Co se skrývá pod pojmem „nedilatovaná hypokinetická kardiomyopatie“?

J. Krejčí

Dilatační kardiomyopatie

- Jde o onemocnění s heterogenní etiologií a patogenezou, jehož prevalence DKMP je odhadována na 1:250-500

- DKMP je charakterizovaná dilatací a systolickou dysfunkcí levé komory /obou komor, která není vysvětlitelná abnormálními hemodynamickými podmínkami nebo koronární nemocí.

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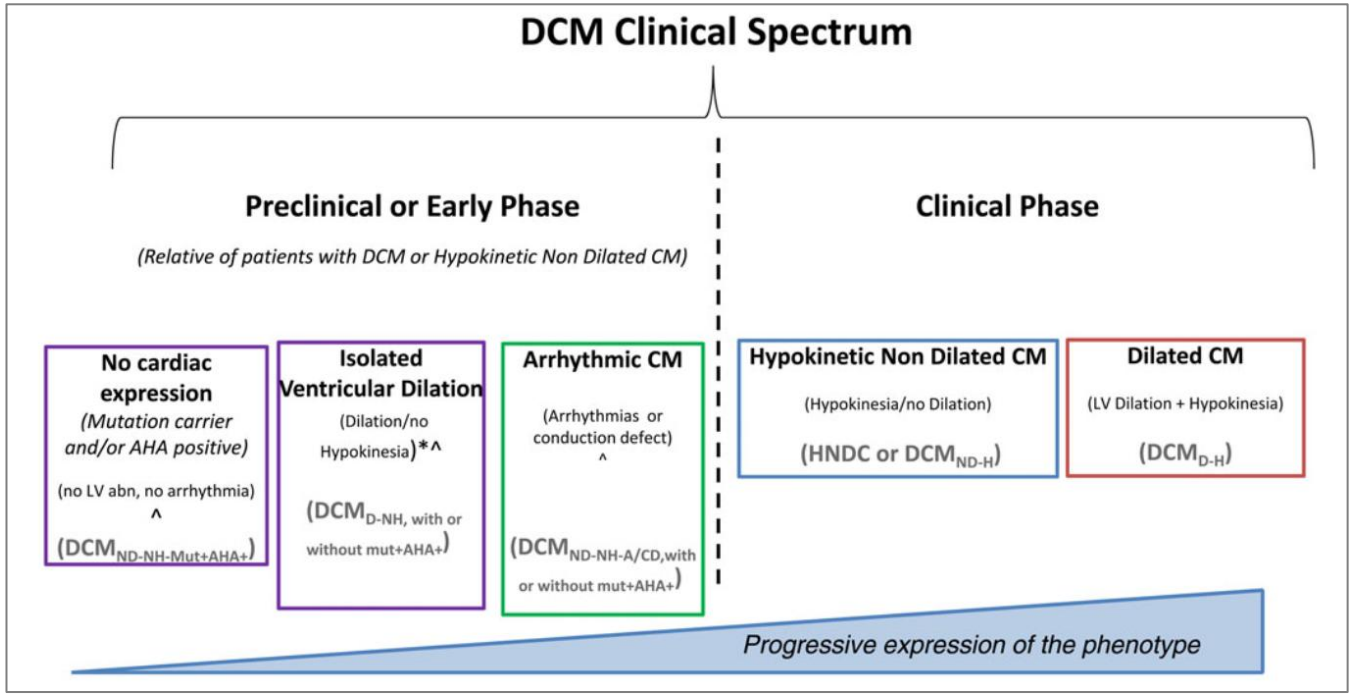
- **Musí být dilatace vždy přítomna? A jak je vůbec definována?**

Proposal for a revised definition of dilated cardiomyopathy, hypokinetic non-dilated cardiomyopathy, and its implications for clinical practice: a position statement of the ESC working group on myocardial and pericardial diseases



Hypokinetic non-dilated cardiomyopathy

Left ventricular or biventricular global systolic dysfunction without dilatation (defined as LVEF < 45%), not explained by abnormal loading conditions or coronary artery disease.

European Heart Journal (2016) **37**, 1850–1858

Definice dilatace levé komory

Left ventricular dilatation is defined by LV end-diastolic (ED) volumes or diameters >2SD from normal according to normograms (Z scores >2 standard deviations) corrected by body surface area (BSA) and age, or BSA and gender.

European Heart Journal (2016) **37**, 1850–1858

Speckle tracking echocardiography in hypokinetic non-dilated cardiomyopathy: comparison with dilated cardiomyopathy

determined by LVEDVI ≤ 75 mL/m^2 for men and LVEDVI ≤ 68 mL/m^2 in women obtained from Iranian subpop-

ESC HEART FAILURE
ESC Heart Failure 2020; **7**: 1909–1916

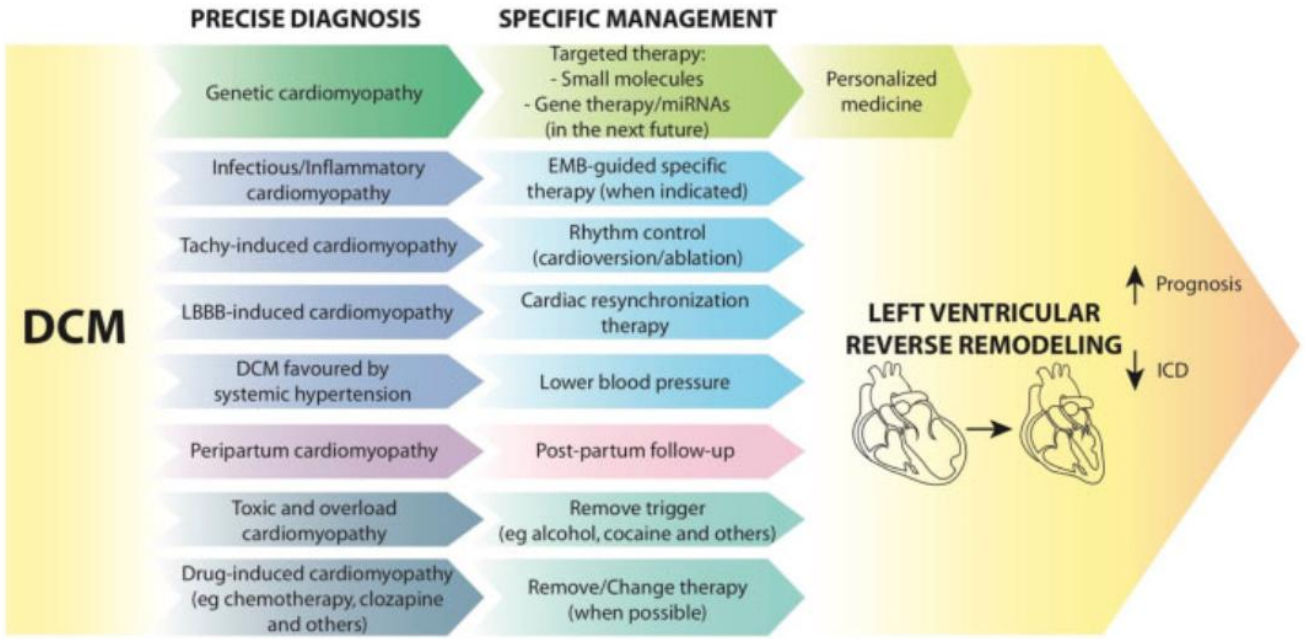
Clinical Phenotype and Genotype Associations With Improvement in Left Ventricular Function in Dilated Cardiomyopathy

DCM defined as LVEF <50% with an indexed LV end diastolic diameter [LVEDDi] >33 mm/m^2 [men] or >32 mm/m^2 [women] measured by echocardiography.

Circ Heart Fail. 2018;11:e005220.

„Dilatační“ kardiomyopatie?

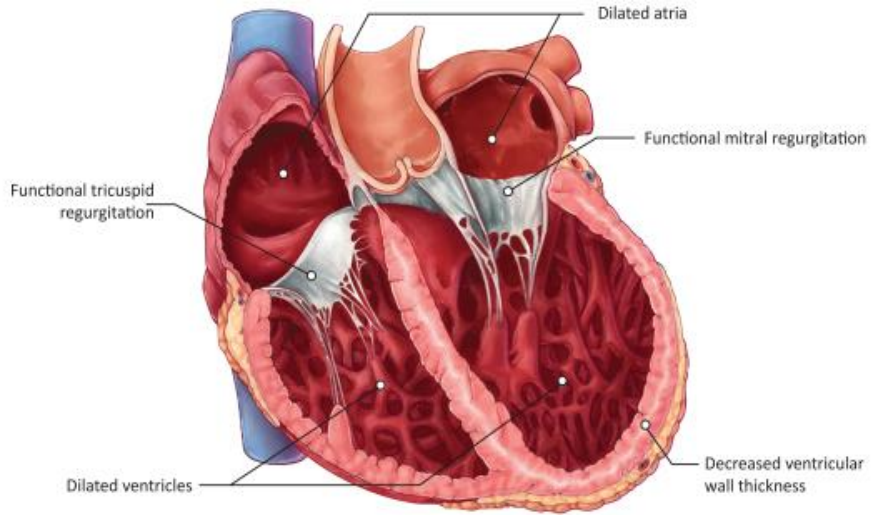
Dilated cardiomyopathy: so many cardiomyopathies!



