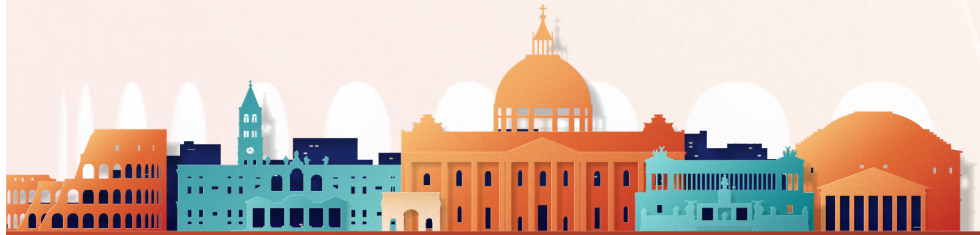
PLACE 2026
PLATFORM OF LABORATORIES FOR ADVANCES IN CARDIAC EXPERIENCE

CONGRESSO
ROMA, 25 - 27 Giugno 2026

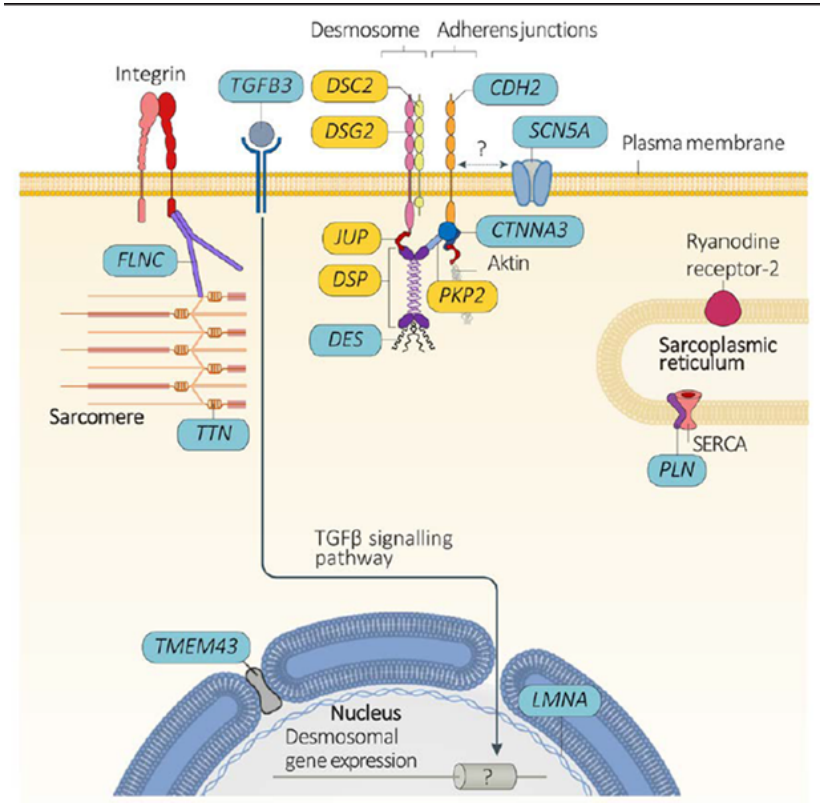
CARDIOMIOPATIA ARITMOGENA: WHAT'S NEW?

Gestione clinica nei pazienti con cardiomiopatia aritmogena con genotipo positivo e fenotipo negativo

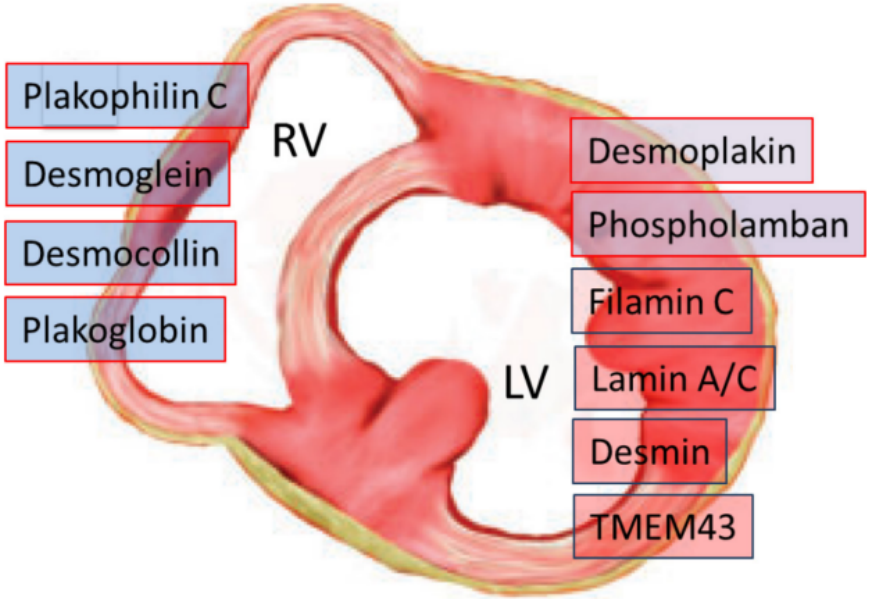
Barbara Bauce, Università degli Studi di Padova



