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pratiques de rythmologie
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Dysplasie Arythmogène du VD

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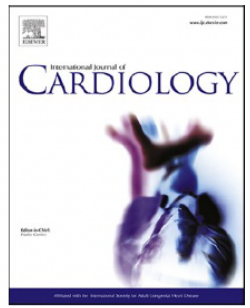

Proposed diagnostic criteria for arrhythmogenic cardiomyopathy: European Task Force consensus report☆

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European Task Force criteria for diagnosis of Arrhythmogenic Cardiomyopathy.

Category	RV Phenotype	LV Phenotype
I. Morpho-functional ventricular abnormalities	***Major*** • Regional RV akinesia, dyskinesia, or aneurysm *plus* one of the following: • global RV dilatation (increase of RV EDV according to the imaging test specific nomograms for age, sex and BSA)* or • global RV systolic dysfunction (reduction of RV EF according to the imaging test specific nomograms for age and sex)* ***Minor*** • Regional RV akinesia, dyskinesia or aneurysm of RV free wall	***Minor*** • Global LV systolic dysfunction, with or without LV dilatation (increase of LV EDV according to the imaging test specific nomograms for age, sex, and BSA)*
II. Structural alterations	***Major*** • Fibrous replacement of the myocardium in ≥1 sample, with or without fatty tissue, at histology ***Minor*** • Unequivocal RV LGE (confirmed in 2 orthogonal views) in ≥1 RV region(s) (excluding tricuspid valve)	***Major*** • "Ring-like" LV LGE (subepicardial or midmyocardial stria pattern) of ≥3 segments (confirmed in 2 orthogonal views), ***Minor*** • LV LGE (subepicardial or midmyocardial stria pattern) of 1 or 2 Bull's Eye segment(s) (in 2 orthogonal views) of the free wall, septum, or both (excluding patchy, focal or septal junctional LGE**)

III. Repolarization abnormalities	***Major*** • Negative T waves in right precordial leads (V_1, V_2, and V_3) or beyond in individuals ≥14 year-old (in the absence of complete RBBB and not preceded by J-point/ST-segment elevation) ***Minor*** • Negative T waves in leads V1 and V2 in males ≥14 year-old (in the absence of RBBB and not preceded by J-point/ST-segment elevation) • Negative T waves beyond V3 in the presence of complete RBBB • Negative T waves beyond V3 in individuals <14 year-old	***Minor*** • Negative T waves in left precordial leads (V_4-V_6) (in the absence of complete LBBB)
IV. Depolarization and conduction abnormalities	***Minor*** • Epsilon wave (reproducible low-amplitude signals between 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)	***Major*** • Low QRS voltages (<0.5 mV peak to peak) in all limb leads in the absence of other causes (e.g. cardiac amyloidosis, obesity, emphysema, or pericardial effusion)
V. Arrhythmias	***Major*** • Frequent ventricular extrasystoles (>500 per 24 h), non-sustained or sustained ventricular tachycardia of LBBB morphology with non-inferior axis ***Minor*** • Frequent ventricular extrasystoles (>500 per 24 h), non-sustained or sustained ventricular tachycardia of LBBB morphology with inferior axis ("RVOT pattern") • History of cardiac arrest due to ventricular fibrillation or sustained ventricular tachycardia of unknown morphology	***Minor*** • Frequent (>500 per 24 h) or exercise-induced ventricular extrasystoles with a RBBB morphology or multiple RBBB morphologies (excluding the "fascicular pattern") • Non-sustained or sustained ventricular tachycardia with a RBBB morphology (excluding the "fascicular pattern") • History of cardiac arrest due to ventricular fibrillation or sustained ventricular tachycardia of unknown morphology

VI. Family history/genetics	***Major*** • Identification of a pathogenic ACM-gene variant in the patient under evaluation • ACM confirmed in a first-degree relative who meets diagnostic criteria • ACM confirmed pathologically at autopsy or surgery in a first-degree relative ***Minor*** • Identification of a likely-pathogenic ACM-gene variant in the patient under evaluation • 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 second-degree relative

Different causes of Arrhythmogenic Cardiomyopathy.

ETIOLOGY		PHENOTYPIC VARIANT(S)	OTHER (POSSIBLE) PHENOTYPIC FEATURES
Genetic causes			
Desmosomal gene defects			
	PKP2 - Plakophilin C	ARVC	
	DSP - Desmoplakin	ALVC-BIV-ARVC	Subtle hair and skin abnormalities
	DSC2 - Desmocollin 2	ARVC-BIV	
	DSG2- Desmoglein 2	ALVC-BIV	
	Cardio-cutaneous syndromes		
	JUP-Plakoglobin (recessive)	ARVC-ALVC	Hair and skin abnormalities (Naxos disease)
	DSP-Desmoplakin (recessive)	ALVC	Hair and skin abnormalities (Carvajal disease)
Non-desmosomal gene defects (genocopies)			
	TMEM 43 (Transmembrane protein 43 - luma)	ARVC	High risk of SCD in males
	PLN (Phospholamban)	ALVC-BIV-ARVC	
	FLNC (Filamin C)	ALVC-BIV	Skeletal myofibrillar myopathy.
	DES (Desmin)	ALVC-BIV	Skeletal myofibrillar myopathy. Conduction system abnormalities
	LMNA (Lamin A/C)	ALVC-BIV	Skeletal muscular dystrophy. Sinus node dysfunction and conduction system abnormalities.
	TGFB3 (transforming growth factor-3)*	ARVC	
	CTNNA3 (alpha-T-catenin)*	ARVC	
	CDH2 (cadherin-2)*	ARVC	
	SCN5A (Sodium channel alpha unit)*	ARVC-ALVC	
	Neuromuscular disorders		
	DMD-Duchenne muscular dystrophy	ALVC	
	DMD-Becker muscular dystrophy	ALVC	
	DMPK-Myotonic dystrophy or Steinert	ALVC	Sinus node dysfunction and/or AV conduction abnormalities
Non-genetic causes (phenocopies)			
	Inflammatory		
	Post-acute or subacute/chronic viral myocarditis	ALVC	
	Cardiac sarcoidosis (chronic granulomatous myocarditis)	ALVC-BIV-ARVC	Multiorgan involvement. Conduction abnormalities (bundle branch block, bifascicular block and AV block)
	Auto-immune multisystem diseases (systemic lupus erythematous; polymyositis /dermatomyositis; scleroderma)	ALVC	Multiorgan involvement. Conduction abnormalities.Vasculitis.
	Parasitic infectious		
	Chagas disease	ARVC-BIV-ALVC	
Unknown cause			
	Idiopathic	ALVC-BIV-ARVC	

* Genes with limited evidence of ACM causality using the Clinical Genome Resource approach to gene-disease curation [56].

