ST-akademin VT 2022
Kardiogenetik

ARVC

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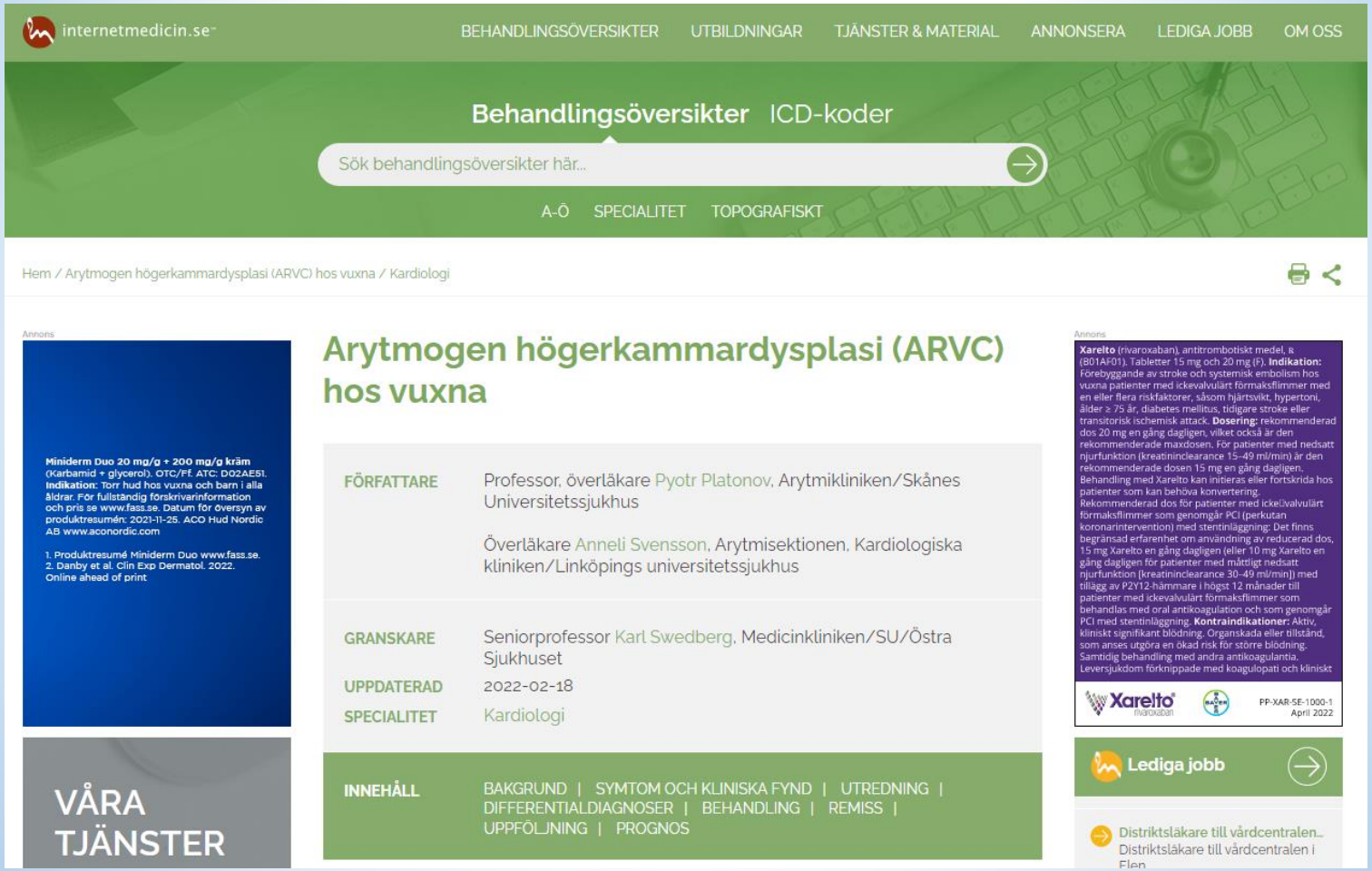
KARDIOLOGISKA KLINIKEN

Universitetssjukhuset i Linköping

Egenreklam…☺



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Bakgrund

- Först beskriven 1982, ärftlig hjärtmuskelsjukdom, kan ge rytmrubbningar och hjärtsvikt
- 1:5000 – 1:2000
- Mikroskopiska skador i myokardiet läker med inlagring av fett och bindväv
- Mycket brett fenotypiskt spektrum
- AC eller ACM – arrhythmogenic cardiomyopathy
- Diagnostiska kriterier presenterades 1994, de reviderade Task Force Criteria 2010 (TFC2010)
  - Imaging, vävnadskaraktäristik, depolarisation o repolarisationsstörningar, arytmier, familjehistoria inkl genetiska fynd

ARVC

CENTRAL ILLUSTRATION: Clinical Approach to Arrhythmogenic Right Ventricular Cardiomyopathy

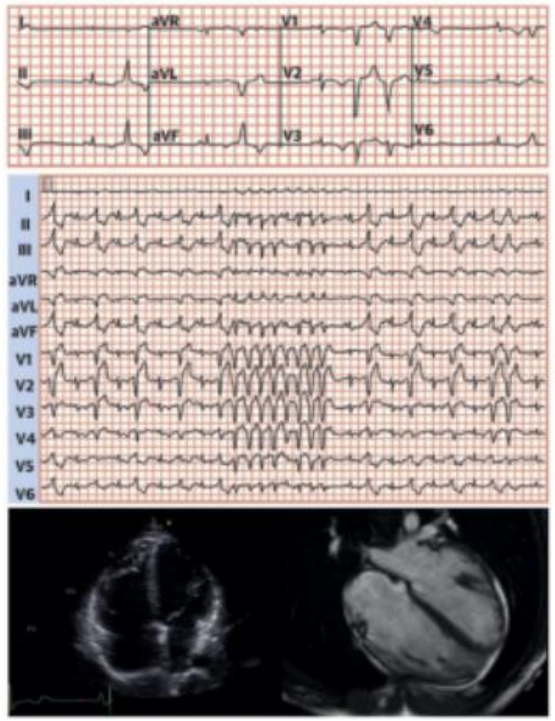
Assessment

Evaluation

- History and pedigree
- ECG, SAECG, Holter
- Echo, MRI
- Genetic testing

Diagnosis

- 2 major TFC
- 1 major + 2 minor TFC
- 4 minor TFC



Risk Stratification

Established Markers

- Cardiac arrest
- VT
- Age
- Male sex
- Cardiogenic syncope
- Extent of TWI
- PVC count
- Reduced LV/RV

Treatment

Conservative

- Exercise restriction

Pharmacological

- β-blockers
- Sotalol, flecainide, amiodarone

Interventional

- ICD
- Ablation

Krahn AD, et al. J Am Coll Cardiol EP. 2022;8(4):533-553.

