Restriktivní kardiomyopatie

Jan Krejčí

FAKULTNÍ NEMOCNICE U SV. ANNY V BRNĚ

I. INTERNÍ KARDIOANGIOLOGICKÁ KLINIKA FNUSA A LF MU

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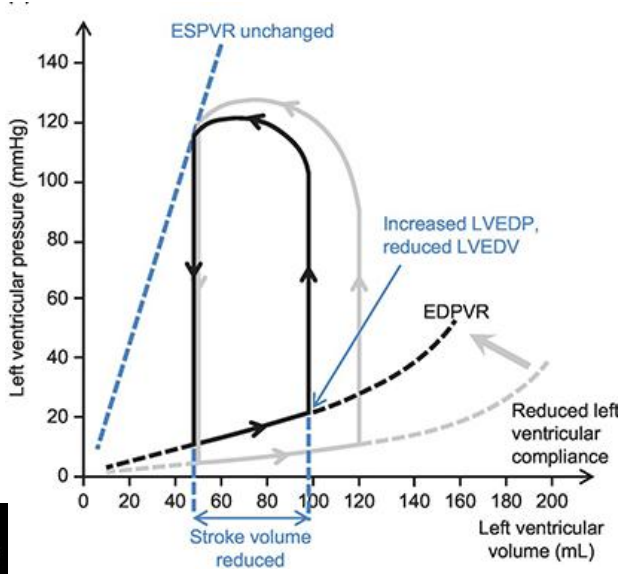
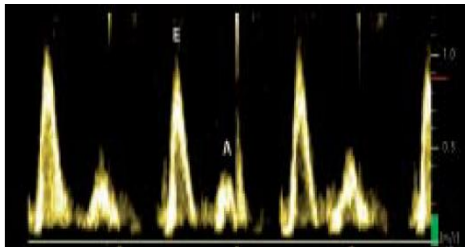
Co o restriktivní kardiomyopatii léta víme…

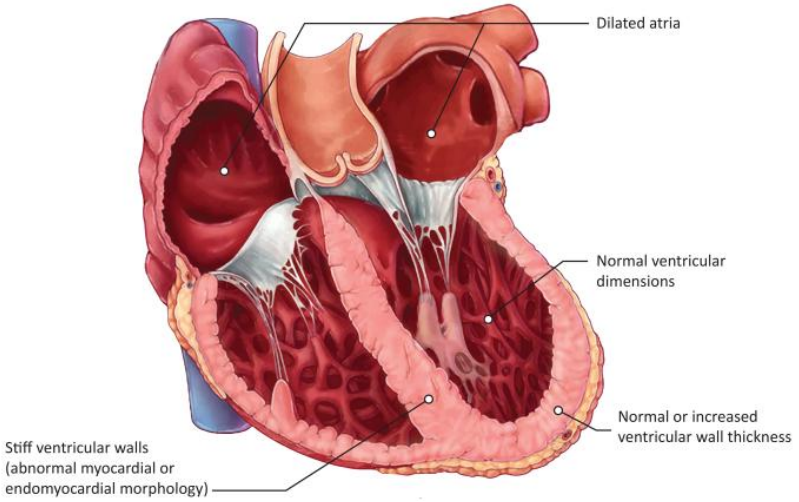
- nejméně častá KMP s nejhorší prognózou
- je charakterizovaná restriktivní hemodynamikou
- typickým rysem je zvýšená tuhost myokardu (snížená kompliance), která může mít původ v myokardu i v endokardu





Co o restriktivní kardiomyopatii léta víme…

- morfologicky jde o onemocnění s normální (či téměř normální) LVEF a normálními/sníženými objemy jedné nebo obou komor
- tloušťka stěn LK by neměla být zvýšená (či jen mírně)
- je přítomna dilatace obou síní



Širší definice RCM dle C. Rapezziho

Restrictive cardiomyopathy: definition and diagnosis

Proposal for a new definition and classification of restrictive cardiomyopathy

The approach we followed was first of all to refine and enhance the nosographic aspect of the classification and, subsequently, to provide useful insights for the diagnosis of individual diseases. The definition of RCM can be slightly modified as follows: RCM is characterized by the coexistence of persistent restrictive pathophysiology, commonly with atrial dilatation, and nondilated ventricles, regardless of ventricular wall thickness and systolic function. Several forms of RCM are predominantly due to endocardial involvement, leading to a similar haemodynamic patterns as for isolated myocardial diseases.

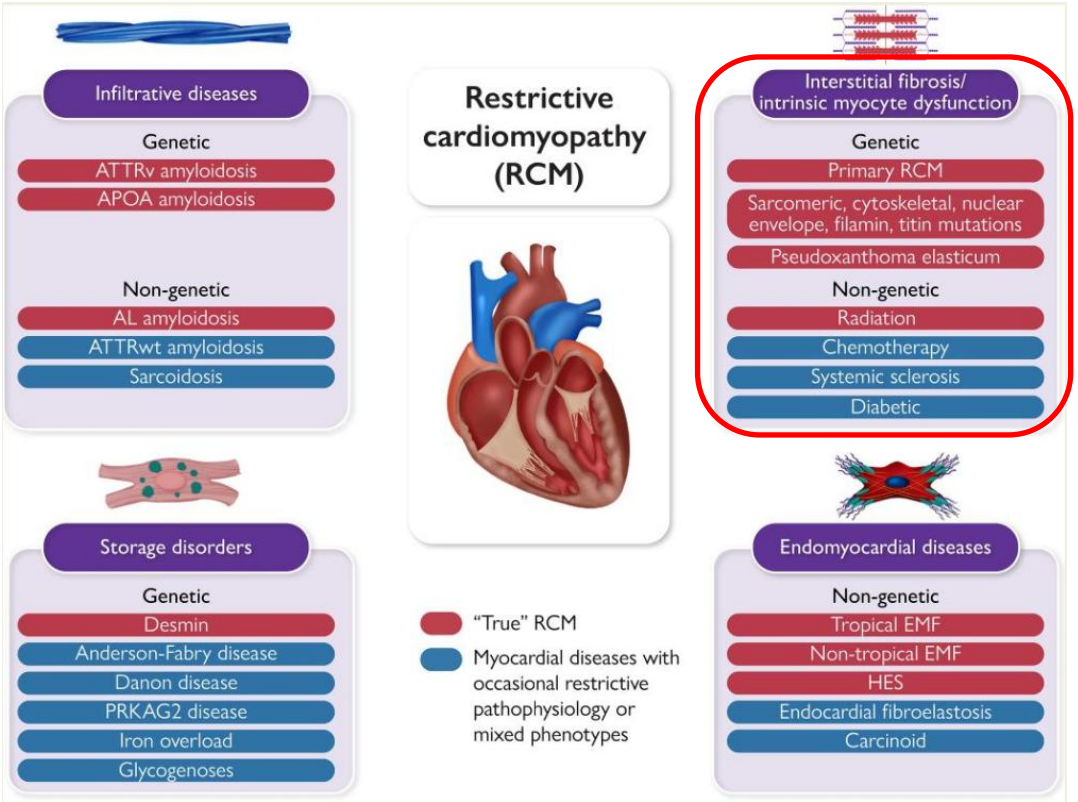
European Heart Journal (2022) **43**, 4679–4693

Dělení RCM podle „místa postižení“

Restrictive cardiomyopathy: definition and diagnosis

European Heart Journal (2022) **43**, 4679–4693

Postižení myocytů / intersticiální fibróza



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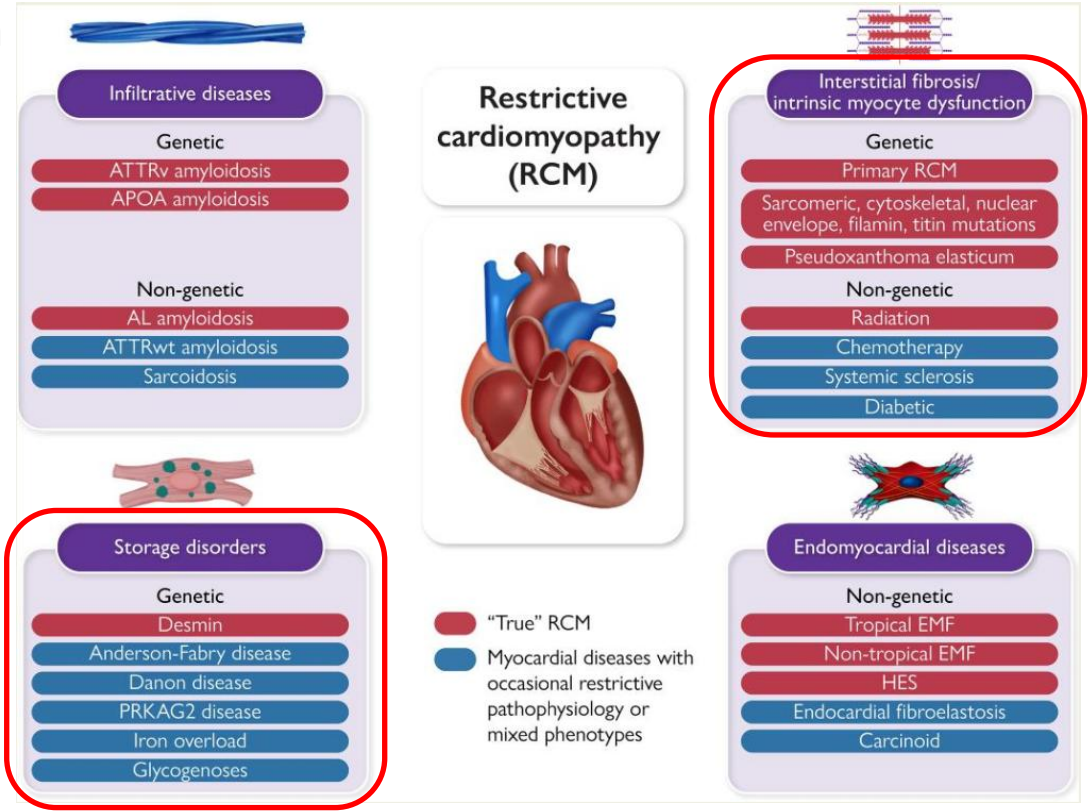

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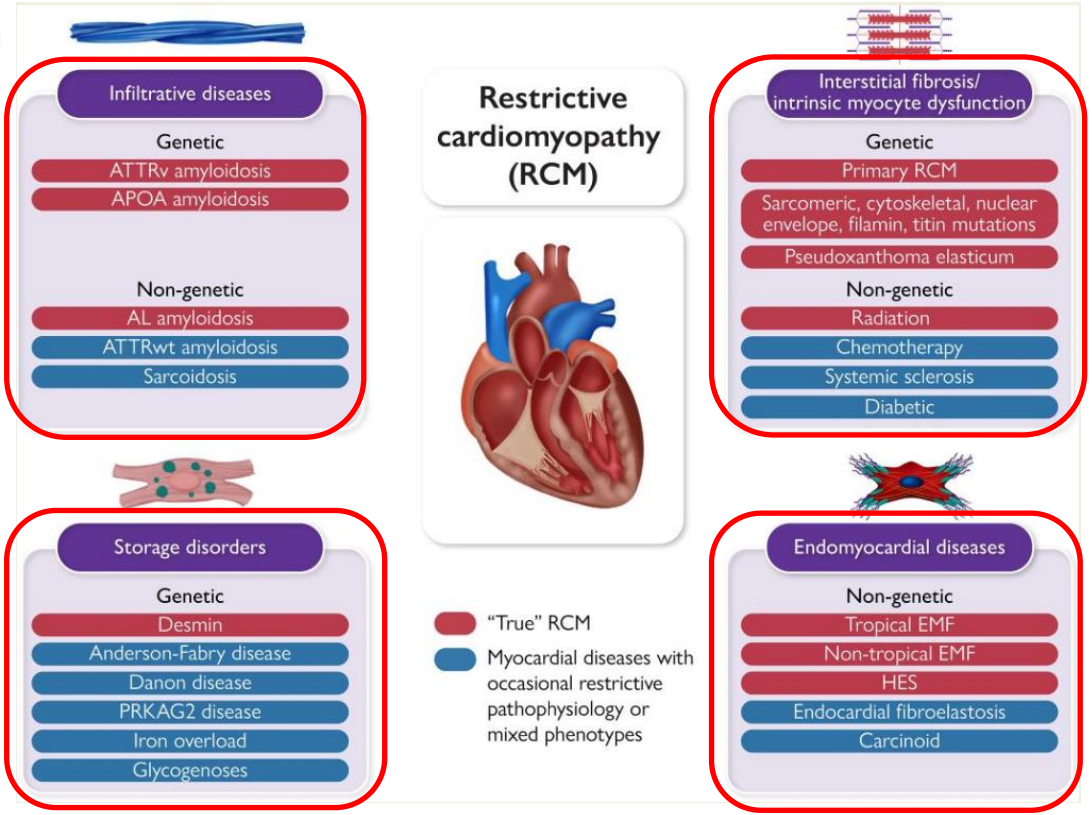

