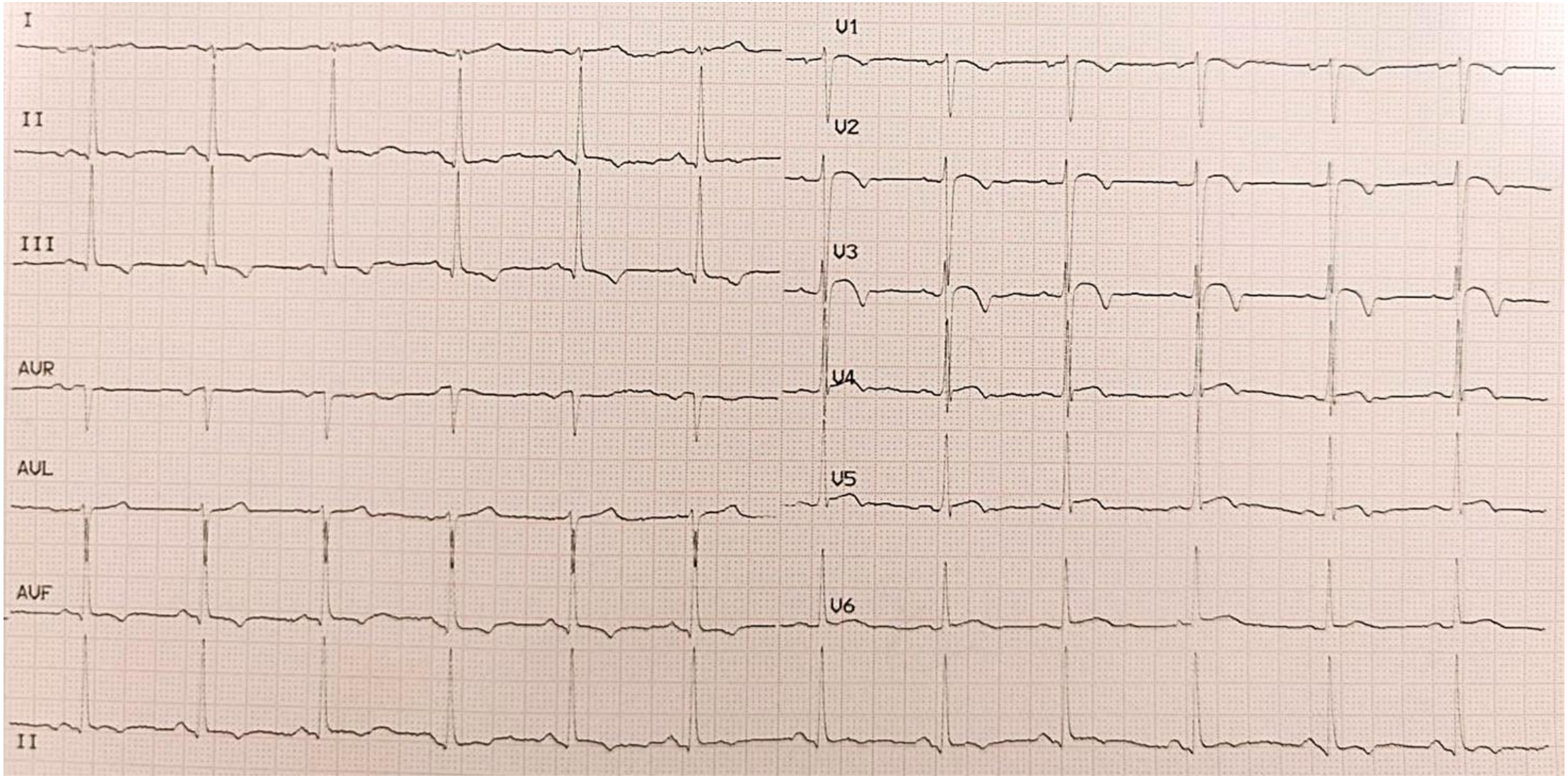
Ke správné diagnóze napříč kardiomyopatiemi

Veronika Gašpárková
Kardiologická klinika 2. LF UK a FN Motol | Praha
ČASS 10.10.2025



11/2011

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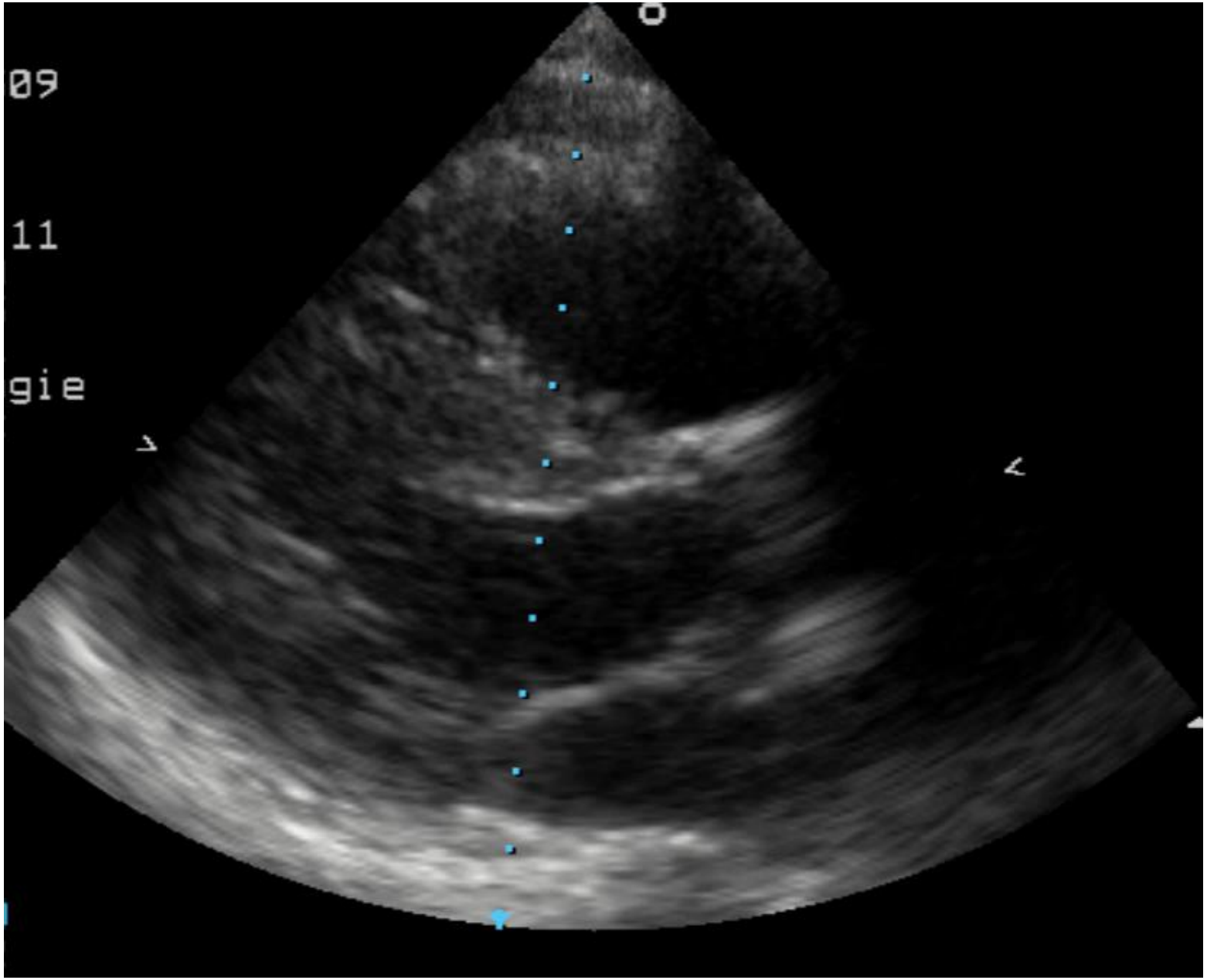