European Heart Journal (2020) **41**, 3784–3786

„Dilatační“ kardiomyopatie není jedna diagnóza

Heart failure in cardiomyopathies: a position paper from the Heart Failure Association of the European Society of Cardiology

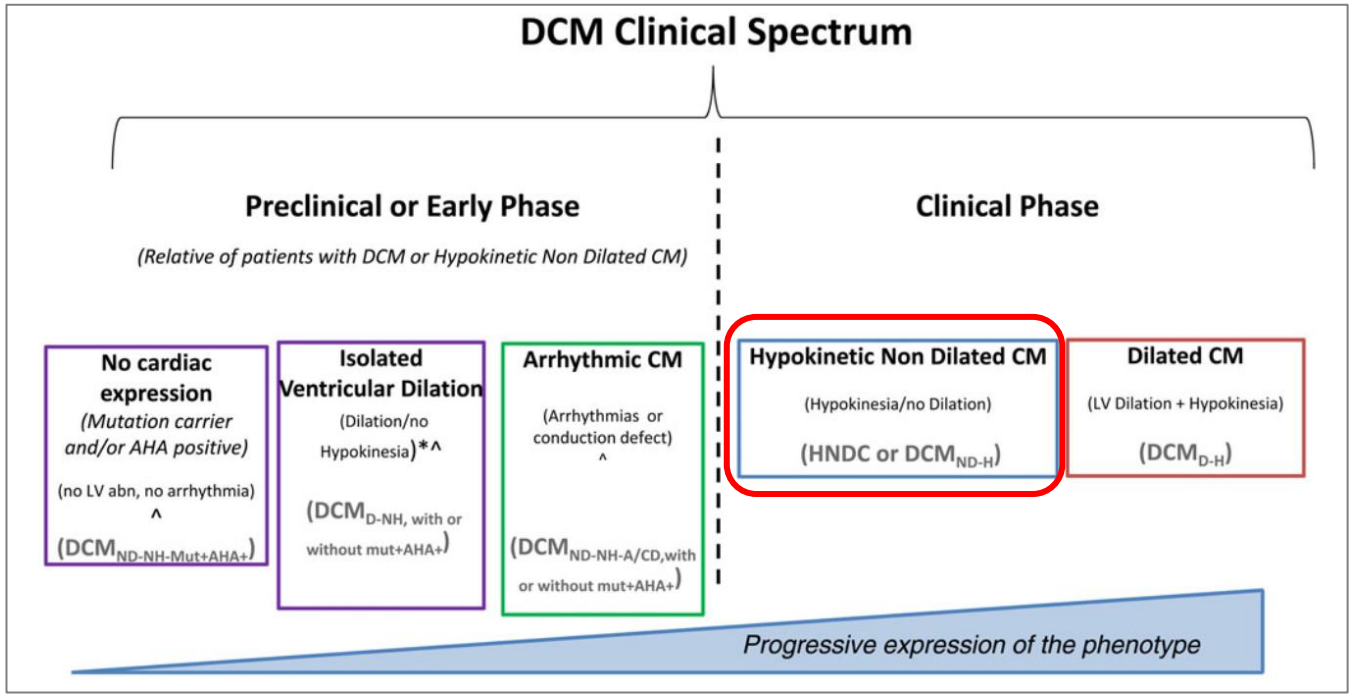


Dilated cardiomyopathy

Genetic	Infection	Systemic immune-mediated disease	Toxic and overload	Drugs	Endocrine/ metabolic	Peripartum
Cardiac phenotype: • titin (TTN) • lamin A/C (LMNA) • myosin heavy chain (MYH7) • troponin T (TNT2) • Myosin binding protein C (MYBPC3) phospholamban (PLN) **Neuromuscular disorders:** • Duchenne muscular dystrophy • Becker muscular dystrophy	**Post-myocarditis:** • Enteroviruses (e.g., Coxsackieviruses), • Parvovirus B19 • Adenoviruses, • Herpes Viruses (Human Cytomegalovirus, Epstein-Barr virus, Human Herpesvirus 6), • Echoviruses, • Hepatitis C Virus, • Influenza A virus **Other:** [e.g. HIV, Chagas]	**Autoimmune**: • Rheumatoid arthritis • Coeliac disease • Systemic lupus erythematosus • Dermatomyositis, polymyositis • Systemic sclerosis • Primary biliary cirrhosis • Vasculitis (some) • Myasthenia gravis • Pemphigus, pemphigoid **Autoinflammatory**: [e.g. Chron disease, ulcerative colitis, gout, reactive arthritis, rare monogenic disorders]	• Alcohol (ethanol) • Cocaine • Amphetamines • Ecstasy • Anabolic steroids • Arsenic • Cobalt • Carbon-monoxide • Lead • Iron-overload/haem ochromatosis • Amyloidosis	**Chemotherapy:** [anthracycline, trastuzumab, antimetabolites, alkylating agents, monoclonal antibodies, tyrosine kinase inhibitors, immunomodulating agents] **Other:** [clozapine, olzapine, chlorpromazine, risperidone, lithium, trycyclic antidepressants, ephedrine all-trans retinoic acid phenothiazines]	• Diabetes mellitus, • Acromegaly • Pheochromocytoma • Thyroid gland dysfunction	Idiopathic cardiomyopathy presenting with HFrEF towards the end of pregnancy, or in the months following delivery, where no other cause of HF is found

European Journal of Heart Failure (2019) **21**, 553–576

Proposal for a revised definition of dilated cardiomyopathy, hypokinetic non-dilated cardiomyopathy, and its implications for clinical practice: a position statement of the ESC working group on myocardial and pericardial diseases



- HNDC nemusí být jen vývojovou fází „klasické“ DCM
- některé etiologické podtypy výrazněji disponují k obrazu HNDC
- odlišný klinický obraz, prognózu i léčbu

European Heart Journal (2016) **37**, 1850–1858

Léčba DCM – „one-size-fits-all“ již není správný přístup!

Dilated cardiomyopathy in the era of precision medicine: latest concepts and developments

Keywords Dilated cardiomyopathy · DCM · Cardiomyopathies · Precision medicine · Inherited cardiac diseases · ARVC · LMNA · FLNC · BAG3 · TMEM43 · PLN · DSP · Athletic heart syndrome · Hypokinetic non-dilated cardiomyopathy · Molecular cardiology · Heart failure

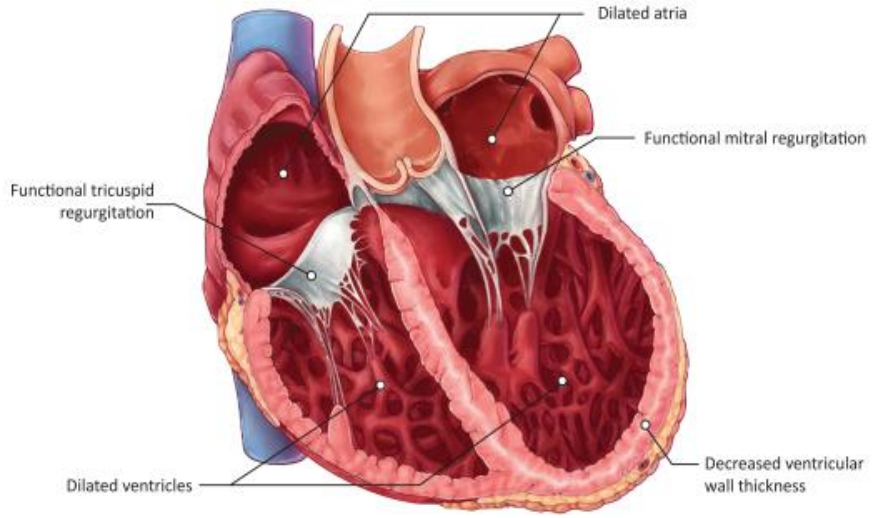
Heart Failure Reviews (2022) 27:1173–1191

Precision medicine

medical care designed to optimize efficiency or therapeutic benefit for particular groups of patients, especially by using genetic or molecular profiling.

Když je DCM spíše HNDC…

Heart failure in cardiomyopathies: a position paper from the Heart Failure Association of the European Society of Cardiology



Dilated cardiomyopathy

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- lamin A/C (LMNA)
- myosin heavy chain (MYH7)
- troponin T (TNT2)
- Myosin binding protein C (MYBPC3)
- phospholamban (PLN)

Neuromuscular disorders:
- Duchenne muscular dystrophy
- Becker muscular dystrophy

Infection

Post-myocarditis:
- Enteroviruses (e.g., Coxsackieviruses),
- Parvovirus B19
- Adenoviruses,
- Herpes Viruses (Human Cytomegalovirus, Epstein-Barr virus, Human Herpesvirus 6),
- Echoviruses,
- Hepatitis C Virus,
- Influenza A virus

Other:
[e.g. HIV, Chagas]

Systemic immune-mediated disease

Autoimmune:
- Rheumatoid arthritis
- Coeliac disease
- Systemic lupus erythematosus
- Dermatomyositis, polymyositis
- Systemic sclerosis
- Primary biliary cirrhosis
- Vasculitis (some)
- Myasthenia gravis
- Pemphigus, pemphigoid

Autoinflammatory:
[e.g. Chron disease, ulcerative colitis, gout, reactive arthritis, rare monogenic disorders]

Toxic and overload
- Alcohol (ethanol)
- Cocaine
- Amphetamines
- Ecstasy
- Anabolic steroids
- Arsenic
- Cobalt
- Carbon-monoxide
- Lead
- Iron-overload/haemochromatosis
- Amyloidosis

Drugs

Chemotherapy:
[anthracycline, trastuzumab, antimetabolites, alkylating agents, monoclonal antibodies, tyrosine kinase inhibitors, immunomodulating agents]

Other:
[clozapine, olzapine, chlorpromazine, risperidone, lithium, trycyclic antidepressants, ephedrine all-trans retinoic acid phenothiazines]

Endocrine/ metabolic
- Diabetes mellitus,
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Peripartum

Idiopathic cardiomyopathy presenting with HFrEF towards the end of pregnancy, or in the months following delivery, where no other cause of HF is found