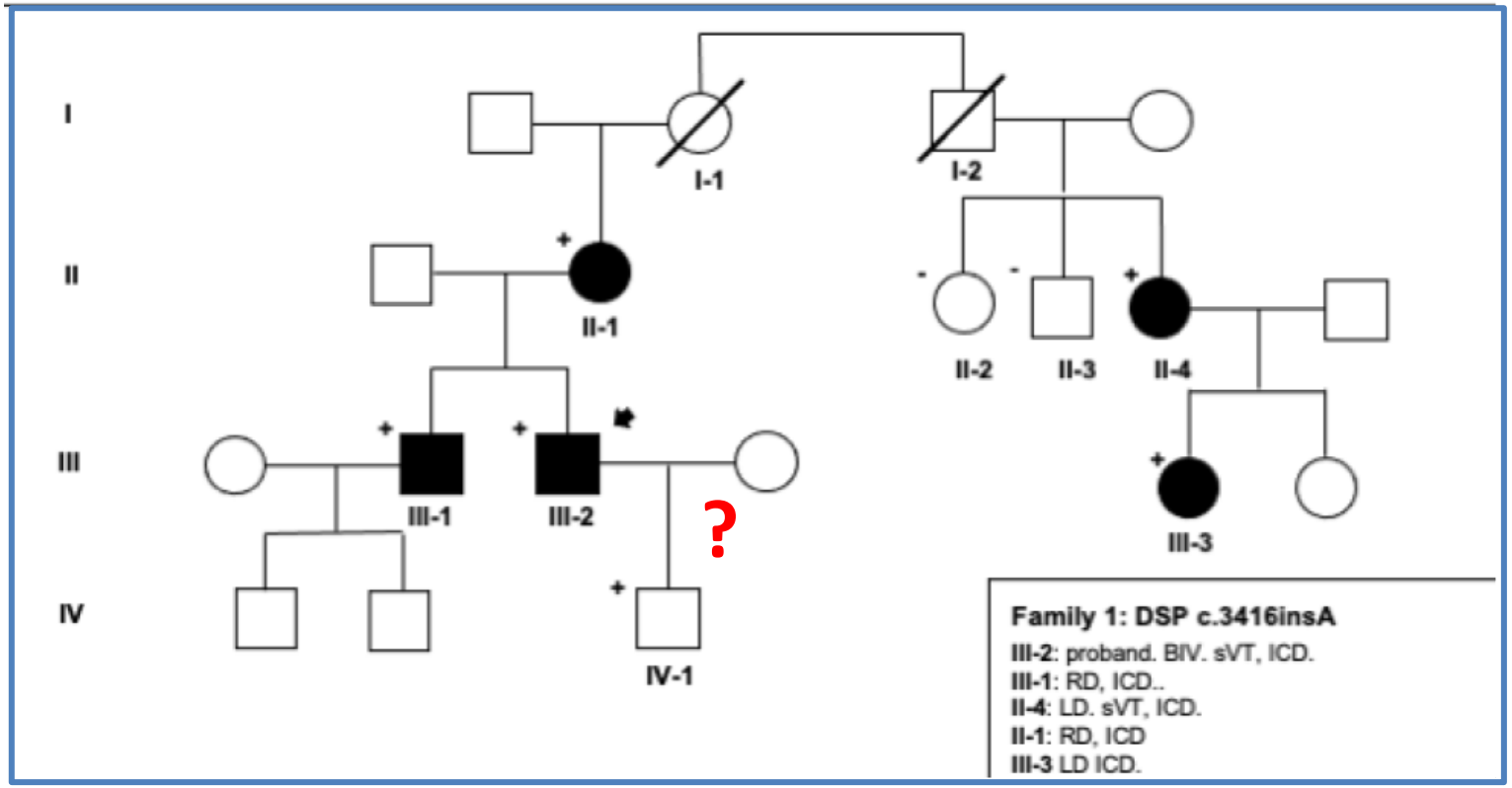
Correlazione genotipo-fenotipo



Asatryan et al, 2021



Corrado et at, Heart 2021



Bariani et al, Heart Rhythm 2022

Circulation

Check for updates

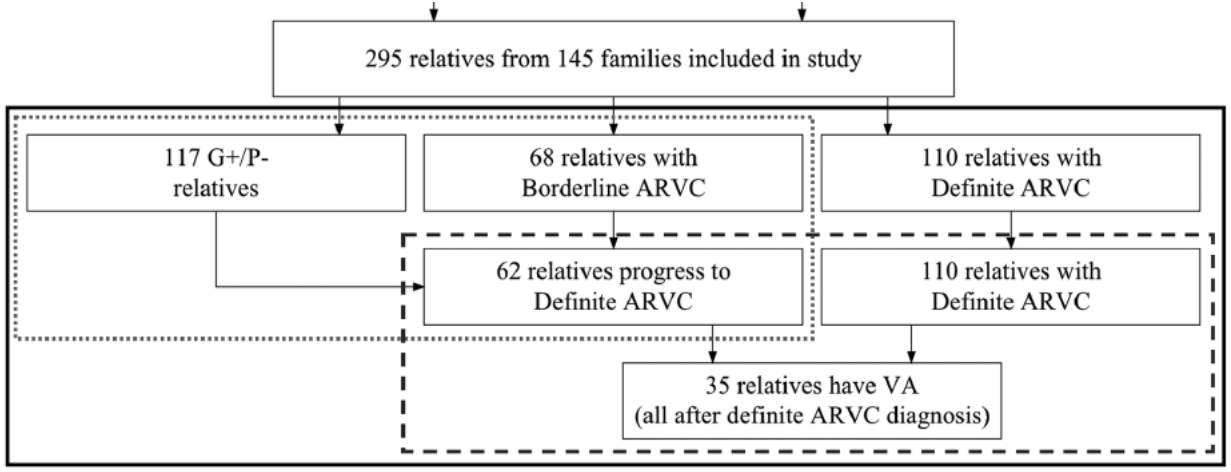
ORIGINAL RESEARCH ARTICLE

Family Screening in Relatives at Risk for Plakophilin-2–Associated Arrhythmogenic Right Ventricular Cardiomyopathy

Steven A. Muller, MD; Babken Asatryan, MD, PhD; Alessio Gasperetti, MD, PhD; Maarten J. Cramer, MD, PhD; Ahmad S. Amin, MD, PhD; Peter Loh, MD, PhD; Richard T. Carrick, MD, PhD; Moniek G.P.J. Cox, MD, PhD; Pim van der Harst, MD, PhD; Marish I.F.J. Oerlemans, MD, PhD; Crystal Tichnell, MGC, RN; Sing-Chien Yap, MD, PhD; Brittney Murray, MS, CGC; Stefan L. Zimmerman, MD; J. Peter van Tintelen, MD, PhD; Hugh Calkins, MD; Anneline S.J.M. te Riele, MD, PhD*; Cynthia A. James, PhD, CGC*

Circulation 2025

Tot 295 relatives
- 110 (37%) affetti
- 68 (23%) bordeline
- 117 (40%) G+/F-



62 (21%) con diagnosi durante FU di 8.5 anni

Tot 172 affetti (58%)

Età dei pazienti G+/F-

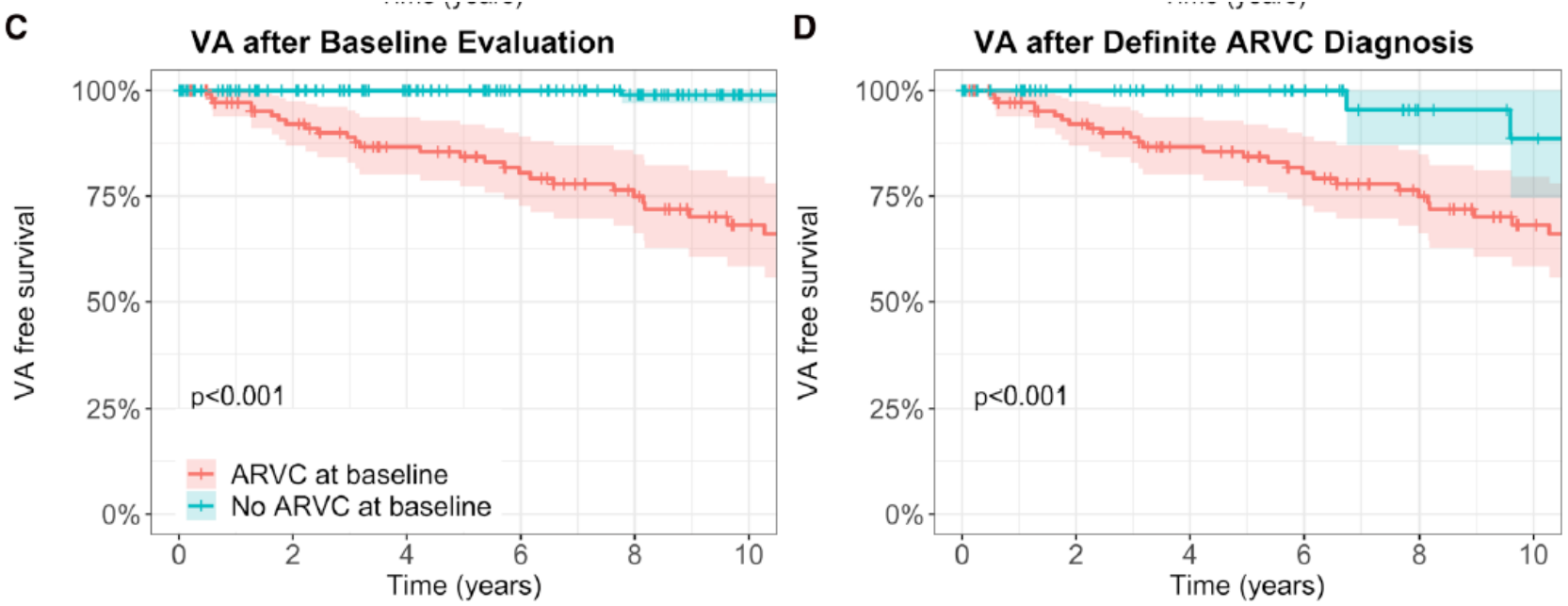
Table 1. Baseline Characteristics

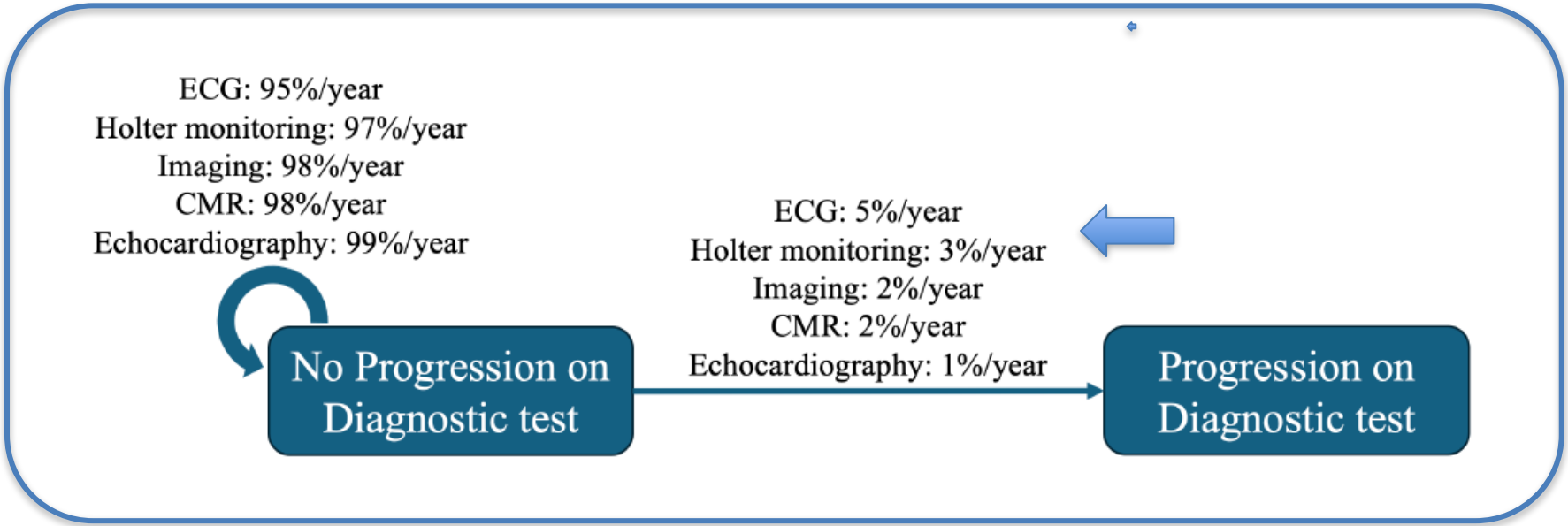
	Complete cohort (N=295)	Nondefinite ARVC			Definite ARVC (n=110)	*P* value, G+/P– vs borderline ARVC	*P* value, definite ARVC vs nondefinite ARVC
		Overall nondefinite ARVC (n=185)	G+/P– (n=117)	Borderline ARVC (n=68)			
Age at presentation, y	30.9 (18.0–47.7)	26.2 (15.2–43.4)	21.3 (13.5–41.3)	32.9 (21.7–47.2)	37.3 (22.2–50.5)	0.001	<0.001
Age (categorical), n (%)						0.004	<0.001
<14 y	38 (12.9)	37 (20.0)	32 (27.4)	5 (7.4)	1 (0.9)		
14–20 y	56 (19.0)	36 (19.5)	25 (21.4)	11 (16.2)	20 (18.2)		
20–30 y	50 (16.9)	29 (15.7)	17 (14.5)	12 (17.6)	21 (19.1)		
30–40 y	46 (15.6)	28 (15.1)	13 (11.1)	15 (22.1)	18 (16.4)		
>40 y	105 (35.6)	55 (29.7)	30 (25.6)	25 (36.8)	50 (45.5)		
Sex, n (%)						0.181	0.222
Male	122 (41.4)	82 (44.3)	47 (40.2)	35 (51.5)	40 (36.4)		
Female	173 (58.6)	103 (55.7)	70 (59.8)	33 (48.5)	70 (63.6)		

