MUNI Masarykova univerzita
Lékařská fakulta
MED

Využití multimodalitního zobrazování pro stratifikaci rizika u pacientů s komorovými arytmiemi

Hypertrofická kardiomyopatie

Hana Poloczková

I.Interní kardioangiologická klinika FN u sv. Anny v Brně
LF Masarykovy univerzity

FAKULTNÍ NEMOCNICE U SV. ANNY V BRNĚ ®

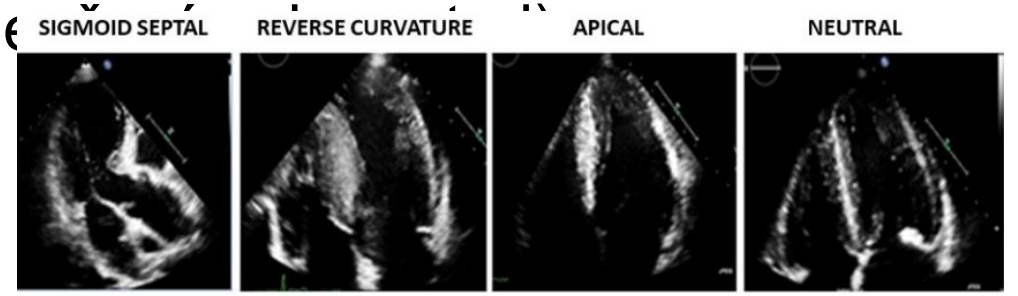
I. INTERNÍ KARDIOANGIOLOGICKÁ KLINIKA FNUSA A LF MU

Hypertrofická kardiomyopatie

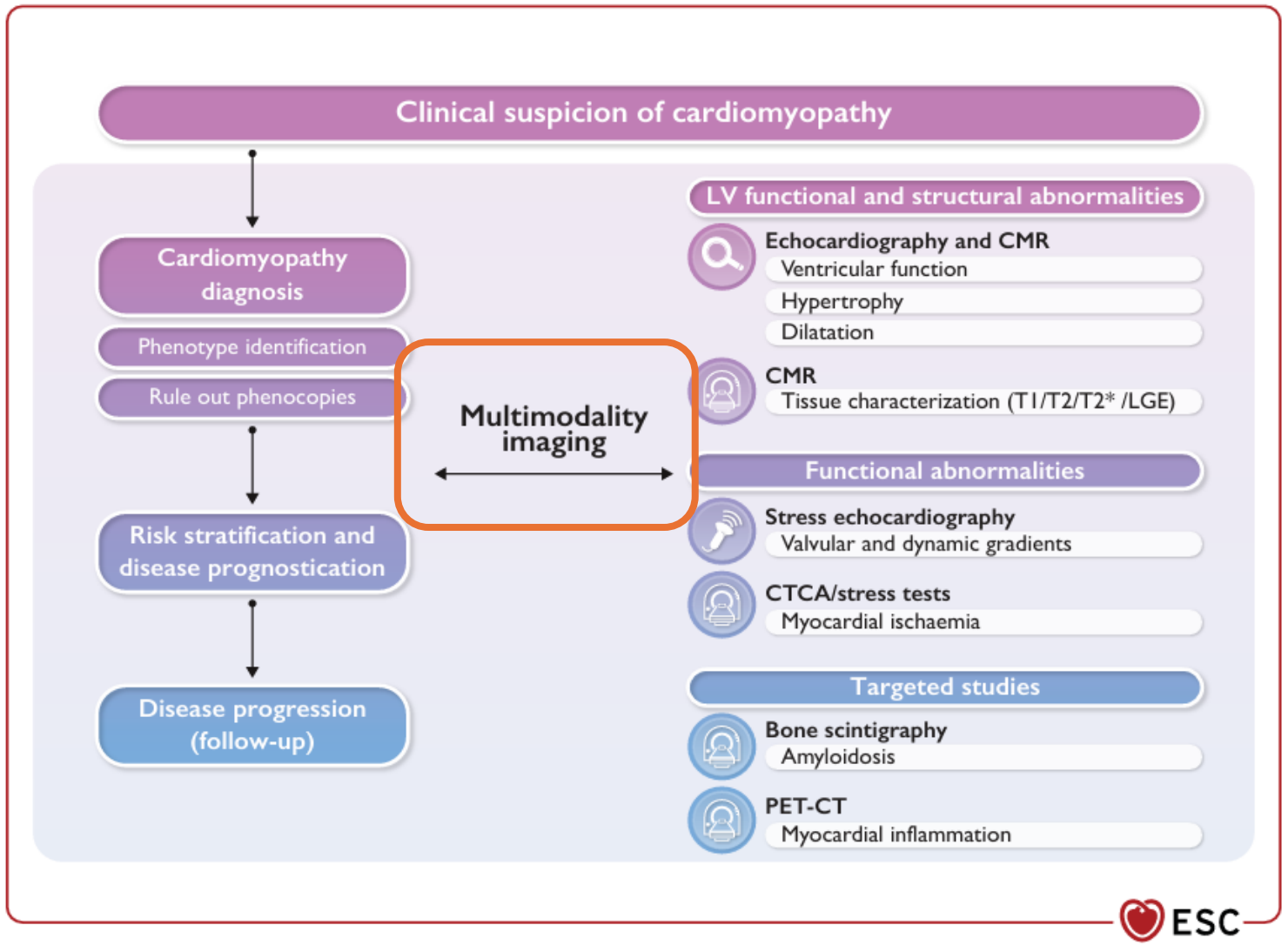
- Definována tloušťkou stěny LK ≥ 15 mm (s/bez hypertrofie stěny PK), kt. není
  vysvětlitelná abnormálním plněním LK

- méně významná hypertrofie LK 13-14 mm – nutno zohlednit další faktory (RA, genetika, EKG abnormity..)

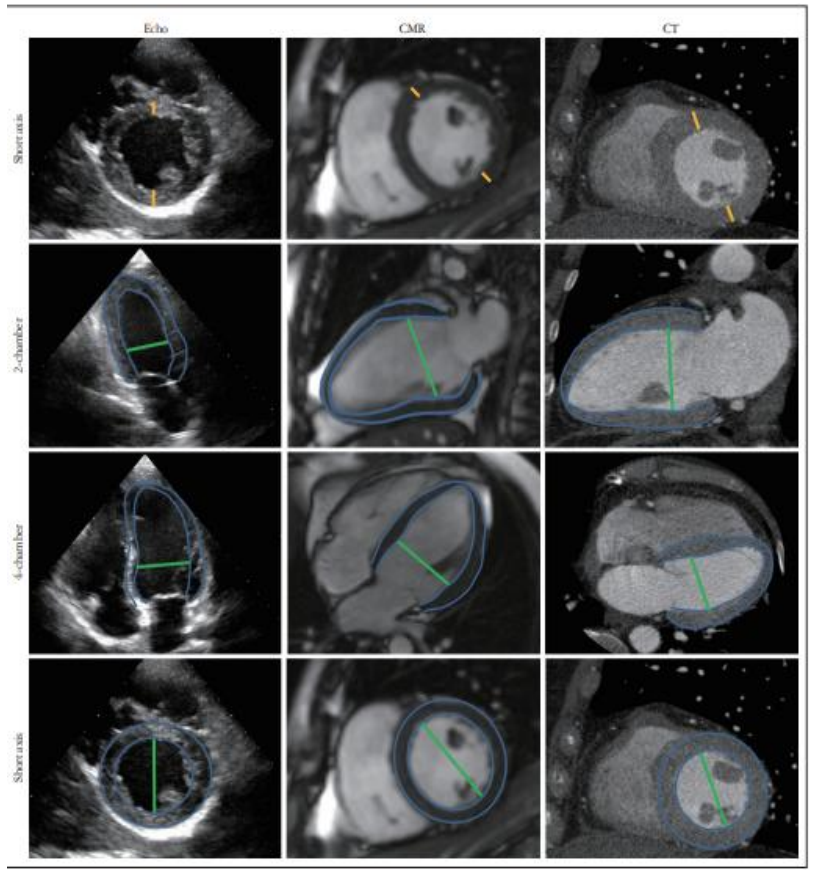
- Výskyt v populaci 1: 500 - 1: 200 (se zl



(Alkema, J Cardiovasc Ultrasound, 2016)



(Arbelo, EHJ, 2023)



(Alkema, J Cardiovasc Ultrasound, 2016)

Management HKMP

MAYO CLINIC PROCEEDINGS

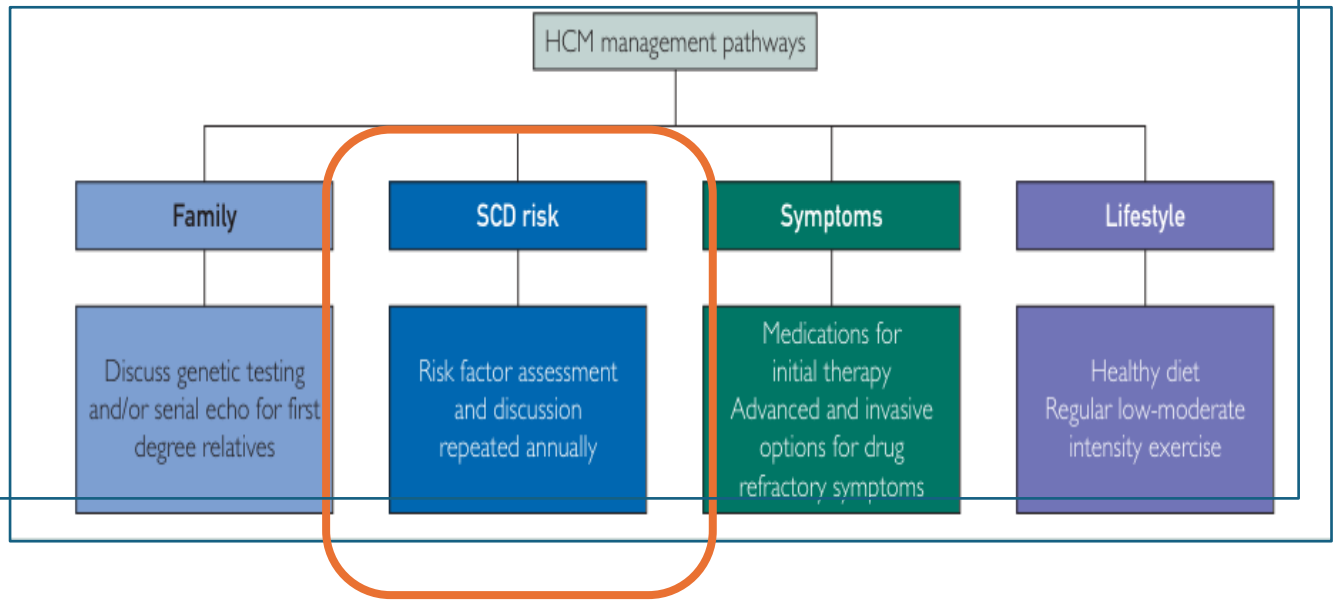
Hypertrophic Cardiomyopathy: State of the Art

Steve R. Ommen, MD; Rick A. Nishimura, MD; Hartzell V. Schaff, MD; and Joseph A. Dearani, MD

- roční mortalita 1-2 %, z důvodu NSS ~ 0,8 % (≤ 60 let), v důsl. komorových arytmií
- nsKT 20-25 % pac. při 24/48h EKG monitoraci, prognost. význam u mladších 30 let
- neprokázán pozit. vliv AA th

