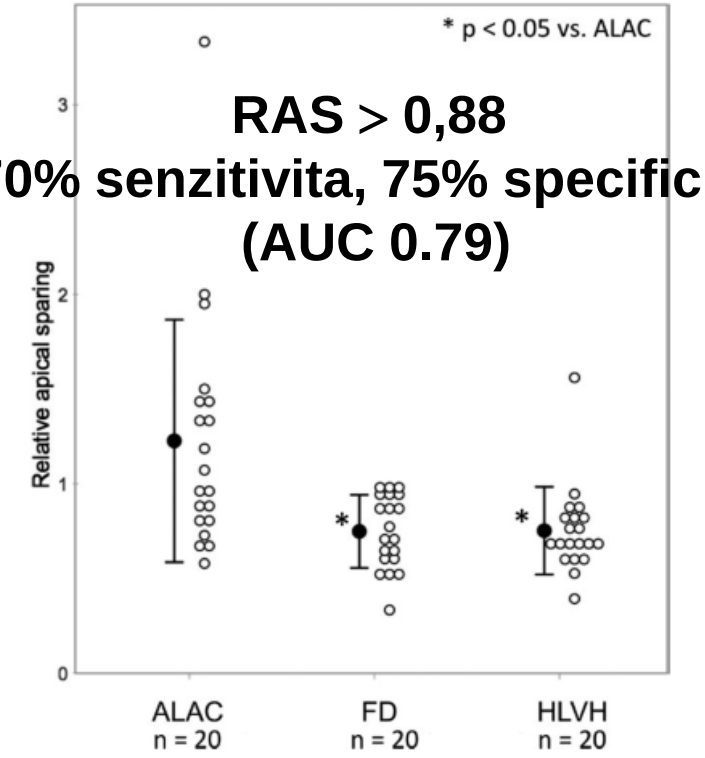


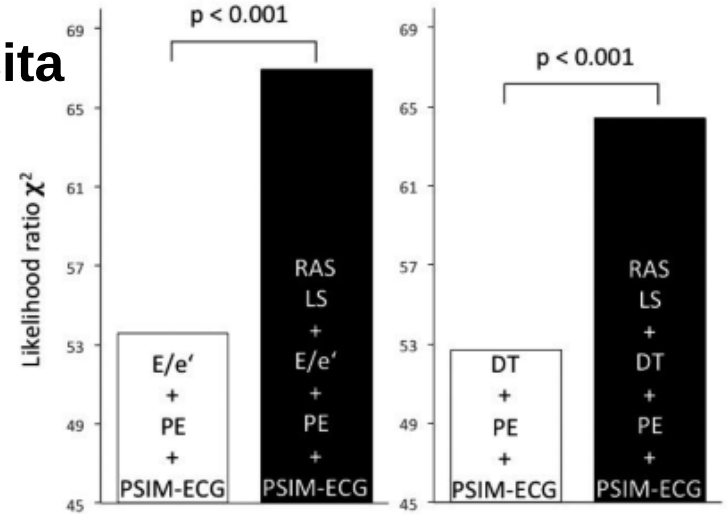
Phelan D, et al. Heart 2012;98

Amyloidóza a fenomén „Relative Apical Sparing“

20 pts s AL amyloidní kardiomyopatií, 20 pts s Fabry kardiomyopatií, 20 pts s hypertenzní LVH