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Key epidemiological metrics in adults and children for the different cardiomyopathy phenotypes

Cardiomyopathy phenotype	Adults	Children
HCM	Prevalence: 0.2%	Childhood incidence: 0.002–0.005% Childhood prevalence: 0.029%
DCM	Prevalence: 0.036–0.400%	Childhood incidence: 0.003–0.006% Childhood prevalence: 0.026% Infantile incidence: 0.038–0.046%
NDLVC	To be determined	To be determined
ARVC	Prevalence: 0.078%	Very rare in infancy and early childhood; to be determined in older children and adolescents
RCM	Rare	Childhood incidence: 0.0003%

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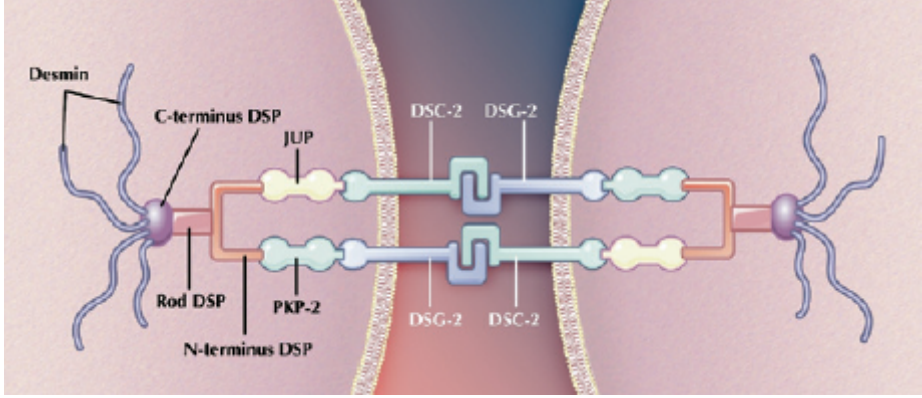
Genetic testing



- The genes underlying ARVC mainly encode proteins of the cardiac desmosome:

  plakophilin-2 (*PKP2*), desmoplakin (*DSP*), desmoglein-2 (*DSG2*), desmocollin-2 (*DSC2*), and plakoglobin (*JUP*).

- In addition to desmosomal genes, P/LP variants have also been described in other genes, including *DES*, *TMEM43* and *PLN*.
- Pathogenic or likely pathogenic variants can be identified in up to 60% of patients with a diagnosis of ARVC.
- The pattern of inheritance in the majority of ARVC families is autosomal dominant.
- The penetrance of the disease in genetic carriers is age, gender, and physical activity dependent.

Examples of signs and symptoms that should raise the suspicion of specific aetiologies, grouped according to cardiomyopathy phenotype (5)

Finding	Cardiomyopathy phenotype				
	HCM	DCM	NDLVC	ARVC	RCM
Angiokeratomata	Anderson–Fabry disease	-	-	-	-
Pigmentation of skin and scars	-	Haemo-chromatosis	-	-	-
Palmoplantar keratoderma and woolly hair	-	Carvajal syndrome	-	Naxos and Carvajal syndromes	-
	-	*DSP* variants	*DSP* variants	*DSP* variants	-

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Examples of electrocardiographic features that should raise the suspicion of specific aetiologies, grouped according to cardiomyopathy phenotype (3)

Cardiomyopathy phenotype	Finding	Specific diseases to be considered
DCM (continued)	Posterolateral infarction pattern	Dystrophinopathy Limb-girdle muscular dystrophy Sarcoidosis
	Extremely low QRS amplitude	*PLN* variant
NDLVC	AV block	Laminopathy Desminopathy
	Extremely low QRS amplitude	*PLN* variant
	Low QRS voltage + atypical RBBB	Desmosomal variants
ARVC	T wave inversion V1-V3 + terminal activation delay +/- low right ventricular voltages +/- atypical RBBB	-
RCM	AV block	Desminopathy Amyloidosis