European Journal of Heart Failure (2019) **21**, 553–576

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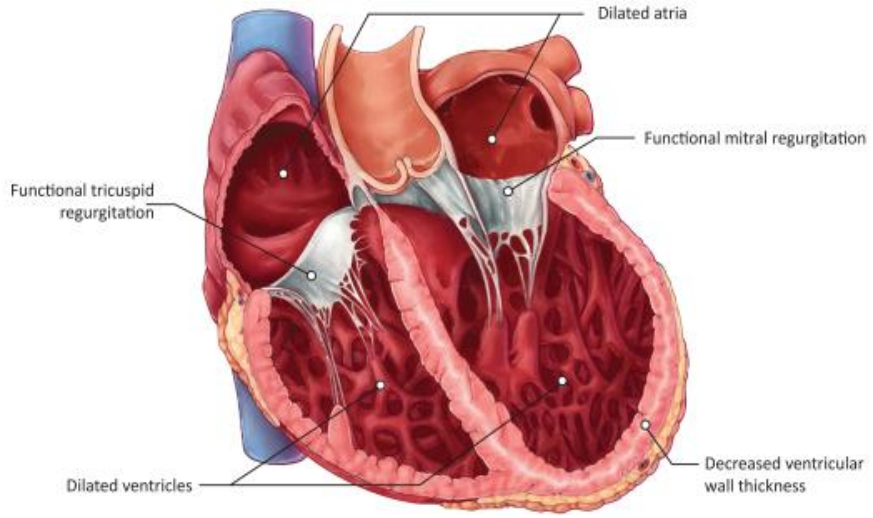
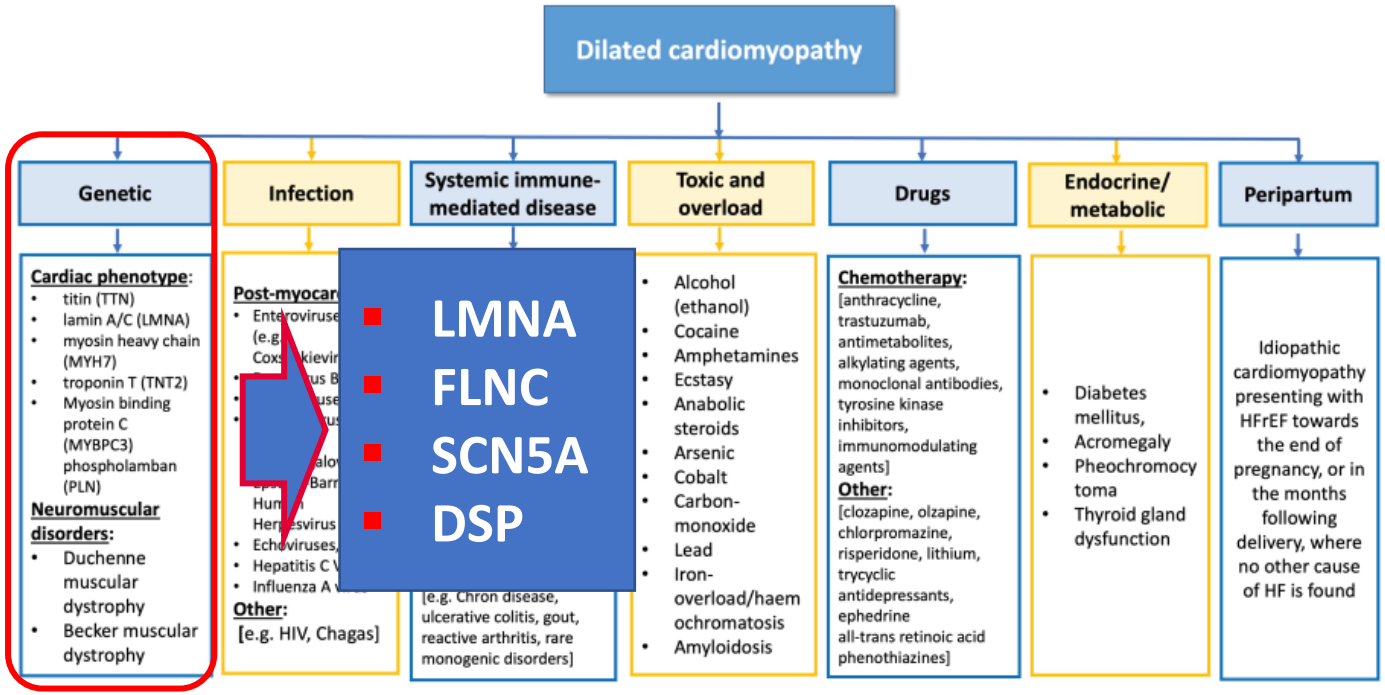
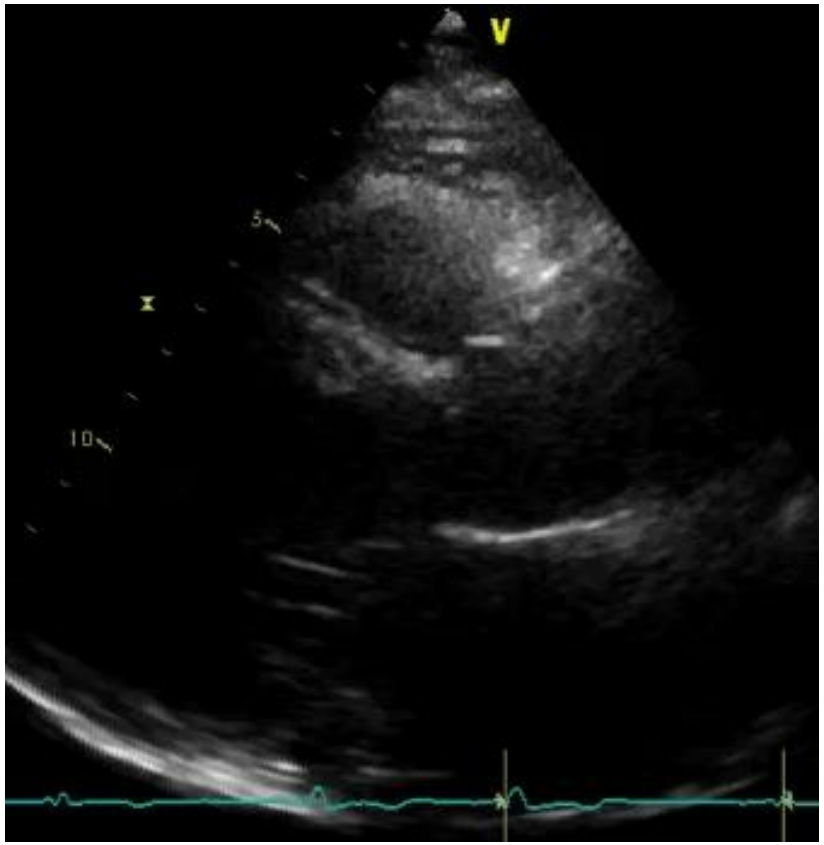
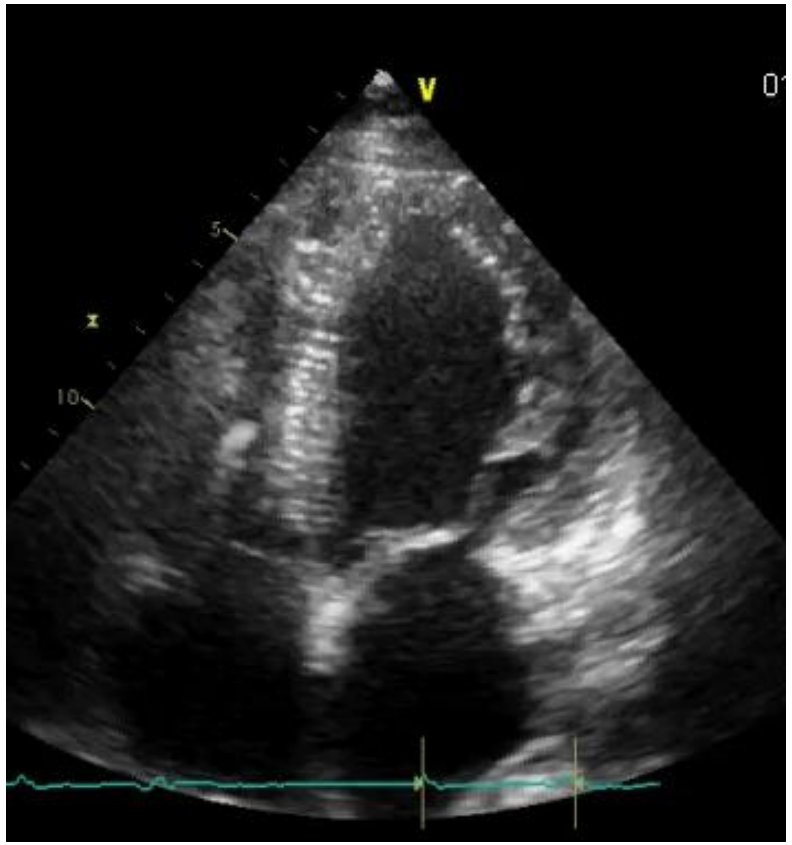




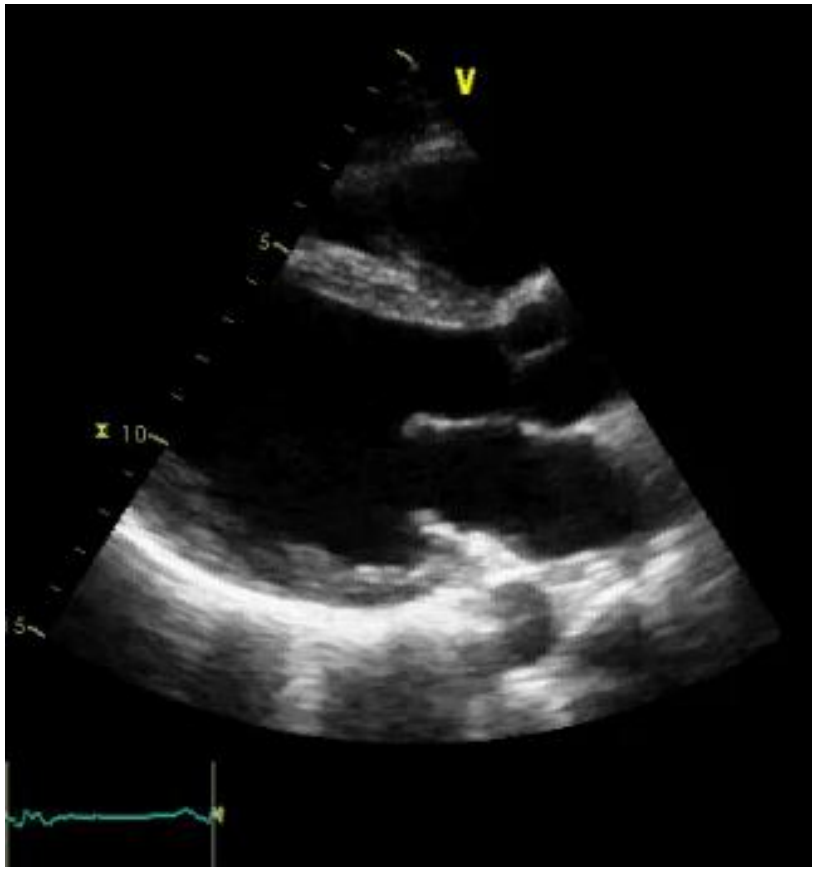
European Journal of Heart Failure (2019) **21**, 553–576

