Αρρυθμιογόνος Δυσπλασία της Δεξιάς Κοιλίας: Διαγνωστικά και Θεραπευτικά Διλλήματα

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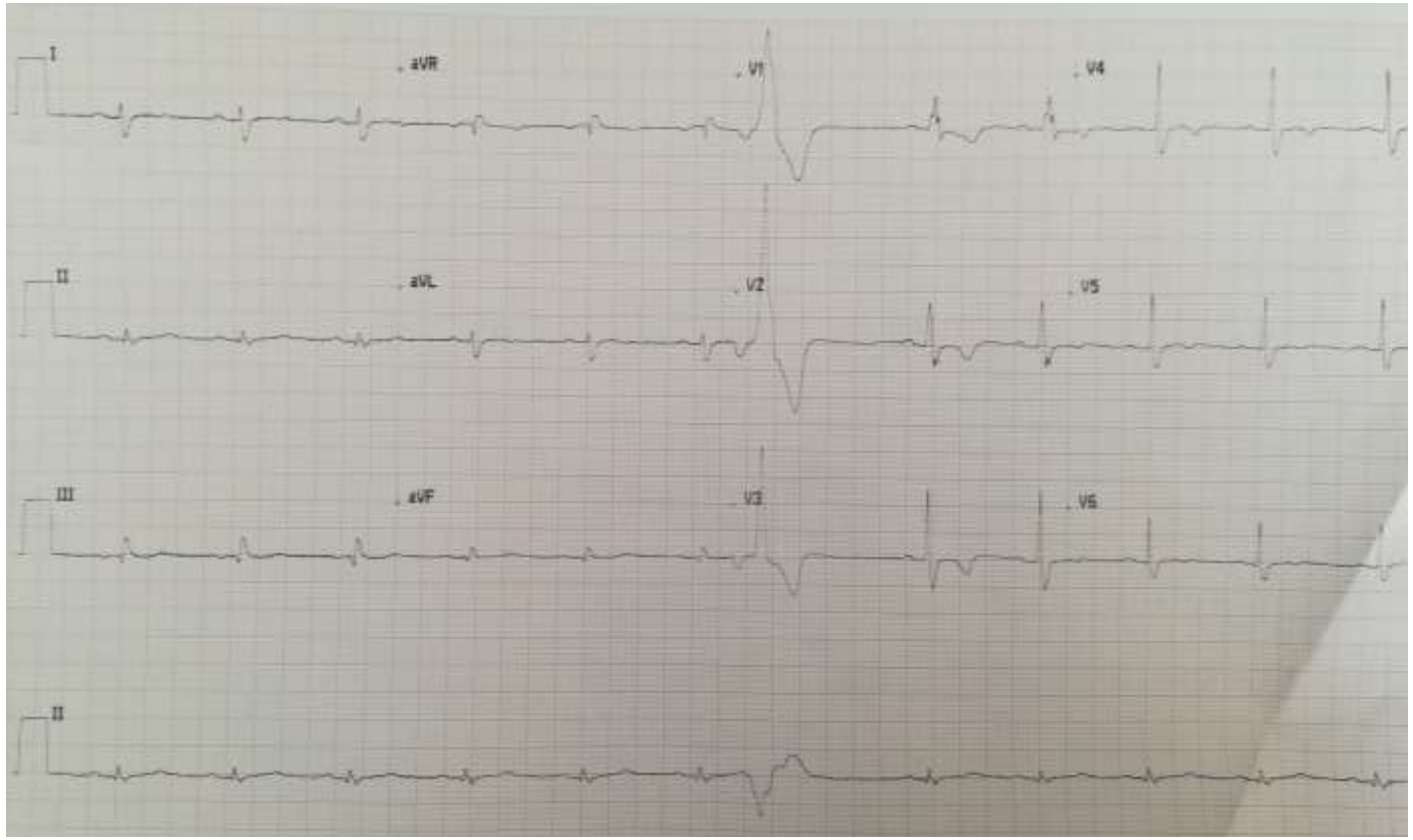
Κλινική Άγιος Λουκάς

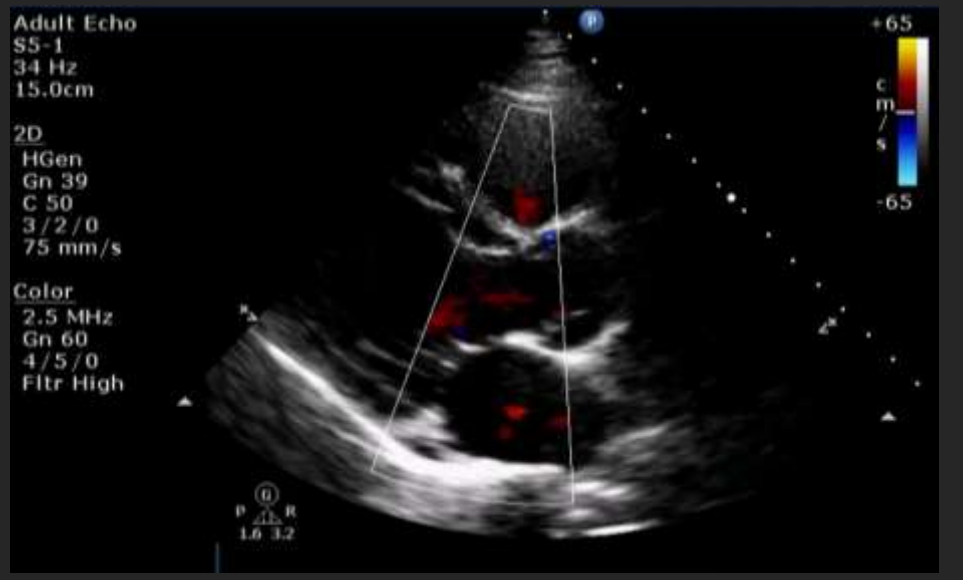
Ημερίδα Κλινικής Καρδιολογίας, Κατερίνη 8 Φεβρουαρίου 2020

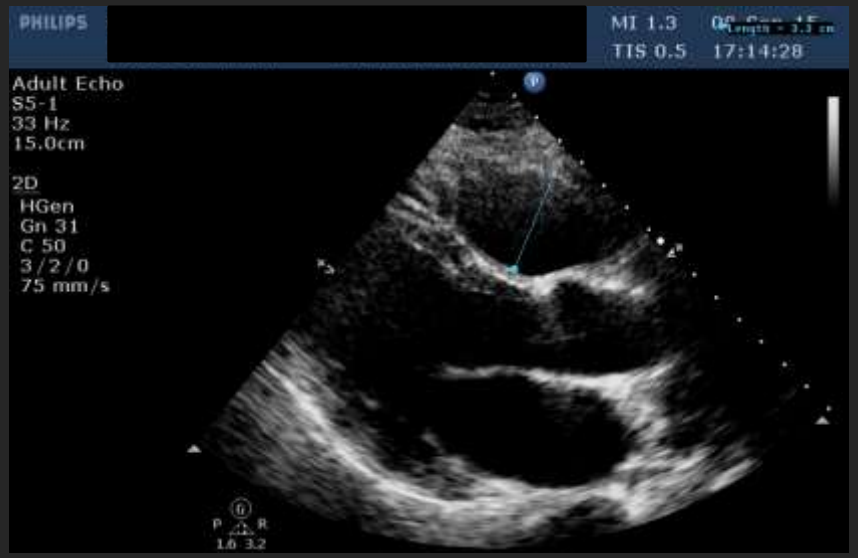
Case

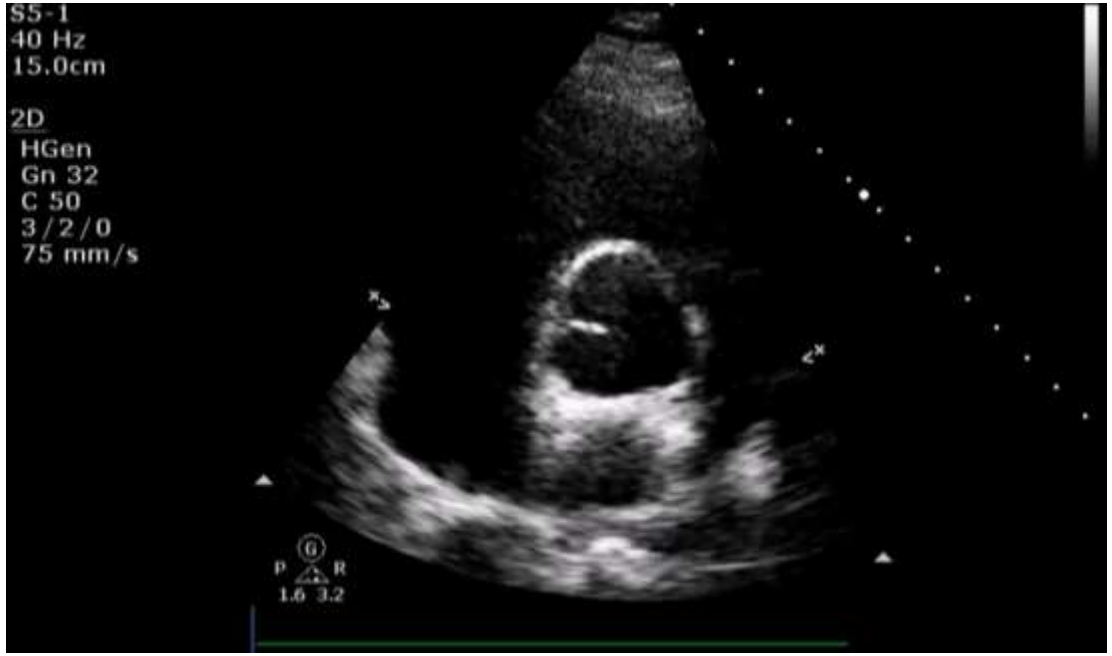
- 38 years old male
- Chest pain
- Heavy smoker
- Family history: coronary artery disease (father)
- Normal lab findings
- ECG: T wave inversions (V1-V6)