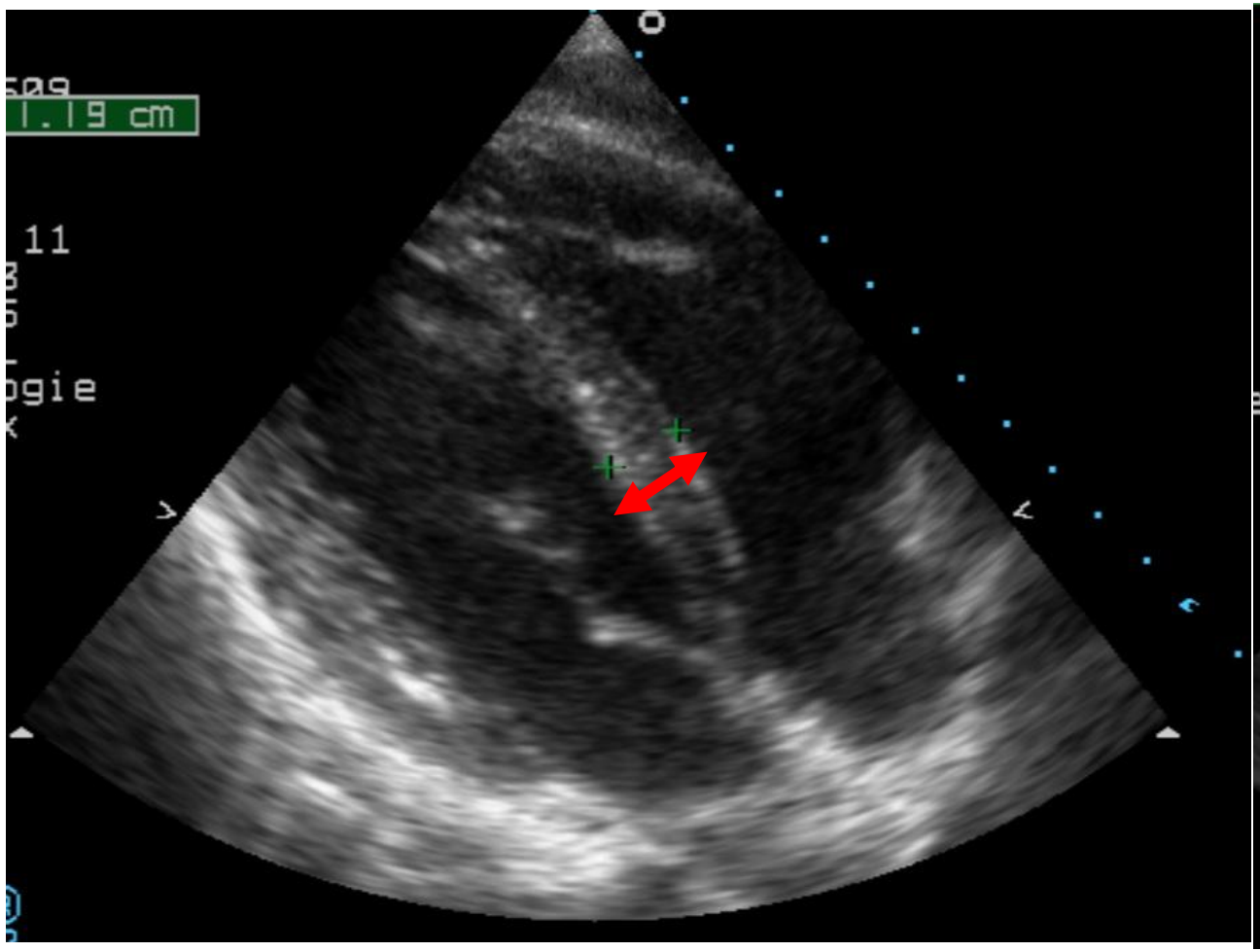

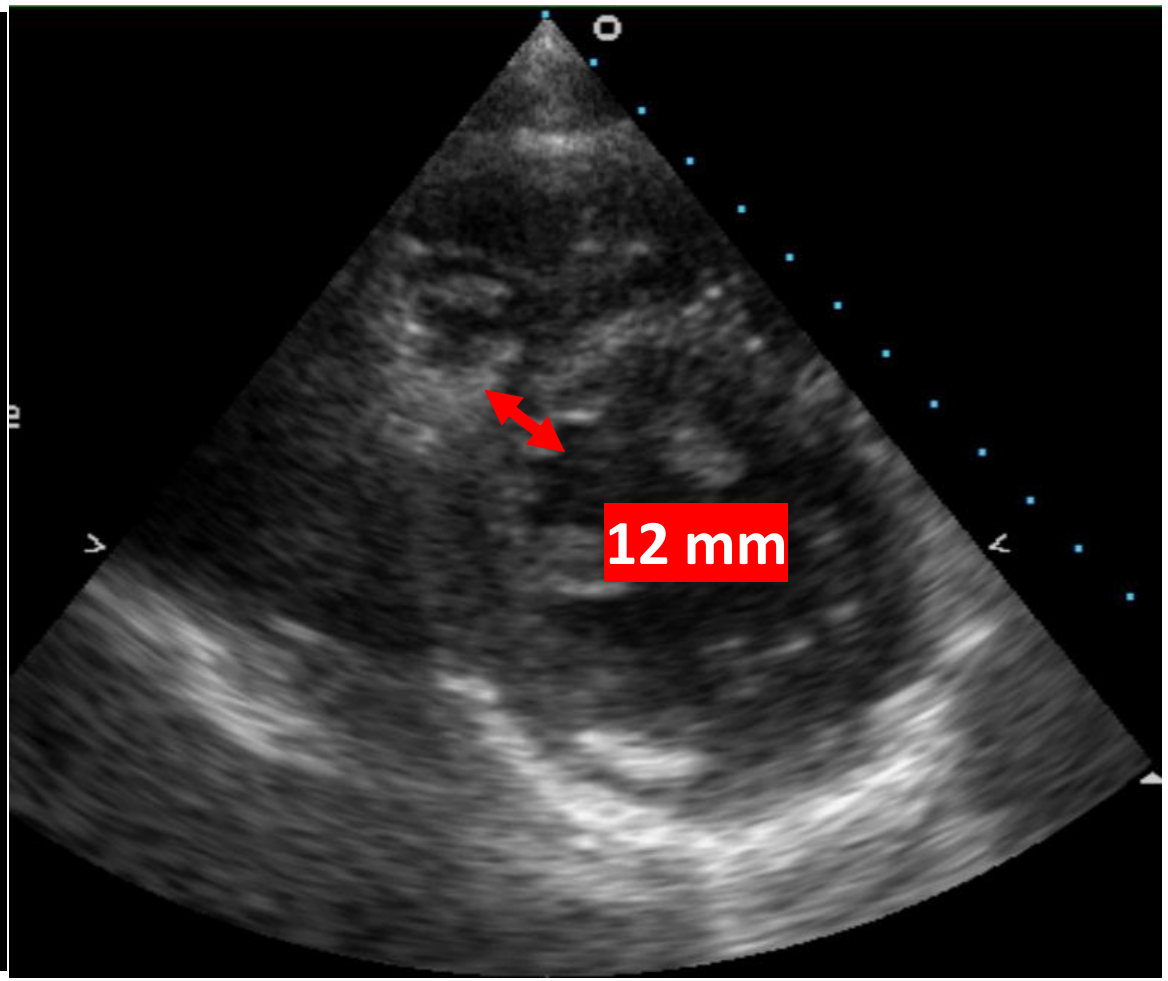

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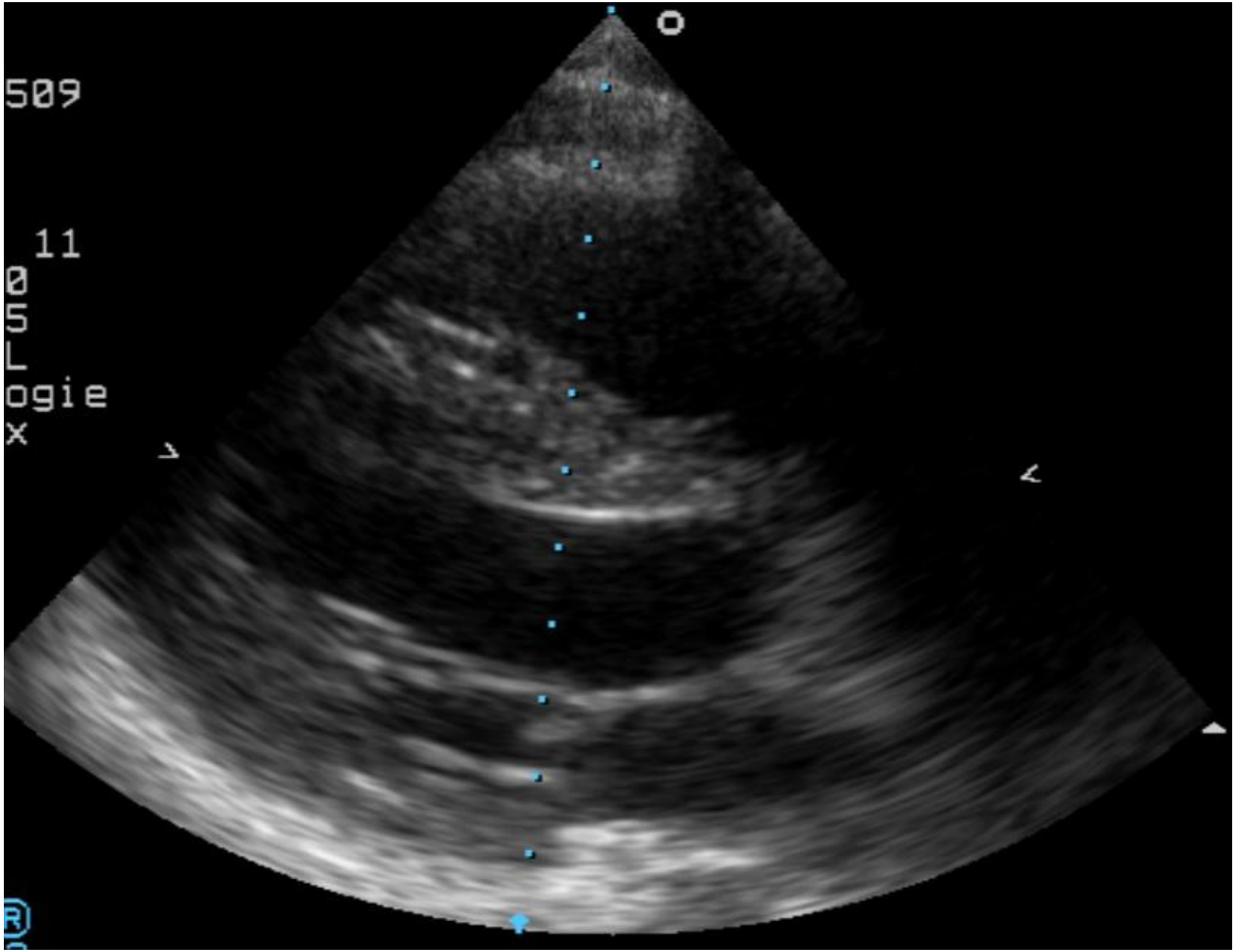
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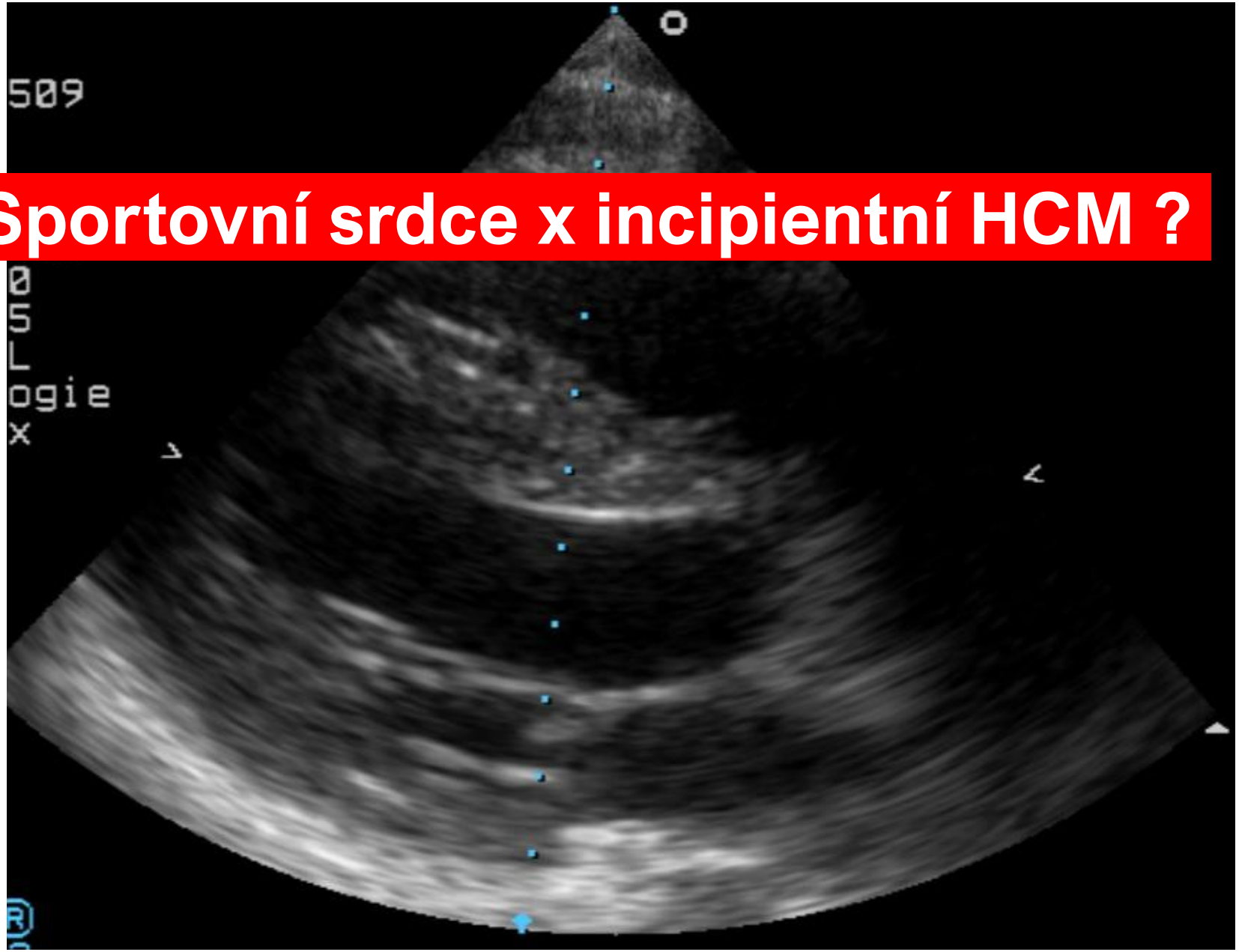
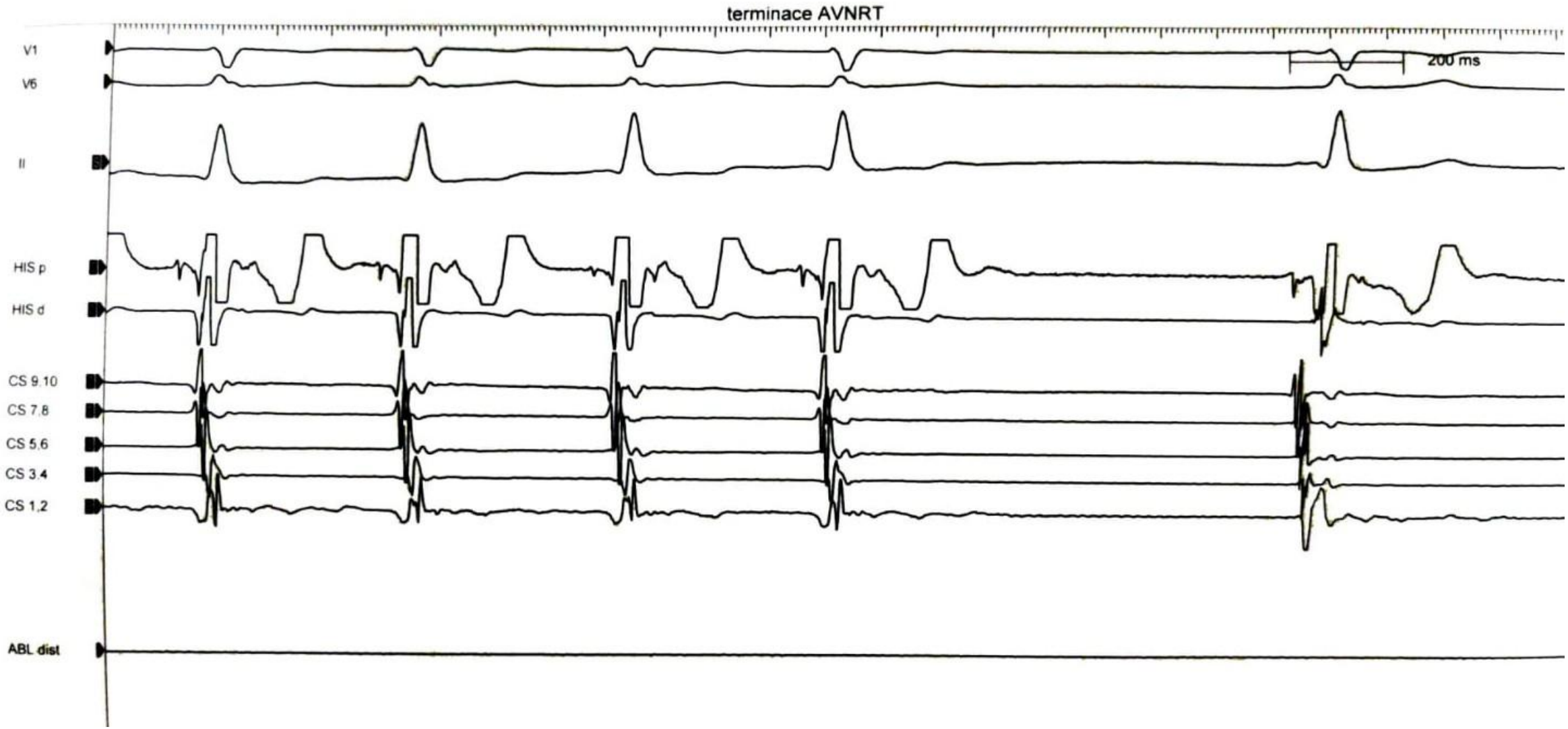
x