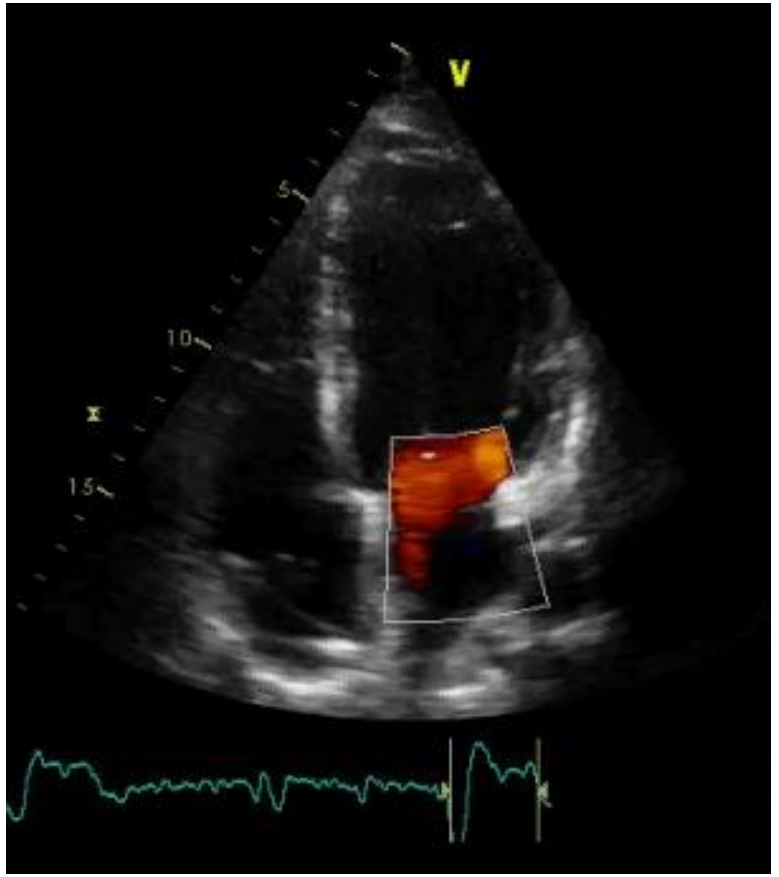
HNDC s mutací LMNA





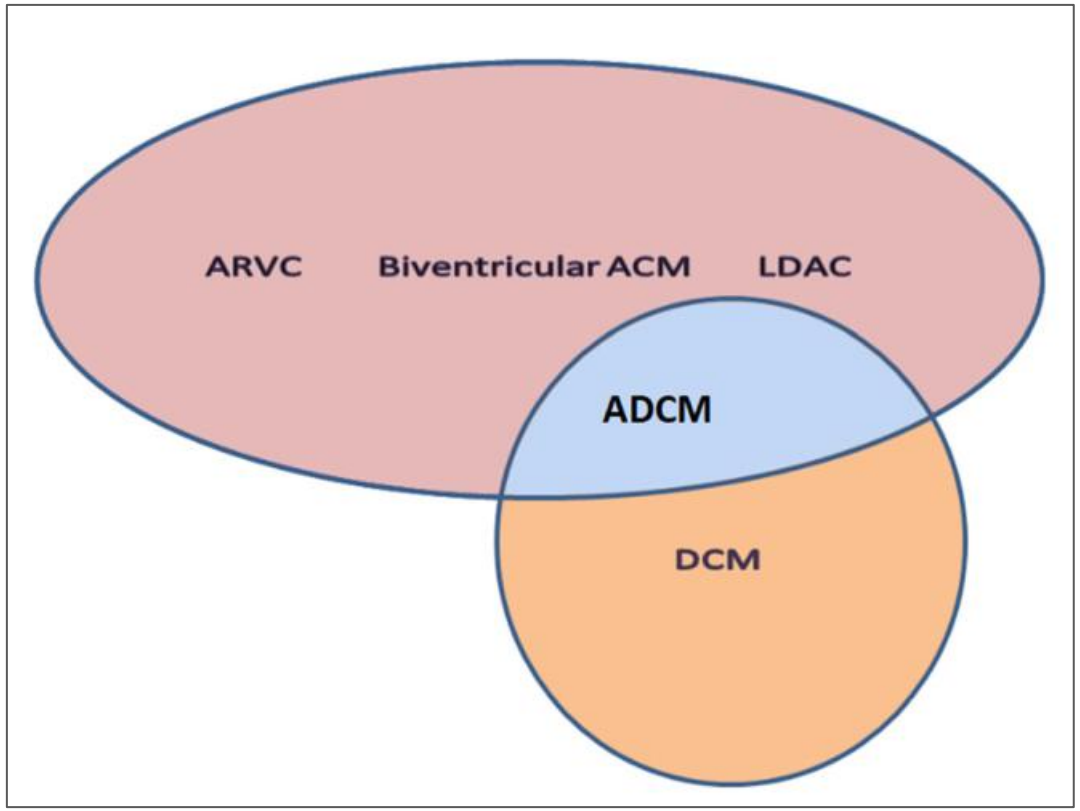
HNDC s mutací DSP





Fenotypový překryv DCM a LVAC

Emerging concepts in arrhythmogenic dilated cardiomyopathy



- Až třetina nemocných s DCM může být *post mortem* reklasifikována na ACM
- Nejasnosti v definici ADCM vs LDAC

Heart Failure Reviews (2021) 26:1219–1229

Terminologické nejasnosti…ADCM a LVAC

Diagnosis of arrhythmogenic cardiomyopathy: The Padua criteria

While the adjective “arrhythmic” refers to the nonspecific outcome of every heart disease which manifests with rhythm and conduction disturbances, the adjective “arrhythmogenic” (the suffix *-genic* according to its etymology from ancient Greek -γενής means “tending to generate” arrhythmias) is disease-specific and denotes the distinctive *propensity* of ACM to develop ventricular arrhythmias, as a clinical reflection of the underlying fibro-fatty myocardial replacement which is the pathologic hallmark of the disease.

D. Corrado et al. / International Journal of Cardiology 319 (2020) 106–114

“Padua criteria” for diagnosis of Arrhythmogenic Cardiomyopathy.

Category	Right ventricle (upgraded 2010 ITF diagnostic criteria)	Left ventricle (new diagnostic criteria)
I. Morpho-functional ventricular abnormalities	*By echocardiography, CMR or angiography:* *Major* • Regional RV akinesia, dyskinesia, or bulging *plus* one of the following: - global RV dilatation (increase of RV EDV according to the imaging test specific nomograms) - global RV systolic dysfunction (reduction of RV EF according to the imaging test specific nomograms) *Minor* • Regional RV akinesia, dyskinesia or aneurysm of RV free wall	*By echocardiography, CMR or angiography:Minor* • Global LV systolic dysfunction (depression of LV EF or reduction of echocardiographic global longitudinal strain), with or without LV dilatation (increase of LV EDV according to the imaging test specific nomograms for age, sex, and BSA) *Minor* • Regional LV hypokinesia or akinesia of LV free wall, septum, or both
II. Structural myocardial abnormalities	*By CE-CMR:Major* • Transmural LGE (stria pattern) of ≥1 RV region(s) (inlet, outlet, and apex in 2 orthogonal views) *By EMB (limited indications):Major* • Fibrous replacement of the myocardium in ≥1 sample, with or without fatty tissue	*By CE-CMR:Major* • LV LGE (stria pattern) of ≥1 Bull's Eye segment(s) (in 2 orthogonal views) of the free wall (subepicardial or midmyocardial), septum, or both (excluding septal junctional LGE)
III. Repolarization abnormalities	*Major* • Inverted T waves in right precordial leads (V_1,V_2, and V_3) or beyond in individuals with complete pubertal development (in the absence of complete RBBB) *Minor* • Inverted T waves in leads V1 and V2 in individuals with completed pubertal development (in the absence of complete RBBB) • Inverted T waves in V1,V2,V3 and V4 in individuals with completed pubertal development in the presence of complete RBBB.	*Minor* • Inverted T waves in left precordial leads (V_4-V_6) (in the absence of complete LBBB)
IV. Depolarization abnormalities	*Minor* • Epsilon wave (reproducible low-amplitude signalsbetween end of QRS complex to onset of the T wave) in the right precordial leads (V1 to V3) • Terminal activation duration of QRS ≥55 ms measured from the nadir of the S wave to the end of the QRS, including R', in V1, V2, or V3 (in the absence of complete RBBB)	*Minor* • Low QRS voltages (<0.5 mV peak to peak) in limb leads (in the absence of obesity, emphysema, or pericardial effusion)
V. Ventricular arrhythmias	*Major* • Frequent ventricular extrasystoles (>500 per 24 h), non-sustained or sustained ventricular tachycardia of LBBB morphology *Minor* • Frequent ventricular extrasystoles (>500 per 24 h), non-sustained or sustained ventricular tachycardia of LBBB morphology with inferior axis (“RVOT pattern”)	*Minor* • Frequent ventricular extrasystoles (>500 per 24 h), non-sustained or sustained ventricular tachycardia with a RBBB morphology (excluding the “fascicular pattern”)
VI. Family history/genetics	*Major* • ACM confirmed in a first-degree relative who meets diagnostic criteria • ACM confirmed pathologically at autopsy or surgery in a first degree relative • Identification of a pathogenic or likely pathogenetic ACM mutation in the patient under evaluation *Minor* • History of ACM in a first-degree relative in whom it is not possible or practical to determine whether the family member meets diagnostic criteria • Premature sudden death (<35 years of age) due to suspected ACM in a first-degree relative • ACM confirmed pathologically or by diagnostic criteria in a second-degree relative	