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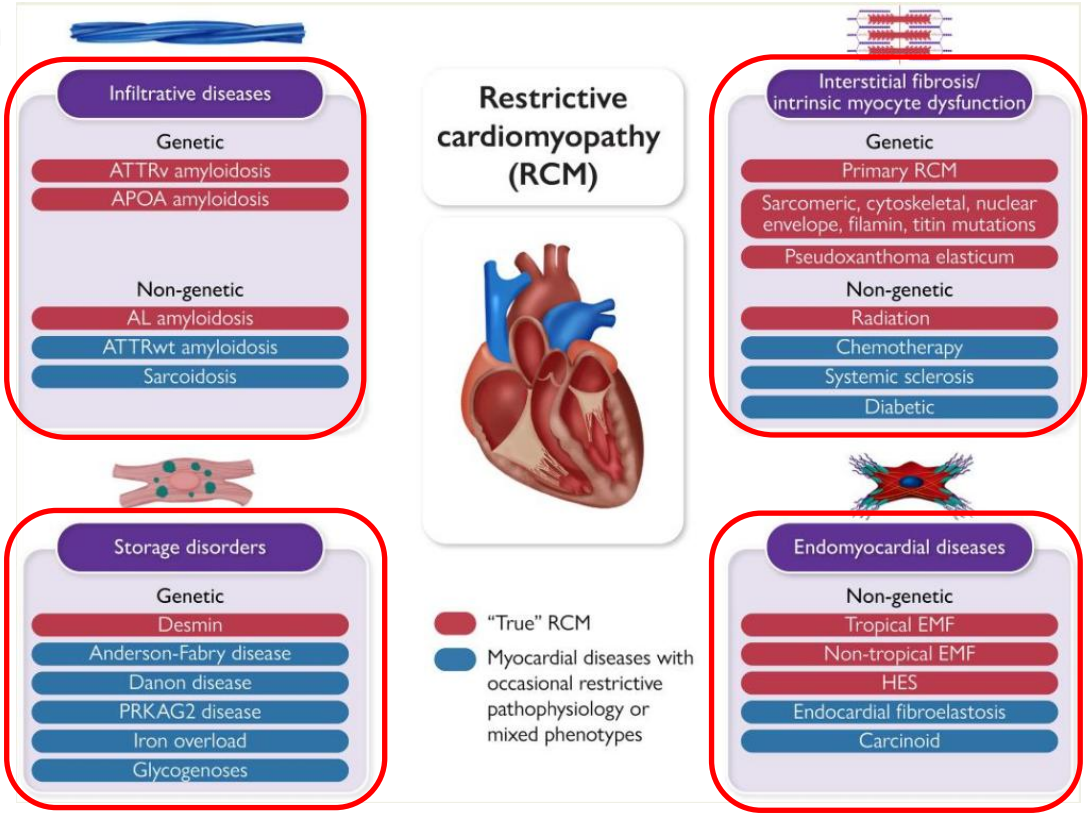
Postižení endo(myo)kardu



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Restriktivní kardiomyopatie

ESC European Society of Cardiology
European Heart Journal (2023) 00, 1–124
https://doi.org/10.1093/eurheartj/ehad194

ESC GUIDELINES

2023 ESC Guidelines for the management of cardiomyopathies

Developed by the task force on the management of cardiomyopathies of the European Society of Cardiology (ESC)

Eur Heart J. 2023 Oct 1;44(37):3503-3626.

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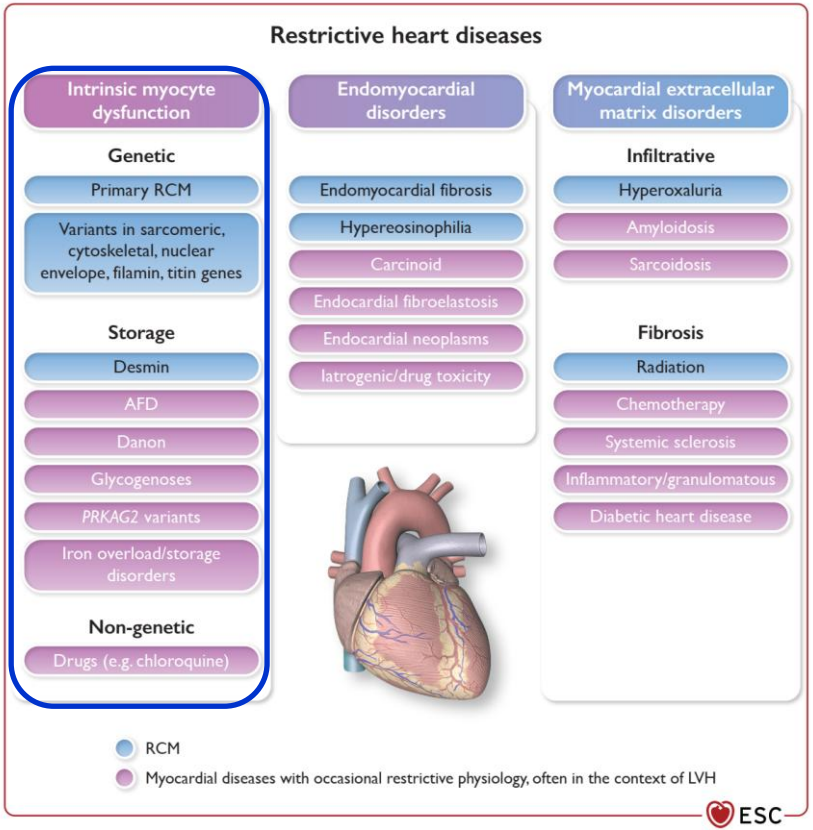

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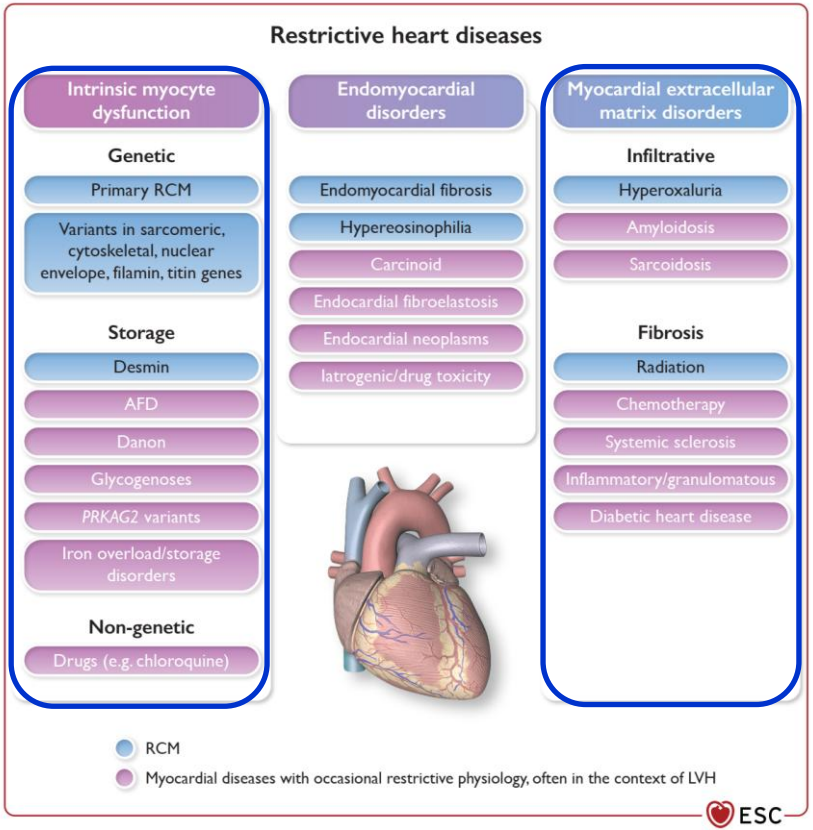

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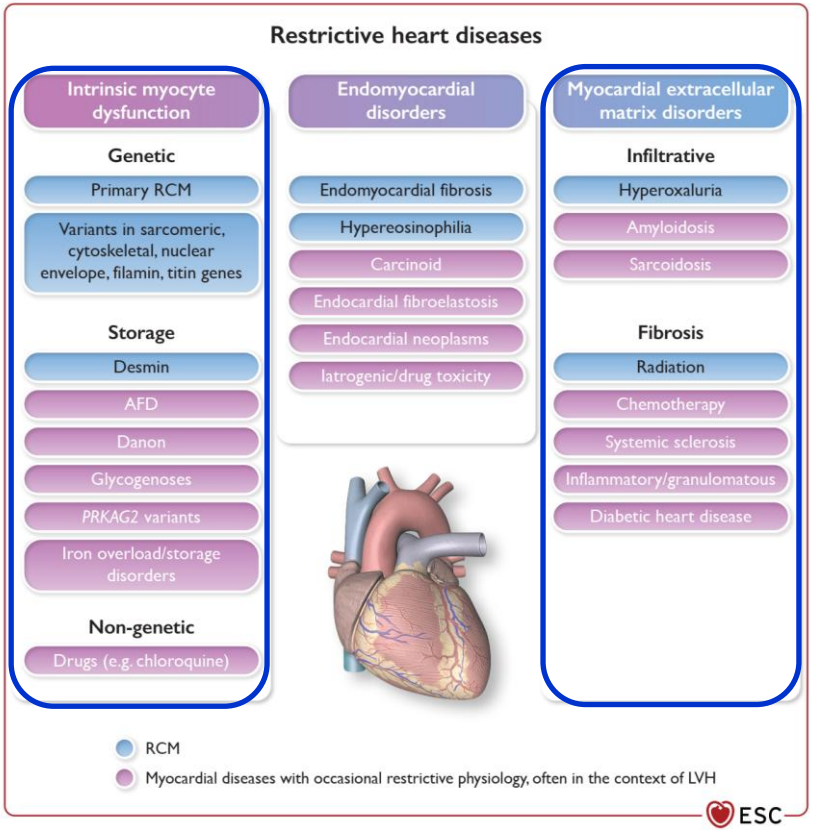

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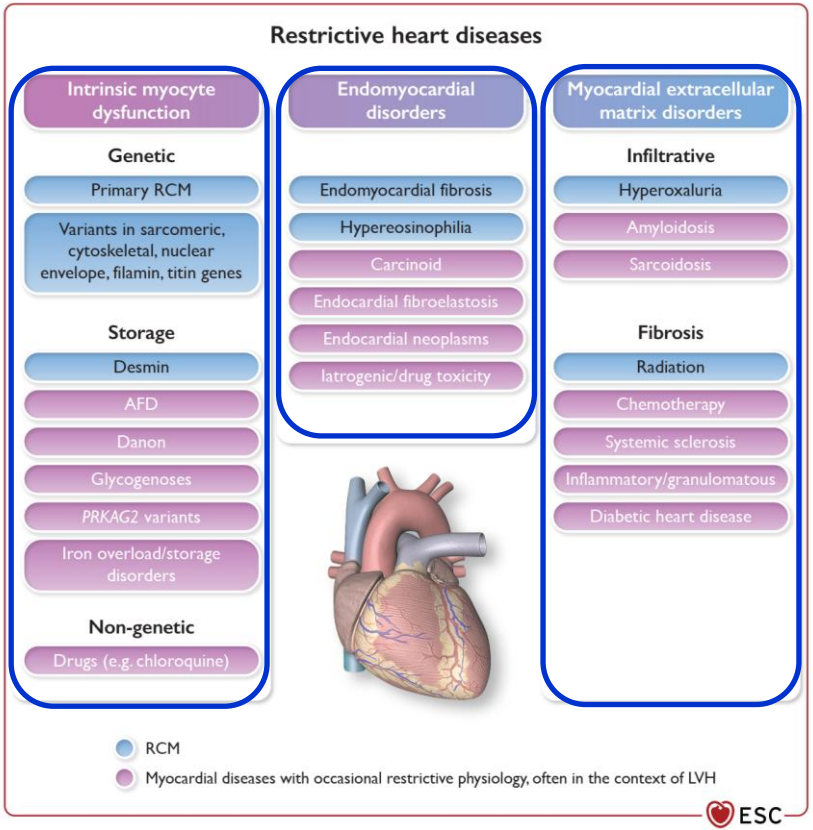

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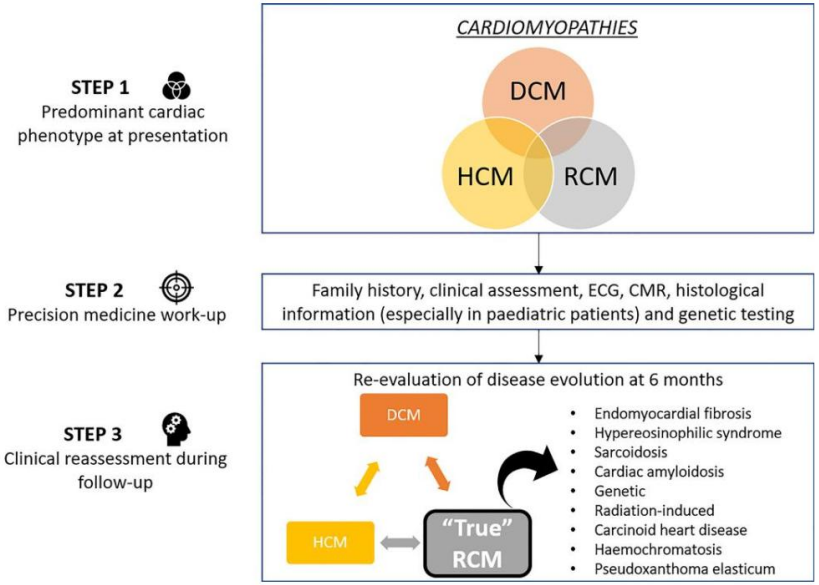
RCM

- RCM je heterogenní skupina kardiomyopatií
- Genotypový překryv s řadou jiných KMP
- Pohyblivý cíl…vývoj klinického obrazu v čase nebo podle dalších okolností (např. volumové situace) – potřeba re-evaluace

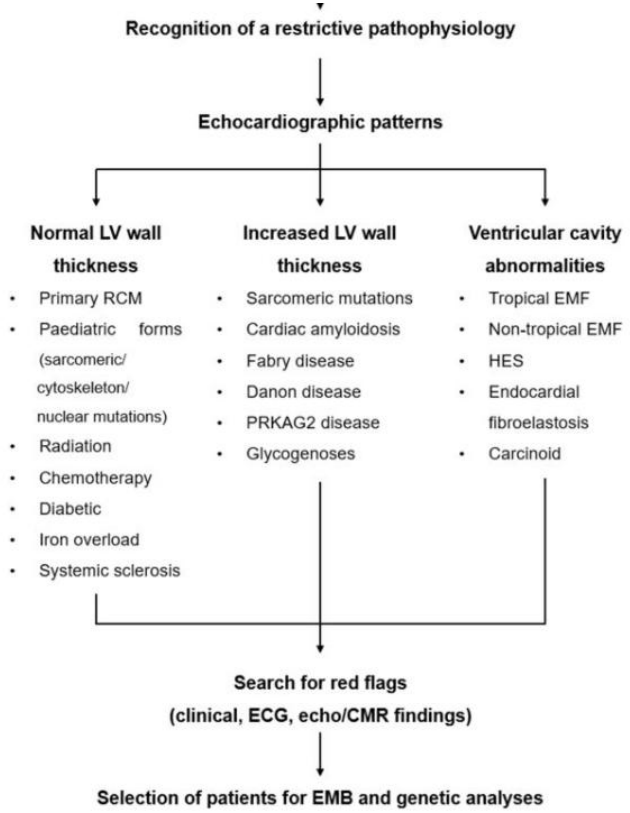


European Heart Journal (2022) **43**, 4679–4693

RCM

- Nesmírně heterogenní skupina kardiomyopatií
- Genotypový překryv s řadou jiných kardiomyopatií
- Pohyblivý cíl…vývoj klinického obrazu v čase nebo podle dalších okolností (např. volumové situace)

- Velmi náročná na diagnostiku a diferenciální dg.