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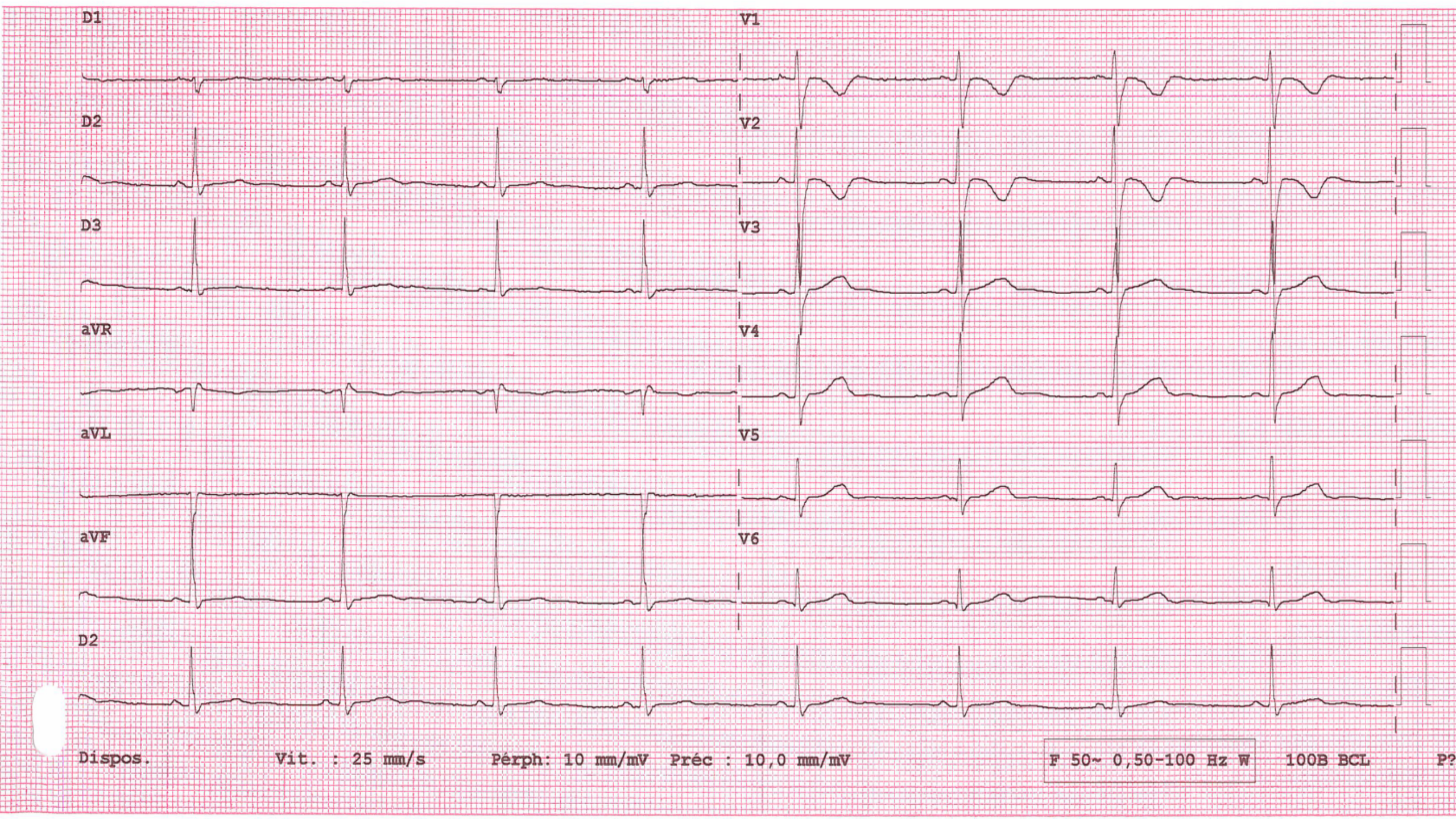
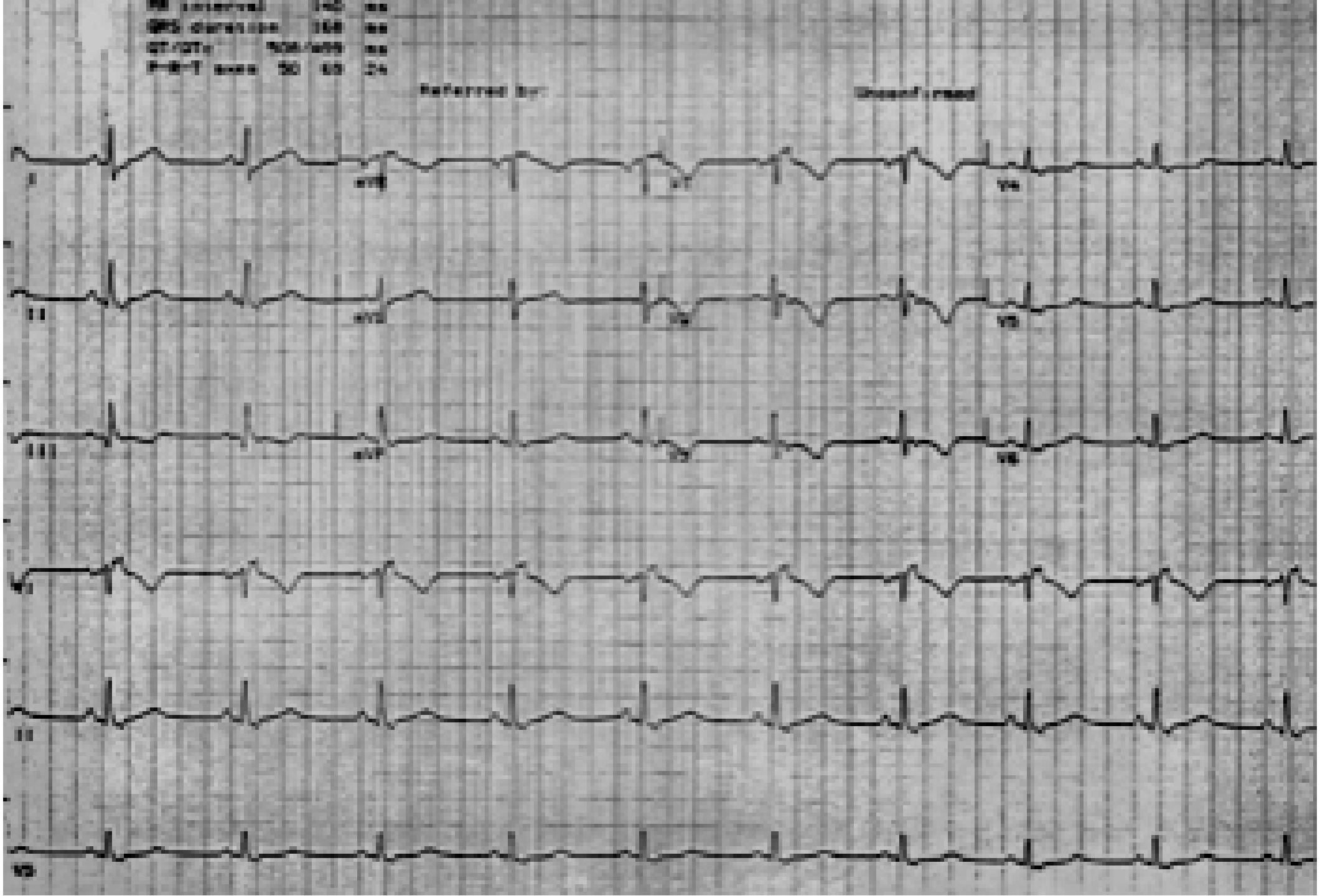
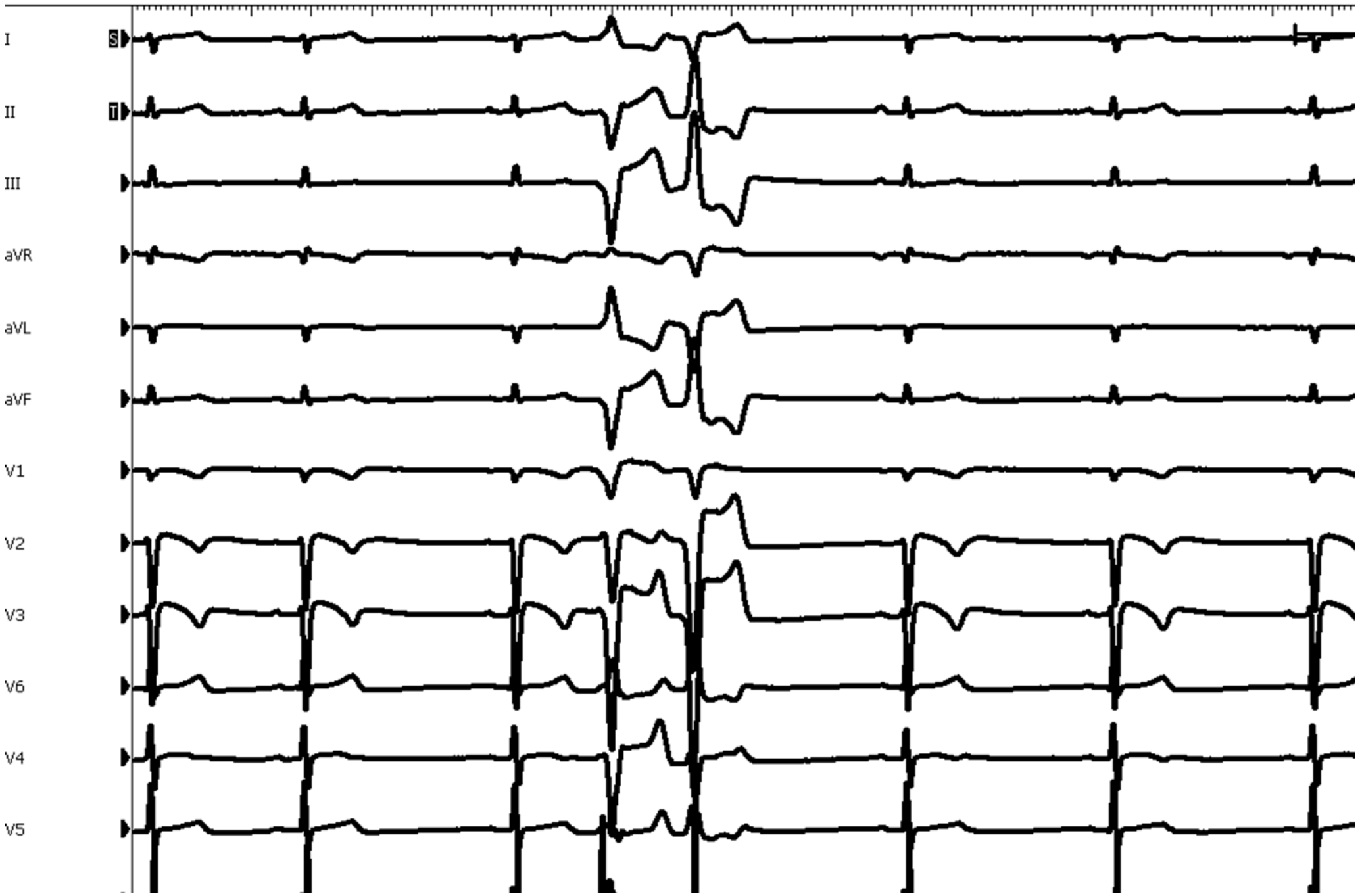

