

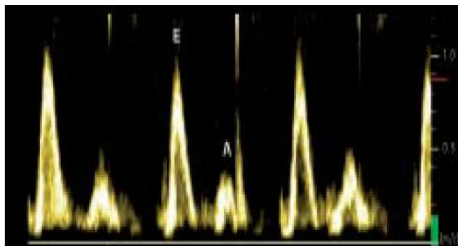
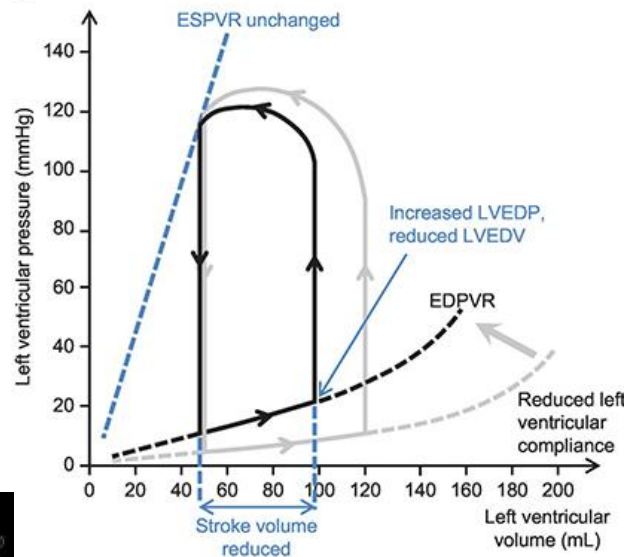
Restriktivní kardiomyopatie

Jan Krejčí



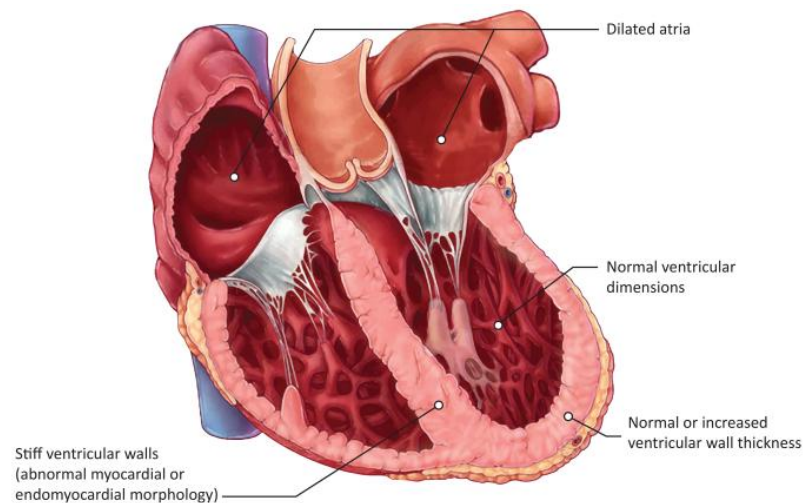
Co o restriktivní kardiomyopatii léta víme...

- 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

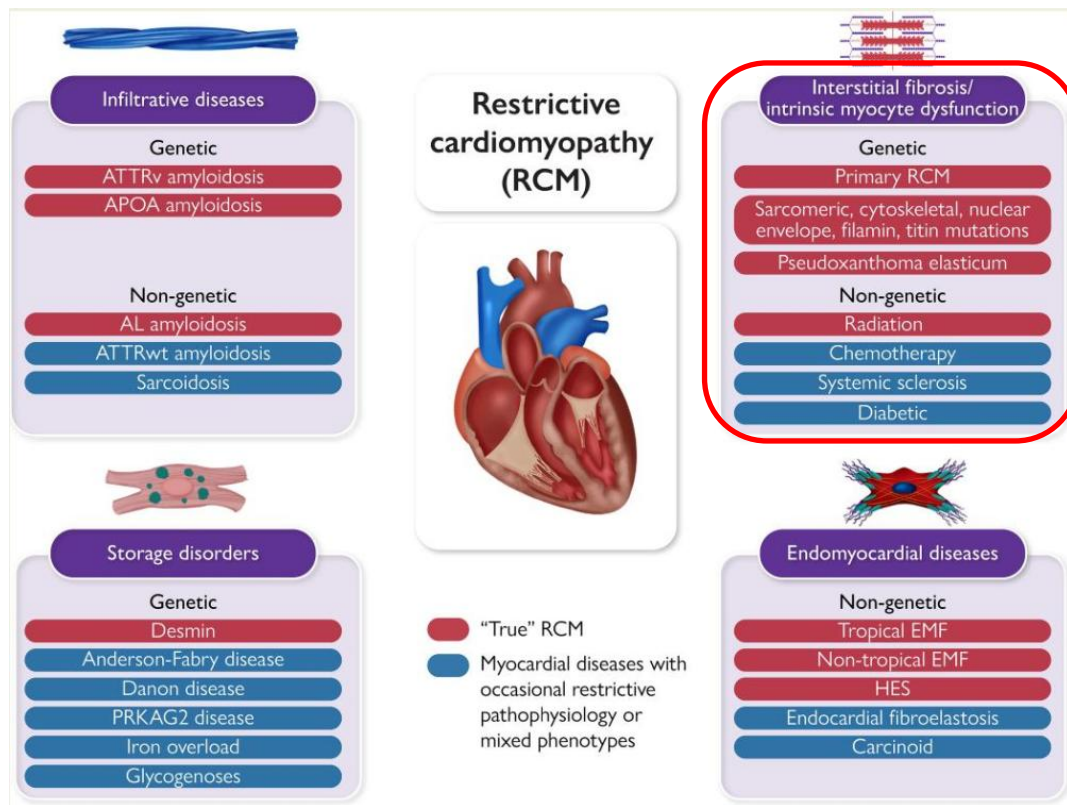
The approach we followed was first of all to refine and enhance the nosographic aspect of the classification and, subsequently, to provide useful insights for the diagnosis of individual diseases. The definition of RCM can be slightly modified as follows: **RCM is characterized by the coexistence of 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.**