Sportovní srdce x incipientní HCM ?





04/2012

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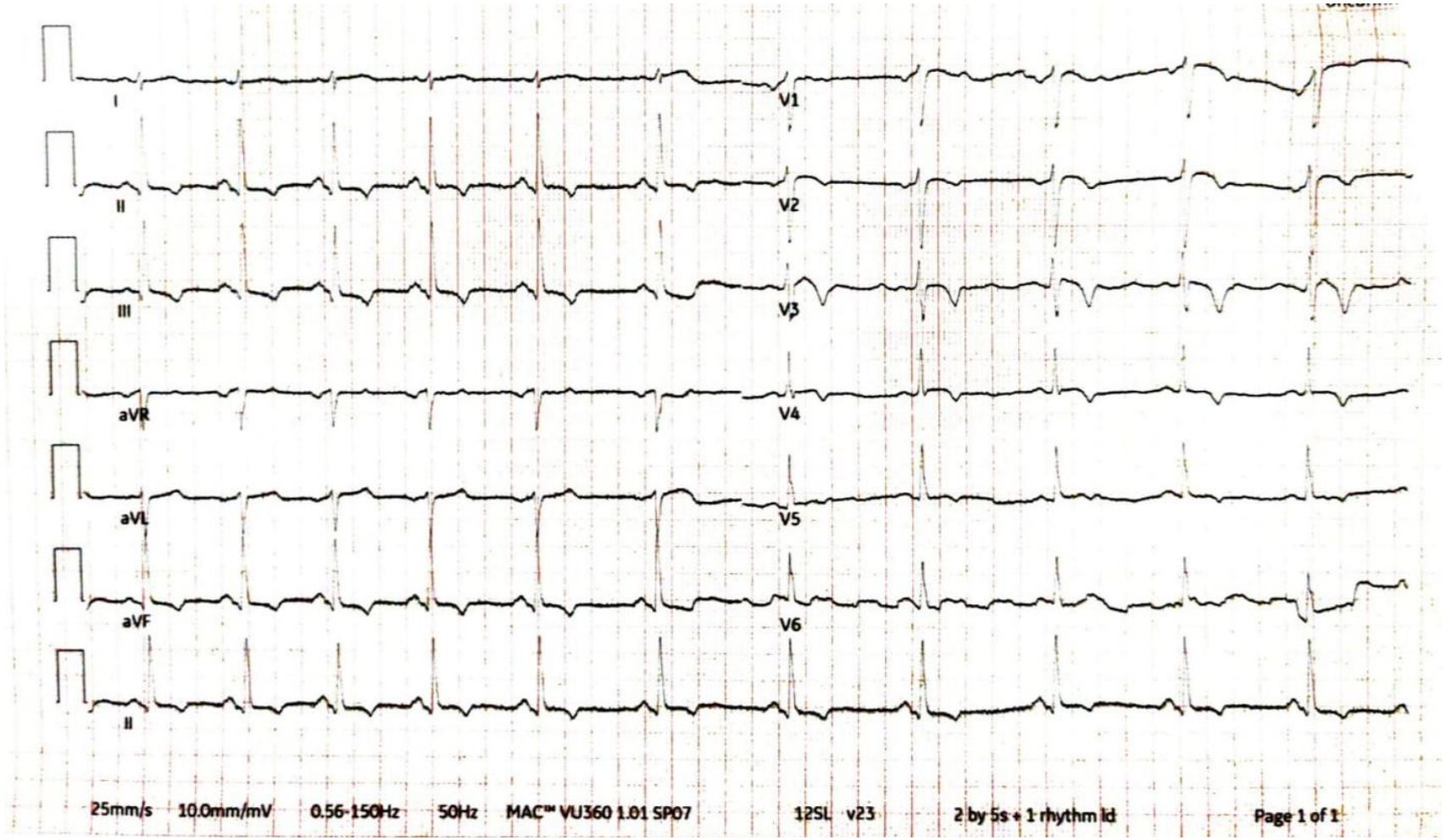
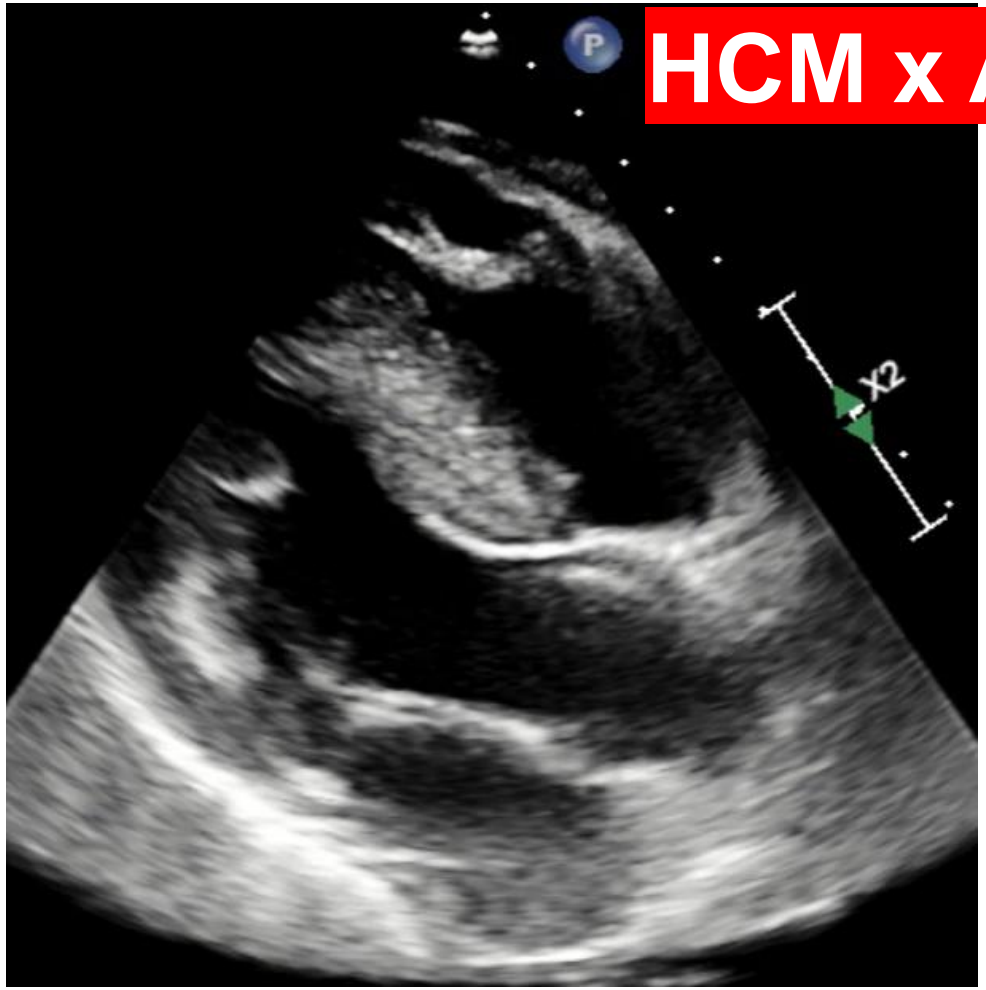
Kontrola po 5 letech…