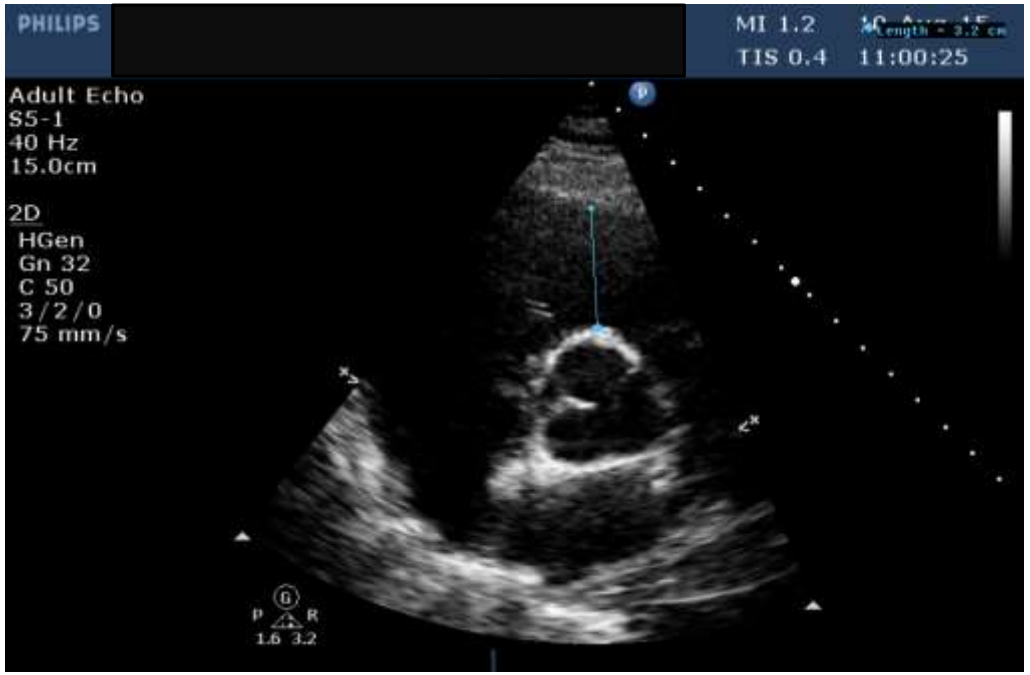


Echocardiography

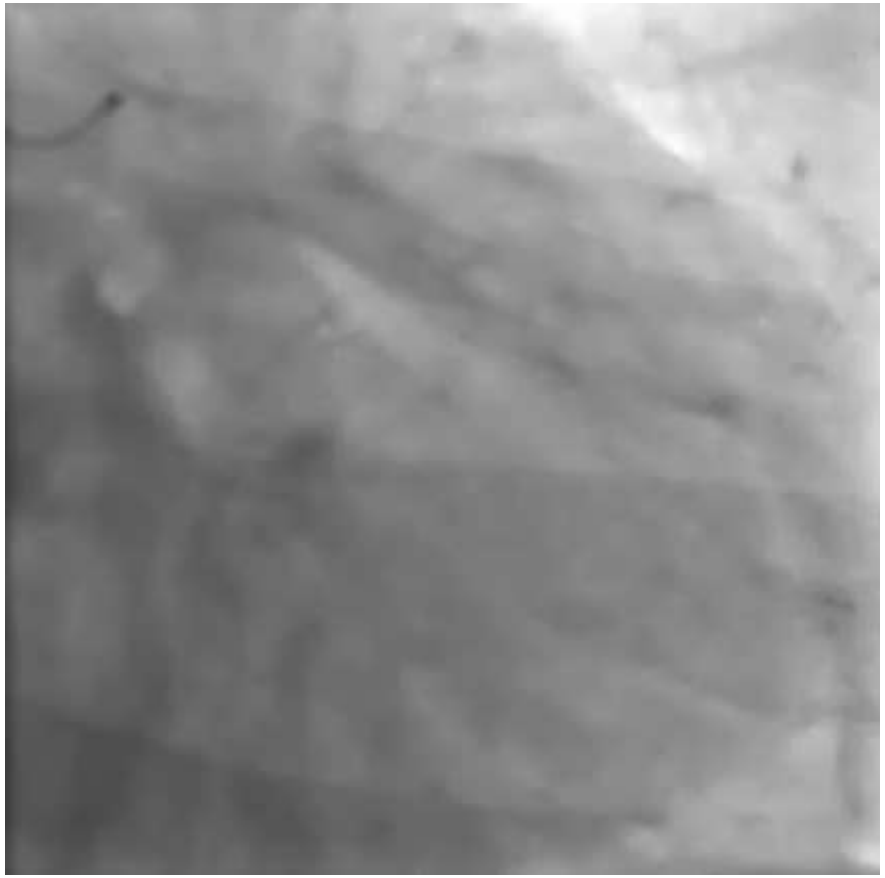


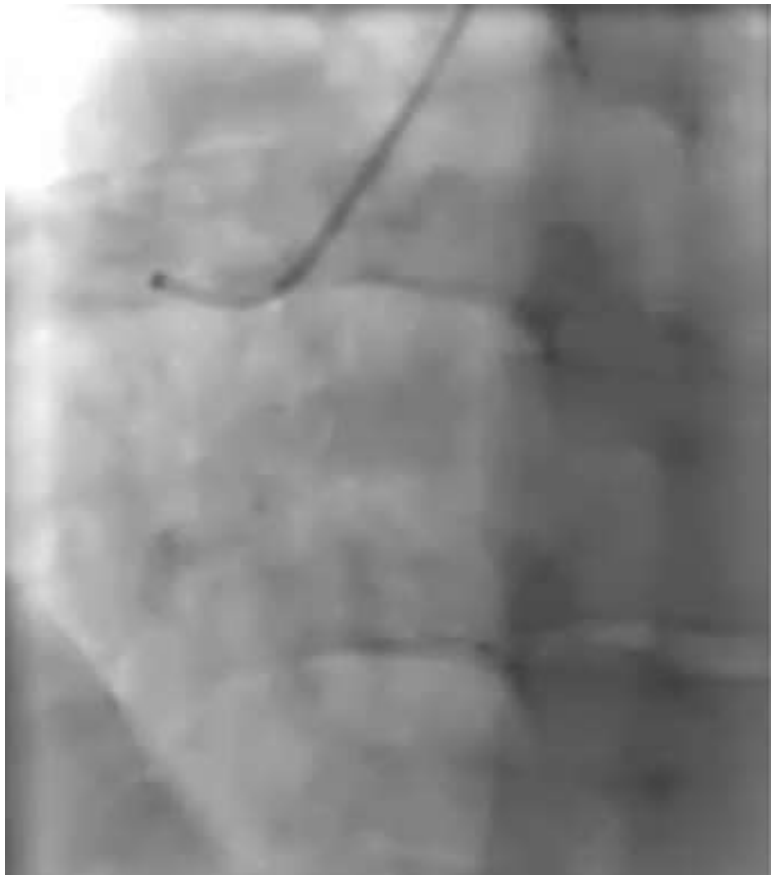






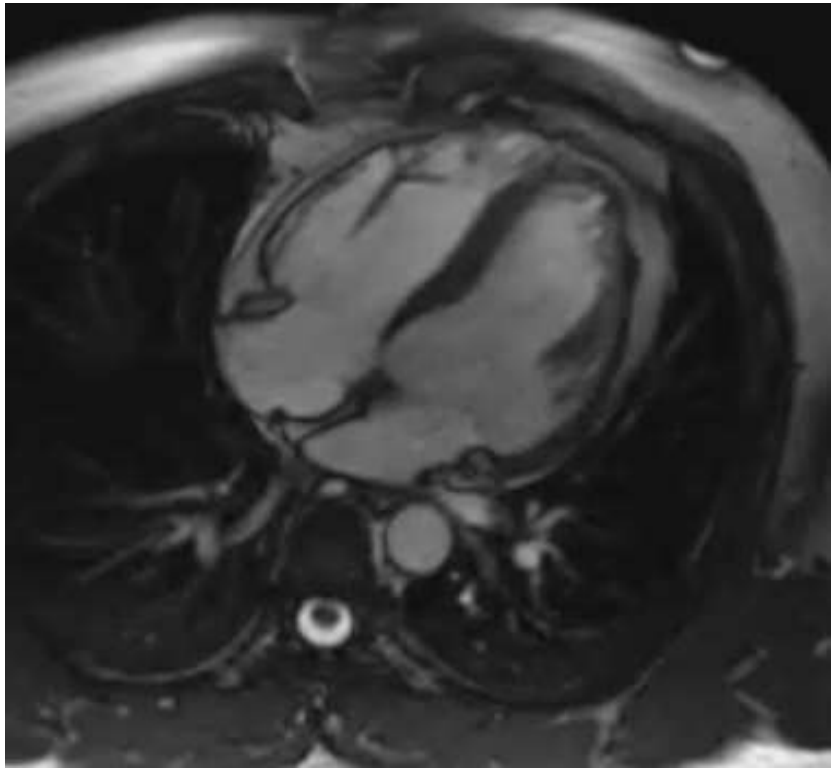
Coronary Angiography

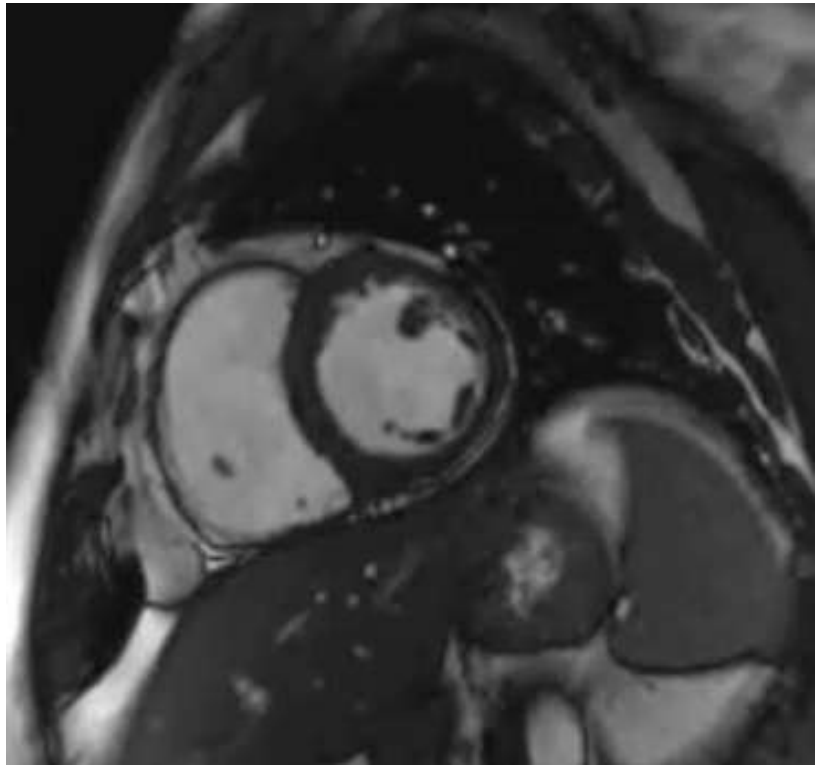


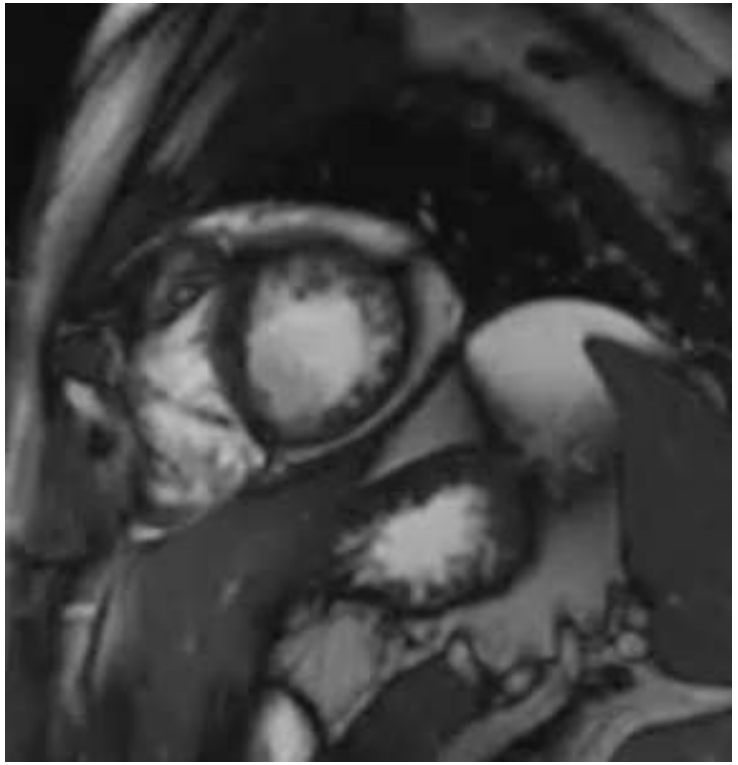


Normal Coronary Angiography

CMR (I)







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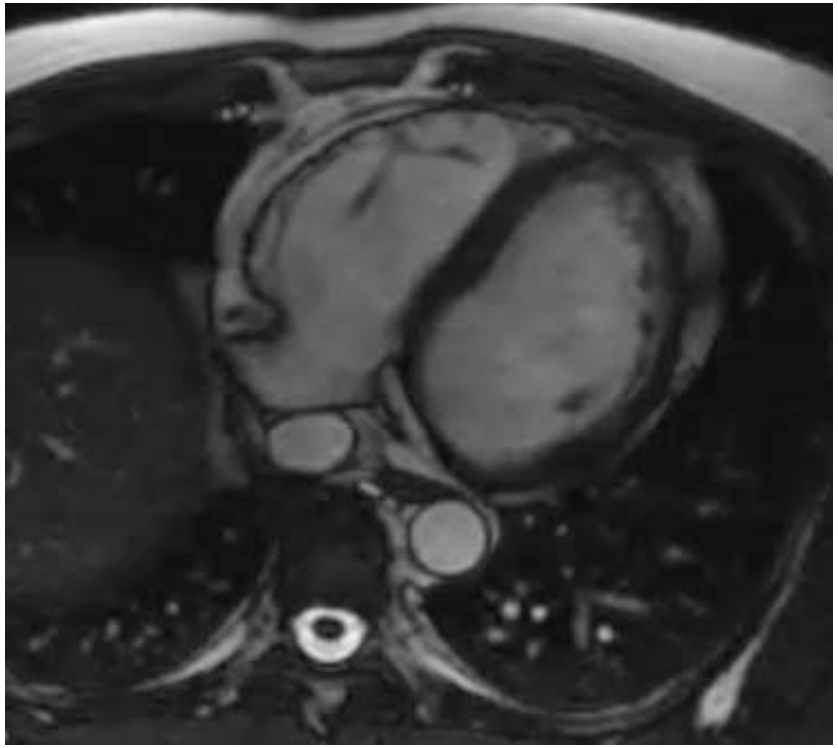
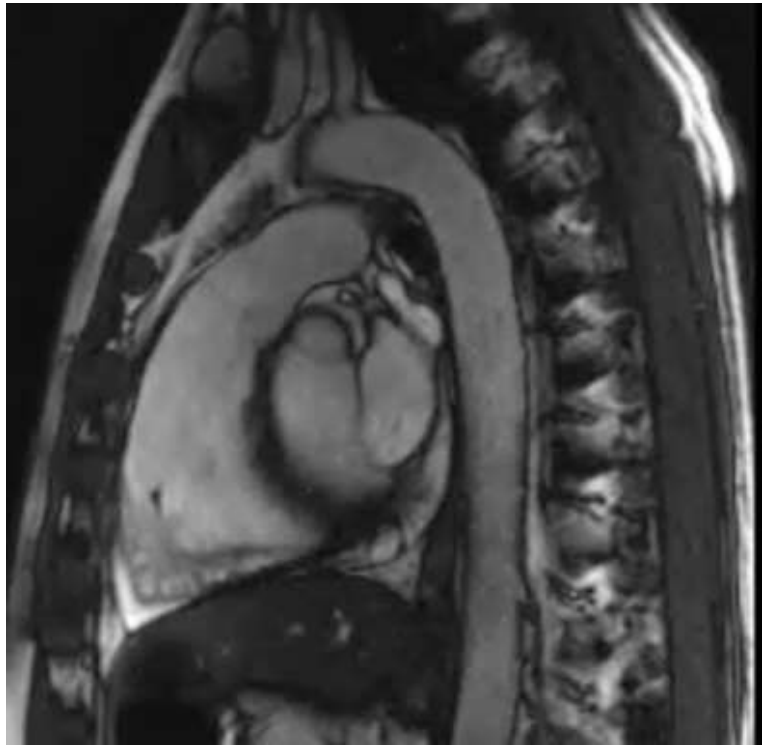
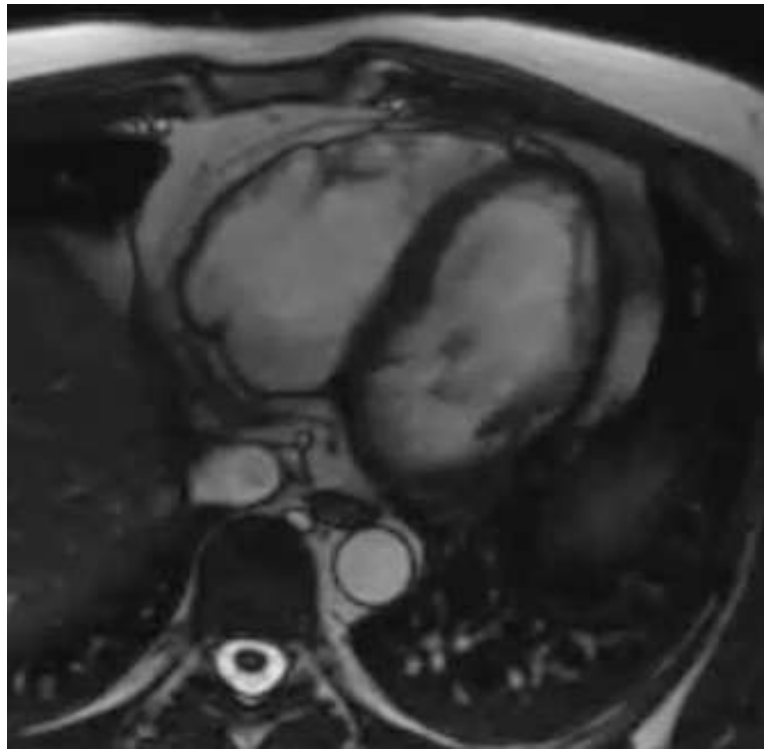
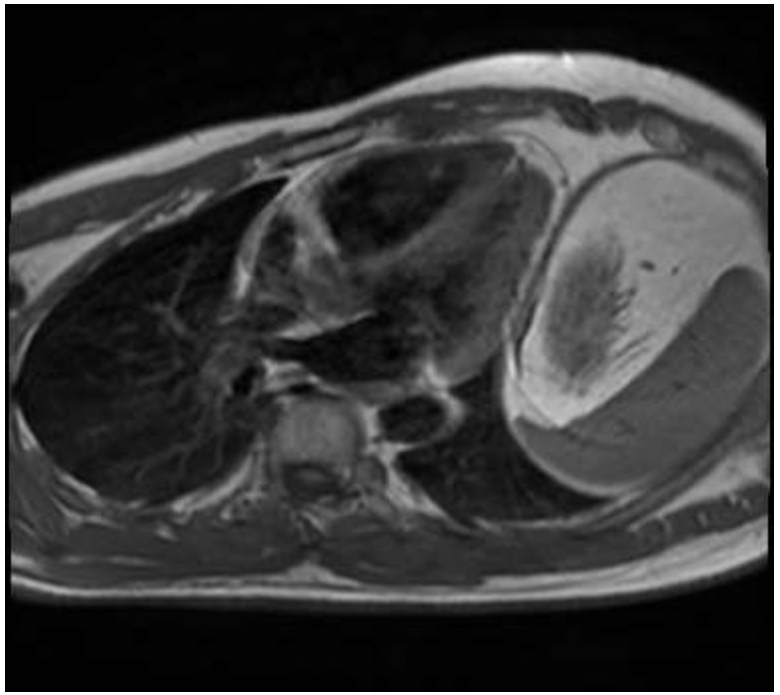


