P.L., female, 14 years-old

Età : 13 Anni
Sesso : M
Peso : 56 kg Altezza : 167.0 cm
n° ID :
Reparto : MED E CARDIOLOGIA DELLO SPORT

Analizzato da :
Data esame (Durata) :

Indicazioni :

FCG Basale
Terapia :

RISULTATI (Tutto)

FREQUENZA CARDIACA : (Totale QRS : 124618) (Durata Ora : 23:53)
Media : 87 bpm
Giorno (08:00 - 23:00) : 95 bpm
Notte (23:00 - 06:00) : 75 bpm
FC Max : 155 bpm a (1)08:39:51
FC Min : 49 bpm a (1)23:36:30
RR Max : 1230 ms a (1)23:01:09
RR Min : 375 ms a (1)08:38:57

BRADICARDIA : 0
PAUSE : 0
BATTITI MANCATI : 3
1/ a (1)14:20:33 RR = 1175ms
2/ a (1)17:49:38 RR = 1055ms
3/ a (1)12:27:38 RR = 965ms

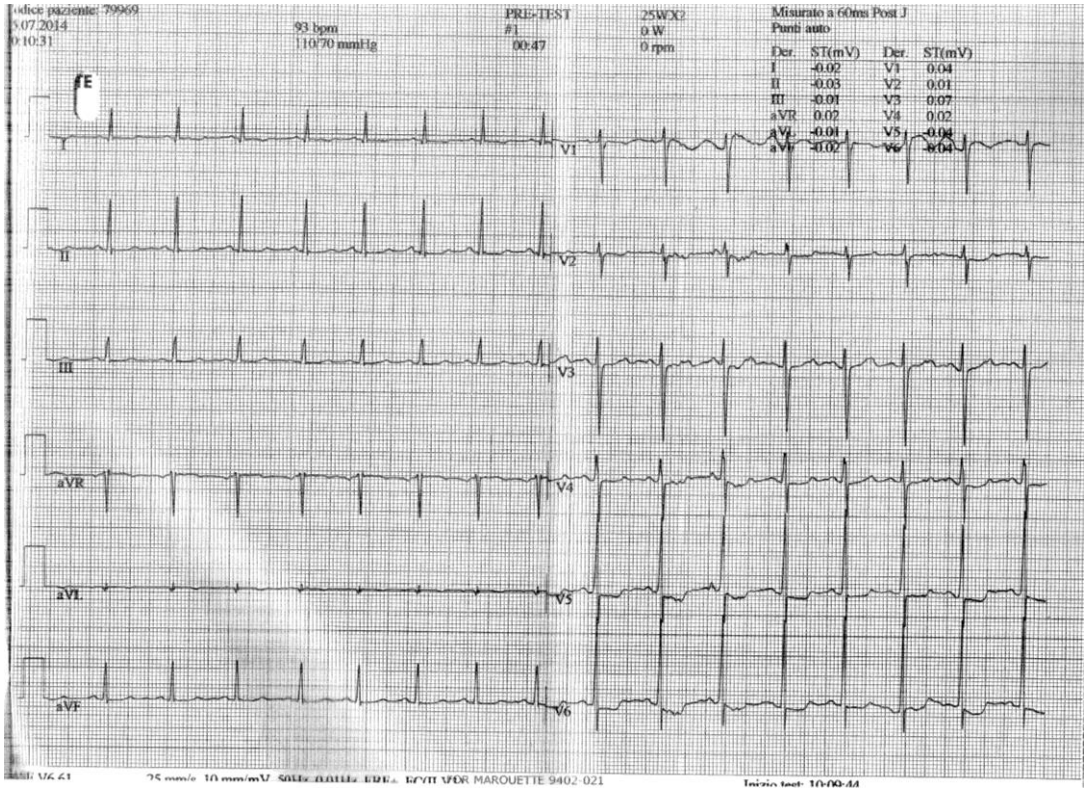
EPISODI VENTRICOLARI :
BATTITI ECTOP. :
Isolati : 846 0.7 %
Coppie : 2 0.0 %
Salve : 2 0.0 %
Totale : 856
BI & TRIGEMIN. : 28 & 2
Durata Totale : (1)00:02:28
1/ a (1)19:45:02 : Durata : (1)00:00:06
2/ a (1)20:00:20 : Durata : (1)00:00:06
3/ a (1)20:00:03 : Durata : (1)00:00:05
TACHICARDIA : 0

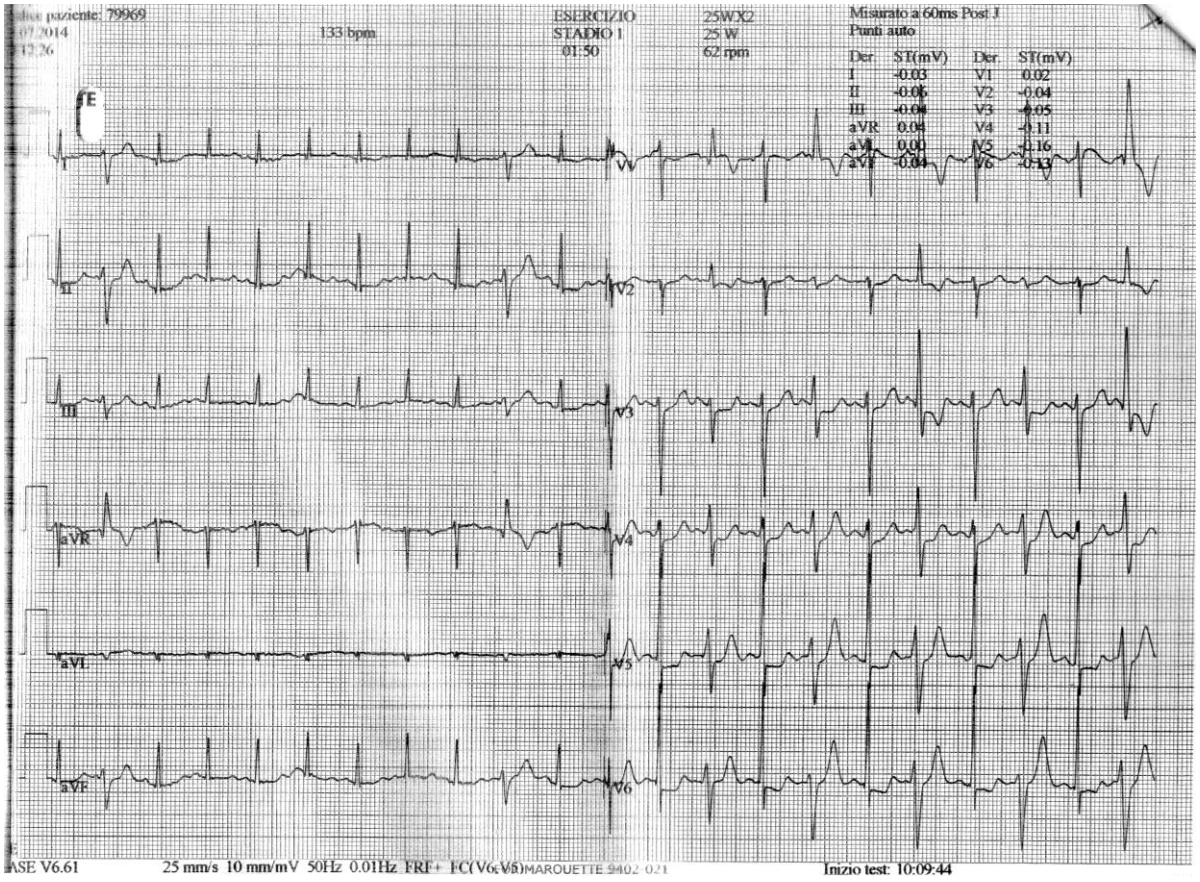
EPISODI SOPRAVENTRICOLARI :
BATTITI ECTOP. :
Isolati : 0 0.0 %
Coppie : 14 0.0 %
Salve : 16 0.0 %
Totale : 116
BI & TRIGEMIN. : 0 & 0
TACHICARDIA : 1
Durata Totale : (1)00:00:05 (0.0%)
1/ a (1)08:18:05 : 155 bpm (1)00:00:05
INSTABILITA' RR : 1
Durata Totale : (1)00:06:56 (0.5%)
1/ a (1)23:35:03 : Durata : (1)00:06:56

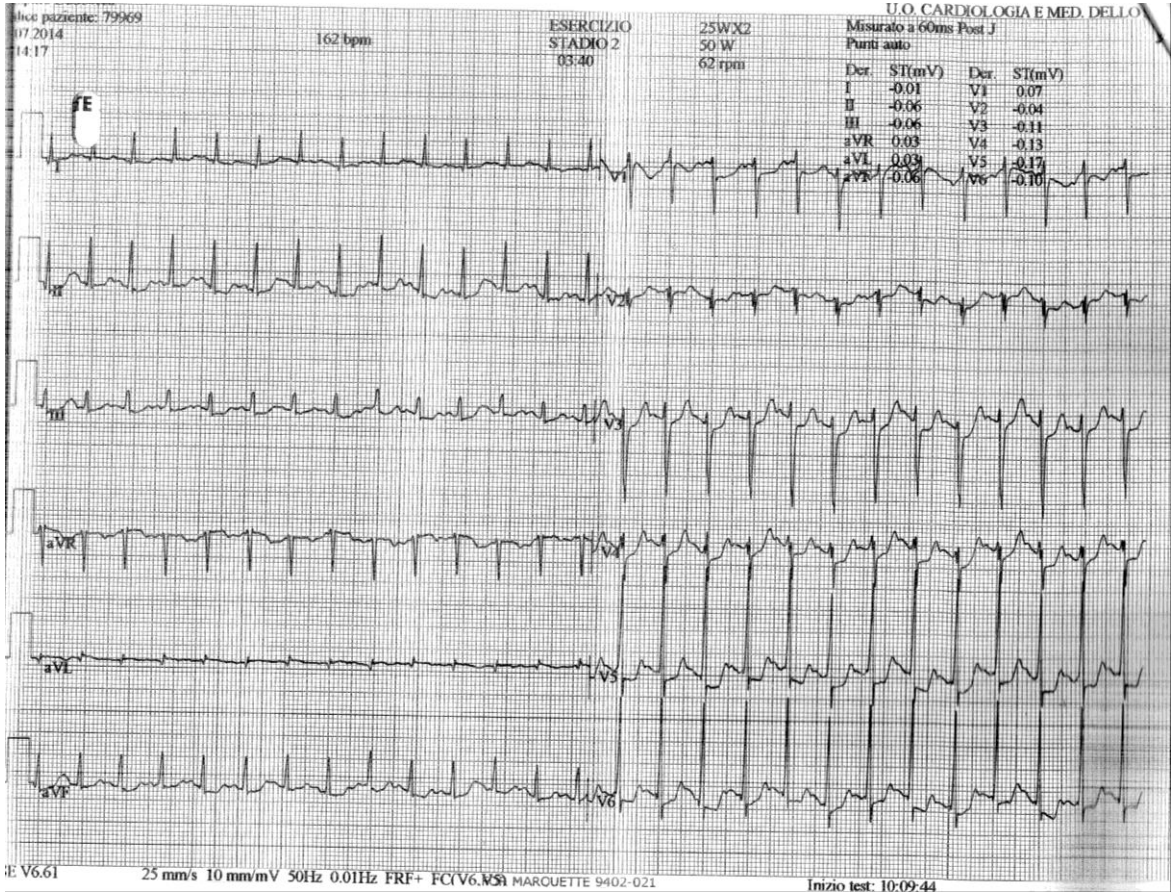
COMMENTI

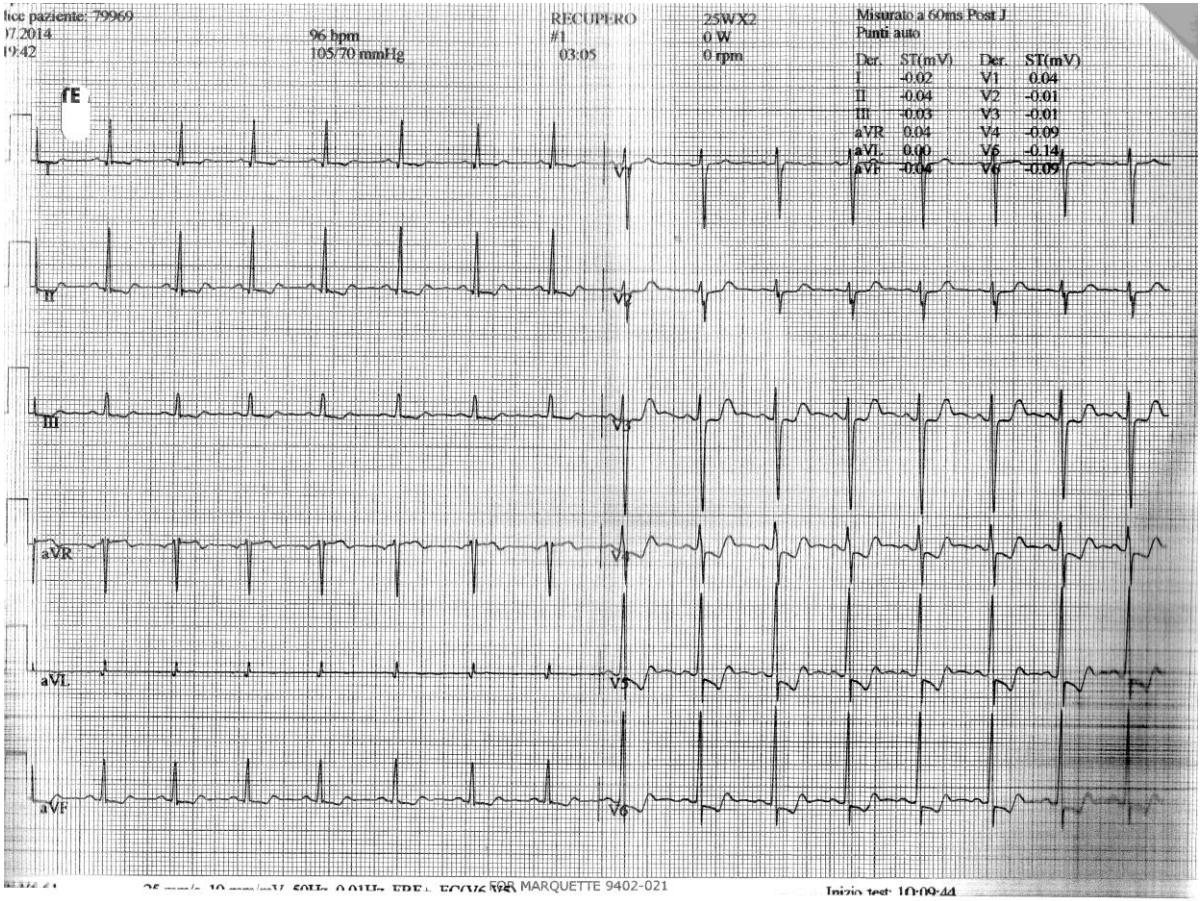
Aritmia extrasistolica ventricolare.Si segnalano alcune coppie di BEV,tratti di bigeminismo e due episodi di TV lenta non sostenuta (max 8 battiti) Alcune aberranze..TRV

ECG stress test 07/14




dice paziente: 79969
07.2014
12:26
133 bpm
ESERCIZIO
STADIO 1
01:50
25WX2
25 W
62 rpm
Misurato a 60ms Post J
Punti auto
Der. ST(mV) Der. ST(mV)
I -0.03 V1 0.02
II -0.05 V2 -0.04
III -0.04 V3 -0.05
aVR 0.04 V4 -0.11
aVL 0.00 V5 -0.16
aVF -0.04 V6 -0.13
FE
I
II
III
aVR
aVL
aVF
V1
V2
V3
V4
V5
V6
ASE V6.61
25 mm/s 10 mm/mV 50Hz 0.01Hz FRF+ FC(V6;V5)MARQUETTE 9402 021
Inizio test: 10:09:44


U.O. CARDIOLOGIA E MED. DELLO
lice paziente: 79969
07.2014
14:17
162 bpm
ESERCIZIO
STADIO 2
03:40
25WX2
50 W
62 rpm
Misurato a 60ms Post J
Punti auto
Der. ST(mV) Der. ST(mV)
I -0.01 V1 0.07
II -0.06 V2 -0.04
III -0.06 V3 -0.11
aVR 0.03 V4 -0.13
aVL 0.03 V5 -0.17
aVF -0.06 V6 -0.10
I
II
III
aVR
aVL
aVF
V1
V2
V3
V4
V5
V6
E V6.61
25 mm/s 10 mm/mV 50Hz 0.01Hz FRF+ FC(V6.MS)
MARQUETTE 9402-021
Inizio test: 10:09:44

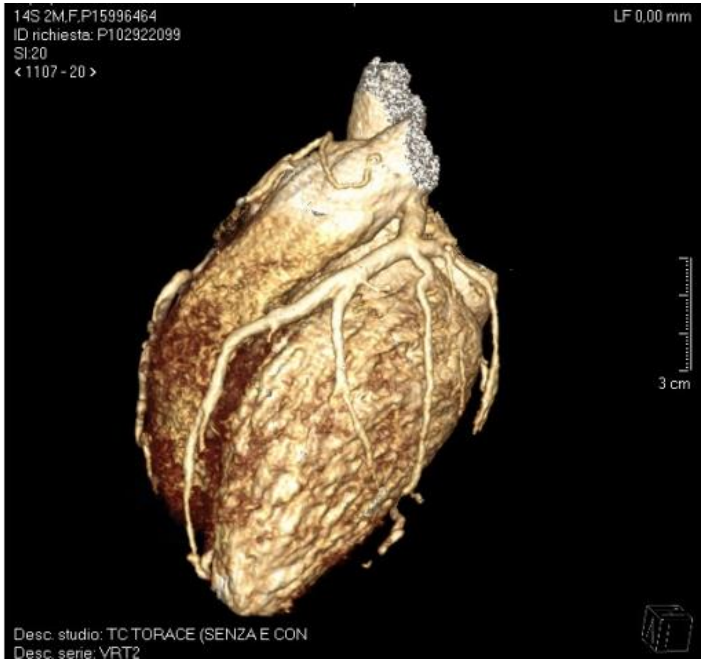

ice paziente: 79969
7.2014
9:42
96 bpm
105/70 mmHg
RECUPERO
#1
03:05
25WX2
0 W
0 rpm
Misurato a 60ms Post J
Punti auto
Der. ST(mV) Der. ST(mV)
I -0.02 V1 0.04
II -0.04 V2 -0.01
III -0.03 V3 -0.01
aVR 0.04 V4 -0.09
aVL 0.00 V5 -0.14
aVF -0.04 V6 -0.09
I
II
III
aVR
aVL
aVF
V1
V2
V3
V4
V5
V6
FOR MARQUETTE 9402-021
Inizio test: 10:09:44

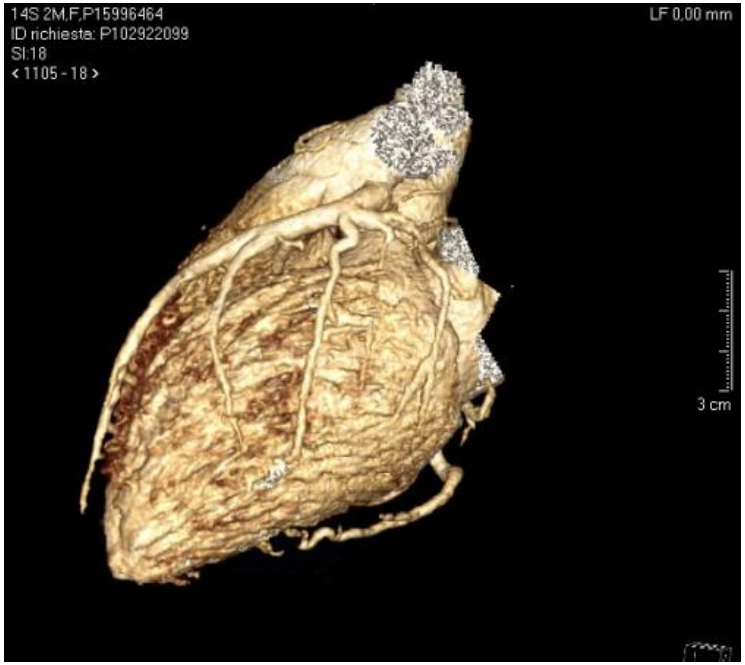
TC CUORE (SENZA E CON CONTRASTO)
TC ADDOME SUP. (SENZA E CON CONTRASTO)
TC TORACE (SENZA E CON CONTRASTO)

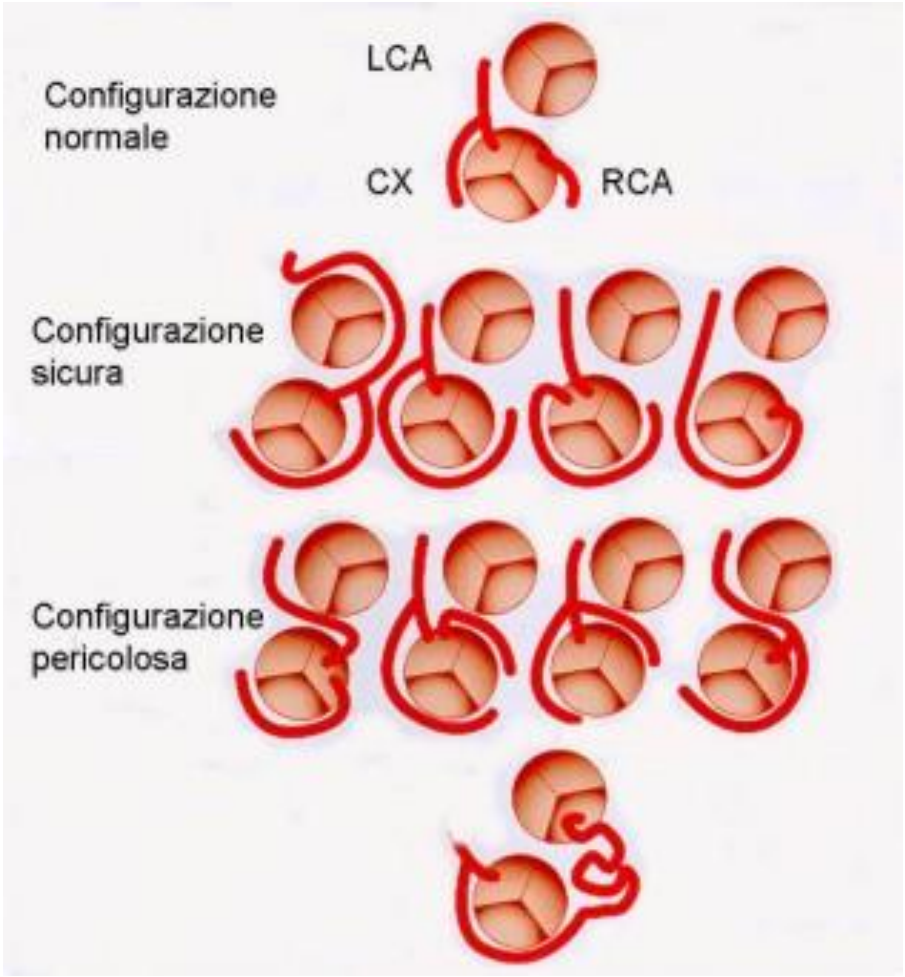
Referto

Esame eseguito con tecnica sequenziale multidetettore in corso di cardiosincronizzazione ECG durante infusione ev a bolo di circa 75 cc di mdc organoiodato (Iopamiro 370). Non complicanze in acuto.
FC durante la scansione compresa tra 75-81.
Non complicanze in acuto.