ARVC utredning

- Bilddiagnostik
  - Ekokardiografi (med mer fokus på höger kammare än vid standardundersökning)
  - MR hjärta
- Vävnadsdiagnostik
  - Hjärtmuskelbiopsi utförs numera sällan på ren diagnostisk indikation
- Repolarisations- och depolarisationsstörningar
  - Vilo-EKG
  - SAECG (*Signal-averaged electrocardiography*, ”sena potentialer”, högfrekvens-EKG)
- Arytmier
  - Holter-EKG/telemetri (för att räkna antal VES per 24 timmar och eventuellt dokumentera icke-ihållande VT)
  - Cykelarbetsprov (ansträngningsutlöst arytmi?)
- Familjeanamnes inklusive genetisk analys

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%
II. Tissue characterization of wall	
Major	
• Fibrofatty replacement of myocardium on endomyocardial biopsy	• Residual myocytes <60% by morphometric analysis (or <50% if estimated), with fibrous replacement of the RV free wall myocardium in ≥1 sample, with or without fatty replacement of tissue on endomyocardial biopsy
Minor	• Residual myocytes 60% to 75% by morphometric analysis (or 50% to 65% if estimated), with fibrous replacement of the RV free wall myocardium in ≥1 sample, with or without fatty replacement of tissue on endomyocardial biopsy
III. Repolarization abnormalities	
Major	• Inverted T waves in right precordial leads (V_1, V_2, and V_3) or beyond in individuals >14 years of age (in the absence of complete right bundle-branch block QRS ≥120 ms)
Minor	
• Inverted T waves in right precordial leads (V_2 and V_3) (people age >12 years, in absence of right bundle-branch block)	• Inverted T waves in leads V_1 and V_2 in individuals >14 years of age (in the absence of complete right bundle-branch block) or in V_4, V_5, or V_6 • Inverted T waves in leads V_1, V_2, V_3, and V_4 in individuals >14 years of age in the presence of complete right bundle-branch block

Continued

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 • Terminal activation duration of QRS ≥55 ms measured from the nadir of the S wave to the end of the QRS, including R′, in V_1, V_2, or V_3, in the absence of complete right bundle-branch block
V. Arrhythmias	
Major	
	• Nonsustained or sustained ventricular tachycardia of left bundle-branch morphology with superior axis (negative or indeterminate QRS in leads II, III, and aVF and positive in lead aVL)
Minor	
• Left bundle-branch block-type ventricular tachycardia (sustained and nonsustained) (ECG, Holter, exercise) • Frequent ventricular extrasystoles (>1000 per 24 hours) (Holter)	• Nonsustained or sustained ventricular tachycardia of RV outflow configuration, left bundle-branch block morphology with inferior axis (positive QRS in leads II, III, and aVF and negative in lead aVL) or of unknown axis • > 500 ventricular extrasystoles per 24 hours (Holter)
VI. Family history	
Major	
• Familial disease confirmed at necropsy or surgery	• ARVC/D confirmed 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‡ 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 • Familial history (clinical diagnosis based on present criteria)	• 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 • 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

PLAX indicates parasternal long-axis view; RVOT, RV outflow tract; BSA, body surface area; PSAX, parasternal short-axis view; aVF, augmented voltage unipolar left foot lead; and aVL, augmented voltage unipolar left arm lead.
Diagnostic terminology for original criteria: This diagnosis is fulfilled by the presence of 2 major, or 1 major plus 2 minor criteria or 4 minor criteria from different groups.
Diagnostic terminology for revised criteria: definite diagnosis: 2 major or 1 major and 2 minor criteria or 4 minor from different categories; borderline: 1 major and 1 minor or 3 minor criteria from different categories; possible: 1 major or 2 minor criteria from different categories.
*Hypokinesis is not included in this or subsequent definitions of RV regional wall motion abnormalities for the proposed modified criteria.
‡A pathogenic mutation is a DNA alteration associated with ARVC/D that alters or is expected to alter the encoded protein, is unobserved or rare in a large non-ARVC/D control population, and either alters or is predicted to alter the structure or function of the protein or has demonstrated linkage to the disease phenotype in a conclusive pedigree.

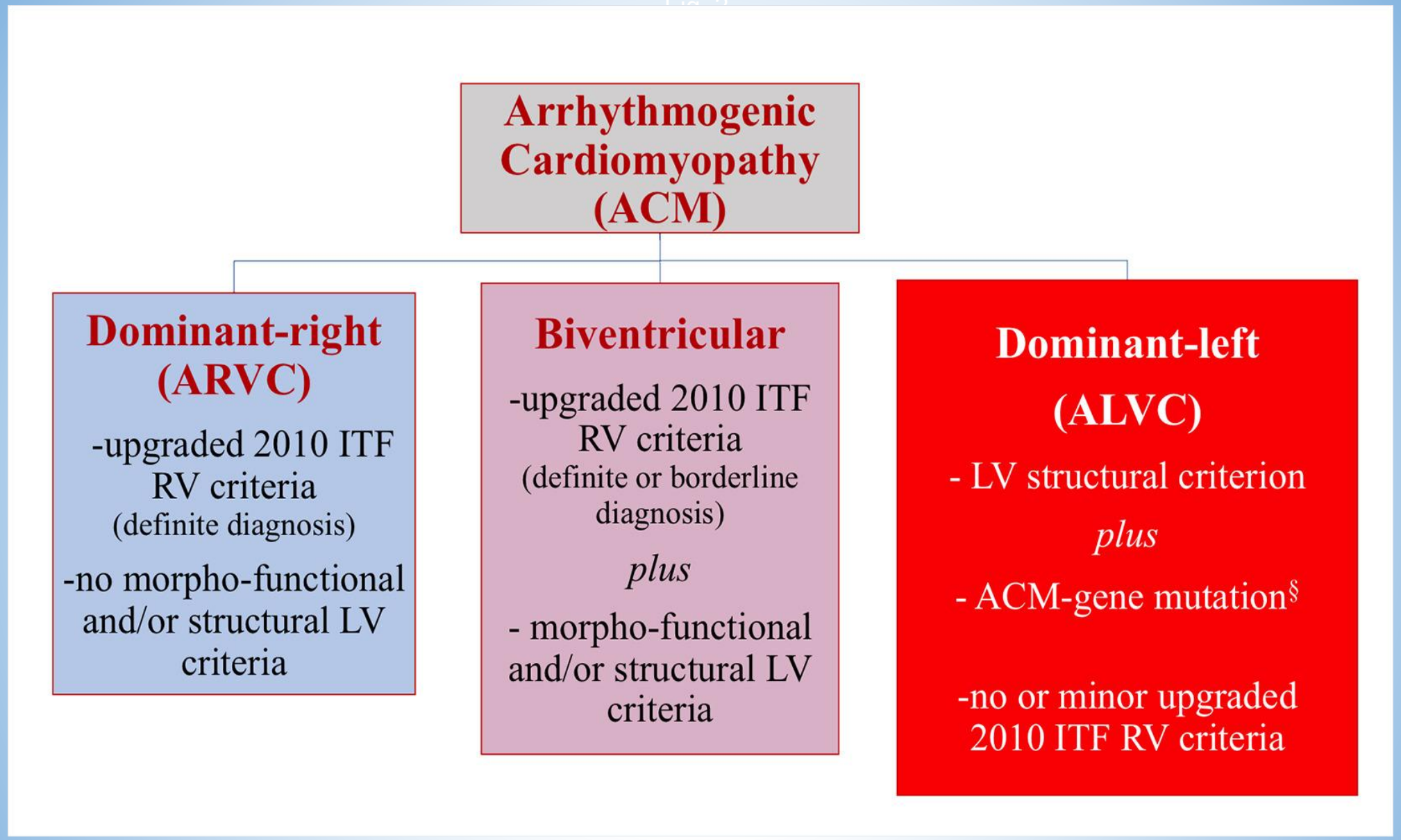
”Diagnosis of arrhythmogenic cardiomyopathy: The Padua criteria”

Corrado et al, 2020

Table 1
"Padua criteria" for diagnosis of Arrhythmogenic Cardiomyopathy.