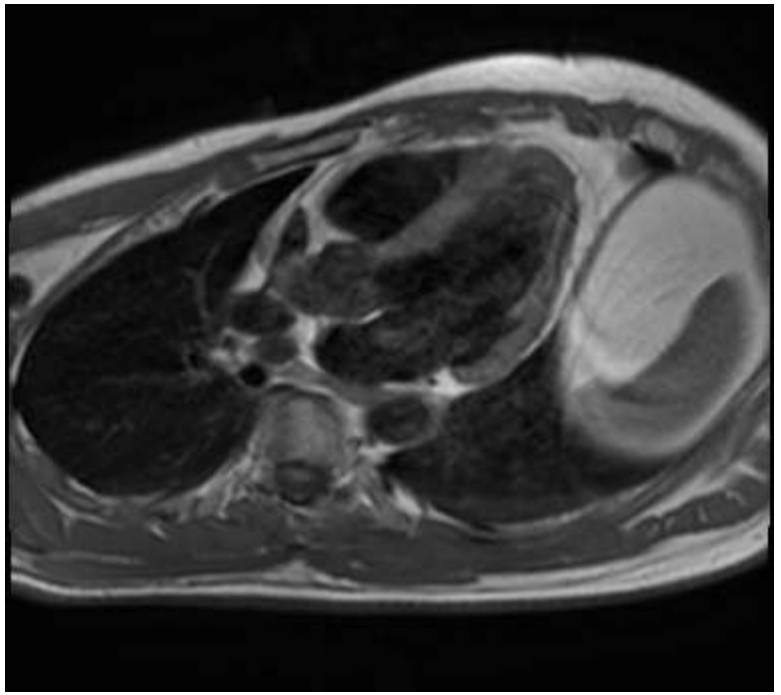


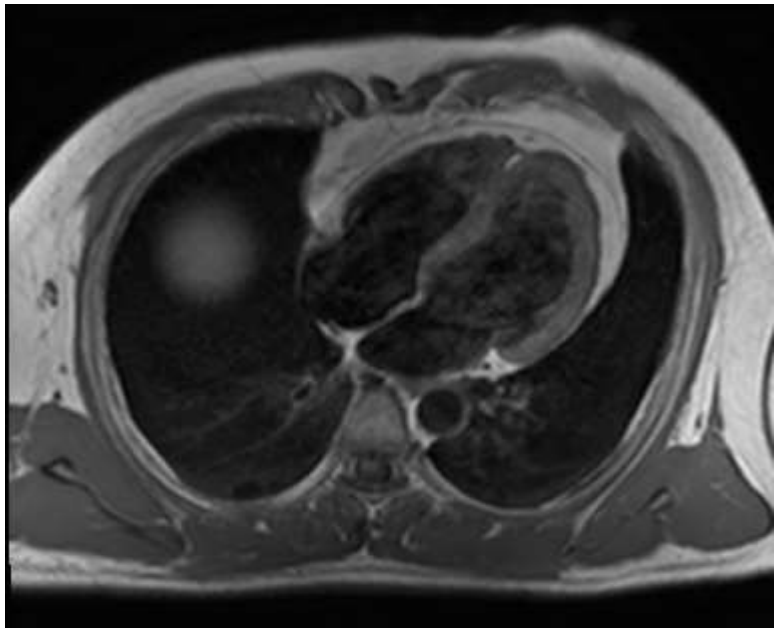


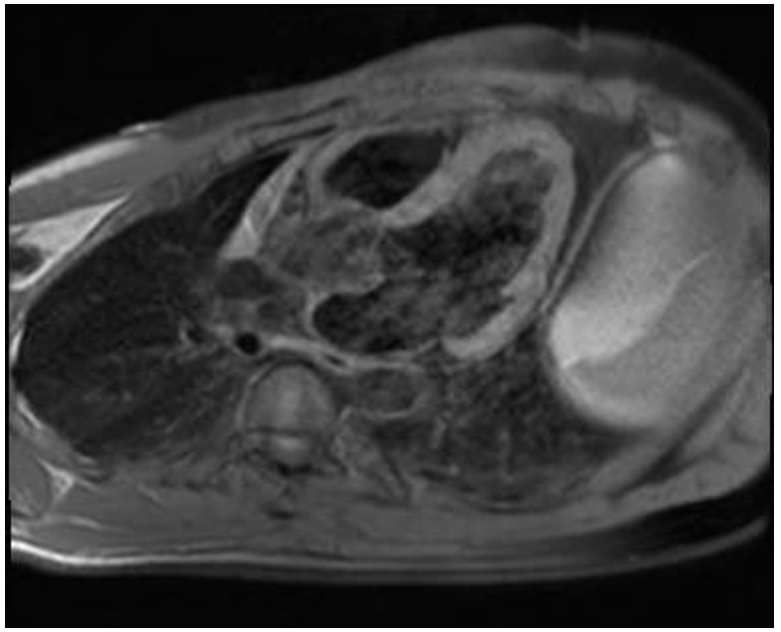
LV EF 49% normal EDV /BSA 82,6ml/m2
RV EF 43% normal EDV/BSA 93ml/m2