EKG

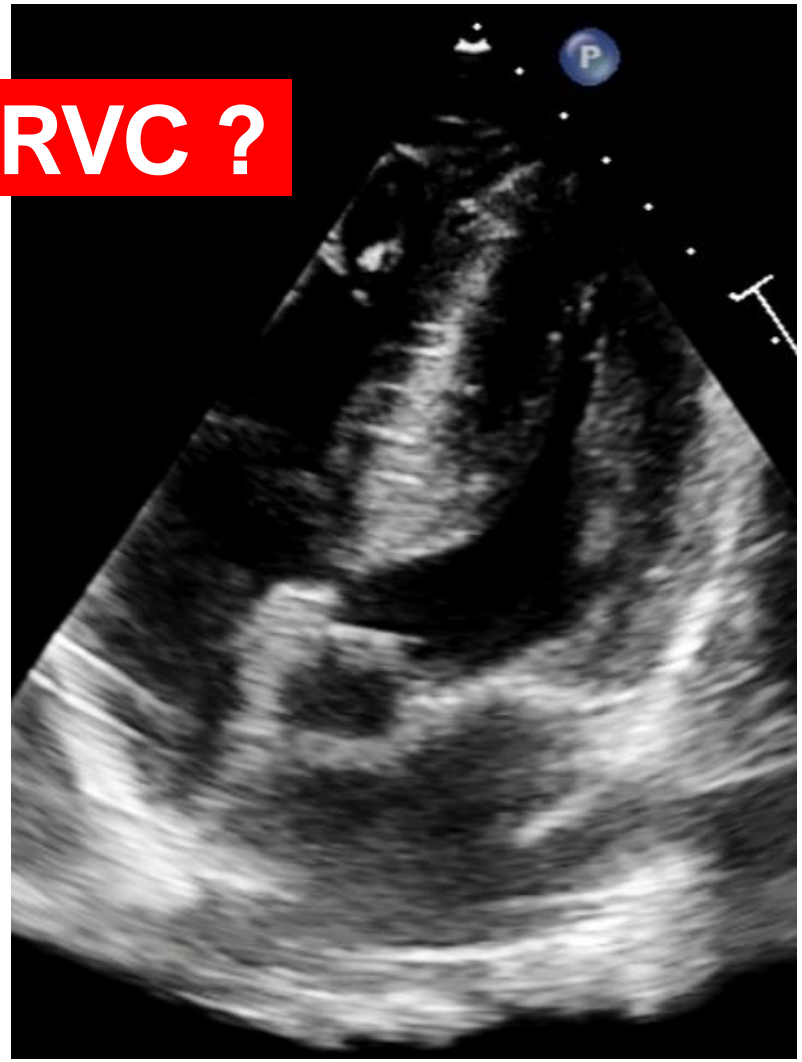

I
II
III
aVR
aVL
aVF
II
V1
V2
V3
V4
V5
V6
25mm/s 10.0mm/mV 0.56-150Hz 50Hz MAC™ VU360 1.01 SP07 12SL v23 2 by 5s + 1 rhythm ld Page 1 of 1

2020

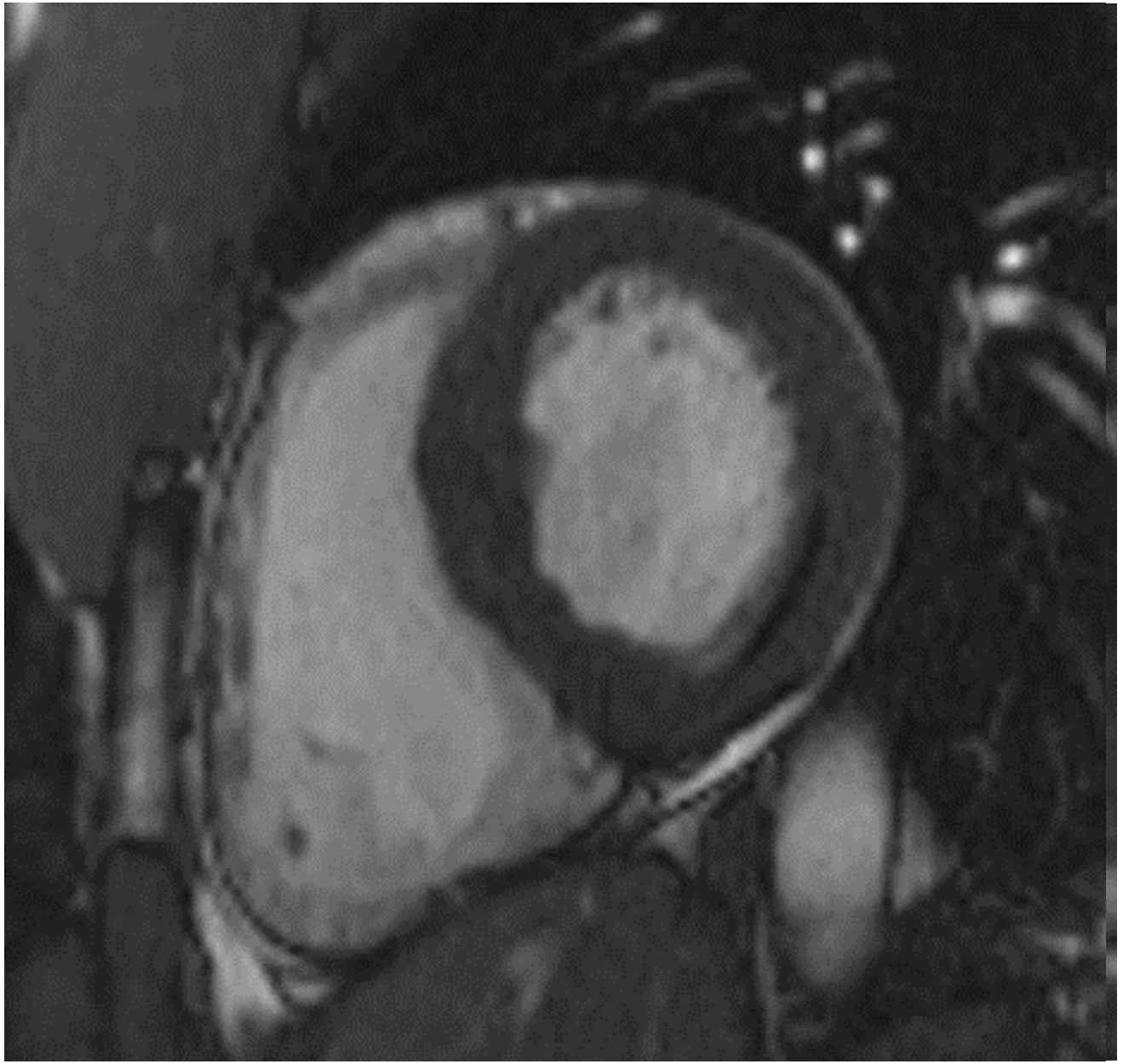
TTE 2020

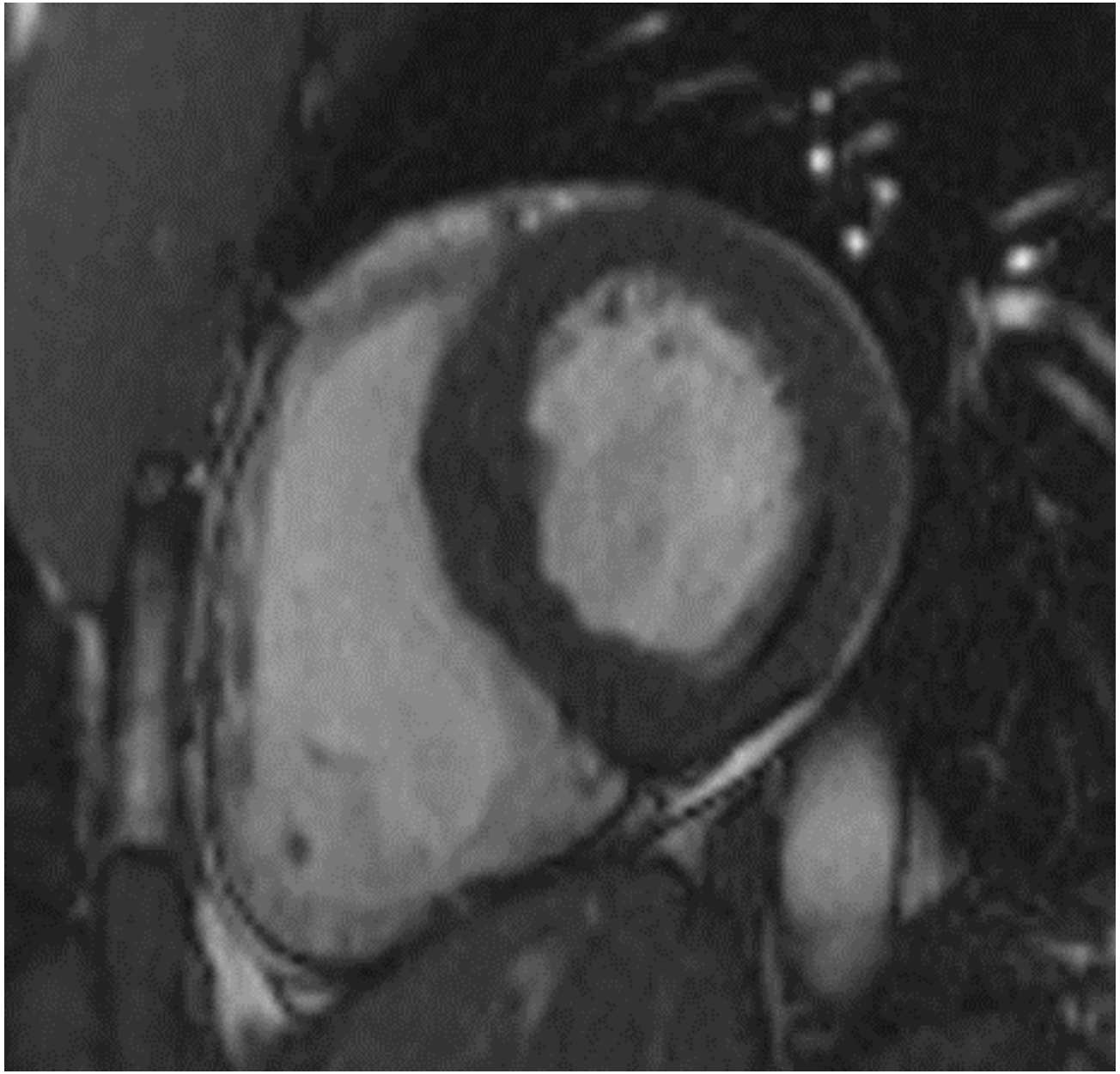


HCM x ARVC ?