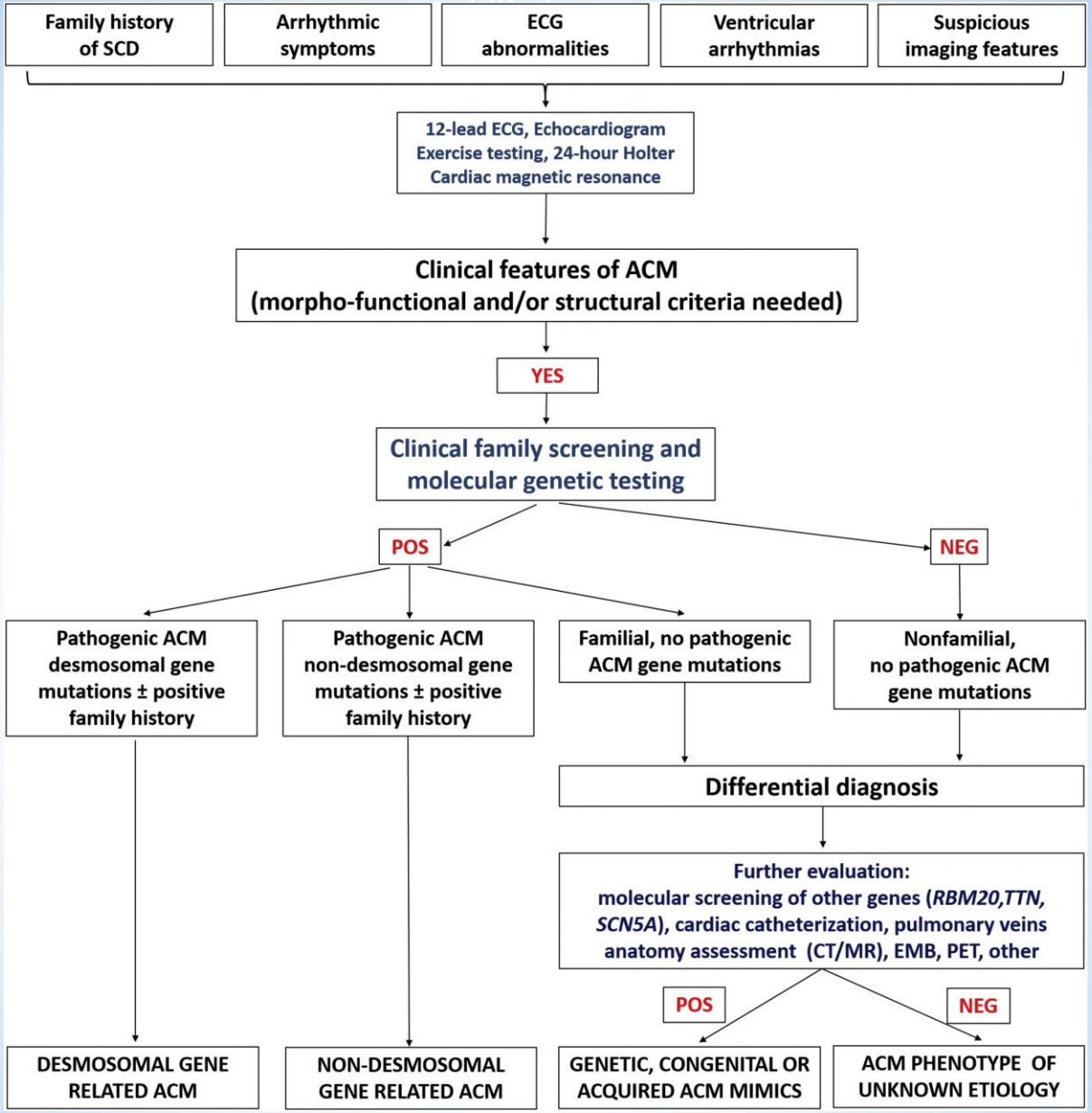
Category	Right ventricle (upgraded 2010 ITF diagnostic criteria)	Left ventricle (new diagnostic criteria)
I. Morpho-functional ventricular abnormalities	*By echocardiography, CMR or angiography:* *Major* • Regional RV akinesia, dyskinesia, or bulging plus one of the following: – global RV dilatation (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	

ACM = arrhythmogenic cardiomyopathy; BSA = body surface area; EDV = end diastolic volume; EF = ejection fraction; ITF = International Task Force; LBBB = left bundle-branch block; LGE = late gadolinium enhancement; LV = left ventricle; RBBB = right bundle-branch block; RV = right ventricle; RVOT = right ventricular outflow tract.



Corrado et al. International Journal of Cardiology 2020 319106-114

ACM diagnos



Corrado et al.
International Journal of Cardiology 2020
319106-114

Diff-diagnoser

Table 1 Most common differential diagnostic considerations for ARVC

Differential diagnosis	Comparison of clinical features
Cardiac sarcoidosis	▶ Similarities with ARVC: focal myocardial lesions, (regional) ventricular dysfunction, arrhythmias and LGE with non-ischaemic pattern. ▶ Contrasting with ARVC: non-familial pattern, atrioventricular conduction delay, extracardiac manifestations and predominant intraventricular septal involvement.
Myocarditis	▶ Similarities with ARVC: non-ischaemic LGE and arrhythmias. ▶ Contrasting with ARVC: history of viral prodromes, imaging findings suggesting myocardial oedema (acute phase) as well as pericardial involvement.
Dilated cardiomyopathy	▶ Similarities with ARVC: familial pattern, phenotype may mimic ARVC/ACM with LV involvement. ▶ Contrasting with ARVC: ventricular arrhythmias predominantly in context of impaired ventricular structure/function, usually preceded by heart failure.
Uhl's anomaly	▶ Similarities with ARVC: loss of RV myocardium and RV dilatation. ▶ Contrasting with ARVC: non-familial, RV birth defect, deficiency of myocardium appearing as 'parchment', symptoms early childhood and primarily heart failure.
Brugada syndrome	▶ Similarities with ARVC: ventricular arrhythmias and pseudo right bundle branch block. ▶ Contrasting with ARVC: ventricular arrhythmias predominantly at rest and structural abnormalities absent.
Athlete's heart	▶ Similarities with ARVC: cardiac remodelling may mimic ARVC and exercise accelerates structural modifications. ▶ Contrasting with ARVC: reversible, balanced biventricular dilatation and hypertrophy, no dysfunction and no regional wall motion abnormalities.
Idiopathic RVOT VT	▶ Similarities with ARVC: VTs with LBBB inferior axis morphology. ▶ Contrasting with ARVC: benign prognosis, curative catheter ablation and structural/ECG abnormalities usually absent.

ACM, arrhythmogenic cardiomyopathy; ARVC, Arrhythmogenic right ventricular cardiomyopathy; LBBB, left bundle branch block; LGE, late gadolinium enhancement; LV, left ventricular; RV, right ventricular; RVOT, RV outflow tract; VT, ventricular tachycardia.