Τ1 ακολουθίες χωρίς και με καταστολή λίπους

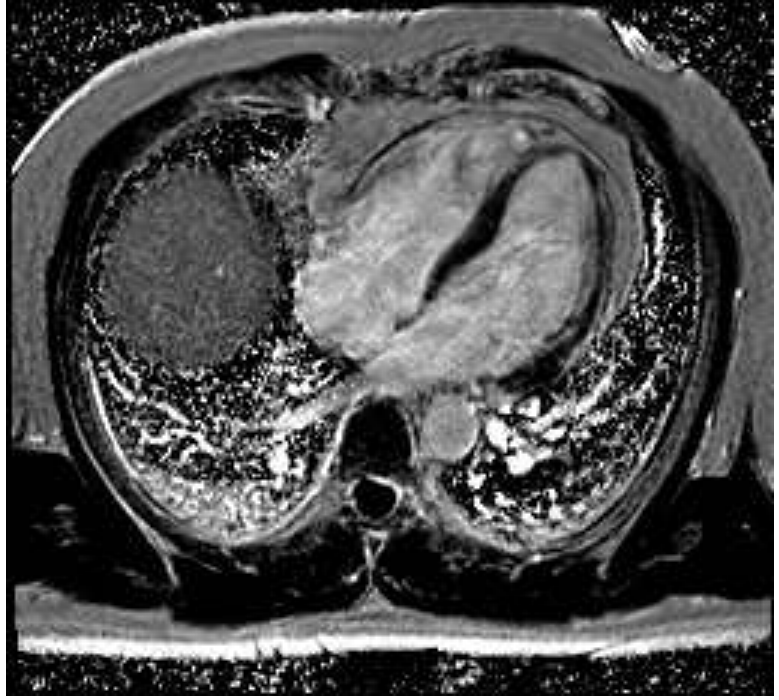


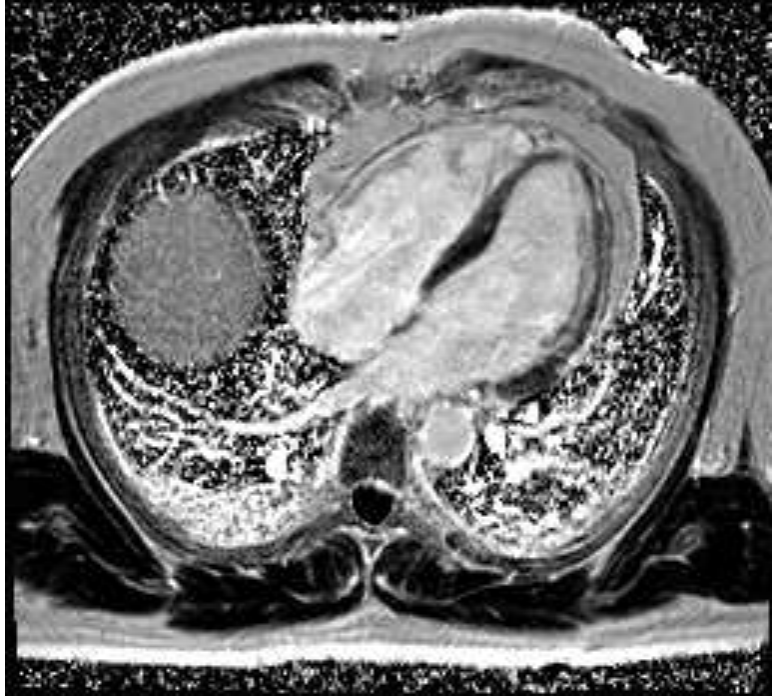


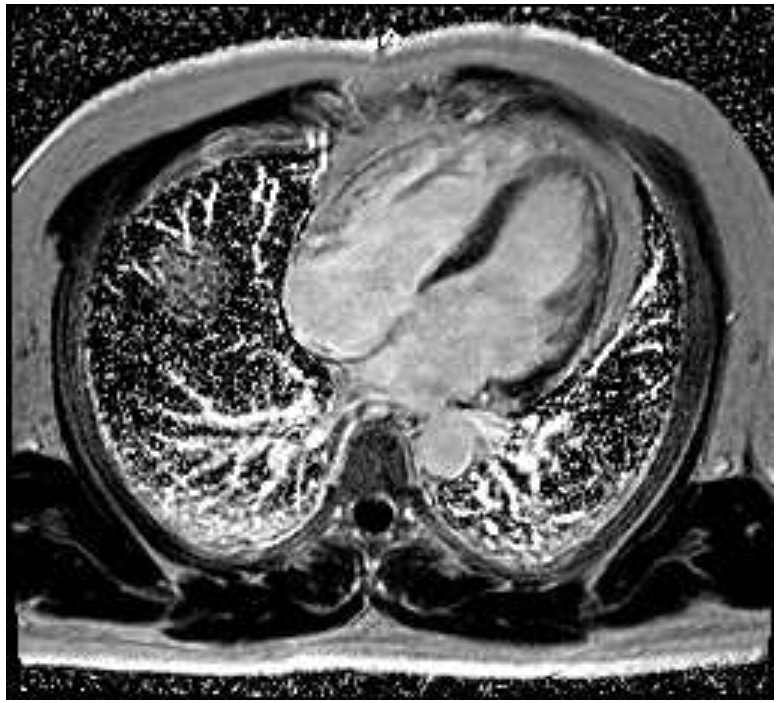


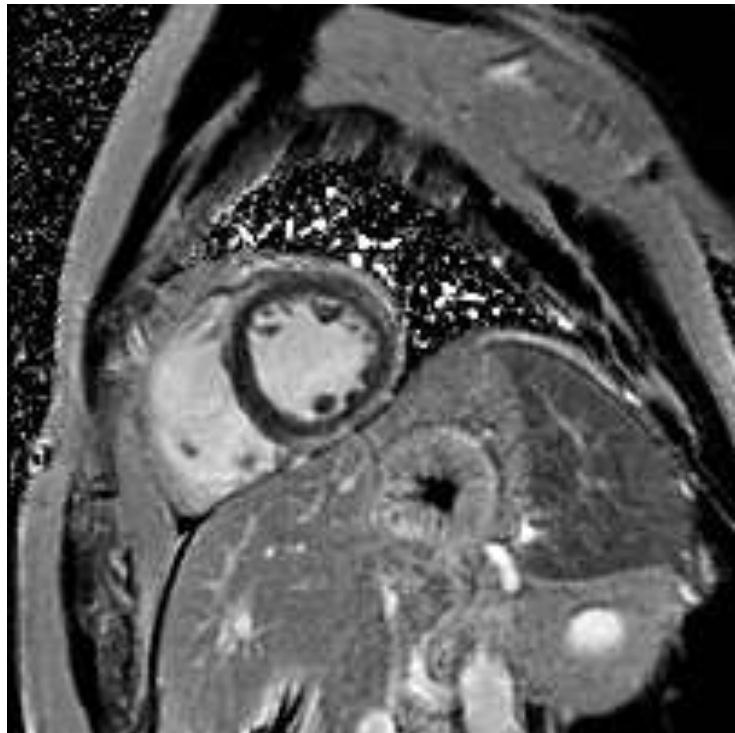


Ακολουθίες καθυστερημένης ενίσχυσης με γαδολίνιο

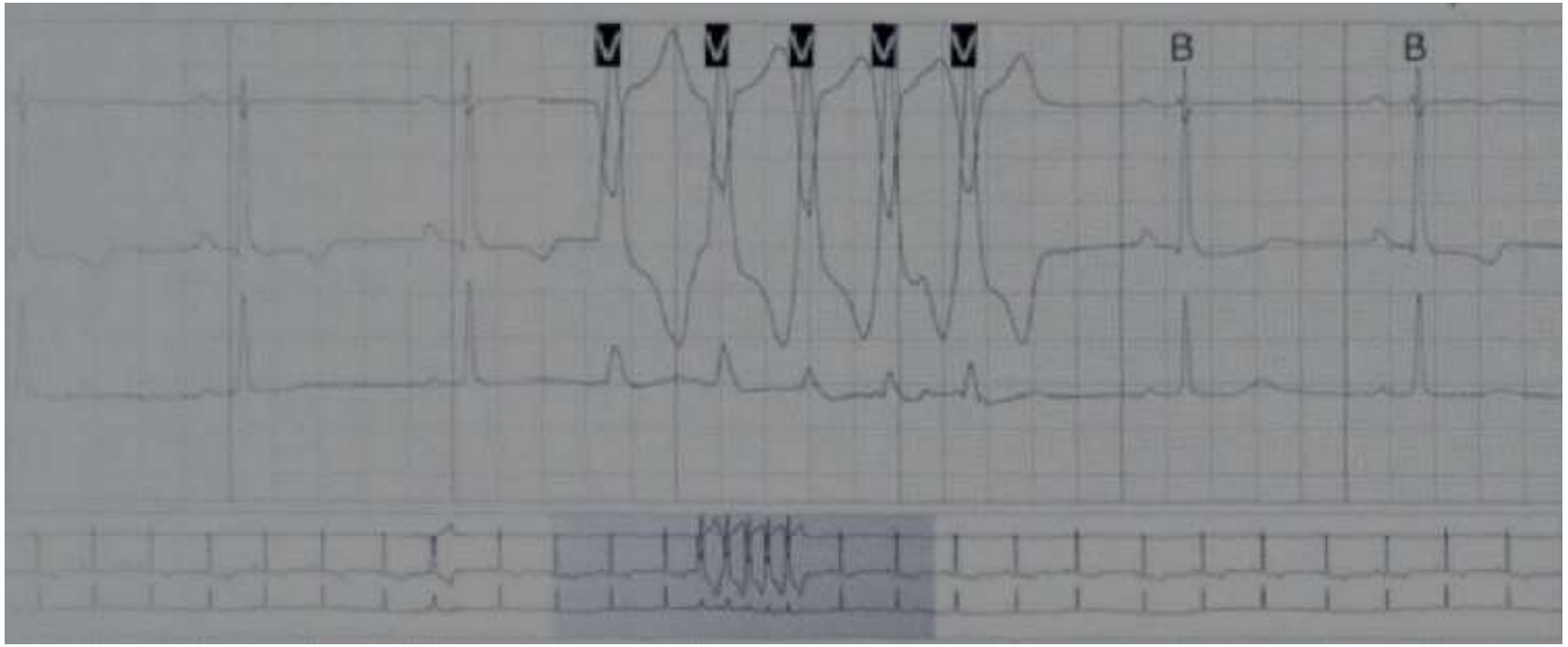








Holter ρυθμού



Revised (2010) Task Force Criteria For Diagnosis Of ARVC/D

Definite diagnosis: **2 major**, or **1 major plus 2 minor criteria** or **4 minor criteria** from different groups.

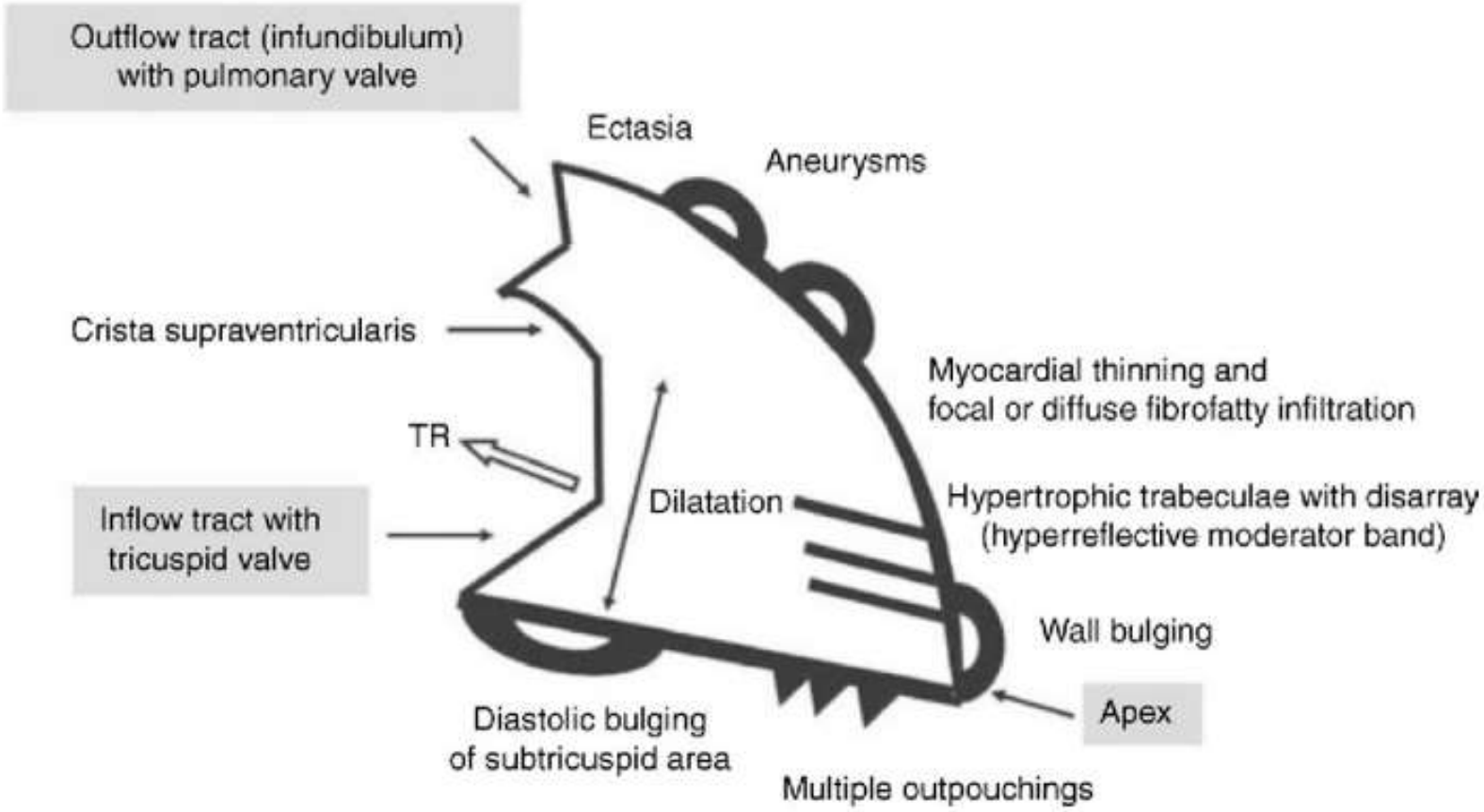
Borderline: diagnosis: **1 major and 1 minor** or **3 minor criteria** from different categories

Possible diagnosis: **1 major** or **2 minor** criteria from different categories.

FI. Marcus etal Circulation 2010

I. Global or regional dysfunction/structural alterations	
Major	***2D TTE***
	Regional RV akinesia, dyskinesia or aneurysm
	and 1 of the following criteria (end diastole):
	- PLAX RVOT ≥32 mm (PLAX/BSA] ≥19 mm/m²)
	- PSAX RVOT ≥36 mm (PSAX/BSA] ≥21 mm/m²)
	or RV fractional area change ≤33 %
	CMR
	Regional RV akinesia, dyskinesia, or dyssynchronous RV contraction
	and 1 of the following criteria (end diastole):
	- RV end-diastolic volume /BSA ≥110 mL/m² (male) or ≥100 mL/m² (female)
	or RV ejection fraction ≤40 %
	RV Angiography
	Regional RV akinesia, dyskinesia or aneurysm
Minor	***2D TTE***
	Regional RV akinesia, or dyskinesia
	and 1 of the following criteria (end diastole):
	- PLAX RVOT ≥29–31 mm ([PLAX/BSA] ≥16–18 mm/m²)
	- PSAX RVOT ≥32–35 mm ([PSAX/BSA] ≥18–20 mm/m²)
	- RV fractional area change >33–39 %
	CMR
	Regional RV akinesia, dyskinesia or dyssynchronous RV contraction
	and 1 of the following criteria (end diastolic):
	- RV end-diastolic volume/BSA ≥100–109 mL/m² (male) or ≥90–99 mL/m² (female)
	or RV ejection fraction >40–44 %
II. Histopathology (endomyocardial biopsy)	
Major	Residual myocytes <60 % by morphometric analysis (or <50 % if estimated), with fibrous replacement of the RV free wall myocardium ≥1 sample, with or without fatty replacement
Minor	Residual myocytes 60–75 % by morphometric analysis (or 50–65 % if estimated), with fibrous replacement of the RV free wall ≥1 sample, with or without fatty replacement
III. Repolarisation abnormalities (14 years of age)	
Major	T-wave inversions V1–3 or beyond (in absence of complete RBBB)
Minor	T-wave inversions V1–2 or V4–6 (in absence of complete RBBB)
	T-wave inversions V1–4, if complete RBBB present
IV. Depolarisation abnormalities	
Major	ε *wave* (reproducible low-amplitude signals between end of QRS complex to onset of the T wave) in V1–3
Minor	Signal-averaged ECG with late potentials (if QRS on standard surface ECG <110 ms)
V. Arrhythmias	
Major	Non-sustained or sustained ventricular tachycardia (VT) of LBBB morphology with superior axis
Minor	Non-sustained or sustained VT of RVOT configuration, LBBB morphology with inferior axis or of unknown axis
	>500 VES per 24 h (Holter)
VI. Family history	
Major	ARVC/D in a first-degree relative who meets current Task Force Criteria
	ARVC/D confirmed pathologically at autopsy or surgery in a first-degree relative
	Identification of a pathogenic mutation categorised as associated with ARVC/D in index patient
Minor	Suspected ARVC/D in a first-degree relative (current Task Force criteria can not be determined)
	Premature SCD (<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 relatives

Summary of the significant imaging findings in patients with ARVC/D ("**Triangle of Dysplasia**")



A Abidov et al (2016). CMR Features of ARVC/D. CMR in Diagnosis, Clinical Management, and Prognosis of ARVC

Arrhythmogenic (right) ventricular cardiomyopathy /dysplasia(ARVC/D)