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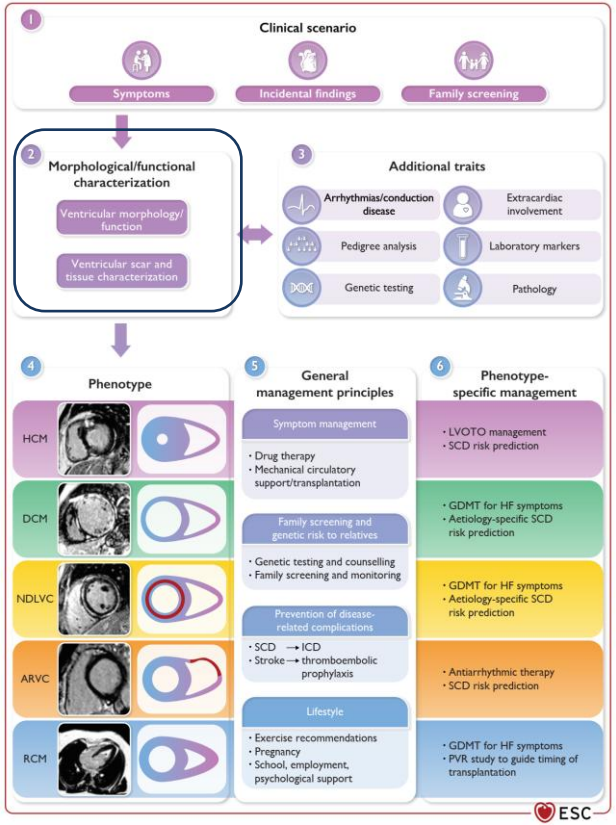
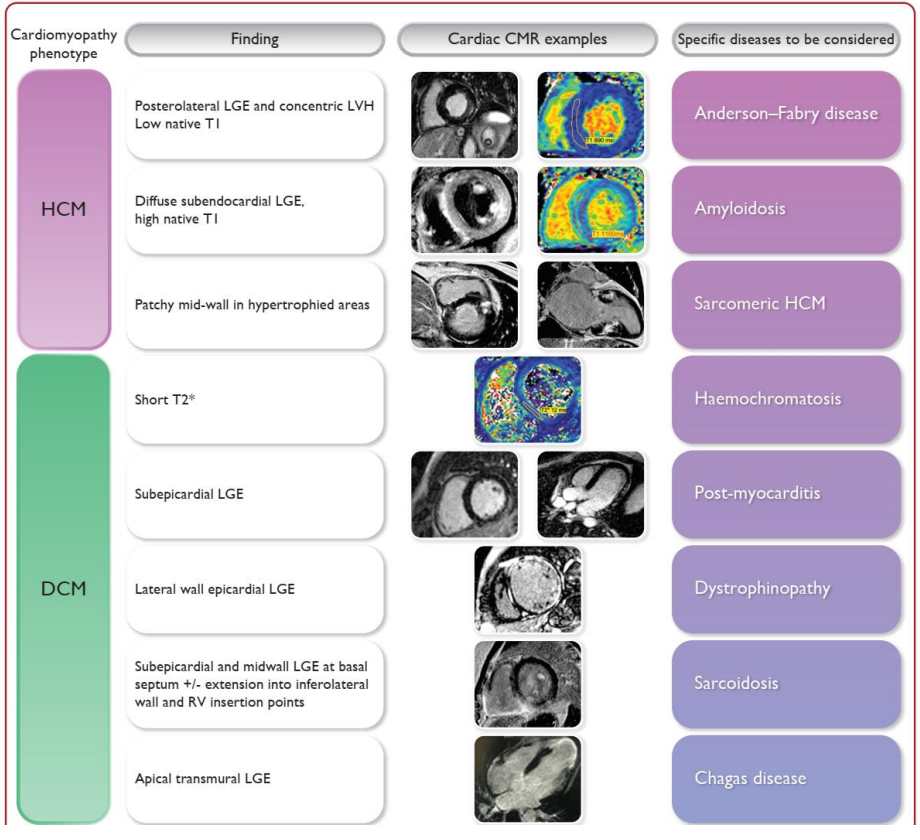
Zásady přístupu k diagnostice KMP dle ESC guidelines

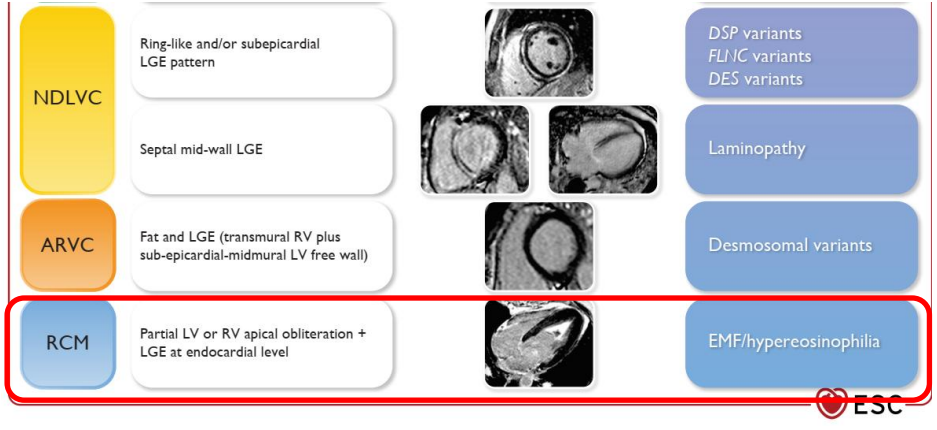
Morphological/functional characterization

- Ventricular morphology/ function
- Ventricular scar and tissue characterization



Heterogenita RCM – přínos CMR





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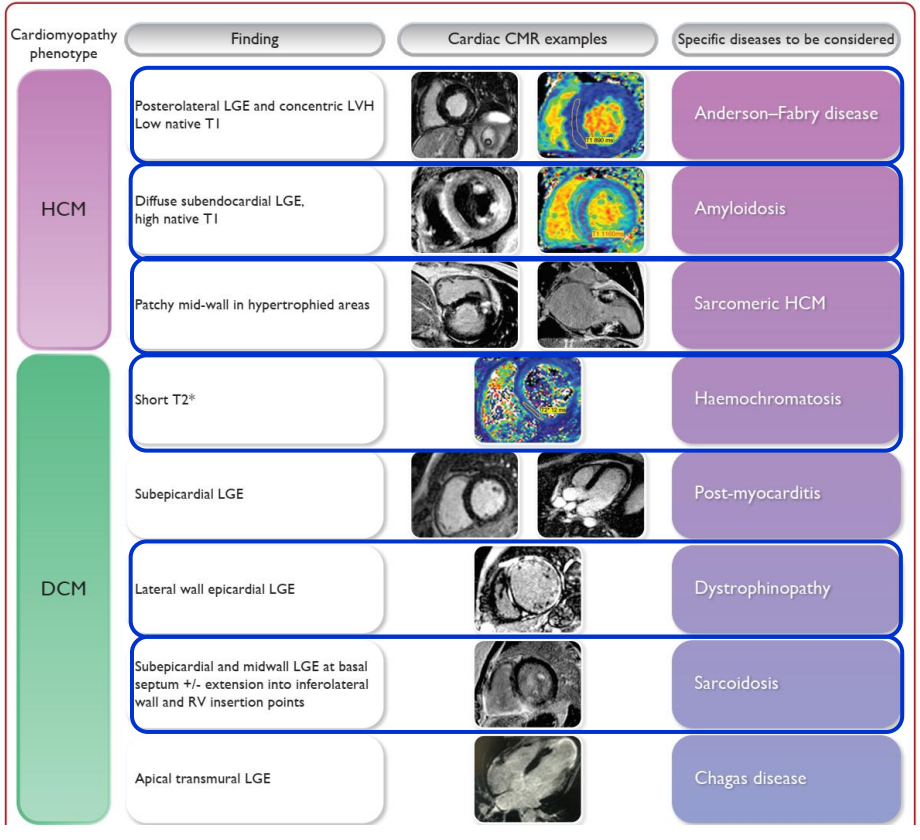

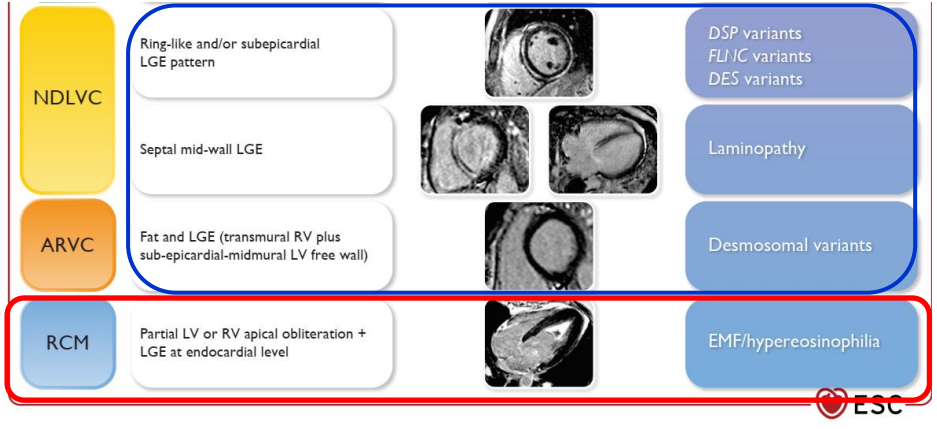

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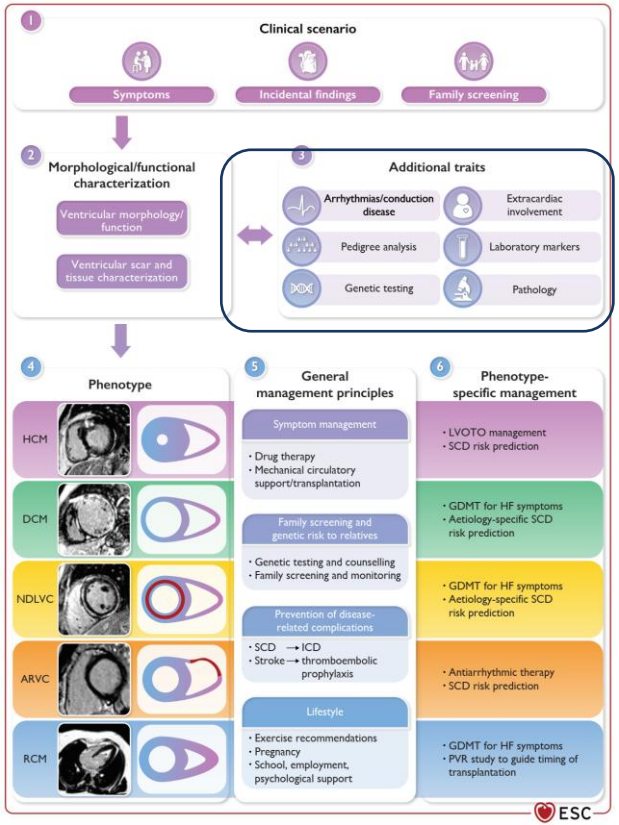
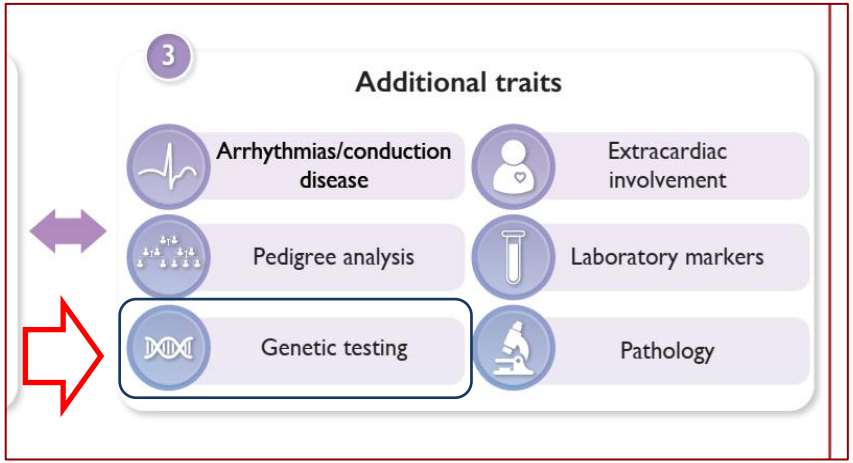
Diagnostika KMP a genetické testování