	Complete cohort (N=295)	Nondefinite ARVC			Definite ARVC (n=110)	*P* value, G+/P– vs borderline ARVC	*P* value, definite ARVC vs nondefinite ARVC
		Overall nondefinite ARVC (n=185)	G+/P– (n=117)	Borderline ARVC (n=68)			
Age at presentation, y	30.9 (18.0–47.7)	26.2 (15.2–43.4)	21.3 (13.5–41.3)	32.9 (21.7–47.2)	37.3 (22.2–50.5)	0.001	<0.001
Depolarization TFC fulfillment, n (%)	65 (22.0)	36 (19.5)	0 (0.0)	36 (52.9)	29 (26.6)	<0.001	0.192
Prolonged TAD	41 (13.9)	22 (11.9)	0 (0.0)	22 (32.4)	19 (17.4)	<0.001	0.222
Late potentials on SAECG	30 (38.0)	16 (29.1)	0 (0.0)	16 (55.2)	14 (58.3)	<0.001	0.027
Holter TFC fulfillment, n (%)	83 (28.1)	17 (9.2)	0 (0.0)	17 (25.0)	66 (60.6)	<0.001	<0.001
PVC count/24h (n=275)	46 (2–681)	5 (1–59)	2 (0–37)	18 (2–411)	831 (178–2813)	<0.001	<0.001
Imaging TFC fulfillment, n (%)	51 (17.3)	2 (1.1)	0 (0.0)	2 (2.9)	49 (45.0)	0.134	<0.001
CMR TFC fulfillment (n=163), n (%)						0.361	<0.001
Minor TFC	14 (7.1)	1 (1.0)	0 (0.0)	1 (2.9)	13 (13.0)		
Major TFC	30 (15.2)	0 (0.0)	0 (0.0)	0 (0.0)	30 (30.0)		
Echocardiographic TFC fulfillment (n=206)						0.387	<0.001
Minor TFC	6 (2.3)	1 (0.6)	0 (0.0)	1 (1.7)	5 (4.9)		
Major TFC	11 (4.3)	0 (0.0)	0 (0.0)	0 (0.0)	11 (10.7)		

	Complete cohort (N=295)	Nondefinite ARVC			Definite ARVC (n=110)	*P* value, G+/P– vs borderline ARVC	*P* value, definite ARVC vs nondefinite ARVC
		Overall nondefinite ARVC (n=185)	G+/P– (n=117)	Borderline ARVC (n=68)			
Age at presentation, y	30.9 (18.0–47.7)	26.2 (15.2–43.4)	21.3 (13.5–41.3)	32.9 (21.7–47.2)	37.3 (22.2–50.5)	0.001	<0.001
Symptoms at baseline presentation (n=236), n (%)						0.894	<0.001
Asymptomatic	159 (67.4)	119 (77.3)	78 (78.8)	41 (74.5)	40 (48.8)		
Palpitations	41 (17.4)	21 (13.6)	12 (12.1)	9 (16.4)	20 (24.4)		
Presyncope	17 (7.2)	5 (3.2)	3 (3.0)	2 (3.6)	12 (14.6)		
Syncope	19 (8.1)	9 (5.8)	6 (6.1)	3 (5.5)	10 (12.2)		
Electrical TFC fulfillment, n (%)	168 (56.9)	66 (35.7)	0 (0.0)	66 (97.1)	102 (92.7)	<0.001	<0.001
Repolarization TFC fulfillment, n (%)	92 (31.2)	13 (7.0)	0 (0.0)	13 (19.1)	79 (71.8)	<0.001	<0.001
T-wave inversions V_1–V_2	34 (11.5)	13 (7.0)	0 (0.0)	13 (20.0)	21 (19.1)	<0.001	0.002
T-wave inversions V_1–V_3	51 (17.2)	0 (0.0)	0 (0.0)	0 (0.0)	51 (49.0)	<0.001	<0.001
T-wave inversions V_4–V_6	5 (1.7)	0 (0.0)	0 (0.0)	0 (0.0)	5 (4.8)	…	0.006
T-wave inversions with CRBBB V_1–V_4	2 (0.7)	0 (0.0)	0 (0.0)	0 (0.0)	2 (1.9)	…	0.253

Instabilità elettrica nei pz G+/P-




ECG: 95%/year
Holter monitoring: 97%/year
Imaging: 98%/year
CMR: 98%/year
Echocardiography: 99%/year
No Progression on Diagnostic test
ECG: 5%/year
Holter monitoring: 3%/year
Imaging: 2%/year
CMR: 2%/year
Echocardiography: 1%/year
Progression on Diagnostic test

Fattori rilevanti nello screening

Table 2. Frailty Cox Proportional Hazard Regression for Definite ARVC Diagnosis

	Analysis for definite ARVC diagnosis, adjusted for baseline clinical phenotype*			
	HR	Lower 95% CI	Upper 95% CI	*P* value
Age (referent: ≥40 y)†	…	…	…	
<14 y	1.33	0.58	3.02	0.500
14–20 y	1.34	0.61	2.97	0.470
20–40 y	2.33	1.21	4.48	0.012
Female sex	2.38	1.39	4.17	0.002
Sibling of the pro-band	1.14	0.62	2.08	0.680
Symptomatic	1.79	0.98	3.25	0.058

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Electrophysiolog

Clinical Profile and Long-term Follow-up of 37 Families With Arrhythmogenic Right Ventricular Cardiomyopathy

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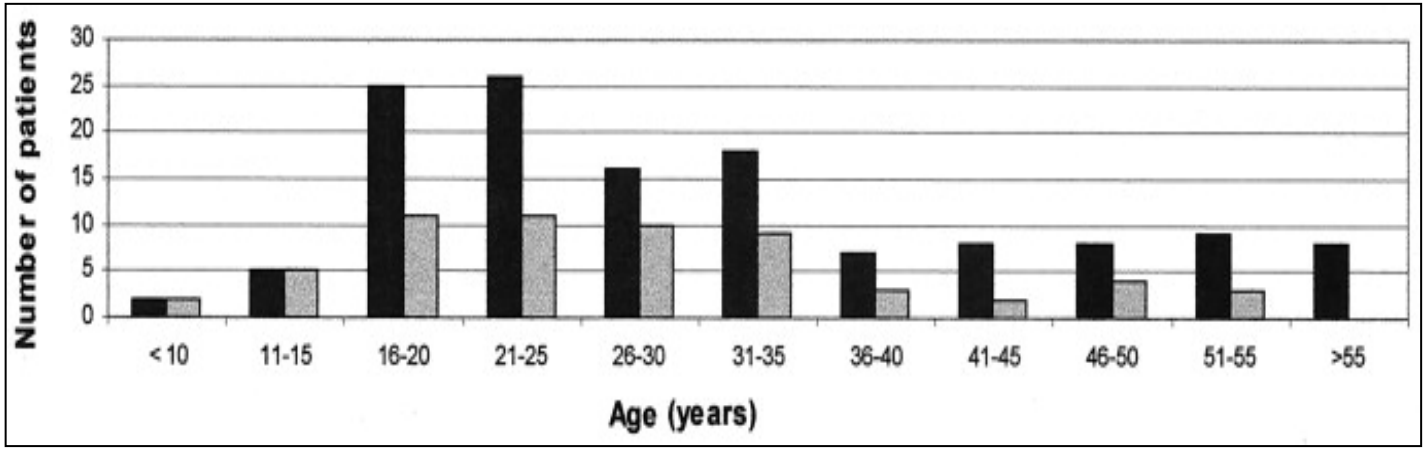
Padua, Italy