D1
V1
D2
V2
D3
V3
aVR
V4
aVL
V5
aVF
V6
D2
Dispos.
Vit. : 25 mm/s
Périph: 10 mm/mV
Préc : 10,0 mm/mV
F 50~ 0,50-100 Hz W
100B BCL
P?




I
II
III
aVR
aVL
aVF
V1
V2
V3
V6
V4
V5

First-level (to be performed in each patient) and second-level (to be performed in selected patients following specialist evaluation to identify specific aetiologies) laboratory tests, grouped by cardiomyopathy phenotype (1)

Level	HCM	DCM	NDLVC	ARVC	RCM
First	CK Liver function NT-proBNP Proteinuria Renal function Troponin	Calcium CK Ferritin Full blood count Liver function NT-proBNP Phosphate Proteinuria Renal function Serum iron Thyroid function Troponin Vitamin D (children)	Calcium CK C-reactive protein Full blood count Liver function NT-proBNP Phosphate Proteinuria Renal function Troponin	C-reactive protein Liver function NT-proBNP Renal function Troponin	CK Ferritin Full blood count Liver function NT-proBNP Proteinuria Renal function Serum angiotensin-converting enzyme Serum iron Troponin Urine and plasma protein immunofixation, free light chains

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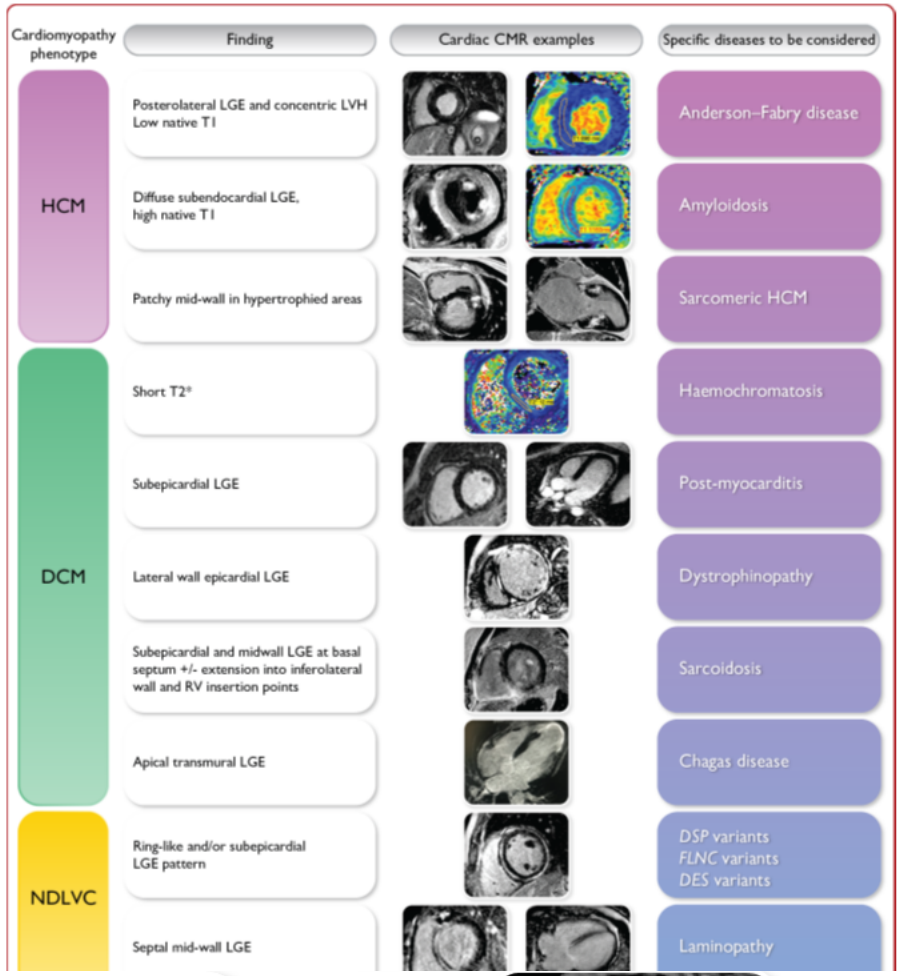
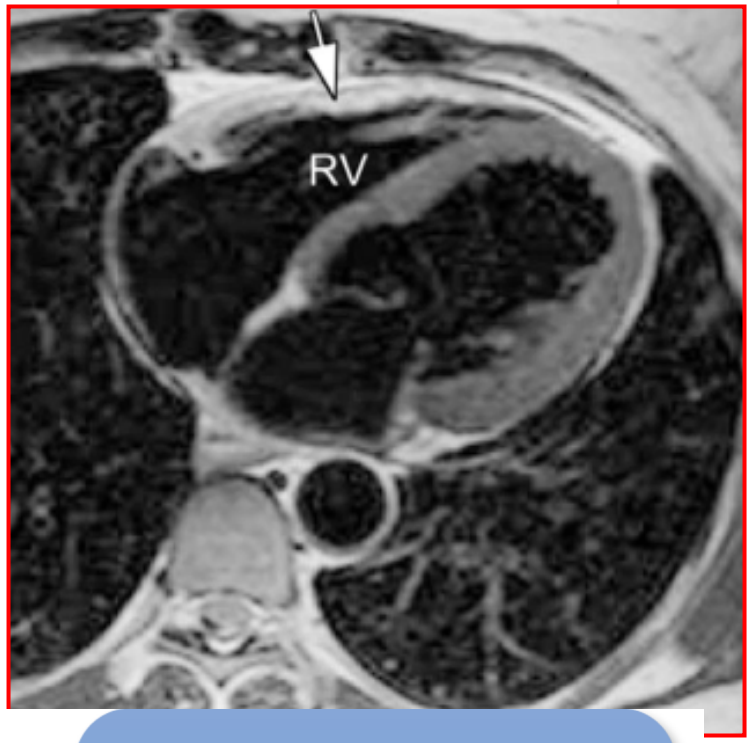
Figure 7