Diferenciální dg DCM a LVAC

Arrhythmogenic right ventricular cardiomyopathy: evaluation of the current diagnostic criteria and differential diagnosis

Table 6 **Dilated cardiomyopathy vs. left-dominant arrhythmogenic right ventricular cardiomyopathy**

	Dilated cardiomyopathy	Left-dominant ARVC
Inheritance	≤35% (AD)	>50% (AD, AR)
Predominant genetic background	Mutations of genes encoding for cytoskeleton, muscular sarcomere, and nuclear envelope proteins	Mutations of genes encoding for desmosomal proteins, PLN, or FLN-C
Main clinical manifestations	Heart failure, cardiac arrest, palpitations	Palpitations, syncope, cardiac arrest
ECG abnormalities	Left ventricular hypertrophy with a strain pattern of ST-segment; left bundle branch block	Low QRS voltages in limb leads; negative T waves in lateral leads; negative T waves in right precordial leads (biventricular form)

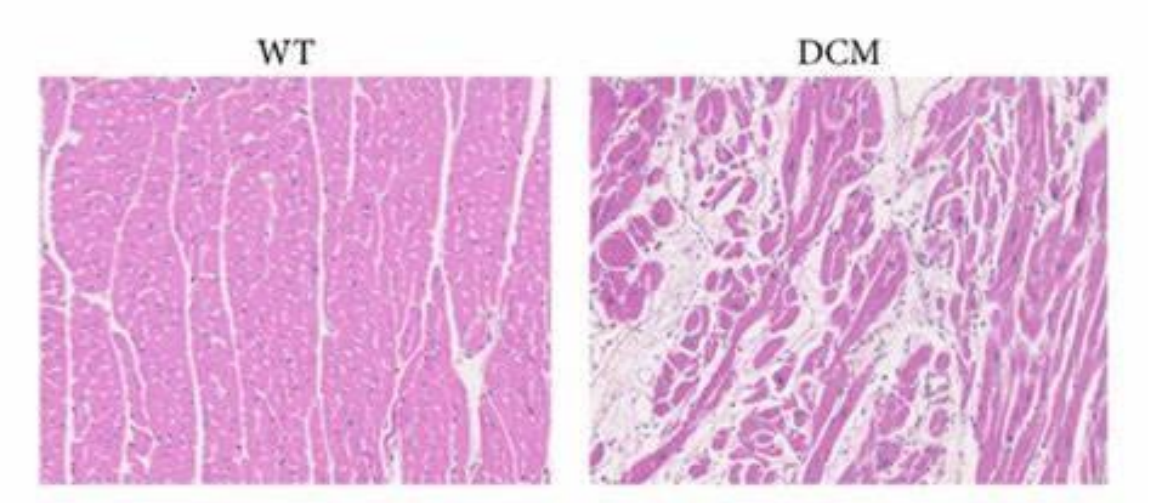
European Heart Journal (2020) **41**, 1414–1427

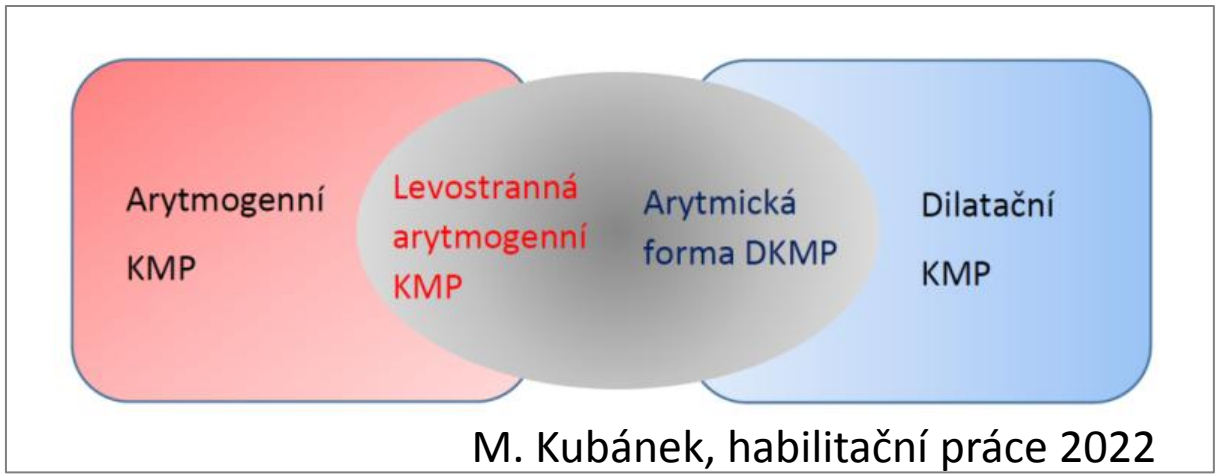
Diferenciální dg DCM/ADCM a LVAC

Echocardiography and cardiac magnetic resonance imaging findings	Dilated and hypokinetic LV with no or patchy non-ischaemic (mid-myocardial) LGE (septum) Regional wall motion abnormalities (uncommon). Systolic LV dysfunction unrelated to the global extent of LGE	Non-dilated and hypokinetic LV with large amount of non-ischemic (subepicardial) LGE (inferolateral LV wall) Regional wall motion abnormalities (common). Systolic LV dysfunction related to the global extent of LGE
EMB features	Non-specific myocardial abnormalities	Fibrofatty myocardial replacement
Types of ventricular arrhythmias	PVBs and NSVT (RBBB pattern); sustained VT(uncommon); VF	PVBs, NSVT, and monomorphic sustained VT (RBBB pattern; both LBBB and RBBB patterns in biventricular form); VF
Mechanism of VT	Scar-related or functional re-entry (branch to branch re-entry)	Scar-related re-entry
Most common site of VT origin	Intramural septum	Subepicardial infero-lateral LV free wall