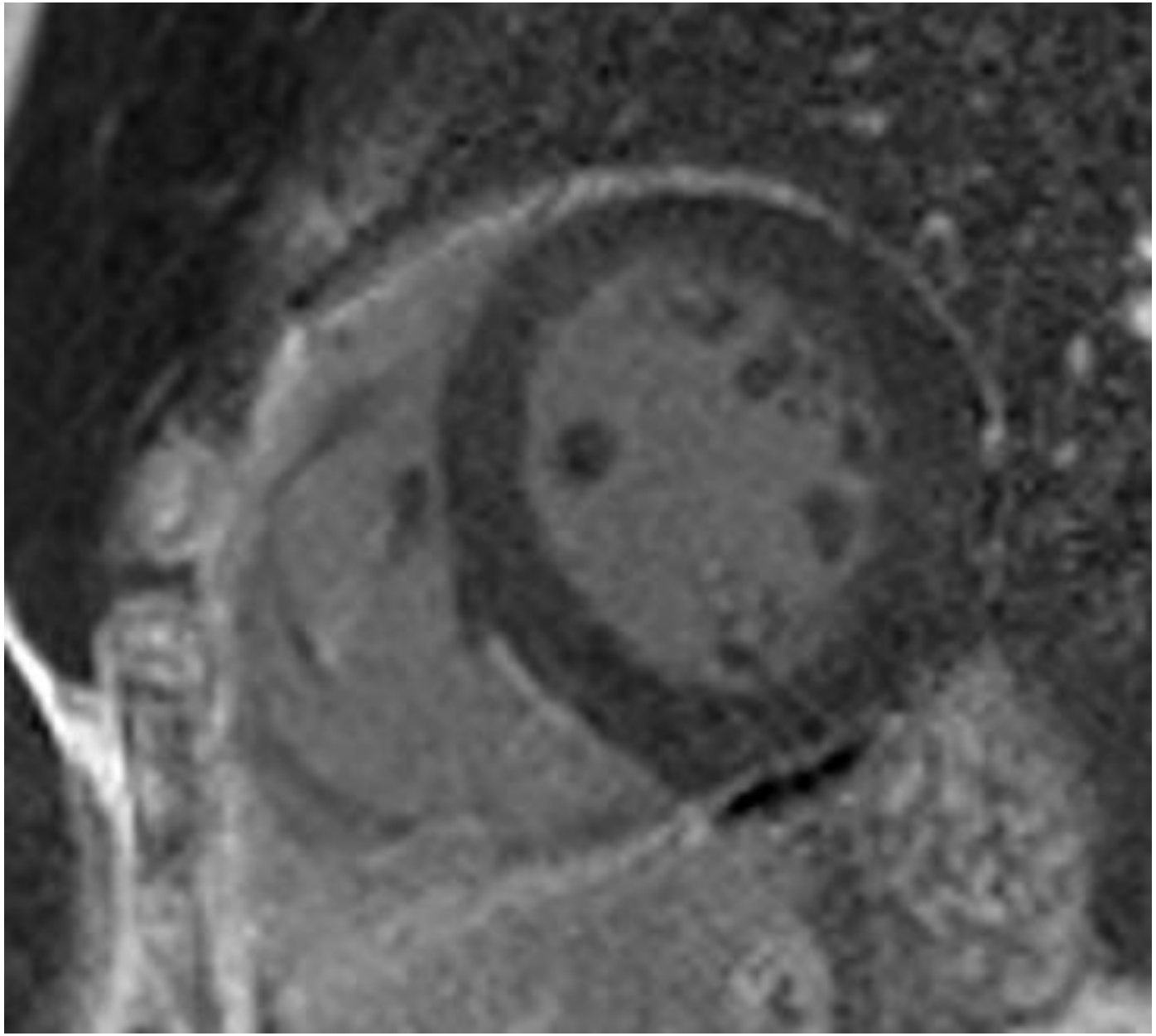


MRI





MRI (LGE)



MRI (LGE)

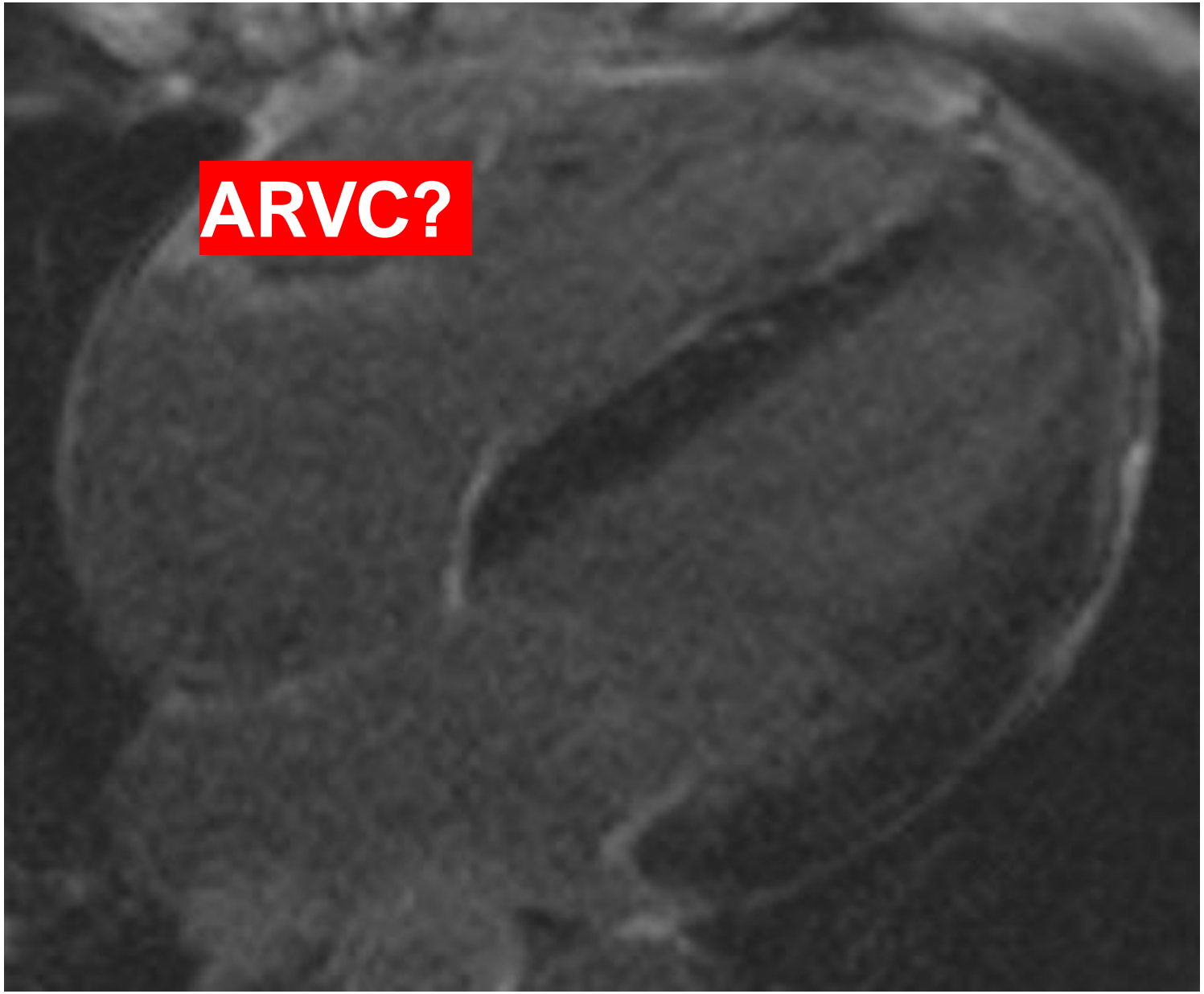
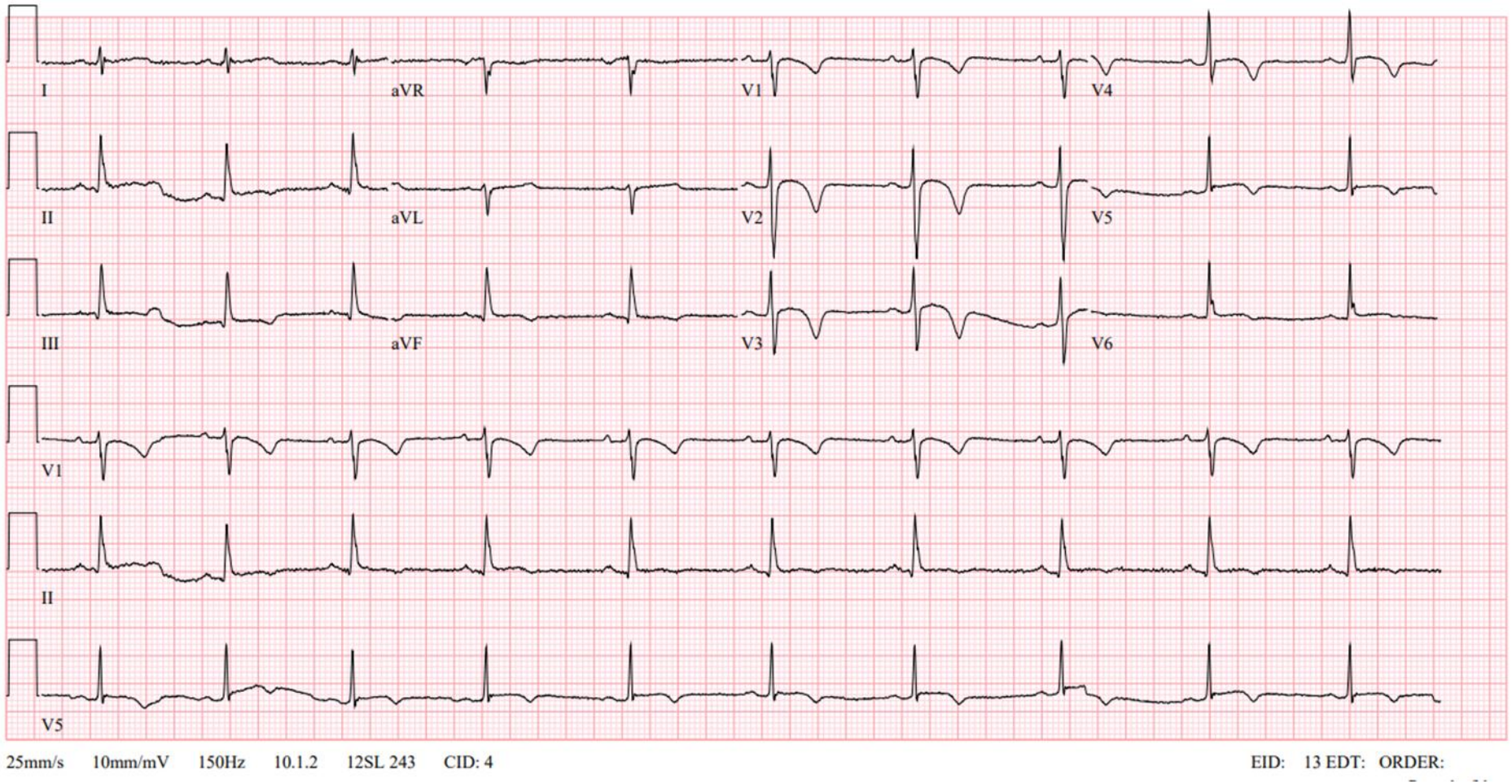


Table I Comparison of original and revised task force criteria

Original task force criteria	Revised task force criteria
I. Global or regional dysfunction and structural alterations*	
Major	
	By 2D echo:
• Severe dilatation and reduction of RV ejection fraction with no (or only mild) LV impairment • Localized RV aneurysms (akinetic or dyskinetic areas with diastolic bulging) • Severe segmental dilatation of the RV	• Regional RV akinesia, dyskinesia, or aneurysm • *and* 1 of the following (end diastole): — PLAX RVOT ≥32 mm (corrected for body size [PLAX/BSA] ≥19 mm/m^2) — PSAX RVOT ≥36 mm (corrected for body size [PSAX/BSA] ≥21 mm/m^2) — *or* fractional area change ≤33%
	By MRI:
	• Regional RV akinesia or dyskinesia or dyssynchronous RV contraction • *and* 1 of the following: — Ratio of RV end-diastolic volume to BSA ≥110 mL/m^2 (male) or ≥100 mL/m^2 (female) — *or* RV ejection fraction ≤40%
	By RV angiography:
	• Regional RV akinesia, dyskinesia, or aneurysm
Minor	
	By 2D echo:
• Mild global RV dilatation and/or ejection fraction reduction with normal LV • Mild segmental dilatation of the RV • Regional RV hypokinesia	• Regional RV akinesia or dyskinesia • *and* 1 of the following (end diastole): — PLAX RVOT ≥29 to <32 mm (corrected for body size [PLAX/BSA] ≥16 to <19 mm/m^2) — PSAX RVOT ≥32 to <36 mm (corrected for body size [PSAX/BSA] ≥18 to <21 mm/m^2) — *or* fractional area change >33% to ≤40%
	By MRI:
	• Regional RV akinesia or dyskinesia or dyssynchronous RV contraction • *and* 1 of the following: — Ratio of RV end-diastolic volume to BSA ≥100 to <110 mL/m^2 (male) or ≥90 to <100 mL/m^2 (female) — *or* RV ejection fraction >40% to ≤45%