- jediná účinná prevence ICD

→ identifikace pacientů s nejvyšším rizikem NSS



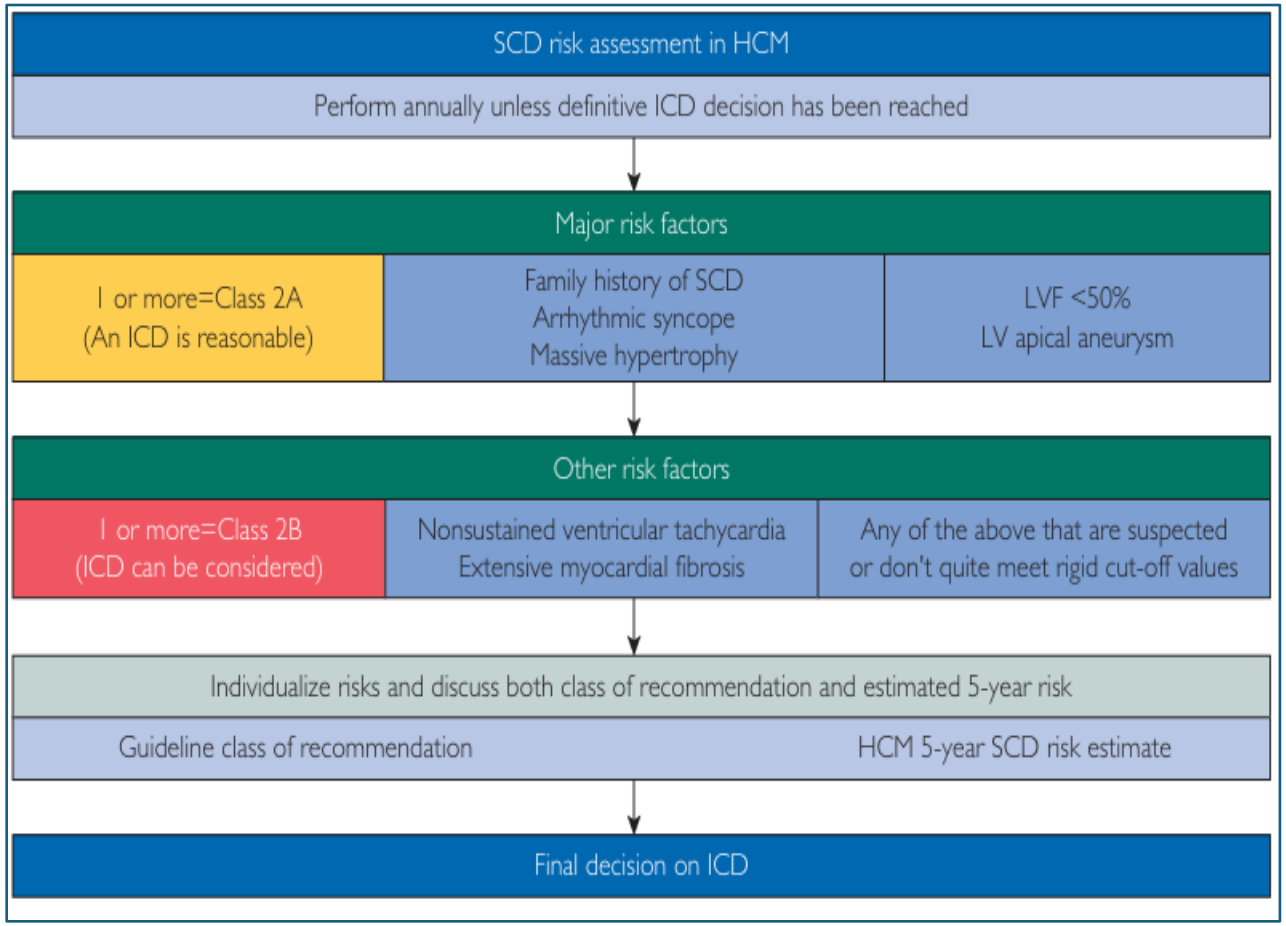
Rizikové faktory NSS – AHA/ACC 2024

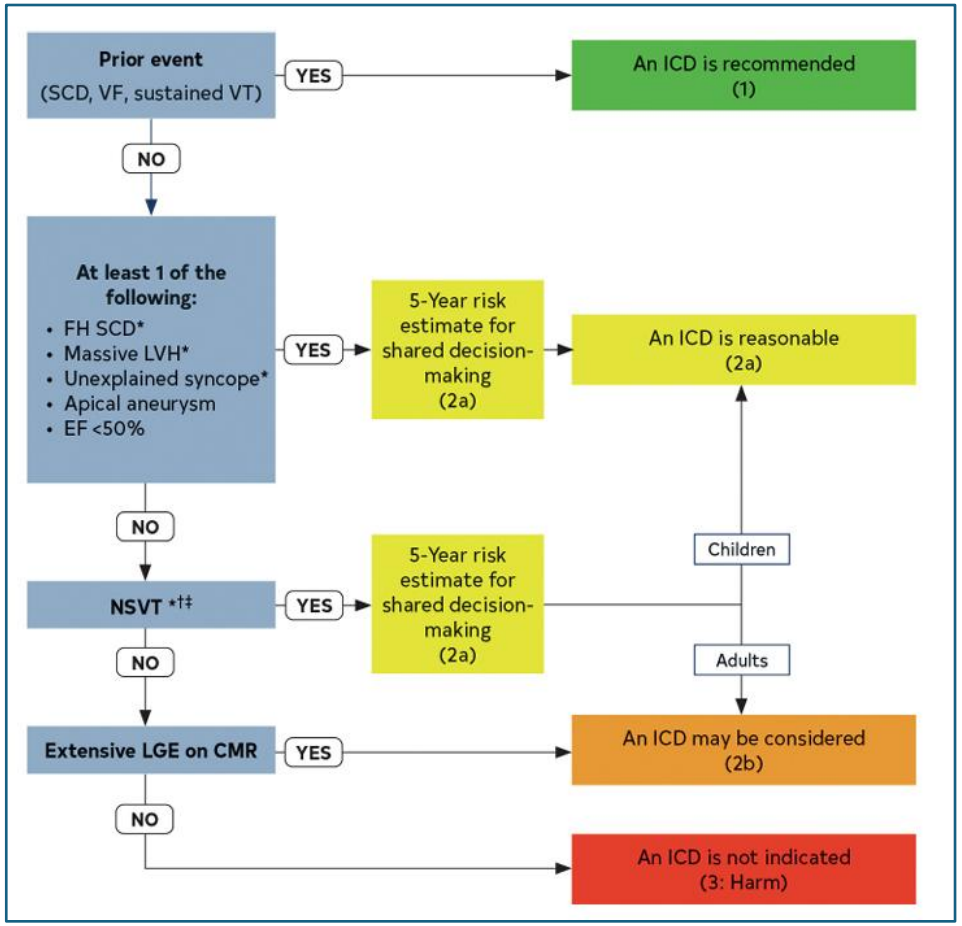
TABLE 8 Clinical Sudden Death Risk Factors for Adults and Children With HCM

Family history of sudden death from HCM	Sudden death judged definitively or likely attributable to HCM in ≥1 first-degree or close relatives who are ≤50 y of age. Close relatives would generally be second-degree relatives; however, multiple SCDs in tertiary relatives should also be considered relevant.[30,31]
Massive LVH	Wall thickness ≥30 mm in any segment within the chamber by echocardiography or CMR imaging; consideration for this morphologic marker is also given to borderline values of ≥28 mm in individual patients at the discretion of the treating cardiologist. For pediatric patients with HCM, an absolute or z-score threshold for wall thickness has not been established; however, a maximal wall thickness that corresponds to a z-score ≥20 (and >10 in conjunction with other risk factors) appears reasonable.[32,33]
Unexplained syncope	≥1 unexplained episodes involving acute transient loss of consciousness, judged by history unlikely to be of neurocardiogenic (vasovagal) etiology, not attributable to LVOTO, and especially when occurring within 6 mo of evaluation (events beyond 5 y in the past do not appear to have relevance).[34]
HCM with LV systolic dysfunction	Systolic dysfunction with EF <50% by echocardiography or CMR imaging.[24,27]
LV apical aneurysm	Apical aneurysm defined as a discrete thin-walled dyskinetic or akinetic segment with transmural scar or LGE of the most distal portion of the LV chamber, independent of size. (In children, apical aneurysm is uncommon, and the risk has not been studied.)[15,16]
Extensive LGE on CMR imaging	Extensive LGE, representing replacement fibrosis, either quantified or estimated by visual inspection, comprising ≥15% of LV mass (extent of LGE conferring risk has not been defined in children).[9-11,20-22,25]
NSVT on ambulatory monitor	≥3 beats at ≥120 bpm has generally been used in studies. It would seem most appropriate to place greater weight on NSVT as a risk marker when runs are frequent (eg, ≥3), longer (eg, ≥10 beats), or faster (eg, ≥200 bpm) occurring usually over 24 to 48 h of monitoring. For pediatric patients, a VT rate that exceeds the baseline sinus rate by >20% is considered significant.[35-37]
Genotype status	Genotype-positive status (ie, harboring a putatively disease-causing pathogenic/likely pathogenic variant) is associated with higher SCD risk in pediatric patients with HCM.[12,14]

(Ommen, Mayo Clin Proc, 2025)

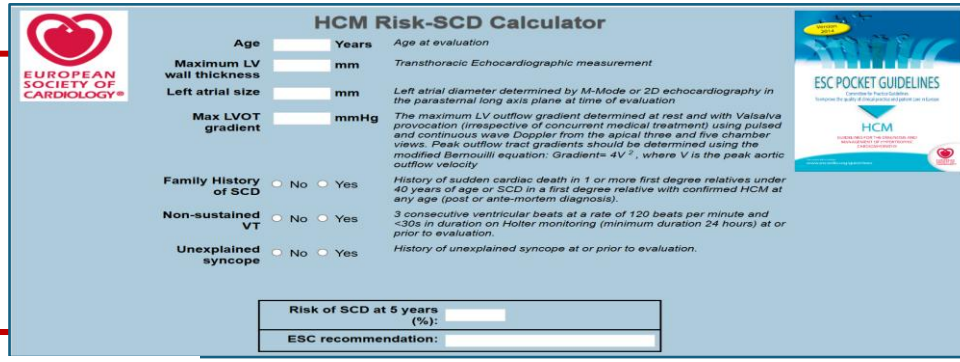
Riziková stratifikace AHA/ACC 2024





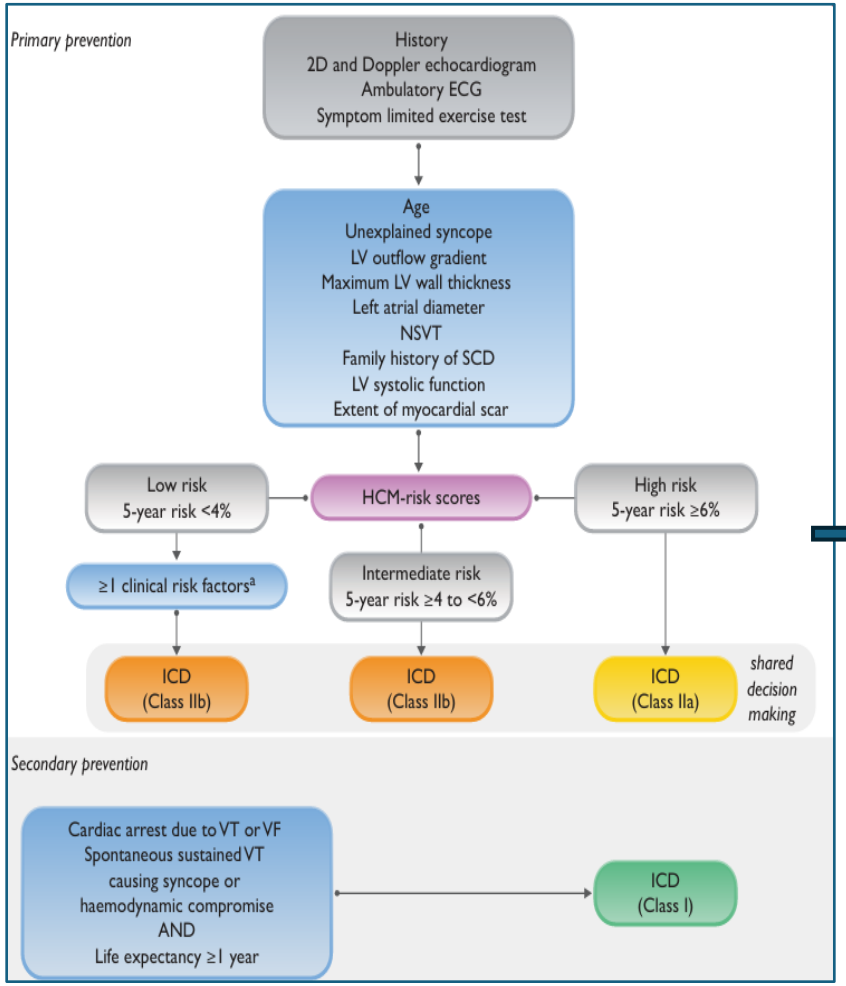
(Ommen, Mayo Clin Proc, 2025)