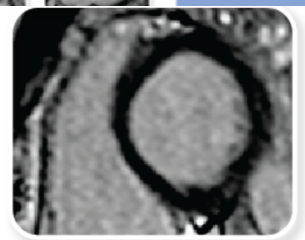
Examples of cardiac magnetic resonance imaging tissue characterization features that should raise the suspicion of specific aetiologies, grouped according to cardiomyopathy phenotype





ARVC

Fat and LGE (transmural RV plus sub-epicardial-midmural LV free wall)



Desmosomal variants

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Frequently encountered actionable results on multimodality imaging

Parameter/Finding	Action
RWMAs on echocardiography or CMR	Raise suspicion of concomitant CAD, myocarditis, ARVC, NDLVC, or sarcoidosis
Systolic impairment on echocardiography or CMR	Assessment of risk in DCM, NDLVC, and ARVC; evaluation of treatment efficacy
Measurement of the wall thickness on echocardiography or CMR	Diagnosis of HCM (when echocardiography is inconclusive); risk stratification in HCM
Diastolic dysfunction on echocardiography	Explain symptoms; evaluation of treatment efficacy
Left atrial size on echocardiography	SCD risk prediction in HCM; systematic screening for AF in case of left atrial enlargement
LVOTO in HCM on resting/exercise echocardiography	Explain symptoms; guide management
Non-invasive evaluation of pulmonary pressures	Explain symptoms; guide management
Tissue characterization on CMR	Diagnosis; risk assessment
Inflammation on CMR or 18F-FDG-PET	Diagnosis; evaluation of treatment efficacy in inflammatory cardiomyopathies

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Recommendations for cardiac magnetic resonance indication in patients with cardiomyopathy

Recommendations	Class	Level
Contrast-enhanced CMR is recommended in patients with cardiomyopathy at initial evaluation.	I	B
Contrast-enhanced CMR should be considered in patients with cardiomyopathy during follow-up to monitor disease progression and aid risk stratification and management.	IIa	C
Contrast-enhanced CMR should be considered for the serial follow-up and assessment of therapeutic response in patients with cardiac amyloidosis, Anderson–Fabry disease, sarcoidosis, inflammatory cardiomyopathies, and haemochromatosis with cardiac involvement.	IIa	C
In families with cardiomyopathy in which a disease-causing variant has been identified, contrast-enhanced CMR should be considered in genotype-positive/phenotype-negative family members to aid diagnosis and detect early disease.	IIa	B
In cases of familial cardiomyopathy without a genetic diagnosis, contrast-enhanced CMR may be considered in phenotype-negative family members to aid diagnosis and detect early disease.	IIb	C

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Atrial fibrillation burden and management in cardiomyopathies (4)

Condition	AF epidemiology		AF management			
	Prevalence	Annual incidence	Anticoagulation	Long-term rate control	Long-term rhythm control	
ARVC	9–30%	2.1–2.8%	According to cardioembolic risk (always if HF or reduced LVEF)	Beta-blockers (preferred) Verapamil or diltiazem (only if LVEF ≥40%) AV node ablation + CRT or physiological pacing	Rhythm control preferred in case of symptoms or/and heart failure or LV dysfunction	
					Flecainide (associated with beta-blockers) Amiodarone, sotalol	Ablation

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Recommendations for management of atrial fibrillation and atrial flutter in patients with cardiomyopathy (1)

Recommendations	Class	Level
Anticoagulation		
Oral anticoagulation in order to reduce the risk of stroke and thromboembolic events is recommended in all patients with HCM or cardiac amyloidosis and AF or atrial flutter (unless contraindicated).	I	B
Oral anticoagulation to reduce the risk of stroke and thromboembolic events is recommended in patients with DCM, NDLVC, or ARVC, and AF or atrial flutter with a CHA_2DS_2-VASc score ≥2 in men or ≥3 in women.	I	B
Oral anticoagulation to reduce the risk of stroke and thromboembolic events should be considered in patients with RCM and AF or atrial flutter (unless contraindicated).	IIa	C
Oral anticoagulation to reduce the risk of stroke and thromboembolic events should be considered in patients with DCM, NDLVC, or ARVC, and AF or atrial flutter with a CHA_2DS_2-VASc score of 1 in men or of 2 in women.	IIa	B

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Recommendations for routine follow-up of patients with cardiomyopathy

Recommendations	Class	Level
It is recommended that all clinically stable patients with cardiomyopathy undergo routine follow-up using a multiparametric approach that includes ECG and echocardiography every 1 to 2 years.	I	C
Clinical evaluation with ECG and multimodality imaging is recommended in patients with cardiomyopathy whenever there is a substantial or unexpected change in symptoms.	I	C

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Recommendation for resting and ambulatory electrocardiogram monitoring in patients with arrhythmogenic right ventricular cardiomyopathy

Recommendation	Class	Level
Annual ambulatory ECG monitoring is recommended in patients with ARVC to aid in diagnosis, management, and risk stratification.	I	C

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Recommendations for the antiarrhythmic management of patients with arrhythmogenic right ventricular cardiomyopathy

Recommendations	Class	Level
Beta-blocker therapy is recommended in ARVC patients with VE, NSVT, and VT.	I	C
Amiodarone should be considered when regular beta-blocker therapy fails to control arrhythmia-related symptoms in patients with ARVC.	IIa	C
Flecainide in addition to beta-blockers should be considered when single agent treatment has failed to control arrhythmia-related symptoms in patients with ARVC.	IIa	C
Catheter ablation with availability for epicardial approach guided by 3D electroanatomical mapping of VT should be considered in ARVC patients with incessant VT or frequent appropriate ICD interventions for VT despite pharmacological therapy with beta-blockers.	IIa	C