Biopsi

- Biopsi del av TFC2010, men görs sällan primärt
- Fria väggen tunn -> risk för perforation
- Septum “tryggare”, men påverkas sent av ARVC
- “Fläckigt” engagemang

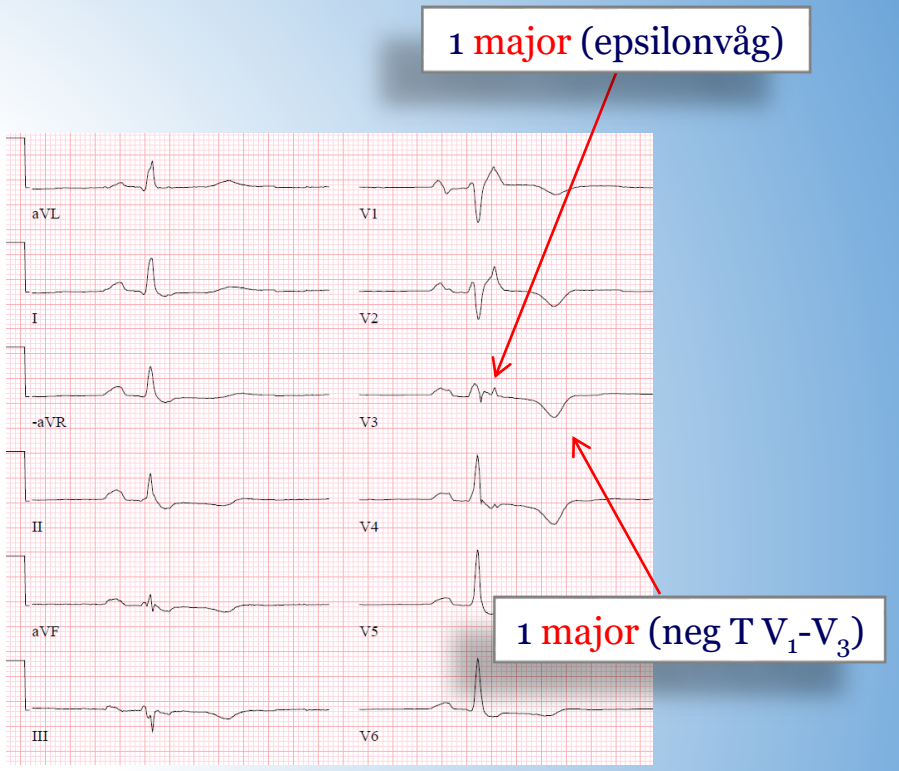
- Som led i diff-diagnostik, helst guidat av elektroanatomisk kartläggning
- Viktigt fortsatt vid obduktion (komplikationsrisken begränsar inte längre)

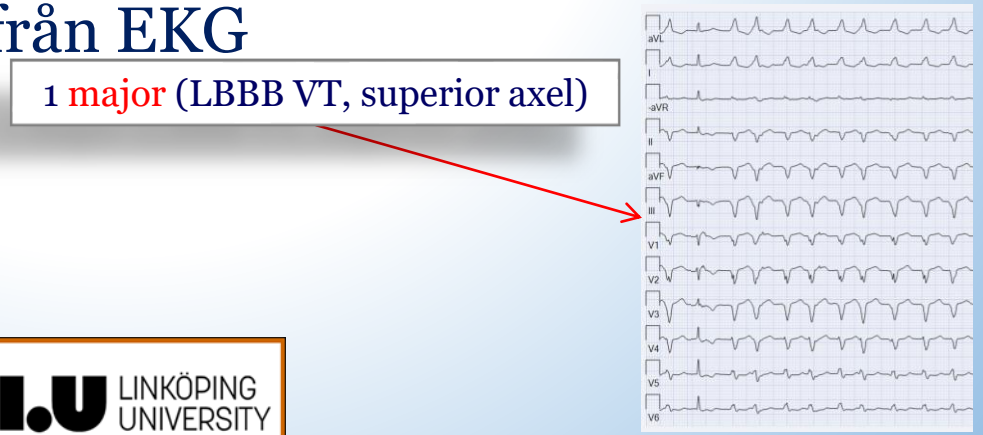
- Utveckling av MR- o CT-tekniker kan förhoppningsvis framgent mäta fibros o fett icke-invasivt

ARVC - ECG

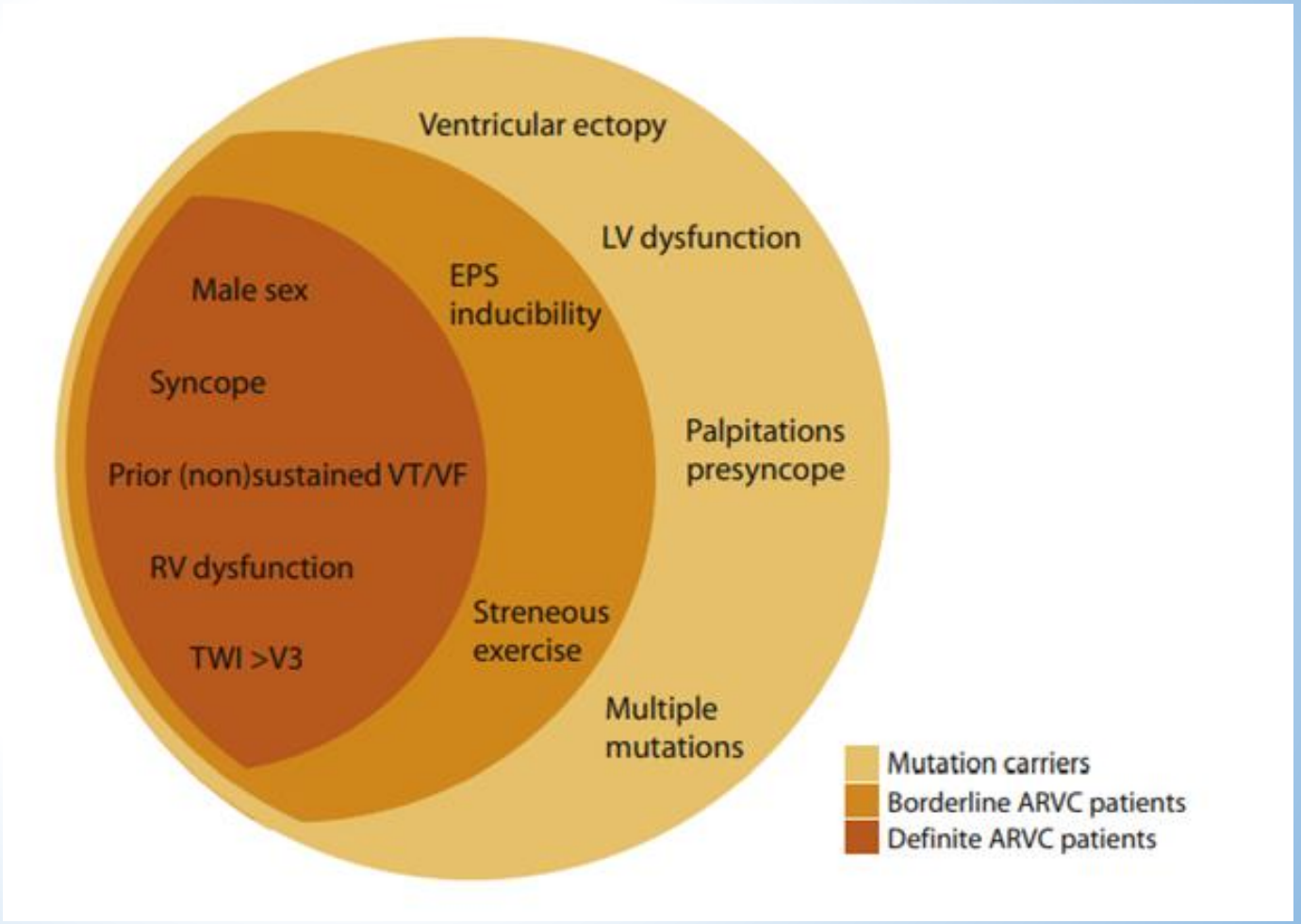
- 12-avlednings-EKG – hörnsten i diagnostiken
- T-vågsnegativitet i avledningarna V1-V3 (TFC2010 major-kriterium)