HCM Risk-SCD Calculator



Risk factor	Comment
Age	• The effect of age on SCD has been examined in a number of studies[86,525,584,760–764] and two have shown a significant association, with an increased risk of SCD in younger patients.[525,584] • Some risk factors appear to be more important in younger patients, most notably NSVT,[765] severe LVH,[766] and unexplained syncope.[584] • Sudden cardiac death is very rare below the age of 6 years,[535,767] and there are some data to suggest a peak of SCD in childhood HCM between 9 and 15 years;[757] however, the association between age at diagnosis and SCD risk in childhood HCM remains unclear.
NSVT	• NSVT (defined as ≥3 consecutive ventricular beats at ≥120 b.p.m. lasting <30 s) occurs in 20–30% of patients during ambulatory ECG monitoring and is an independent predictor of SCD.[81,525,535,590,764,765,768–773] • There is no evidence that the frequency, duration, or rate of NSVT influences the risk of SCD.[765,774] • NSVT occurring during or immediately following exercise is very rare, but may be associated with a high risk of SCD.[768]
Maximum LV wall thickness	• The severity and extent of LVH measured by TTE are associated with the risk of SCD.[81,535,592,593,763,765,770–772,775–780] • Several studies have shown the greatest risk of SCD in patients with a maximum wall thickness of ≥30 mm; however, there are few data in patients with extreme hypertrophy (≥35 mm).[525,592,763,765,769,781–784]
Family history of sudden cardiac death at a young age	• While definitions vary,[525,592,762,782] a family history of SCD is usually considered clinically significant when one or more first-degree relatives have died suddenly aged <40 years with or without a diagnosis of HCM, or when SCD has occurred in a first-degree relative at any age with an established diagnosis of HCM. • Family history of SCD does not appear to be an independent risk factor for SCD in childhood HCM.[81,535] This may be due to a higher prevalence of *de novo* variants in childhood HCM, the inclusion of non-sarcomeric disease, and/or under-reporting of family history in paediatric cohorts.
Syncope	• Syncope is common in patients with HCM but is challenging to assess, as it has multiple causes.[785] • Non-neurocardiogenic syncope for which there is no explanation after investigation is associated with an increased risk of SCD.[81,525,535,584,590,755,761,768,769,781,786–788] • Episodes within 6 months of evaluation may be more predictive of SCD.[584]
Left atrial diameter	• Several studies have reported a positive association between LA size and SCD.[81,525,535,584,772,789] There are no data on the association between SCD and LA area or volume. Measurement of LA size is also important in assessing the risk of AF (see *Section 6.10.3*).
LV outflow tract obstruction	• A number of studies have reported a significant association between LVOTO and SCD risk.[86,525,590,762,768,790] Several unanswered questions remain, including the prognostic importance of provocable LVOTO and the impact of treatment (medical or invasive) on SCD. • In childhood HCM, there are conflicting data on the association between LVOTO and SCD risk.[81,535,772,777]

ESC 2023 guidelines



TTE

Diametr LS

LVOTG

Max. tloušťka stěny LK

Systol. funkce LK

Rozsah LGE

MRI

Primary prevention		
The HCM Risk-SCD calculator is recommended as a method of estimating risk of sudden death at 5 years in patients aged ≥16 years for primary prevention.[525,821–824]	I	B
Validated paediatric-specific risk prediction models (e.g. HCM Risk-Kids) are recommended as a method of estimating risk of sudden death at 5 years in patients aged <16 years for primary prevention.[81,833]	I	B
It is recommended that the 5-year risk of SCD be assessed at first evaluation and re-evaluated at 1–2 year intervals or whenever there is a change in clinical status.[525]	I	B
Implantation of an ICD should be considered in patients with an estimated 5-year risk of sudden death of ≥6%, following detailed clinical assessment that considers: (i) the lifelong risk of complications; (ii) competing mortality risk from the disease and comorbidities; AND (iii) the impact of an ICD on lifestyle, socio-economic status, and psychological health.[81,521,525,726,832,833]	IIa	B

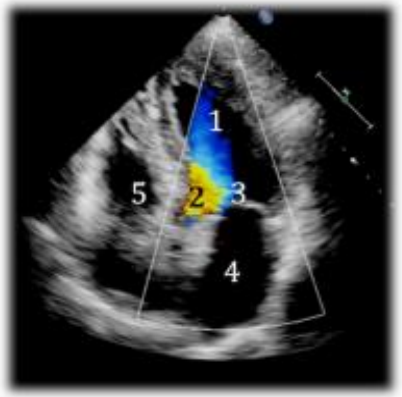
(Arbelo, EHJ, 2023)

Echokardiografie

Hypertrophic cardiomyopathy

Unexplained maximal WT ≥15 mm in any myocardial segment.
(≥13 mm in first degree relatives of HCM patients)

1. Left ventricle
- ✓ Wall thickness
  - *Involved segments*
  - *Symmetric/asymmetric*
  - *Maximal thickness*
- ✓ Systolic function
  - *Ejection fraction*
  - *Longitudinal strain*
- ✓ Diastolic function



2. LVOTO
- ✓ Presence of LVOTO: > 30 mmHg
  - *Excercise echo in symptomatic patients with < 50 mmHg at rest or with bedside manoeuvres (Valsalva, standing…)*
- ✓ Assess midventricular obstruction

3. Mitral valve
- ✓ SAM : confirm contact with septum and duration
- ✓ Evaluate leaflets, chordae and papillary muscle abnormalities
- ✓ Consider concurrent organic disease

4. Left atria
- ✓ Anteroposterior diameter
- ✓ Indexed volume

5. Right ventricle
- ✓ Evaluate hypertrophy
- ✓ Assess RVOT obstruction

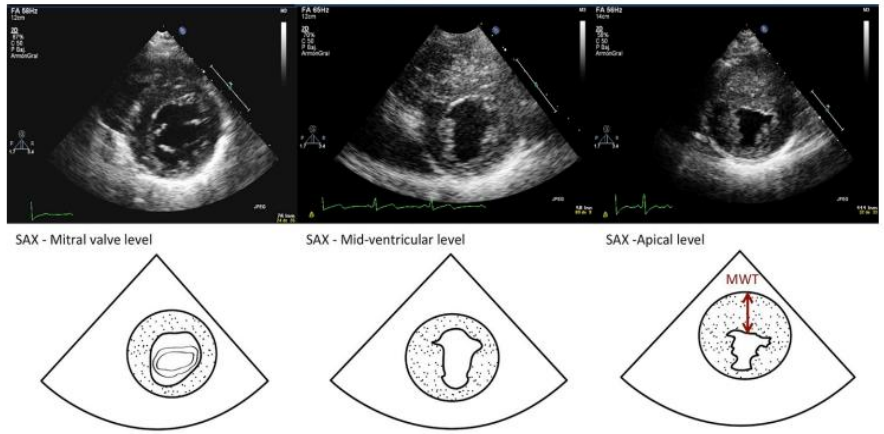
(Dominquez, BMJ, 2018)

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
Transoesophageal echocardiography should be considered in patients with HCM and LVOTO if the mechanism of obstruction is unclear or when assessing the mitral valve apparatus before a septal reduction procedure, or when severe mitral regurgitation caused by intrinsic valve abnormalities is suspected.[599–602]	IIa	C

(Arbelo, EHJ, 2023)

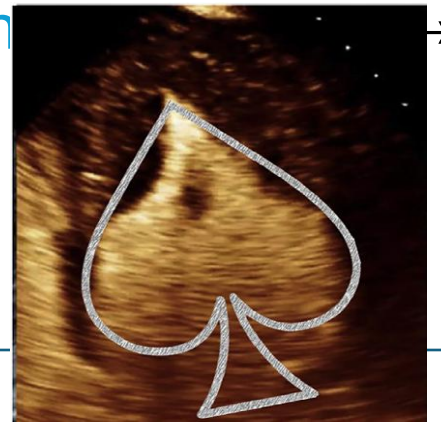
Hodnocení hypertrofie

- Hodnocení tloušťky stěn LK – měření v end-diastole, preferenčně PSAX
  - hodnocení všech segmentů od baze k apexu
  - nejčastěji asymetrická hypertrofie septa
- Tíže a rozsah hypertrofie -riziko NSS → vysoké riziko - hypertrofie > 30 mm



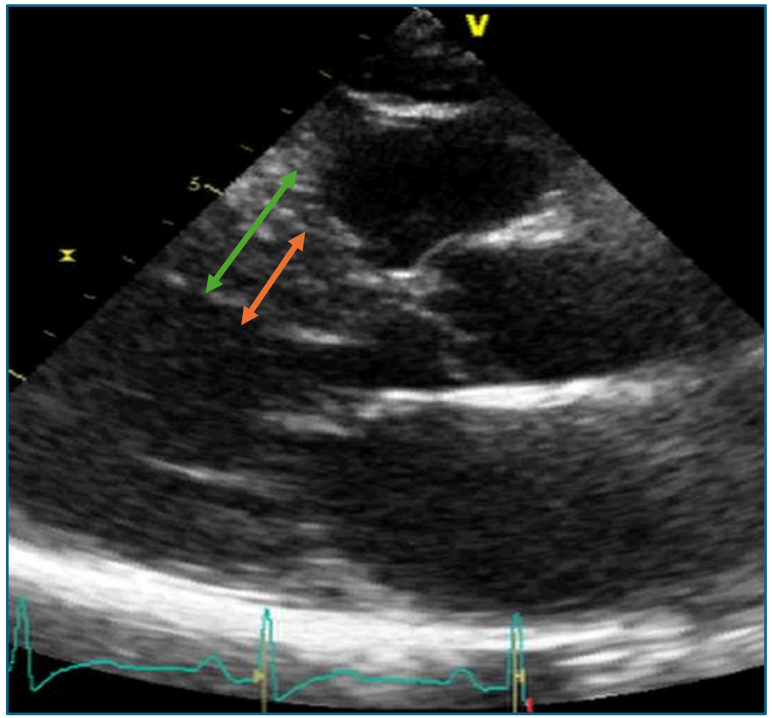
(Dominquez, BMJ, 2018)

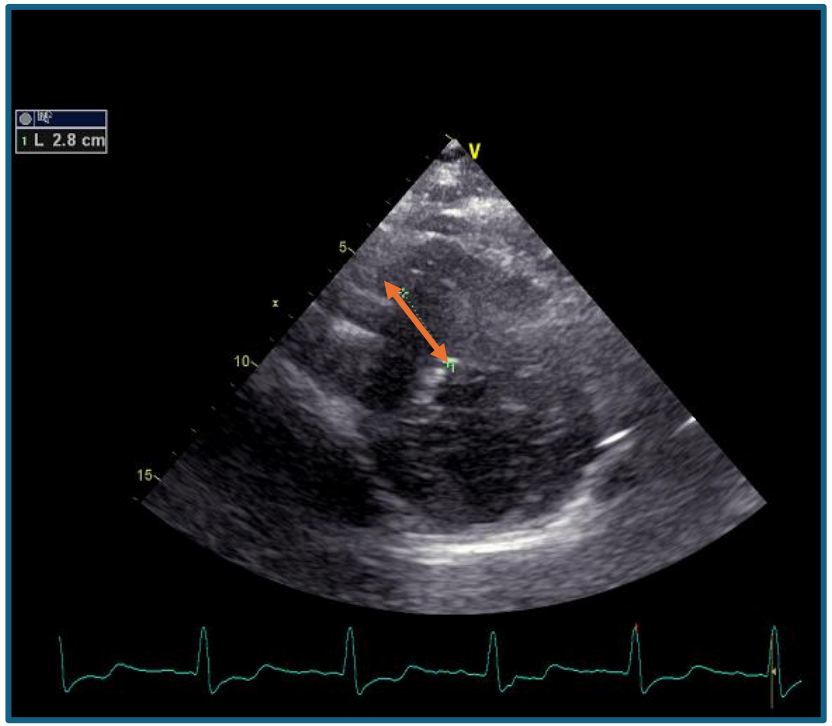
apikáln →



(Abraham, Am J Cardiol, 2024)