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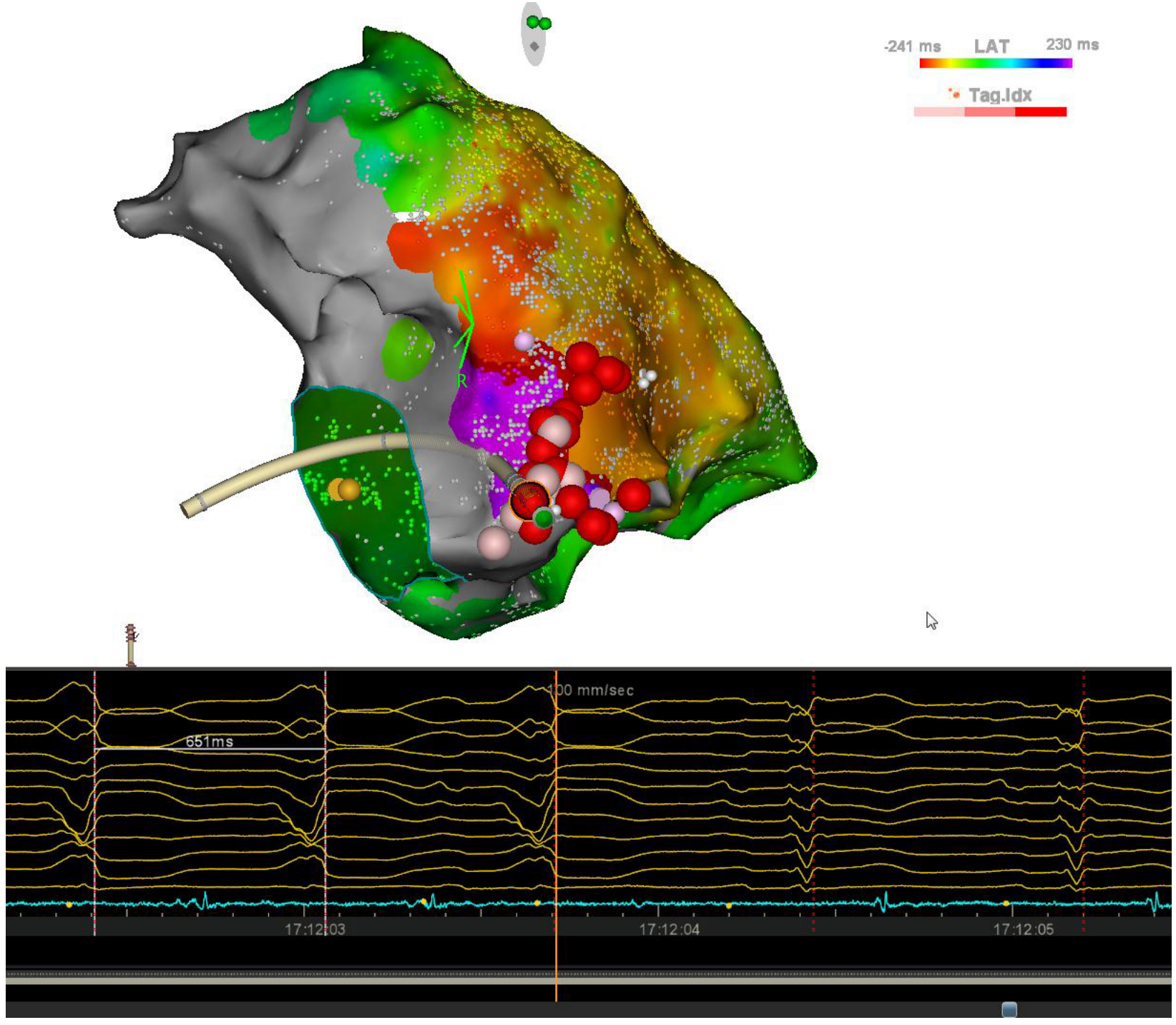
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Recommendations for sudden cardiac death prevention in patients with arrhythmogenic right ventricular cardiomyopathy

Recommendations	Class	Level
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.	I	A
An ICD should be considered in ARVC patients who have suffered a haemodynamically tolerated VT.	IIa	B
Primary prevention		
High-risk features should be considered to aid individualized decision-making for ICD implantation in patients with ARVC.	IIa	B
The updated 2019 ARVC risk calculator should be considered to aid individualized decision-making for ICD implantation in patients with ARVC.	IIa	B

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ARVC Risk Calculator

ARVC Risk Calculator v3.0

The Arrhythmogenic Right Ventricular Cardiomyopathies (ARVC) Risk Calculator estimates the risk of ventricular arrhythmias (VA) within 5 years for patients with a *definite* ARVC diagnosis. Please read all definitions below carefully, as well as our general disclaimer, to avoid invalid interpretation of results.

This calculator provides 1-, 2- or 5-year risk estimations for:

Primary prevention patients only, i.e. those without prior sustained VA:

– Risk of the *first* sustained VA (including all types)*

– Risk of the *first* sustained VA, adjusted for programmed ventricular stimulation (PVS) results

All patients with definite ARVC:

– Risk of *fast* sustained VA (only ventricular tachycardia (VT) >250bpm; ventricular fibrillation/flutter (VF); sudden cardiac arrest/death (SCA/SCD))

*External validation studies available

NB: ALL FIELDS BELOW must be filled in, any missing value will render the calculation invalid.

Structured Graphical Abstract

Jordà P, Eur Heart J. 2022

Key Question

Does the published arrhythmogenic right ventricular cardiomyopathy (ARVC) risk calculator (ARVCrisk.com) adequately predicting sustained ventricular arrhythmia (VA) in a distinct geographically diverse cohort?.

Key Finding

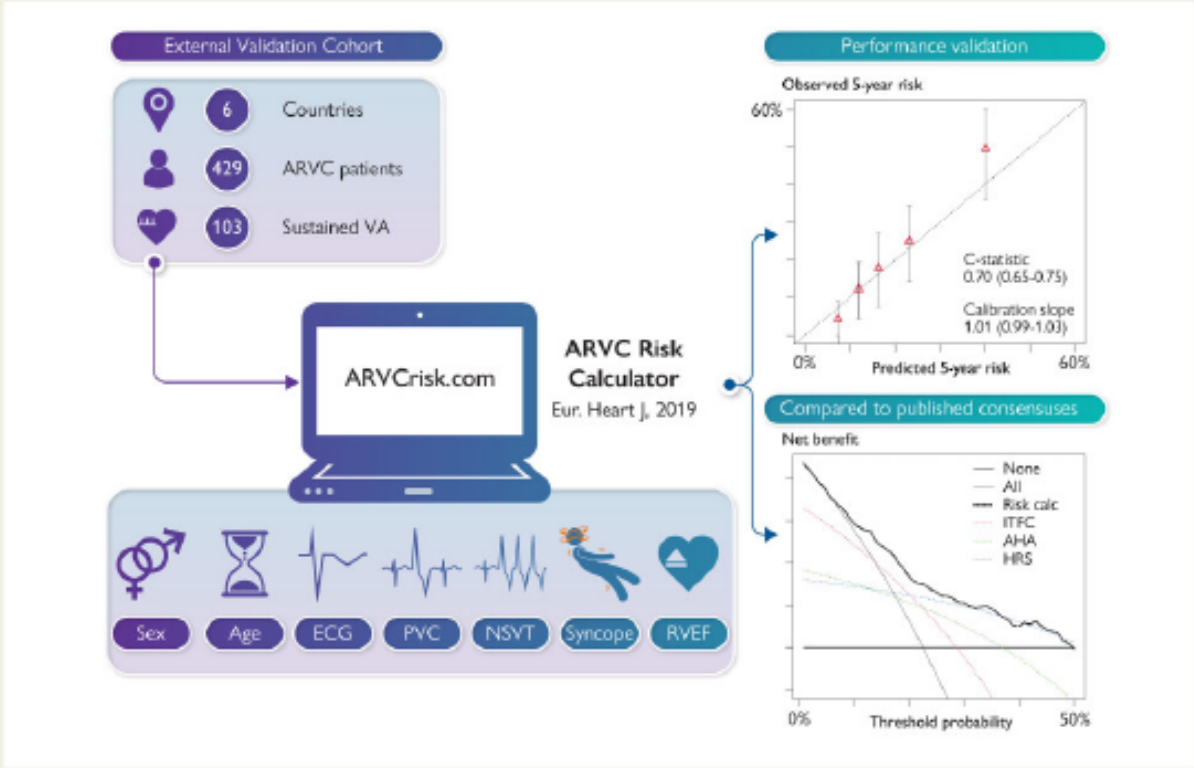
The ARVC risk calculator:

1) Predicted adequately with good discrimination and calibration.