Dělení RCM podle „místa postižení“

Restrictive cardiomyopathy: definition and diagnosis

European Heart Journal (2022) 43, 4679–4693

Postižení myocytů / intersticiální fibróza



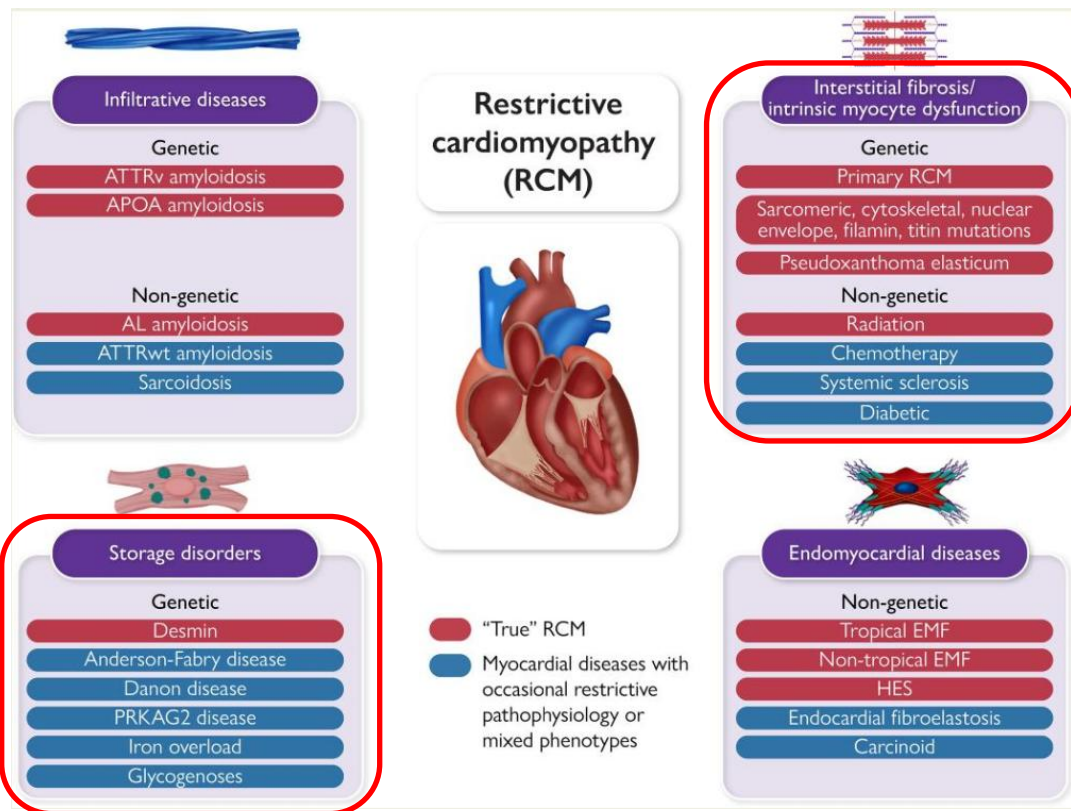
Dělení RCM podle „místa postižení“

Restrictive cardiomyopathy: definition and diagnosis

European Heart Journal (2022) 43, 4679–4693

Postižení myocytů / intersticiální fibróza

Postižení při intracelulární akumulaci patologických substátů



Dělení RCM podle „místa postižení“

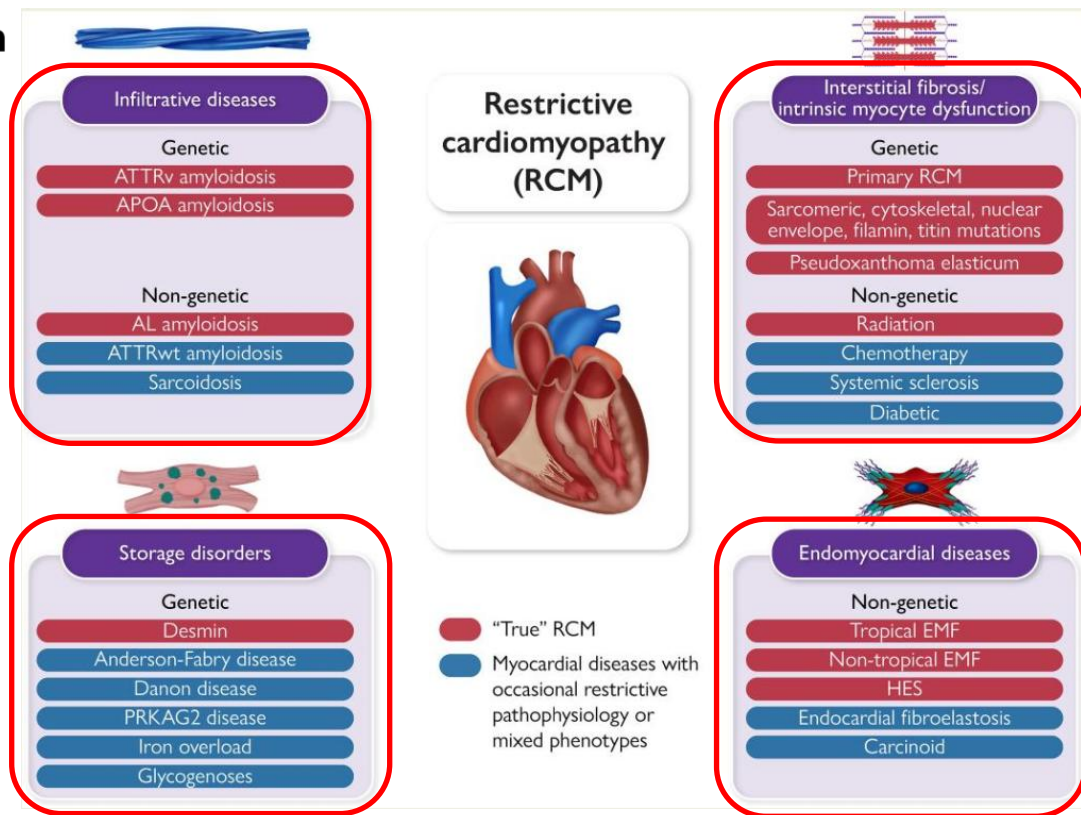
Restrictive cardiomyopathy: definition and diagnosis

European Heart Journal (2022) 43, 4679–4693

Postižení myocytů / intersticiální fibróza

Postižení při intracelulární akumulaci patologických substátů

Postižení při extracelulární akumulaci patologických substátů



Dělení RCM podle „místa postižení“

Restrictive cardiomyopathy: definition and diagnosis

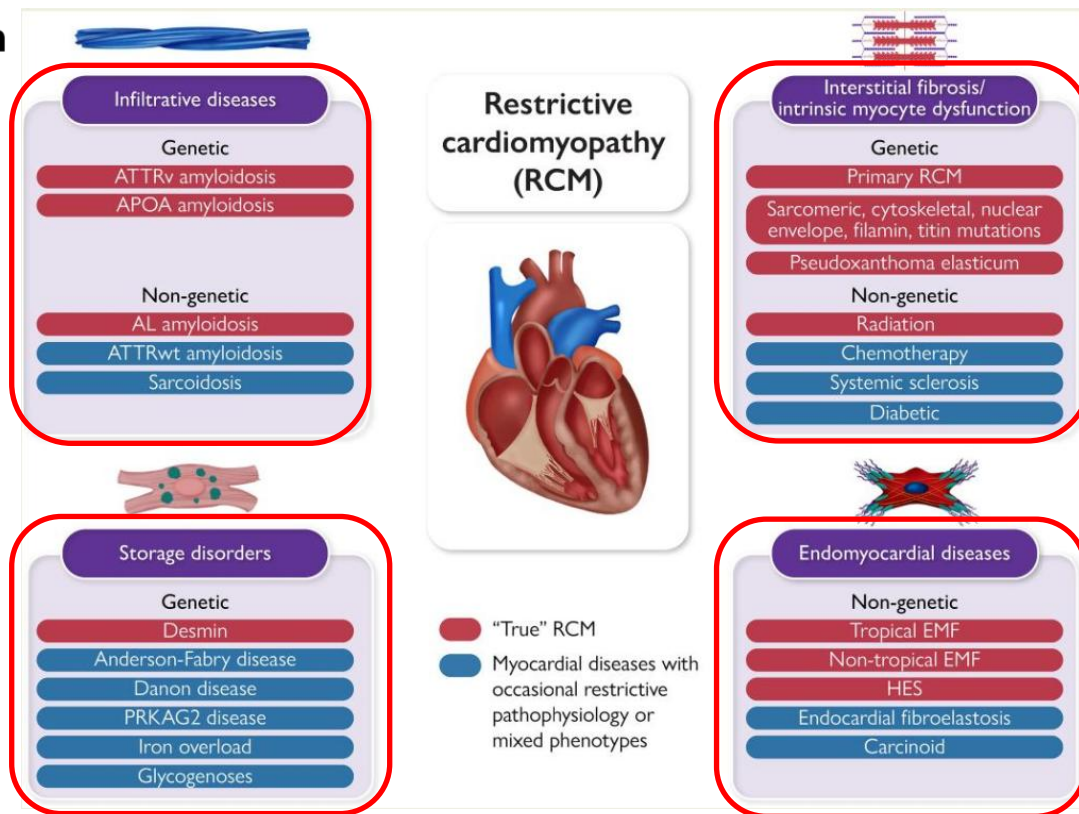
European Heart Journal (2022) 43, 4679–4693

Postižení myocytů / intersticiální fibróza

Postižení při intracelulární akumulaci patologických substátů

Postižení při extracelulární akumulaci patologických substátů

Postižení endo(myo)kardu



Restriktivní kardiomyopatie

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.

Postižení myocytů

Restrictive heart diseases

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

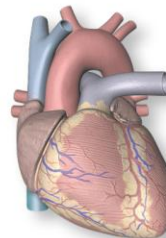
Radiation

Chemotherapy

Systemic sclerosis

Inflammatory/granulomatous

Diabetic heart disease



● RCM

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

Restriktivní kardiomyopatie

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.

Postižení myocytů

Postižení extracelulární matrix

Restrictive heart diseases

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

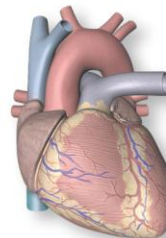
Radiation

Chemotherapy

Systemic sclerosis

Inflammatory/granulomatous

Diabetic heart disease



● RCM

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

Restriktivní kardiomyopatie

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.

Postižení myocytů

Postižení extracelulární matrix

Restrictive heart diseases

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

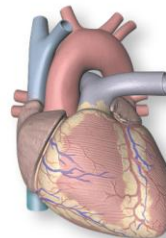
Radiation

Chemotherapy

Systemic sclerosis

Inflammatory/granulomatous

Diabetic heart disease



● RCM

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

Restriktivní kardiomyopatie

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.

Postižení myocytů

Postižení extracelulární matrix

Postižení endo(myo)kardu

Restrictive heart diseases

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

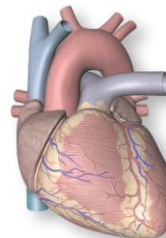
Radiation

Chemotherapy

Systemic sclerosis

Inflammatory/granulomatous

Diabetic heart disease

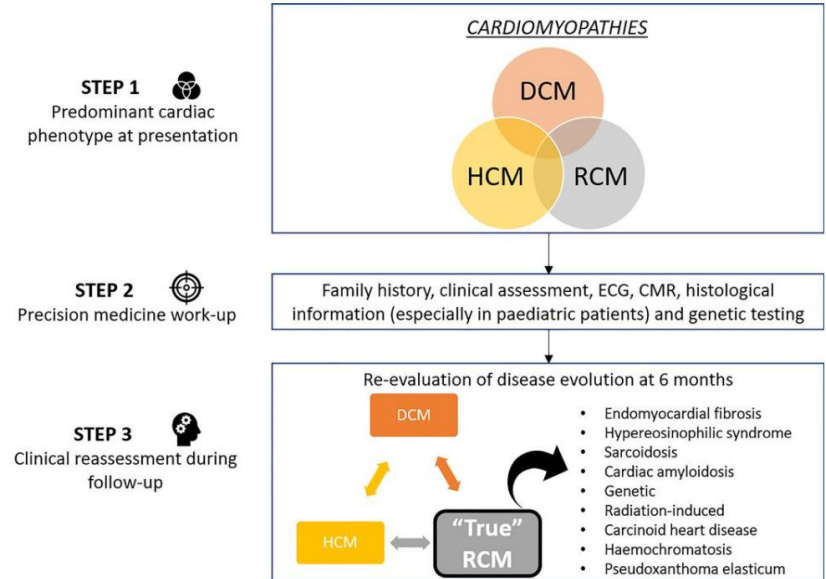


● RCM

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

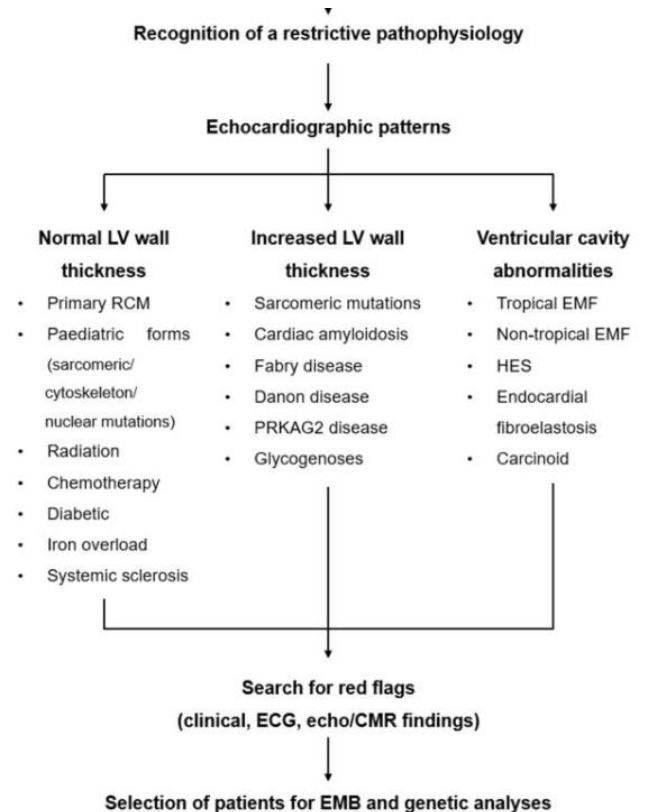
RCM

- 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



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.