- Depolarisations- och repolarisationsstörningarna kan variera över tid och även vara reversibla
- Diagnosen kan ställas “bara” utifrån EKG





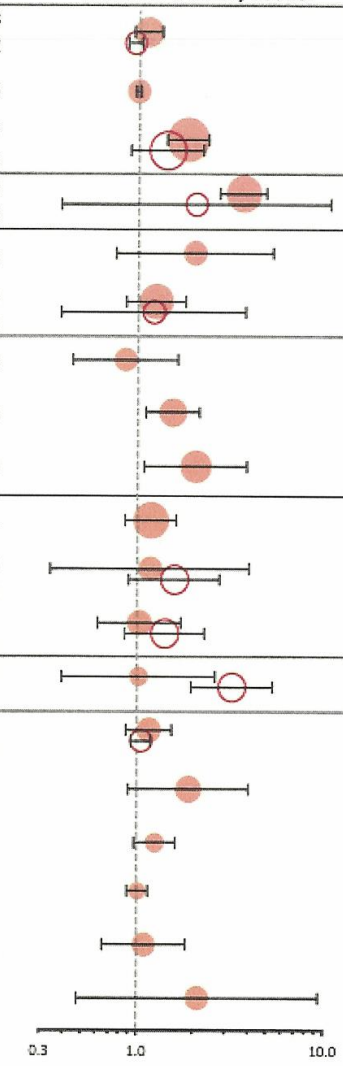
ARVC riskvärdering



Bosman LP, Heart Rhythm 2018

ARVC riskvärdering

Risk factor		Size / events	Studies	Pooled Hazard Ratio, random-effects, 95%CI	p-value	I^2	References
Demographics	Age, 5 yrs increase	180 / 89	3	1.12 [0.94-1.33]	0.196	15.5	S6, S19, S38
		133 / 56	2	0.95 [0.87-1.03]	0.239	0.0	S11, S37
	Age <35 yrs	170 / 58	3	0.99 [0.96-1.02]	0.550	0.0	S11, S22, S23
	Male sex	617 / 194	7	1.83 [1.41-2.37]	<0.001	0.0	S6, S11, S19, S22, S23, S24, S38
		342 / 154	4	1.42 [0.91-2.23]	0.124	18.5	S3, S8, S9, S37
Symptoms	Unexplained syncope	509 / 136	5	3.67 [2.75-4.9]	<0.001	0.0	S6, S11, S22, S24, S38
		147 / 59	2	2.04 [0.39-10.74]	0.401	85.8	S3, S9
Family history	Proband status	293 / 60	2	2.01 [0.76-5.33]	0.159	82.4	S24, S25
	Family SCD <35 yrs	483 / 123	4	1.25 [0.86-1.8]	0.237	0.0	S6, S11, S24, S39
		147 / 59	2	1.21 [0.39-3.8]	0.741	65.0	S3, S9
Arrhythmia	>1000 PVC/24h	299 / 59	2	0.86 [0.45-1.64]	0.640	0.0	S24, S38
	Prior non-sustained VT	405 / 84	3	1.54 [1.10-2.15]	0.011	0.0	S11, S24, S38
	Prior sustained VT/VF	406 / 104	3	2.05 [1.08-3.88]	0.027	54.5	S19, S24, S25
ECG	TWI V1-3	489 / 132	4	1.18 [0.86-1.62]	0.305	0.0	S11, S17, S22, S24
	Epsilon wave	116 / 60	2	1.17 [0.34-4.01]	0.801	59.9	S17, S22
		190 / 91	2	1.58 [0.90-2.77]	0.109	0.0	S3, S37
	SAECG LPs, ≥1 criteria	184 / 64	2	1.03 [0.61-1.72]	0.920	0.0	S6, S11, S17
		190 / 91	2	1.40 [0.86-2.30]	0.177	0.0	S3, S36
EPS	VA inducible at EPS	138 / 37	2	1.02 [0.39-2.64]	0.968	0.0	S11, S37
		209 / 89	3	3.24 [1.95-5.39]	<0.001	0.0	S3, S6, S35
Imaging	LVEF, 5% reduction	182 / 62	4	1.16 [0.87-1.54]	0.306	50.2	S2, S23, S25, S28
		133 / 56	2	1.05 [0.93-1.19]	0.414	0.0	S9, S37
	RVEF, 5% reduction	185 / 74	4	1.89 [0.90-3.99]	0.092	87.1	S2, S6, S28, S37
	RVFAC, 5% reduction	107 / 39	2	1.25 [0.97-1.61]	0.090	1.9	S23, S25
	RVEDV, 5 mL/m2	110 / 30	2	1.01 [0.89-1.15]	0.890	2.5	S2, S25
	TFC minor or major	108 / 58	2	1.09 [0.65-1.84]	0.737	0.0	S19, S23
	TFC major	116 / 60	2	2.12 [0.48-9.41]	0.323	84.6	S17, S22