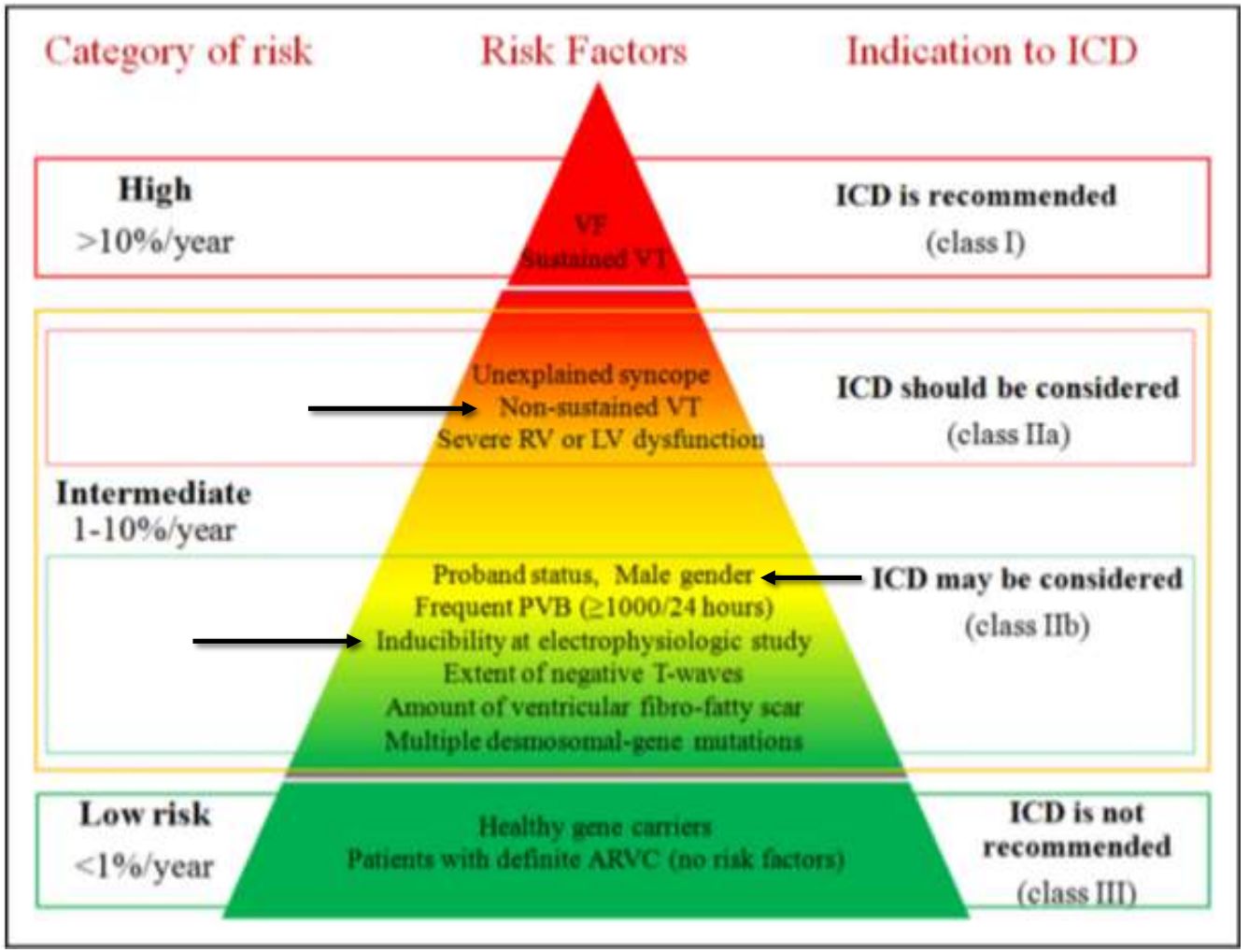
- ARVC/D is an inherited cardiomyopathy clinically characterized by ventricular arrhythmias and ventricular dysfunction.
- ARVC/D is a genetic disease that is characterized by (i) fibro-fatty replacement, (ii) myocardial atrophy, (iii) fibrosis, and (iv) chamber dilation and aneurysms.
- Progression to more diffuse RV disease and LV involvement, typically affecting the posterior lateral wall, is common.
- ARVC/D is considered to be familial with autosomal dominant inheritance, although there are recessive forms (eg, Naxos disease, Carvajal syndrome) that are associated with a cutaneous phenotype
- The prevalence of ARVC/D in the general population has been reported to be 1:5000, affecting men more frequently than women with a ratio of 3:1.

FI. Marcus etal Circulation 2010

ACE-inhibitors + Beta Blockers

Risk Stratification in ARVC

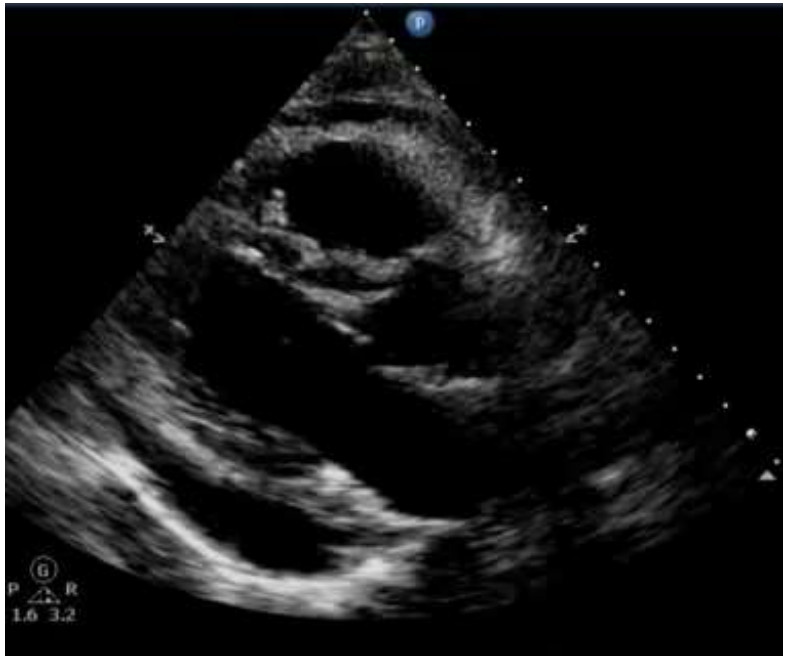
Because electrical abnormalities can precede structural abnormalities with SCD as the first tragic and lethal manifestation, especially in young adults with many potential years of life lost, optimizing screening strategies is of paramount importance.

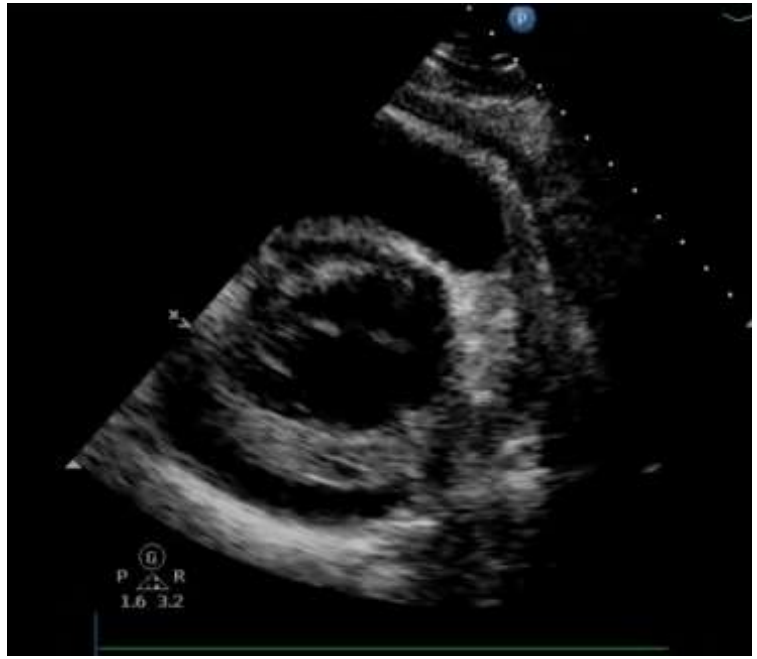


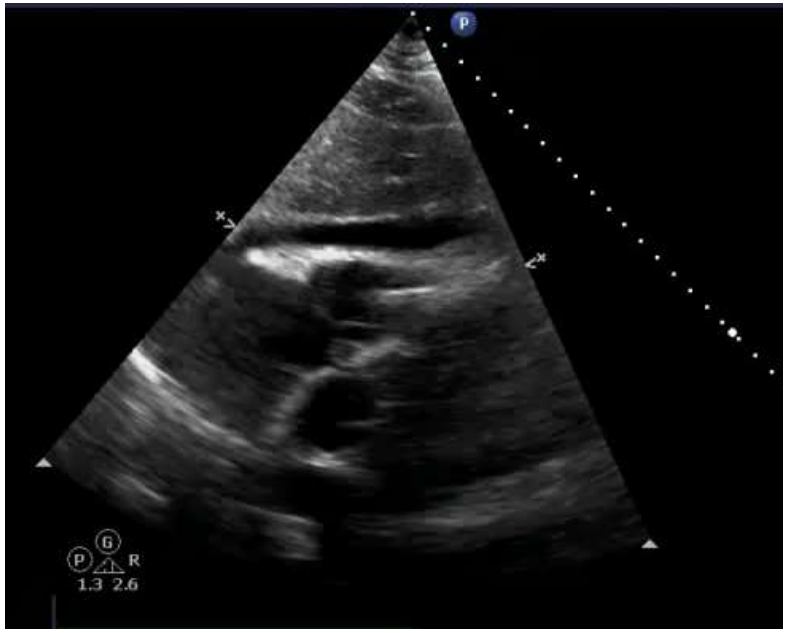
D Corrado Circ Res 2017

1 month later after ICD implantation

- the patient admitted with chest pain dyspnea and fever.
- ECHO revealed moderate pericardial effusion
- NSAID’s + Colchicine