Hodnocení hypertrofie





(Archív I.IKAK)

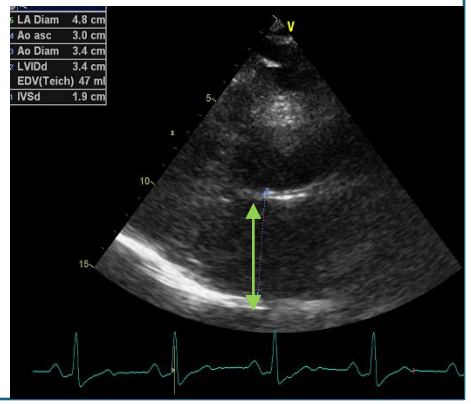
Hodnocení velikosti levé síně

- dilatace LS u většiny pacientů ← mitrální regurgitace, vysoké plnicí tlaky
- Diametr LS v PLAX – pozit. asociace s rizikem NSS

  (riziko rozvoje fibrilace síní, kardioembolizace)

  - diametr > 45 mm

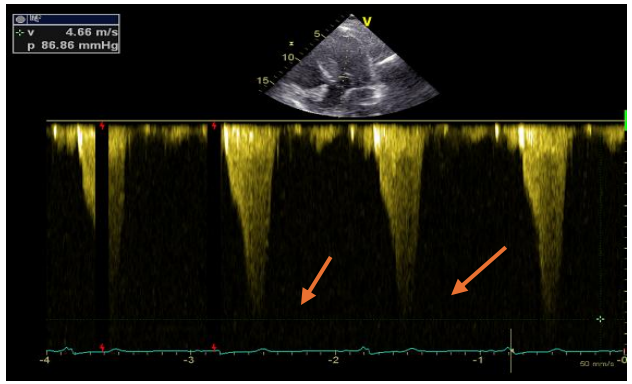
- Velikost plochy či objemu LS – není asoc. s rizikem NSS



(Archív I.IKAK)

Přítomnost obstrukce

- Obstrukce u 50 – 70 % pacientů, u ½ jen provokovaná
  - LVOT - v důsledku SAM, ve 2. polovině systoly
  - midventrikulárně – asi u 10 %
- Obstrukční/neobstrukční forma HKMP
  - grad ≥ 30 mmHg
  - hemodynam. významná ≥ 50 mmHg

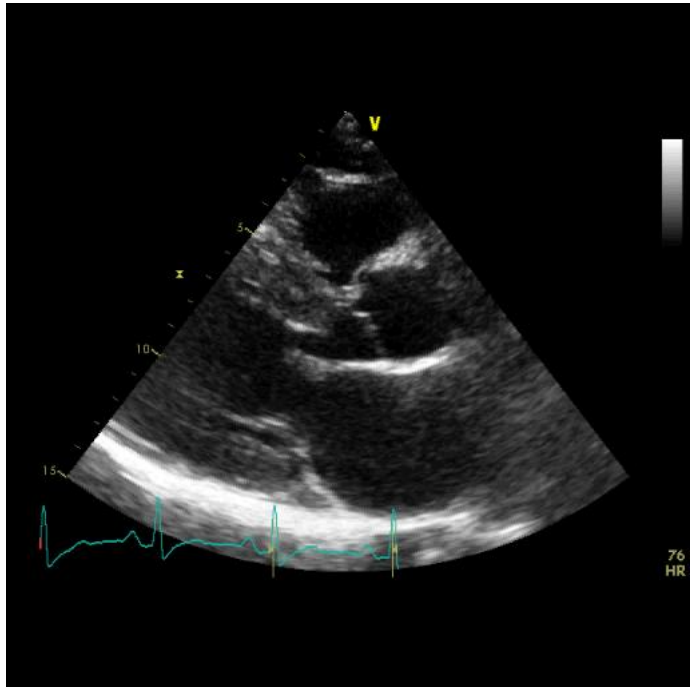


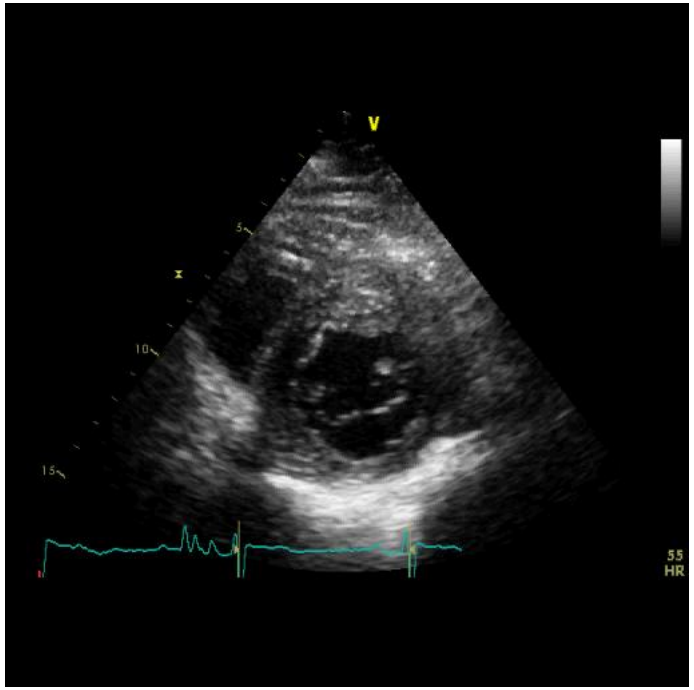
- Asociace LVOTG s rizikem NSS

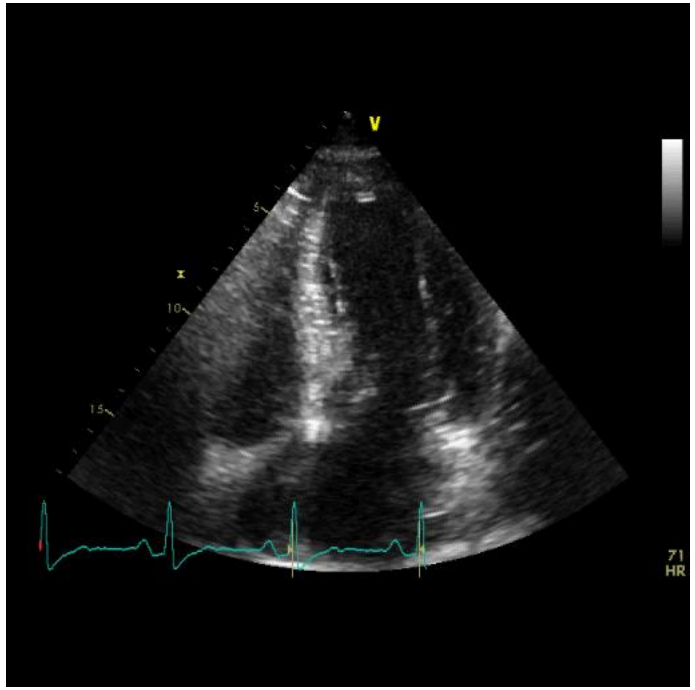
(Archív I.IKAK)

Hodnocení systolické funkce LK

- hodnota EF LK, FS – obvykle normální až supranormální
  (díky zachování radiální kontraktility)







(Archív I.IKAK)

Systolická funkce LK

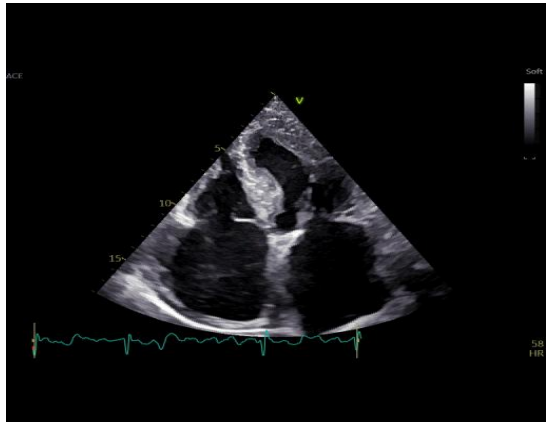
- 5 – 10 % pacientů → „burnt out“ fáze → „end stage HCM“

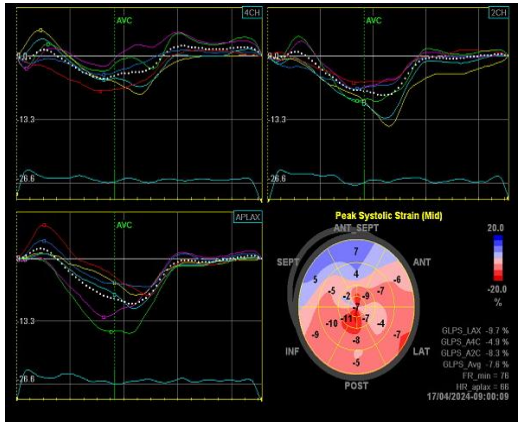
  - dilatace LK, regrese hypertrofie

- spojena s rizikem NSS (7-20 % ve srov. s běžnou populací)

pokles hodnoty EF → EF < 50 %

= těžká systolická dysfunkce !





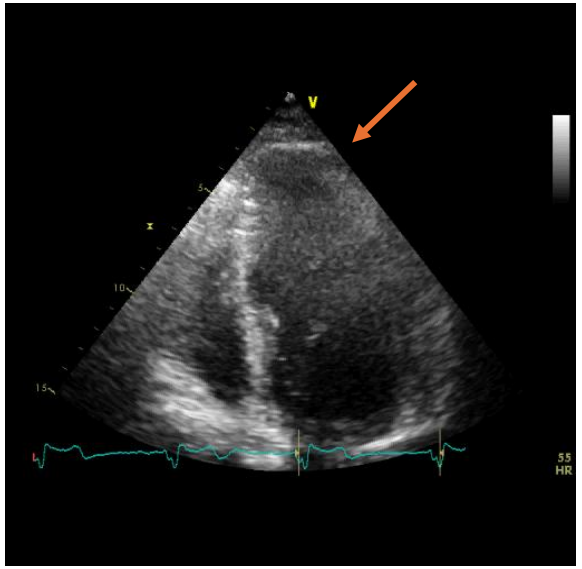
(Archív I.IKAK)