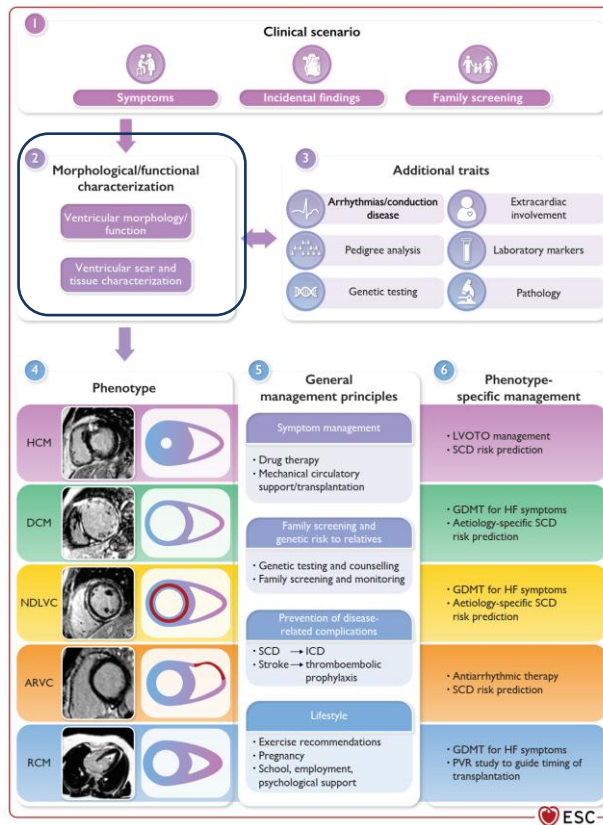
European Heart Journal (2022) 43, 4679–4693

Zásady přístupu k diagnostice KMP dle ESC guidelines

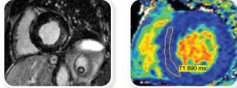
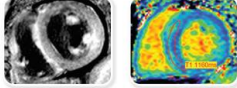
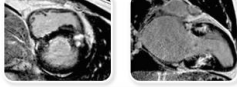
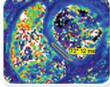
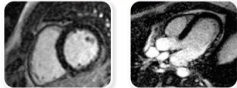
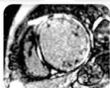
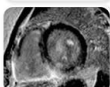
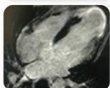
Morphological/functional characterization


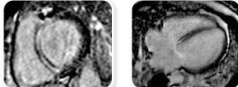
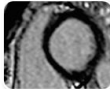
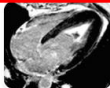
Ventricular morphology/
function

Ventricular scar and
tissue characterization



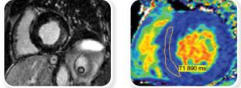
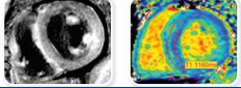
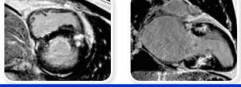
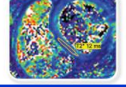
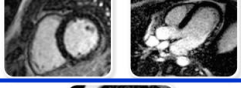
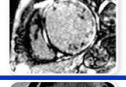
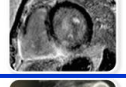

Heterogenita RCM – přínos CMR

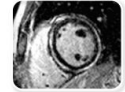
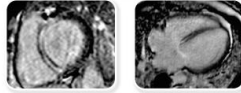
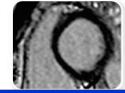
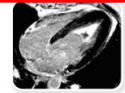
Cardiomyopathy phenotype	Finding	Cardiac CMR examples	Specific diseases to be considered
HCM	Posterolateral LGE and concentric LVH Low native T1		Anderson–Fabry disease
	Diffuse subendocardial LGE, high native T1		Amyloidosis
	Patchy mid-wall in hypertrophied areas		Sarcomeric HCM
DCM	Short T2*		Haemochromatosis
	Subepicardial LGE		Post-myocarditis
	Lateral wall epicardial LGE		Dystrophinopathy
	Subepicardial and midwall LGE at basal septum +/- extension into inferolateral wall and RV insertion points		Sarcoidosis
	Apical transmural LGE		Chagas disease

NDLVC	Ring-like and/or subepicardial LGE pattern		DSP variants FLNC variants DES variants
	Septal mid-wall LGE		Laminopathy
ARVC	Fat and LGE (transmural RV plus sub-epicardial-midmural LV free wall)		Desmosomal variants
RCM	Partial LV or RV apical obliteration + LGE at endocardial level		EMF/hypereosinophilia



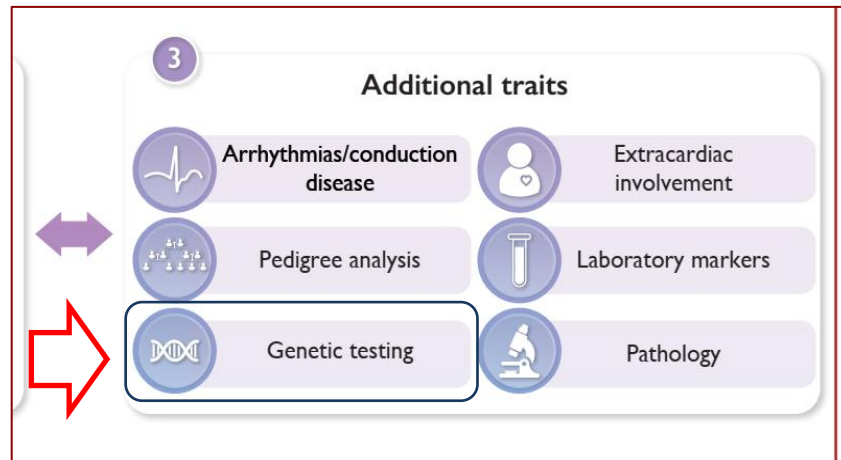
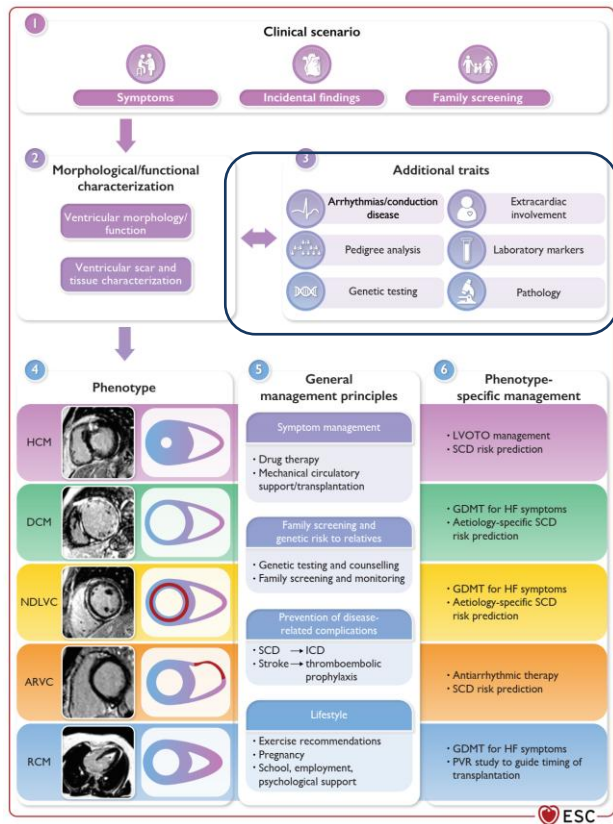
Heterogenita RCM – přínos CMR

Cardiomyopathy phenotype	Finding	Cardiac CMR examples	Specific diseases to be considered
HCM	Posterolateral LGE and concentric LVH Low native T1		Anderson–Fabry disease
	Diffuse subendocardial LGE, high native T1		Amyloidosis
	Patchy mid-wall in hypertrophied areas		Sarcomeric HCM
	Short T2*		Haemochromatosis
DCM	Subepicardial LGE		Post-myocarditis
	Lateral wall epicardial LGE		Dystrophinopathy
	Subepicardial and midwall LGE at basal septum +/- extension into inferolateral wall and RV insertion points		Sarcoidosis
	Apical transmural LGE		Chagas disease

NDLVC	Ring-like and/or subepicardial LGE pattern		DSP variants FLN/C variants DES variants
	Septal mid-wall LGE		Laminopathy
ARVC	Fat and LGE (transmural RV plus sub-epicardial-midmural LV free wall)		Desmosomal variants
RCM	Partial LV or RV apical obliteration + LGE at endocardial level		EMF/hypereosinophilia