Si documenta origine anomala della coronaria sinistra dall'arteria polmonare (ALCAPA) a livello dell parete inferiore del tronco.
Normale origine e decorso della coronaria destra dominante che appare marcatamente diffusament dilatata e tortuosa e dalla quale originano grossolani e ubiquitari circoli collaterali per il sistema di sinistra sia epicardici e sia intramuscolari (alcuni dei quali a decorso perforante nel setto e a livello della trabecolatura del ventricolo destro).
Si segnala ramo per il nodo seno-atriale con origine dalla circonflessa.
Cavità ventricolare sinistra di dimensioni lievemente aumentate.
Non ulteriori reperti TC di rilievo nel volume esaminato.



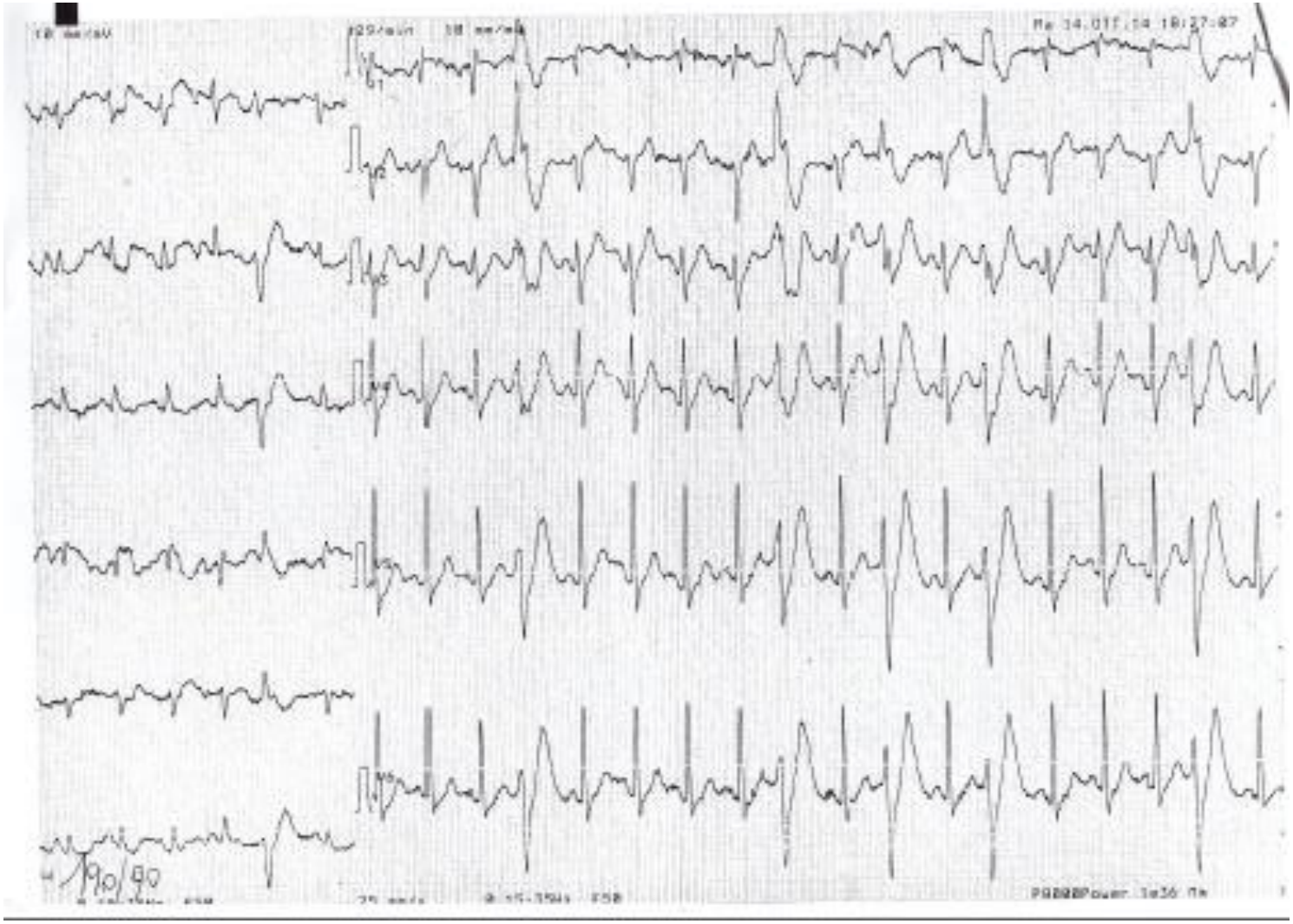




Benign (80%)	Incidence (%)	Anomalies (%)
Separate or adjacent LCX and LAD ostia	0.4	30
Separate or adjacent LCX and LAD ostia	0.4	30
LCX from PSV	<0.01	0.3
Absent LCX	0.003	0.2
Clinically Significant (20%)		
LMCA from RSV	0.02	1
LAD from RSV	0.03	2
RCA from LSV	0.10	10
LMCA from PA	<0.01	<1
LAD or RCA from PA	<0.01	<1

Mayo Clinic Practice of Cardiology. 3rd ed. St Louis, MO: Mosby; 1996:1815-1836.

L.G. 48 anni
Maschio
Podista agonista
Familiarità per cardiopatia ischemica
Ipertensione arteriosa
Pregressa meniscectomia



Extrasistolia ventricolare non soppressa dallo sforzo al test ergometrico.

RISULTATI (Tutto)

FREQUENZA CARDIACA : (Totale QRS : 95560) (Durata Ora : 22:38)

Media : 70 bpm
Giorno (08:00 - 21:00) : 70 bpm
Notte (23:00 - 06:00) : 67 bpm

FC Max : 125 bpm a (1)16:36:21
FC Min : 53 bpm a (1)18:36:06

RR Max : 1365 ms a (1)02:36:52
RR Min : 475 ms a (1)16:36:19

BRADICARDIA : 0

PAUSE : 0

BATTITI MANCATI : 3
1/a (1)10:44:11 RR = 1265ms
2/a (1)04:18:42 RR = 1240ms
3/a (1)02:24:43 RR = 1115ms

EPISODI VENTRICOLARI :

BATTITI ECTOP. :
Isolati : 4521 4.7 %
Coppie : 71 0.1 %
Salve : 0 0.0 %
Totale : 4663

BI & TRIGEMIN. : 7 & 72
Durata Totale : (1)00:12:08
1/a (1)00:40:32 : Durata : (1)00:00:17
2/a (1)00:51:38 : Durata : (1)00:00:16
3/a (1)23:06:13 : Durata : (1)00:00:14

TACHICARDIA : 0

EPISODI SOPRAVENTRICOLARI :

BATTITI ECTOP. :
Isolati : 60 0.1 %
Coppie : 1 0.0 %
Salve : 0 0.0 %
Totale : 62

BI & TRIGEMIN. : 0 & 0

TACHICARDIA : 0

INSTABILITA' RR : 0

COMMENTI

Sono state analizzate 23 ore di registrazione.
Il ritmo è stato sinusale con FC oscillante tra 125-53 bpm. media diurna 70 bpm, notturna 67 bpm.
Rari BESV isolati, 1 coppia.
Frequenti BEV isolati, prevalentemente monomorfi, 71coppie.
Limitatamente alle 3 derivazioni esplorate e alla Fc max raggiunta non sono state evidenziate modificazioni della ripolarizzazione ventricolare.

Referto:

INDICAZIONI

Aritmia Ventricolare.Bev.

TECNICA

Qualità: Buono

Sequenza: Ssfp, T2bb, Dyn fee, Irtfe,.

Peso:81,00, Altezza:175,00, Bsa:1,98, Bmi:26,45..

VALUTAZIONE FUNZIONALE VENTRICOLO SINISTRO

Lvdev: ml 157,00, Lves: ml 45,00, LVEDVind: ml/m2 79,00, LVESind: ml/m2 23,00, Lvsv: ml112,00, Pc: litri/min 8,30, Diam. telediag.: mm 53,00, Diam. telesist.: mm 28,00, Lvef: % 71

VALUTAZIONE FUNZIONALE VENTRICOLO DESTRO

Rvedv: ml 149,00, Rves: ml 42,00, Rvedvind: ml/m2 75,00, Rvesind: ml/m2 21,00, Rvsv: ml/m2 108,00, Pc: litri/min 8,00, Rvef: % 72.

MORFOLOGIA CARDIACA

Ventricolo Sinistro: Volumi, dimensioni e spessori parietali nei limiti. Funzione sistolica globale conservata,

Ventricolo Destro: volumi e funzione sistolica globale nei limiti,

Valvola Mitrale:jet di rigurgito

Atrio sinistro: 22 cmq

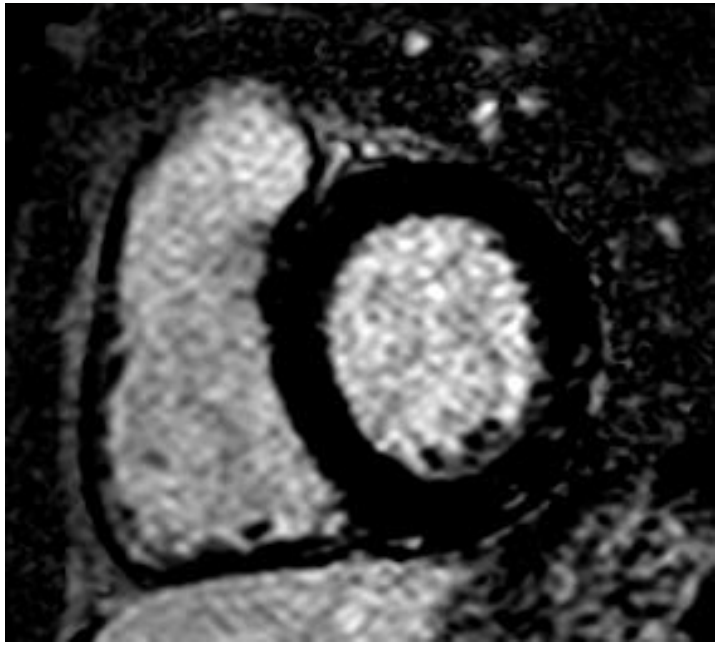
Atrio destro: 20 cmq

POTENZIAMENTO MIOCARDICO

Dopo somministrazione di mdc si osserva puntiformi aree di potenziamento subepicardiche ed intramiocardiche in sede corrispodenza della parete inferiroe in sede basale.

PERICARDIO

Minima falda fluida in sede infero-laterale



CORONAROGRAFIA

Pressioni Aorta: 130 / 90 () **Pressioni Vsin:** / ()

Circolo Coronarico: Dominanza destra

Coronaria Sinistra:
Tronco comune: esente da alterazioni
Arteria discendente anteriore: grossolane irregolarità nel tratto medio; stenosi al 65-70% all'origine del primo ramo diagonale (FFR 0.88)
Arteria circonflessa: stenosi ostiale all'80% (FFR 0.67).

Coronaria Destra:
Esente da significative alterazioni

Diagnosi:
Coronarie normali.
Malattia di un vaso coronarico.

Conclusioni:
Aterosclerosi coronarica con impegno critico di un ramo principale (arteria circonflessa) e subcritico di un ramo secondario (ramo diagonale). Eseguita angioplastica con stent medicato su arteria circonflessa.

Indirizzo Terapeutico: Angioplastica

ANGIOPLASTICA CORONARICA

N° Lesioni Trattate: 1 CFx Prox

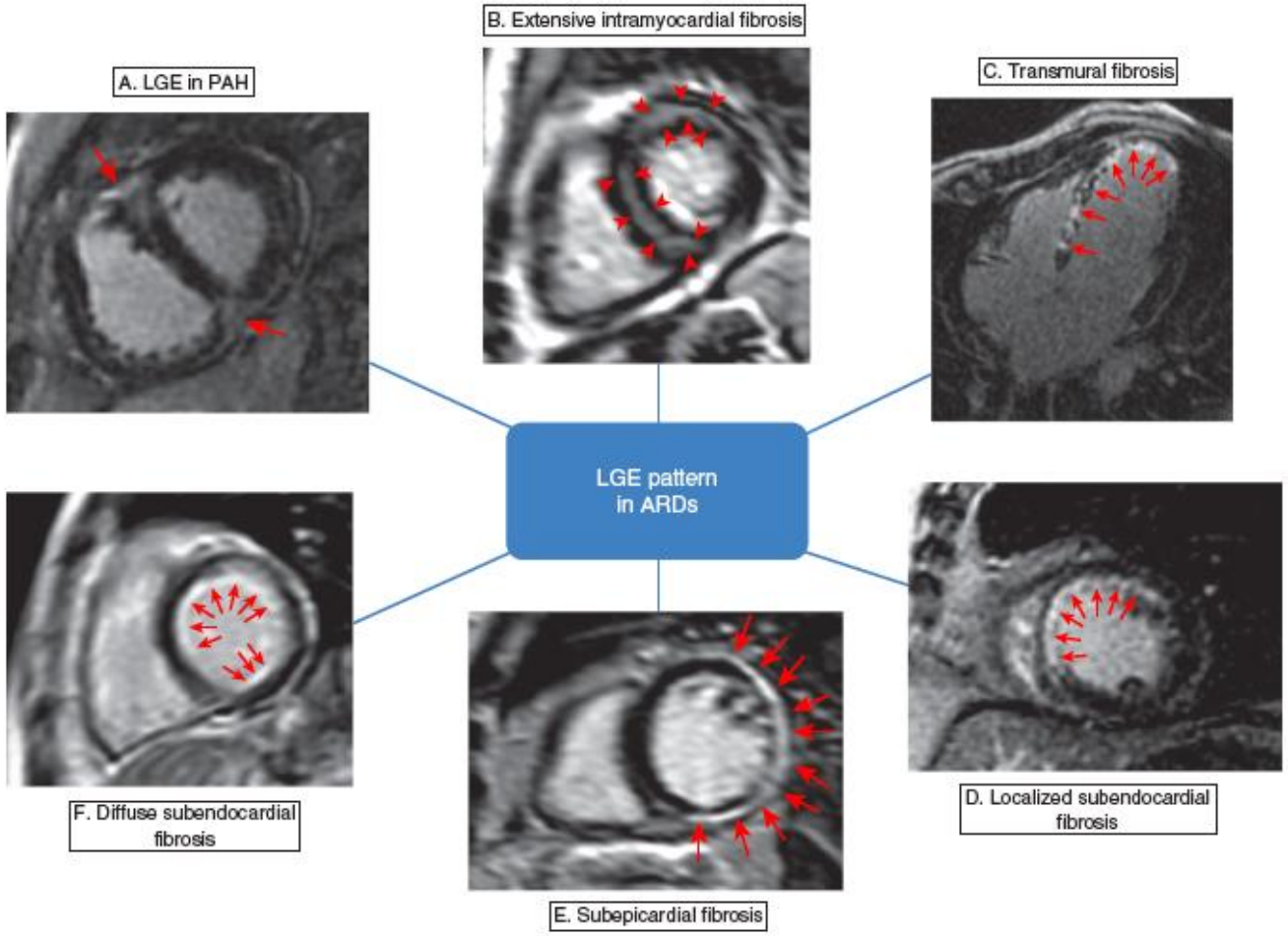
Efficacia: Efficace

Conclusioni:
Si cannula l'ostio della coronaria sinistra con catetere guida CLS 3,5 (Mach1, Boston) e si supera con guida da 0.014" (Verrata Pressure Wire, Volcano) la stenosi critica all'ostio dell'arteria circonflessa. Si impianta direttamente stent medicato 3,0 x 18 mm espanso a 18 atmosfere e postdilatato con pallone non copmpliante 3,25 x 8 mm (Quantum, Boston) espanso a 18 atmosfere. Buon risultato angiografico finale con flusso TIMI III.

LESIONI TRATTATE

CFx Prox **Trattamento:** Stenosi	**STENOSI: PRE** 80 % **POST** 0 %	**TIMI: PRE** 3 **POST** 3

TIPOLOGIA	Ellis B2
Dati Tecnici	Stent: Post-dilatazione Guidina: Lesione Facilmente Superata Stent: Direct Stenting Efficace


B. Extensive intramyocardial fibrosis
A. LGE in PAH
C. Transmural fibrosis
LGE pattern
in ARDs
F. Diffuse subendocardial
fibrosis
D. Localized subendocardial
fibrosis
E. Subepicardial fibrosis

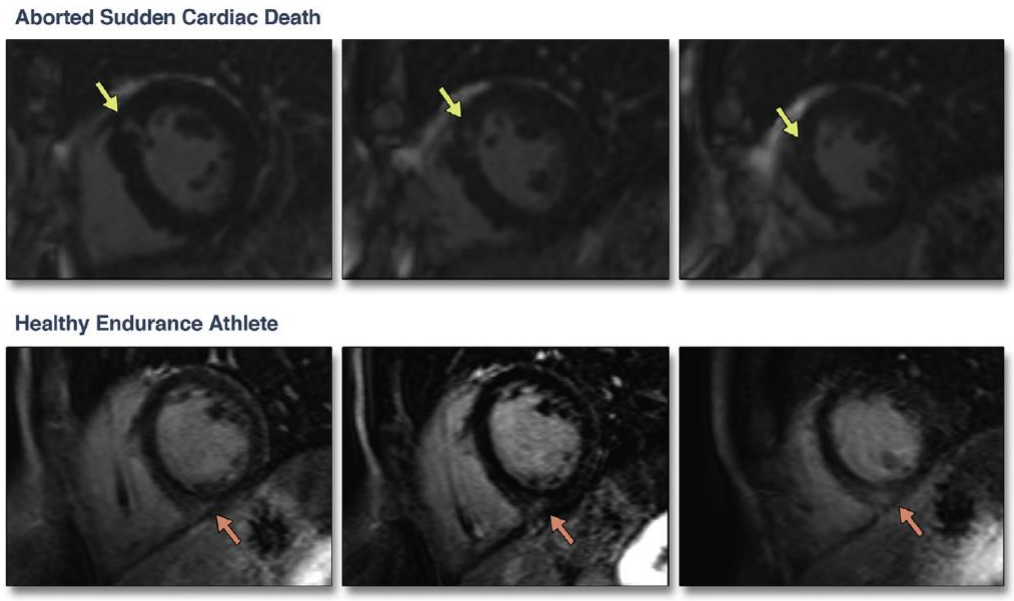


Figure 4. Delayed Gadolinium Enhancement in 2 Competitive Runners With Very Different Clinical Outcomes

Cardiac magnetic resonance images of consecutive short-axis slices following gadolinium contrast are demonstrated. The 3 images are taken from a case report of an athlete who survived sudden cardiac death. The authors speculated that the patch of delayed gadolinium enhancement in the anteroseptal wall of the left ventricle (LV) **(yellow arrows)** may represent pro-arrhythmic myocardial fibrosis resulting from extreme exercise training (65). On the other hand, similar findings have been found in 12 to 50% of well-trained endurance athletes (33,60,61). In the case of the apparently healthy 36-year-old marathon runner undergoing screening in this review, there is a similar small patch of delayed gadolinium enhancement in the posteroseptum of the LV. Given the uncertainty of the significance of these findings, identification of small patches of delayed gadolinium enhancement on screening may be of limited clinical value in older endurance athletes.

Images in Cardiovascular Medicine

Nonischemic Left Ventricular Scar

Sporadic or Familial? Screen the Genes, Scan the Mutation Carriers

Kalliopi Pilichou, PhD; Massimiliano Mancini, MD; Ilaria Rigato, MD, PhD; Elisabetta Lazzarini, BSc; Benedetta Giorgi, MD; Elisa Carturan, PhD; Barbara Bauce, MD, PhD; Giulia d'Amati, MD, PhD; Martina Perazzolo Marra, MD, PhD; Cristina Basso, MD, PhD

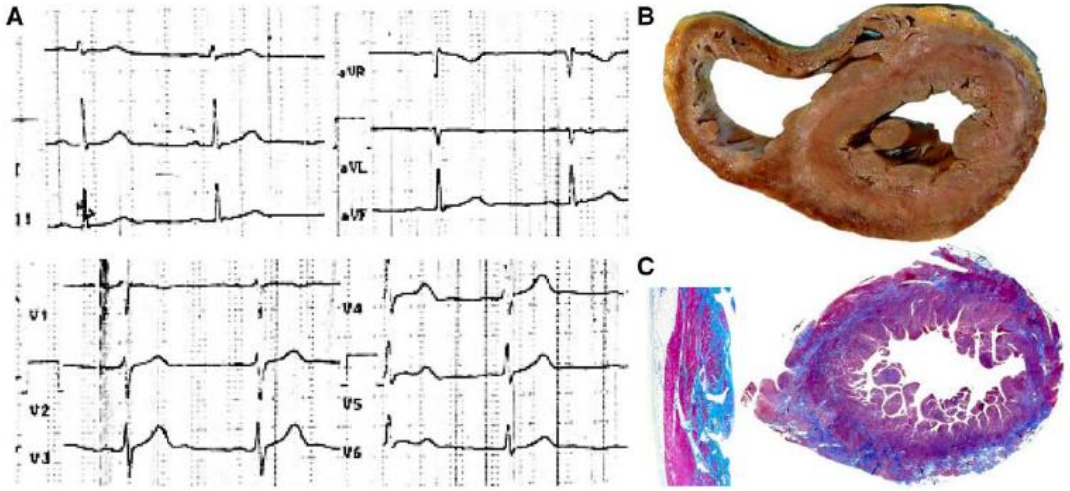


Figure 1. A, Basal 12-lead ECG at annual preparticipation screening showing normal findings. **B**, Transverse section of the heart showing a subepicardial scar-like grey rim in the anterolateral and posterior LV free wall and in the septum, in the absence of wall thinning and aneurysm formation. **C**, Histological examination revealed focal RV involvement (on the left) and extensive circumferential, subepicardial, and intramural fibrous replacement of the LV free wall (on the right). LV indicates left ventricle; and RV, right ventricle.

(*Circulation*. 2014;130:e180-e182.)