Marcus et al., *Eur Heart J*, 2010

MRI



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Marcus et al., *Eur Heart J*, 2010

Table I *Continued*

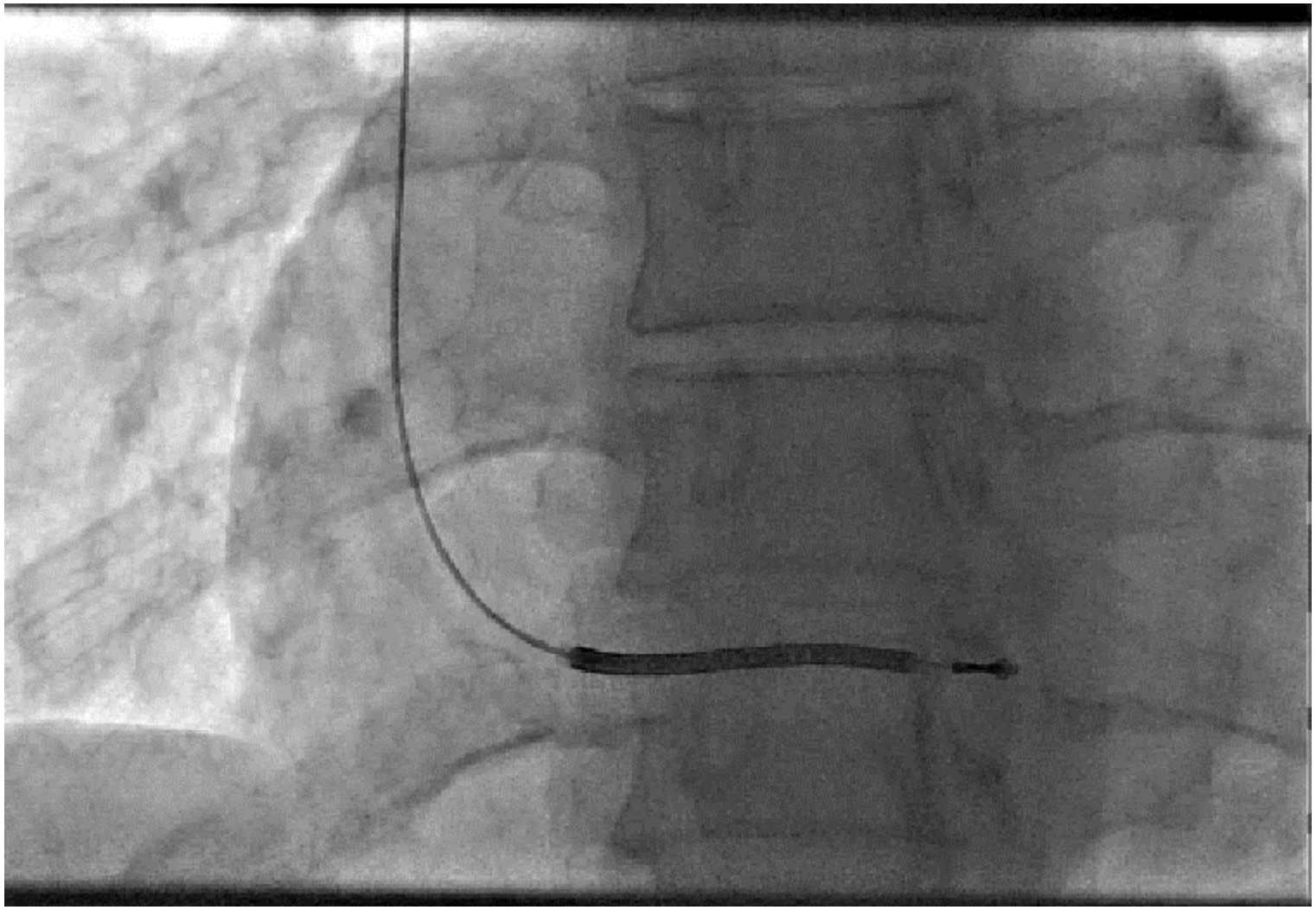
Original task force criteria	Revised task force criteria
IV. Depolarization/conduction abnormalities	
Major	
• Epsilon waves or localized prolongation (>110 ms) of the QRS complex in right precordial leads (V_1 to V_3)	• Epsilon wave (reproducible low-amplitude signals between end of QRS complex to onset of the T wave) in the right precordial leads (V_1 to V_3)
Minor	
• Late potentials (SAECG)	• Late potentials by SAECG in ≥1 of 3 parameters in the absence of a QRS duration of ≥110 ms on the standard ECG • Filtered QRS duration (fQRS) ≥114 ms • Duration of terminal QRS <40 μV (low-amplitude signal duration) ≥38 ms • Root-mean-square voltage of terminal 40 ms ≤20 μV

Molekulárně genetické vyšetření (NGS):
DSC2 c.1263+2T>G - **patogenní**
DCS2 c.595C>T p.(Arg199Cys) - VUS

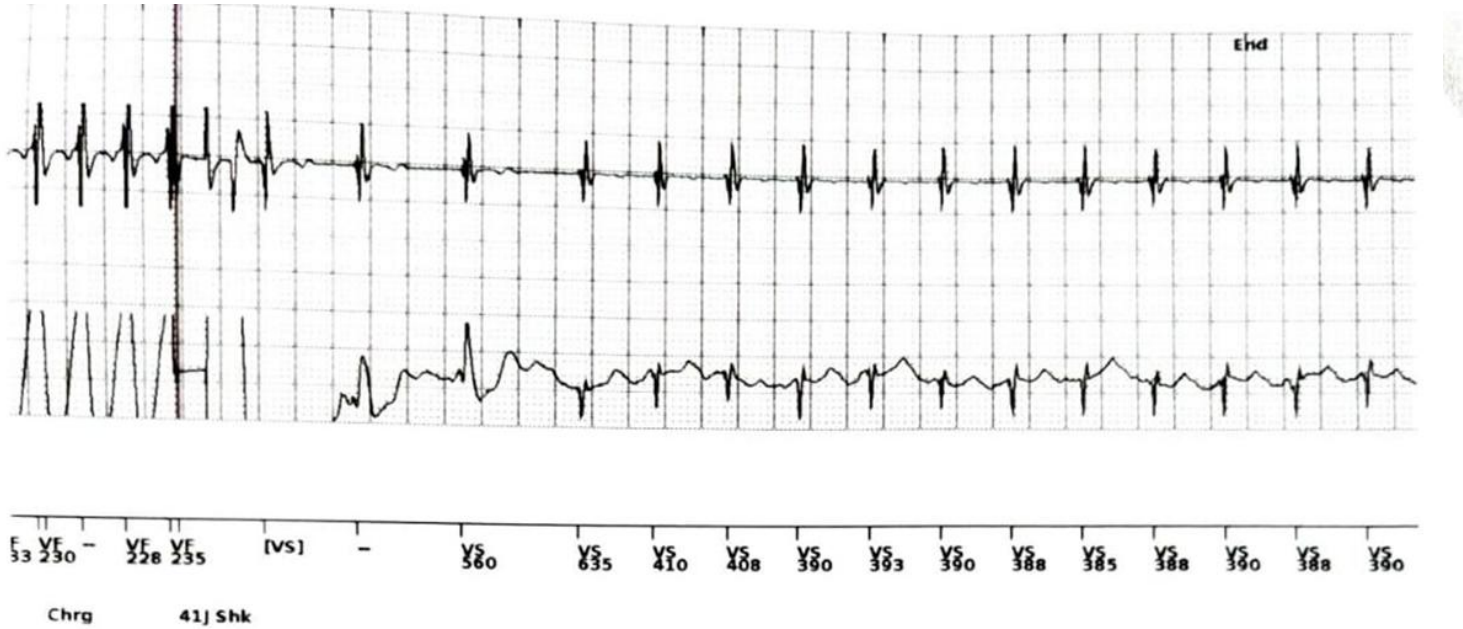
	• Identification of a pathogenic mutation* categorized as associated or probably associated with ARVC/D in the patient under evaluation
Minor	
• Family history of premature sudden death (<35 years of age) due to suspected ARVC/D	• History of ARVC/D in a first-degree relative in whom it is not possible or practical to determine whether the family member meets current Task Force criteria
• Familial history (clinical diagnosis based on present criteria)	• Premature sudden death (<35 years of age) due to suspected ARVC/D in a first-degree relative
	• ARVC/D confirmed pathologically or by current Task Force Criteria in second-degree relative