2) Performed better than other consensus-based implantable cardioverter defibrillator (ICD) implantation algorithms.

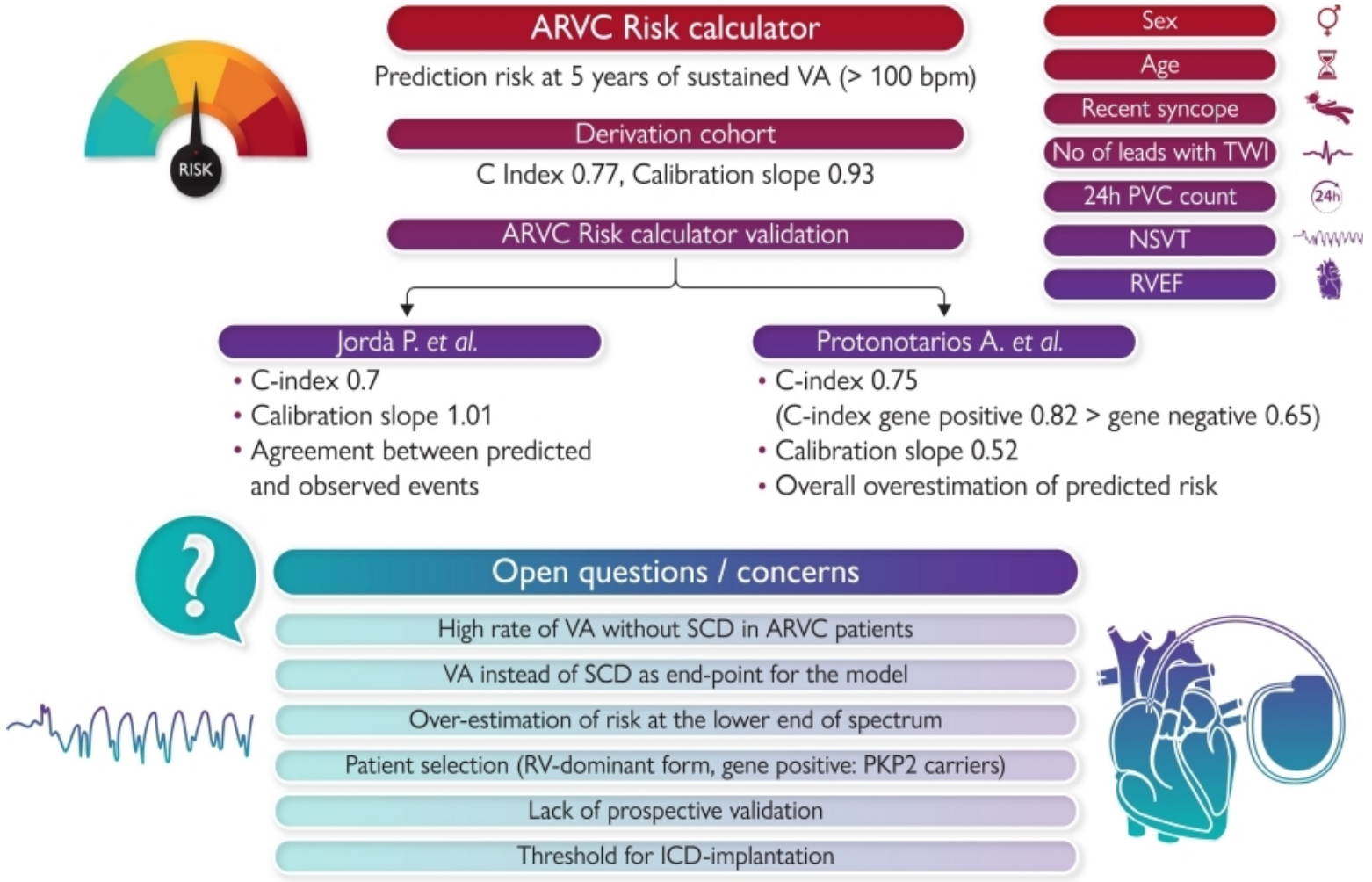
Take Home Message

The ARVC risk calculator provides reliable information and can facilitate shared decision-making regarding ICD implantation in primary prevention ARVC.



Validation of the arrhythmogenic right ventricular cardiomyopathy (ARVC) risk calculator in a distinct cohort. AHA, American Heart Association; ECG, electrocardiogram; HRS, Heart Rhythm Society; ICD, implantable cardioverter-defibrillator; ITFC, International Task Force Criteria; NSVT, non-sustained ventricular tachycardia; PVC, premature ventricular complex; VA, ventricular arrhythmia.