Truncating *FLNC* Mutations Are Associated With High-Risk Dilated and Arrhythmogenic Cardiomyopathies

Martín F. Ortiz-Genga, MD,[a,b] Sofía Cuenca, MD, PhD,[c] Matteo Dal Ferro, MD,[d] Esther Zorio, MD, PhD,[e] Ricardo Salgado-Aranda, MD,[f] Vicente Climent, MD,[g] Laura Padrón-Barthe, PhD,[h] Iria Duro-Aguado, MD,[i] Juan Jiménez-Jáimez, MD, PhD,[j] Víctor M. Hidalgo-Olivares, MD,[k] Enrique García-Campo, MD,[l] Chiara Lanzillo, MD, PhD,[m] M. Paz Suárez-Mier, MD, PhD,[n] Hagith Yonath, MD,[o] Sonia Marcos-Alonso, MD, PhD,[p] Juan P. Ochoa, MD,[b] José L. Santomé, BSc,[b] Diego García-Giustiniani, MD,[b] Jorge L. Rodríguez-Garrido, MD,[b,p] Fernando Domínguez, MD,[c] Marco Merlo, MD,[d] Julián Palomino, MD, PhD,[i] María L. Peña, MD,[q] Juan P. Trujillo, MD, PhD,[b] Alicia Martín-Vila, PharmD,[i] Davide Stolfo, MD,[d] Pilar Molina, MD, PhD,[r] Enrique Lara-Pezzi, PhD,[h,s] Francisco E. Calvo-Iglesias, MD, PhD,[i] Eyal Nof, MD,[o] Leonardo Calò, MD,[m] Roberto Barriales-Villa, MD, PhD,[a,p] Juan R. Gimeno-Blanes, MD, PhD,[t] Michael Arad, MD, PhD,[o] Pablo García-Pavía, MD, PhD,[c,u] Lorenzo Monserrat, MD, PhD[a,b]

ABSTRACT

BACKGROUND Filamin C (encoded by the *FLNC* gene) is essential for sarcomere attachment to the plasmatic membrane. *FLNC* mutations have been associated with myofibrillar myopathies, and cardiac involvement has been reported in some carriers. Accordingly, since 2012, the authors have included *FLNC* in the genetic screening of patients with inherited cardiomyopathies and sudden death.

OBJECTIVES The aim of this study was to demonstrate the association between truncating mutations in *FLNC* and the development of high-risk dilated and arrhythmogenic cardiomyopathies.

METHODS *FLNC* was studied using next-generation sequencing in 2,877 patients with inherited cardiovascular diseases. A characteristic phenotype was identified in probands with truncating mutations in *FLNC*. Clinical and genetic evaluation of 28 affected families was performed. Localization of filamin C in cardiac tissue was analyzed in patients with truncating *FLNC* mutations using immunohistochemistry.

RESULTS Twenty-three truncating mutations were identified in 28 probands previously diagnosed with dilated, arrhythmogenic, or restrictive cardiomyopathies. Truncating *FLNC* mutations were absent in patients with other phenotypes, including 1,078 patients with hypertrophic cardiomyopathy. Fifty-four mutation carriers were identified among 121 screened relatives. The phenotype consisted of left ventricular dilation (68%), systolic dysfunction (46%), and myocardial fibrosis (67%); inferolateral negative T waves and low QRS voltages on electrocardiography (33%); ventricular arrhythmias (82%); and frequent sudden cardiac death (40 cases in 21 of 28 families). Clinical skeletal myopathy was not observed. Penetrance was >97% in carriers older than 40 years. Truncating mutations in *FLNC* cosegregated with this phenotype with a dominant inheritance pattern (combined logarithm of the odds score: 9.5). Immunohistochemical staining of myocardial tissue showed no abnormal filamin C aggregates in patients with truncating *FLNC* mutations.

CONCLUSIONS Truncating mutations in *FLNC* caused an overlapping phenotype of dilated and left-dominant arrhythmogenic cardiomyopathies complicated by frequent premature sudden death. Prompt implantation of a cardiac defibrillator should be considered in affected patients harboring truncating mutations in *FLNC*.
(J Am Coll Cardiol 2016;68:2440–51) © 2016 by the American College of Cardiology Foundation.

Left-Dominant Arrhythmogenic Cardiomyopathy

An Under-Recognized Clinical Entity

Srijita Sen-Chowdhry, MBBS, MD (Cantab), MRCP,*‡ Petros Syrris, PhD,*
Sanjay K. Prasad, MD, MRCP,‡ Siân E. Hughes, MBBS, PhD, MRCPath,†
Robert Merrifield, PhD,§ Deirdre Ward, MBBS, MRCPI,* Dudley J. Pennell, MD, FACC,‡
William J. McKenna, MD, DSc, FACC*

London, United Kingdom

Objectives We sought to investigate the clinical-genetic profile of left-dominant arrhythmogenic cardiomyopathy (LDAC).

Background In the absence of coronary disease and left ventricular (LV) systolic dysfunction, lateral T-wave inversion and arrhythmia of LV origin are often considered benign. Similarly, chest pain with enzyme release might be attributed to viral myocarditis. We hypothesized that these abnormalities might be manifestations of the "left-dominant" subtype of arrhythmogenic right ventricular cardiomyopathy.

Methods The 42-patient cohort was established through clinical evaluation of individuals with unexplained (infero)lateral T-wave inversion, arrhythmia of LV origin, and/or proven LDAC/idiopathic myocardial fibrosis in the family.

Results Patients presented from adolescence to age >80 years with arrhythmia or chest pain but not heart failure. Desmosomal mutations were identified in 8 of 24 families (15 of 33 patients). Magnetic resonance findings included LV late-enhancement in a subepicardial/midwall distribution, corresponding to fibrofatty replacement and fibrosis on histopathology. Fifty percent had previously been misdiagnosed with viral myocarditis, dilated cardiomyopathy (DCM), hypertrophic cardiomyopathy, or idiopathic ventricular tachycardia. Arrhythmic events included presentation with ventricular fibrillatory arrest in 1 patient and 2 instances of sudden cardiac death during follow-up.

Conclusions Arrhythmogenic cardiomyopathy is distinguished from DCM by a propensity towards arrhythmia exceeding the degree of ventricular dysfunction. The left-dominant subtype is under-recognized owing to misattribution to other disorders and lack of specific diagnostic criteria. Clinicians are alerted to the possibility of LDAC in patients of any age with unexplained arrhythmia of LV origin, (infero)lateral T-wave inversion, apparent DCM (with arrhythmic presentation), or myocarditis (chest pain and enzyme rise with unobstructed coronary arteries). (J Am Coll Cardiol 2008;52:2175–87) © 2008 by the American College of Cardiology Foundation

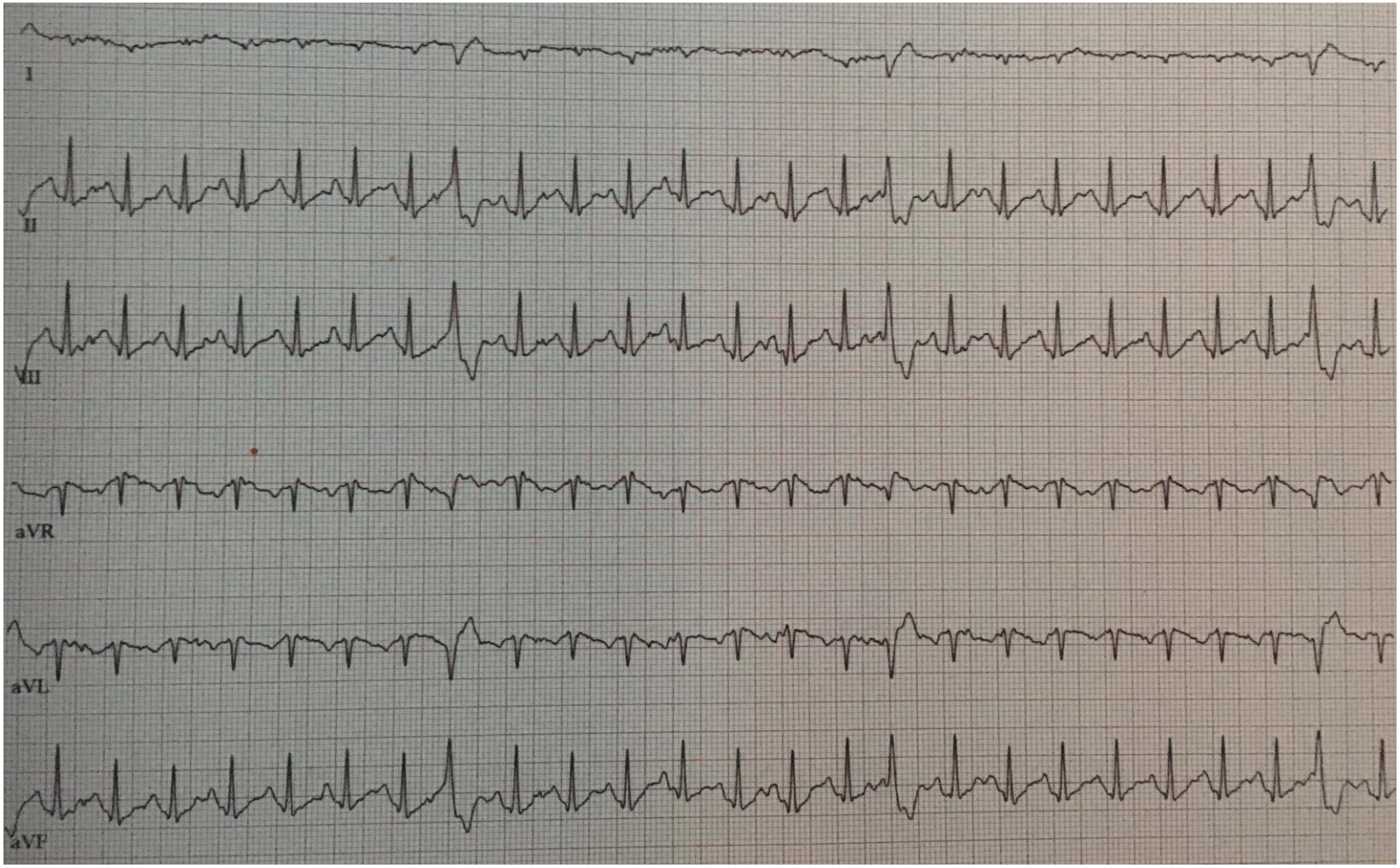
Genetic Variation in Titin in Arrhythmogenic Right Ventricular Cardiomyopathy–Overlap Syndromes

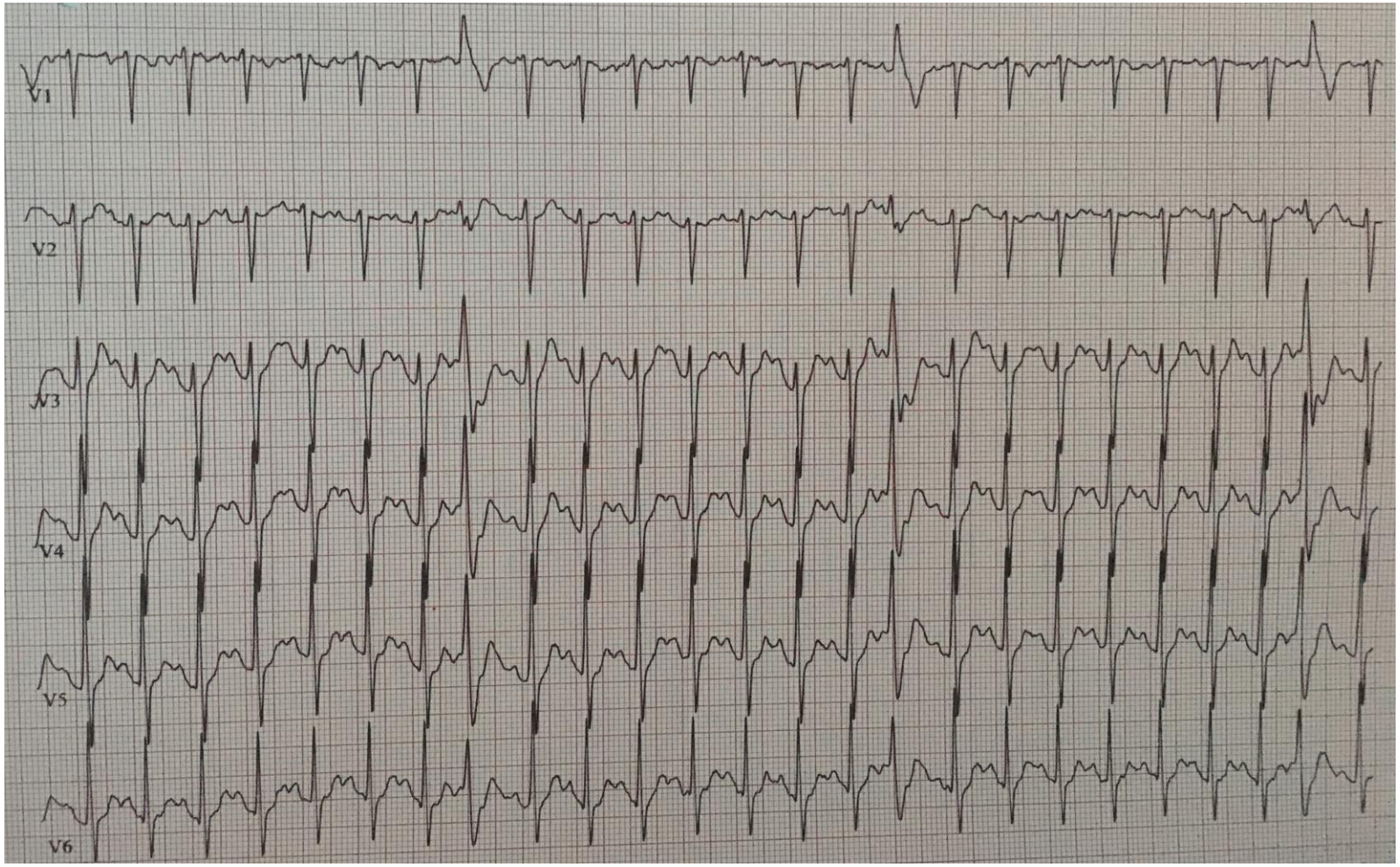
Matthew Taylor, MD, PhD; Sharon Graw, PhD; Gianfranco Sinagra, MD; Carl Barnes, MD; Dobromir Slavov, PhD; Francesca Brun, MD; Bruno Pinamonti, MD; Ernesto E. Salcedo, MD; William Sauer, MD; Stylianos Pyxaras, MD; Brian Anderson; Bernd Simon, PhD; Julius Bogomolovas, PhD; Siegfried Labeit, MD; Henk Granzier, PhD; Luisa Mestroni, MD

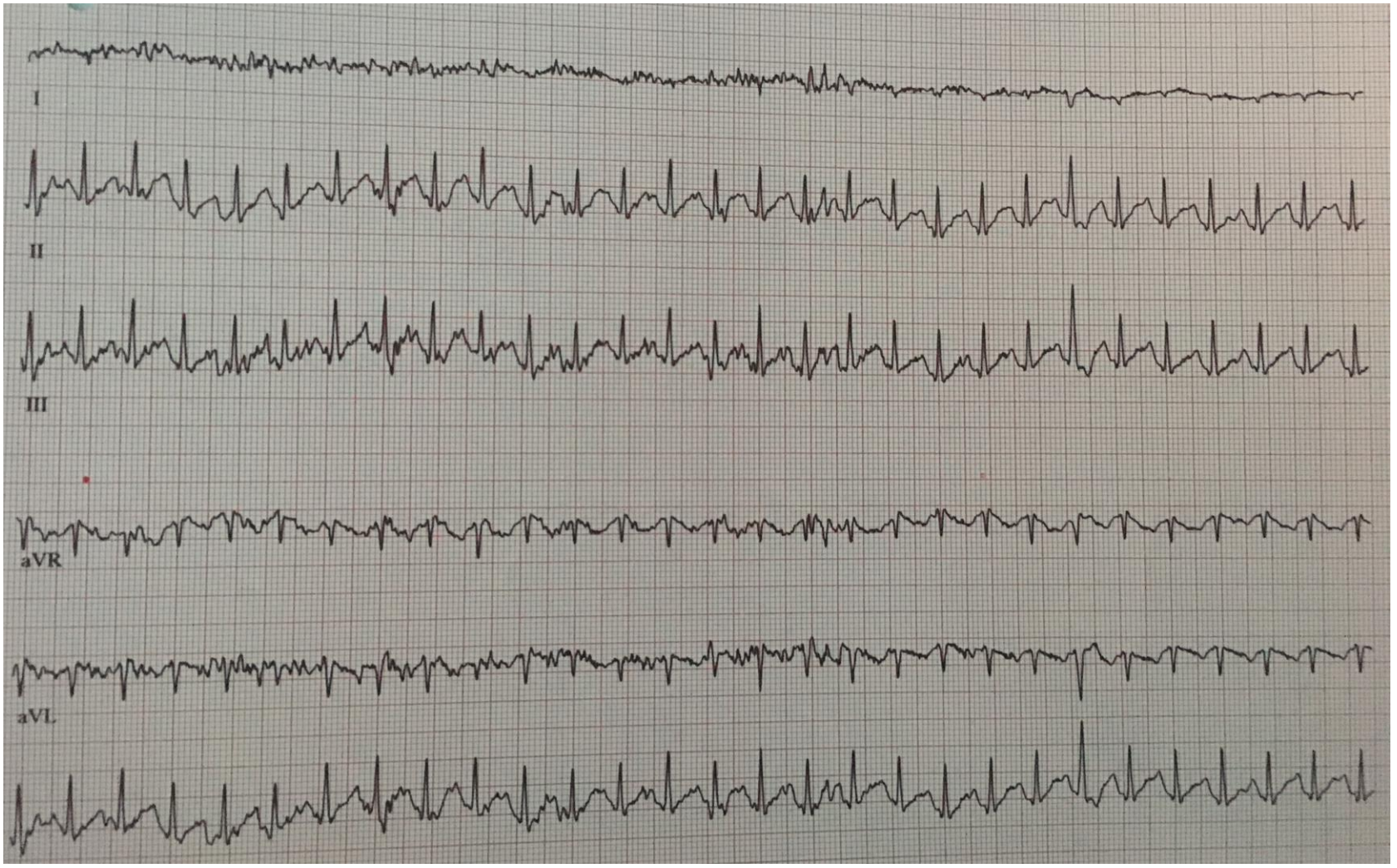
Background—Arrhythmogenic right ventricular cardiomyopathy (ARVC) is an inherited genetic myocardial disease characterized by fibrofatty replacement of the myocardium and a predisposition to cardiac arrhythmias and sudden death. We evaluated the cardiomyopathy gene titin (*TTN*) as a candidate ARVC gene because of its proximity to an ARVC locus at position 2q32 and the connection of the titin protein to the transitional junction at intercalated disks.

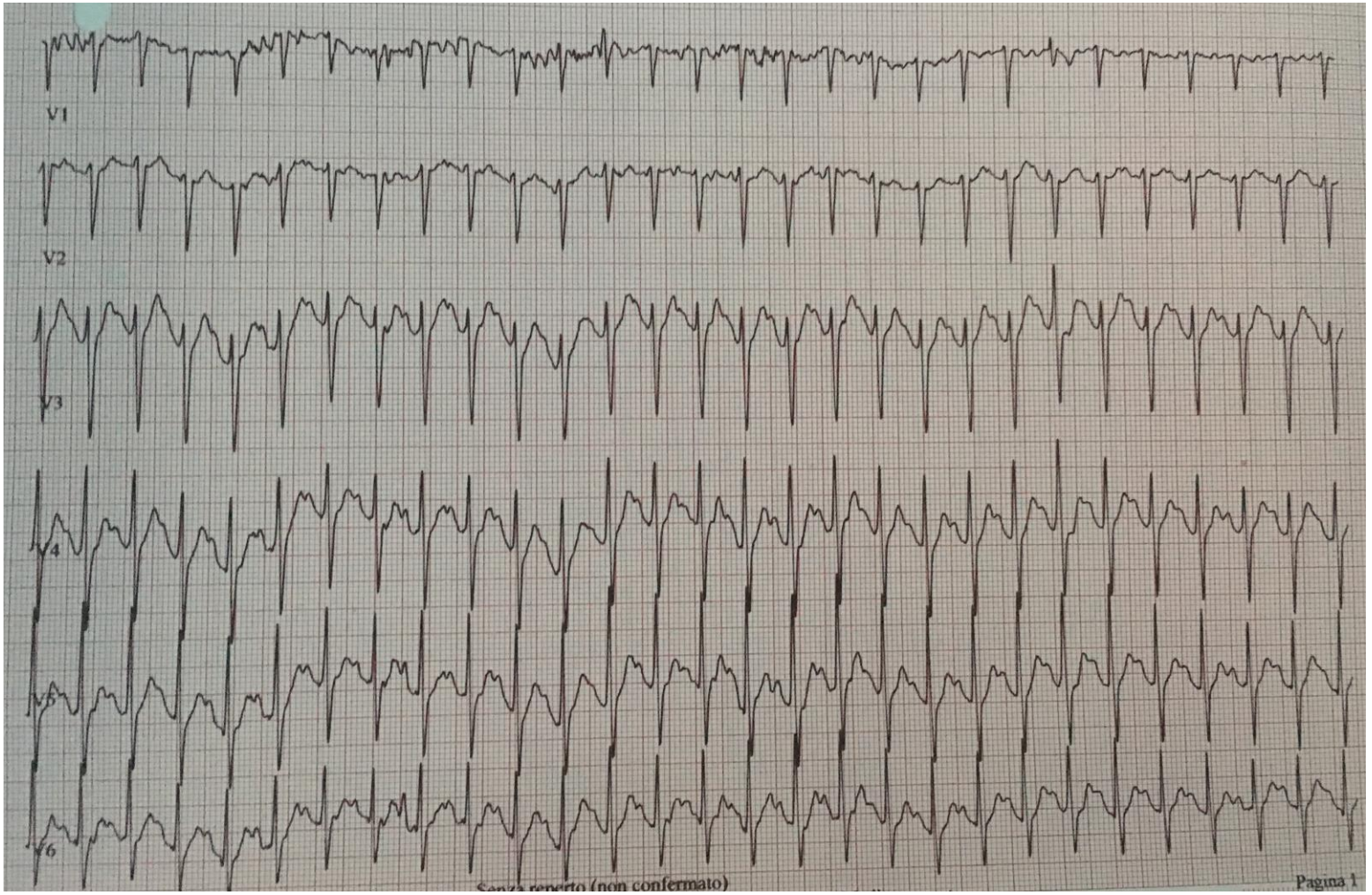
Methods and Results—All 312 titin exons known to be expressed in human cardiac titin and the complete 3′ untranslated region were sequenced in 38 ARVC families. Eight unique *TTN* variants were detected in 7 families, including a prominent Thr2896Ile mutation that showed complete segregation with the ARVC phenotype in 1 large family. The Thr2896Ile mutation maps within a highly conserved immunoglobulin-like fold (Ig10 domain) located in the spring region of titin. Native gel electrophoresis, nuclear magnetic resonance, intrinsic fluorescence, and proteolysis assays of wild-type and mutant Ig10 domains revealed that the Thr2896Ile exchange reduces the structural stability and increases the propensity for degradation of the Ig10 domain. The phenotype of *TTN* variant carriers was characterized by a history of sudden death (5 of 7 families), progressive myocardial dysfunction causing death or heart transplantation (8 of 14 cases), frequent conduction disease (11 of 14), and incomplete penetrance (86%).