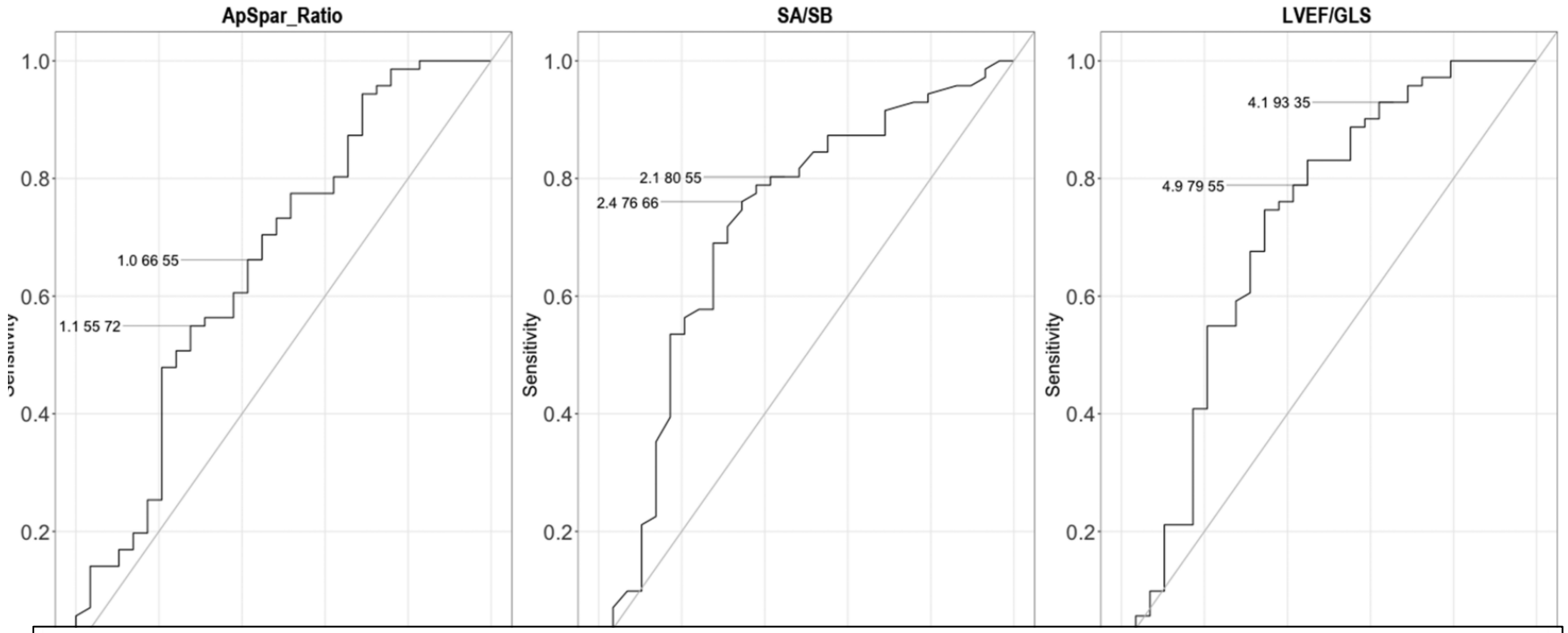




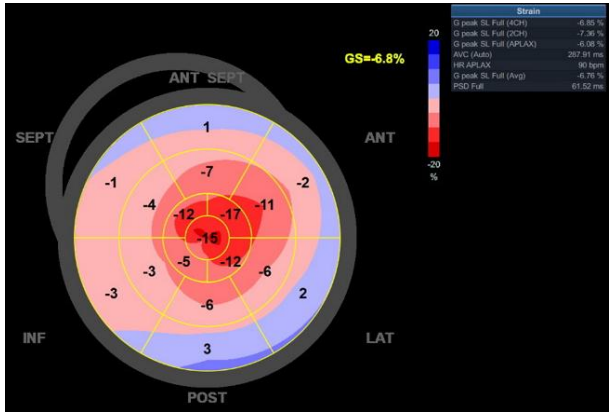


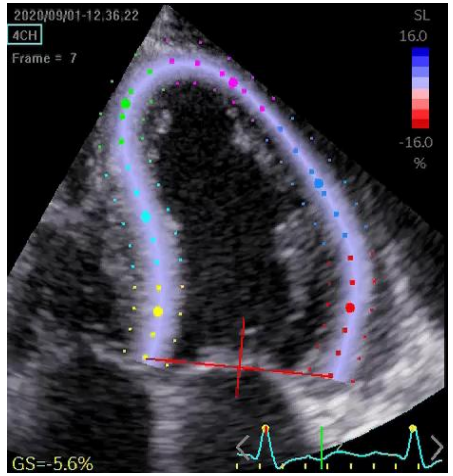
Fikrle, Paleček et al., Echocardiography 2018;35

Amyloidóza a fenomén „Relative Apical Sparing“



	Optimal cut-off[a]	Sensitivity (95% CI)	Specificity (95% CI)	Positive predictive value	Negative predictive value	AUC (95% CI)	*P*-value
ApSpar_Ratio	1.13	-	-	-	-	0.66 (0.54–0.79)	0.011
ApSpar_Ratio > 1.00	-	66 (54–77)	59 (39–76)	80 (71–86)	41 (31–53)	0.62 (0.52–0.73)	0.023
ApSpar_Ratio > 1.13	-	55 (43–67)	72 (53–87)	83 (72–90)	40 (32–48)	0.64 (0.54–0.74)	<0.01
LVEF/GLS	4.95	-	-	-	-	0.72 (0.59–0.84)	<0.001
LVEF/GLS > 4.10	-	93 (84–98)	38 (21–58)	79 (73–83)	69 (46–85)	0.65 (0.56–0.75)	0.001

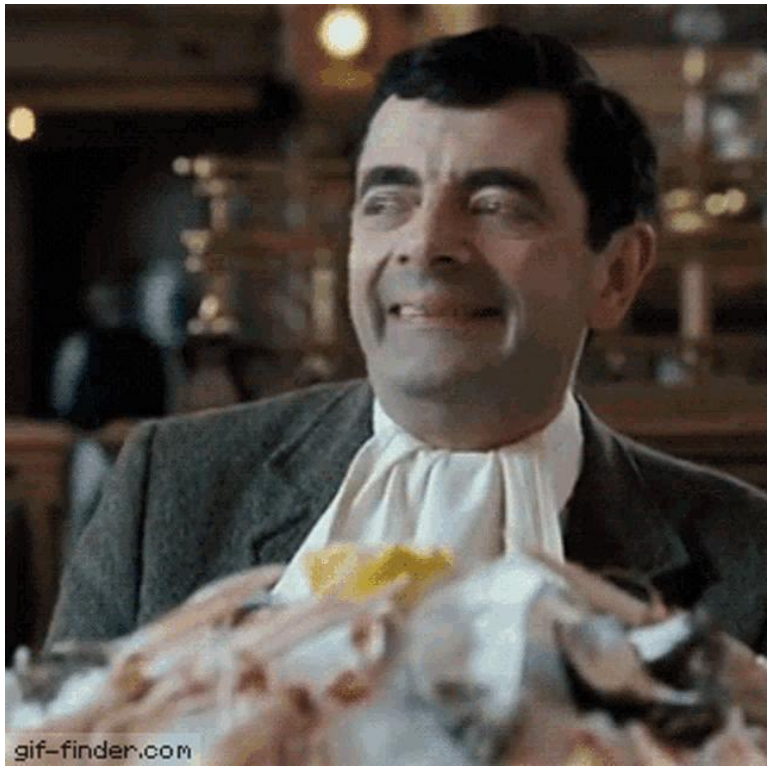




Kyrouac D, Gorcsan J et al., ESC Heart Failure 2022;9

Závěr

Echokardigrafie je základní zobrazovací metodou v managementu kardiomyopatií



KOMPLEXNÍ KARDIO VASKULÁRNÍ CENTRUM VFN Praha

Děkuji za pozornost !