Age of patients at time of ARVC diagnosis (black bars) and at time of onset of arrhythmias (gray bars)

Clinical profile and long-term follow-up of a cohort of patients with desmoplakin cardiomyopathy

Heart Rhythm 2022

Riccardo Bariani, MD,* Marco Cason, PhD,* Ilaria Rigato, MD, PhD,*
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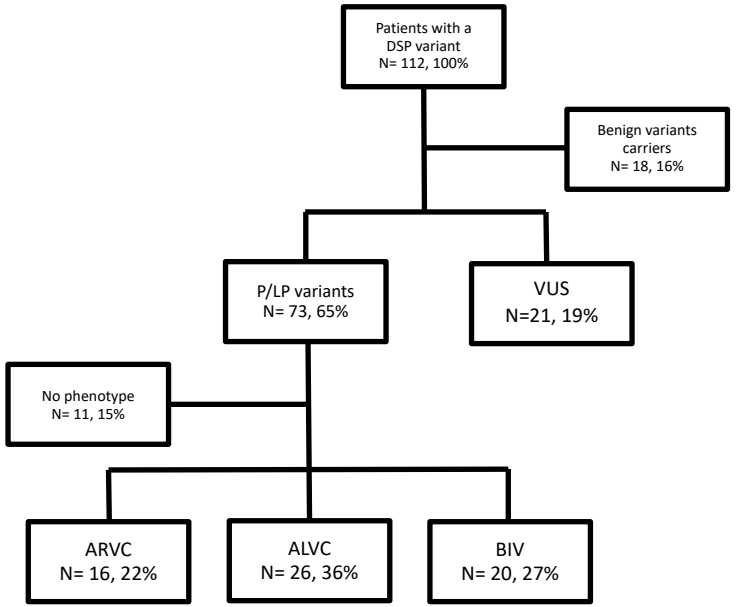
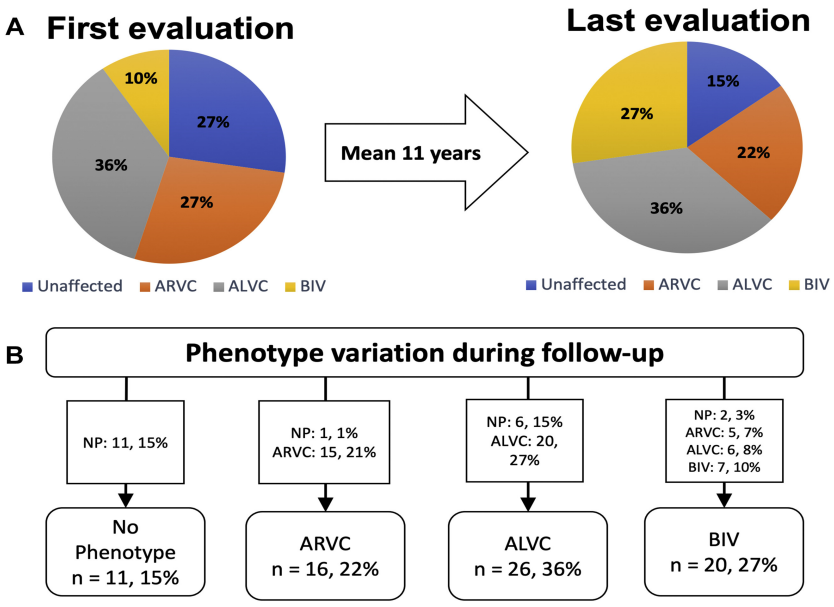
Cristina Basso, MD, PhD,* Kalliopi Pilichou, MsC, PhD,*[1] Barbara Bauce, MD, PhD*[1]





Phenotypic Expression and Clinical Outcomes in Patients With Arrhythmogenic Cardiomyopathies

Riccardo Bariani, MD, PhD, Ilaria Rigato, MD, PhD, Rudy Celeghin, PhD, Maria Bueno Marinas, PhD, Alberto Cipriani, MD, Alessandro Zorzi, MD, PhD, Valeria Pergola, MD, Sabino Iliceto, MD, Cristina Basso, MD, PhD, Martina Perazzolo Marra, MD, PhD, Domenico Corrado, MD, PhD, Dario Gregori, PhD, Kalliopi Pilichou, PhD, Barbara Bauce, MD, PhD

Tot 628 pts

- 337 (54%) con diagnosi ARVC
- 106 (17%) diagnosi bordeline/ possible
- 185 (29%) non affetti

TABLE 1 Clinical Features

	Total (N = 446; 100%)	ARVC (n = 196; 44%)	ALVC (n = 104; 23%)	BIV (n = 146; 33%)	q Value
Female	167 (37)	79 (40)	44 (42)	44 (30)	0.310
2010 TFC					<0.001
None	3 (1)	1 (1)	1 (1)	1 (1)	
Certain	337 (76)	164 (84)	34 (33)	139 (95)	
Borderline	56 (13)	20 (10)	33 (32)	3 (2)	
Possible	50 (11)	11 (6)	36 (35)	3 (2)	

JACC 2024

Caso clinico

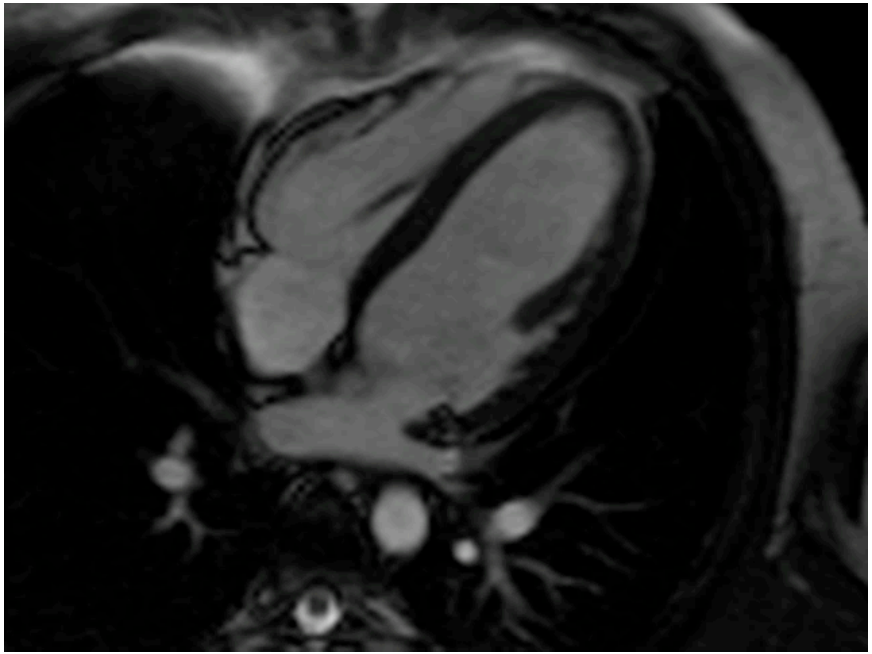
- Maschio, 21 anni
- Anamnesi familiare: madre e nonno paterno con diagnosi di Cardiomiopatia Aritmogena