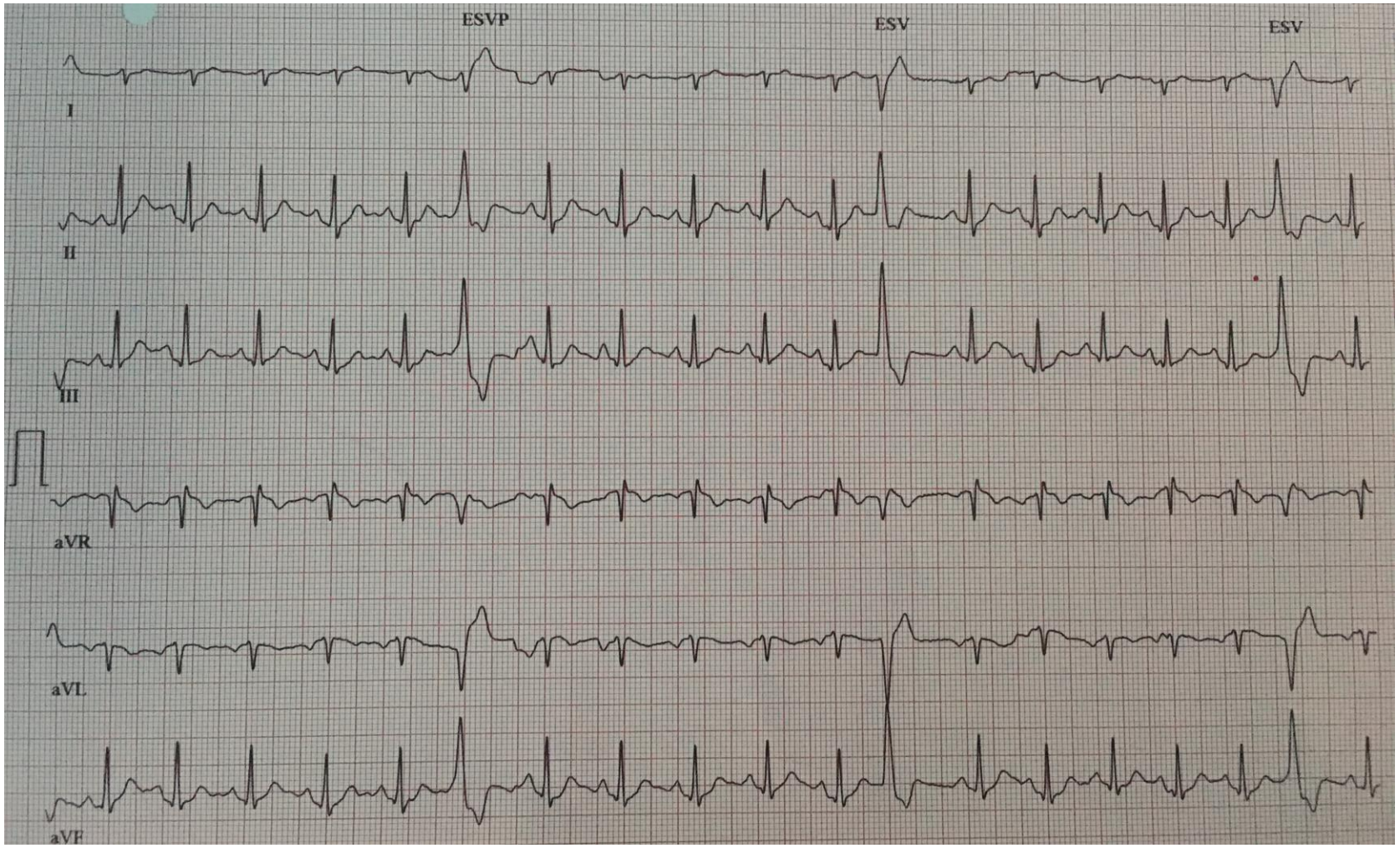
Conclusions—Our data provide evidence that titin mutations can cause ARVC, a finding that further expands the origin of the disease beyond desmosomal proteins. Structural impairment of the titin spring is a likely cause of ARVC and constitutes a novel mechanism underlying myocardial remodeling and sudden cardiac death. (***Circulation*. 2011;124:876-885.**)