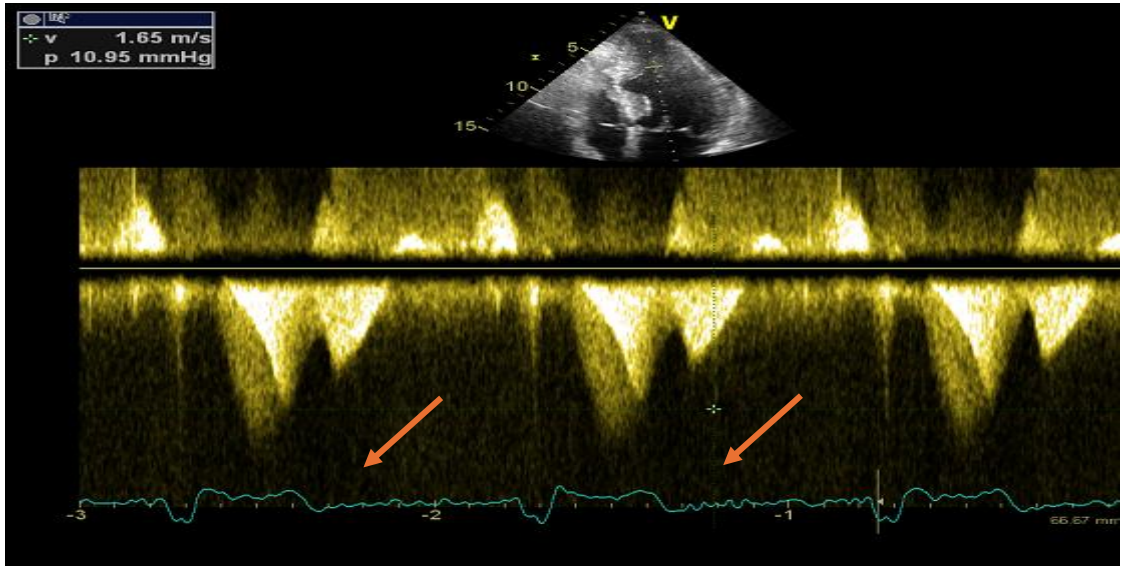
Apikální aneurysma

- apikální aneurysma – prevelance ~ 3 %

  často midventrikulární obstrukce – u cca 10 %- „lobster-claw“

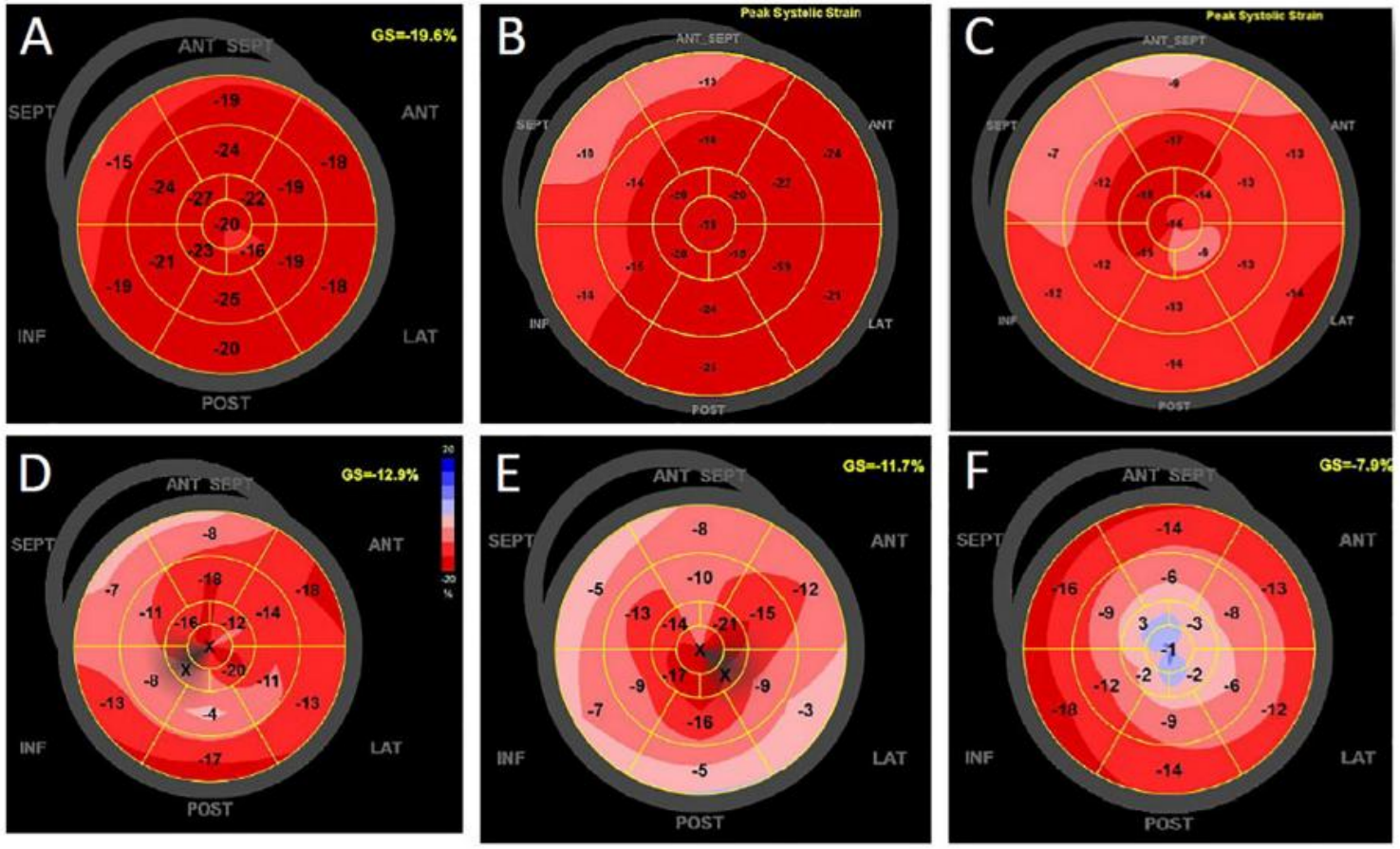
  - často výrazné symptomy, spojeno s monomorf KT





(Archív I.IKAK)

Hodnocení systolické funkce



(Abraham, Am J Cardiol, 2024)

Hodnocení longitudinálního strainu – vyšší senzitivita ve srovnání s hodnocením EF

Snížení hodnot GLS i přes normální hodoty EF LK již v časných stadiích

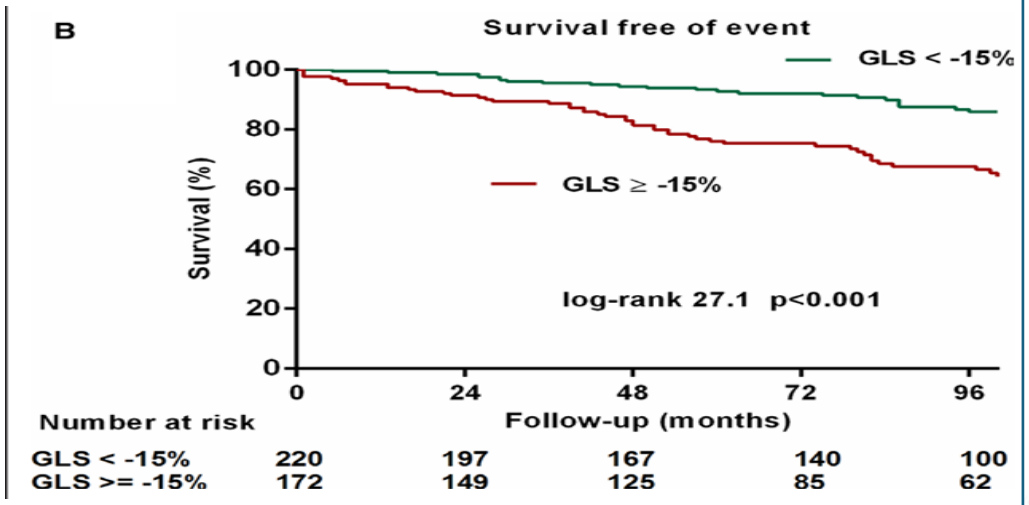
Patologické hodnoty strainu v oblastech hypertrofie

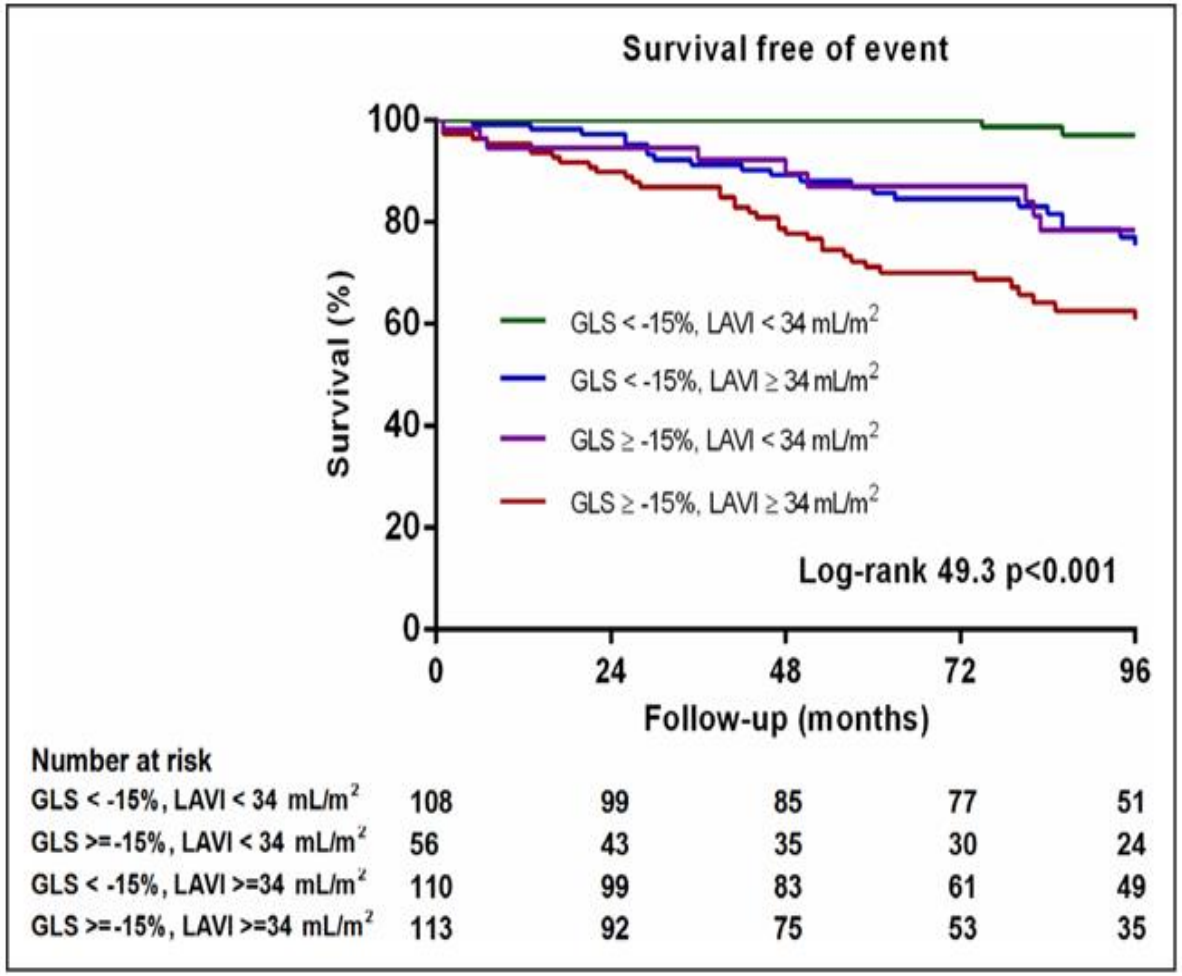
Patologické hodnoty i u genotyp+/fenotyp- osob