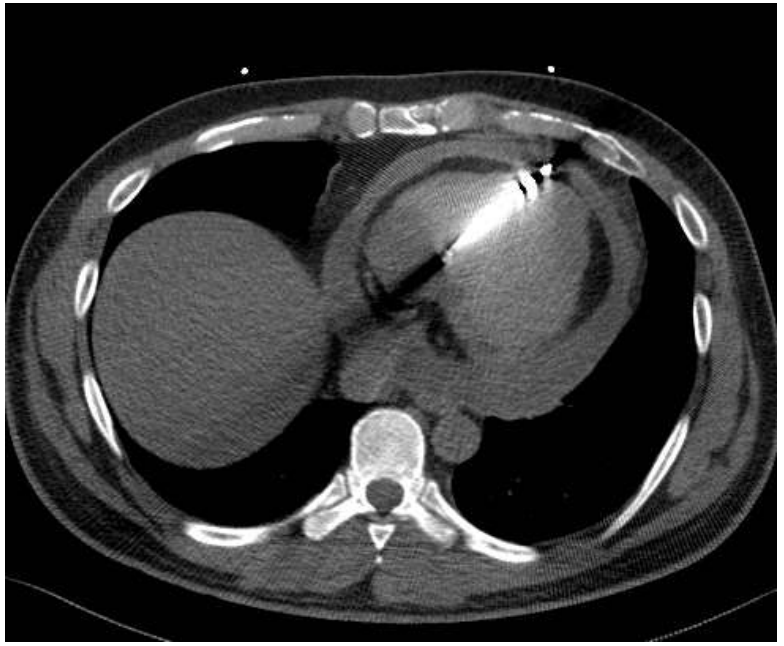


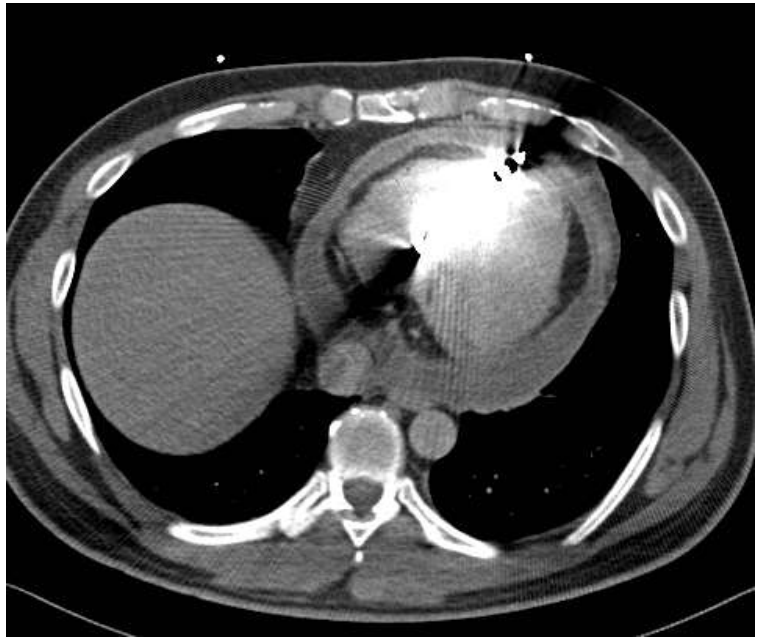


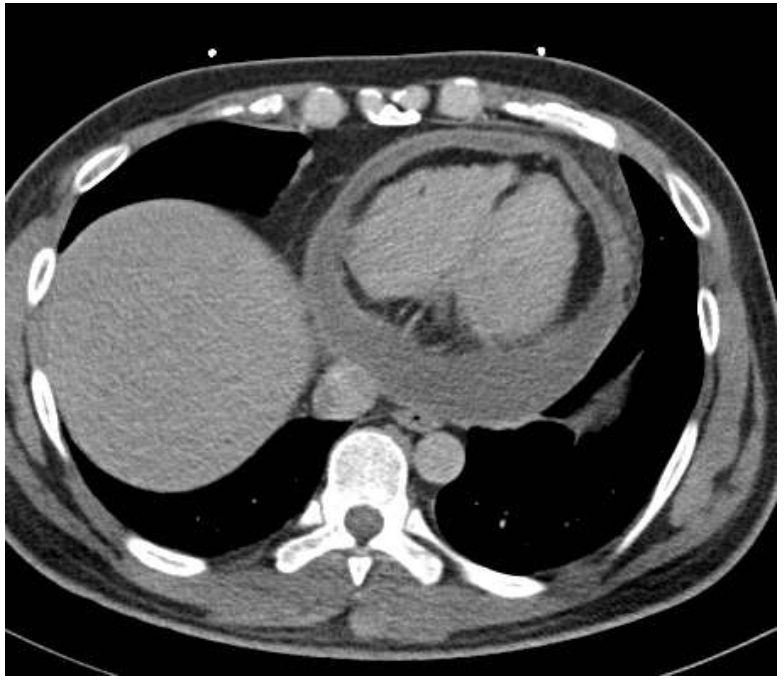


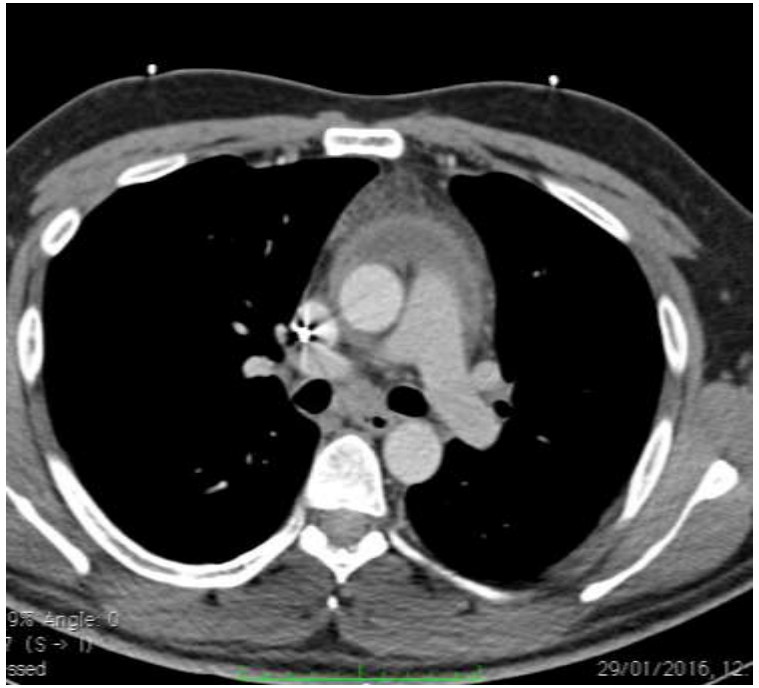
2 weeks later
Cardiac CT

Cardiac Operation



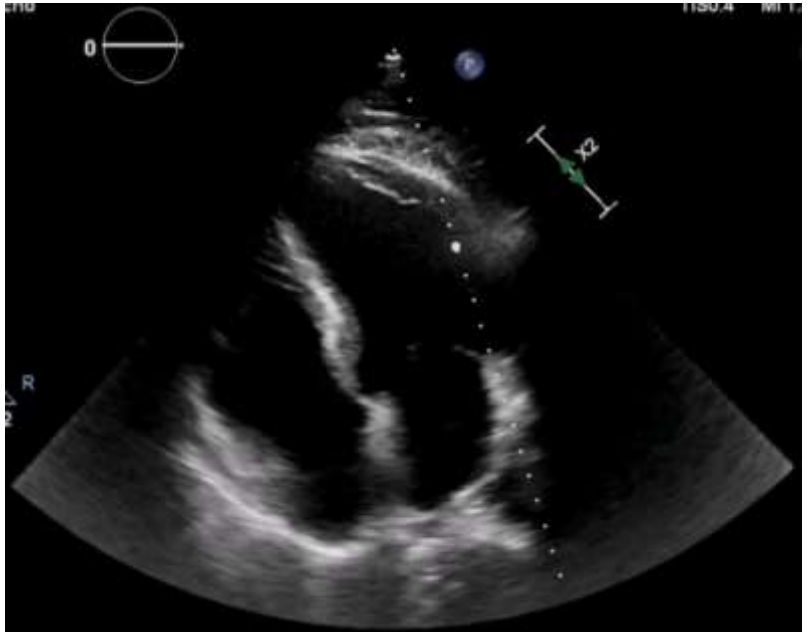


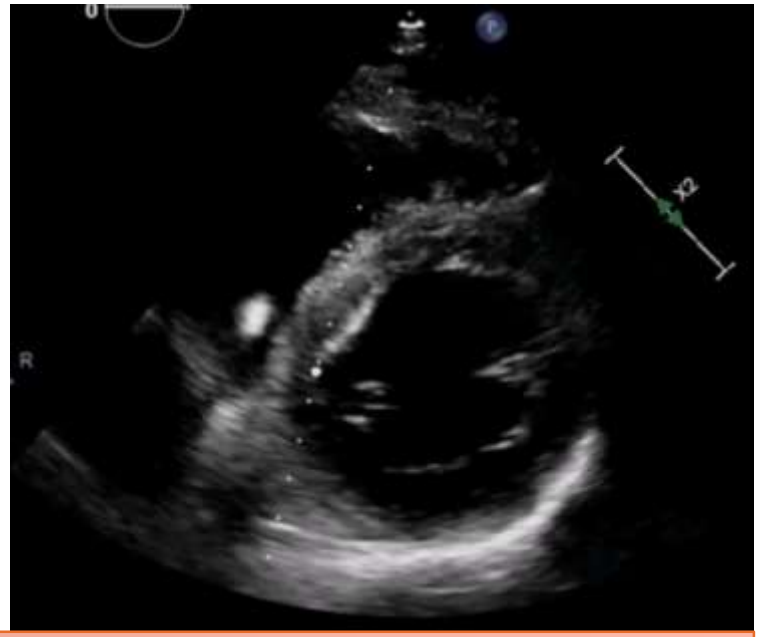




2 Χρόνια αργότερα

- Ο/Σ συσφικτικό άλγος
- Αυξημένες τιμές μυοκαρδιακών ενζύμων
- ΗΚΓ χωρίς αλλαγές
- MSCTA: χωρίς ευρήματα





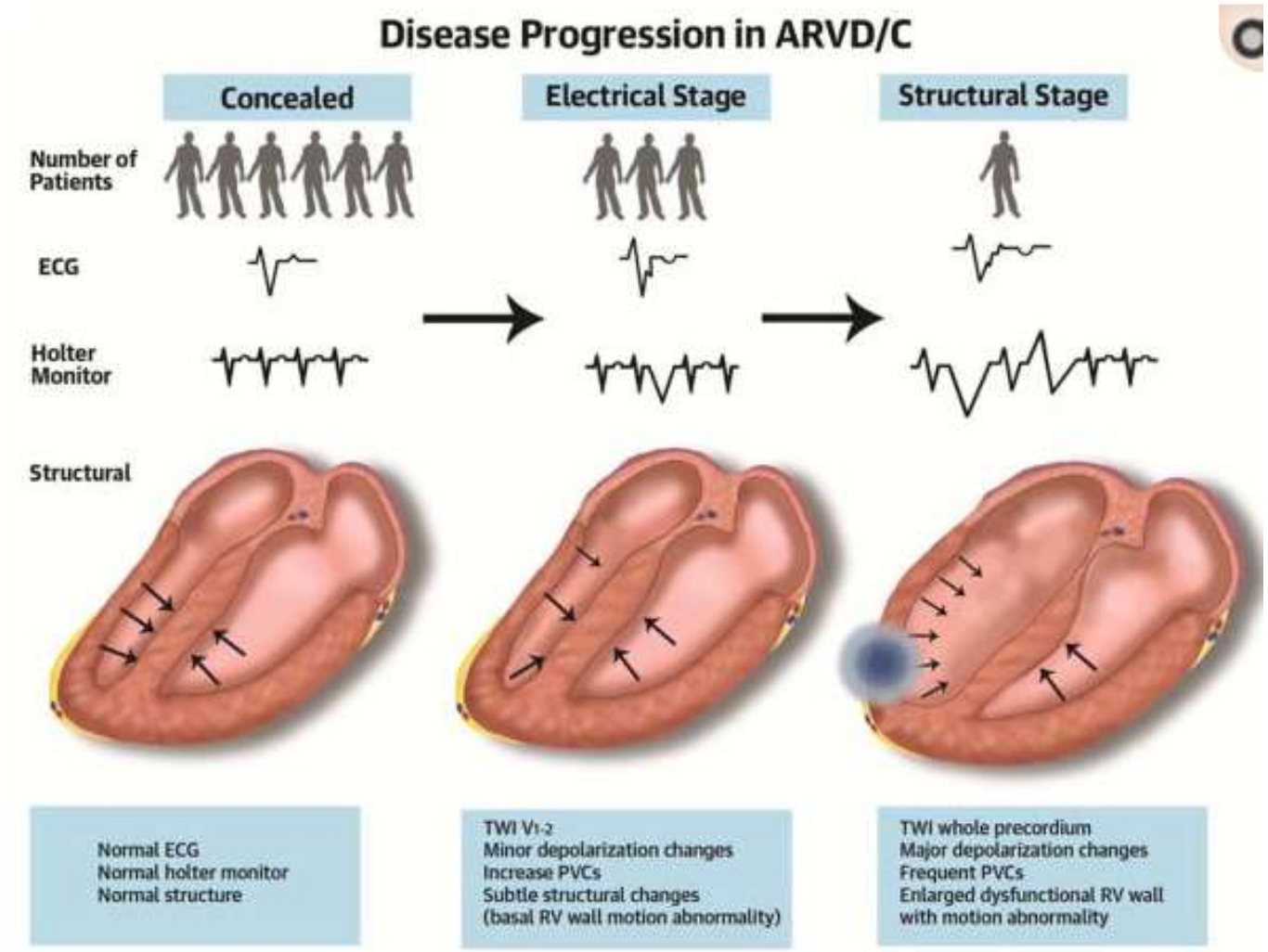
Periodic exacerbations of a previously quiescent disease may be triggered by such inflammatory episodes and are called "**hot phases**" of ARVC. Occasionally, these phases may clinically present with **chest pain**, **increased cardiac troponin level**, **dynamic ECG changes** and **increased arrhythmic activity**.

There are certain times – known as '**hot phases**' – when the disease process becomes more active, increasing the risk of sudden cardiac death. For example, unexplained dizzy spells, sustained palpitations, or blackouts can be a sign of a hot phase.. Hot phases may come and go, but they could be a sign that the disease is progressing.