Diagnostika KMP a genetické testování



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




Role genetického testování u RCM

Gene	Cardiomyopathy phenotype					Associated phenotype
	HCM	DCM	NDLVC	ARVC	RCM	
ABCC9	●	○				* Cantu syndrome
ACTA1	○					
ACTC1	●	●	●	○	○	
ACTN2 ^a	●	●	●			
ALPK3	●					
ANKRD1	○	○				
BAG3	●	●			○	* Myofibrillar myopathy
CACNA1C	●					† Timothy syndrome
CACNB2	○					
CALR3	○					
CASQ2	○					
CAV3	●					* Caveolinopathy
CDH2				○		
COX15	●					* Leigh syndrome
CRYAB	●					* Alpha-B crystallinopathy
CSRP3	●	○				
CTF1		○				
CTNNA3				○		
DES	●	●	●	●	○	† Desminopathy
DMD		●	●			† X-linked progressive MD
DMPK			●			
DSC2				●	●	
DSG2		○				
DSP	○	●	●	●	●	
DTNA		○	●			
EYA4		○				
FHL1	●					† Emery-Dreifuss MD
FLNC	●	●	●		○	† Myofibrillar myopathy
FHOD3	○					
FXN	●					* Friedreich ataxia
GAA	●					* Pompe disease

GATA4				○		
GATAD1			○			
GLA	●					† Anderson-Fabry disease
HCN4				○		
ILK			○		○	
JPH2	●	●				
JUP					●	† Naxos disease (cardiocutaneous syndrome)
KCNQ1	○					
KLF10	○					
LAMA4			○			
LAMP2	●					† Danon disease
LDB3	●		○		○	† Myofibrillar myopathy
LMNA			●	●	○	
LRRC10			○			
MIB1					○	
MTBP3	●	●	○		○	○
MTFH6		○				
MTFH7	●	●	●	○	○	○
MYL2	●	●	○		○	○
MYL3	●	○			○	○
MYLK2						○
MYOM1		○				
MYOZ2		○				
MYRN		○				○
NEBL			○			
NEXN		○	○			
NOK2-5					○	
NNT					○	
NOXD					○	
NPPA			○			
OBSCN	○		○		○	
PDLIM3	○					
PKP2					●	●
PLEKHA2			○			
PLN ^a	●	●		○	○	

PRDM16			○		○	
PRKAG2	●					† PRKAG2 cardiomyopathy
PSEN1			○			
PSEN2			○			
PTPN11	●					† Noonan syndrome
RAF1	●					† Noonan syndrome
RBM20		●	●		○	
RIT1	●					† Noonan syndrome
RYR2	○				○	
SCN5A			●		○	
SGCD			○			
SLC25A4	●					† Mitochondrial disease
TAZ					○	
TBX5					○	
TBX20			○		○	
TCAP	○		○			
TGFB3					○	
TJP1					○	
TMEM43			○		○	●
TMEM70					○	
TMPO	○		○			
TNNC1	○		○			○
TNNI3	●	●	○			○
TNNI3K			○			
TNNT2	●	●	●	●	○	○
TPM1	●	○	○		○	○
TRIM63	○		○		○	
TTN	○	●	●	●	○	○
TTR	●					○
VCL	○	○				

Role genetického testování v dif. dg. HCM/RCM

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

Restriktivní kardiomyopatie - I

Restrictive heart diseases

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

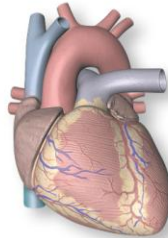
Radiation

Chemotherapy

Systemic sclerosis

Inflammatory/granulomatous

Diabetic heart disease



● RCM

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

Restriktivní kardiomyopatie

Restrictive heart diseases

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

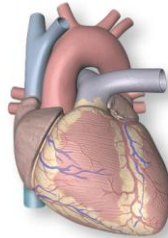
Radiation

Chemotherapy

Systemic sclerosis

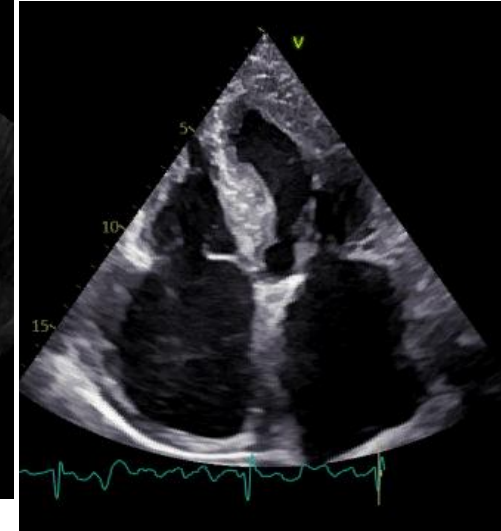
Inflammatory/granulomatous

Diabetic heart disease

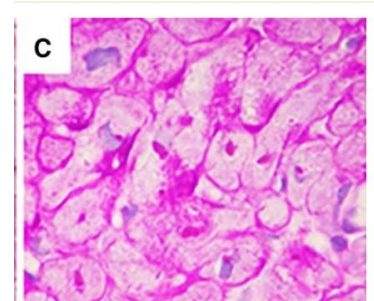
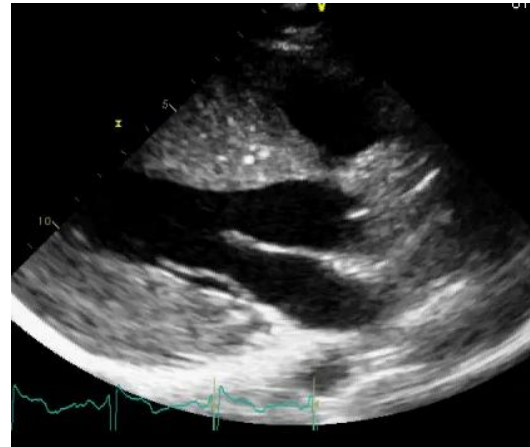
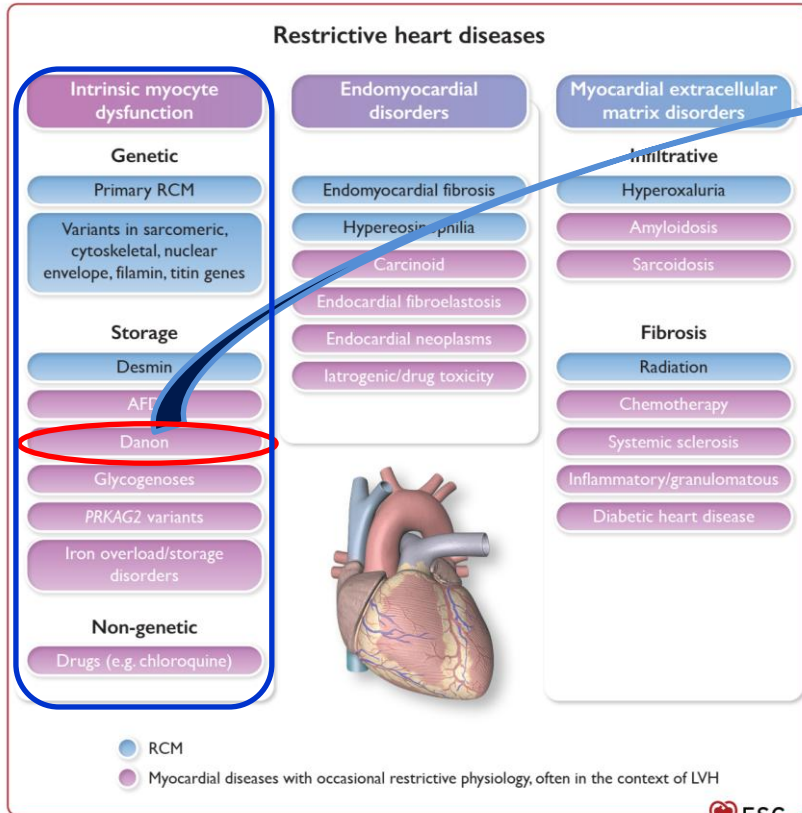


● RCM

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



Danonova nemoc - RCM nebo HCM?