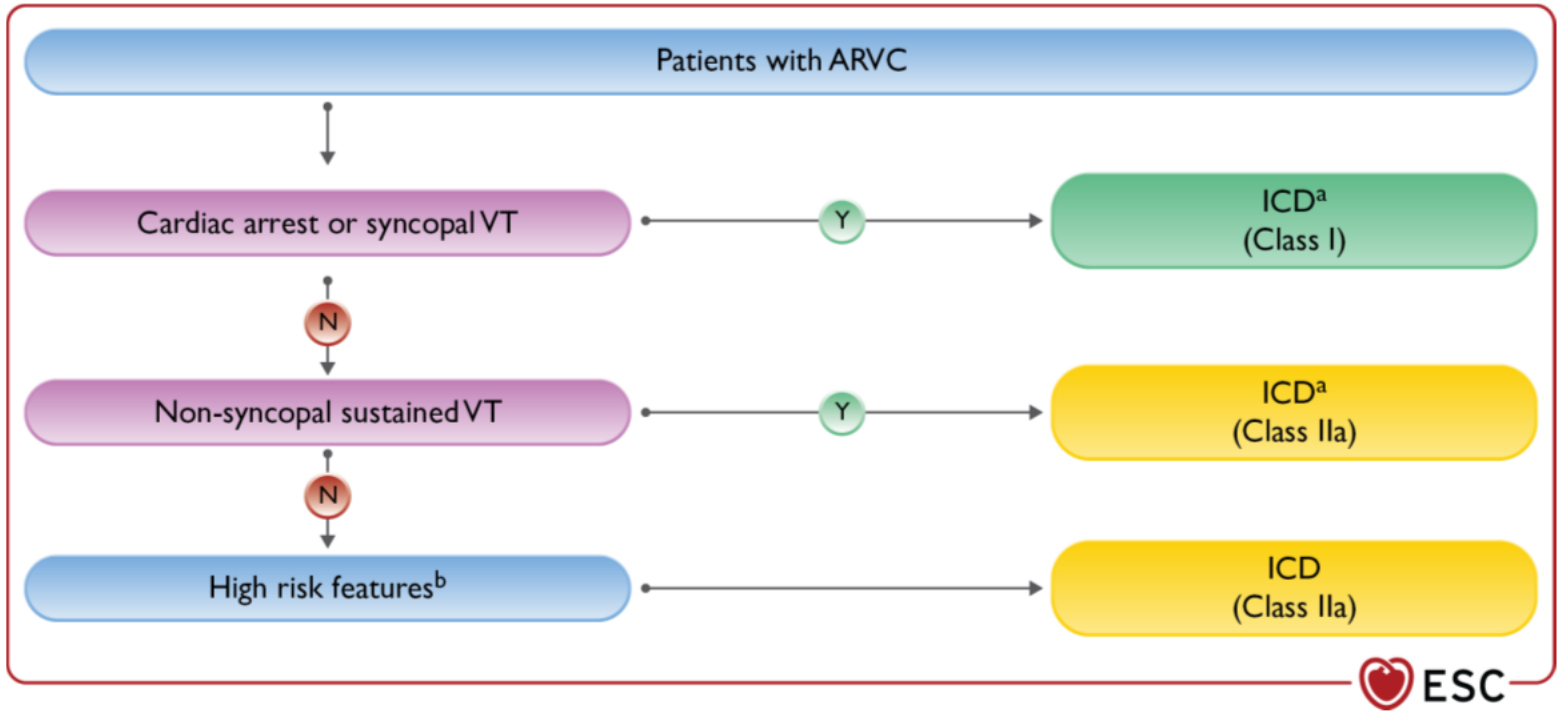
Graphical Abstract NSVT: non-sustained ventricular tachycardia; PVC: premature ventricular contractions; RV: right ventricle; ...



Eur Heart J, Volume 43, Issue 32, 21 August 2022, Pages 3068–3070, https://doi.org/10.1093/eurheartj/ehac324

Figure 18

Algorithm to approach implantable cardioverter defibrillator decision-making in patients with arrhythmogenic right ventricular cardiomyopathy



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Exercise recommendations for patients with cardiomyopathy (2)

Recommendations	Class	Level
HCM (continued)		
High-intensity exercise, including competitive sport, is not recommended in high-risk individuals and in individuals with left ventricular outflow tract obstruction and exercise-induced complex ventricular arrhythmias.	III	C
ARVC		
Avoidance of high-intensity exercise, including competitive sport, may be considered in genotype-positive/phenotype-negative individuals in families with ARVC.	IIb	C
Moderate- and/or high-intensity exercise, including competitive sport, is not recommended in individuals with ARVC.	III	B
DCM and NDLVC		
Moderate- and high-intensity exercise should be considered in individuals who are gene positive and phenotype-negative (with the exception of pathogenic variants in *LMNA* and *TMEM43*) who seek to do so.	IIa	C

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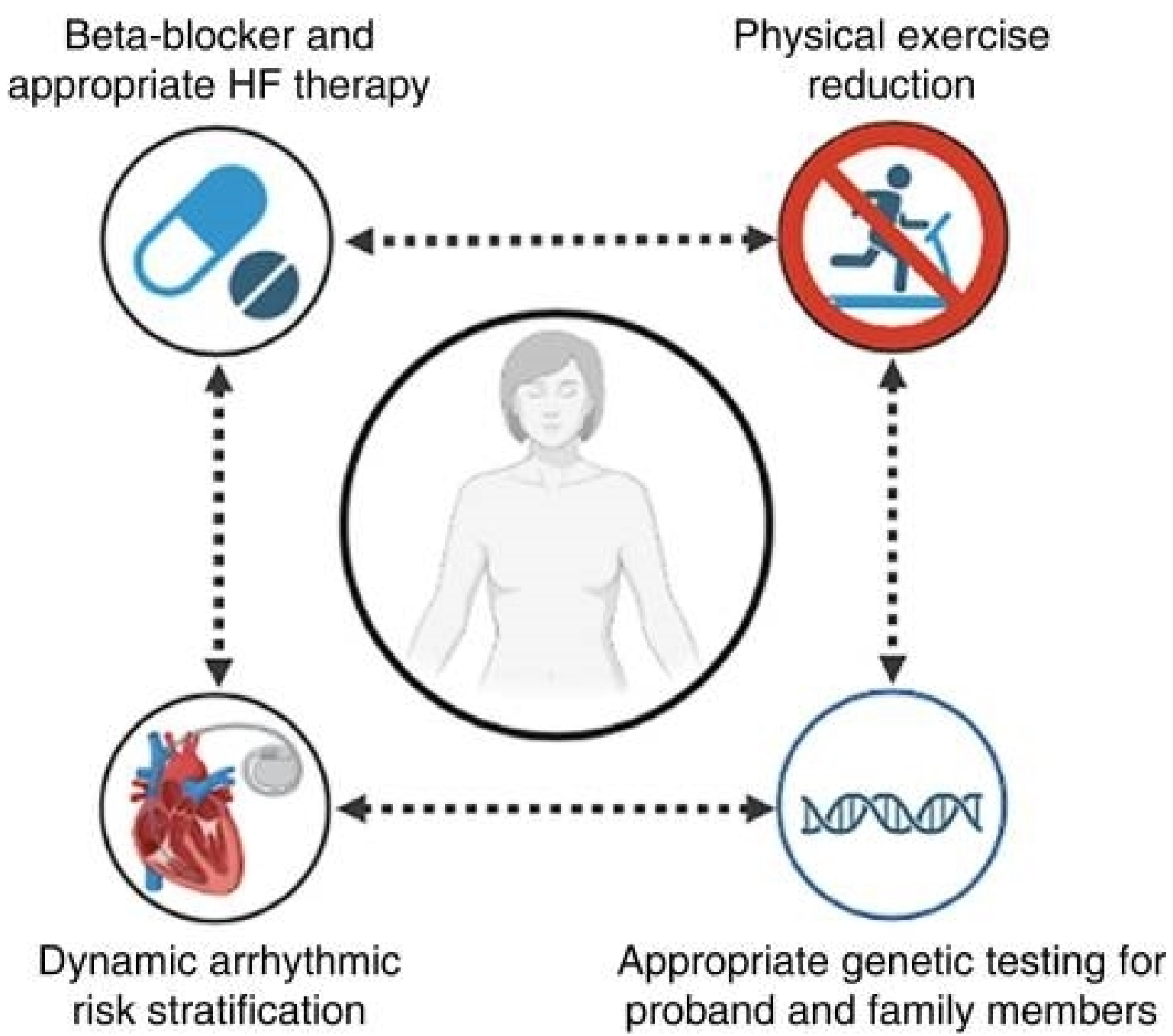

Gasperetti A, Europace 2023