AM Saguner et al WJC 2014

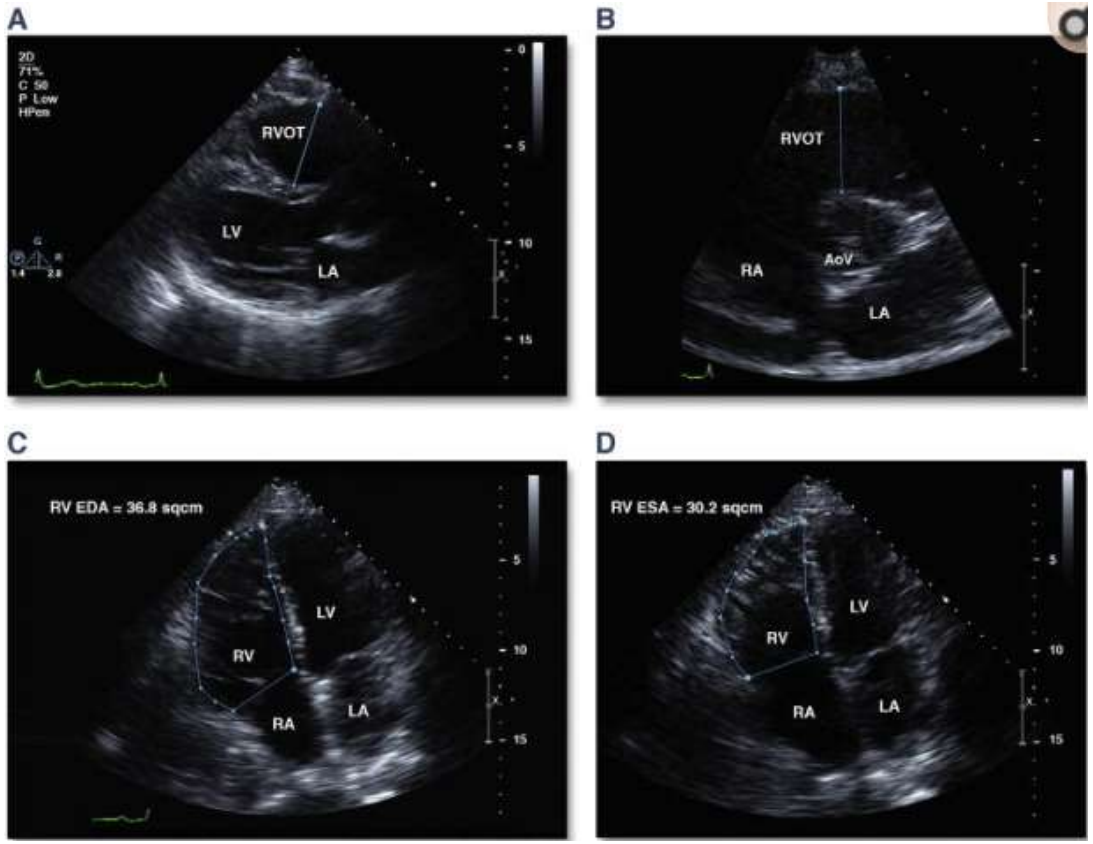
Disease Progression in ARVD/C

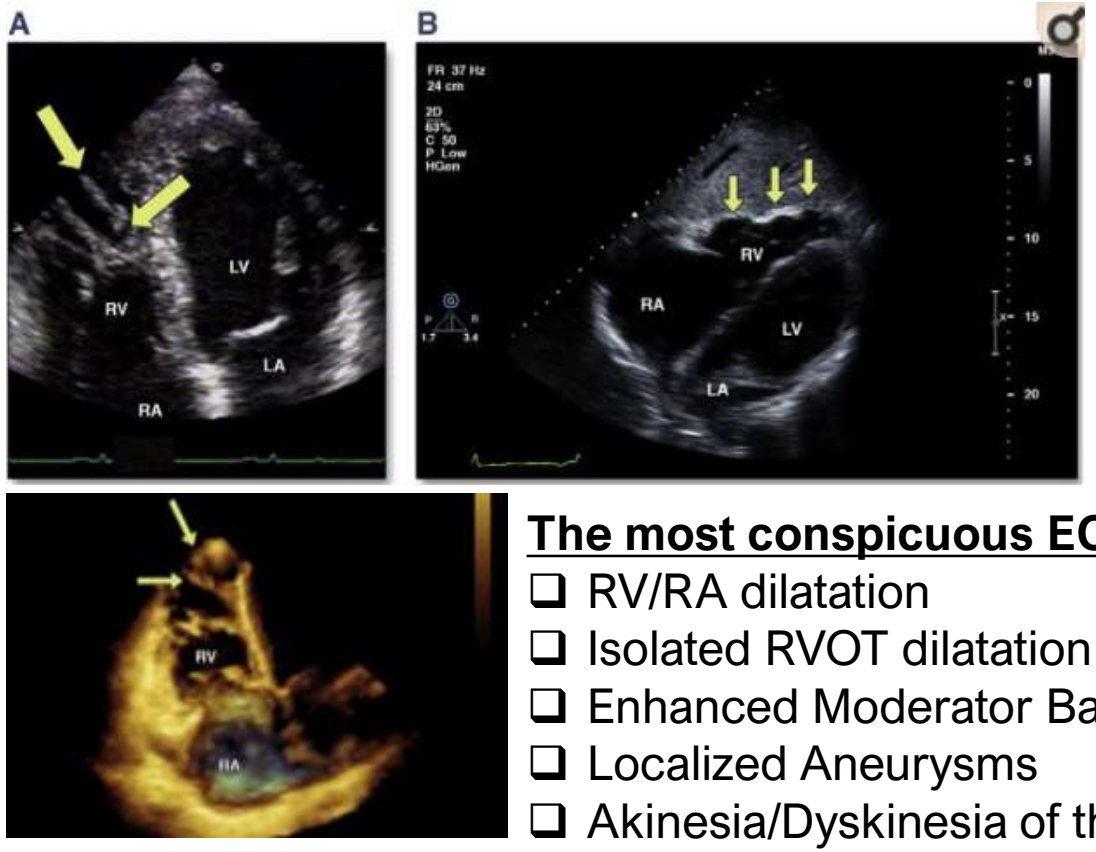
- Family members of an ARVC/D proband have a long latent stage without signs or symptoms of disease (concealed stage).
- Electrical changes on electrocardiogram or Holter monitoring are usually the first sign of disease, and structural abnormalities may be observed later.
- Disease progression is typically slow.



A Riele et al JACC 2014

Transthoracic Echocardiogram in ARVC/D



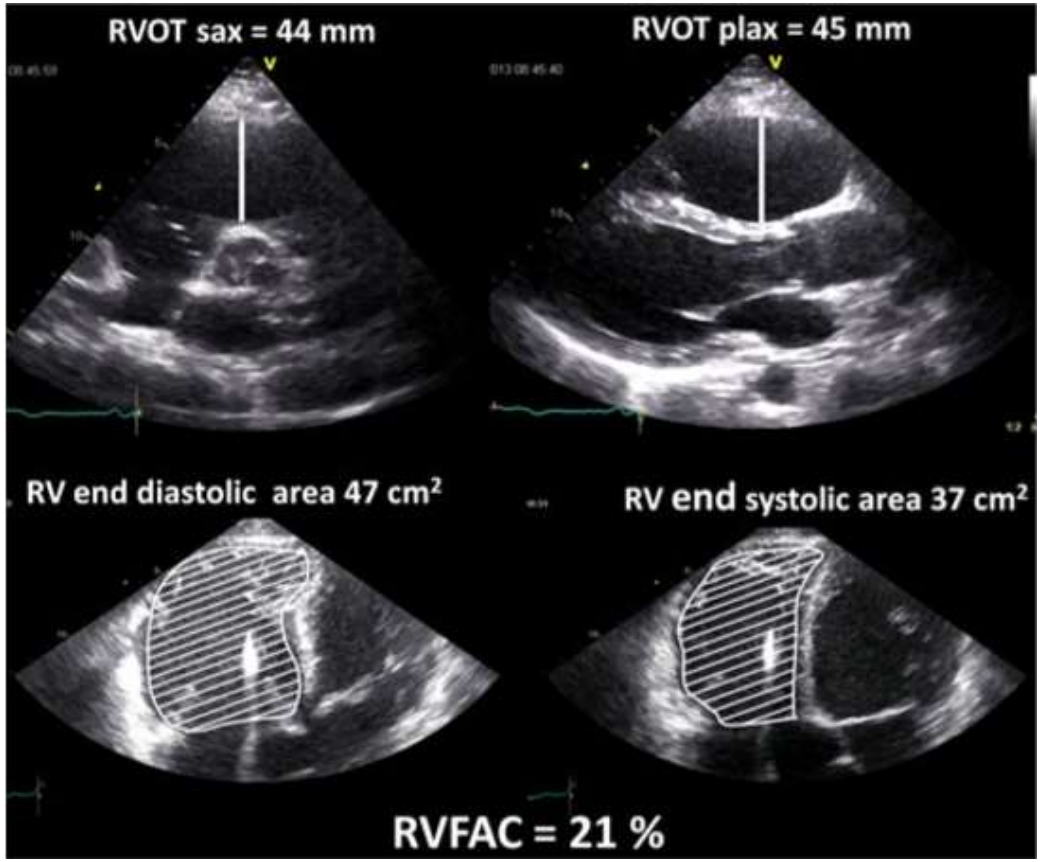


The most conspicuous ECHO findings:

- ❑ RV/RA dilatation
- ❑ Isolated RVOT dilatation
- ❑ Enhanced Moderator Band
- ❑ Localized Aneurysms
- ❑ Akinesia/Dyskinesia of the inferior wall and RV apex

A Riele JACC Card Imag 2015

Echocardiographic measurements in a patient with ARVC/D (Task Force Criteria 2010)

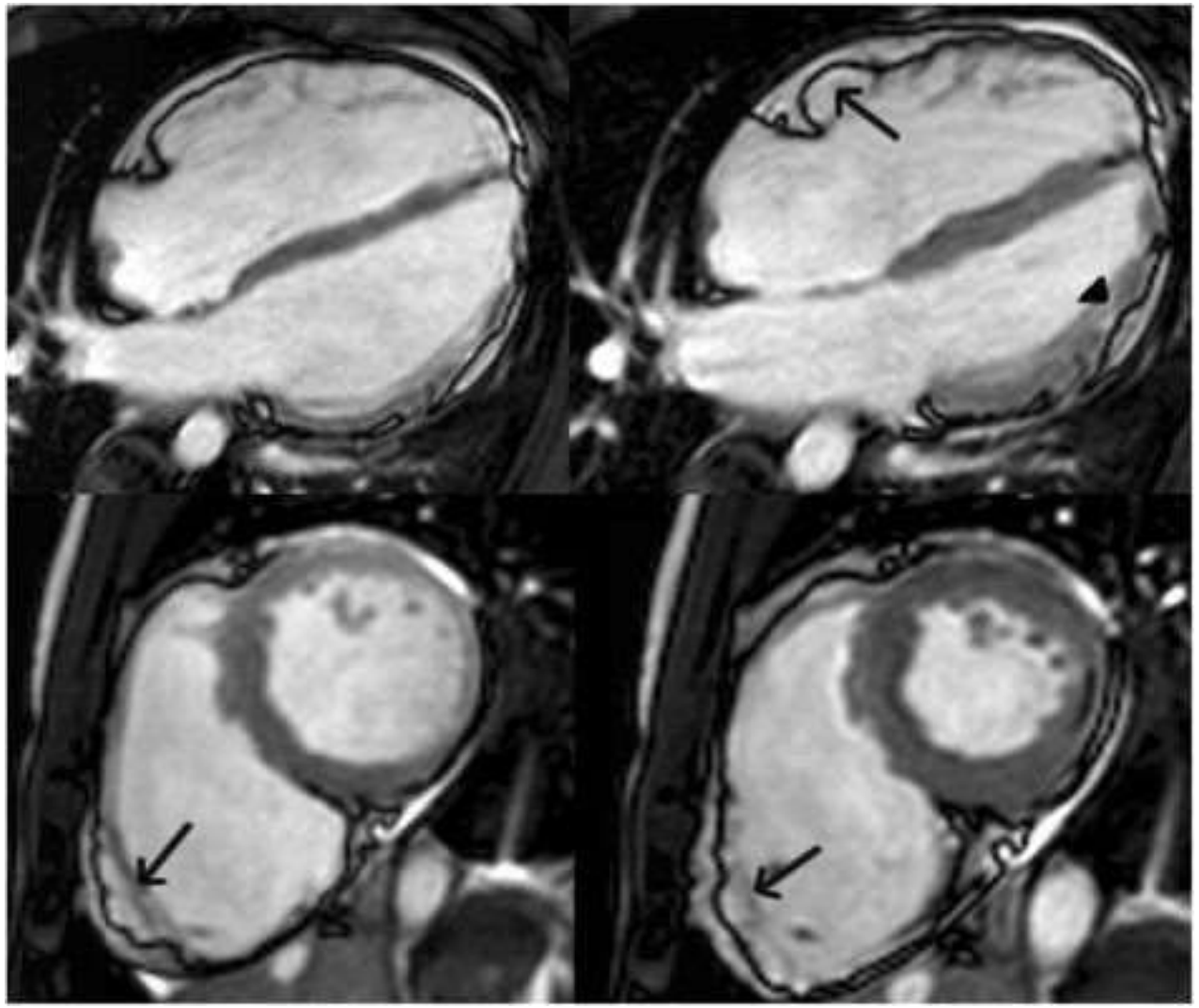


- Echocardiography is the first line imaging modality in ARVC, and the most commonly used imaging tool for follow-up of ARVC patients.
- To diagnose ARVC by echocardiography is challenging and requires high expertise.
- Quantitative assessment of RV function is challenging due to complex anatomy and load dependency.

KH. Haugaa et al Europace 2015

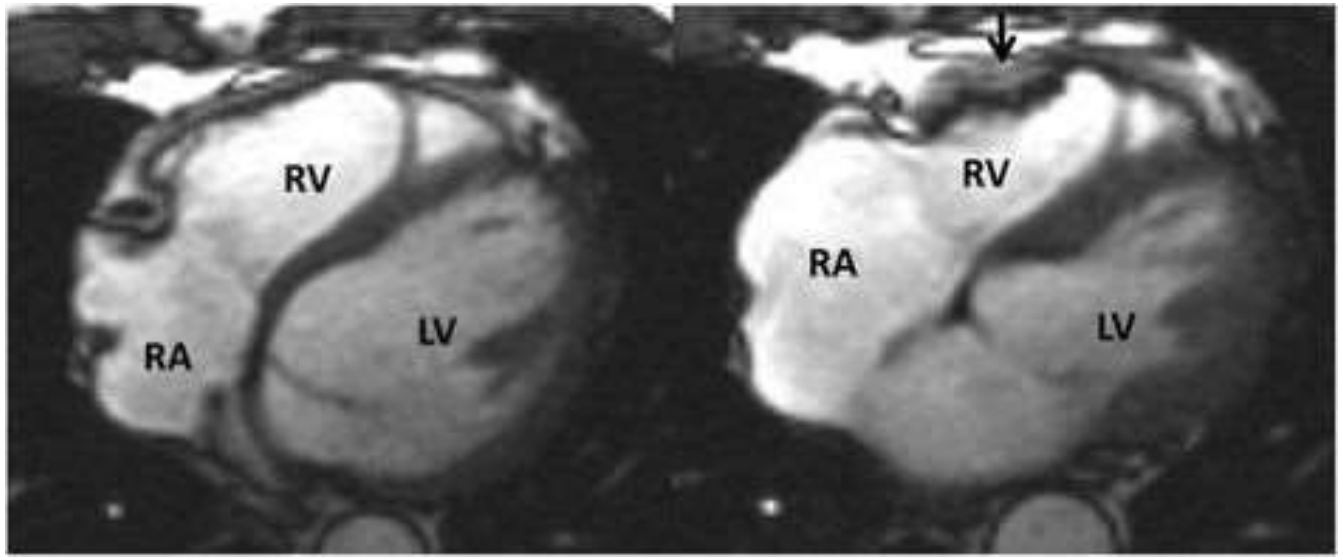
Four-chamber and short-axis bright blood images in an ARVC subject

End-diastolic images are shown in the left panels, end-systolic images in the right panels. Note **subtricuspid dyskinesia** in the end-systolic four-chamber image (arrow), and **right ventricular free wall aneurysms** (i.e. both systolic and diastolic bulging) in the short-axis image (arrows).