Restriktivní kardiomyopatie - II

Restrictive heart diseases

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

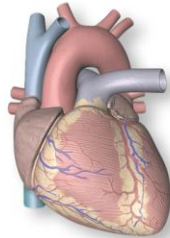
Radiation

Chemotherapy

Systemic sclerosis

Inflammatory/granulomatous

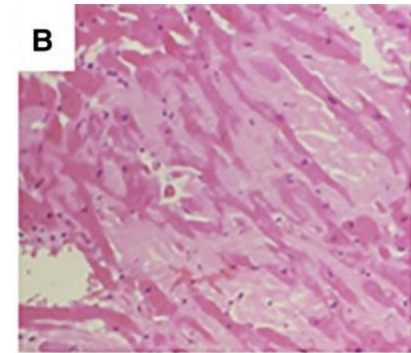
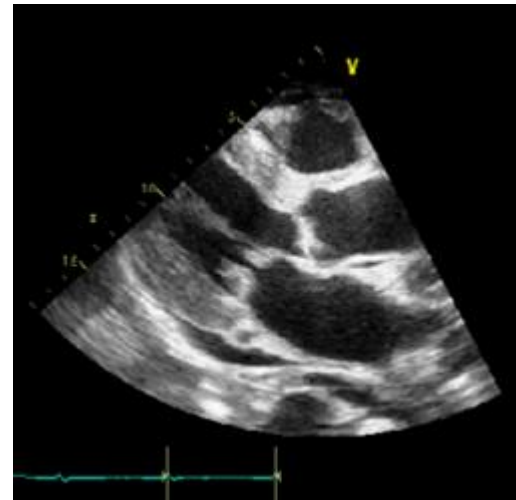
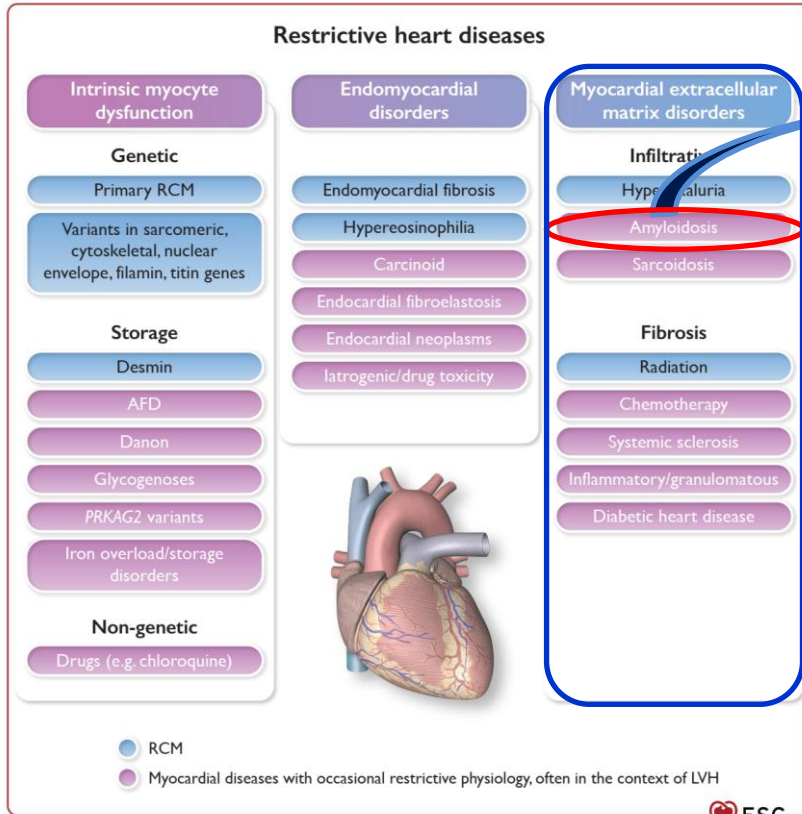
Diabetic heart disease



● RCM

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

Srdeční amyloidóza - RCM nebo HCM?



Srdeční amyloidóza - RCM nebo HCM?

Restrictive heart diseases

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

Hypertension

Amyloidosis

Sarcoidosis

Fibrosis

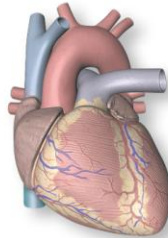
Radiation

Chemotherapy

Systemic sclerosis

Inflammatory/granulomatous

Diabetic heart disease

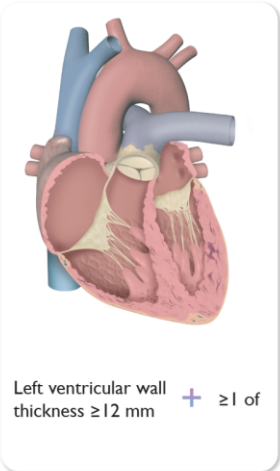


● RCM

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



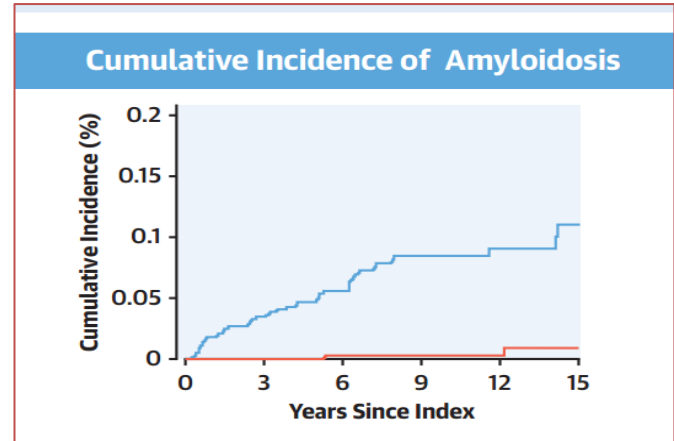
Dif. dg. RCM (HCM) – srdeční amyloidóza



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

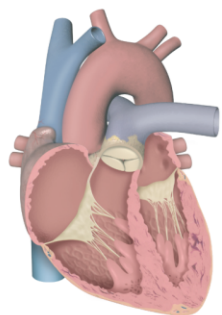


Fosbol et al. JACC 2019

Syndrom karpálního tunelu

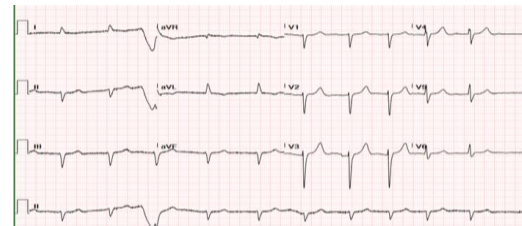
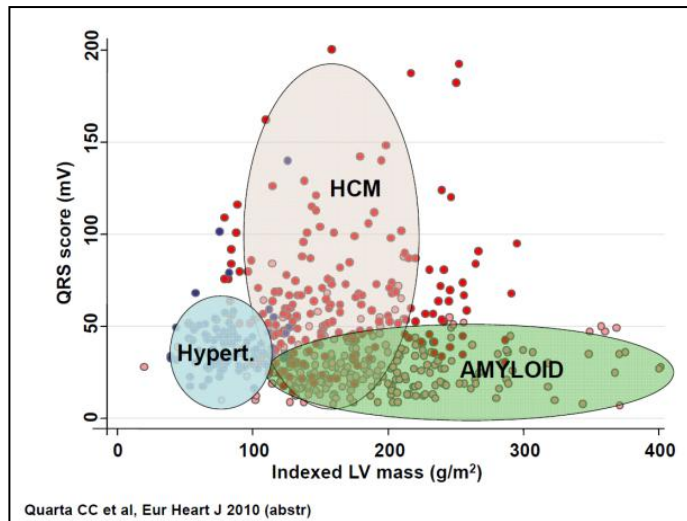
- předchází dg 5-15 let, často oboustranný až u 50% nemocných
- **12x vyšší riziko srdeční amyloidosy**