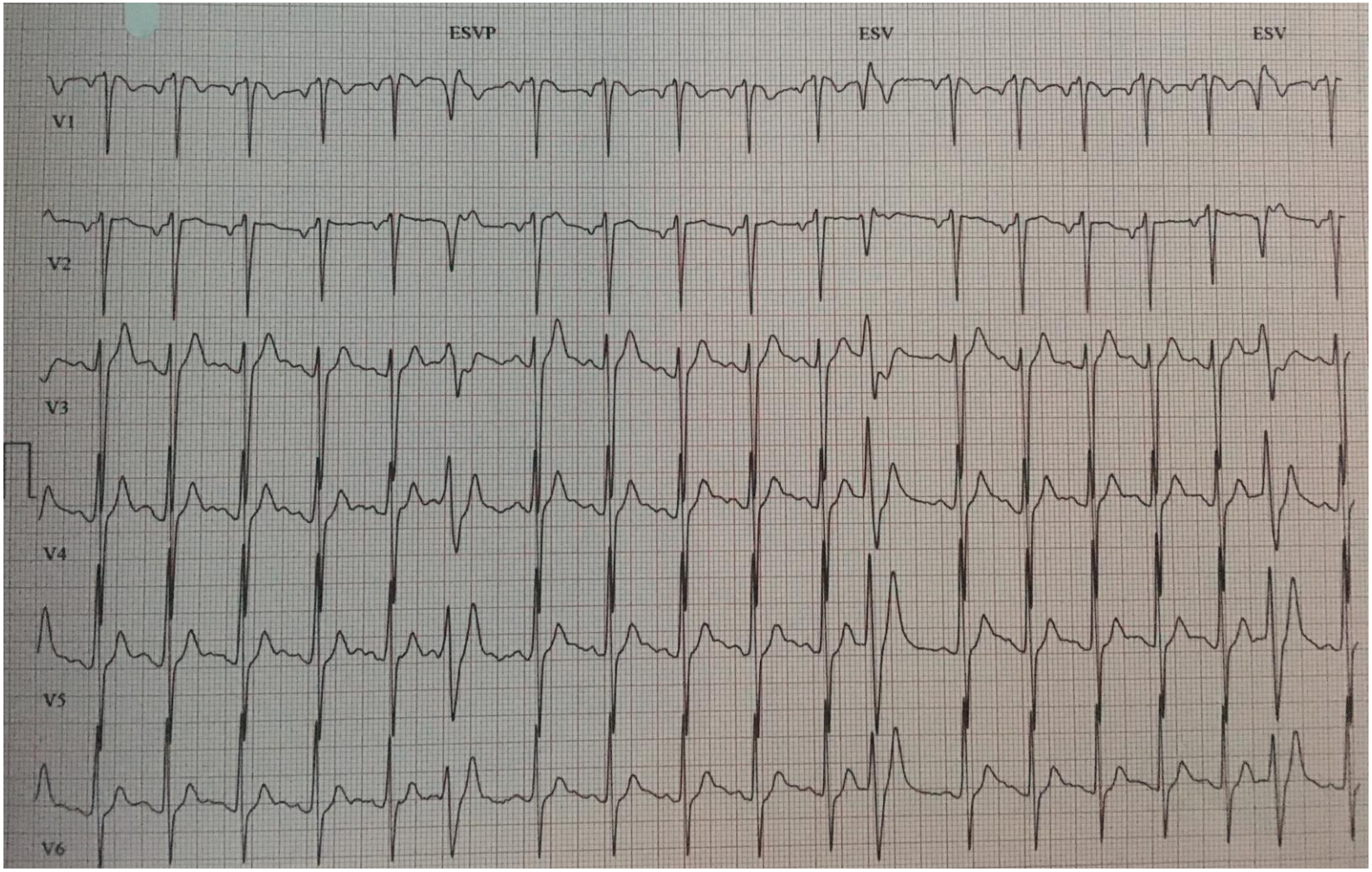

I
II
III
aVR
aVL
aVF

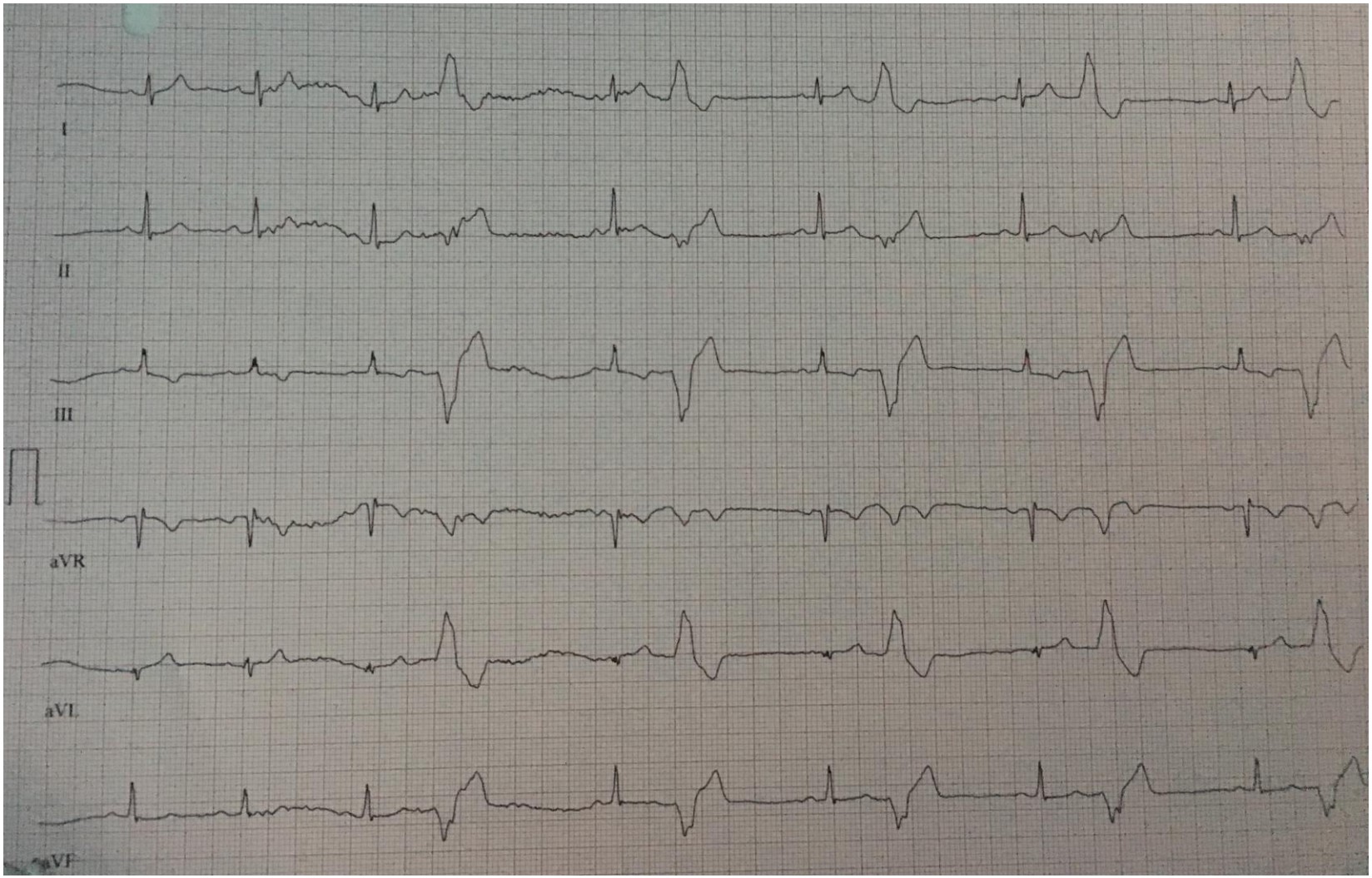

V1
V2
V3
V4
V5
V6

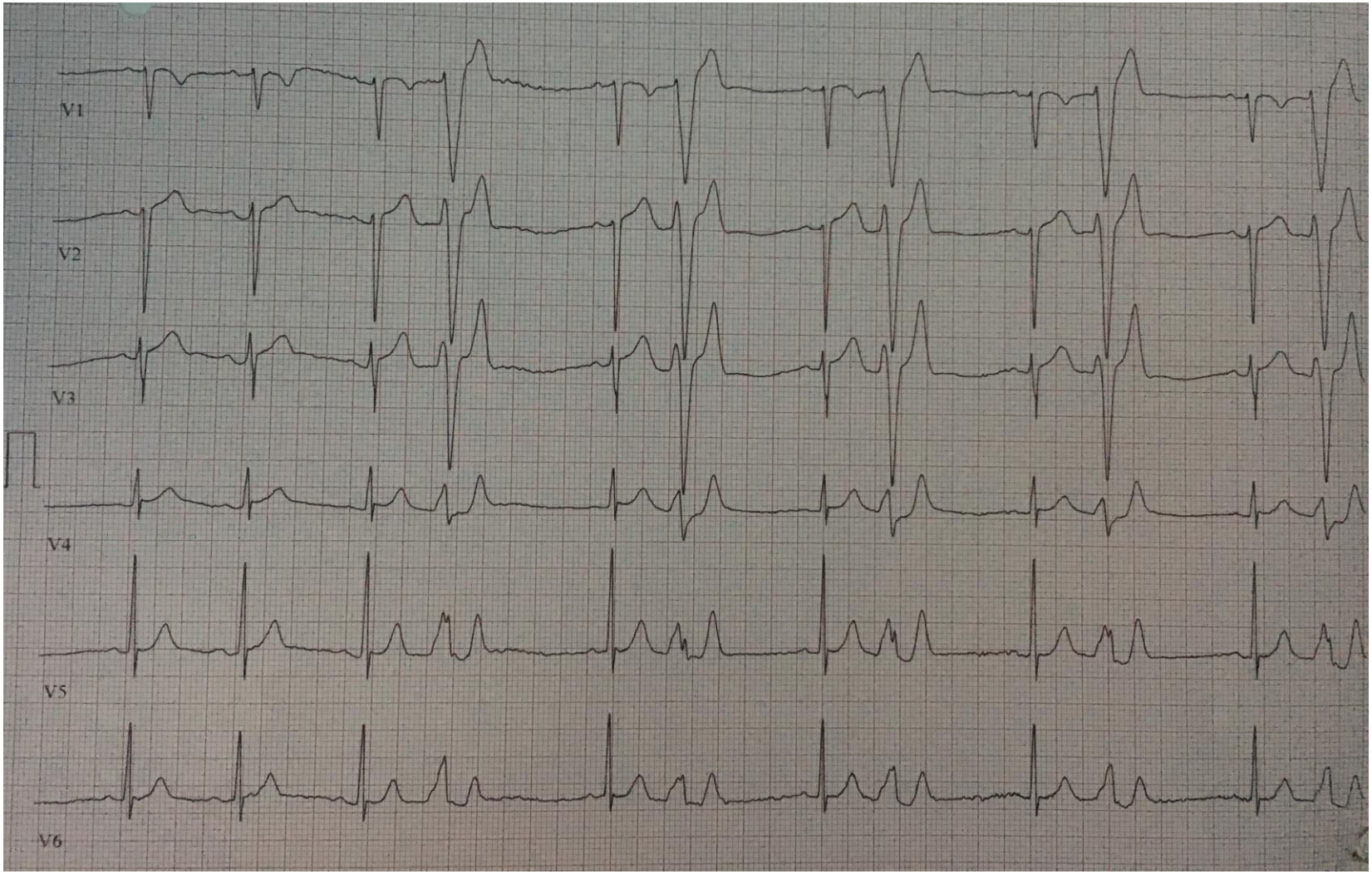

I
II
III
aVR
aVL

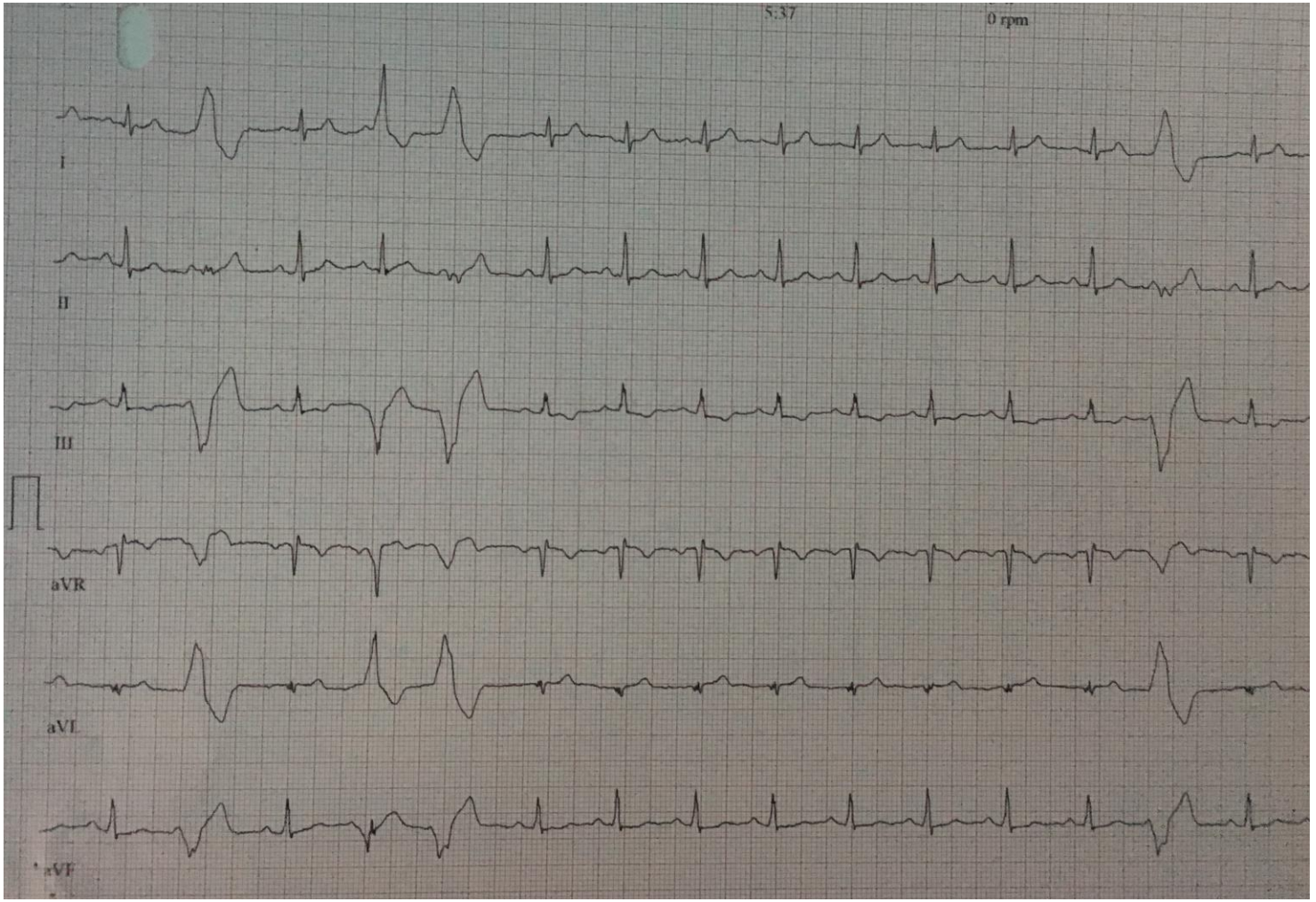

V1
V2
V3
V4
V5
V6
Senza reperto (non confermato)
Pagina 1

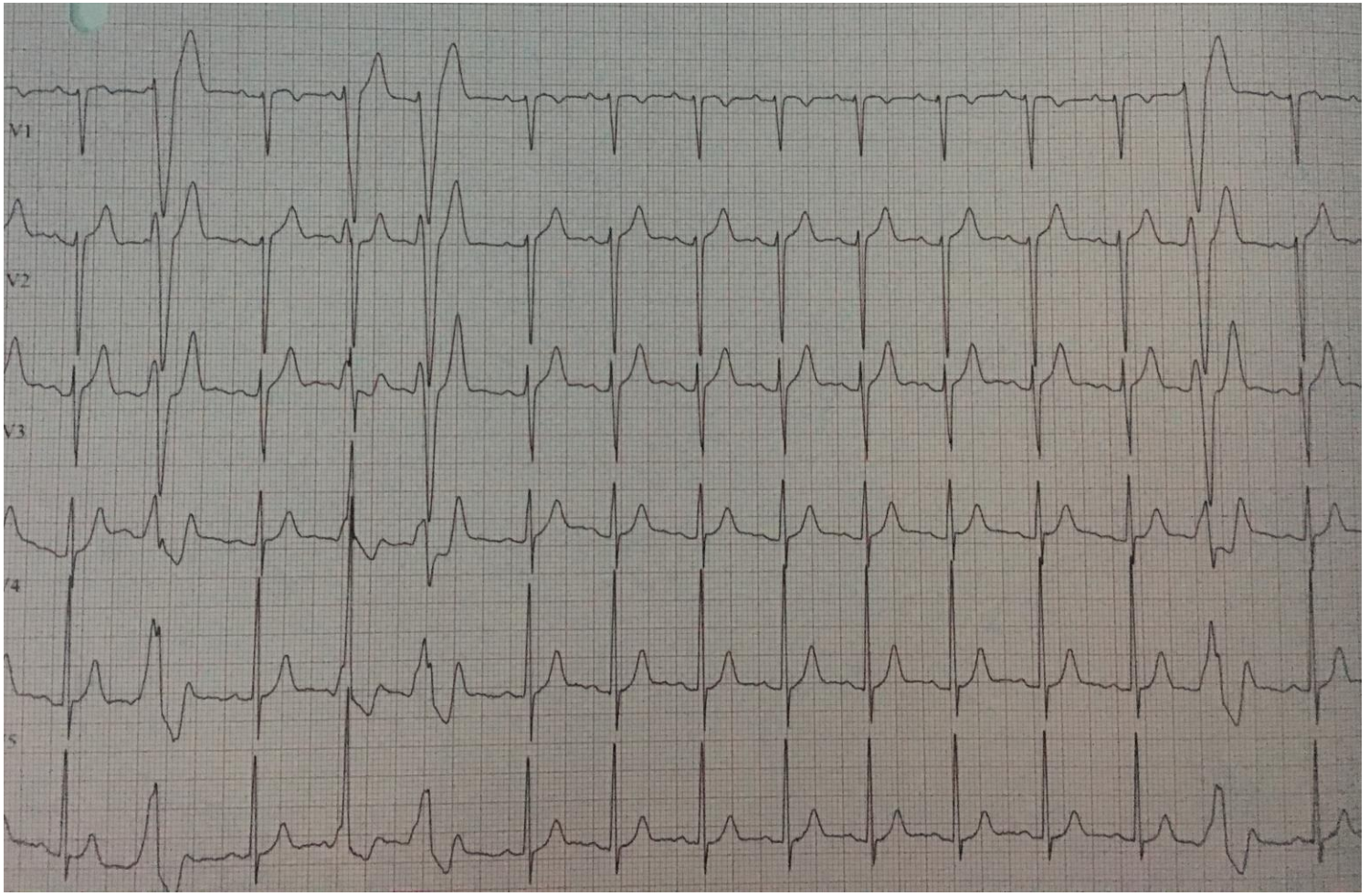

ESVP
ESV
ESV
I
II
III
aVR
aVL
aVF

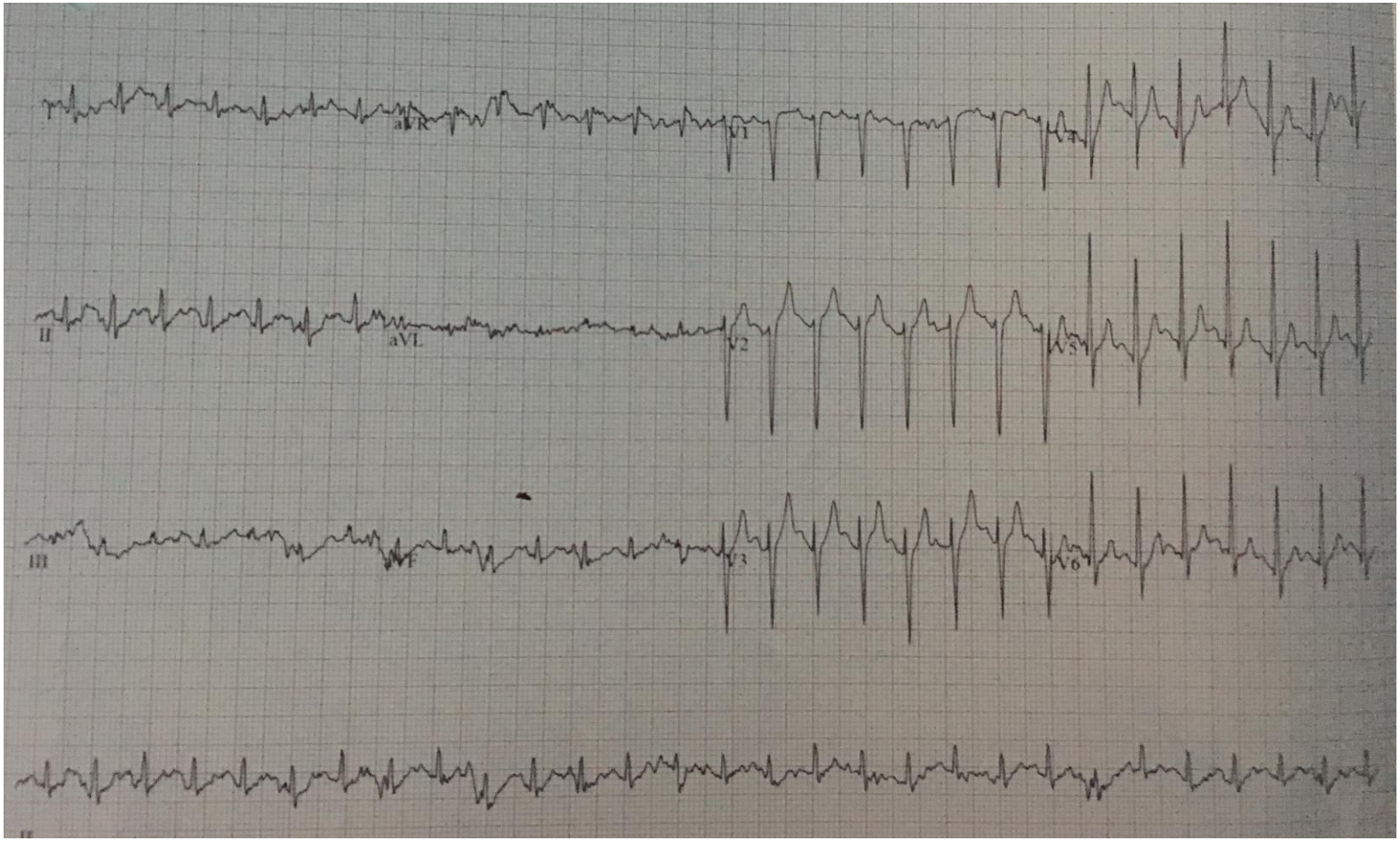

ESVP
ESV
ESV
V1
V2
V3
V4
V5
V6

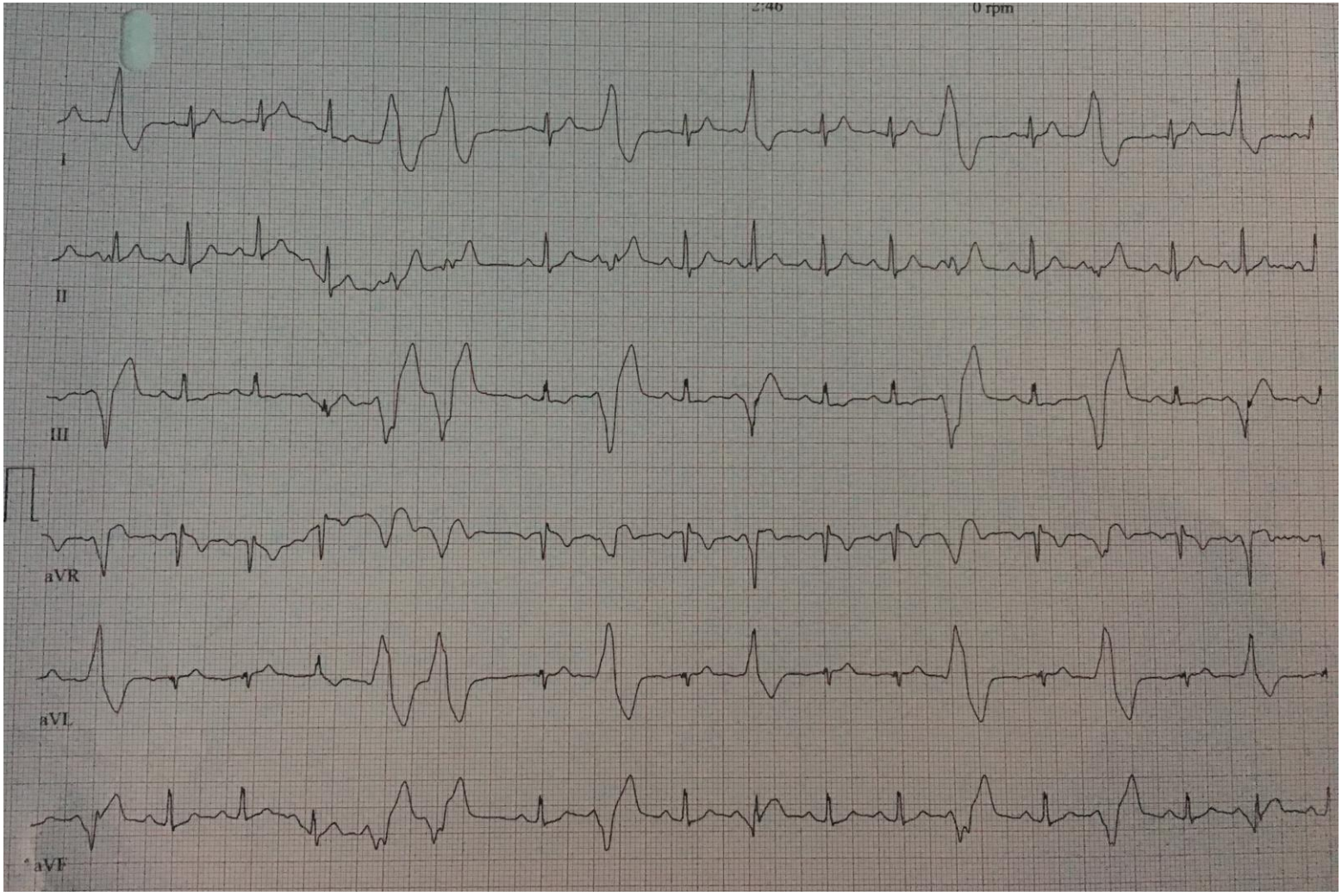

I
II
III
aVR
aVL
aVF

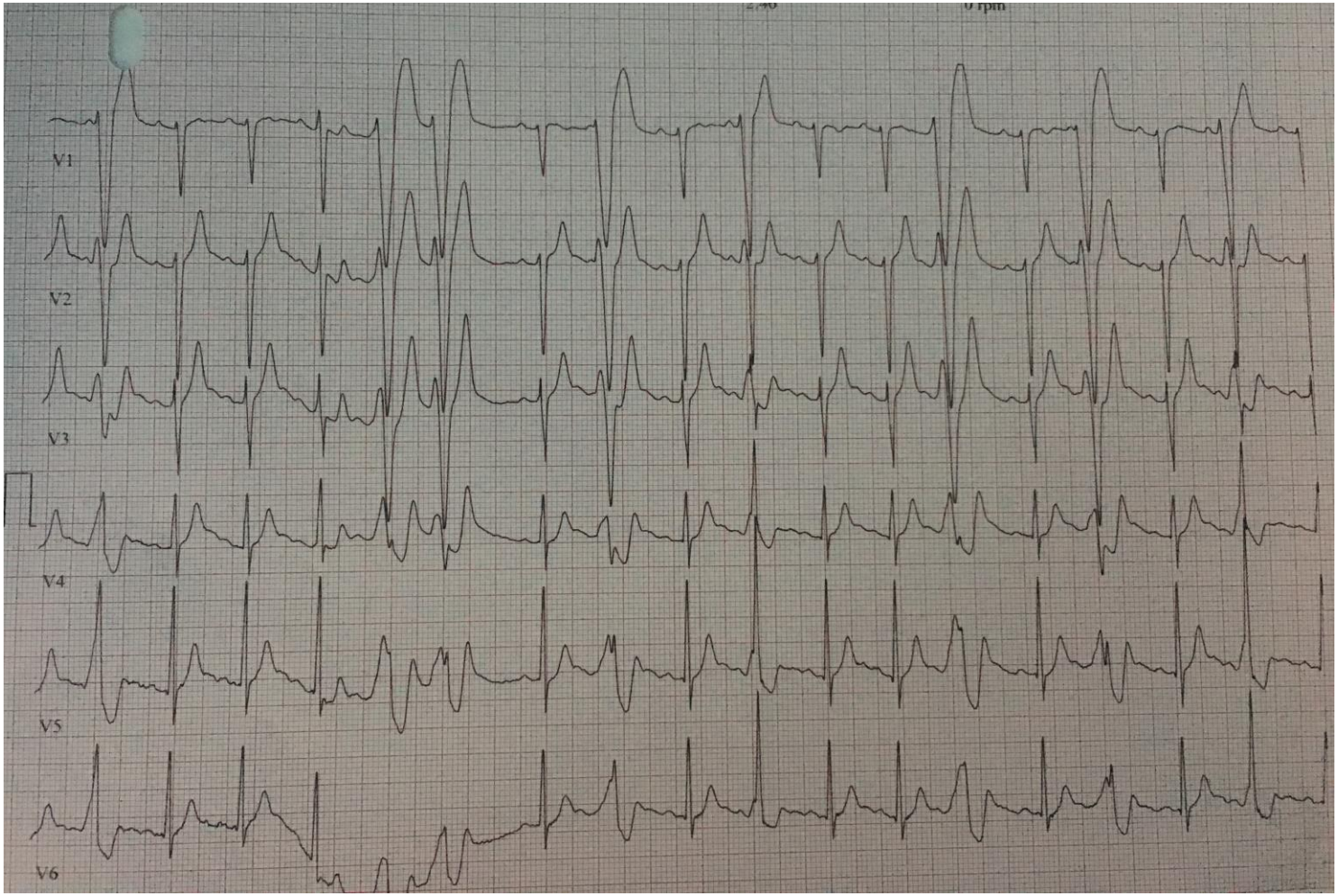

V1
V2
V3
V4
V5
V6


5:37
0 rpm
I
II
III
aVR
aVL
aVF


V1
V2
V3

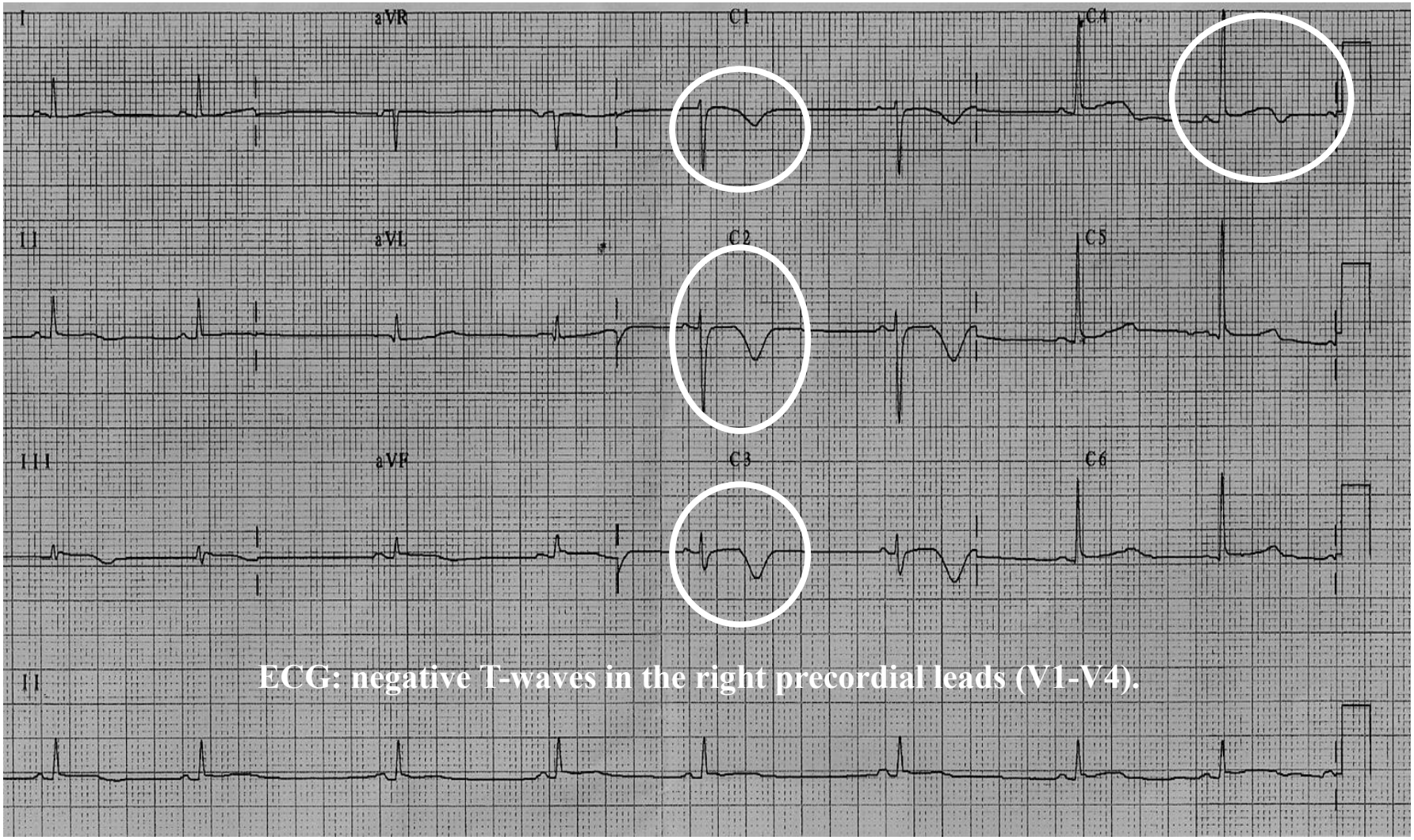

aVR
V1
V4
II
aVL
V2
V5
III
aVF
V3
V6
II


I
II
III
aVR
aVL
aVF

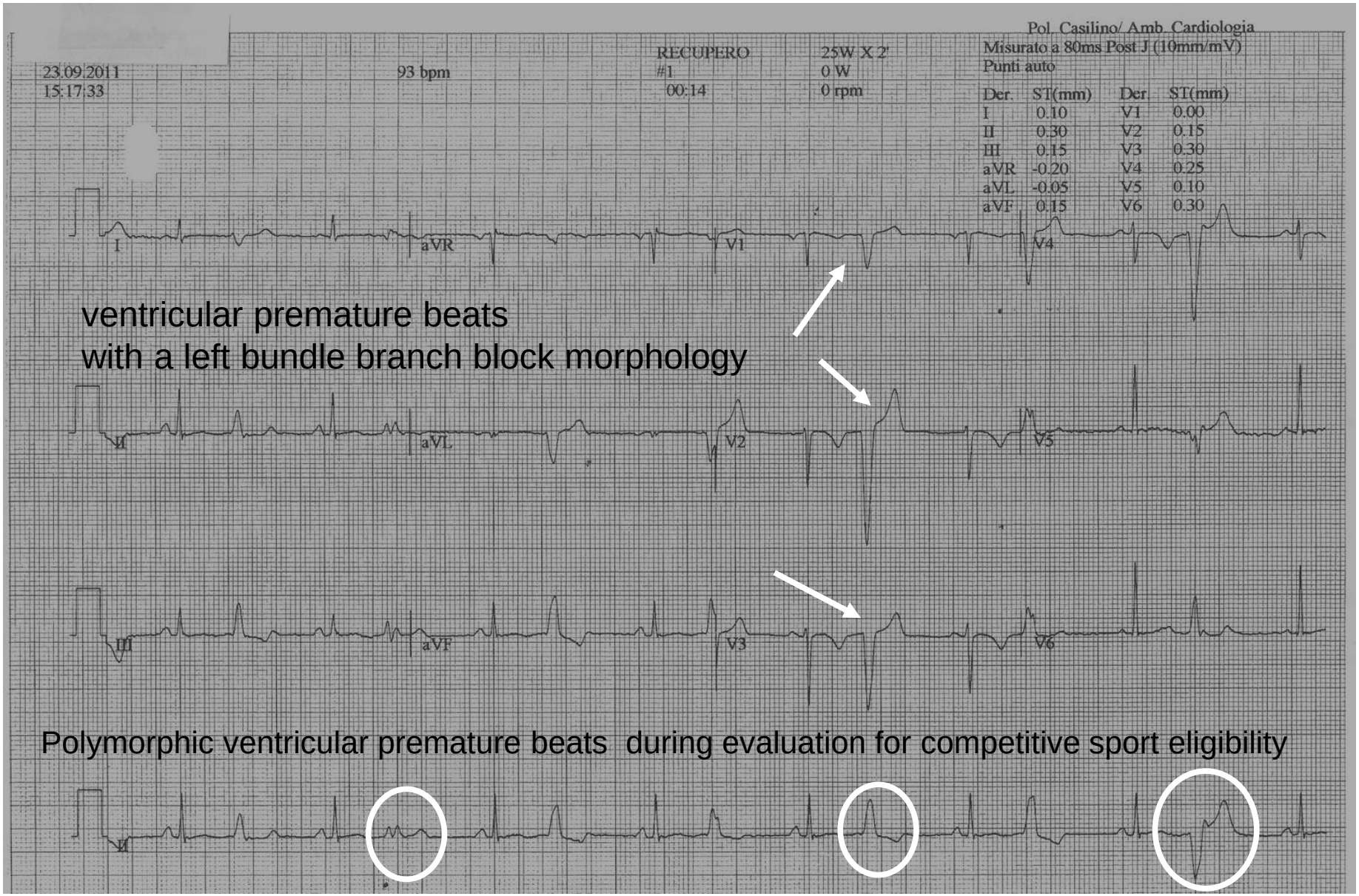

V1
V2
V3
V4
V5
V6

Female 17 year old volleyball player (september 2011)

Asymptomatic for syncope and palpitations.
Family history negative for heart disease or premature sudden cardiac death.