A Riele et al J Cardiovasc Magn Res 2014

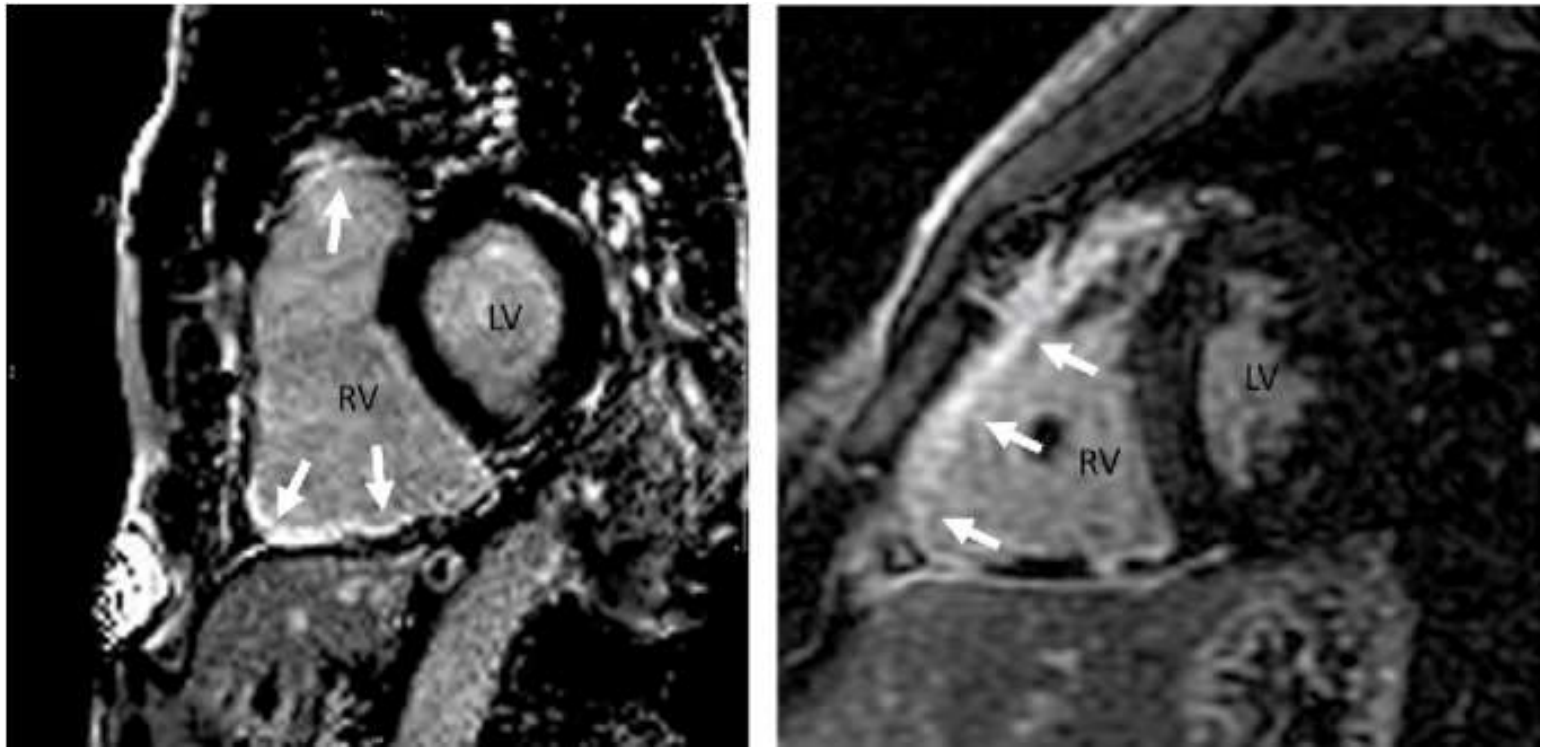
Regional contraction abnormality in the sub-tricuspid region



End diastolic (left) and end systolic image (right) show the so-called "**accordion sign**" in an ARVC mutation carrier. Regional dyssynchronous contraction in the sub-tricuspid region is a readily recognized qualitative pattern of abnormal RV contraction.

A Riele et al J Cardiovasc Magn Res 2014

Delayed enhancement CMR imaging



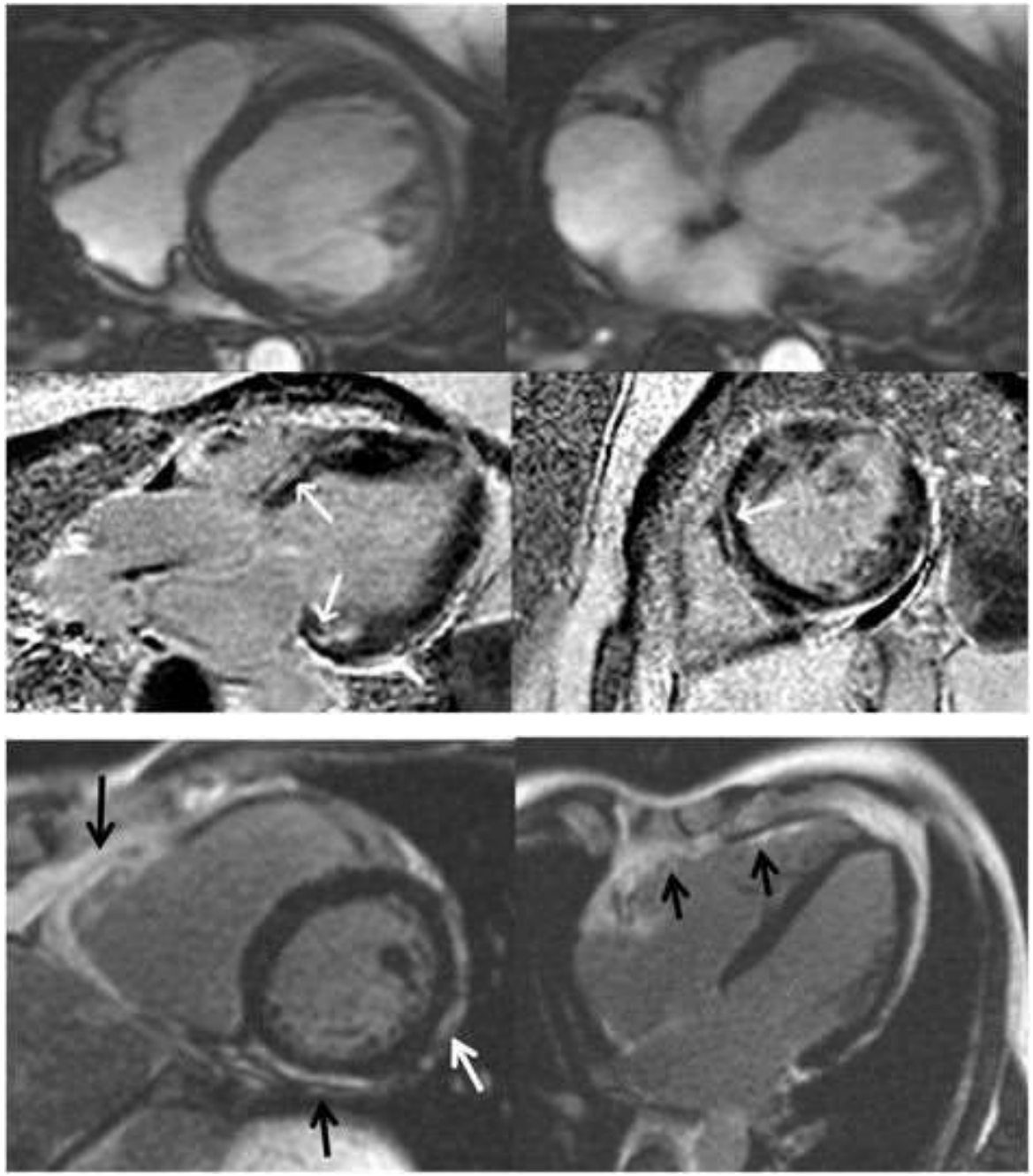
Evidence of the extensive RV fibrotic changes (white arrows) in patients with advanced ARVC/D on the delayed enhancement CMR imaging.

A Abidov et al (2016). CMR Features of ARVC/D. CMR in Diagnosis, Clinical Management, and Prognosis of ARVC

CMR in ARVC subject with predominant LV involvement

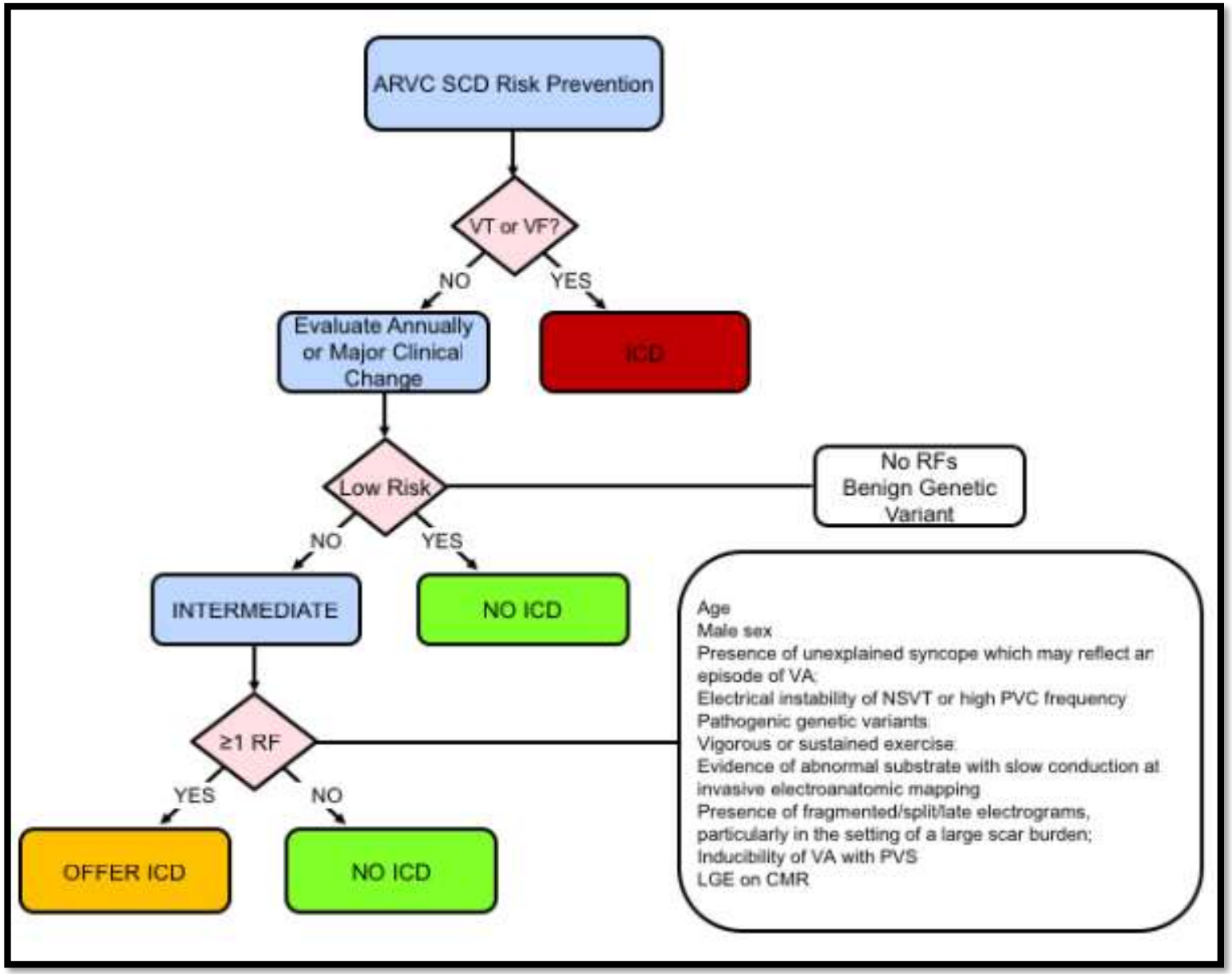
Note a dilated left ventricle in the bright blood images. Late enhancement is observed in a mid-myocardial pattern in the basal septum and basal lateral wall (arrows, bottom panels).

The lateral wall of the LV shows thinning due to fatty replacement that was confirmed on T1-weighted images. The long axis view (right) shows diffuse LGE involving the free wall of the RV.



A Riele et al J Cardiovasc Magn Res 2014

Risk Stratification in ARVC



A Chahal et al JACC 2019

Cardiac Perforation From Implantable Cardioverter-Defibrillator Lead Placement

- 440 251 first-time ICD recipients in the ICD Registry implanted between 2006 and 2011.
- **Cardiac perforation occurred in 625 patients (0.14%)**
- After multivariable adjustment, **older age**, **female sex**, **left bundle branch block**, **worsened heart failure class**, **higher left ventricular ejection fraction**, and **non–single-chamber ICD implant** were associated with a greater odds of perforation.

J Hsu et al Circulation 2018

But what happens in ARVC patients?

- 106 consecutive patients with ARVC who received an ICD
- **Cardiac perforation occurred in 3 patients (2.83%)**

D Corrado et al Circulation 2010

ΕΥΧΑΡΙΣΤΩ ΠΟΛΥ