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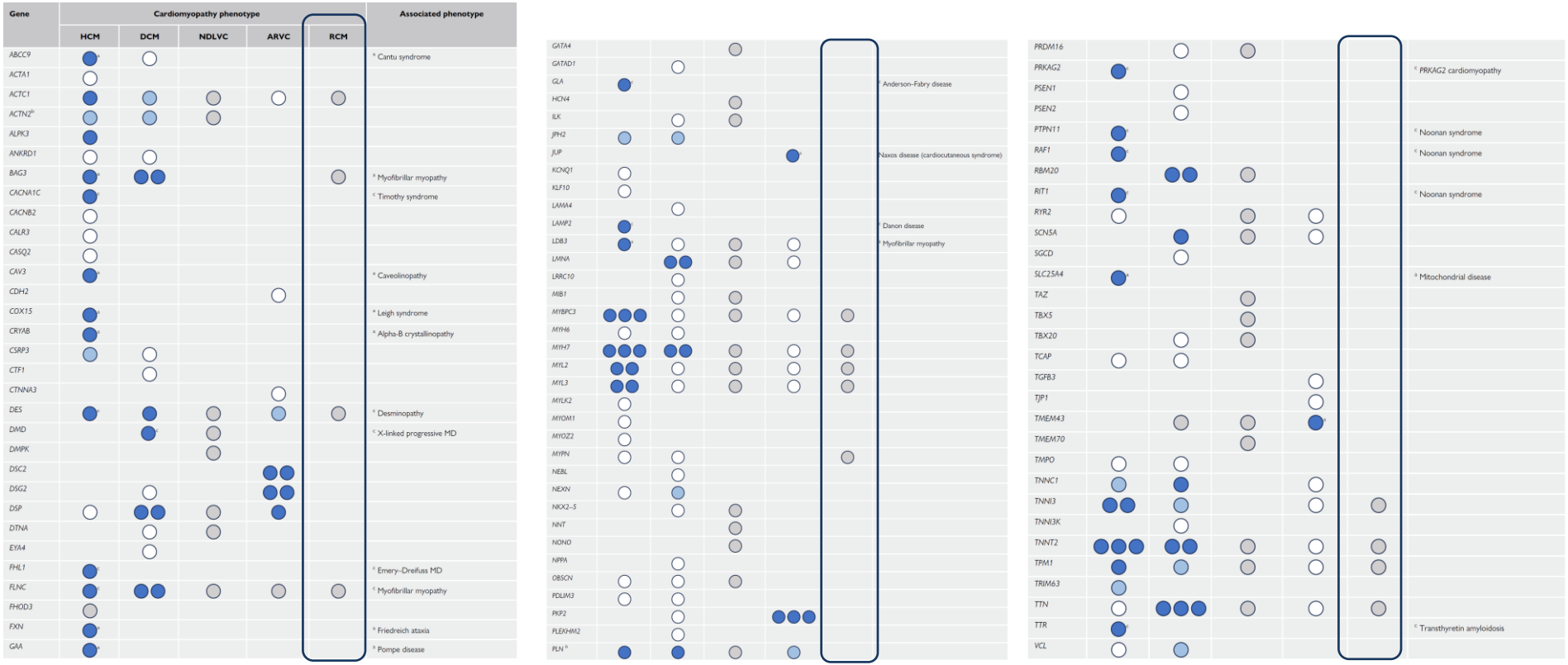
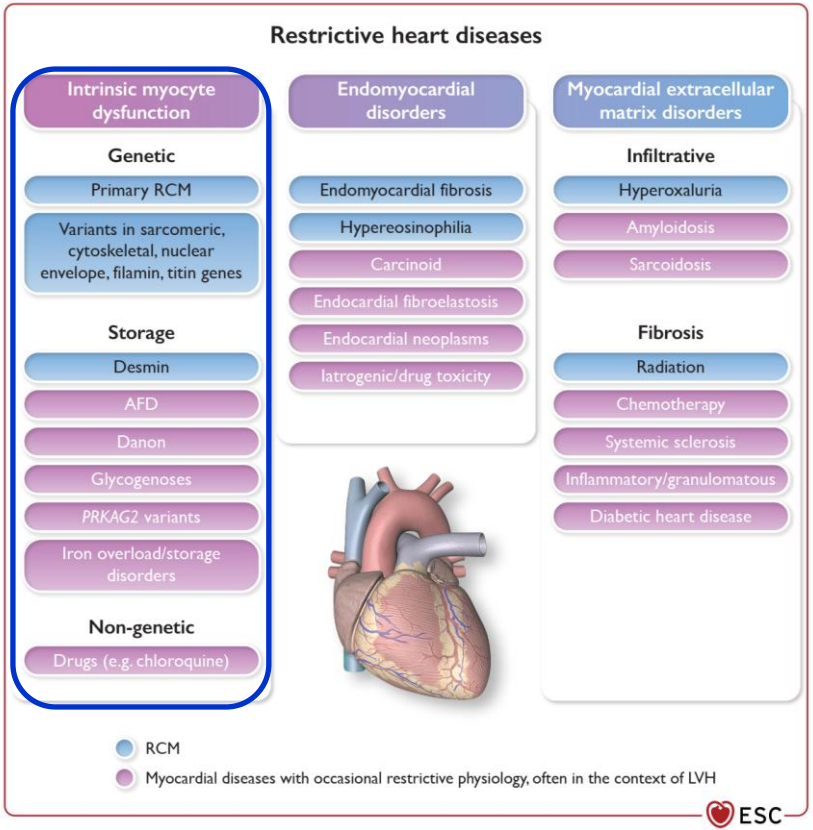
Role genetického testování u RCM



Role genetického testování v dif. dg. HCM/RCM

Gene	Cardiomyopathy phenotype					Associated phenotype
	HCM	DCM	NDLVC	ARVC	RCM	
GLA	 [c]					[c] Anderson–Fabry disease
LAMP2	 [c]					[c] Danon disease
TTR	 [c]					[c] Transthyretin amyloidosis

Restriktivní kardiomyopatie - I



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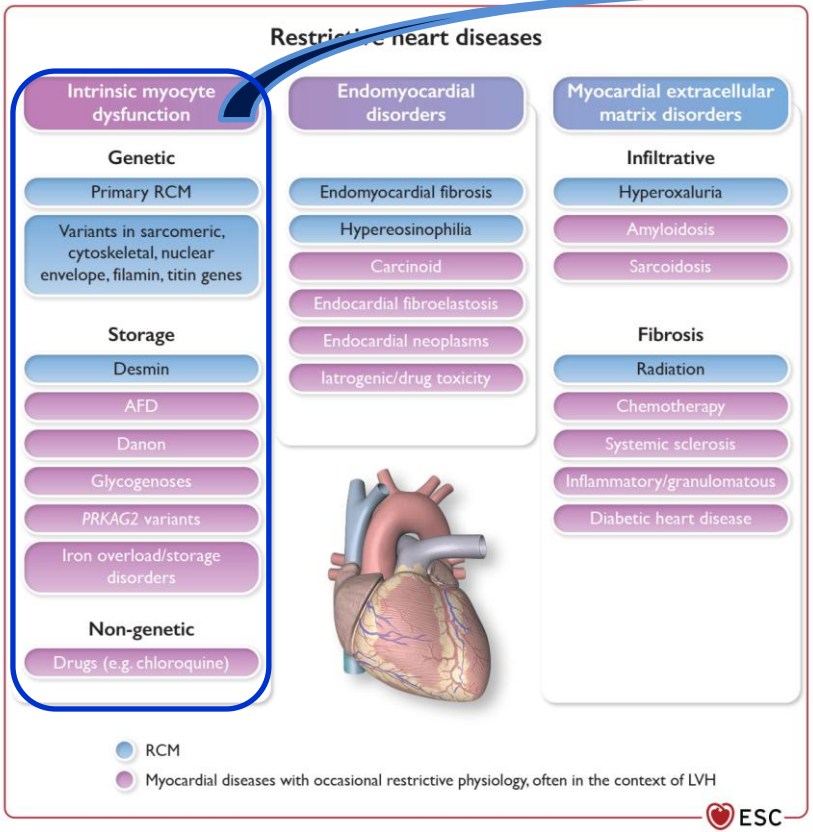

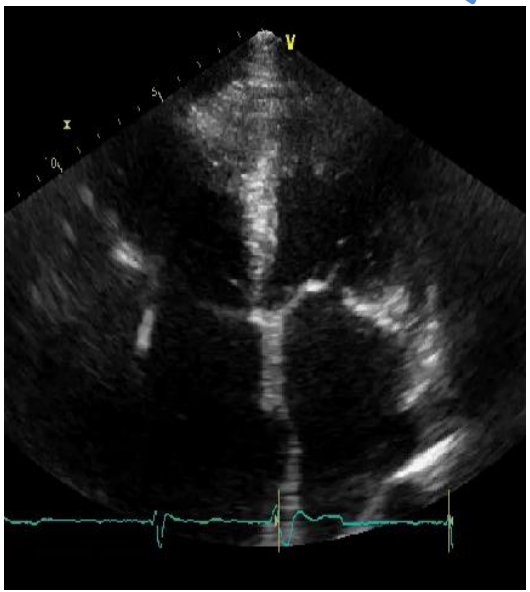

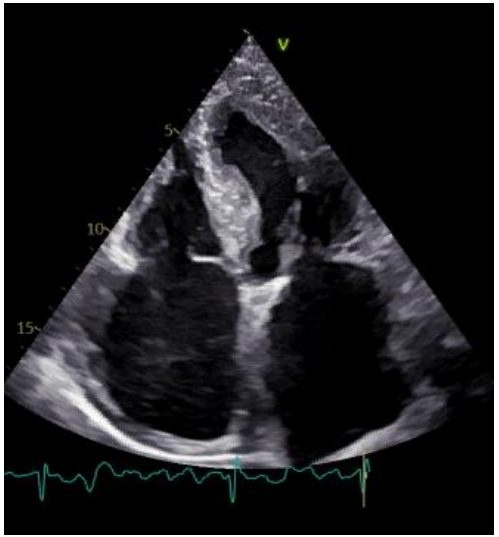

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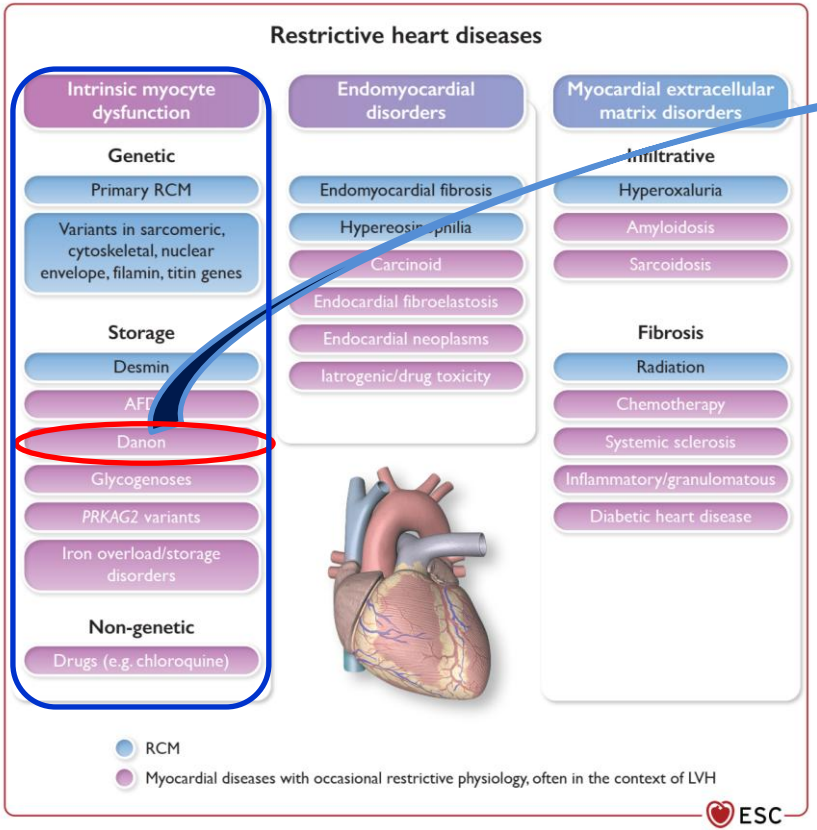
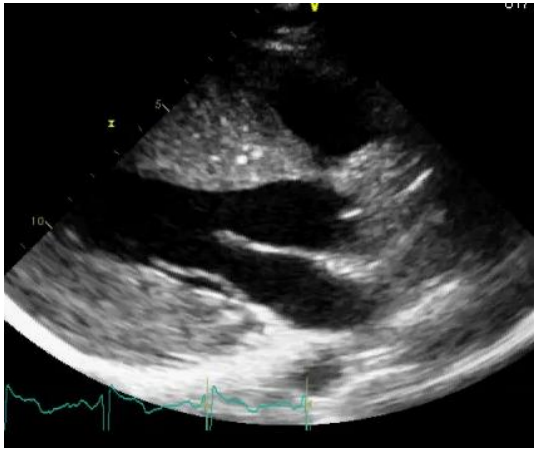
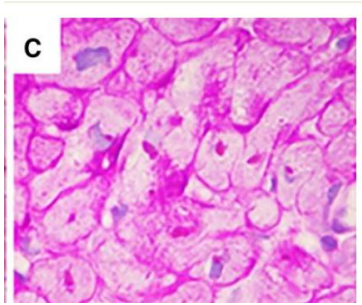
Danonova nemoc - RCM nebo HCM?