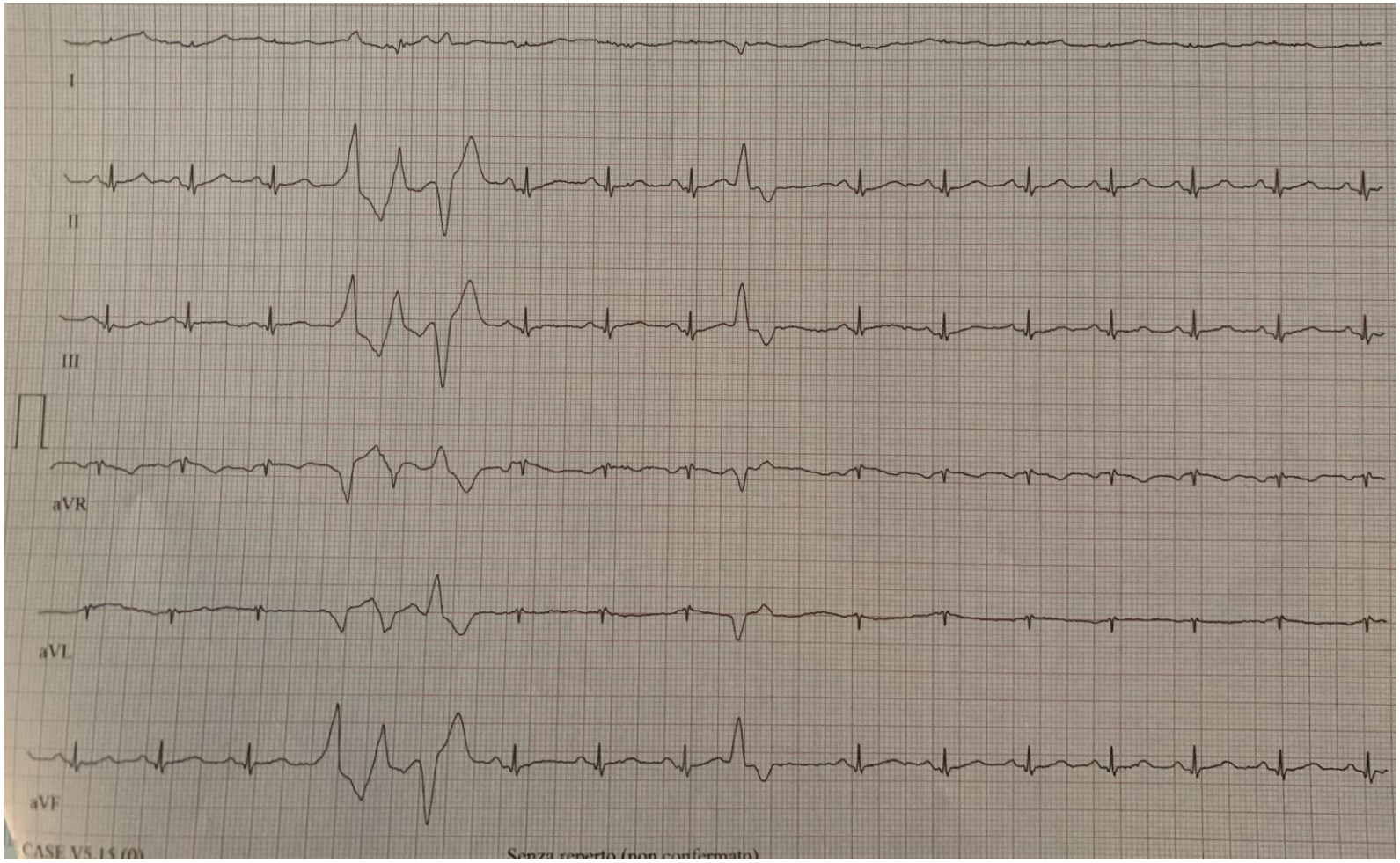


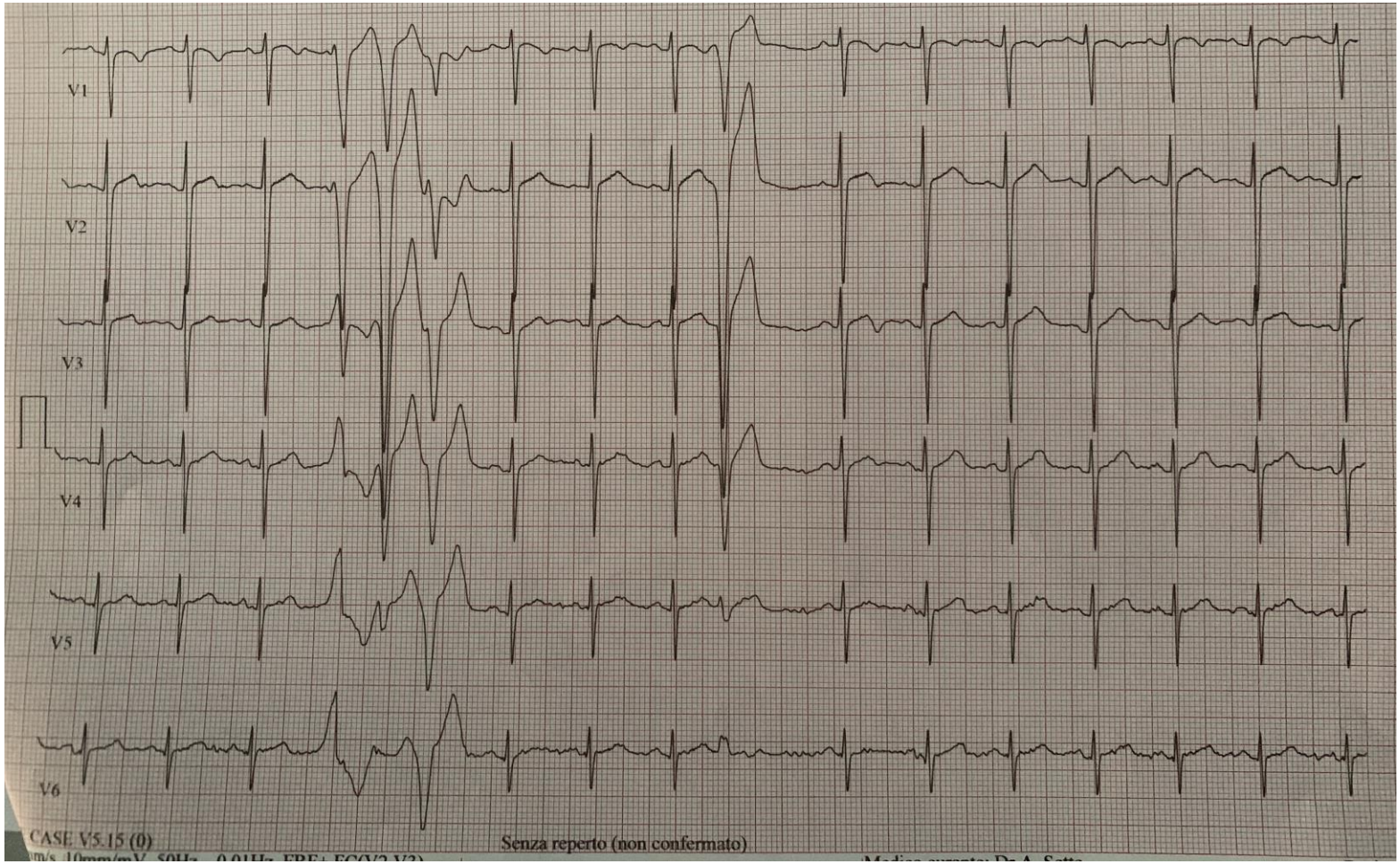
ECG: negative T-waves in the right precordial leads (V1-V4).

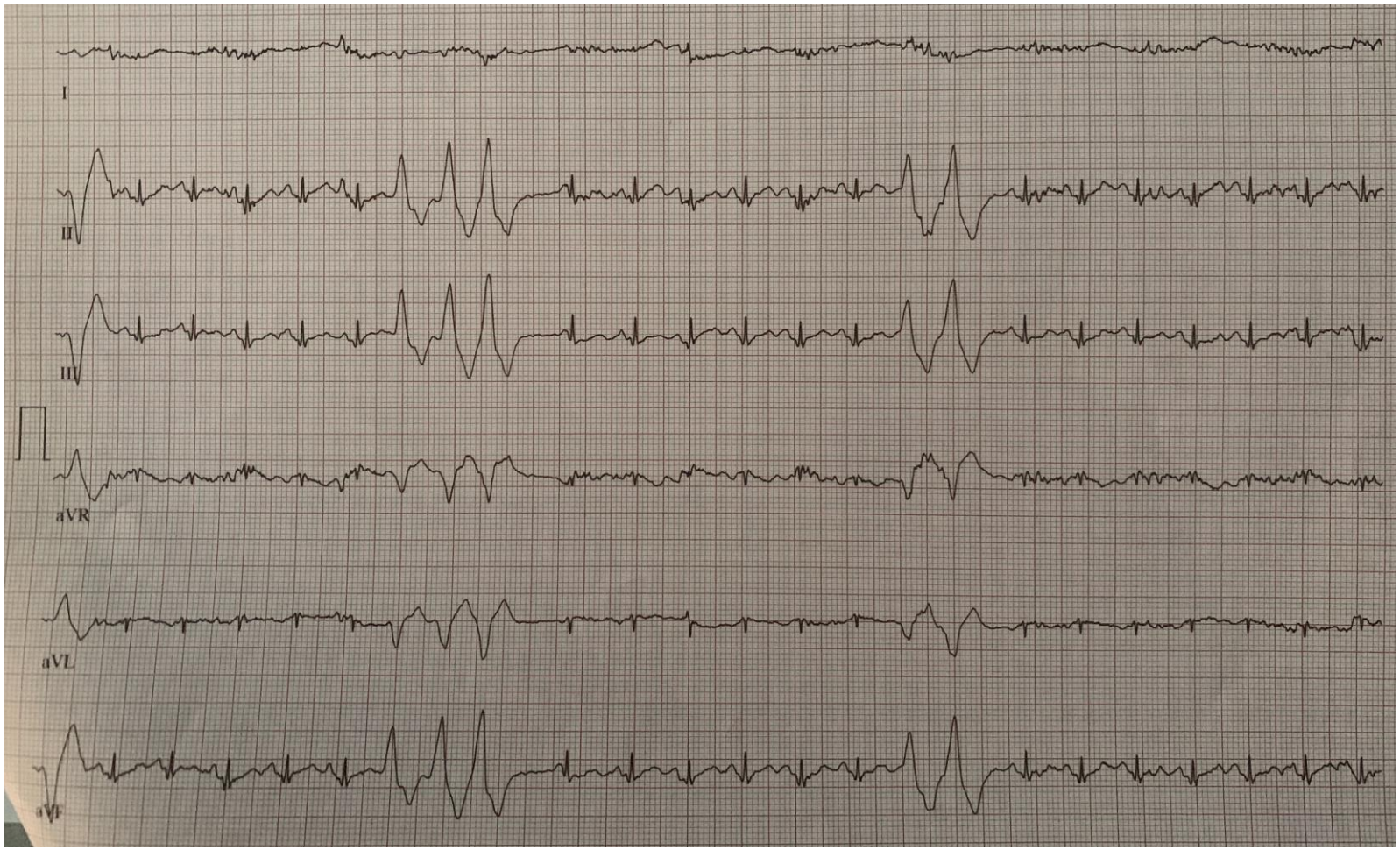


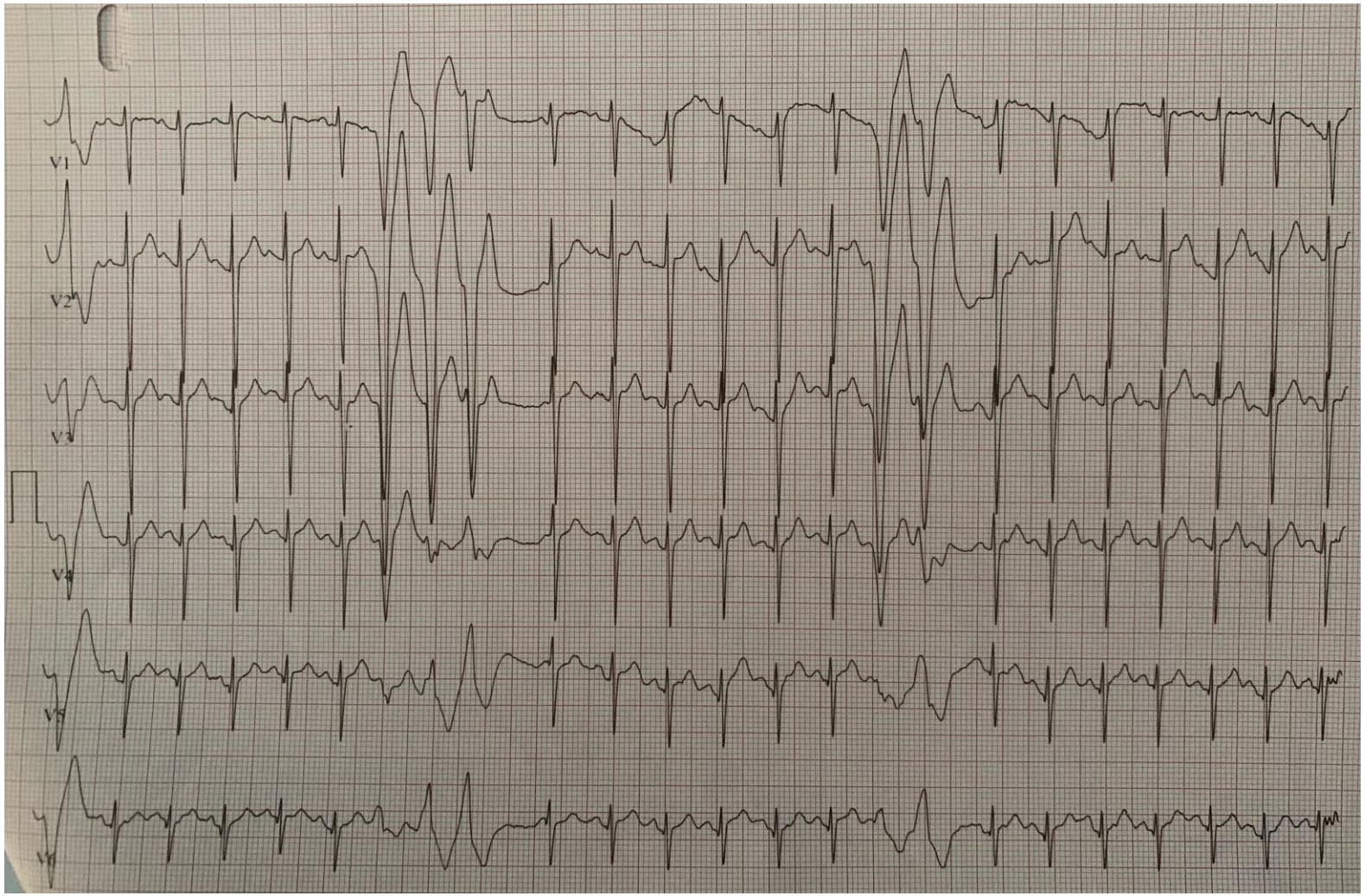
ventricular premature beats
with a left bundle branch block morphology

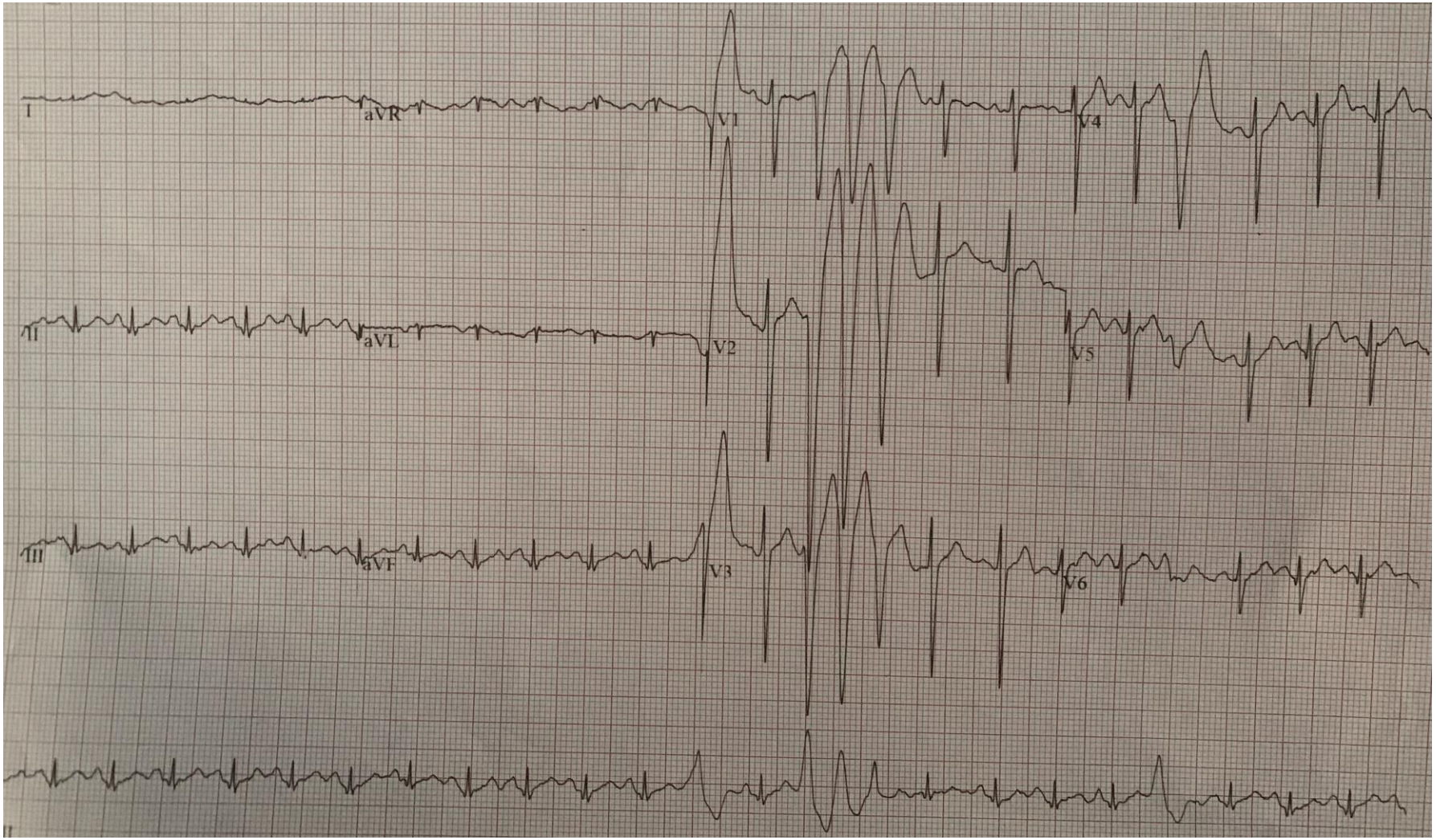
Polymorphic ventricular premature beats during evaluation for competitive sport eligibility


I
II
III
aVR
aVL
aVF
CASE V5.15 (0)
Senza reperto (non confermato)


V1
V2
V3
V4
V5
V6
CASE V5.15 (0)
Senza reperto (non confermato)


I
II
III
aVR
aVL
aVF


V1
V2
V3


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

Totale QRS: **89002** Durata Registrazione: **24 ore, 0 min** Dati analizzati: **24 ore, 0 min**

Report Freq. Card.:

F.C. Min.:	**39**	**alle 05:12:16**
F.C. Max.:	**163**	**alle 22:32:02**
F.C. Media:	**62**	

Episodi di tachicardia/bradicardia:

Tachicardia più lunga:	**0:10:45, 143 BPM Med alle 22:22:45**
Tachicardia più veloce:	**0:10:45, 143 BPM Med alle 22:22:45**
Bradicardia più lunga:	**0:09:40, 46 BPM Med alle 04:58:35**
Bradicardia più lenta:	**0:09:40, 46 BPM Med alle 04:58:35**

Battiti Sopraventricolari:

Singoli:	**0**
Coppie:	**0**
Runs:	**0**
Totale:	**0**

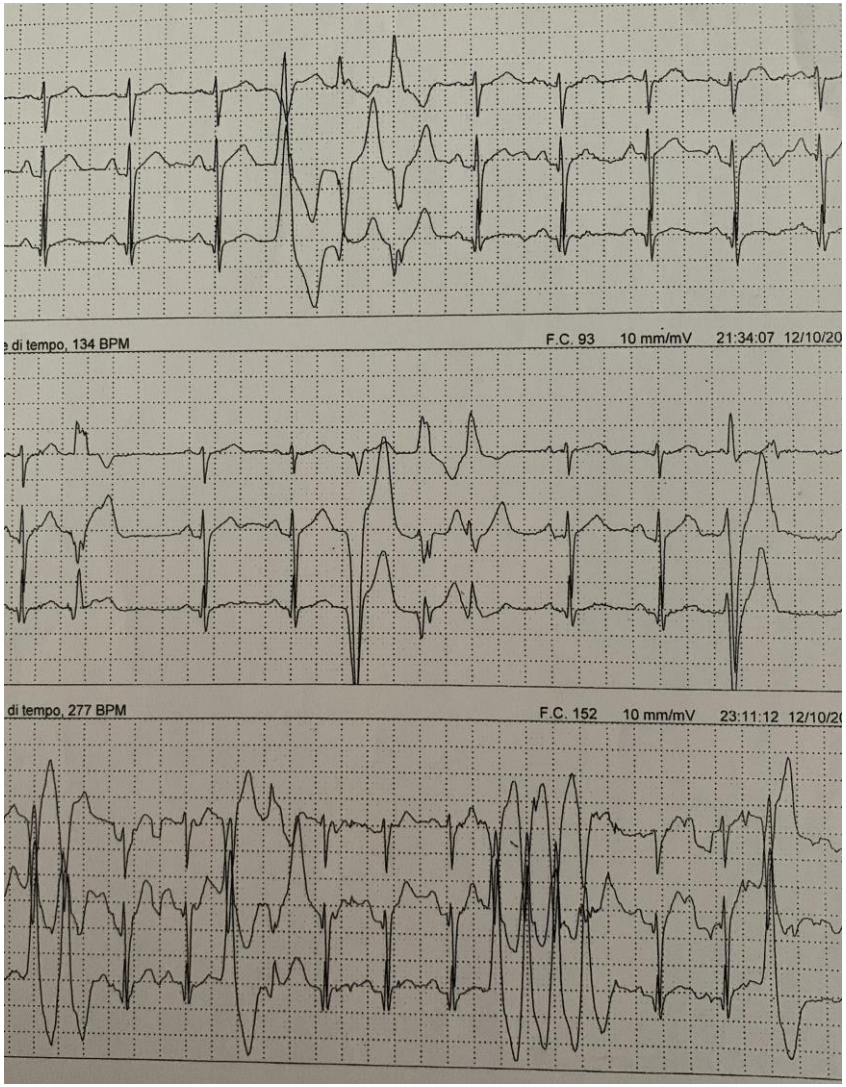
Battiti Ventricolari:

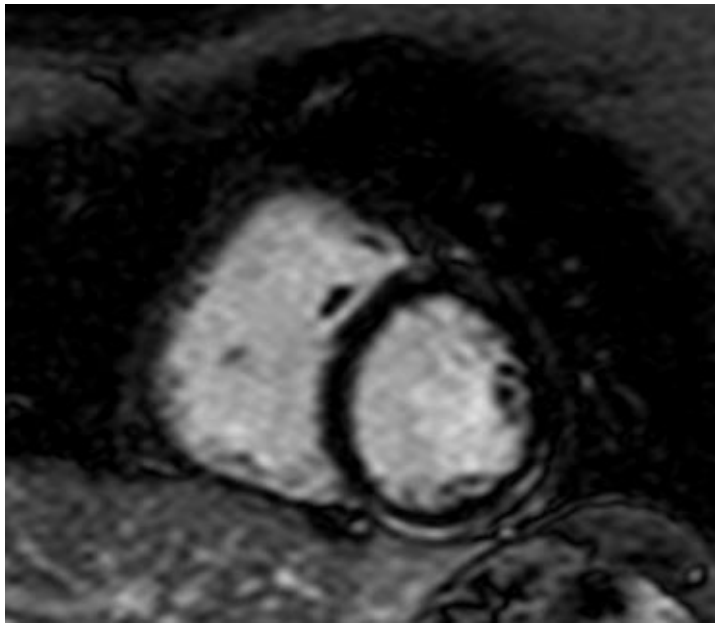
Singoli:	**2195**
Coppie:	**21**
Runs:	**2**
Run più veloce:	**149 BPM alle 22:27:27**
Run più lunga:	**5 alle 22:27:27**
R su T:	**6**
Totale:	**2251**

Variabilità RR:

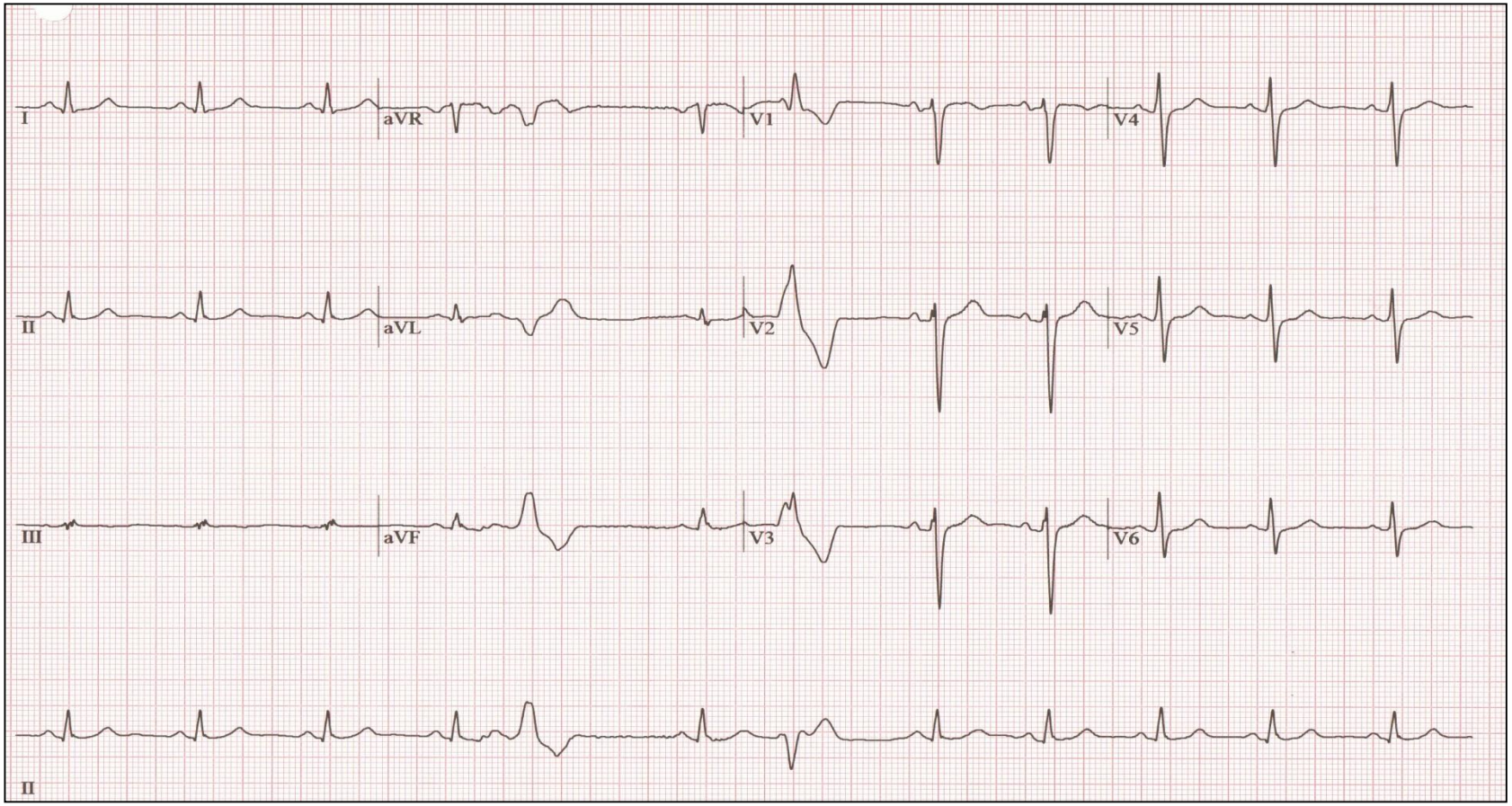
%RR>50:	**20%**
rms-SD:	**60 ms**
Magid SD:	**92 ms**
Kleiger SD:	**192 ms**