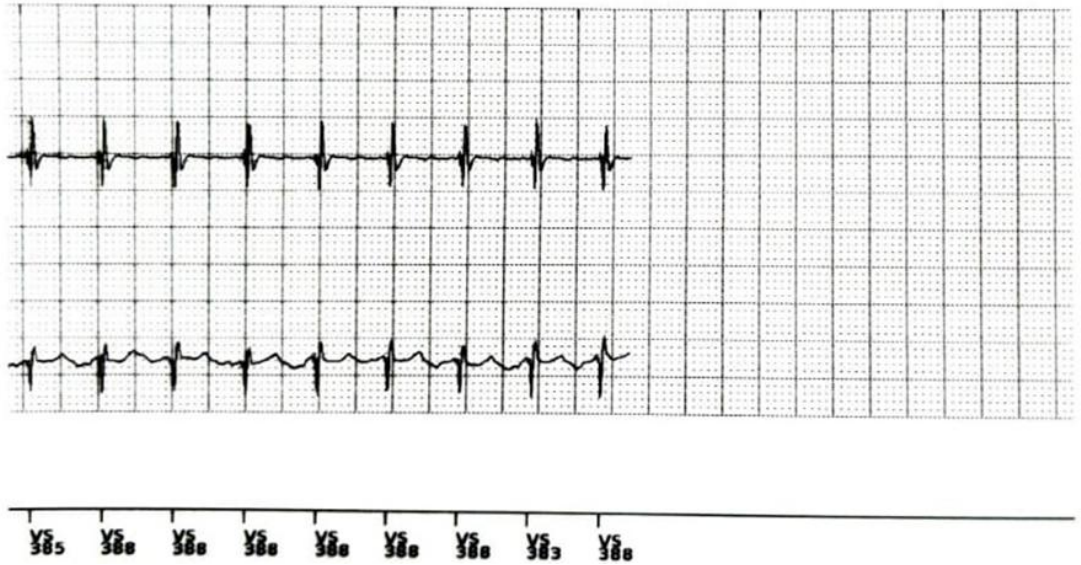
Marcus et al., *Eur Heart J*, 2010

2020



Kontrola po 5 letech…





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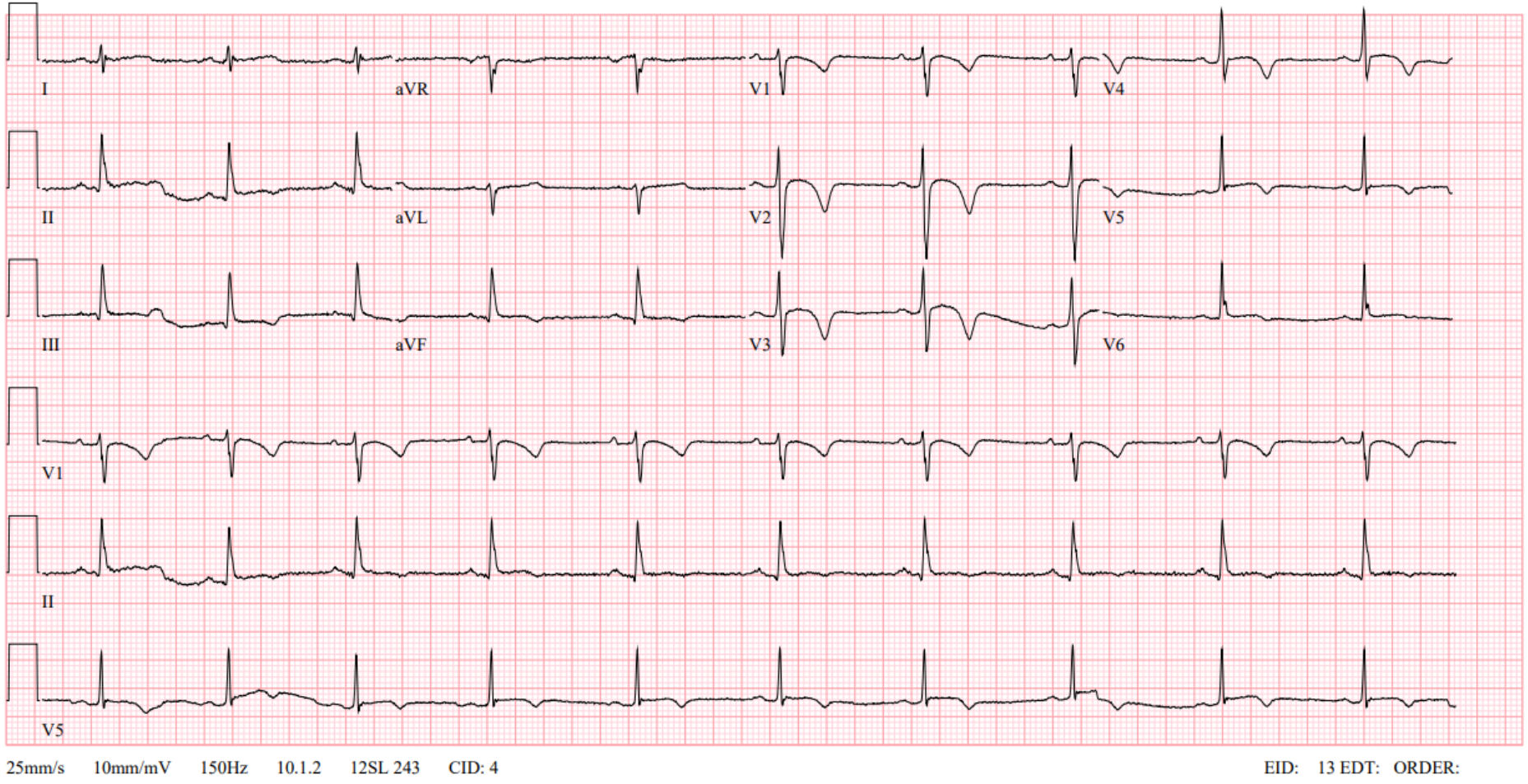
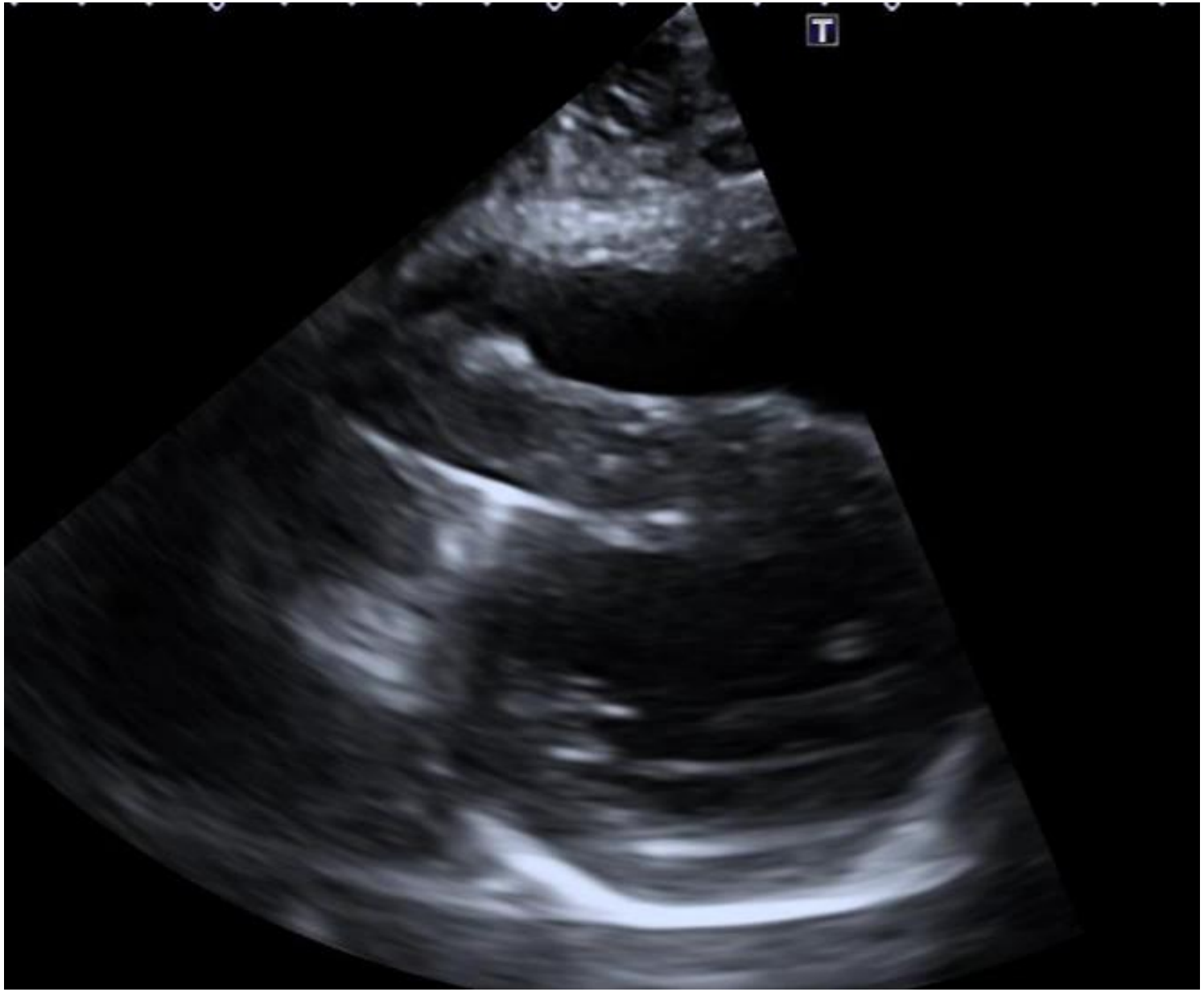
EKG


I
aVR
V1
V4
II
aVL
V2
V5
III
aVF
V3
V6
V1
II
V5
25mm/s 10mm/mV 150Hz 10.1.2 12SL 243 CID: 4
EID: 13 EDT: ORDER:

09/2025

TTE 2025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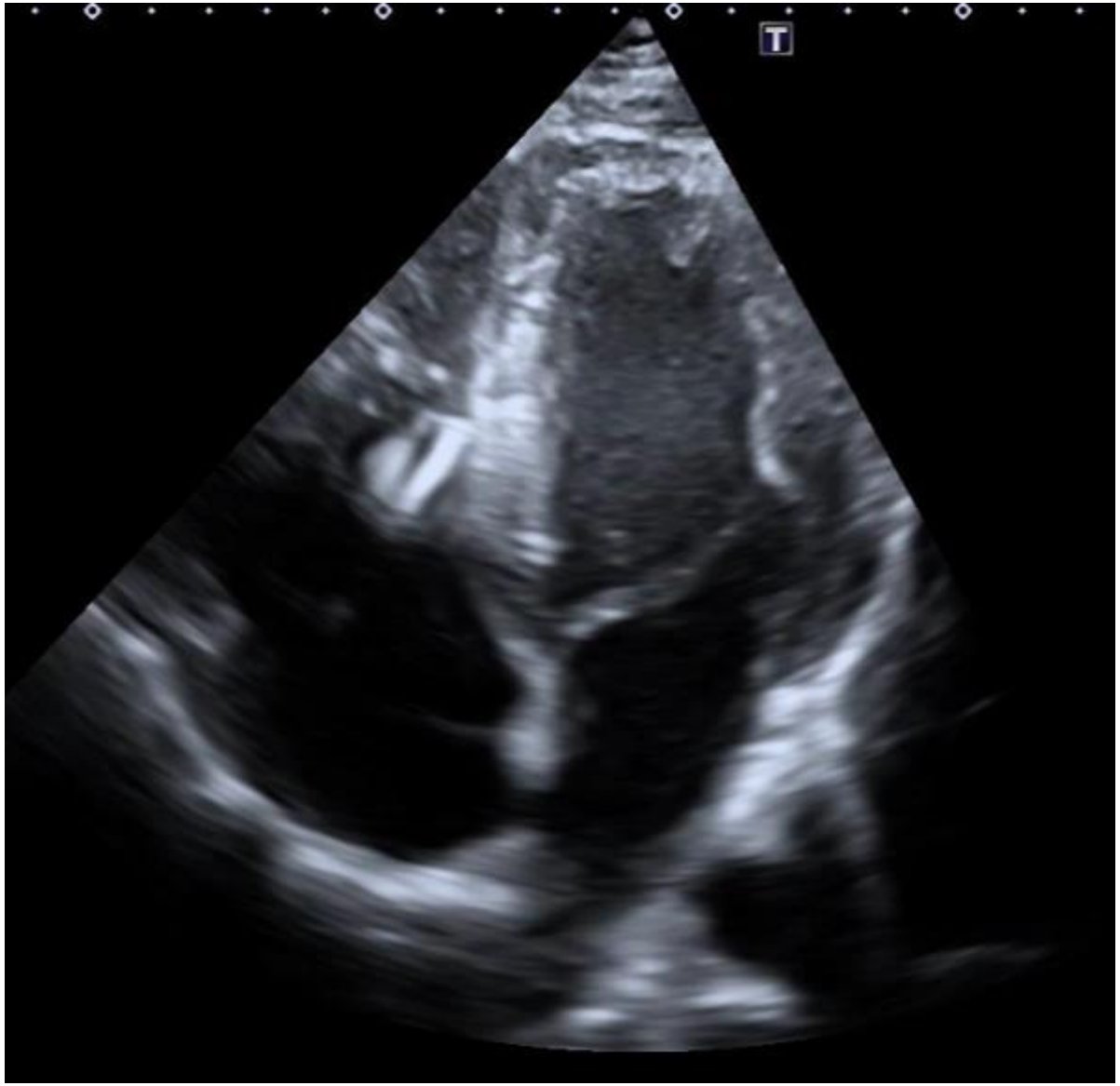