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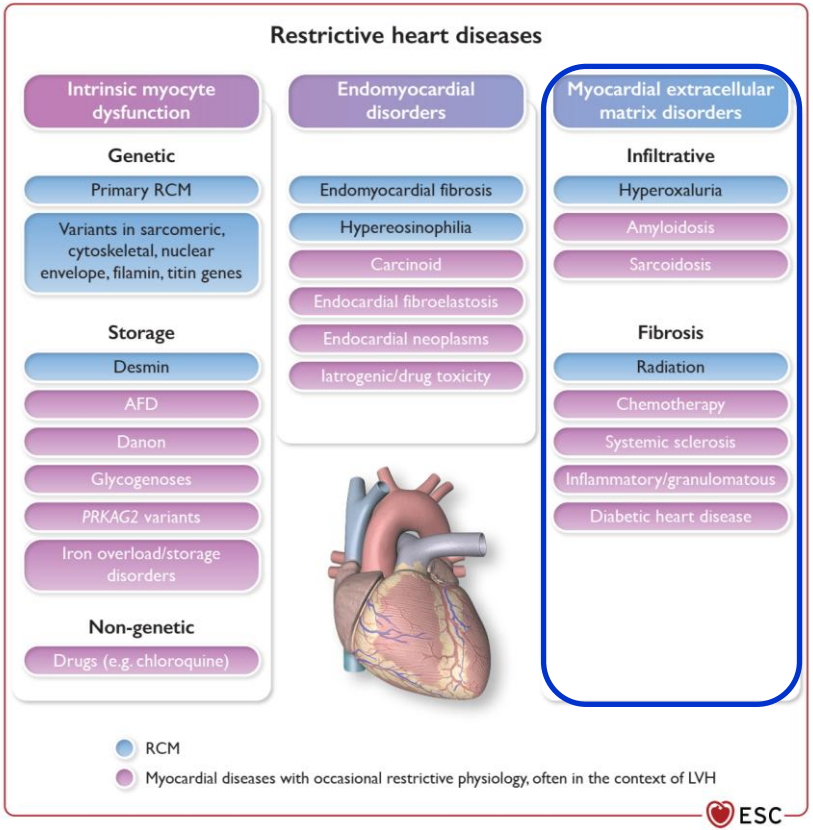
Restriktivní kardiomyopatie - II



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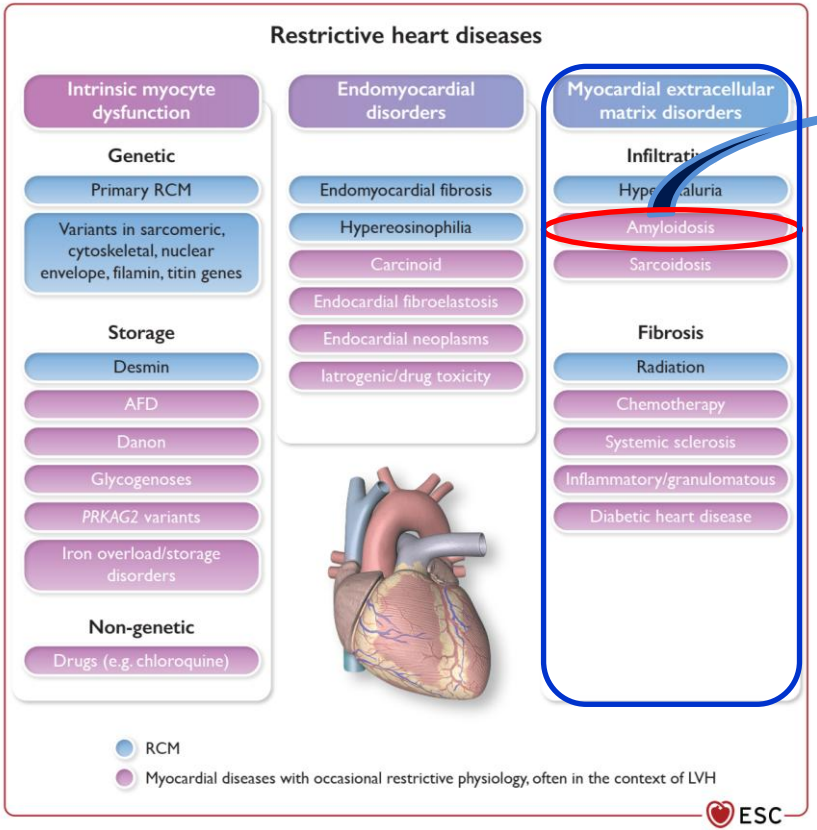
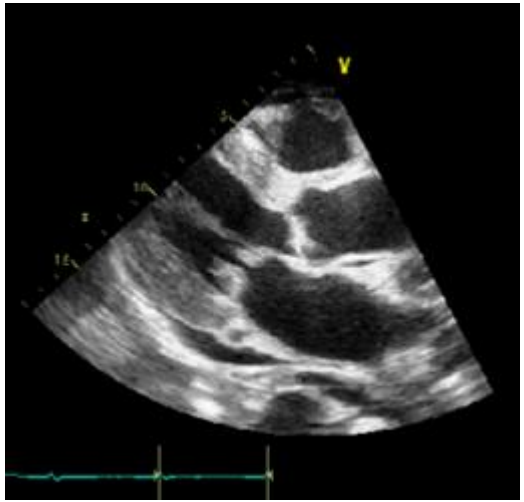
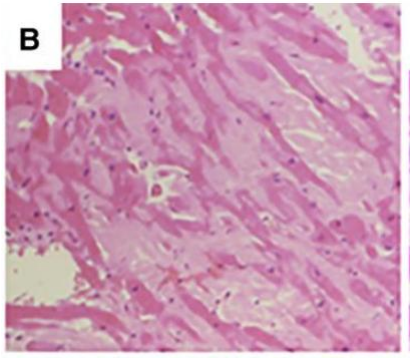
Srdeční amyloidóza - RCM nebo HCM?







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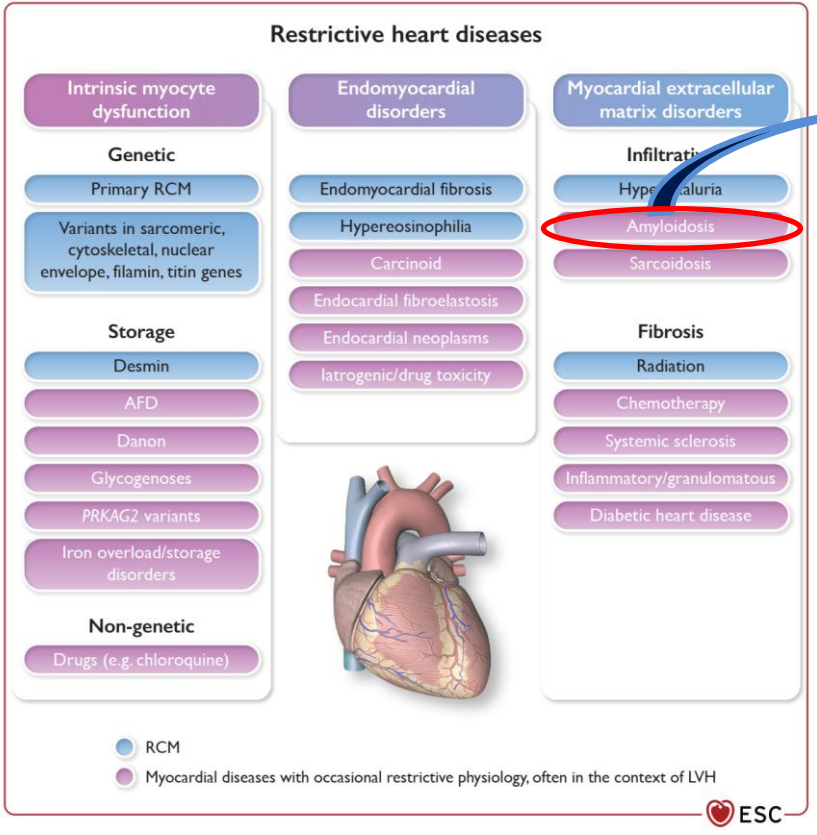
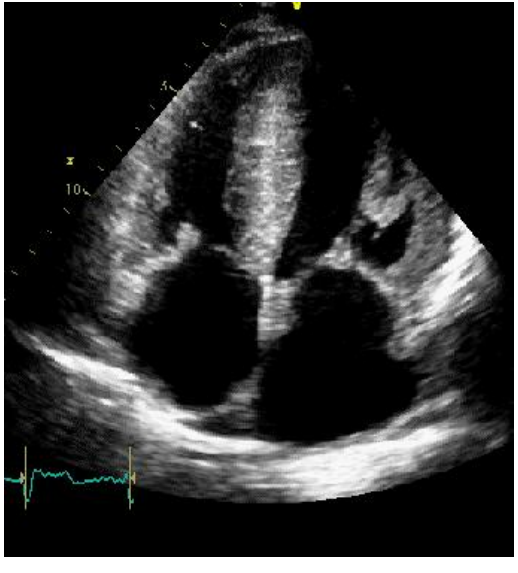
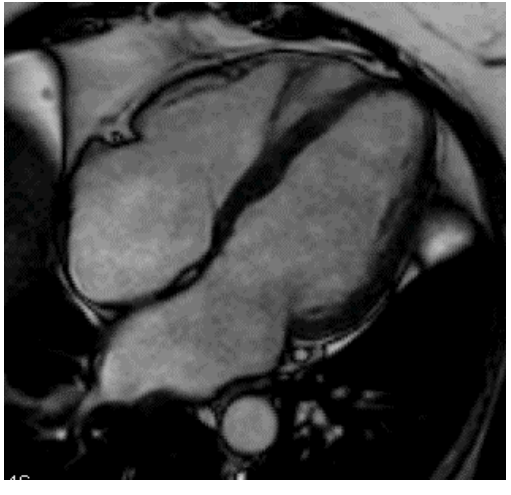
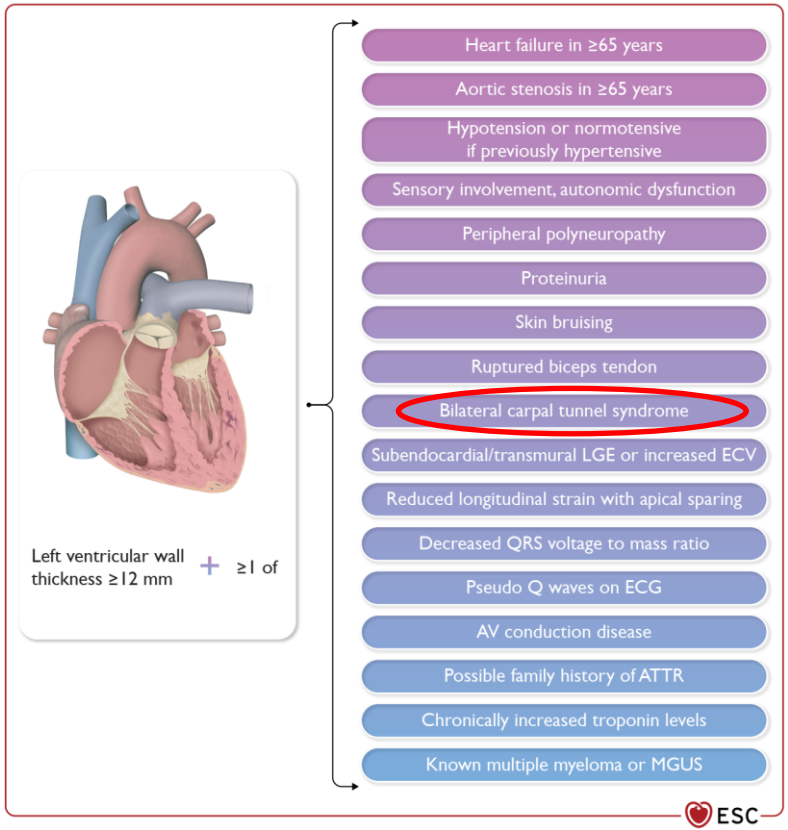
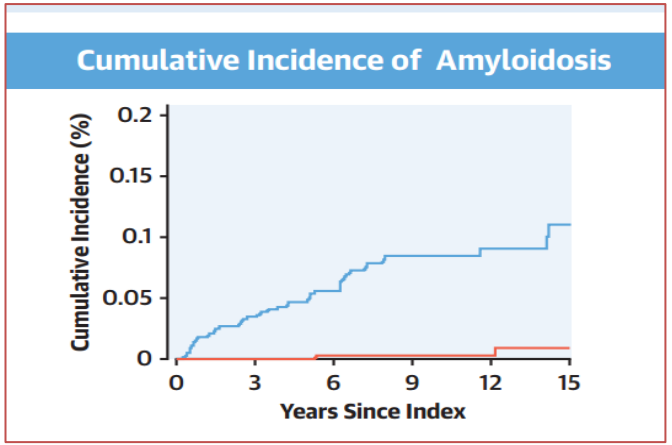