Variazione ST:	**II/Ore:**	**V/Ore:**
Max Sottosliv. (µV):	/	/
Max Soprasliv. (µV):	**125 / 22:56:15**	**37 / 20:21:15**

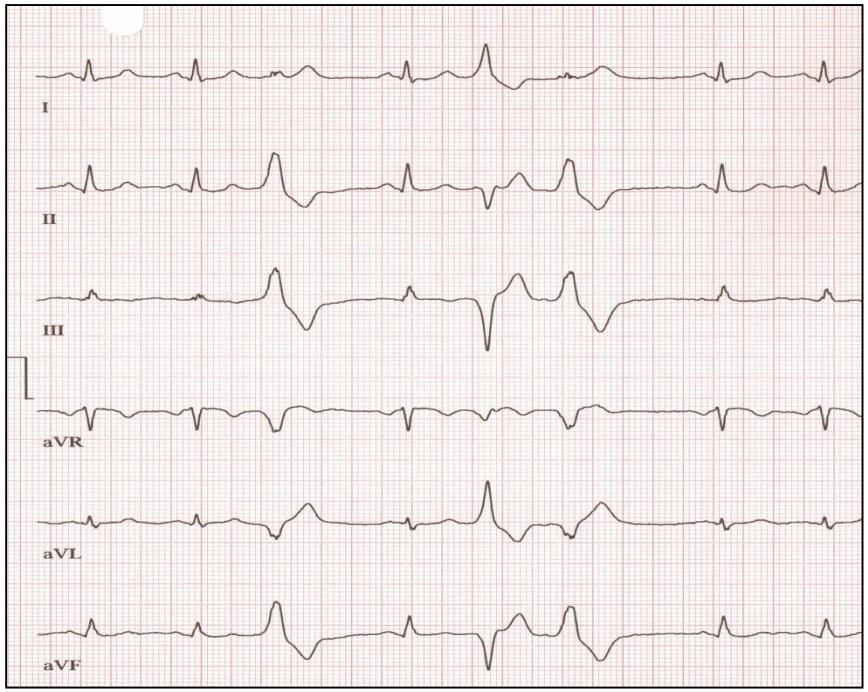

di tempo, 134 BPM
F.C. 93 10 mm/mV 21:34:07 12/10/20
di tempo, 277 BPM
F.C. 152 10 mm/mV 23:11:12 12/10/2

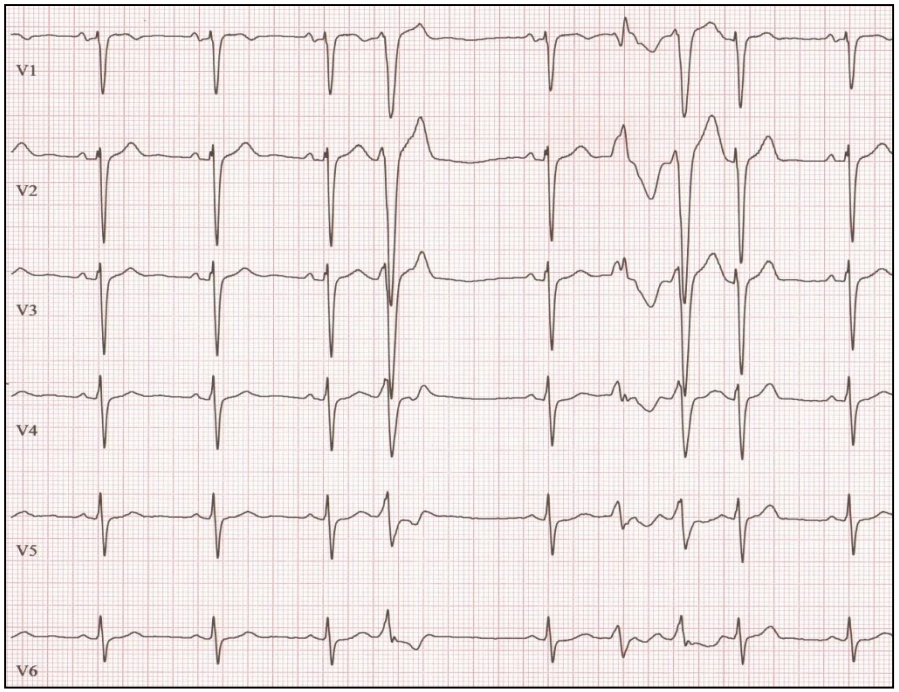


Athlete 45 y.o. male caucasian with 15000 polymorphic PVC, isolated, 228 couplets, 15 NSVT 4 beats



Athlete 45 y.o. male caucasian with 15000 polymorphic PVC, isolated, 228 couplets, 15 NSVT 4 beats

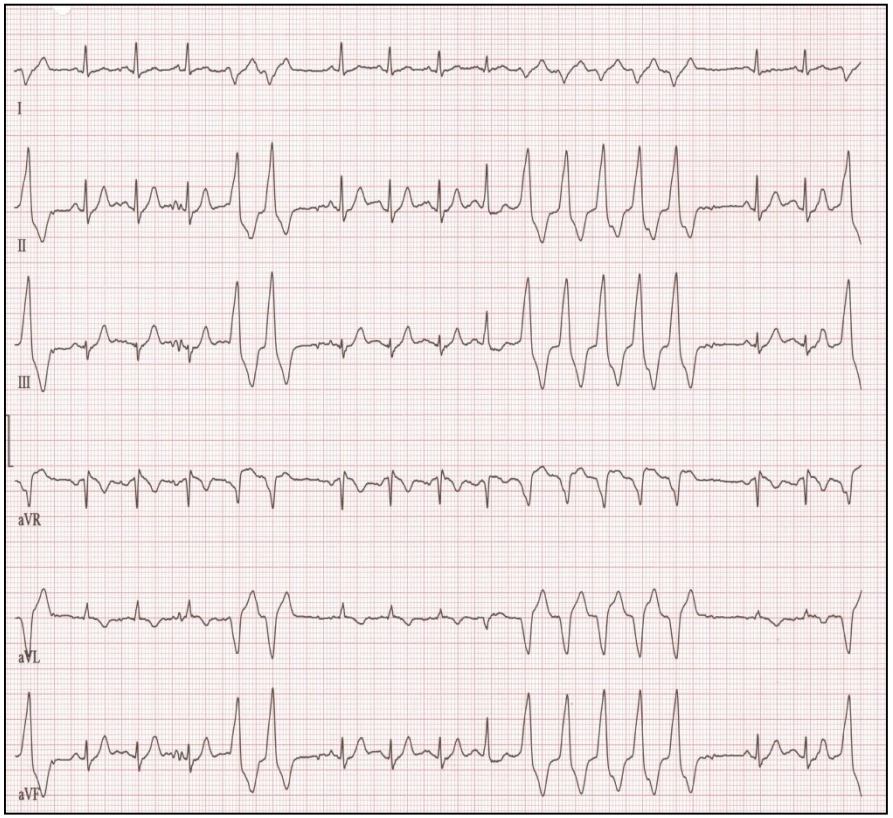


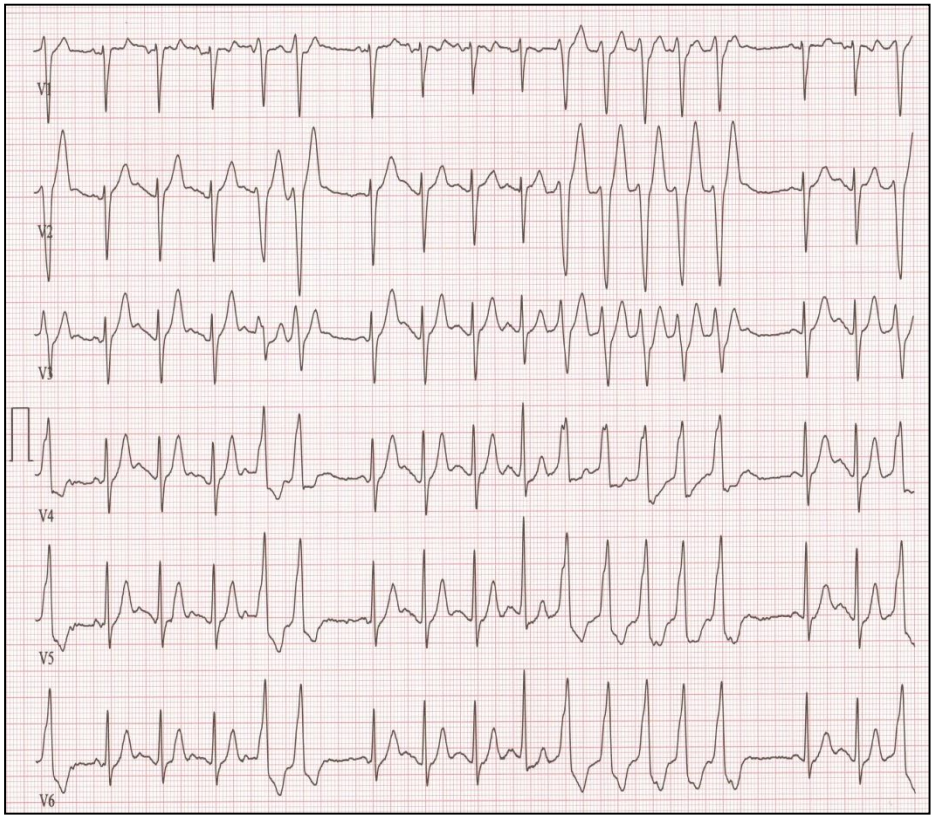


Athlete 45 y.o. male caucasian with 15000 polymorphic PVC, isolated, 228 couplets, 15 NSVT 4 beats

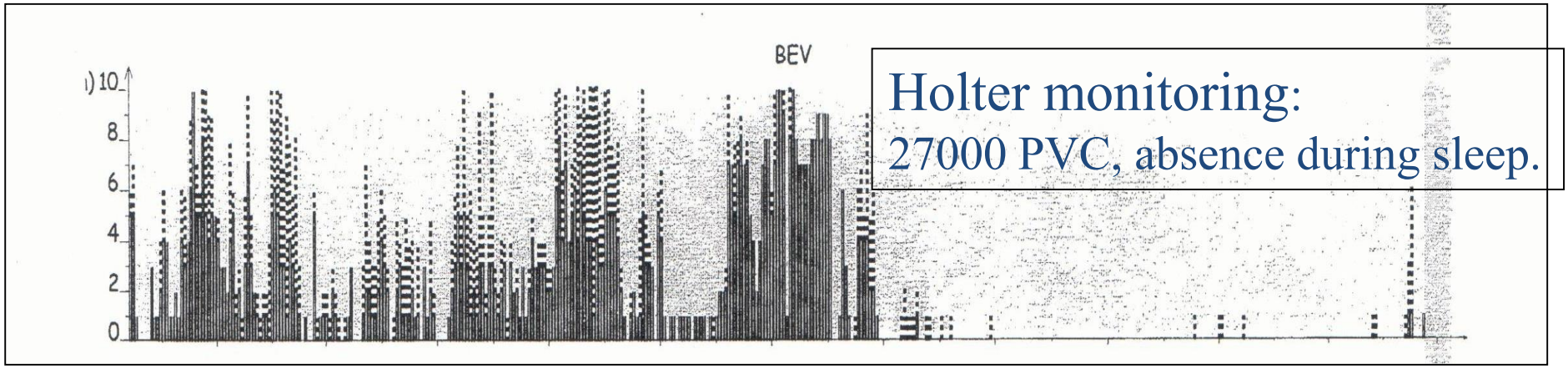
MRI late enhancement inferior basal wall

Athlete 34 y.o. male caucasian with repetitive monomorphic NSVT
No family history of heart disease
Symptomatic for palpitations, no syncope



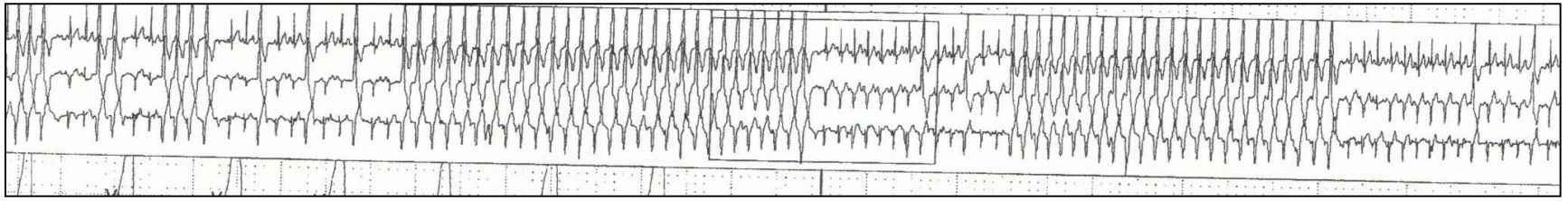


Athlete 34 y.o. male caucasian with repetitive monomorphic NSVT (GALLAVARDIN)
MRI: absence of cardiomyopathy

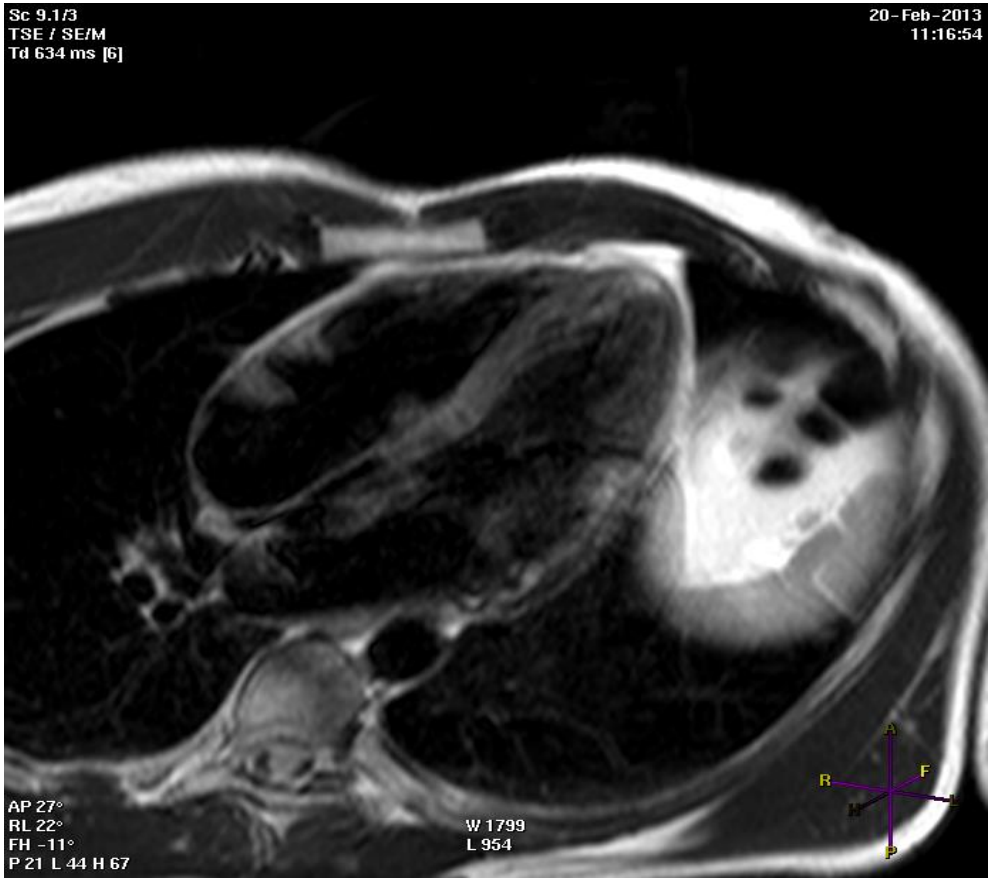


Holter monitoring:
27000 PVC, absence during sleep.

NSVT during Holter monitoring.



Electrophysiological study:
mapping of right ventricular outflow tract (RVOT), inducibility of arrhythmias, RF ablation



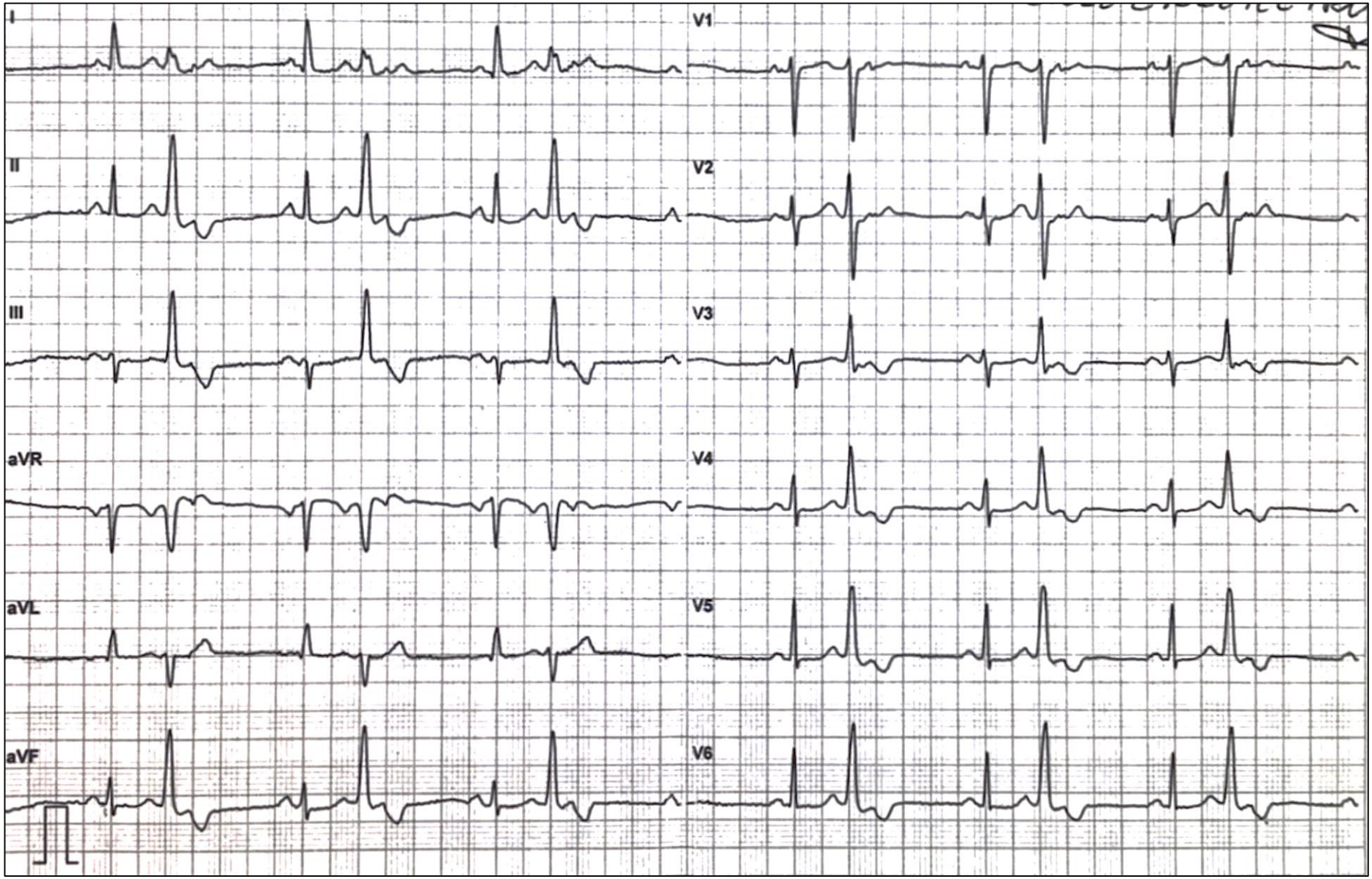
MRI absence of heart disease

EPS no inducibility of arrhythmias
No low voltage areas during electroanatomical mapping

Case #1

- 71 yo female
- Negative medical history
- Symptomatic for palpitation
- No current medications
- Normal echocardiogram findings