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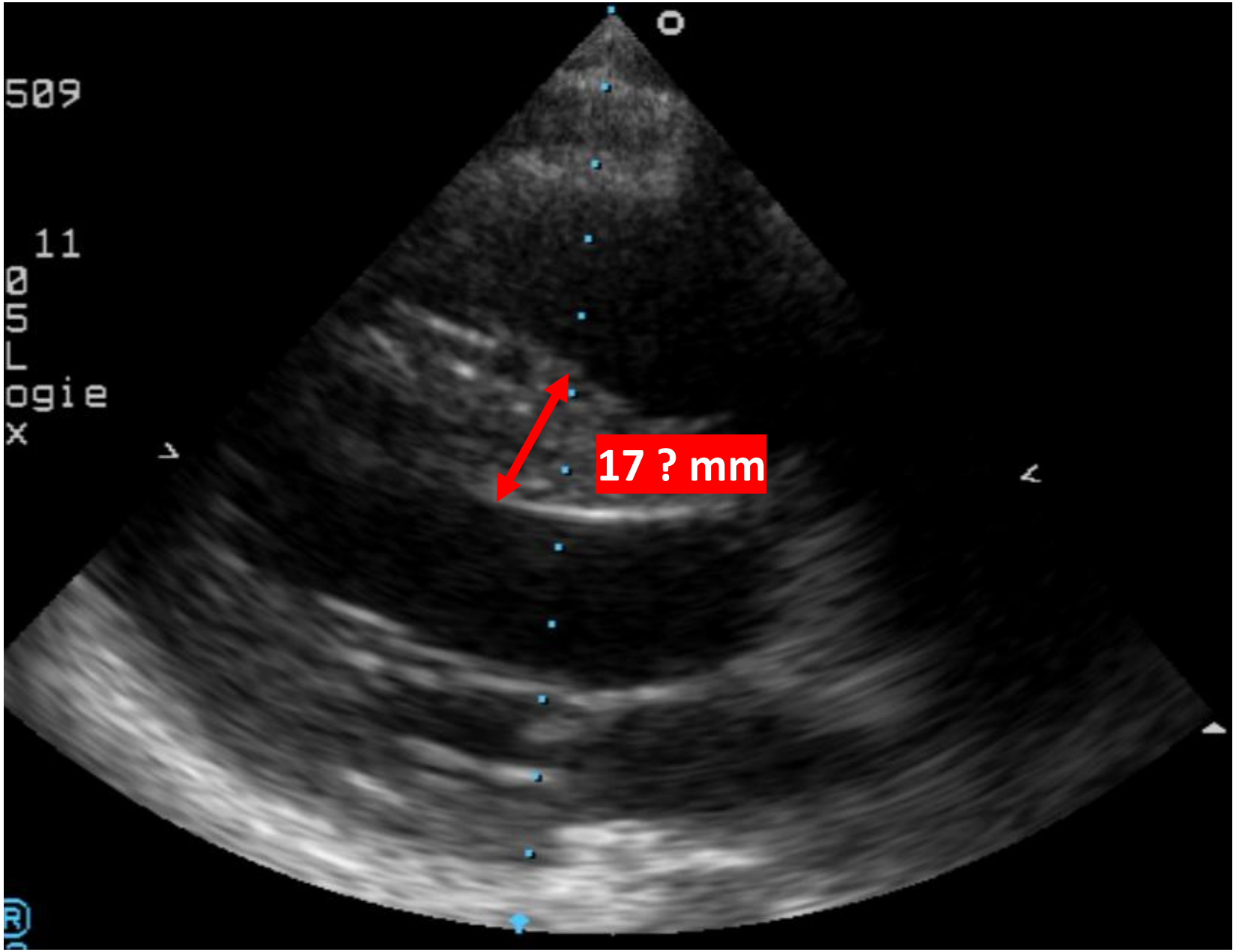
Mohla být dg. časněji?



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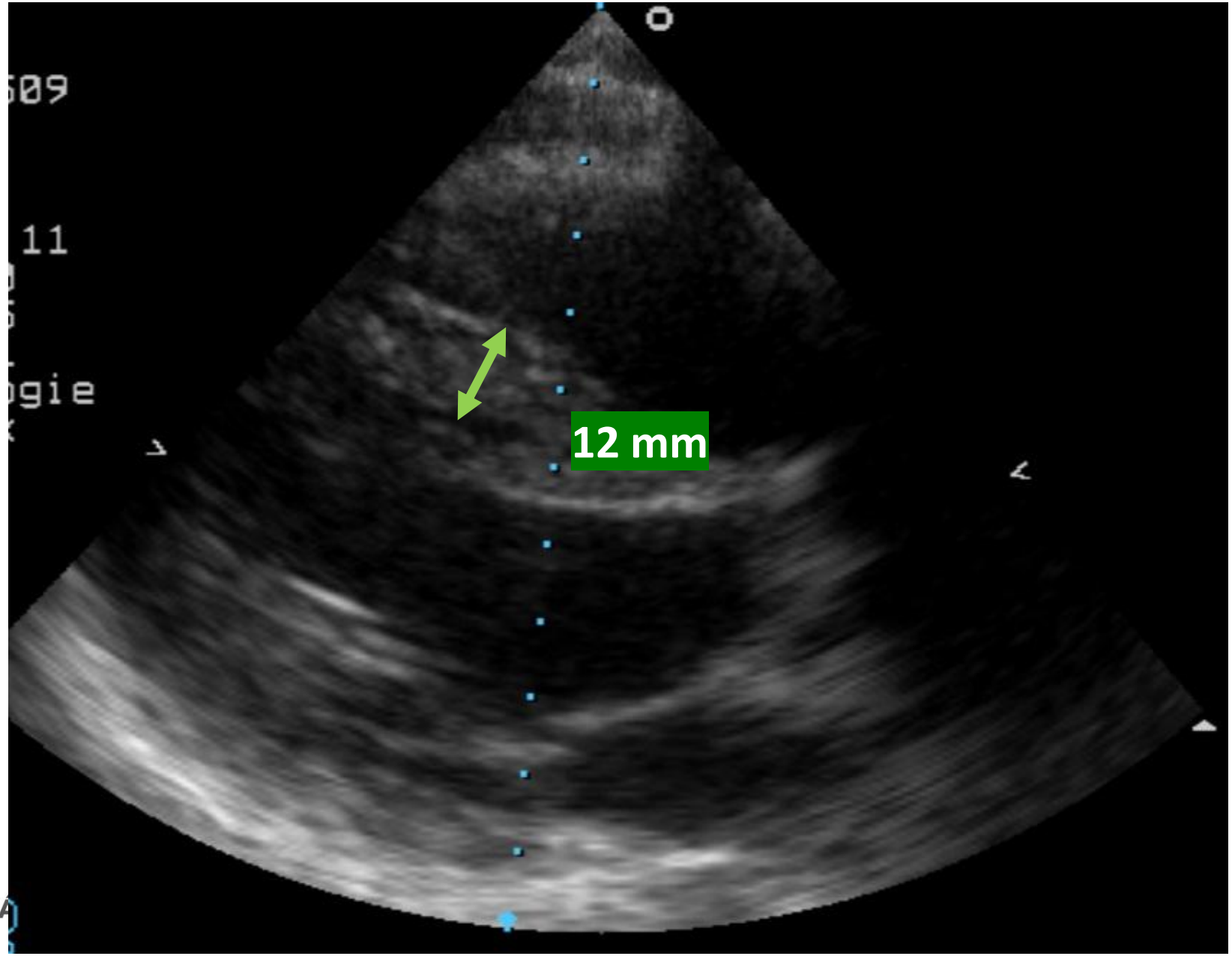

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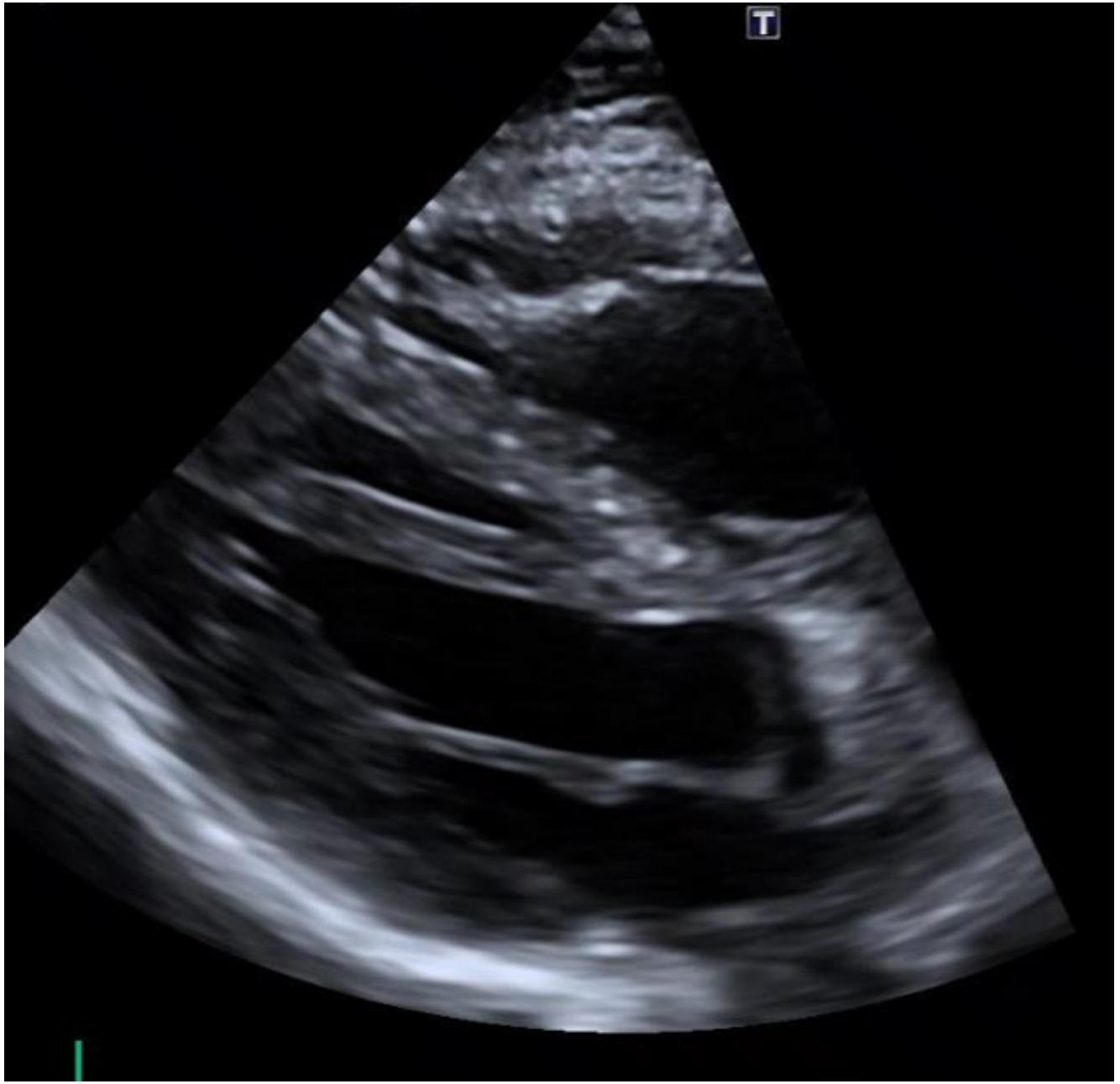
T

Take home message

- Multimodální přístup
- Pečlivá OA, RA a screening příbuzných
  - EKG, holter EKG
  - TTE a/nebo MRI
  - genetické vyšetření
- Dlouhodobý pravidelné sledování
- Restrikce intenzivní sportovní aktivity
  - riziko progrese fenotypu
- Prevence arytmické SCD implantací ICD
  - sekundární prevence
  - primární prevence

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

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Děkuji za pozornost!

Veronika Gašpárková

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