Dif. dg. RCM (HCM) – srdeční amyloidóza





Fosbol et al. JACC 2019

Syndrom karpálního tunelu

- předchází dg 5-15 let, často oboustranný až u 50% nemocných
- **12x vyšší riziko srdeční amyloidosy**

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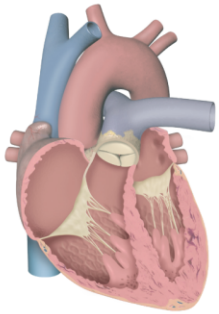
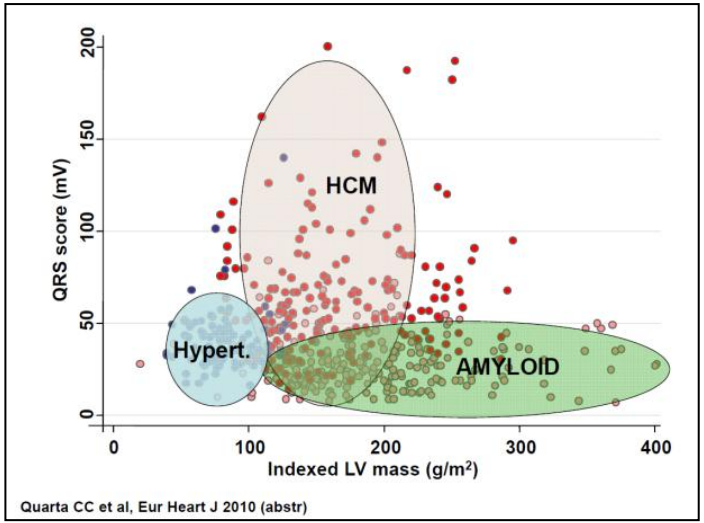
Dif. Dg. RCM (HCM) – srdeční amyloidóza



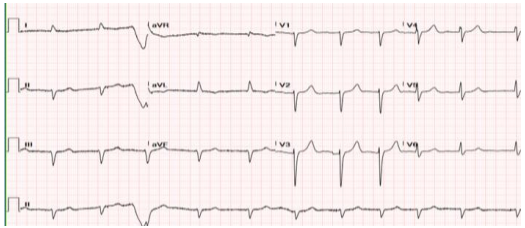
Left ventricular wall thickness ≥12 mm + ≥1 of

- 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

ESC



Quarta CC et al, Eur Heart J 2010 (abstr)



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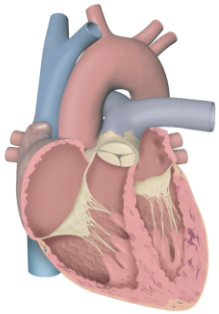
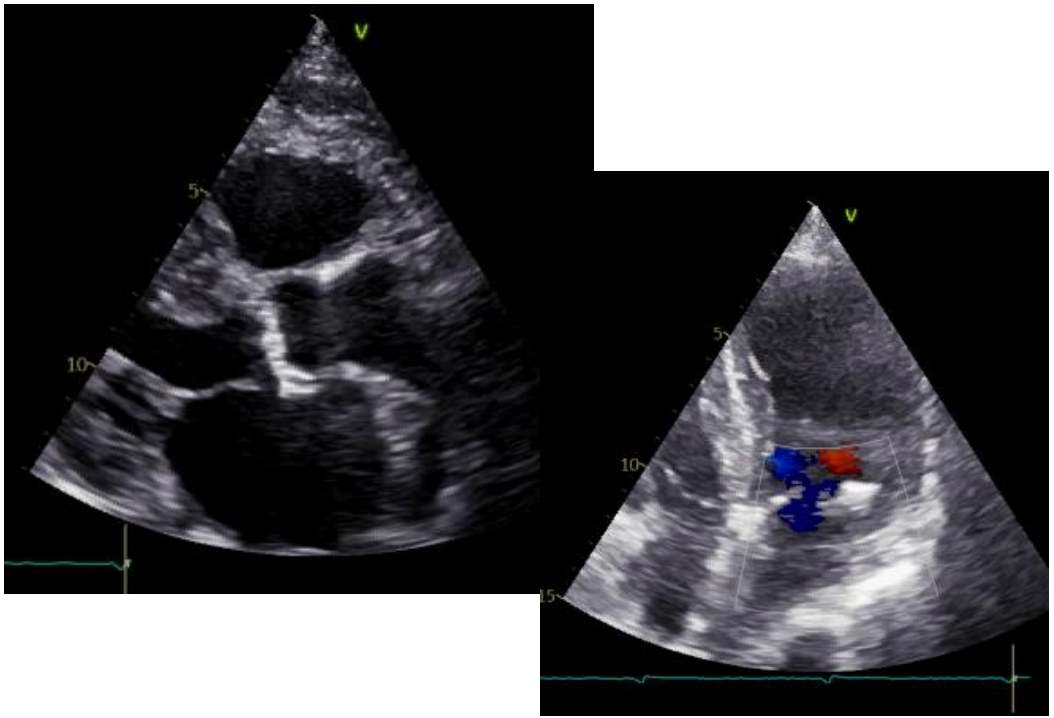


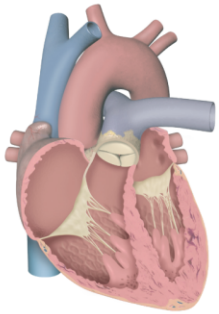
Left ventricular wall thickness ≥12 mm + ≥1 of

- 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

ESC



Dif. Dg. RCM (HCM) – srdeční amyloidóza



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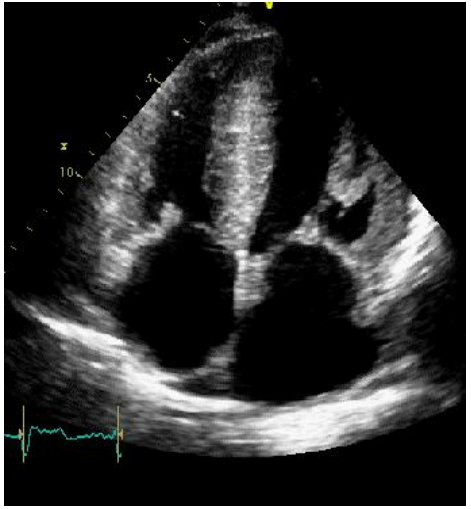

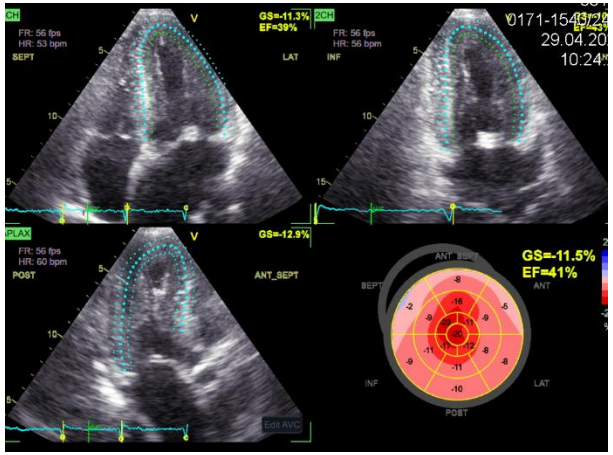

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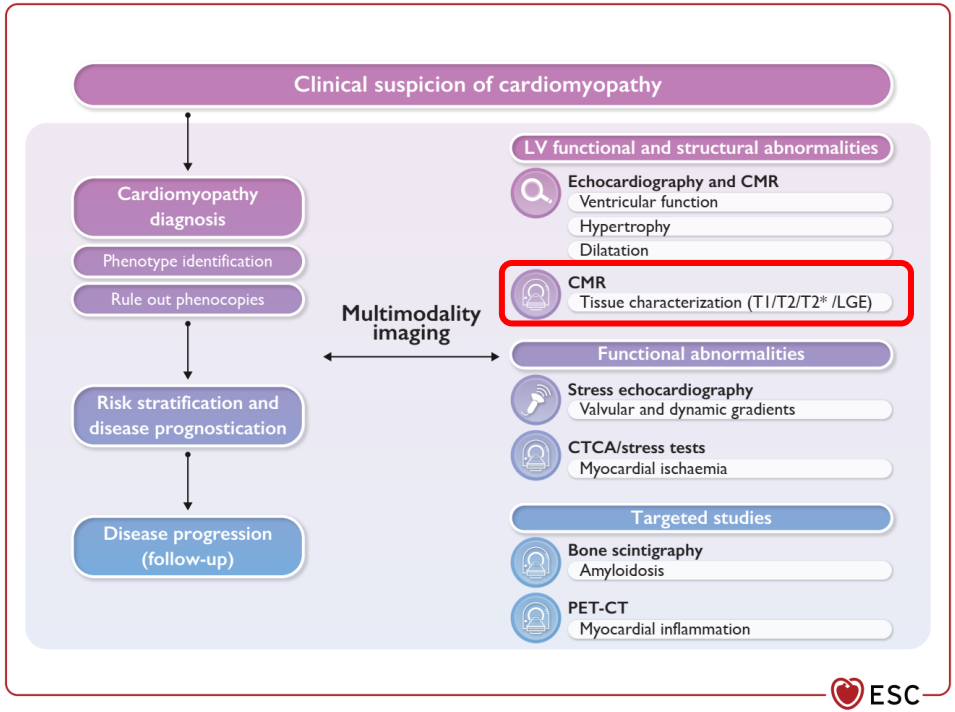
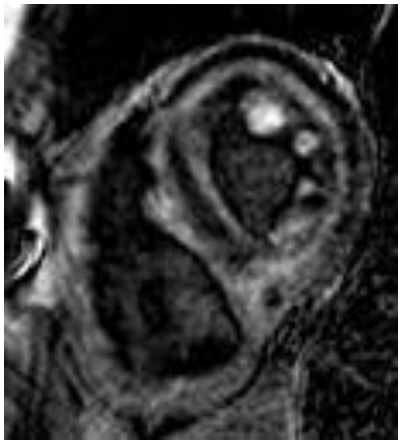
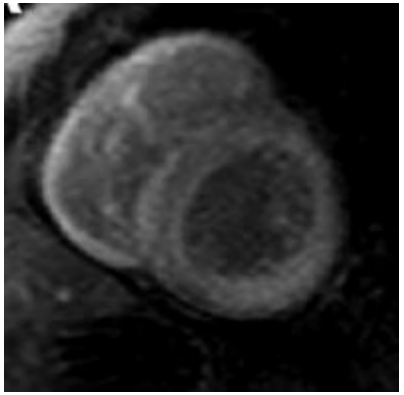
Diagnostika a diferenciální dg. RCM/HCM a srdeční amyloidózy

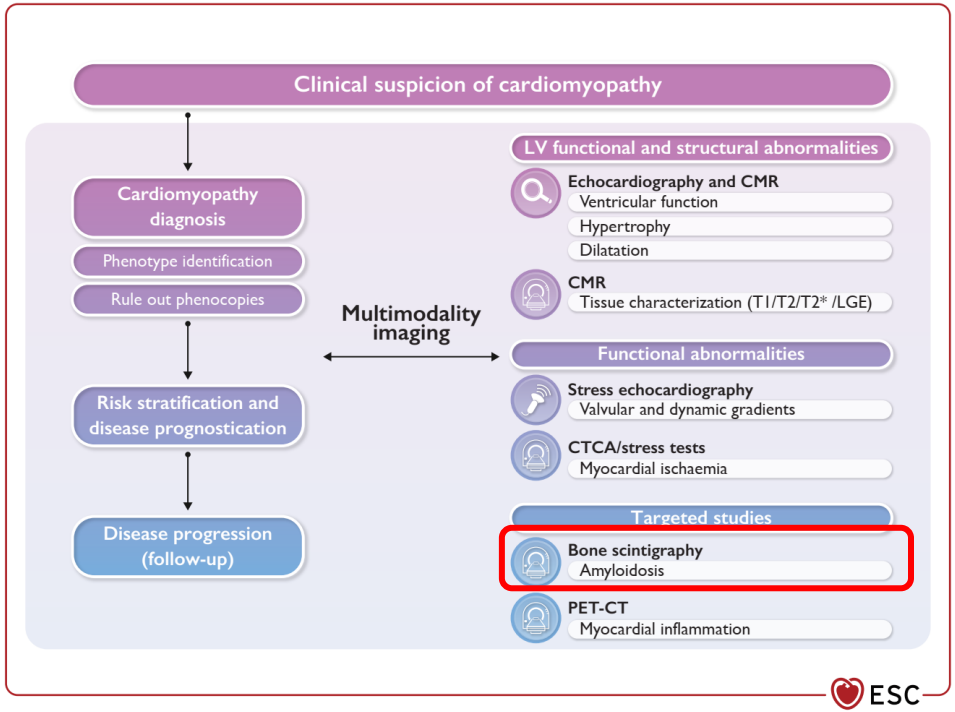


Recommendations	Class[a]	Level[b]
Contrast-enhanced CMR is recommended in patients with cardiomyopathy at initial evaluation.[10,90,116,119–143]	I	B

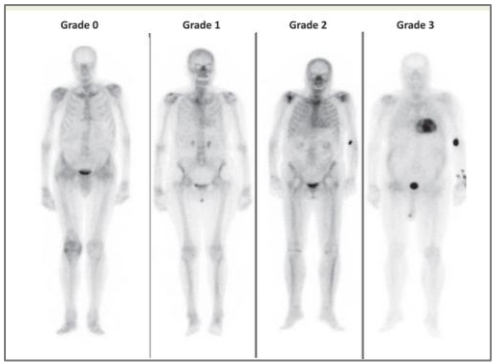




Diagnostika a diferenciální dg. RCM/HCM a srdeční amyloidózy



Recommendations	Class[a]	Level[b]
DPD/PYP/HMDP bone-tracer scintigraphy is recommended in patients with suspected ATTR-related cardiac amyloidosis to aid diagnosis.[166–168]	I	B