Global longitudinal strain, objem levé síně

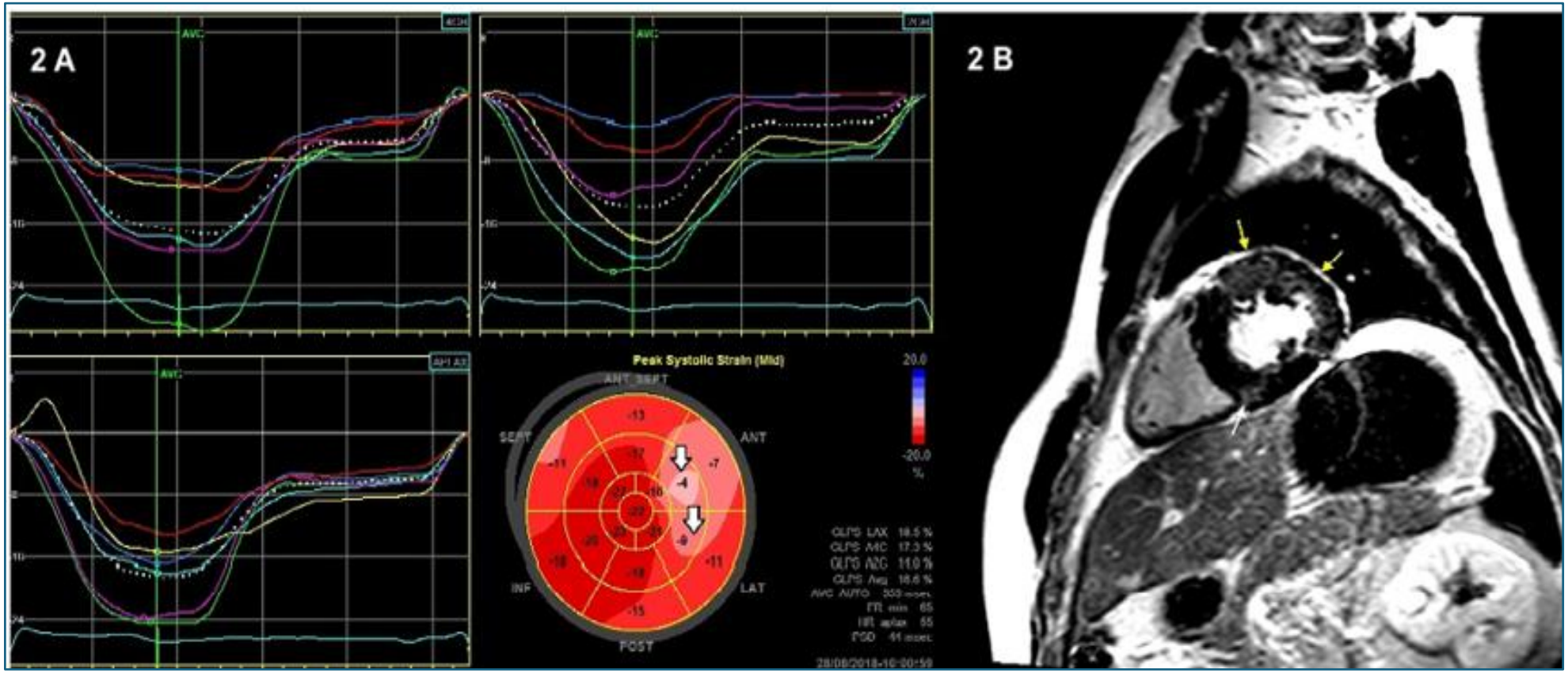
Global Longitudinal Strain and Left Atrial Volume Index Provide Incremental Prognostic Value in Patients With Hypertrophic Cardiomyopathy

Yasmine L. Hiemstra, MD; Philippe Debonnaire, MD, PhD; Marianne Bootsma, MD, PhD; Erik W. van Zwet, MD, PhD; Victoria Delgado, MD, PhD; Martin J. Schalij, MD, PhD; Douwe E. Atsma, MD, PhD; Jeroen J. Bax, MD, PhD; Nina Ajmone Marsan, MD, PhD





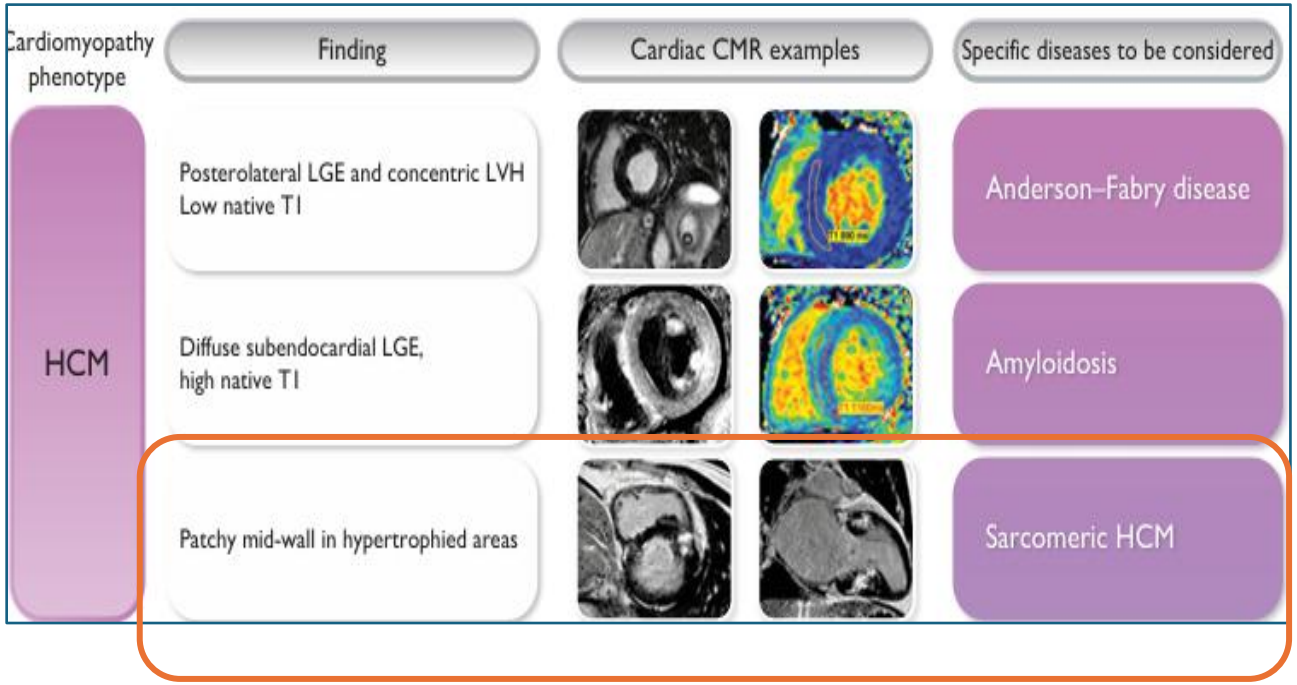
Segmentární longitudinální strain



(Wabich et all, J of Cardiol, 2021)

Segment. longitud. strain cut-off -12,5 % → odp. ložiskům LGE na MRI

Magnetická rezonance

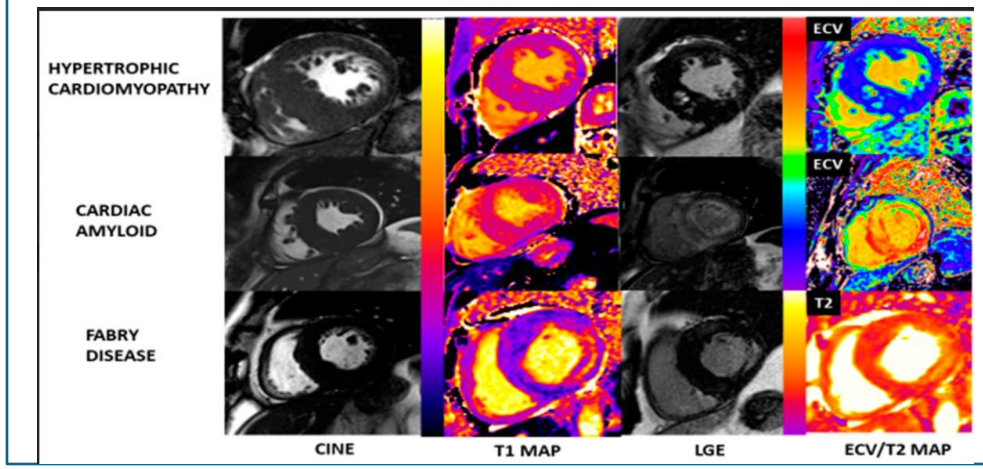


(Arbelo, EHJ, 2023)

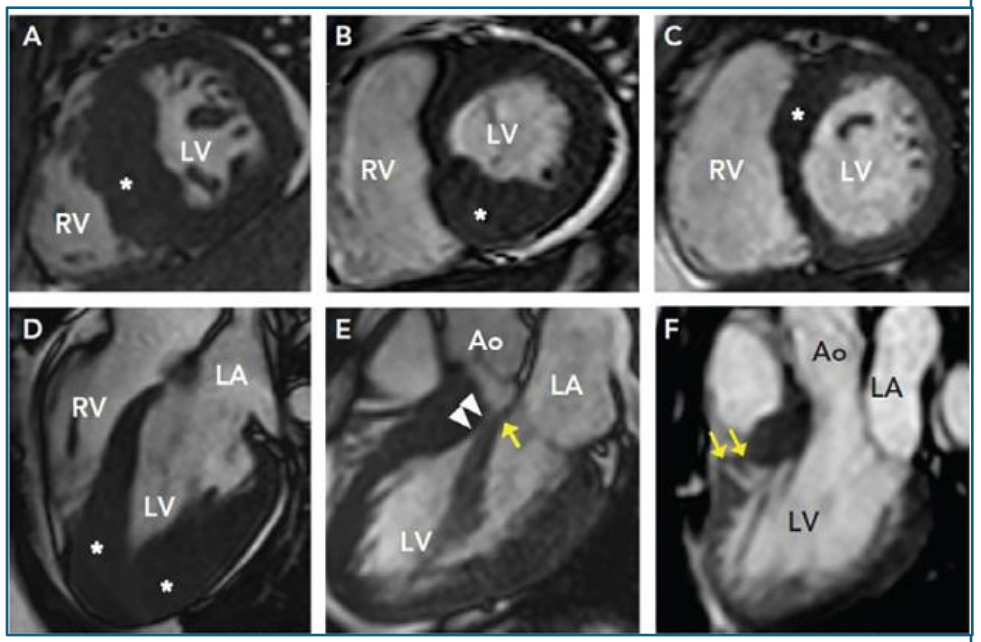
Recommendations	Class[a]	Level[b]
Contrast-enhanced CMR is recommended in patients with cardiomyopathy at initial evaluation.[10,90,116,119–143]	I	B
Contrast-enhanced CMR should be considered in patients with cardiomyopathy during follow-up to monitor disease progression and aid risk stratification and management.[89,90,120–122,127,129,136–147]	IIa	C
Contrast-enhanced CMR should be considered for the serial follow-up and assessment of therapeutic response in patients with cardiac amyloidosis, Anderson–Fabry disease, sarcoidosis, inflammatory cardiomyopathies, and haemochromatosis with cardiac involvement.[148–152]	IIa	C
In families with cardiomyopathy in which a disease-causing variant has been identified, contrast-enhanced CMR should be considered in genotype-positive/phenotype-negative family members to aid diagnosis and detect early disease.[10,122,126,128,129,135–143,145,153–159]	IIa	B
In cases of familial cardiomyopathy without a genetic diagnosis, contrast-enhanced CMR may be considered in phenotype-negative family members to aid diagnosis and detect early disease.[10,128]	IIb	C

MRI – hodnocení hypertrofie

- „zlatý standard“
- anterolaterální, apikální hypertrofie
- rozsah a char. hypertrofie, LGE
- ddg fenokopií

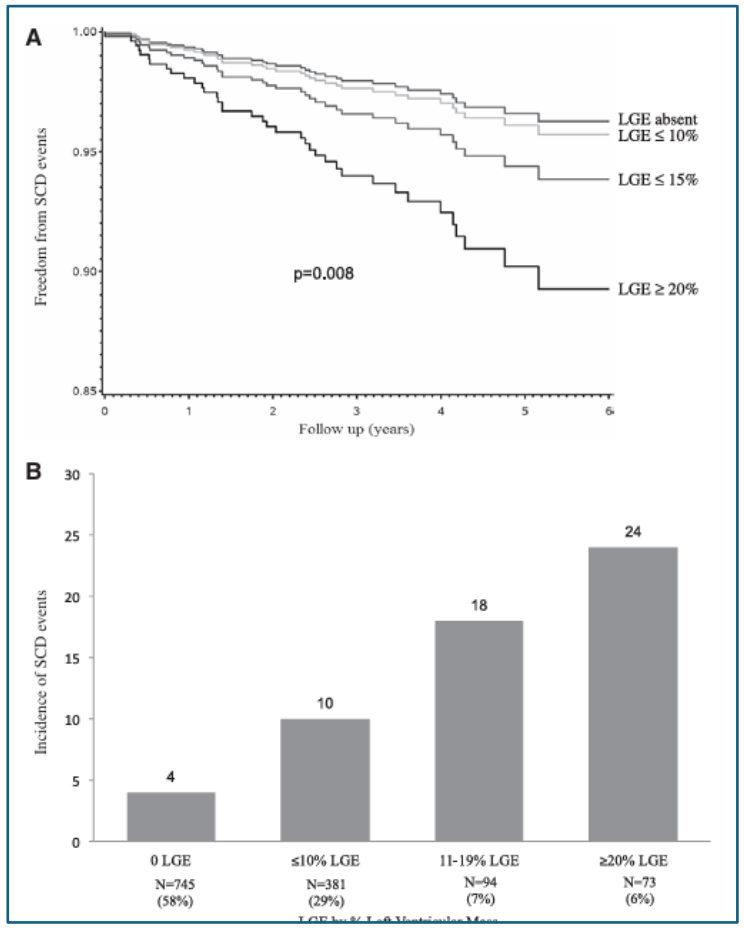


(Goldie, J Clin Med, 2024)