European Heart Journal (2020) **41**, 1414–1427

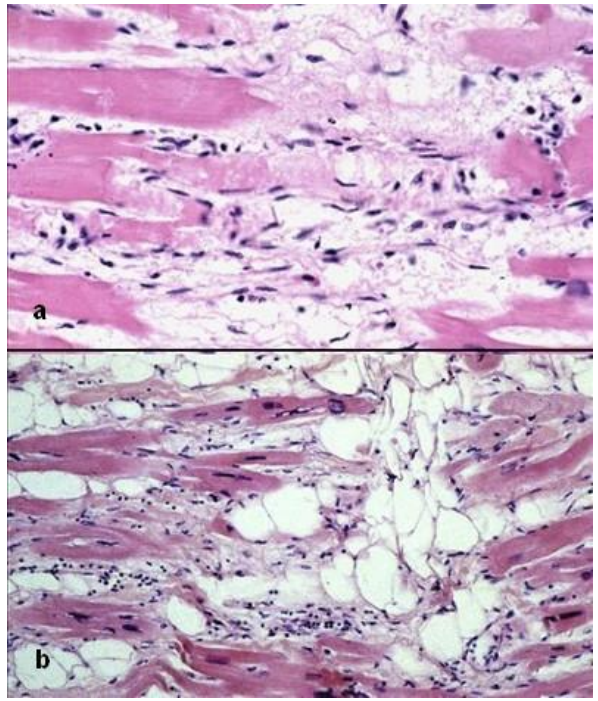
Diferenciální dg DCM a LVAC



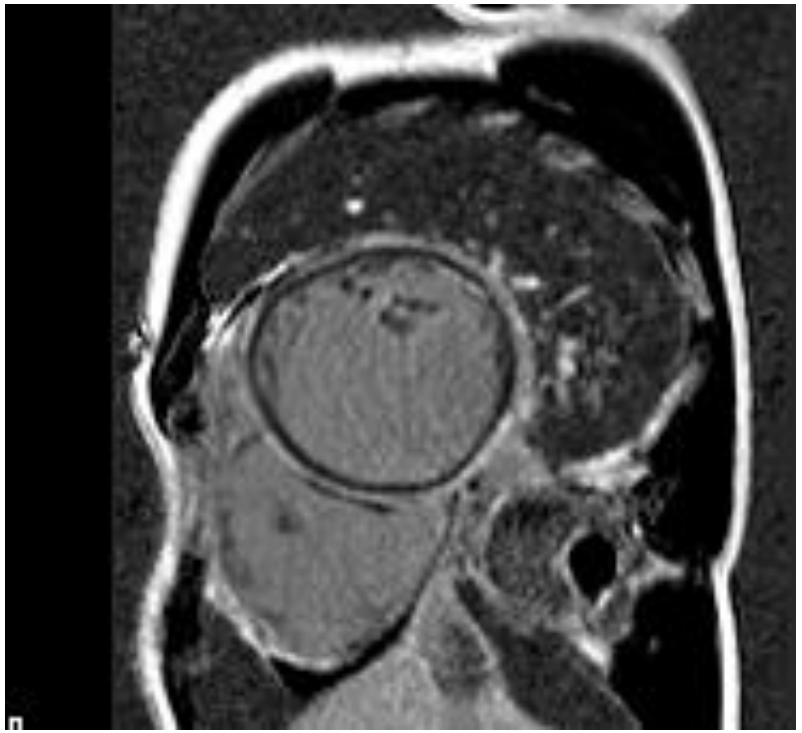


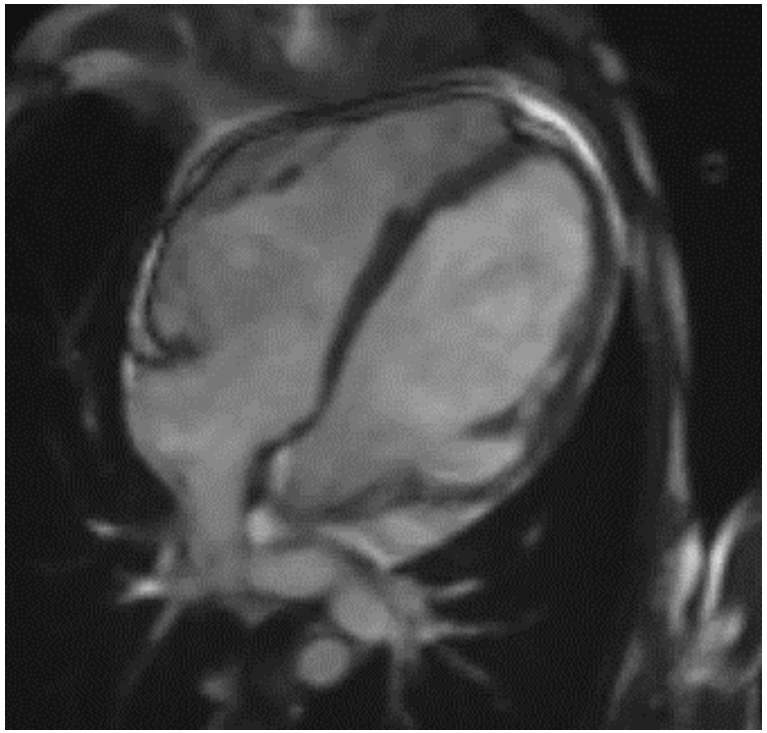
M. Kubánek, habilitační práce 2022

ACM/LVAC



Diferenciální dg DCM a LVAC





Když je DCM spíše ADCM…

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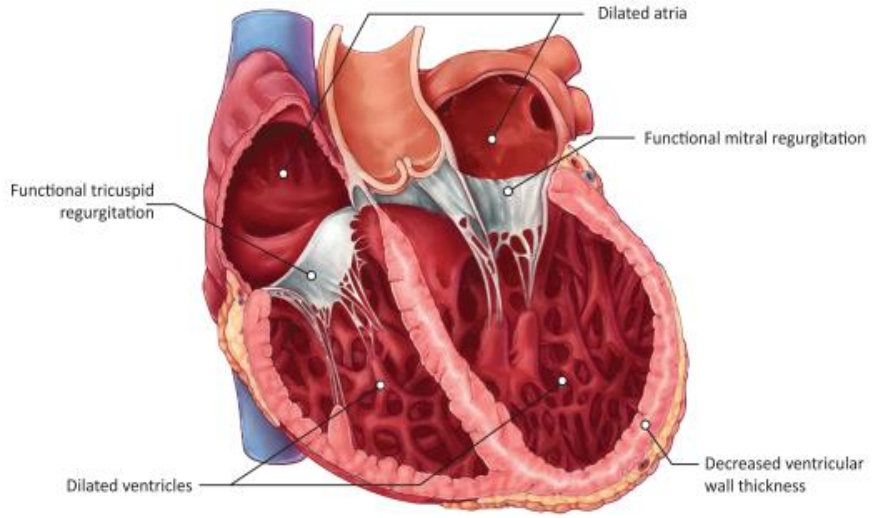

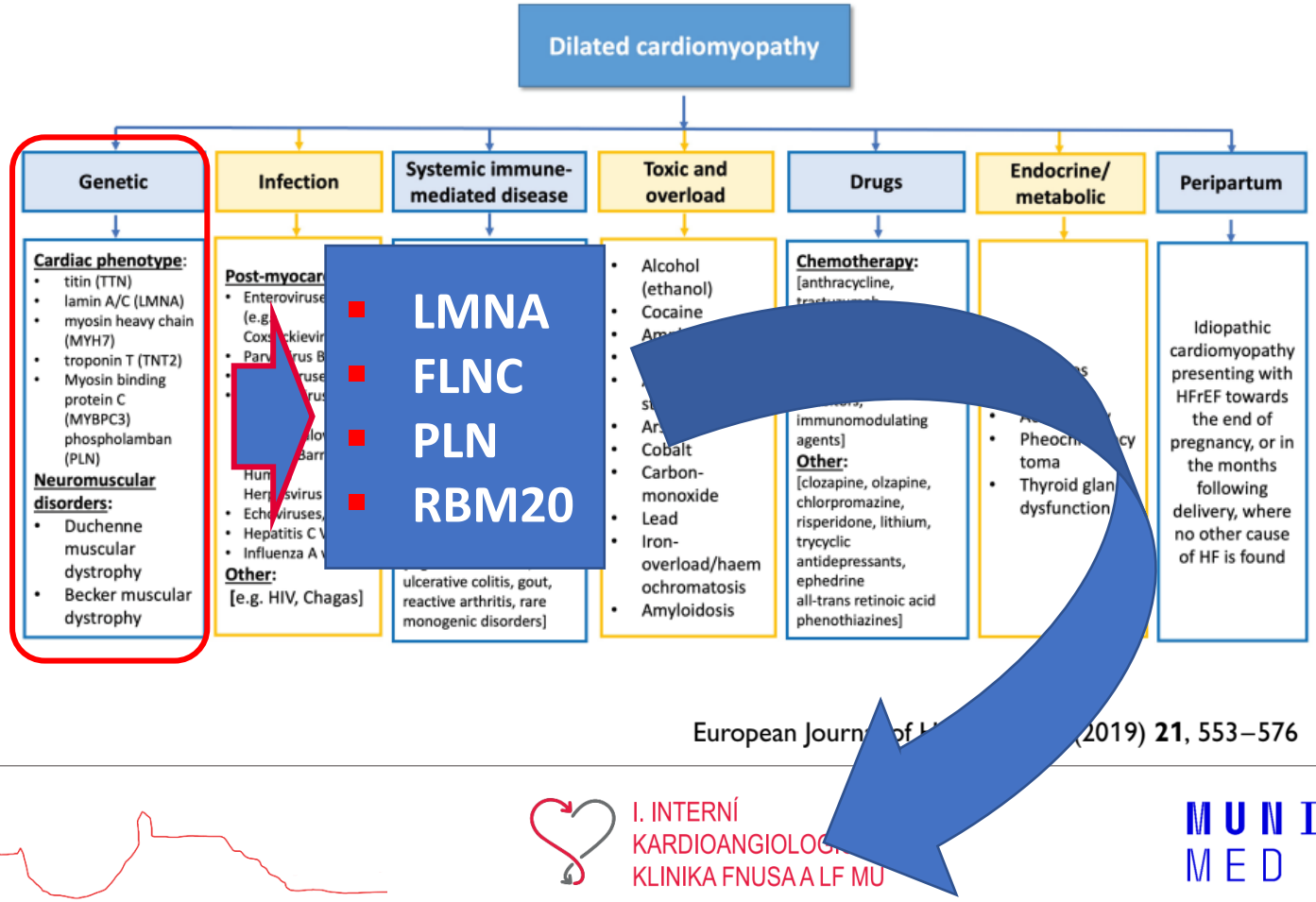

European Journ of (2019) **21**, 553–576

Prevence SCD a genetika

2022 ESC Guidelines for the management of patients with ventricular arrhythmias and the prevention of sudden cardiac death

DCM/HNDCM	
Genetic testing (including at least *LMNA*, *PLN*, *RBM20*, and *FLNC* genes) is recommended in patients with DCM/HNDCM and AV conduction delay at <50 years, or who have a family history of DCM/HNDCM or SCD in a first-degree relative (at age <50 years).	I
Genetic testing (including at least *LMNA*, *PLN*, *RBM20*, and *FLNC* genes) should be considered for risk stratification in patients with apparently sporadic DCM/HNDCM, who present at young age or with signs suspicious for an inherited aetiology.	IIa
ICD implantation should be considered in DCM/HNDCM patients with an LVEF <50% and ≥2 risk factors (syncope, LGE on CMR, inducible SMVT at PES, pathogenic mutations in LMNA, *PLN*, *FLNC*, and *RBM20* genes).	IIa

European Heart Journal (2022) **43**, 3997–4126

Když je DCM spíše HNDC…

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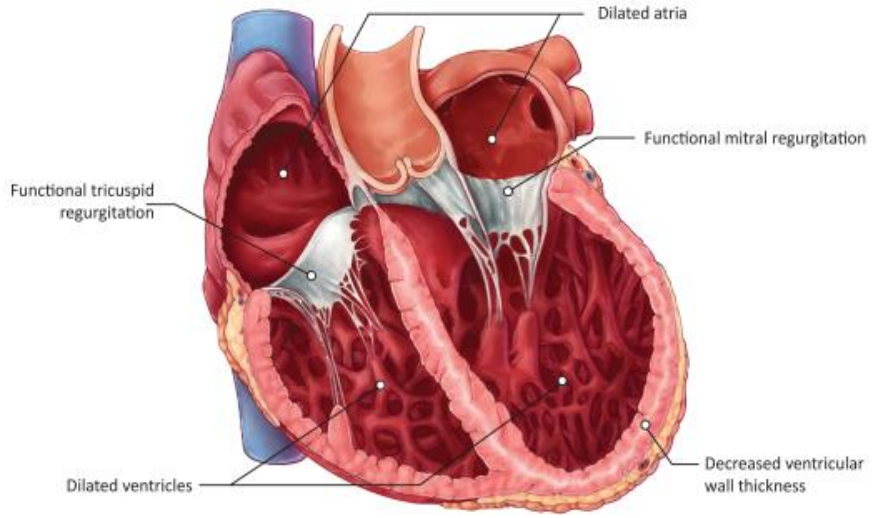