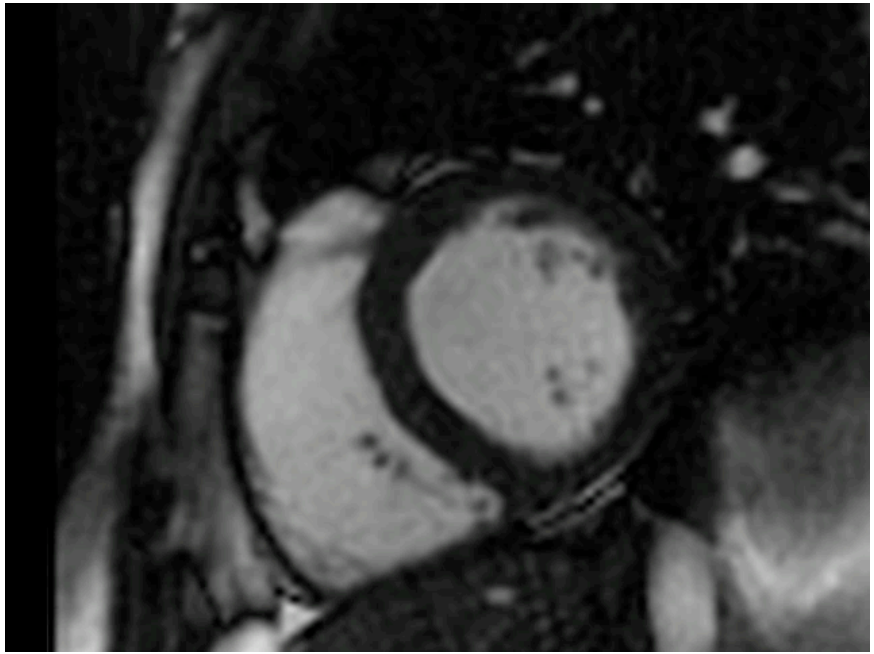
  Non familiarità per morte improvvisa
- Madre con impianto di ICD in prevenzione primaria a 55 anni e presenza di variante genetica PKP2 (classe 4)
- Non pratica attività sportiva

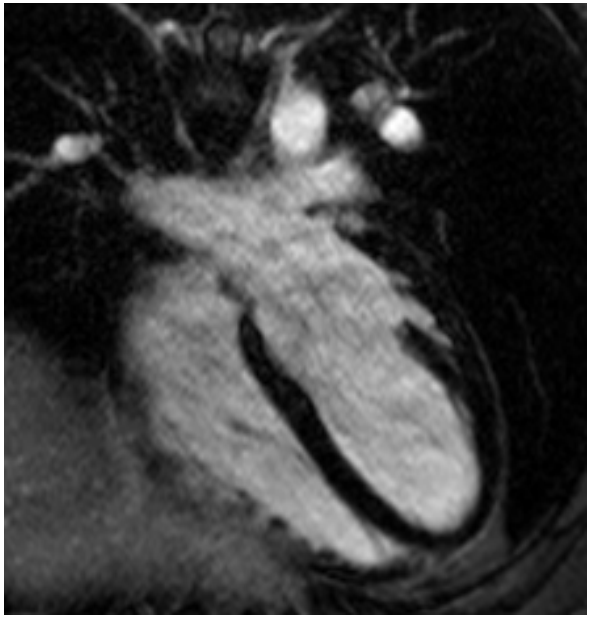
- Screening cardiologico 2008:
  - Paziente asintomatico, classe NYHA I^, buon compenso emodinamico
  - ECG: normale
  - Ecocardiogramma: nei limiti di norma
  - RM cardiaca: ventricoli di normali dimensioni e funzione, non LGE
  - ECG Holter: rari BEV monomorfi
  - Test genetico: variante PKP2 presente anche nella madre e nel nonno