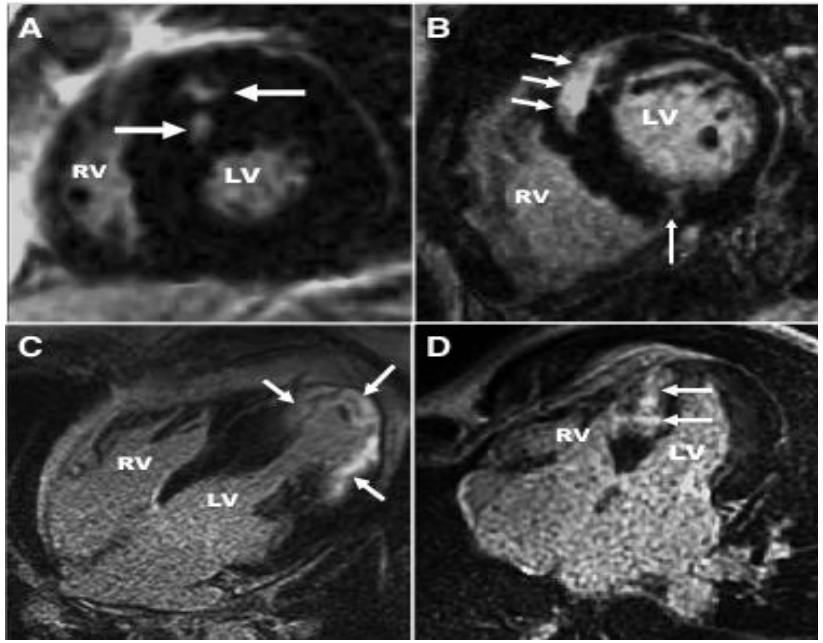
(Abraham, Am J Cardiol, 2024)

LGE v predikci NSS



Prognostic Value of Quantitative Contrast-Enhanced Cardiovascular Magnetic Resonance for the Evaluation of Sudden Death Risk in Patients With Hypertrophic Cardiomyopathy

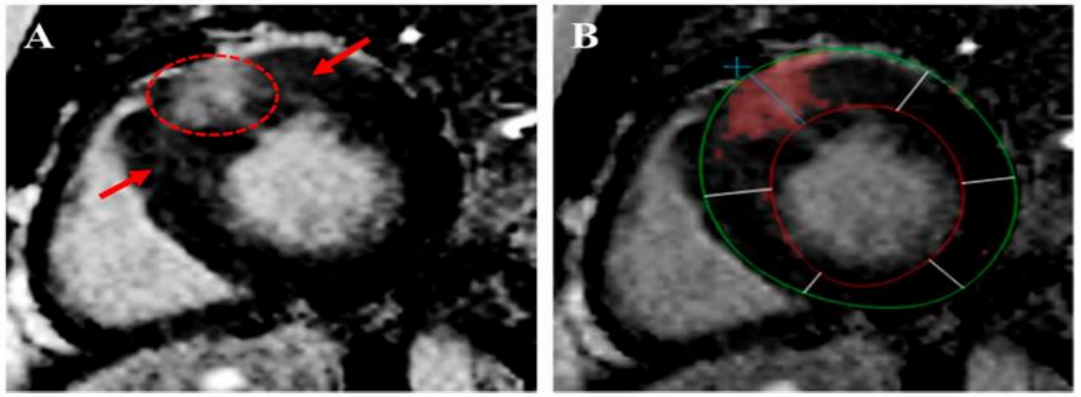
Raymond H. Chan, MD, MPH; Barry J. Maron, MD; Iacopo Olivotto, MD; Michael J. Pencina, PhD; Gabriele Egidy Assenza, MD; Tammy Haas, RN; John R. Lesser, MD; Christiane Gruner, MD; Andrew M. Crean, MD; Harry Rakowski, MD; James E. Udelson, MD; Ethan Rowin, MD; Massimo Lombardi, MD; Franco Cecchi, MD; Benedetta Tomberli, MD; Paolo Spirito, MD; Francesco Formisano, MD; Elena Biagini, MD; Claudio Rapezzi, MD; Carlo Nicola De Cecco, MD; Camillo Autore, MD; E. Francis Cook, PhD; Susie N. Hong, MD; C. Michael Gibson, MD, MS; Warren J. Manning, MD; Evan Appelbaum, MD; Martin S. Maron, MD



LGE v predikci NSS

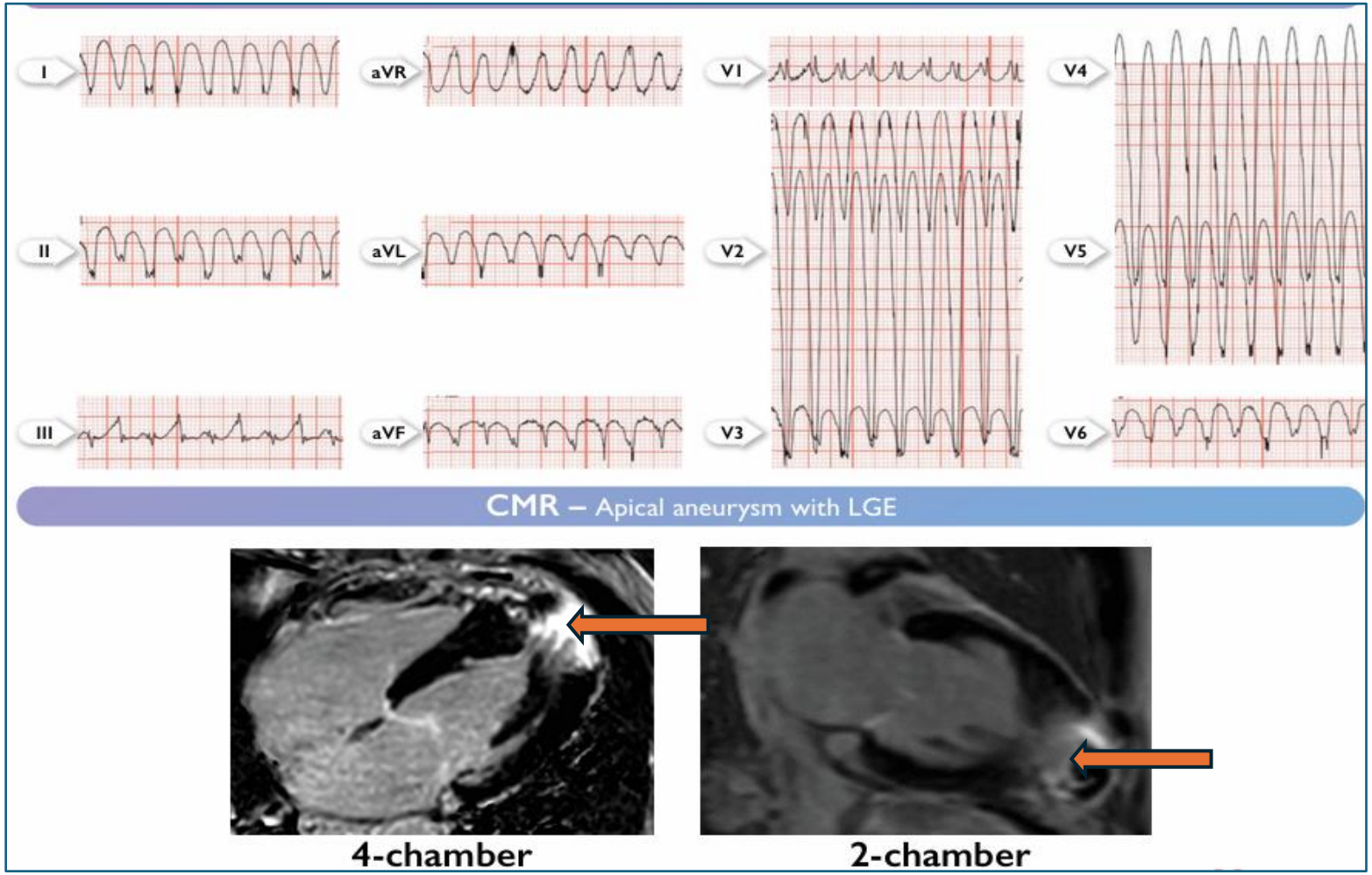
- přítomnost LGE koreluje s rozsahem fibrózy
- ≥ 15 % - dobrá korelace s rizikem NSS
- v dospělosti prevalence LGE 45-90 %, s věkem narůstá
- Riziko NSS < 6 % → LGE 10 – 15 % význ. zvýšení rizika komorových

  arytmií, adekv výboje ICD a NSS



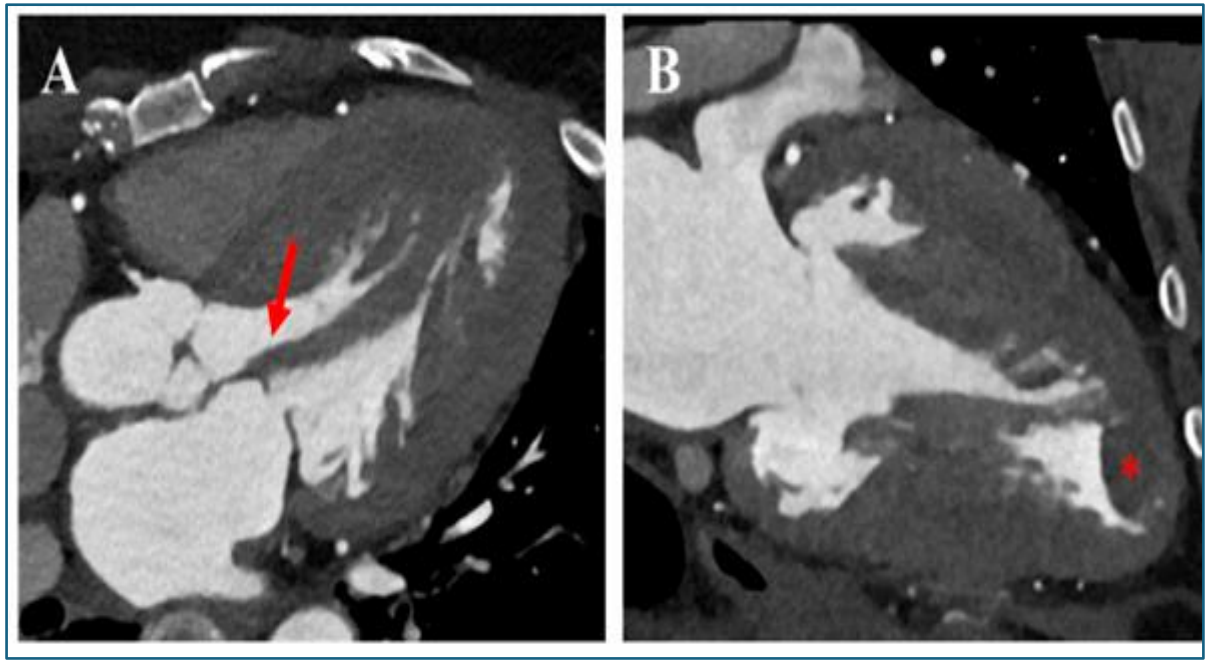
(Dalsania, J Clin Med, 2025)

Apikální aneurysma



(Arbelo, EHJ, 2023)

CT vyšetření



Indikováno u pacientů nevyšetřitelných echokardiograficky,
v případě nedostupnosti nebo KI MRI

(Dalsania, J Clin Med, 2025)