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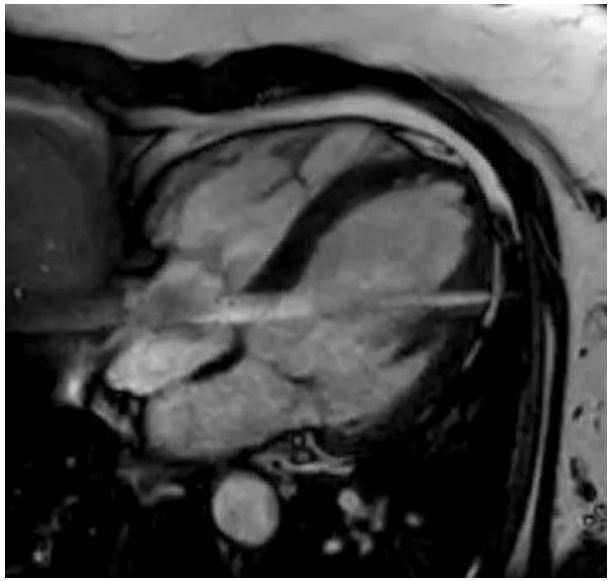
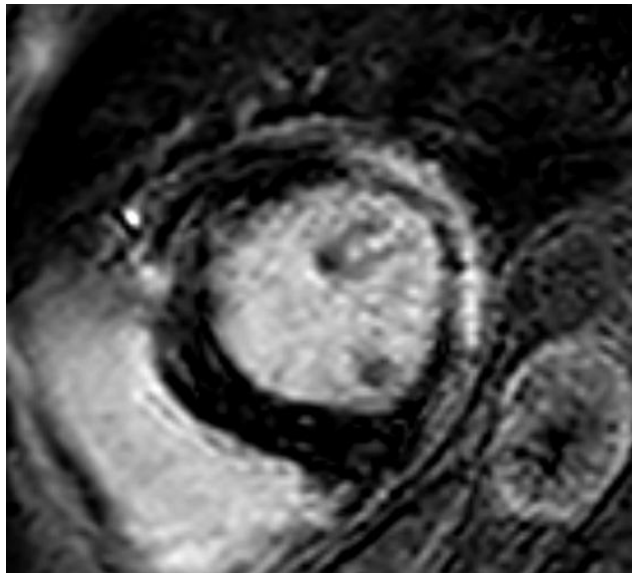
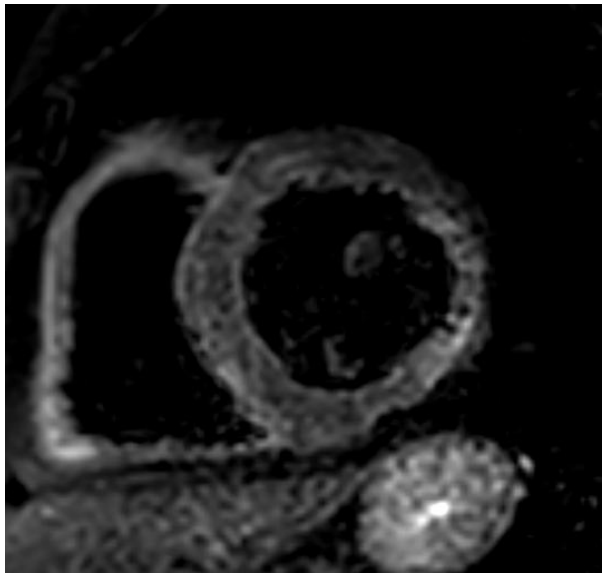
European Journal of Heart Failure (2019) **21**, 553–576

HNDC - DCMi / ZKMP



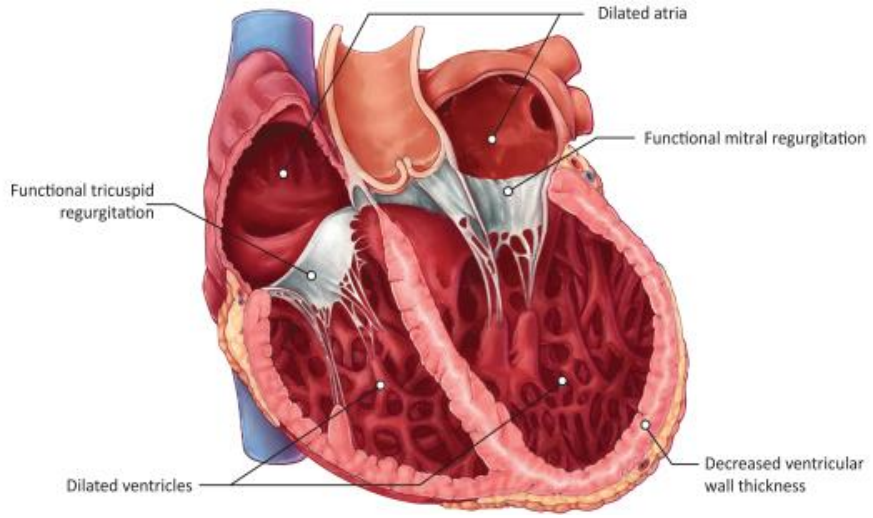




- Zejména u fulminantní myokarditidy nemusí být vyjádřena dilatace LK
- Může být přítomna dokonce „hypertrofie stěn“ LK daná edémem tkáně
- V našem souboru více než 250 nemocných s RODCM s provedením EMB bylo prokázáno, že u ZKMP je statisticky menší LVEDD než u DKMP

Když je DCM spíše HNDC…

Heart failure in cardiomyopathies: a position paper from the Heart Failure Association of the European Society of Cardiology



Dilated cardiomyopathy

Genetic	Infection	Systemic immune-mediated disease	Toxic and overload	Drugs	Endocrine/ metabolic	Peripartum
Cardiac phenotype: • titin (TTN) • lamin A/C (LMNA) • myosin heavy chain (MYH7) • troponin T (TNT2) • Myosin binding protein C (MYBPC3) phospholamban (PLN) **Neuromuscular disorders:** • Duchenne muscular dystrophy • Becker muscular dystrophy	**Post-myocarditis:** • Enteroviruses (e.g., Coxsackieviruses), • Parvovirus B19 • Adenoviruses, • Herpes Viruses (Human Cytomegalovirus, Epstein-Barr virus, Human Herpesvirus 6), • Echoviruses, • Hepatitis C Virus, • Influenza A virus **Other:** [e.g. HIV, Chagas]	**Autoimmune**: • Rheumatoid arthritis • Coeliac disease • Systemic lupus erythematosus • Dermatomyositis, polymyositis • Systemic sclerosis • Primary biliary cirrhosis • Vasculitis (some) • Myasthenia gravis • Pemphigus, pemphigoid **Autoinflammatory**: [e.g. Chron disease, ulcerative colitis, gout, reactive arthritis, rare monogenic disorders]	• Alcohol (ethanol) • Cocaine • Amphetamines • Ecstasy • Anabolic steroids • Arsenic • Cobalt • Carbon-monoxide • Lead • Iron-overload/haem ochromatosis • Amyloidosis	**Chemotherapy:** [anthracycline, trastuzumab, antimetabolites, alkylating agents, monoclonal antibodies, tyrosine kinase inhibitors, immunomodulating agents] **Other:** [clozapine, olzapine, chlorpromazine, risperidone, lithium, trycyclic antidepressants, ephedrine all-trans retinoic acid phenothiazines]	• Diabetes mellitus, • Acromegaly • Pheochromocytoma • Thyroid gland dysfunction	Idiopathic cardiomyopathy presenting with HFrEF towards the end of pregnancy, or in the months following delivery, where no other cause of HF is found

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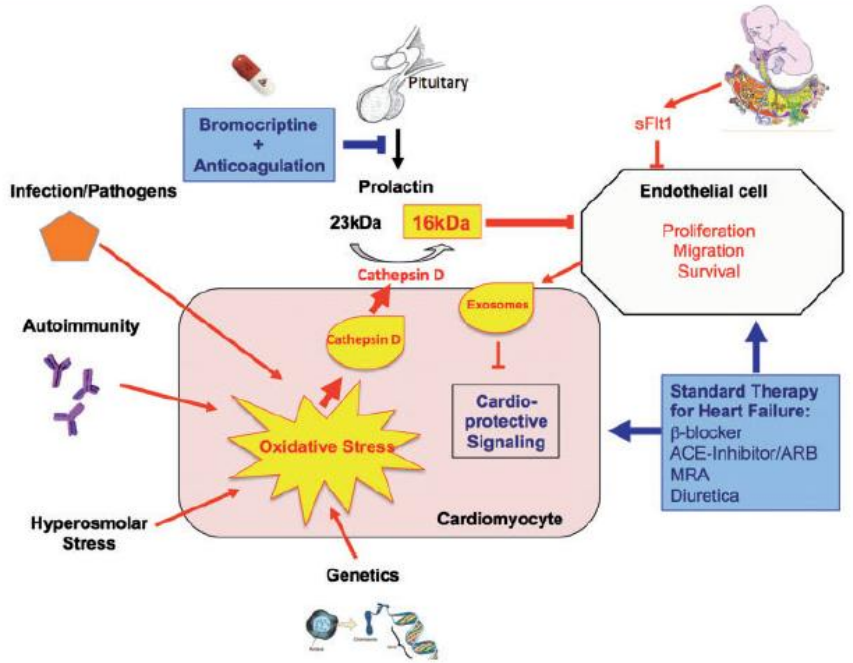
Peripartální kardiomyopatie



PPCM jako vaskulární a hormonální onemocnění

- Natriuretické peptidy, troponin (míra postižení myokardu, prognóza)
- **Echokardiografie (EF LK ≤ 45%, LVEDD může být v normě)**
- **V EMB** může být v 9 - 62% nález myokarditidy.

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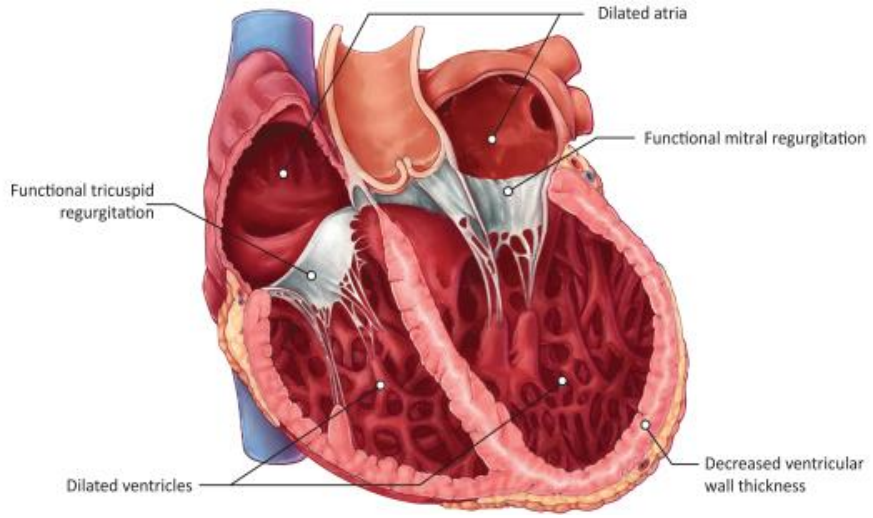