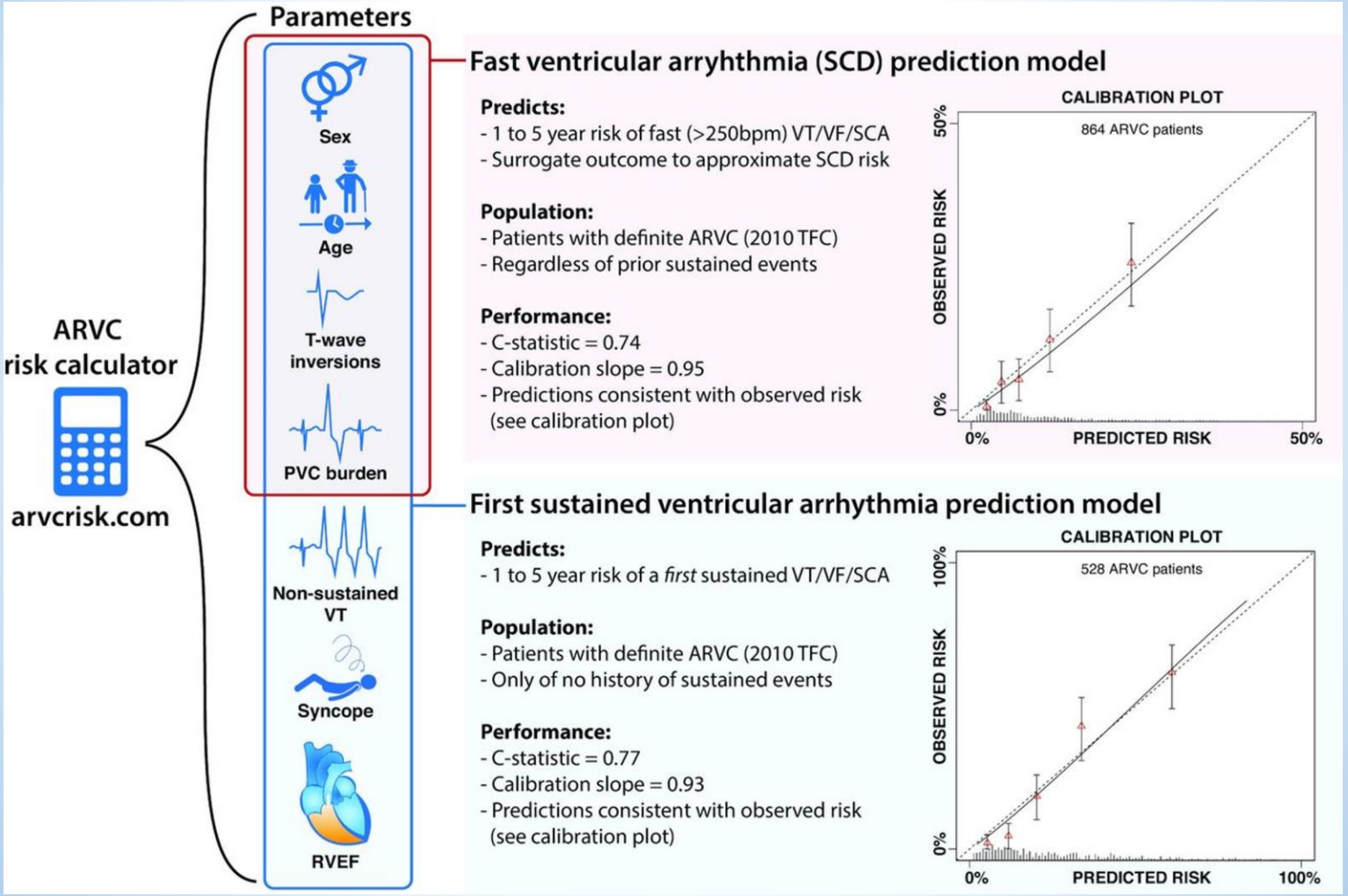


● = cohort with definite ARVC patients only (TFC ≥4) ○ = cohort with at least borderline ARVC patients (TFC ≥3)

Bosman LP, Heart Rhythm 2018

ARVC riskvärdering

arvcrisk.com



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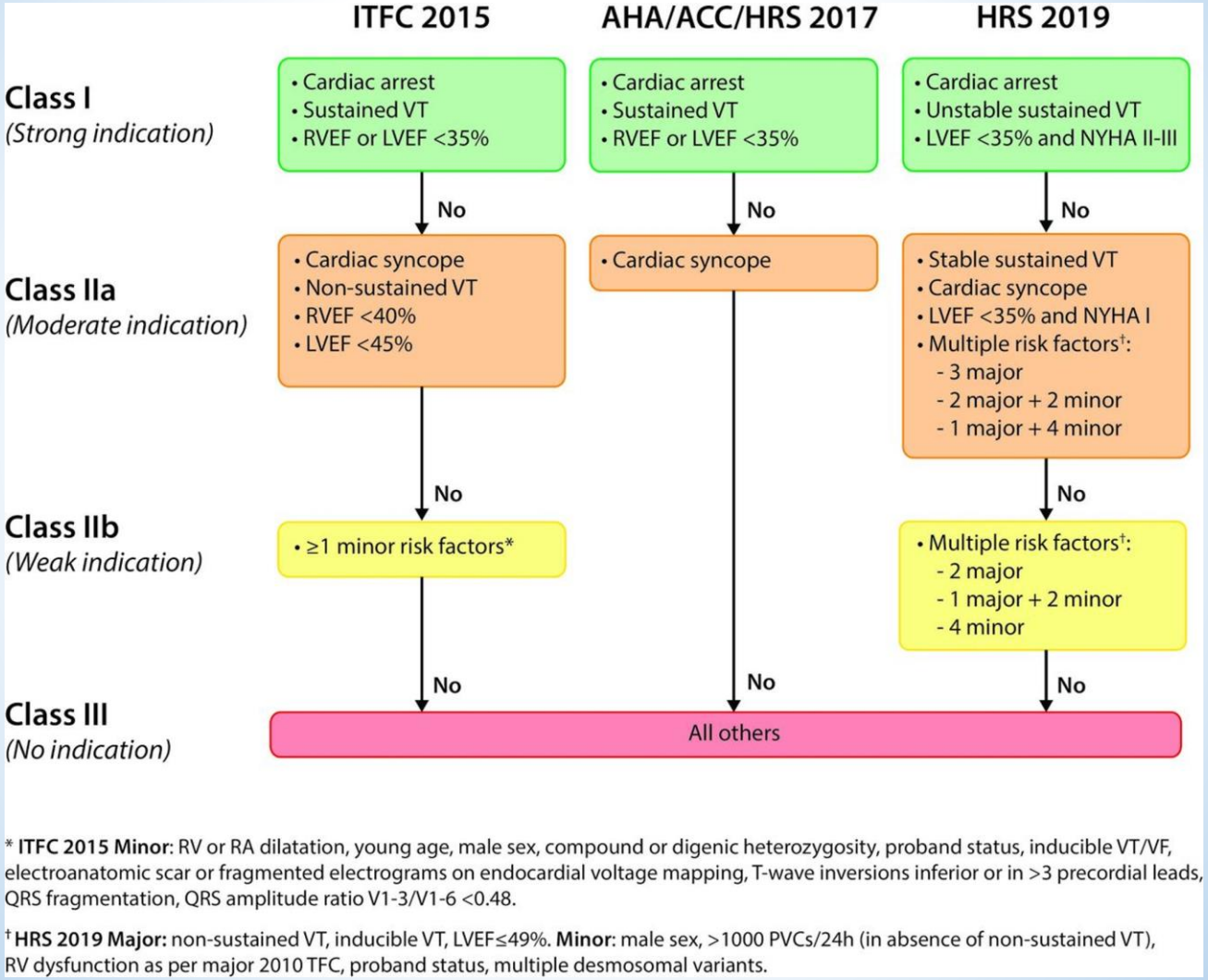
Cadrin-Tourigny J, et al. Eur Heart J. 2019
Cadrin-Tourigny J, et al. Eur Heart J. 2022

ICD-indikation?



* **ITFC 2015 Minor**: RV or RA dilatation, young age, male sex, compound or digenic heterozygosity, proband status, inducible VT/VF, electroanatomic scar or fragmented electrograms on endocardial voltage mapping, T-wave inversions inferior or in >3 precordial leads, QRS fragmentation, QRS amplitude ratio V1-3/V1-6 <0.48.

† **HRS 2019 Major:** non-sustained VT, inducible VT, LVEF≤49%. **Minor**: male sex, >1000 PVCs/24h (in absence of non-sustained VT), RV dysfunction as per major 2010 TFC, proband status, multiple desmosomal variants.