FAKULTNÍ NEMOCNICE U SV. ANNY V BRNĚ

I. INTERNÍ KARDIOANGIOLOGICKÁ KLINIKA FNUSA A LF MU

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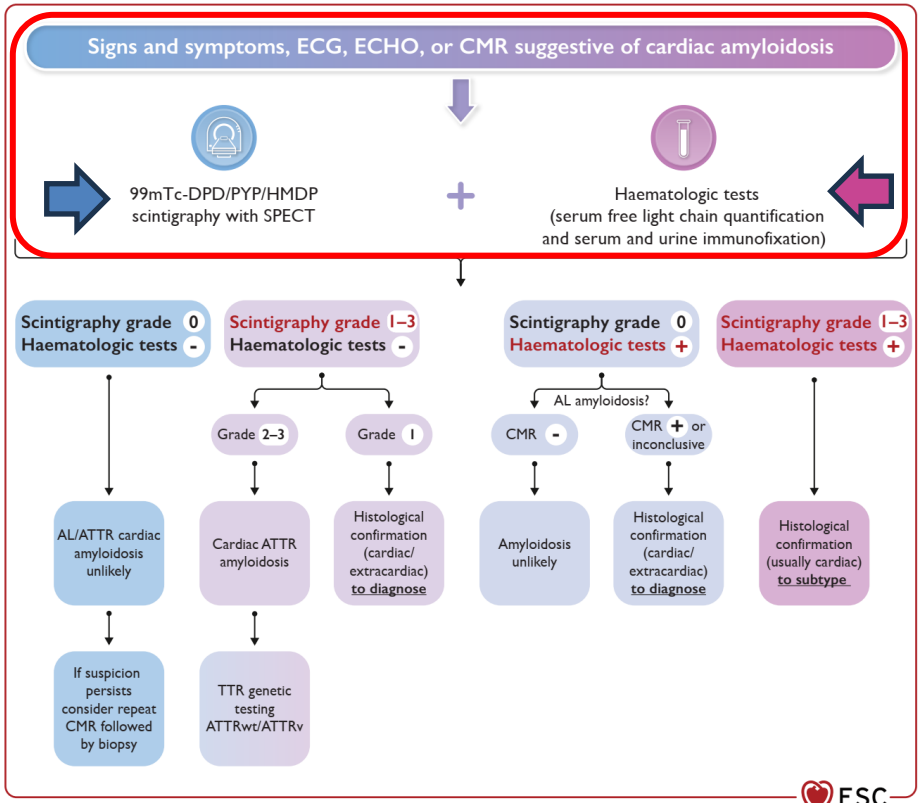
Diagnostika a dif df. srdečních amyloidóz



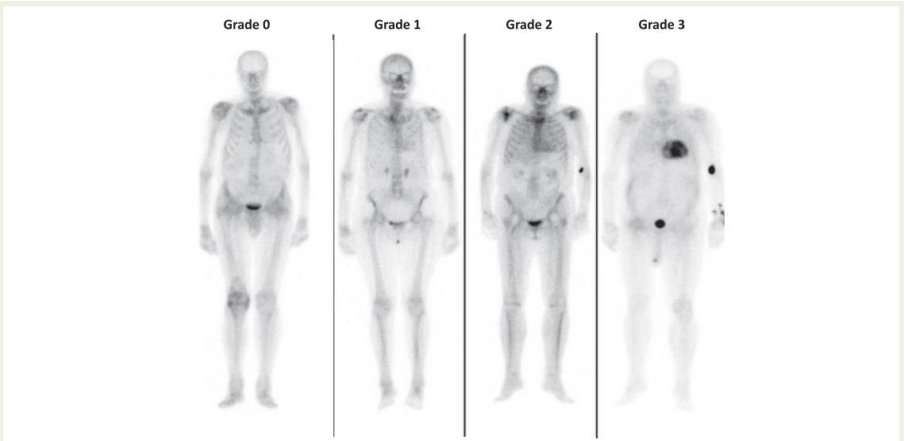


Figure 2 Cardiac uptake grading in bisphosphonate scintigraphy. Grade 0: absence of tracer myocardial uptake and normal bone uptake; Grade 1: myocardial uptake in a lower degree than at bone level; Grade 2: similar myocardial and bone uptake; and Grade 3: myocardial uptake greater than bone with reduced/absent bone uptake.

Endomyocardial biopsy should be considered in patients with RCM to exclude specific diagnoses (including iron overload, storage disorders, mitochondrial cytopathies, amyloidosis, and granulomatous myocardial diseases) and to diagnose restrictive myofibrillar disease caused by desmin variants.	IIa	C

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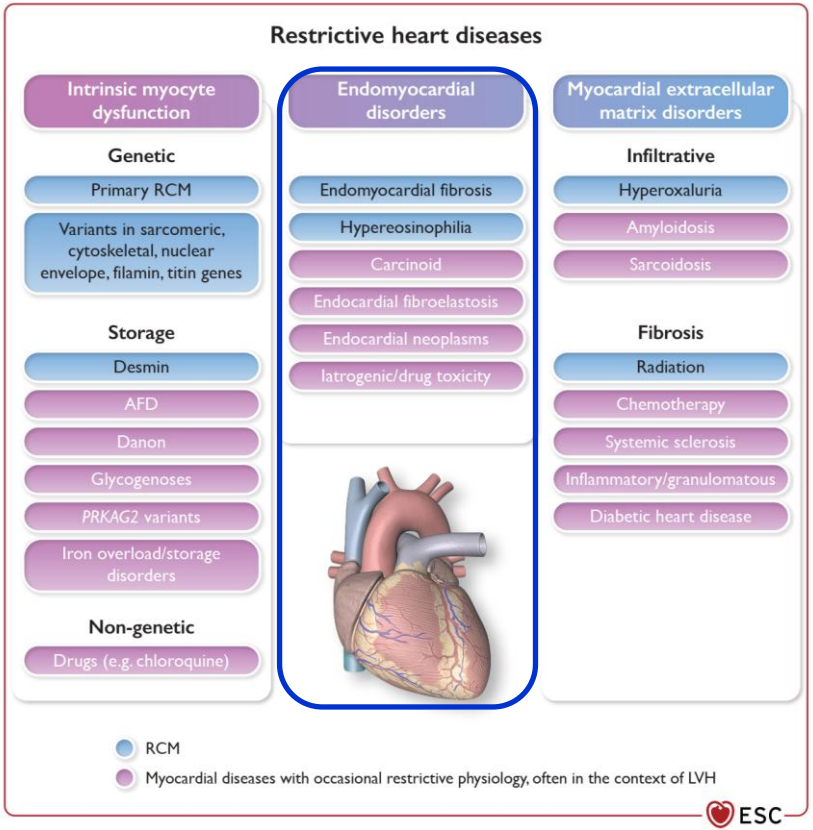
Restriktivní kardiomyopatie - III



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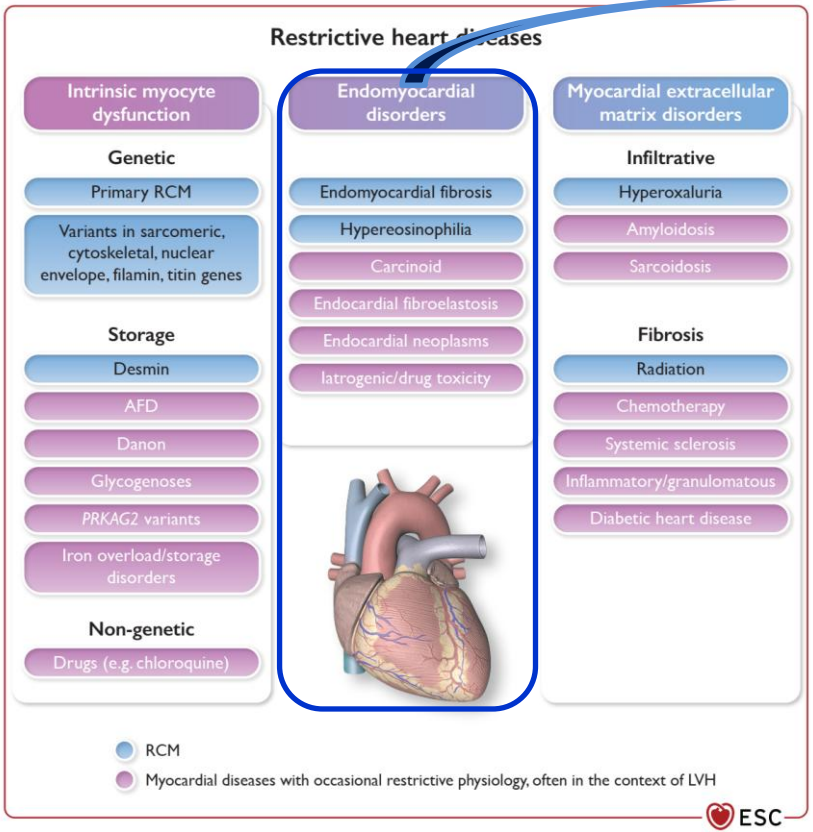
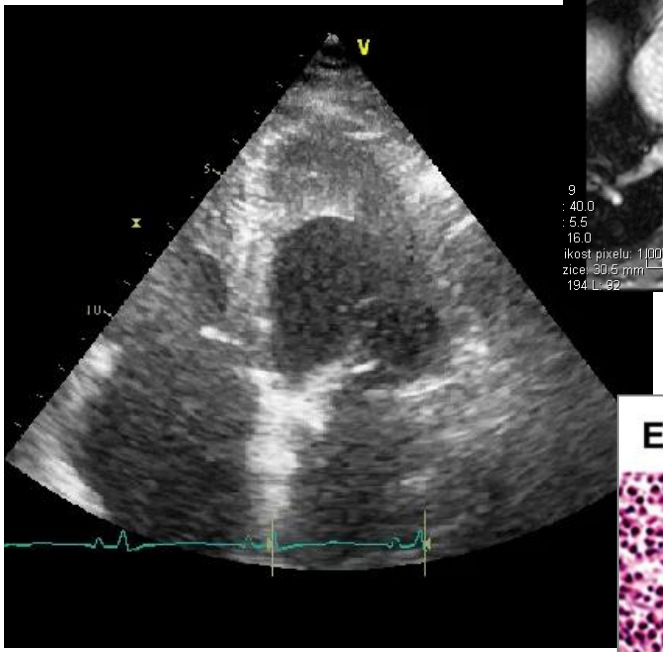
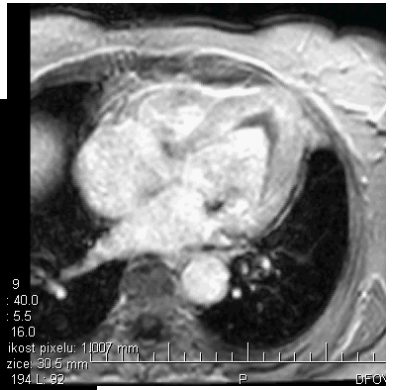
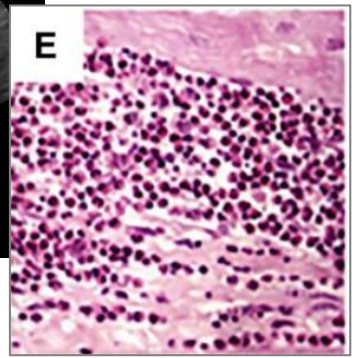
Restriktivní kardiomyopatie



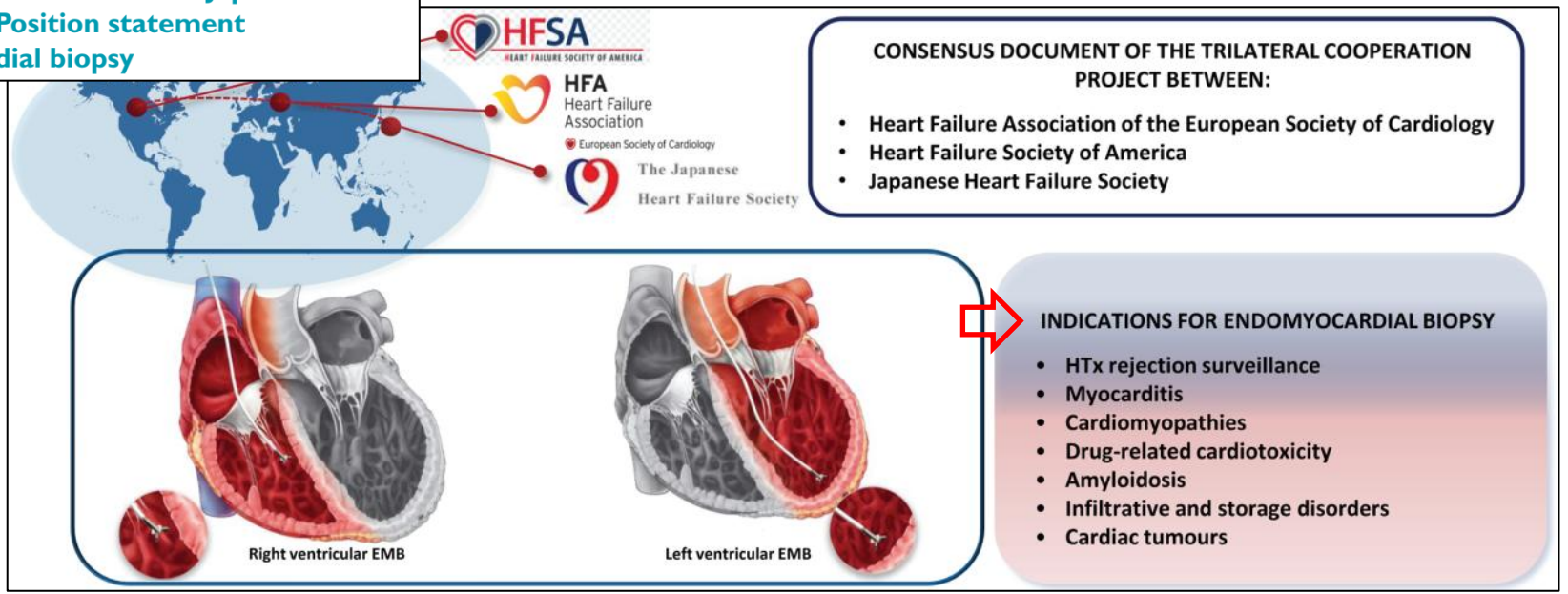






RCM - invazivní diagnostika

Heart Failure Association of the ESC, Heart Failure Society of America and Japanese Heart Failure Society Position statement on endomyocardial biopsy