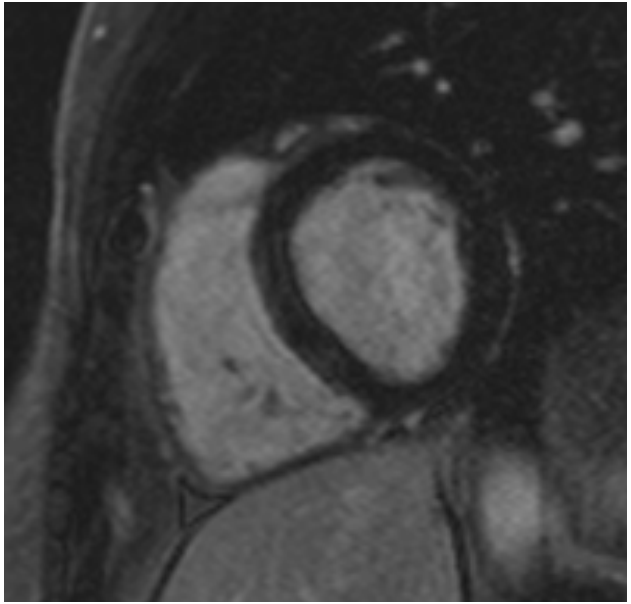
  - Terapia: bisoprololo 1,25 mg/die

RM cardiaca 2011



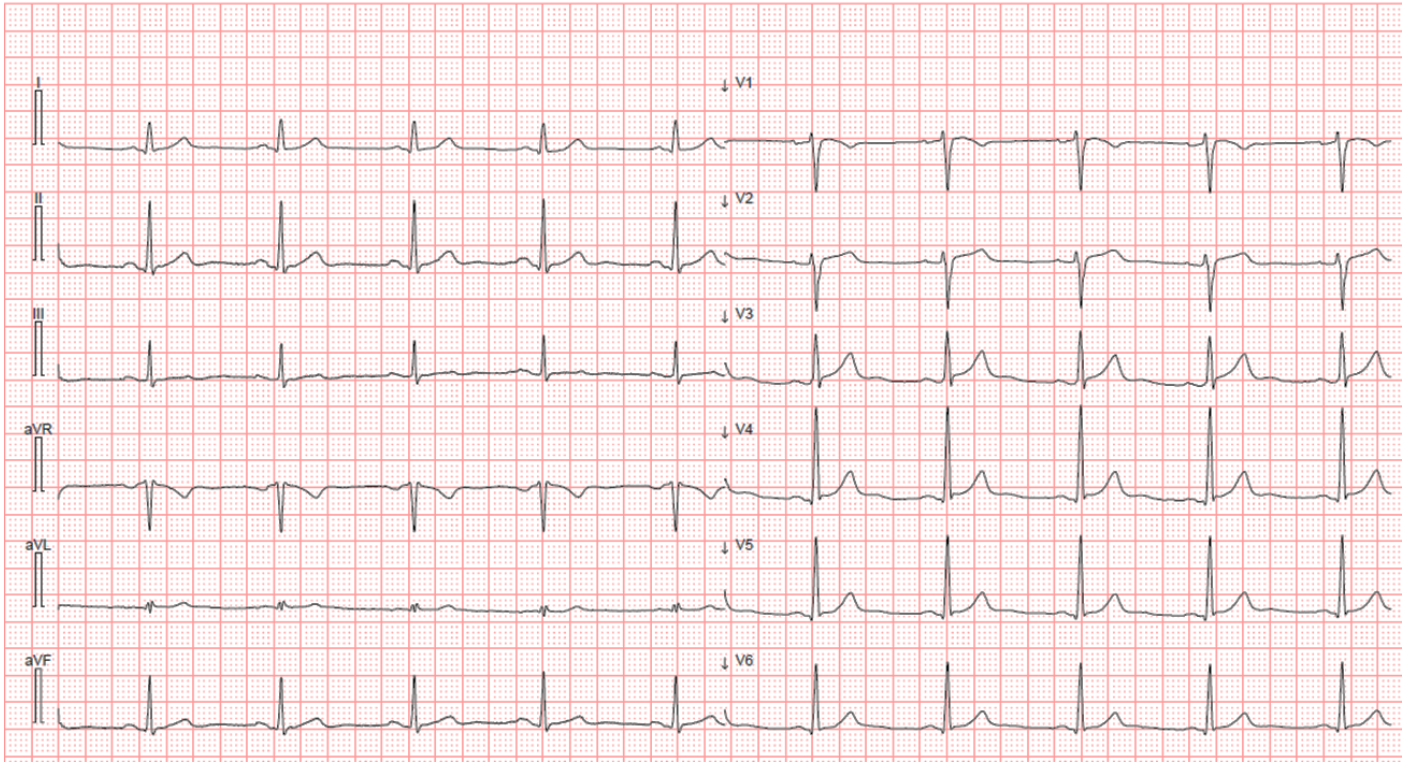


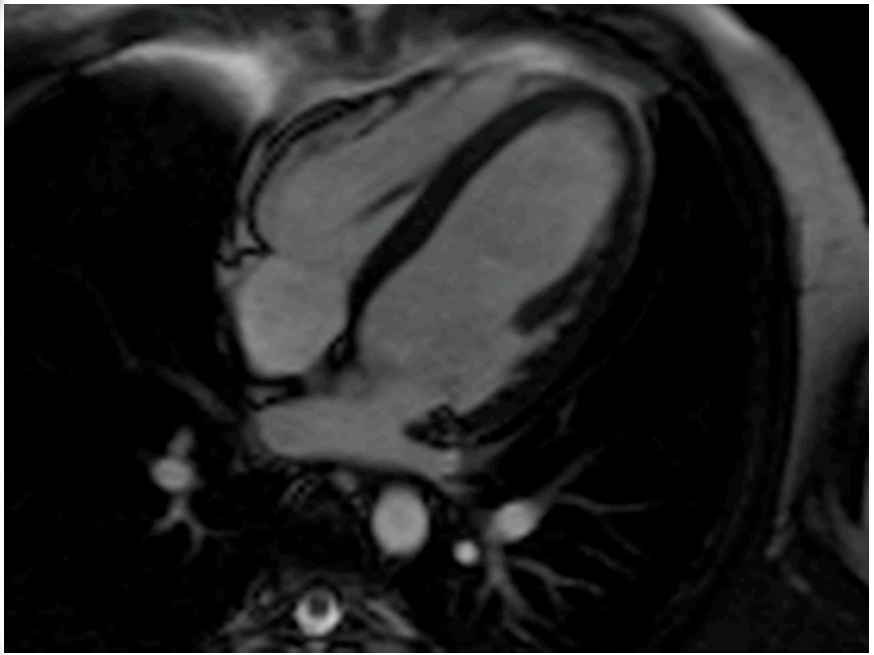




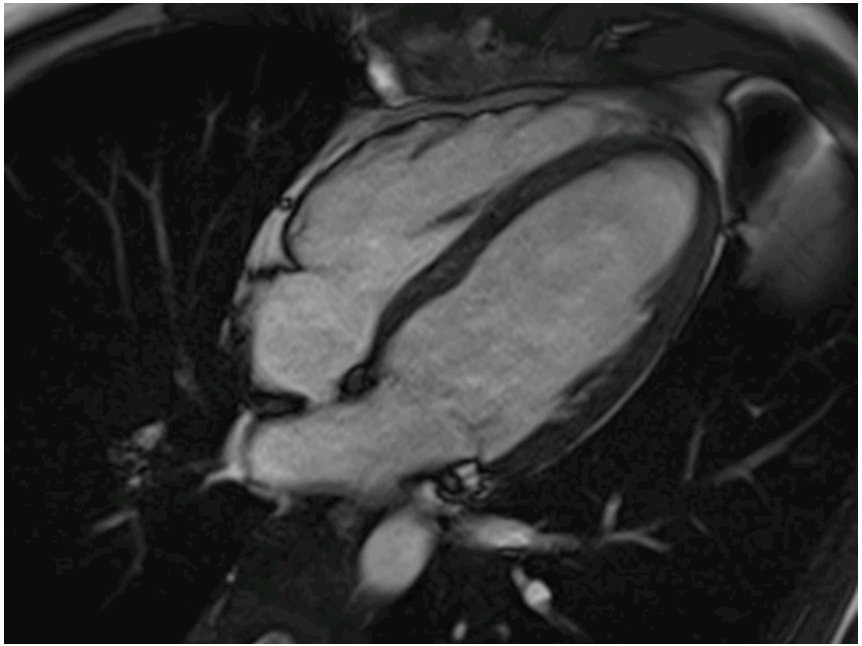
Valutazione Cardiologica 2022

- Età paziente: 32 anni
- Riferisce saltuario cardiopalmo; terapia: bisoprololo 2,5 mg/die





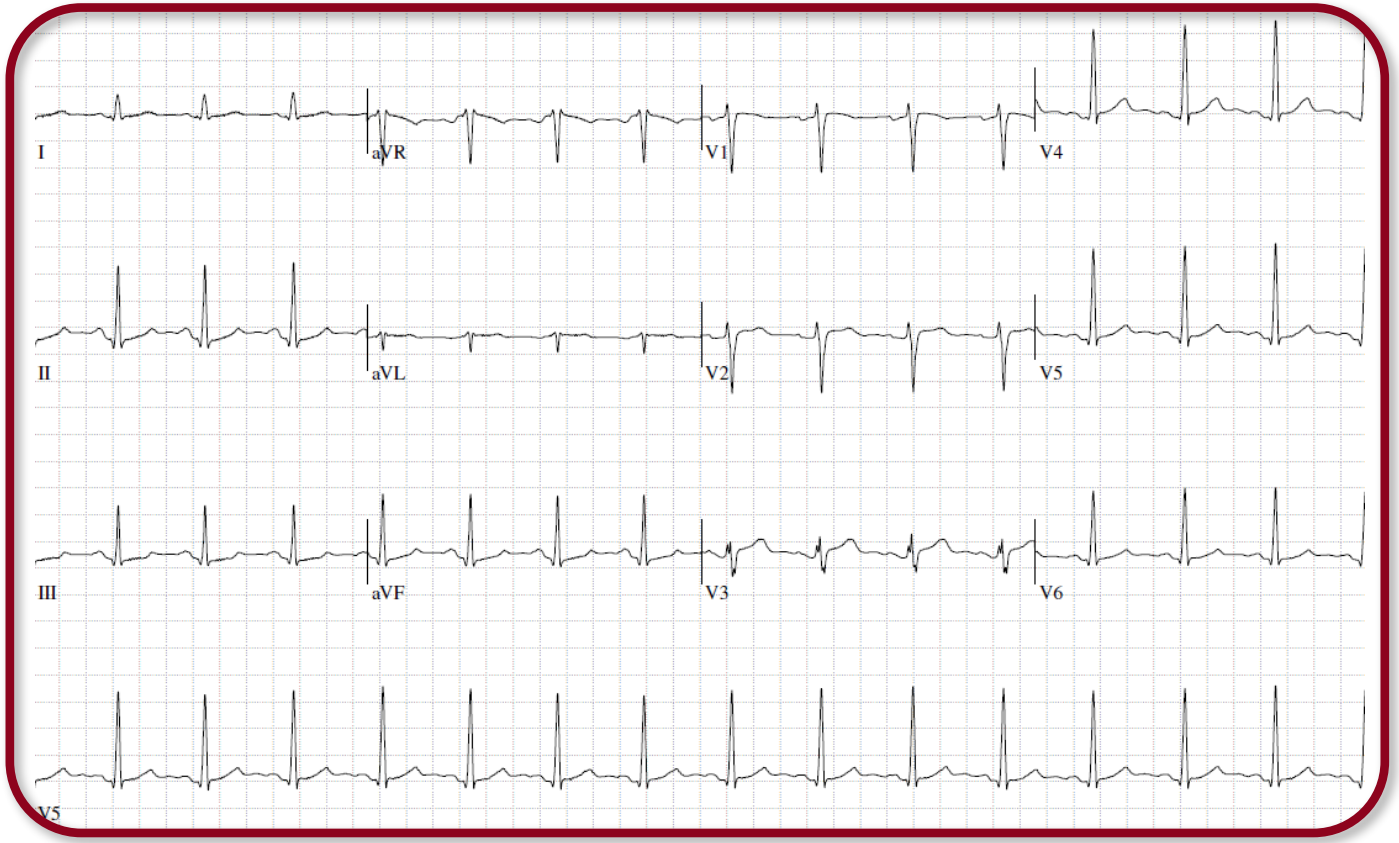
Eta’ 21 anni (2011)