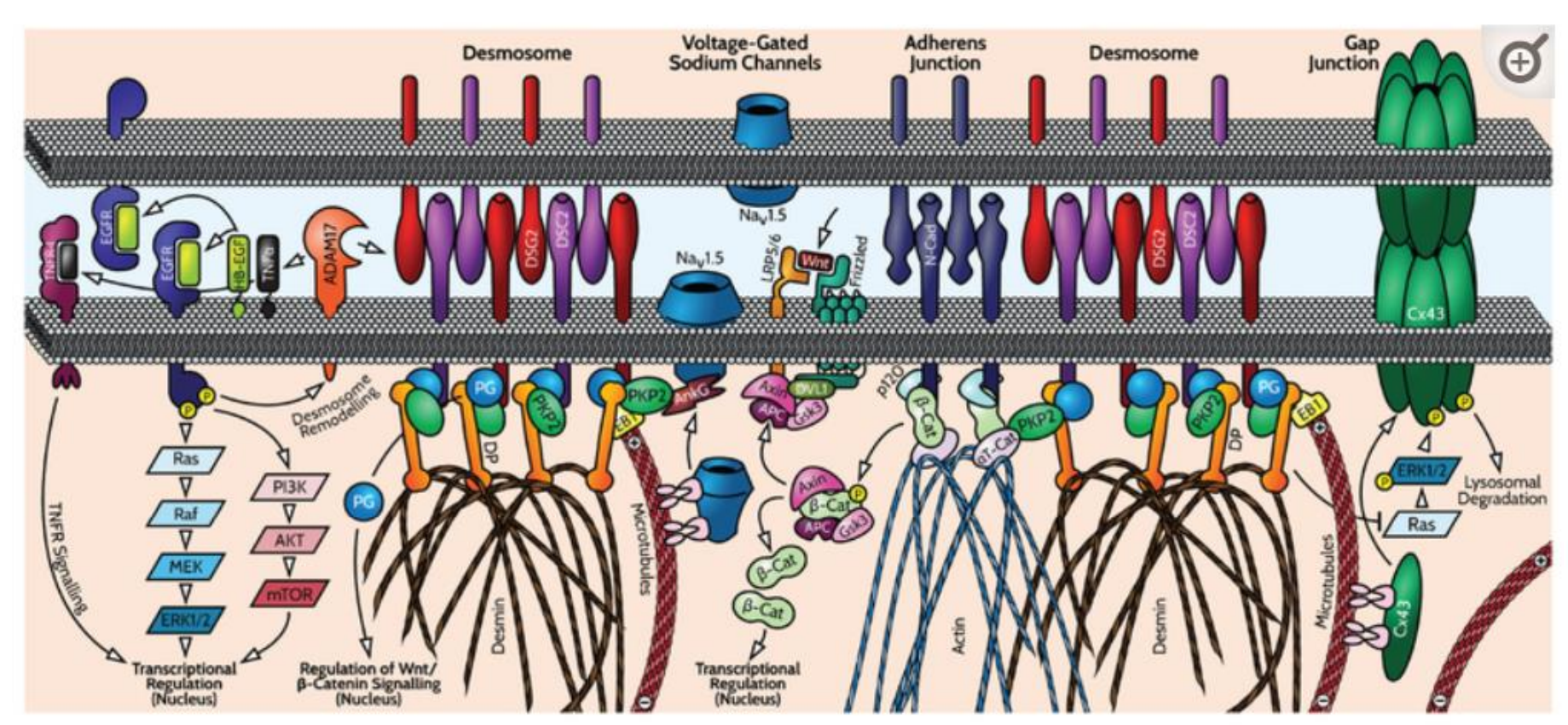
Bosman LP, Heart 2022

ARVC handläggning/behandling

- Arytmier
  - betablockad
  - Antiarytmika (amiodarone, sotalol)
  - ICD?
  - VT-ablation?
- Vid hjärtsvikt – sedvanlig behandling för detta
- I uttalade fall hjärttransplantation – i regel på sviktindikation
- Begränsa fysisk aktivitet – men exakt till vilken grad är okänt

- Graviditet och förlossning tolereras i regel väl

ARVC genetik

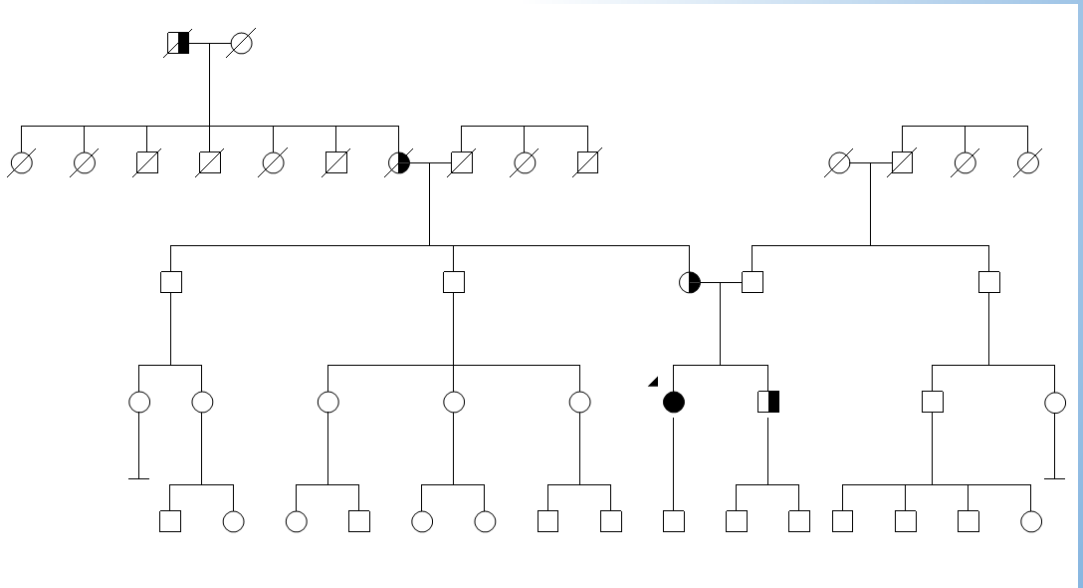


Elliott et al, Eur J Heart Fail. 2019

ARVC genetik, forts

- Autosomalt dominant ärftlighetsgång (mestadels)
- Indexpatienten (probanden) testas initial, sedan diagnosen ställts
  - Pedigree/ familjeträd
  - Kaskadtestning

- Tyvärr inte sällan mer svårtolkade fynd



Användbarhet för genetiska fynd

Table 5 Impact of genetic testing for the proband

Disease	Diagnostic	Prognostic	Therapeutic
Arrhythmia syndromes			
Long QT syndrome	+++	+++	+++
CPVT	+++	+	+
Brugada syndrome	+	+	+
Progressive cardiac conduction disease	+	+	+
Short QT syndrome	+	+	+
Sinus node disease	–	+	–
Atrial fibrillation	–	+	–
Early repolarization syndrome	–	–	–
Cardiomyopathies			
Hypertrophic cardiomyopathy	+++	++	++
Dilated cardiomyopathy	++	+++	++
Arrhythmogenic cardiomyopathy	+++	++	++
Left ventricular non-compaction	+	+	–
Restrictive cardiomyopathy	+	+	+
Congenital heart disease			
Syndromic CHD	+++	+	–
Non-syndromic CHD	+	–	–
Familial CHD	++	–	–

+++ : is recommended/is indicated or useful.
++ : can be recommended/can be useful.
+ : may be considered/may be useful.
– : is not recommended/is not indicated nor useful.