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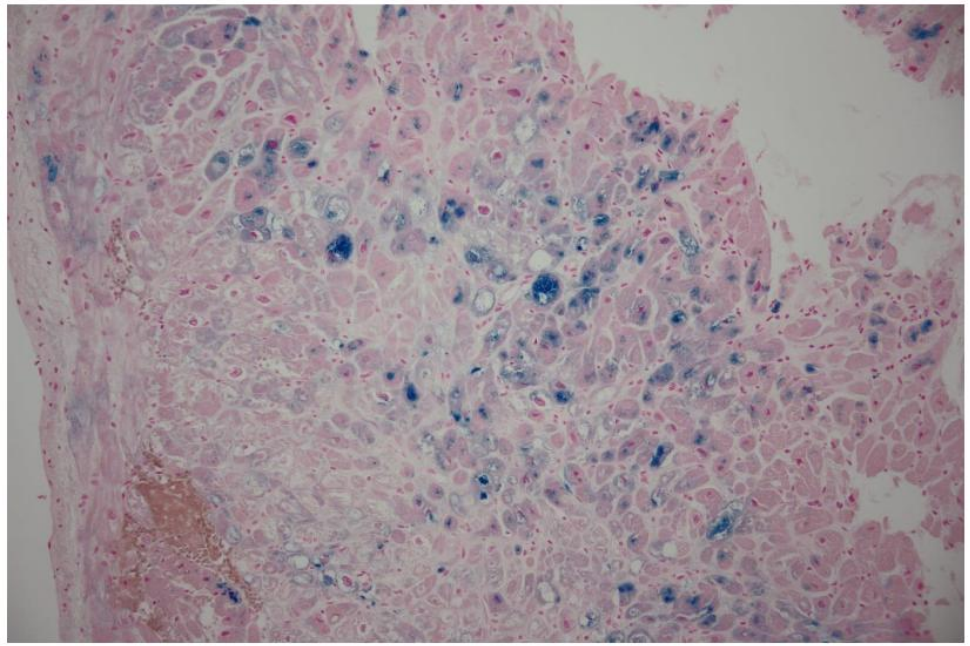
Case Reports > Can J Cardiol. 2016 Dec;32(12):1574.e1-1574.e3. doi: 10.1016/j.cjca.2016.07.589.
Epub 2016 Jul 28.

Successful Treatment of Iron-Overload Cardiomyopathy in Hereditary Hemochromatosis With Deferoxamine and Deferiprone

Lydie Tauchenová [1], Barbora Křížová [1], Miloš Kubánek [2], Soňa Fraňková [3], Vojtěch Melenovský [1], Jaroslav Tintěra [4], Dana Kautznerová [4], Jana Malušková [5], Milan Jirsa [6], Josef Kautzner [1]

Obrázek 6: Vzorek z endomyokardiální biopsie s pozitivním barvením na železo (Berlínská modř) u pacienta s primární hemochromatózou. Z archivu IKEM.



- Obraz HNDC či RCM
- TSAT nad 45% a ferritin nad 300 ug/L
- Detekce patogenních variant HFE
- Typický nález na MRI se zkrácením T2* relaxačního času
- Typický nález v EMB

M. Kubánek, habilitační práce 2022

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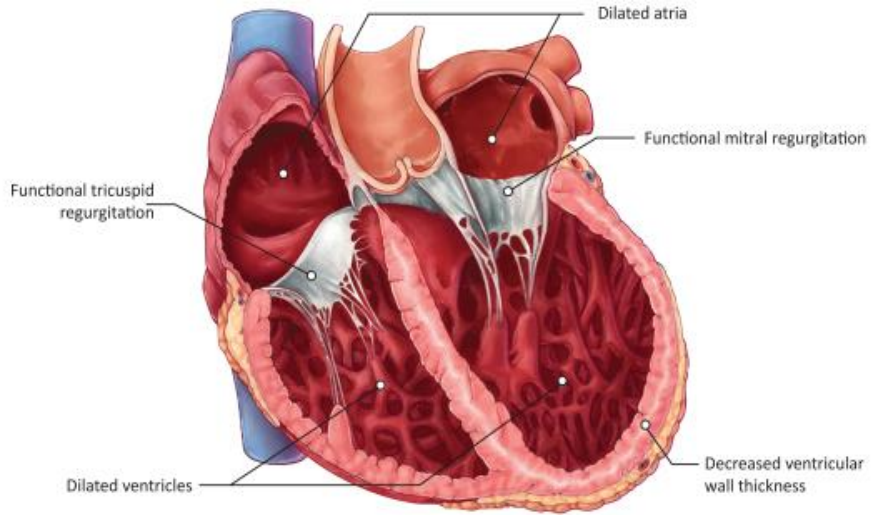

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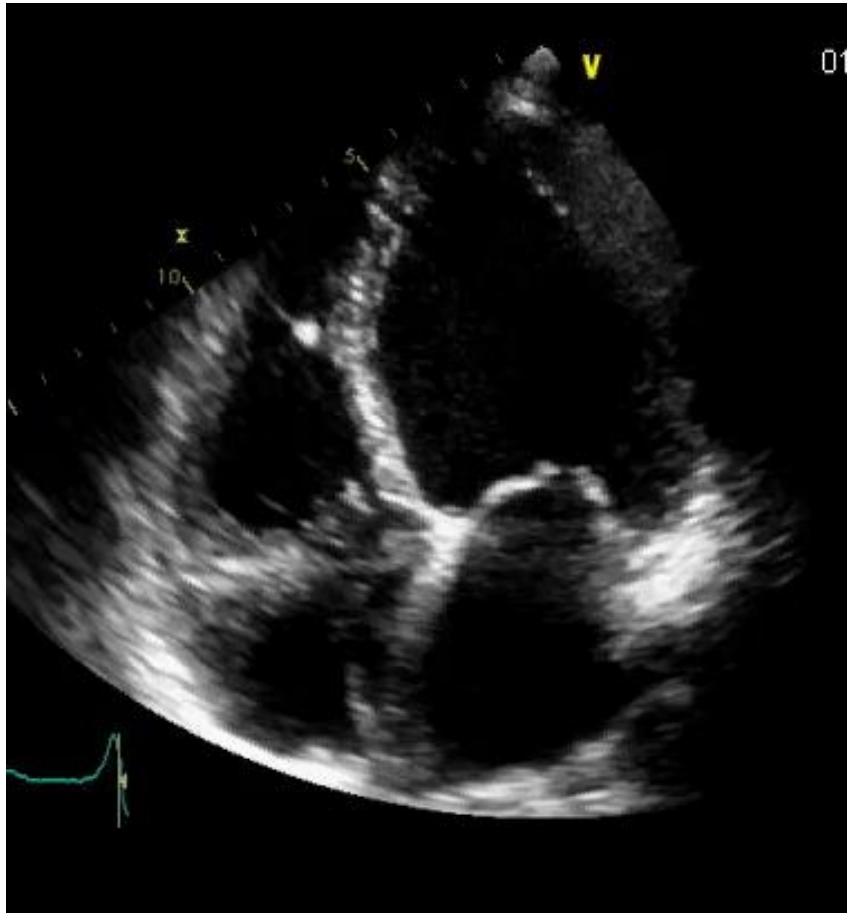
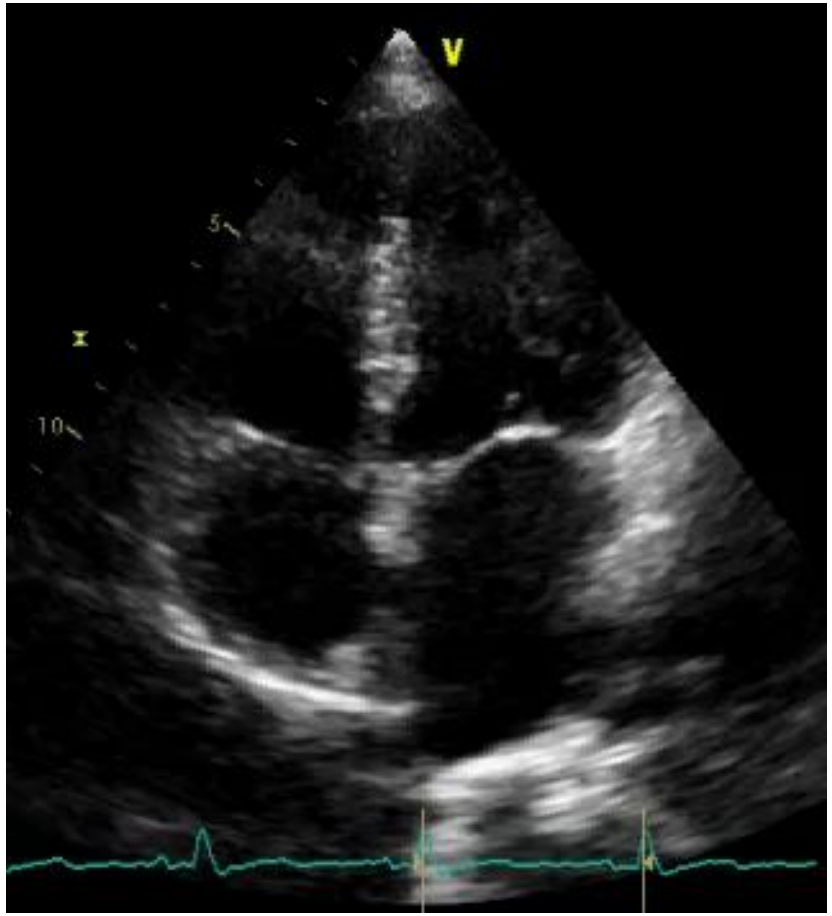
Post-antracyklinová DCM





Závěry

- HNDC může být přechodné stadium ve vývoji klasického fenotypu DCM – a potom je spojena s větší šancí na LVRR s následnou lepší prognózou.
- Fenotyp HNDC však může být spojen s řadou podtypů DCM, které mají horší prognózu než běžná DCM.
- Terapeutické přístupy u těchto podtypů mohou být odlišné zejména při identifikované specifické příčině.
- Nejasnosti ohledně definic LVAC a ADCM, genotypový i fenotypový překryv obou diagnóz.

Děkuji za pozornost!