Dif. Dg. RCM (HCM) – srdeční amyloidóza

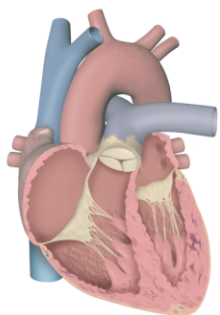


Left ventricular wall thickness ≥ 12 mm + ≥ 1 of

- 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



Dif. dg. RCM (HCM) – srdeční amyloidóza a AoS



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

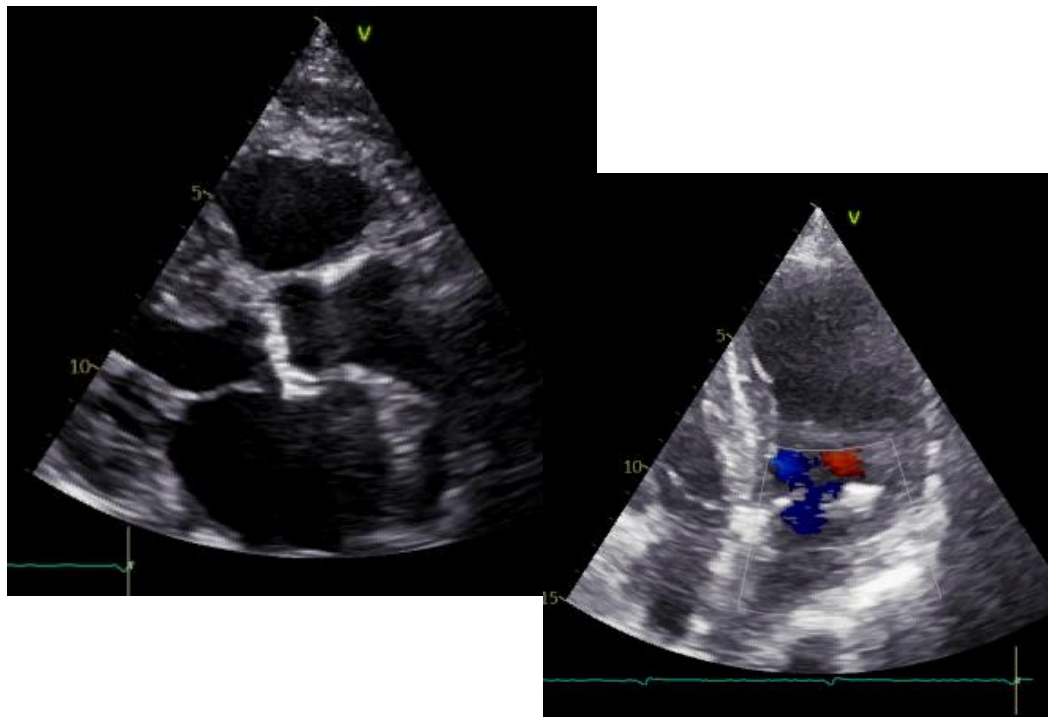
Pseudo Q waves on ECG

AV conduction disease

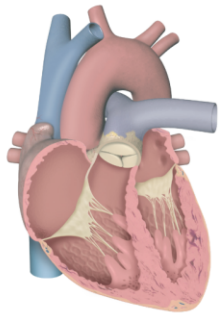
Possible family history of ATTR

Chronically increased troponin levels

Known multiple myeloma or MGUS

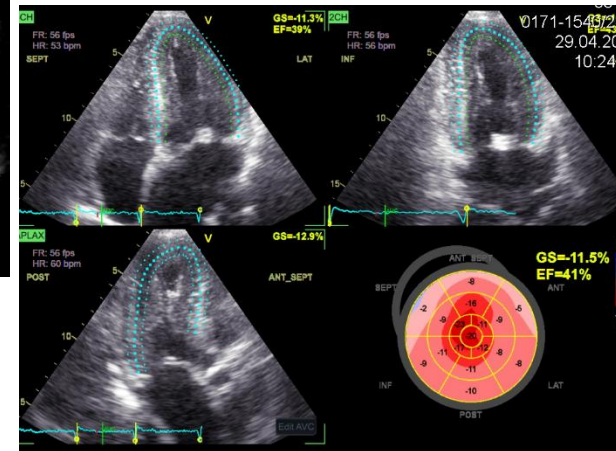
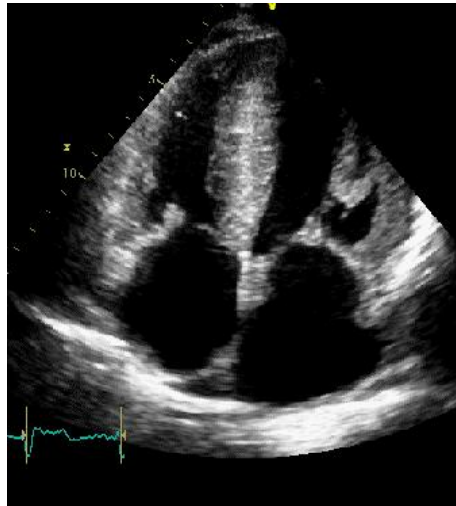


Dif. Dg. RCM (HCM) – srdeční amyloidóza

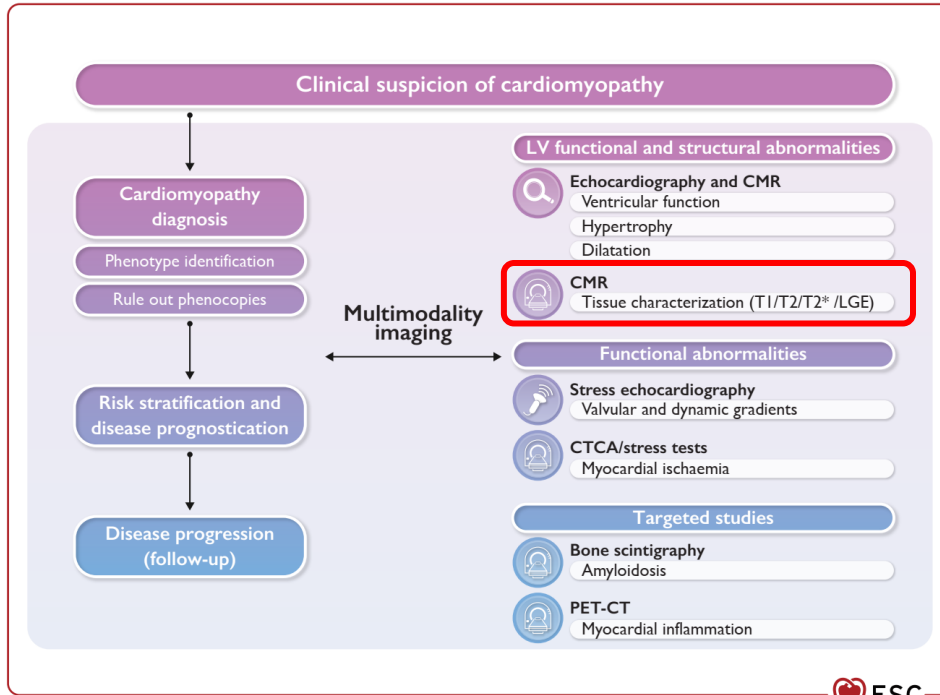


Left ventricular wall thickness ≥ 12 mm + ≥ 1 of

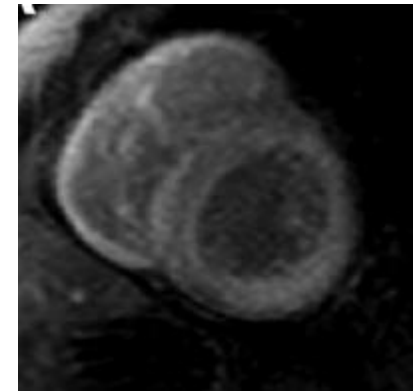
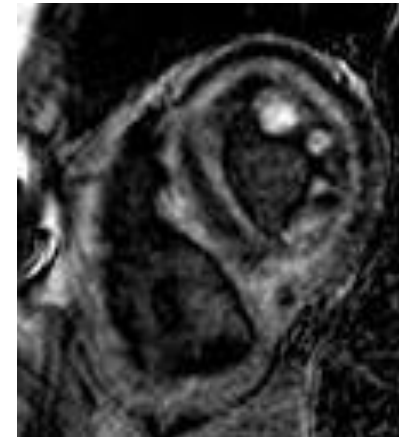
- Heart failure in ≥ 65 years
- Aortic stenosis in ≥ 65 years
- Hypotension or normotensive if previously hypertensive
- Sensory involvement, autonomic dysfunction
- Peripheral polyneuropathy
- Proteinuria
- Skin bruising
- Ruptured biceps tendon
- Bilateral carpal tunnel syndrome
- Subendocardial/transmural 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



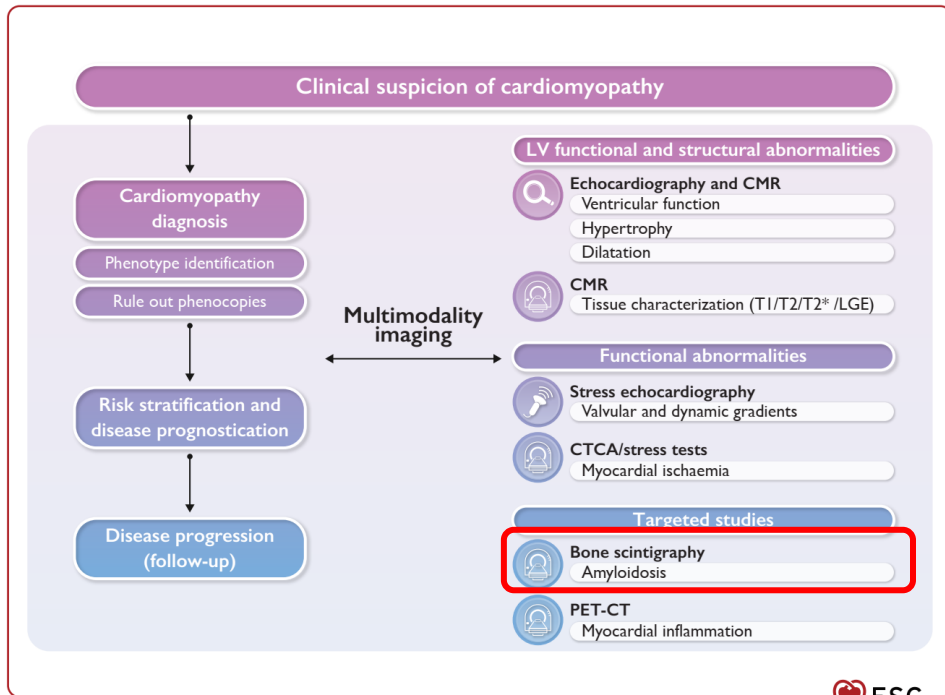
Diagnostika a diferenciální dg. RCM/HCM a srdeční amyloidózy



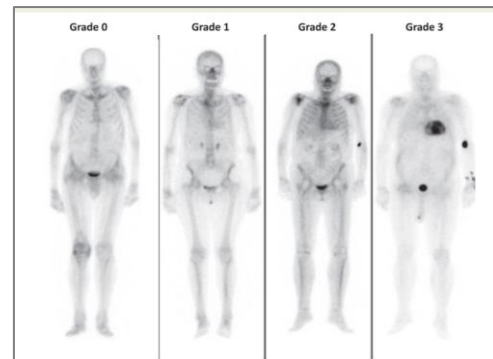
Recommendations	Class ^a	Level ^b
Contrast-enhanced CMR is recommended in patients with cardiomyopathy at initial evaluation. ^{10,90,116,119-143}	I	B