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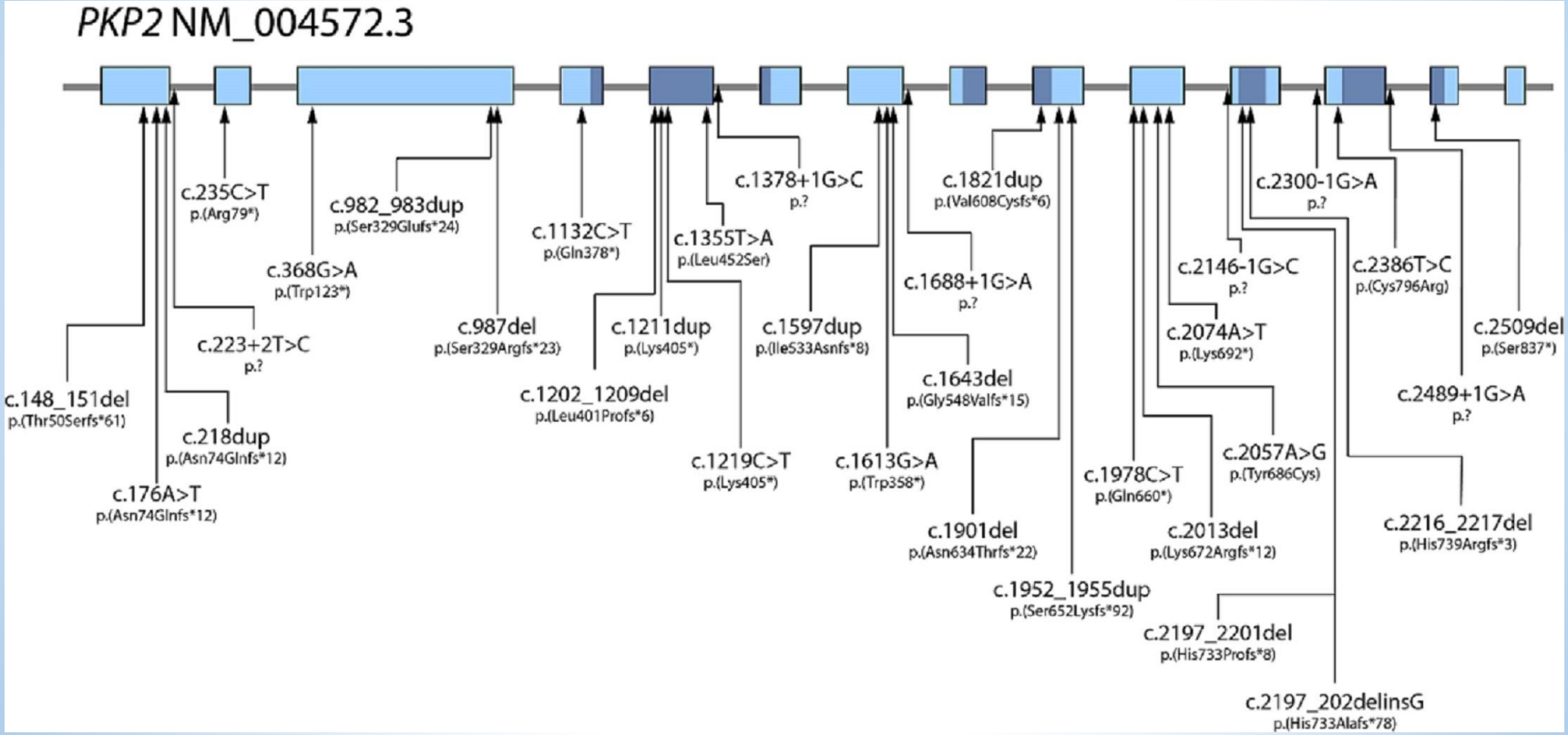
European Heart Rhythm Association (EHRA)/Heart Rhythm Society (HRS)/Asia Pacific Heart Rhythm Society (APHRS)/Latin American Heart Rhythm Society (LAHRS) Expert Consensus Statement on the Q2Q1 State of Genetic Testing for Cardiac Diseases. Wilde et al. 2022

PKP2 en gen – många varianter



Genetisk analys

Arrhythmogenic cardiomyopathy

Disease	Diagnostic	Prognostic	Therapeutic
ACM	+++	++	++

Recommendations	Consensus statement instruction	Ref.
Comprehensive genetic testing is recommended for all patients with consistent phenotypic features of ACM, including those cases diagnosed post-mortem, whatever familial context.		370
Genetic testing of first tier definitive disease-associated genes (currently *PKP2, DSP, DSG2, DSC2, JUP, TMEM43, PLN, FLNC, DES, LMNA*) is recommended.		370,371
Owing to the possibility of complex genotypes, in families with multiple affected members, the case with the more severe and/or earlier phenotype may be considered the 'genetic proband' and be tested first.		362
In patients with a borderline ACM phenotype, comprehensive genetic testing may be considered. The identification of a LP/P genetic variant would be useful to confirm the diagnosis.		372
Variant-specific genetic testing is recommended for family members and appropriate relatives following the identification of the disease-causative variant.		370,373
Predictive genetic testing in related children is recommended in those aged >10–12 years.		370,374
Predictive genetic testing in related children aged below 10–12 years may be considered, especially where there is a family history of early-onset disease.		Expert opinion

(*Continued*)

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State-of-the-Art Review

Arrhythmogenic Right Ventricular Cardiomyopathy

Andrew D. Krahn MD [a], Arthur A.M. Wilde MD, PhD [b, c], Hugh Calkins MD [d], Andre La Gerche MBBS, PhD [e], Julia Cadrin-Tourigny MD [f], Jason D. Roberts MD, MAS [g], Hui-Chen Han MBBS, PhD [a, h]

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Universitetssjukhuset i Linköping

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Review

Arrhythmogenic right ventricular cardiomyopathy: a focused update on diagnosis and risk stratification

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Predicting arrhythmic risk in arrhythmogenic right ventricular cardiomyopathy: A systematic review and meta-analysis

Laurens P. Bosman, MD • Arjan Sammani, BSc • Cynthia A. James, PhD, ScM • ...
Richard N.W. Hauer, MD, PhD • Folkert W. Asselbergs, MD, PhD • Anneline S.J.M. te Riele, MD, PhD •
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ARVC – take home message

- **Misstänk vid**
- T-negativitet i V1-V3 utan annan bakomliggande orsak
- Vid dilaterad högerkammare / nedsatt högerkammarfunktion
- Ventrikulära arytmier, främst hos unga patienter

- **Behandling**
- Arytmier – betablockad, antiarytmika, ev ICD, ev VT-ablation
- Vid hjärtsvikt – sedvanlig behandling för detta
- I uttalade fall hjärttransplantation
- Begränsa fysisk aktivitet – men exakt till vilken grad är okänt

ARVC – take home message

- **Genetik**
- Efter ställd diagnos
- För familjeutredning, inte riskvärdering hos individen

Tack!