Validace ESC 2023 a ACC/AHA 2024 guidelines

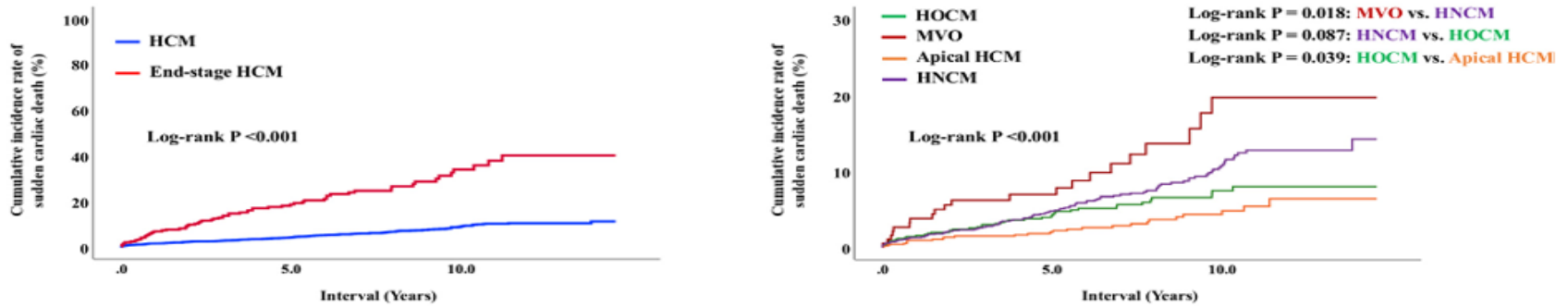
JACC: HEART FAILURE VOL. ■, NO. ■, 2025

ORIGINAL RESEARCH

Validation of Guideline Recommendation on Sudden Cardiac Death Prevention in Hypertrophic Cardiomyopathy

Masashi Amano, MD, PhD,[a] Hiroaki Kitaoka, MD, PhD,[b] Yusuke Yoshikawa, MD, PhD, MPH,[c] Yasushi Sakata, MD, PhD,[d] Kaoru Dohi, MD, PhD,[e] Yukichi Tokita, MD, PhD,[f] Takao Kato, MD, PhD,[g] Shouji Matsushima, MD, PhD,[h] Takeshi Kitai, MD, PhD,[a] Atsushi Okada, MD, PhD,[a] Yutaka Furukawa, MD, PhD,[i] Toshihiro Tamura, MD, PhD,[j] Akihiro Hayashida, MD,[k] Haruhiko Abe, MD,[l] Kenji Ando, MD,[m] Satoshi Yuda, MD, PhD,[n] Moriaki Inoko, MD, PhD,[o] Kazushige Kadota, MD, PhD,[p] Yukio Abe, MD, PhD,[q] Katsuomi Iwakura, MD, PhD,[r] Tetsuya Kitamura, MD, PhD,[s] Jun Masuda, MD, PhD,[t] Takahiro Ohara, MD, PhD,[u] Takashi Omura, MD, PhD,[v] Takashi Tanigawa, MD, PhD,[w] Kenji Nakamura, MD, PhD,[x] Kunihiro Nishimura, MD, PhD,[y] Chisato Izumi, MD, PhD,[z] the REVEAL-HCM Investigators

FIGURE 2 Kaplan-Meier Curves for SCD Events by HCM Phenotypes

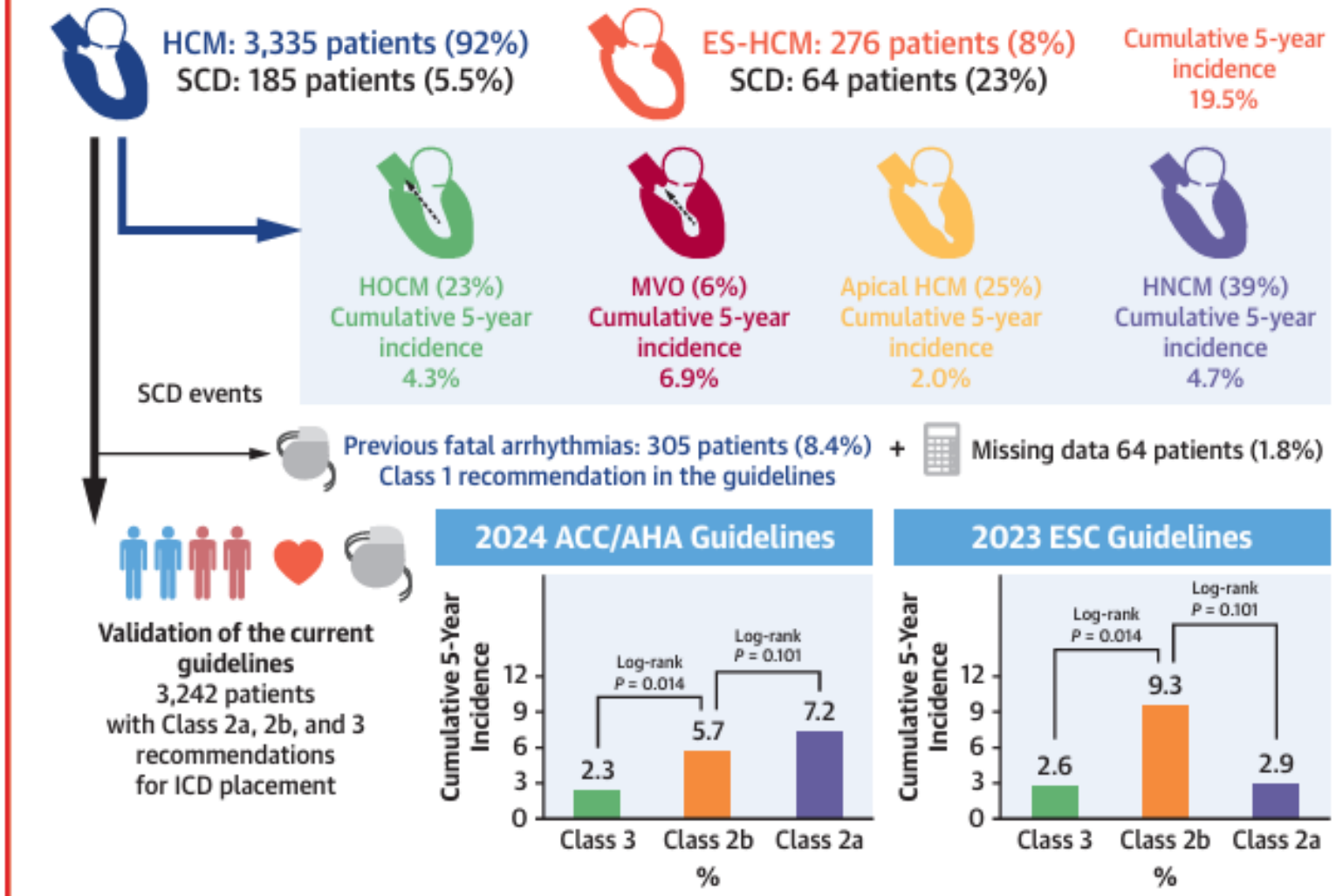


Interval	0 days	1-year	3-year	5-year	7-year	10-year
HCM						
N of patients at risk	3335	2853	2483	1936	1439	775
N of patients with SCD		42	76	108	138	174
Cumulative incidence rate		1.4%	2.6%	4.0%	5.7%	8.9%
End-stage HCM						
N of patients at risk	276	227	192	143	100	43
N of patients with SCD		17	33	43	52	60
Cumulative incidence rate		6.7%	13.6%	18.5%	24.5%	33.9%

Interval	0 days	1-year	3-year	5-year	7-year	10-year
HOCM						
N of patients at risk	838	738	649	503	365	192
N of patients with SCD		12	22	30	36	41
Cumulative incidence rate		1.5%	2.9%	4.3%	5.6%	7.4%
MVO						
N of patients at risk	206	167	136	107	74	29
N of patients with SCD		7	11	12	16	21
Cumulative incidence rate		3.8%	6.2%	6.9%	11.0%	19.7%
Apical HCM						
N of patients at risk	892	758	651	509	381	207
N of patients with SCD		7	11	14	18	24
Cumulative incidence rate		0.9%	1.4%	2.0%	2.8%	4.8%
HNCM						
N of patients at risk	1399	1190	1047	817	619	347
N of patients with SCD		16	32	52	68	88
Cumulative incidence rate		1.2%	2.6%	4.7%	6.8%	10.7%

Comparisons between HCM and ES-HCM (A) and among HCM phenotypes excluding ES-HCM (B). Abbreviations as in Figure 1.

CENTRAL ILLUSTRATION **A Large-Scale Retrospective Multicenter Cohort of Japanese Patients With HCM**



The incidence of SCD events could not be stratified well between Class 2a and 2b in HCM patients.

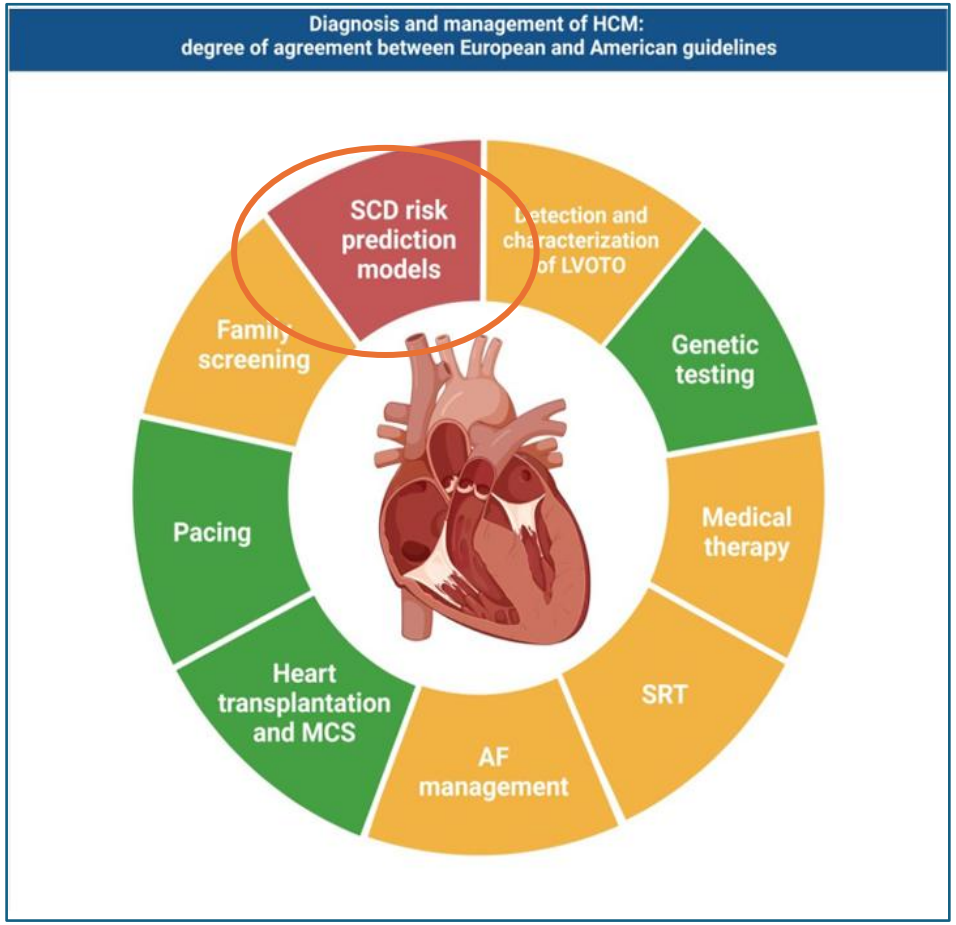
Amano M, et al. JACC Heart Fail. 2025;■(■):■-■.

Závěr

- Multimodalitní imaging nezbytný pro stratifikaci rizika NSS (TTE, MRI, ev CT)

- Follow-up TEE á 1-2 roky
- Follow-up MRI á 3-5 let
- 12 –svodové EKG,
  24/48 h EKG Holter á 1-2 roky

- Stratifikační systémy → identifikace vysoce rizikových pacientů → ICD



(Aimo, Heart Failure Rev, 2025)


FAKULTNÍ NEMOCNICE U SV. ANNY

-
-