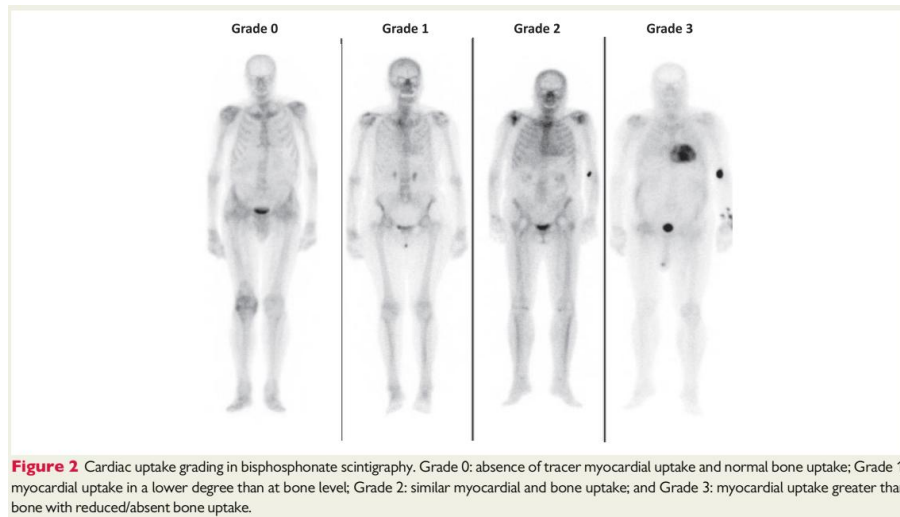
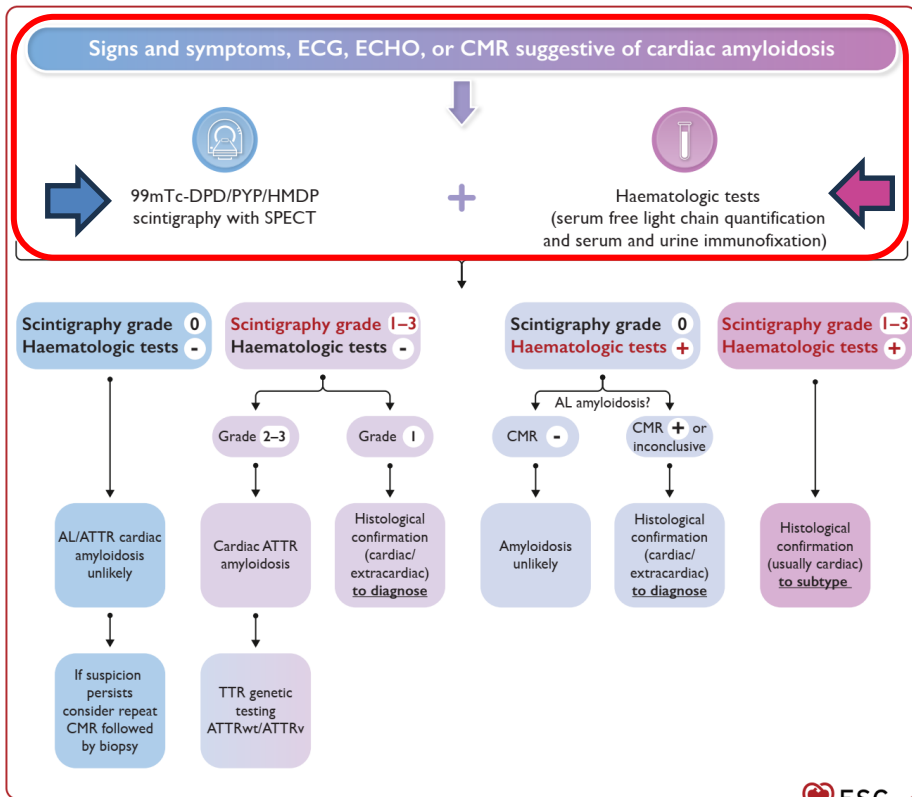
Diagnostika a diferenciální dg. RCM/HCM a srdeční amyloidózy



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



Diagnostika a dif. df. srdečních amyloidóz



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.



Restriktivní kardiomyopatie - III

Restrictive heart diseases

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

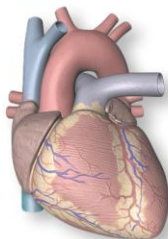
Hypereosinophilia

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

● RCM

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

Restriktivní kardiomyopatie

Restrictive heart diseases

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

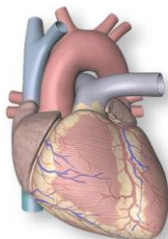
Hypereosinophilia

Carcinoid

Endocardial fibroelastosis

Endocardial neoplasms

Iatrogenic/drug toxicity



Myocardial extracellular matrix disorders

Infiltrative

Hyperoxaluria

Amyloidosis

Sarcoidosis

Fibrosis

Radiation

Chemotherapy

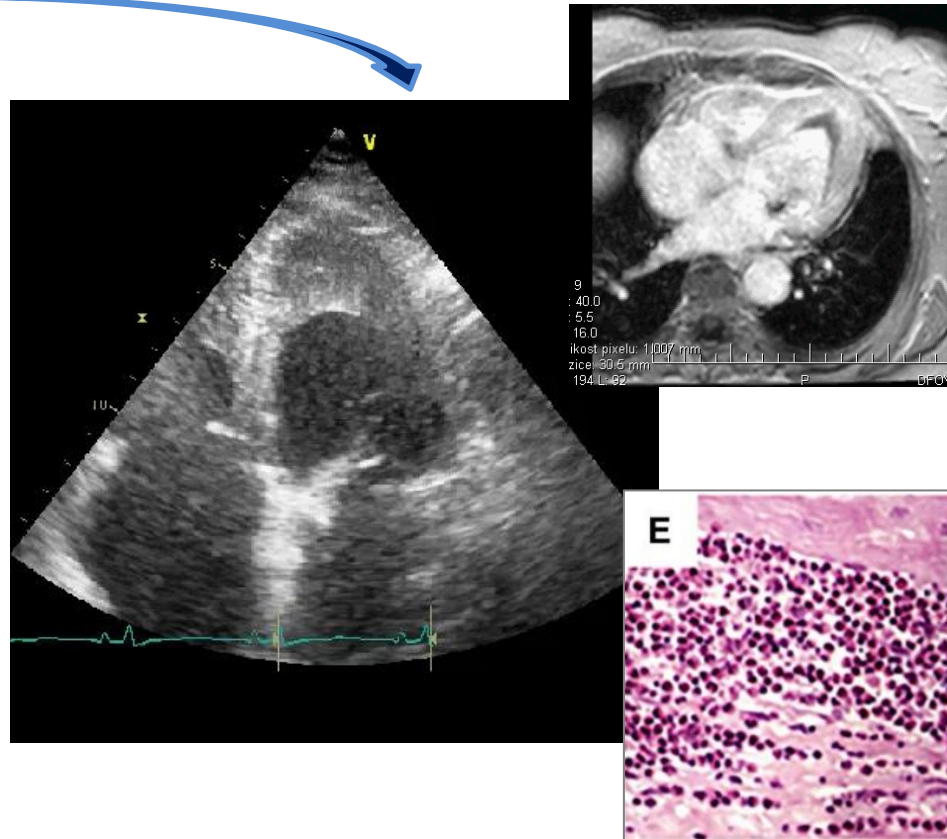
Systemic sclerosis

Inflammatory/granulomatous

Diabetic heart disease

● RCM

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



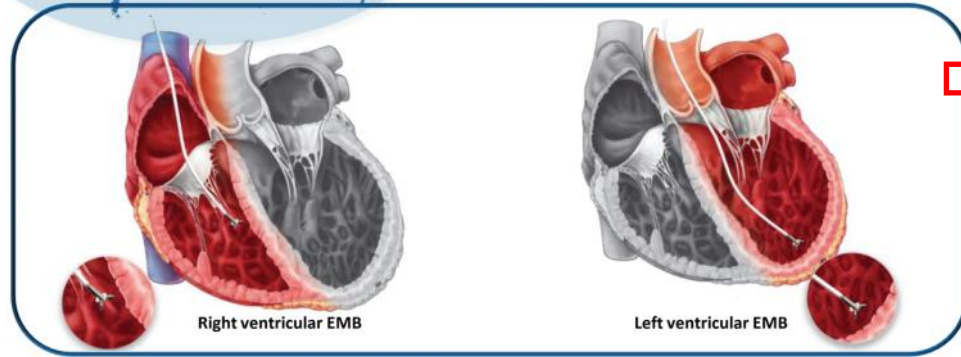
RCM - invazivní diagnostika

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



CONSENSUS DOCUMENT OF THE TRILATERAL COOPERATION PROJECT BETWEEN:

- Heart Failure Association of the European Society of Cardiology
- Heart Failure Society of America
- Japanese Heart Failure Society



INDICATIONS FOR ENDOMYOCARDIAL BIOPSY

- HTx rejection surveillance
- Myocarditis
- Cardiomyopathies
- Drug-related cardiotoxicity
- Amyloidosis
- Infiltrative and storage disorders
- Cardiac tumours

EMB v diagnostice RCM - histologie

Restrictive cardiomyopathy: definition and diagnosis

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

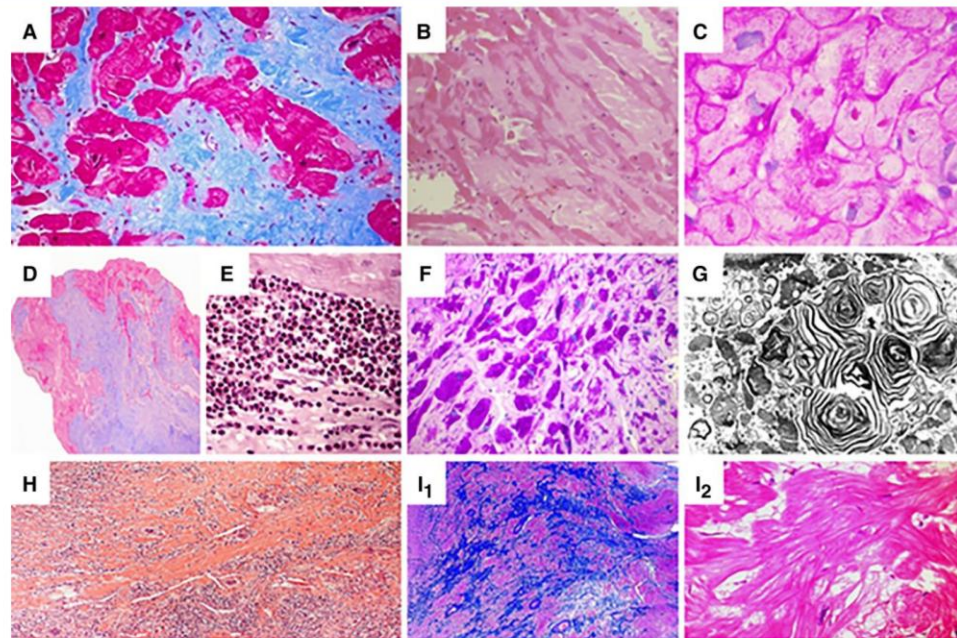


Figure 2 Myocardial tissue in nine different forms of restrictive cardiomyopathy (A) idiopathic restrictive cardiomyopathy; (B) cardiac amyloidosis (with enlargement of the extracellular spaces by amyloid fibres); (C) Danon disease (with intracellular glycogen deposits); (D) endomyocardial fibrosis (with 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