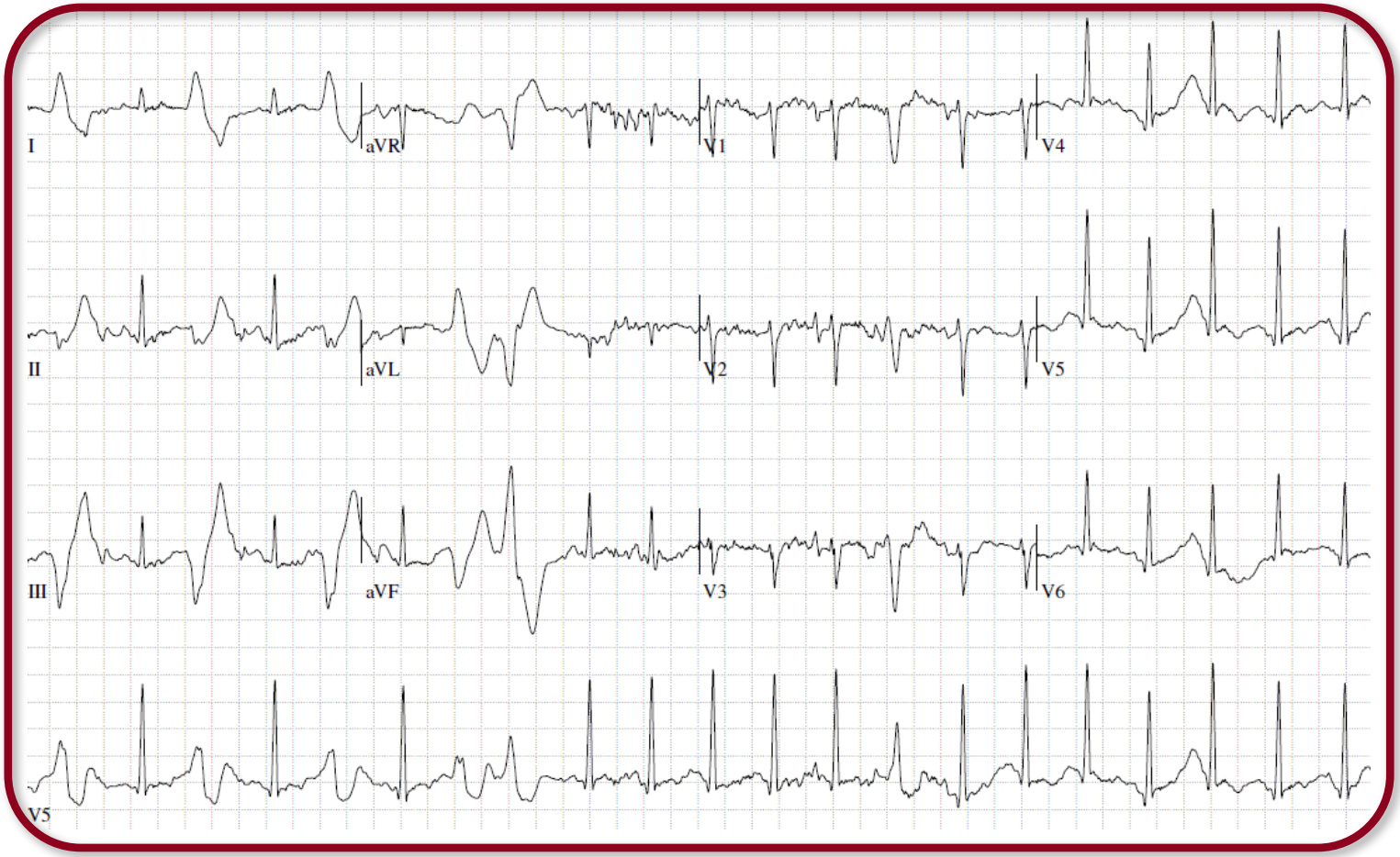


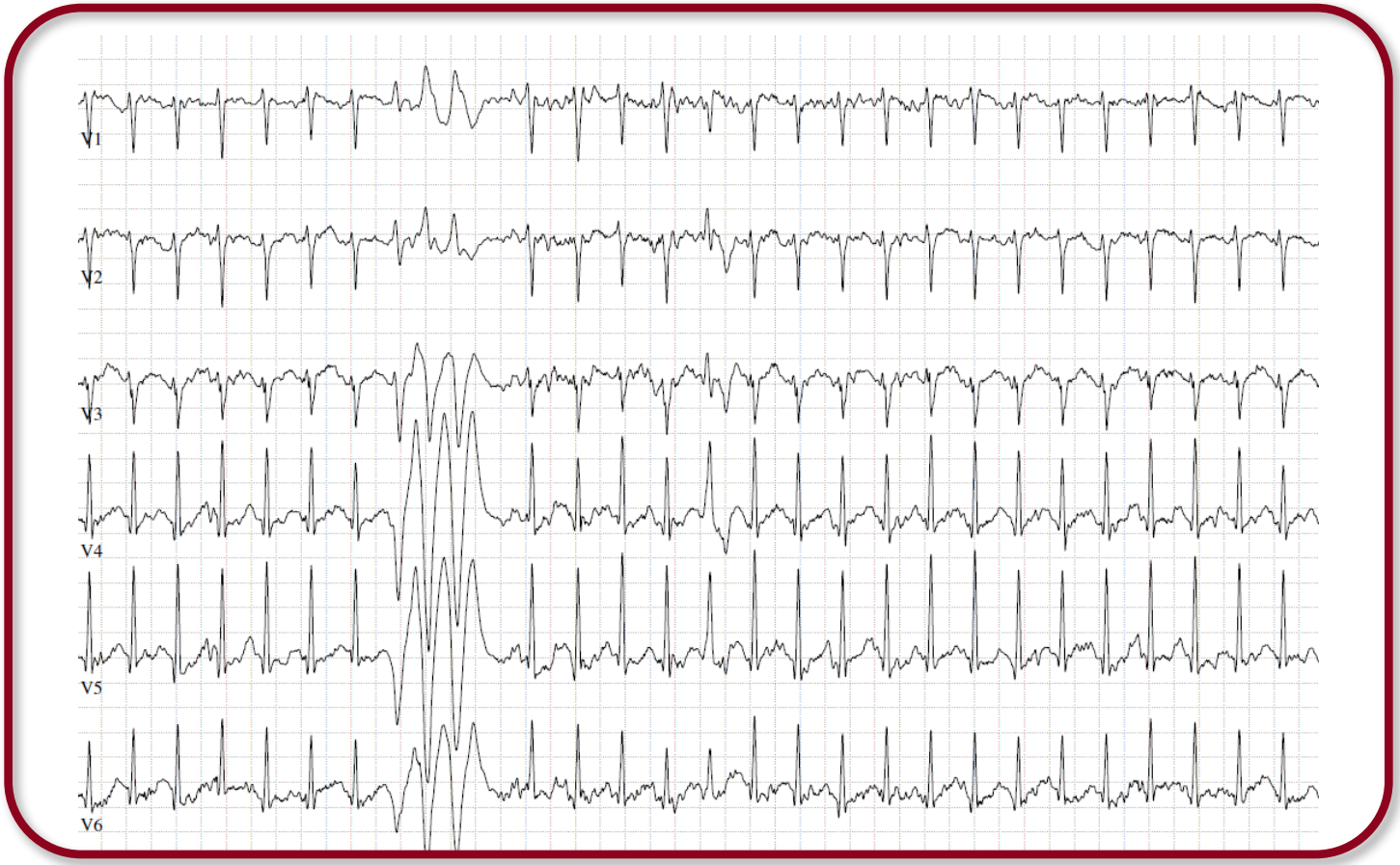
Eta’ 32 anni (2022)

- Ecocardiogramma:
  - ventricolo destro di normali dimensioni e funzione sistolica complessiva conservata, dilatazione tratto di efflusso ed alterazioni della cinetica regionale in regione sottotricuspidalica
- ECG Holter: alcuni BEV monomorfi

Test da sforzo




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



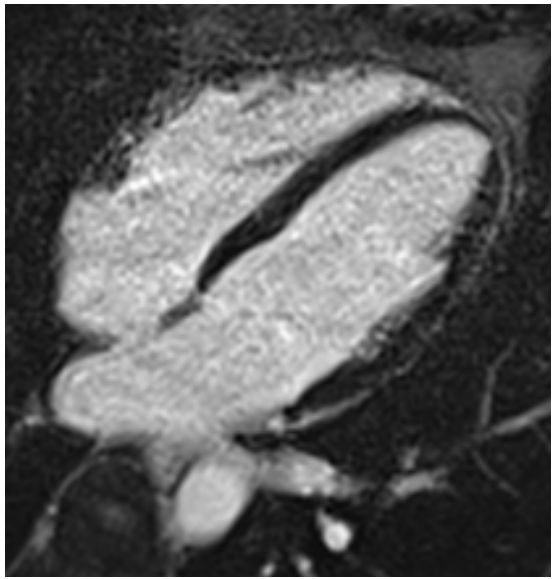
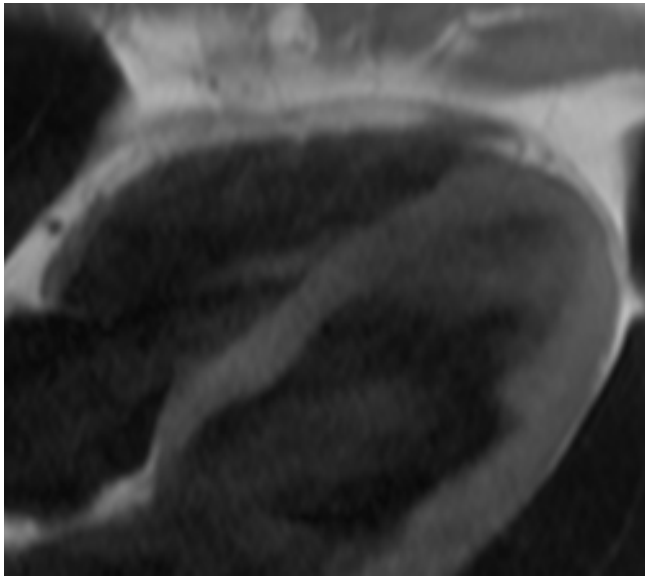
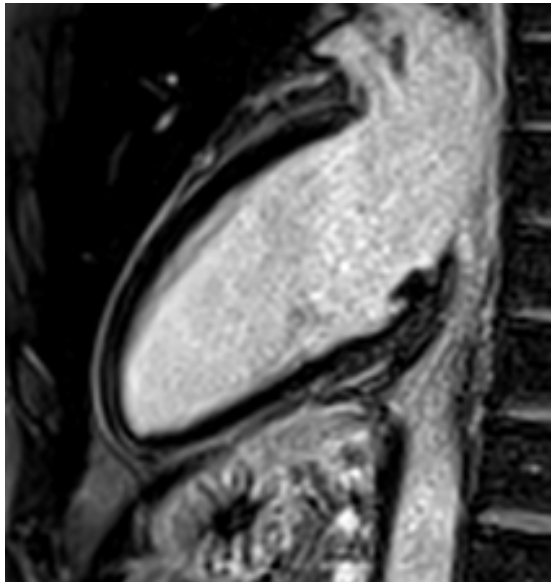
Consigliato metoprololo 100 mg/die