European Journal of Heart Failure (2021) **23**, 854–871

EMB v diagnostice RCM - histologie

Restrictive cardiomyopathy: definition and diagnosis

European Heart Journal (2022) **43**, 4679–4693

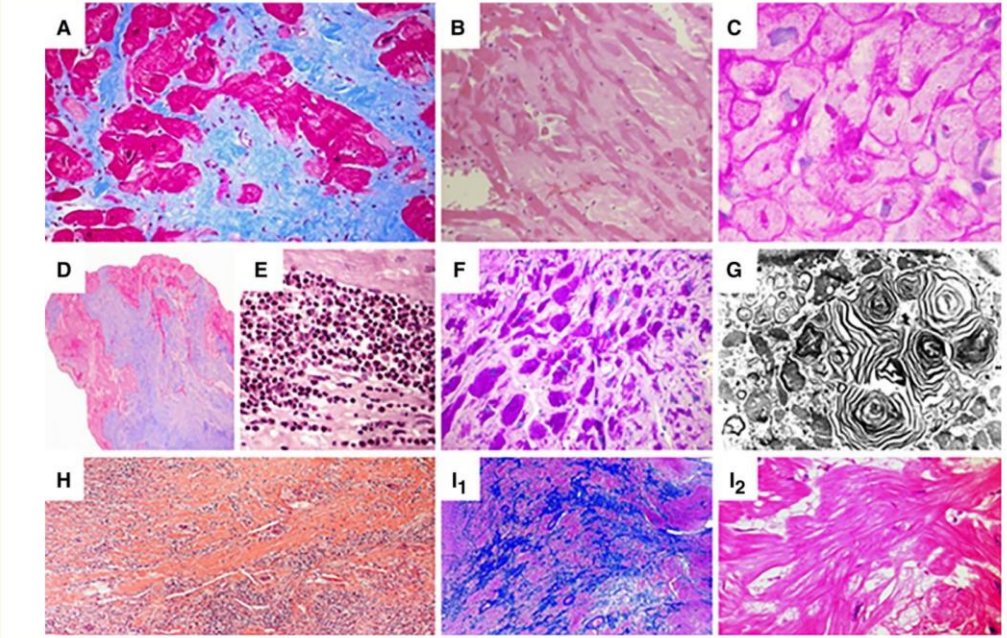


Figure 2 Myocardial tissue in nine different forms of restrictive cardiomyopathy (*A*) idiopathic restrictive cardiomyopathy; (*B*) cardiac amyloidosis (with enlargement of the extracellular spaces by amyloid fibres); (*C*) Danon disease (with intracellular glycogen deposits); (*D*) endomyocardial fibrosis (with extensive fibrosis in the endocardium and myocardium); (*E*) hypereosinophilic syndrome (with tissue accumulation of eosinophils); (*F*) glycogenosis (with tissue accumulation of glycogen); (*G*) Anderson–Fabry disease (with lipid deposits as seen through electron microscopy); (*H*) sarcoidosis (with tissue granulomas); (*I*) end-stage hypertrophic cardiomyopathy (with extensive fibrosis). Courtesy of Dr Ornella Leone, Bologna, Italy.

Obecné terapeutické zásady u RCM

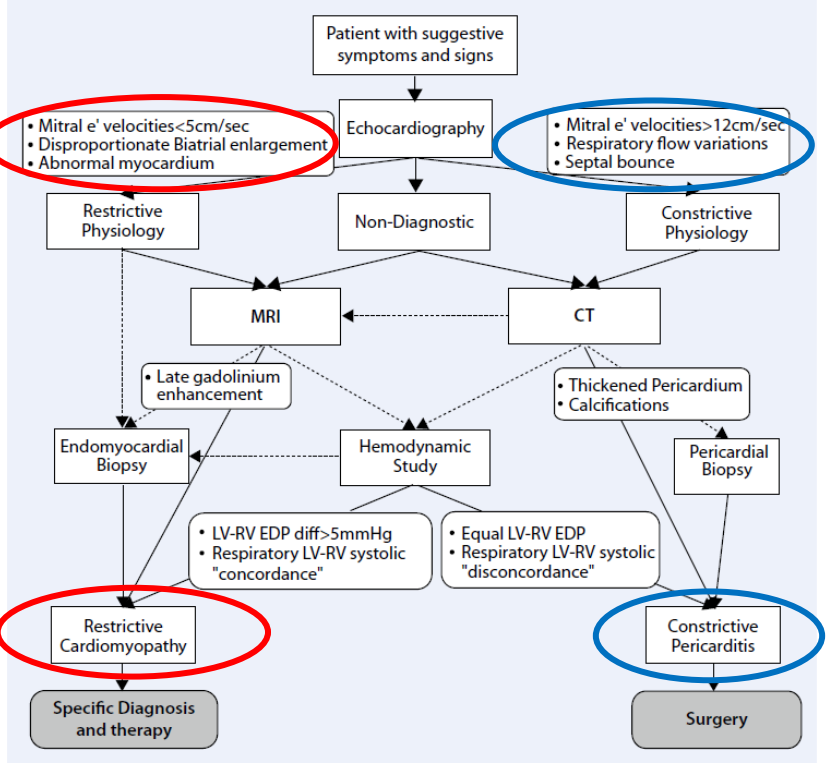
- Zvládnutí kongesce (cave dostatečný plnící tlak k udržení CO)
- Reverzní remodelace není cílem léčby (žádná prognostická data pro RAASi)
- CO je závislý na tepové frekvenci (prodloužení diastoly nezvyšuje EDV!)
  – spíše negativní efekt BB
- Antikoagulace při fi síní
- Kauzální terapie, je-li dostupná

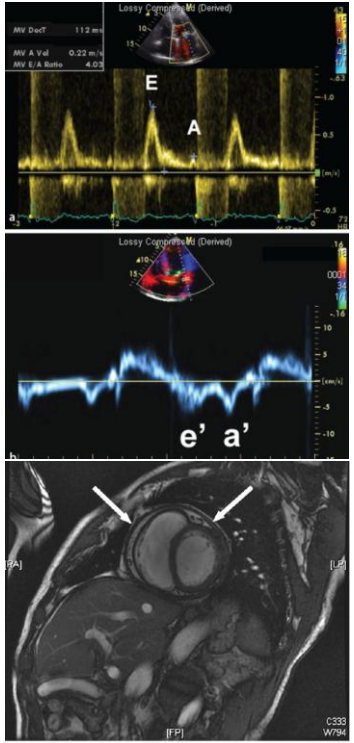
Diferenciální dg konstriktivní perikarditidy a restriktivní kardiomyopatie

Herz 2012 · 37:664–674
DOI 10.1007/s00059-012-3663-4
Published online: 1 September 2012

D.R. Zwas · I. Gotsman · D. Admon · A. Keren
Heart Failure Center, Heart Institute, Hadassah University Hospital, Jerusalem

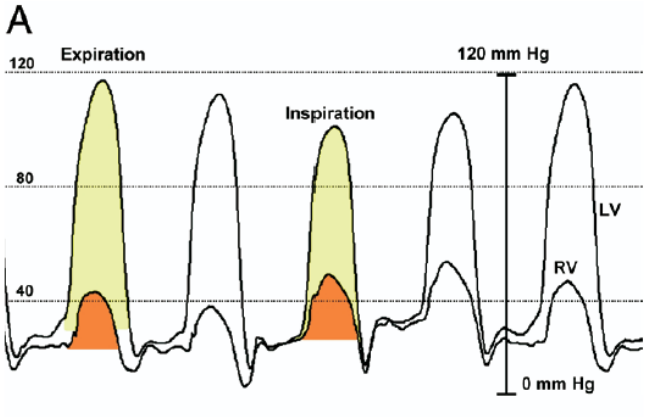
Advances in the differentiation of constrictive pericarditis and restrictive cardiomyopathy





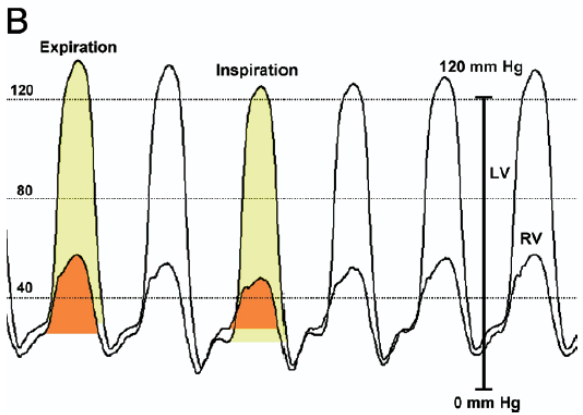
Diferenciální dg konstriktivní perikarditidy a restriktivní kardiomyopatie

Konstriktivní perikarditis



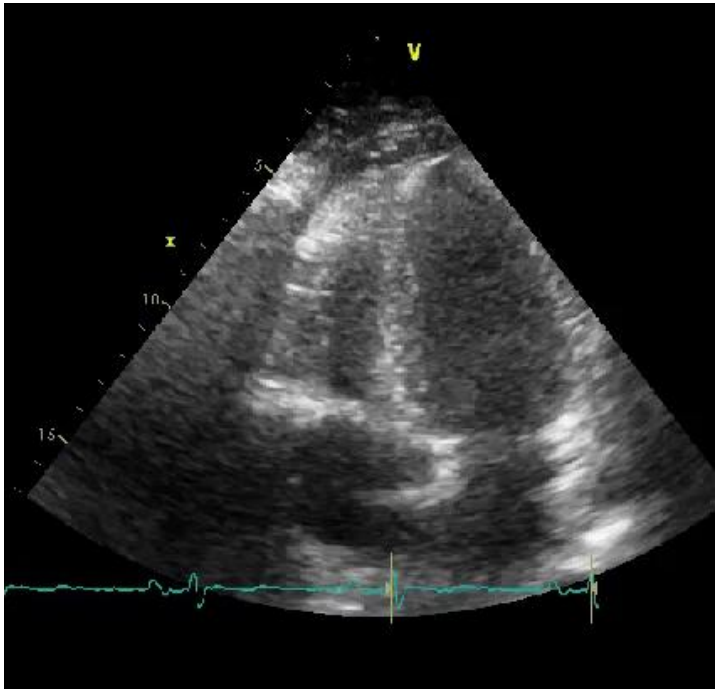
Diskordance systolických komorových tlaků
Zvýrazněná interventrikulární dependence

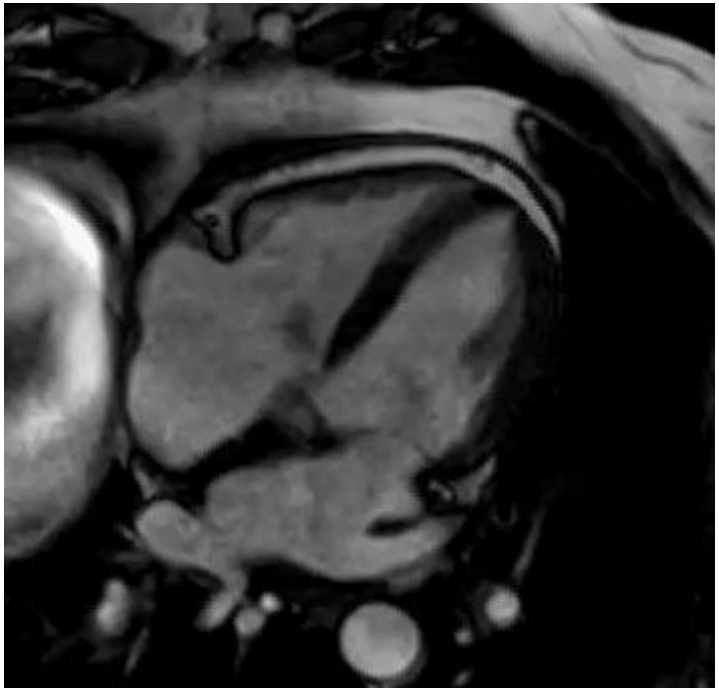
Restriktivní kardiomyopatie



Konkordance systolických komorových tlaků

Diferenciální dg CP a RCM – septal shift a septal bounce





ZÁVĚR

RCM je velmi heterogenní skupina onemocnění, jejichž správná diagnostika a diferenciální diagnostika v řadě případů umožňuje kauzální léčbu!

Děkuji za pozornost!