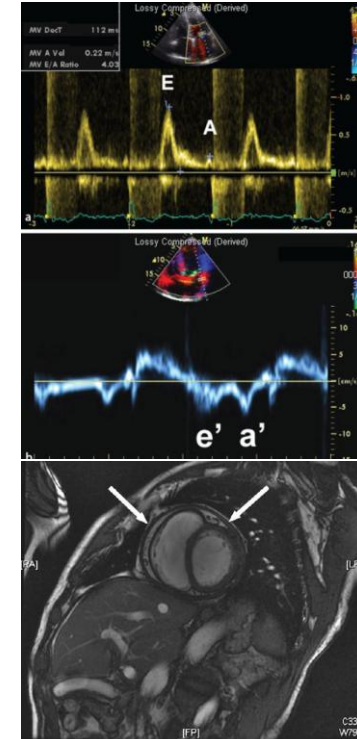
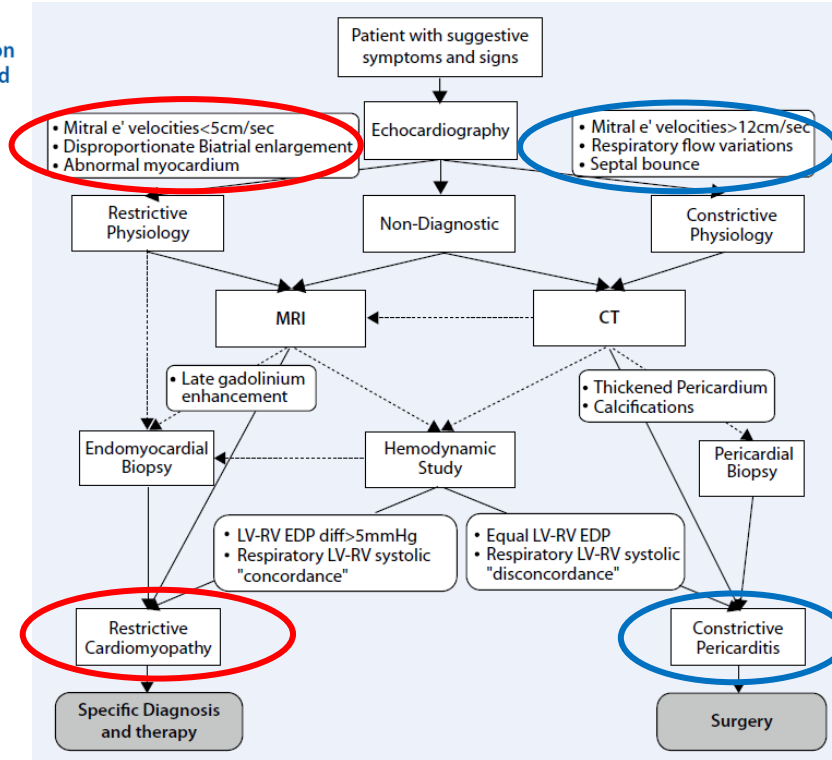
- Zvládnutí kongesce (cave dostatečný plnicí tlak k udržení CO)
- Reverzní remodelace není cílem léčby (žádná prognostická data pro RAASi)
- CO je závislý na tepové frekvenci (prodloužení diastoly nezvyšuje EDV!)
 - spíše negativní efekt BB
- Antikoagulace při fi síní
- Kauzální terapie, je-li dostupná

Diferenciální dg konstrikivní perikarditidy a restriktivní kardiomyopatie

Heiz 2012, 37:664-674
 DOI 10.5002/00059-012-3663-4
 Published online 1 September 2012
 © Urban & Vogel 2012

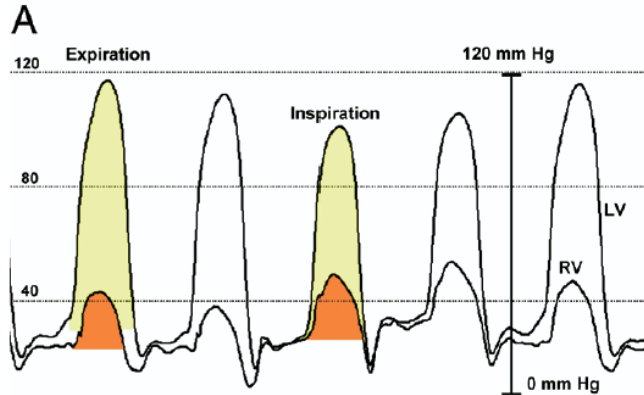
D.R. Zwas · I. Gotsman · D. Admon · A. Keren
 Heart Failure Center, Heart Institute, Hadassah University Hospital, Jerusalem

Advances in the differentiation of constrictive pericarditis and restrictive cardiomyopathy



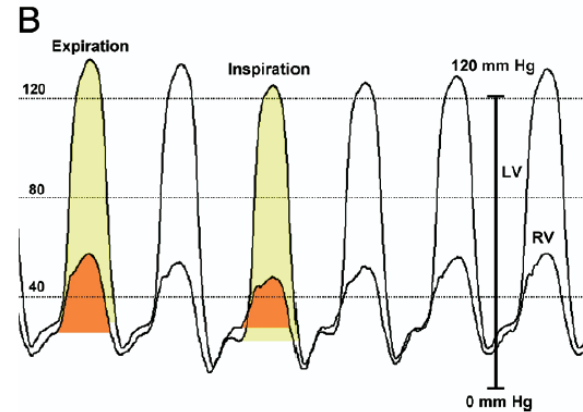
Diferenciální dg konstriktivní perikarditidy a restriktivní kardiomyopatie

Konstriktivní perikarditis



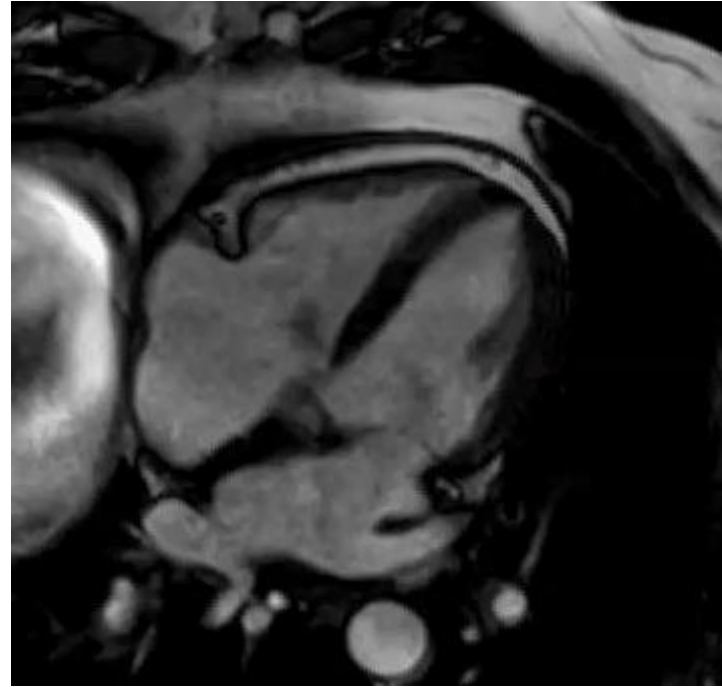
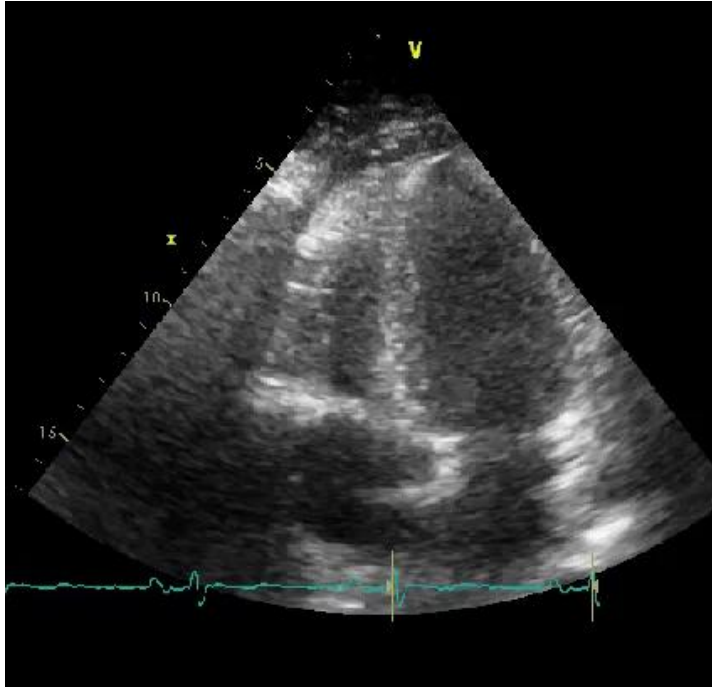
Diskordance systolických komorových tlaků
Zvýrazněná interventrikulární dependence

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!