RM CARDIACA

Valutazione morfofunzionale a riposo
Ventricolo sinistro di normali dimensioni con conservata funzione di pompa (EDV 81 ml/mq, ESV 32 ml/mq, FE 61 %, massa 53 g/mq, SV 92 ml), in assenza di alterazioni della cinetica regionale.
Ventricolo destro di normali dimensioni con conservata funzione di pompa (EDV 89 ml/mq, ESV 38 ml/mq, FE 57 %, SV 93 ml), in presenza di irregolarità parietali sistoliche del profilo anteriore del corpo e del tratto d¿efflusso. A livello di quest¿ultimo e della parete diaframmatica si apprezzano focali aree di discinesia.
Normali dimensioni biatriali; atrio sinistro (area 20 cm2); atrio destro (area 15 cm2).
Non significativi shunt intracardiaci (Qp:Qs 1.1).
Non valvulopatie di rilievo.
Assente versamento pericardico.

Caratterizzazione tissutale
Alle sequenze TSE T1W non segni di infiltrazione adiposa del ventricolo sinistro. Si apprezzano tuttavia iperintensità di segnale lungo il profilo anteriore dell¿RVOT.
Normali i valori di T1 mapping (1031 ms, VN 970 +/- 70 ms) valutati a livello settale medio.
Dopo somministrazione di MdC in fase precoce ("first pass"), non deficit di perfusione.
Normale reperimento del TI.
Nelle sequenze post contrastografiche tardive non aree di enhancement ventricolare sinistro. Tenui iperintensità di segnale omosede alle aree discinetiche del ventricolo destro.

Sintesi interpretativa: anomalie di cinetica e tissutali del ventricolo destro suggestive per cardiomiopatia aritmogena del ventricolo destro. Normali dimensioni e funzione biventricolari.

Follow-up nei pz G+/F-

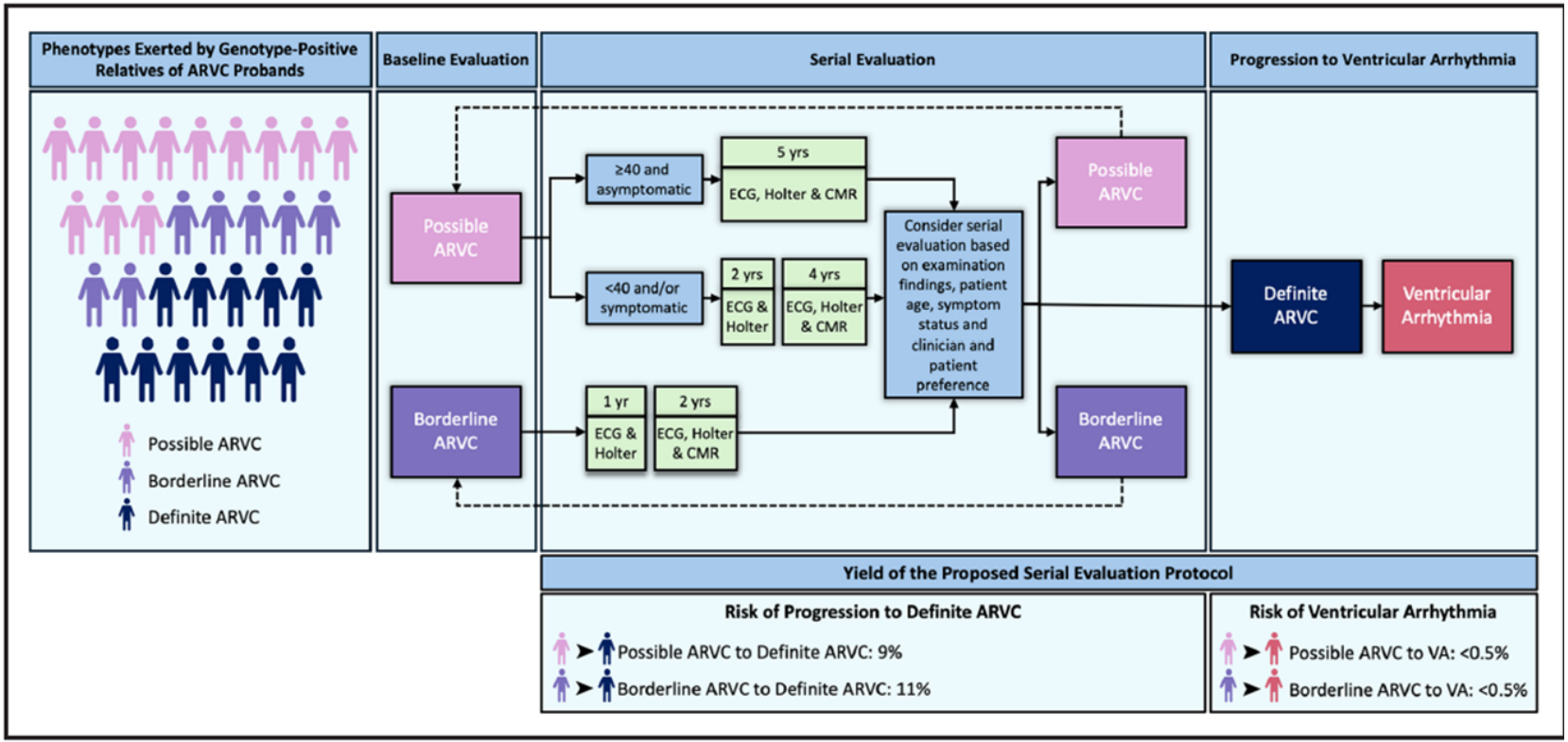


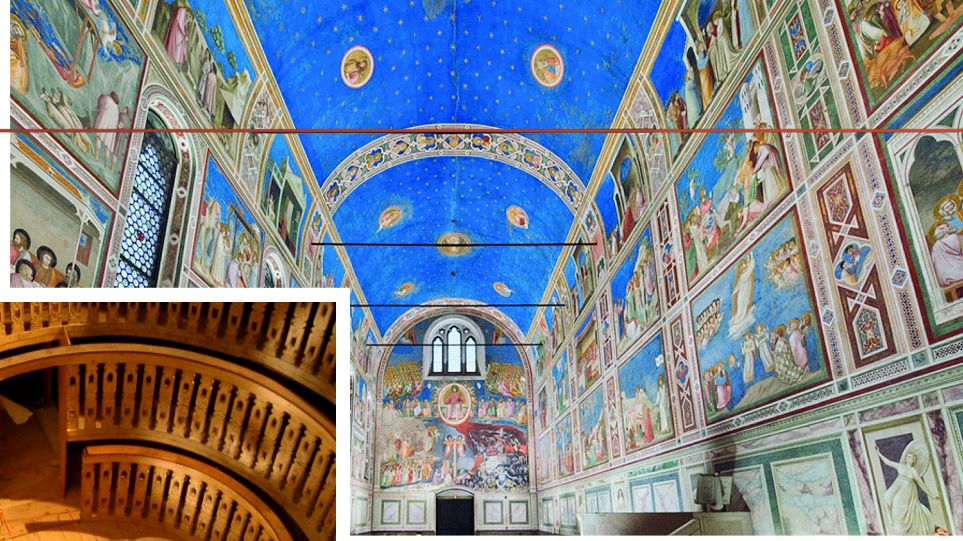
Figure 6. Longitudinal screening algorithm for at-risk pathogenic or likely pathogenic *PKP2* carriers.

Paziente G+/F- nella ACM

- Nella Cardiomiopatia Aritmogena la presenza di una variante genetica è rilevata nel 50-60% dei casi
- Nei familiari portatori di varianti **P/LP** del gene **PKP2**, i familiari più giovani presentano un rischio maggiore di progressione della malattia
- Le anomalie correlate alla malattia sono generalmente rilevate per la prima volta all'ECG e al monitoraggio Holter
- Il follow-up dei pazienti va programmato in base all'età ed alla presenza di sintomi

PLACE 2026

PLATFORM OF LABORATORY AND ADVANCES IN CARDIAC EXPERIENCE



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