12 lead-ECG

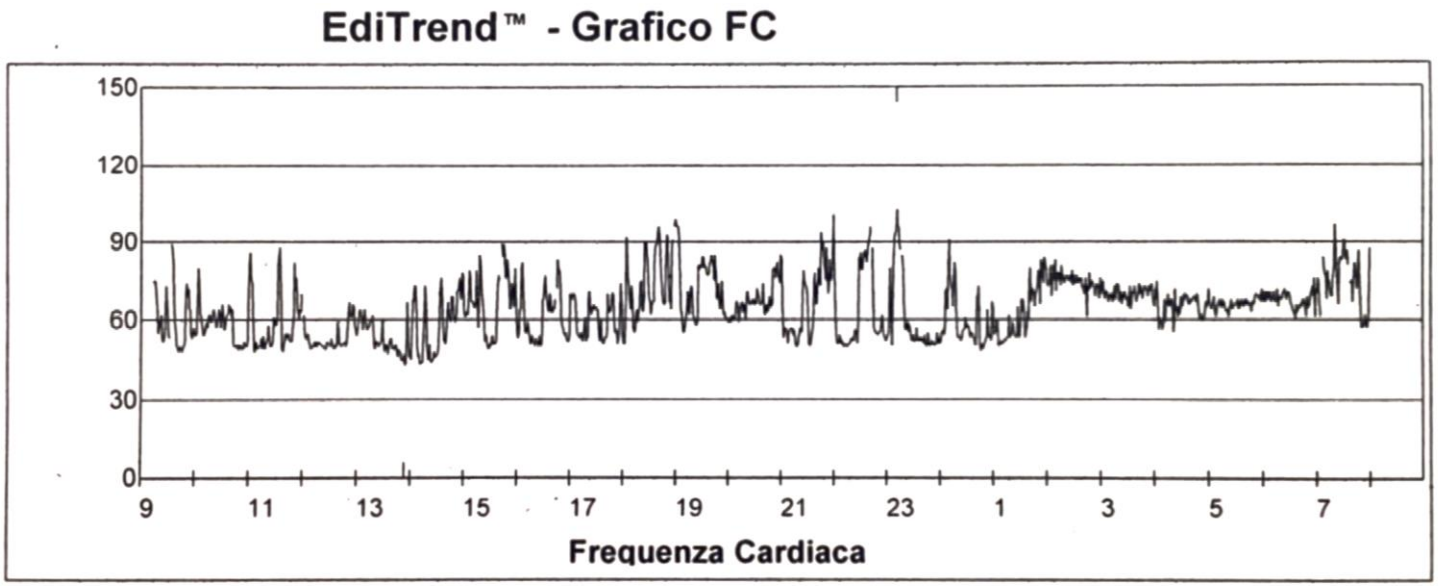


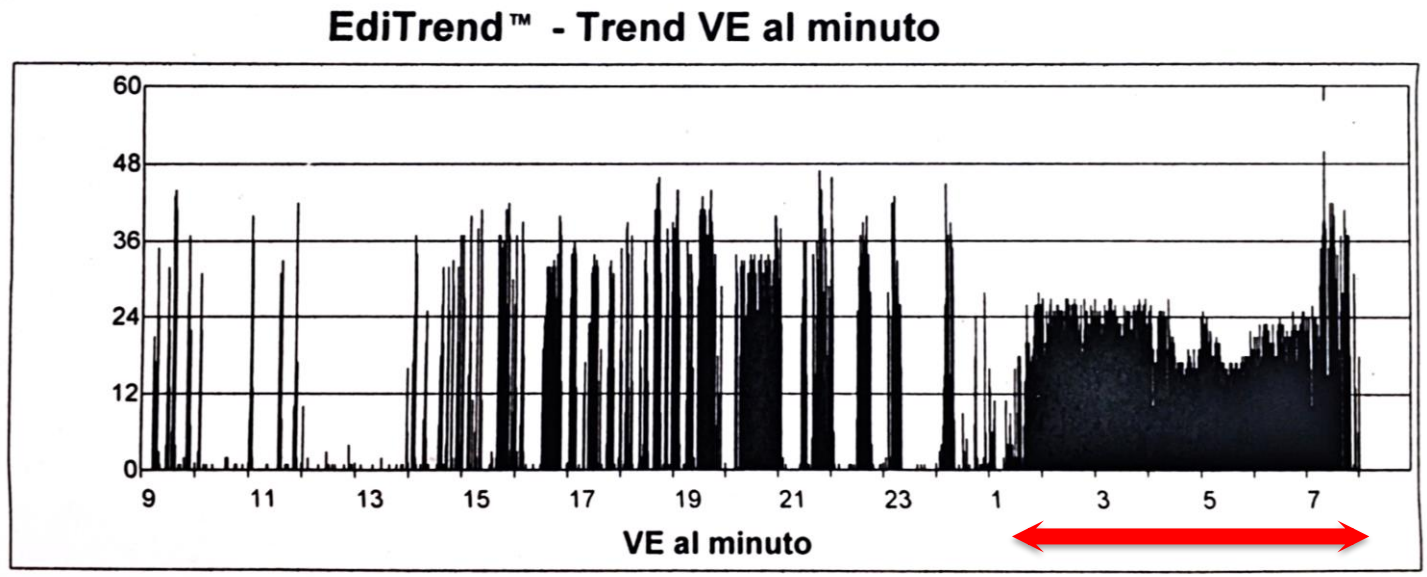
24-h Holter monitoring

Frequenza				Eventi frequenza dipendenti							
Min	43	BPM alle	13.55.00-1	salve Bradicardia	429				Pause	2	
Max	103	BPM alle	23.15.00-1	Piu lunga	161	batt. alle	14.14.51-1		Piu lunga	2,0 sec	
Med	64	BPM		Freq. min.	38	BPM alle	14.26.46-1			alle 06.41	

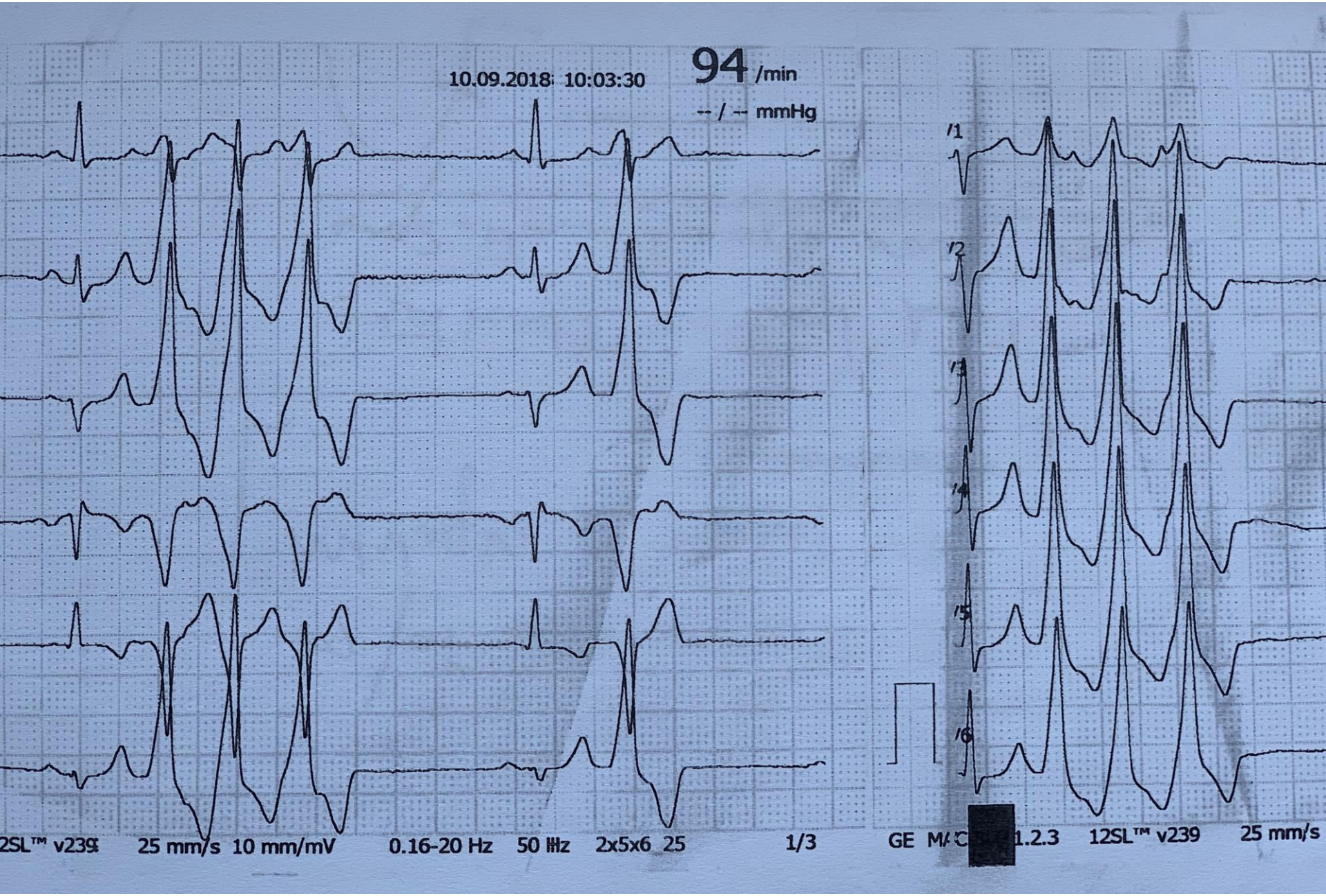
Eventi Ventricolari					Eventi Sopreventricolari				
Battiti Totali	18419		Coppie	174	Battiti Totali	0	Coppie		
% battiti	21,69		Triplette	25	% battiti	0,00			
Forme			Salve bigeminia	299					
Salve AIVR/IVR	0								
Piu lunga	0	batt. alle							
Freq. Min	0	BPM							
Salve Tachi V	26				salve Tachi SV	0			
Piu lunga	4	batt. alle	02.03.19-2		Piu lunga	0	batt. alle		
Freq. Max	164	BPM	02.03.19-2		Freq. Max	0	BPM alle		
VE/minuto max	47	batt. alle	21.48.00-1		SVE/minuto max	0	batt. alle		
VE/ora max	1459	beatt.	07.00.00-2		SVE/ora max	0	beatt.		
VE/ora medio	837,2				SVE/ora medio	0,0			
VE/1000	216,9				SVE/1000	0,0			

Trend


EdiTrend™ - Grafico FC
150
120
90
60
30
0
9
11
13
15
17
19
21
23
1
3
5
7
Frequenza Cardiaca


EdiTrend™ - Trend VE al minuto
60
48
36
24
12
0
9
11
13
15
17
19
21
23
1
3
5
7
VE al minuto

Flecainide therapy was started showing a decrease in PVC burden …9/24 h!

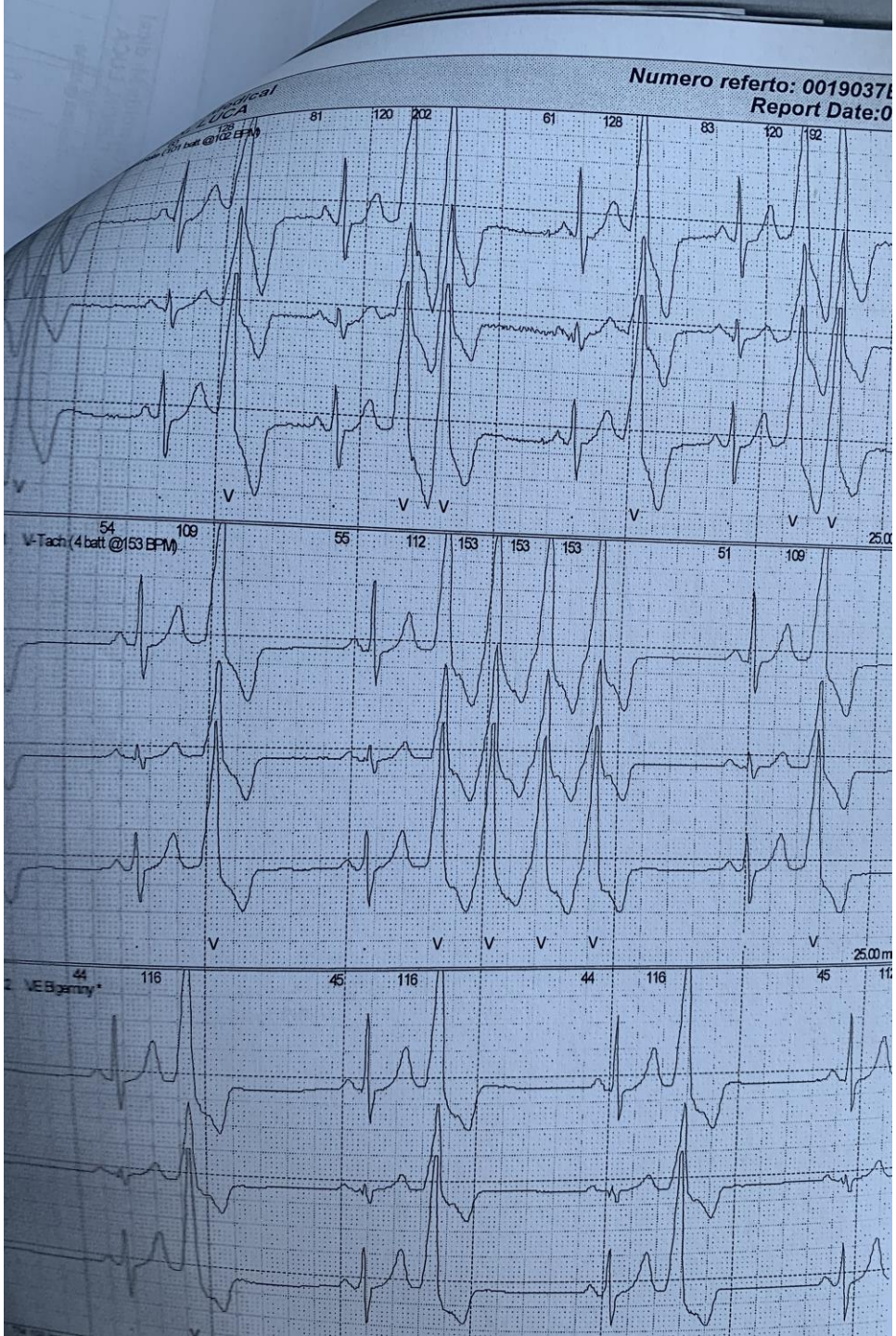

10.09.2018 10:03:30
94 /min
-- / -- mmHg
I
II
III
aVR
aVL
aVF
V1
V2
V3
V4
V5
V6
12SL™ v239
25 mm/s 10 mm/mV
0.16-20 Hz 50 Hz 2x5x6 25
1/3
GE M/C
1.2.3
12SL™ v239
25 mm/s

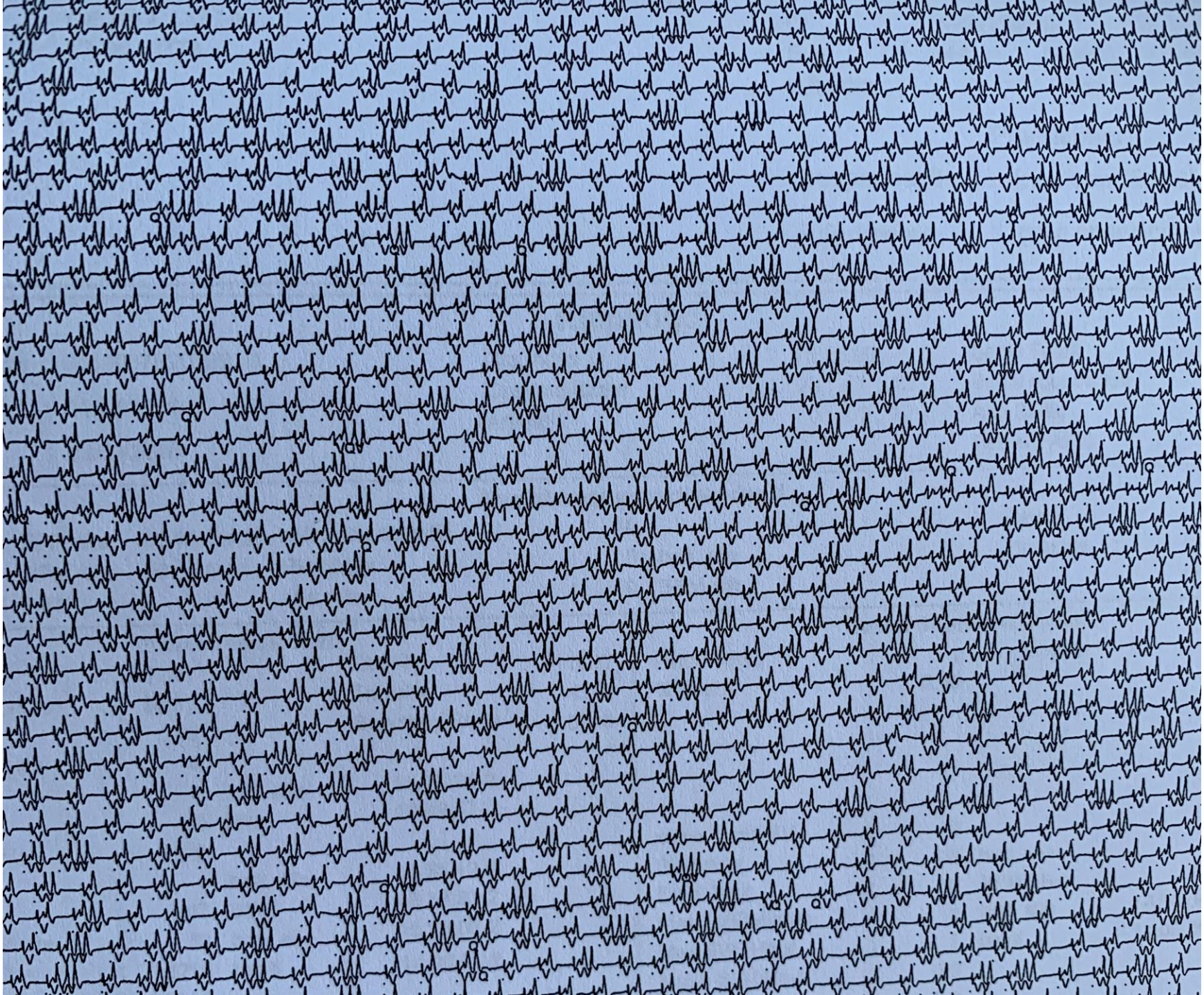
Eventi Ventricolari

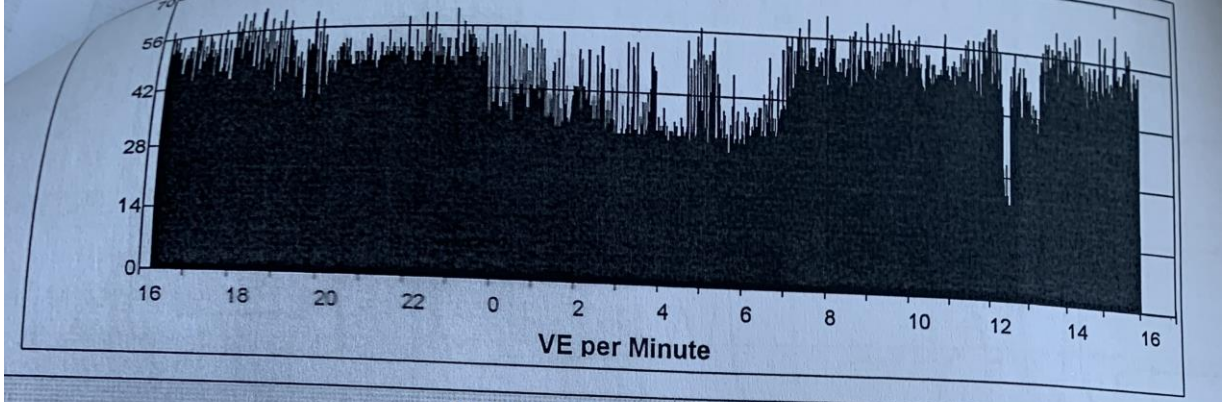
:	67724		Coppie	:	11002
:	58,31		Triplette	:	4370
:	243		Bigeminy Runs	:	5518
:	4				
:	4	batt. alle	15.29.25-2		
:	86	BPM	13.49.37-2		
:	4422				
:	4	batt. alle	18.35.12-1		
:	207	BPM	08.38.33-2		
:	63	batt. alle	15.23.00-2		
:	3210	beatt.	14.00.00-2		
:	2944,5				
:	583,1				

Eventi Sopreventricolari

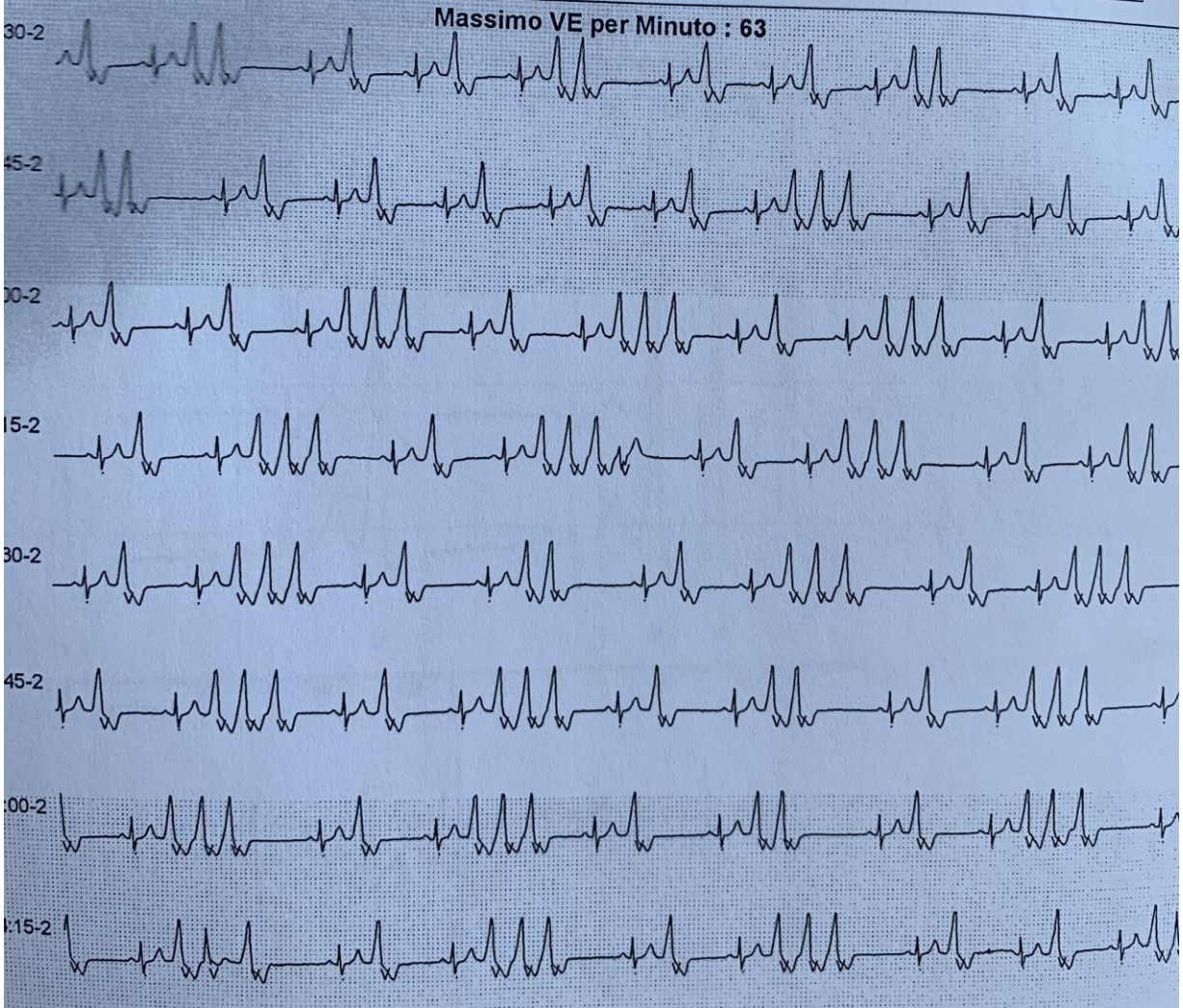
Battiti Totali	:	1	Coppie	:	0	
% Beats	:	0,00				
salve Tachi SV	:	0				
Piu lunga	:	0	batt. alle			
Freq. Max	:	0	BPM alle			
Max SVE/Minute	:	1	batt. alle	14.56.00-2		
Max SVE/Hour	:	1	beatt.	14.00.00-2	-	
Mean SVE/Hour	:	0,0				
SVE/1000	:	0,0				

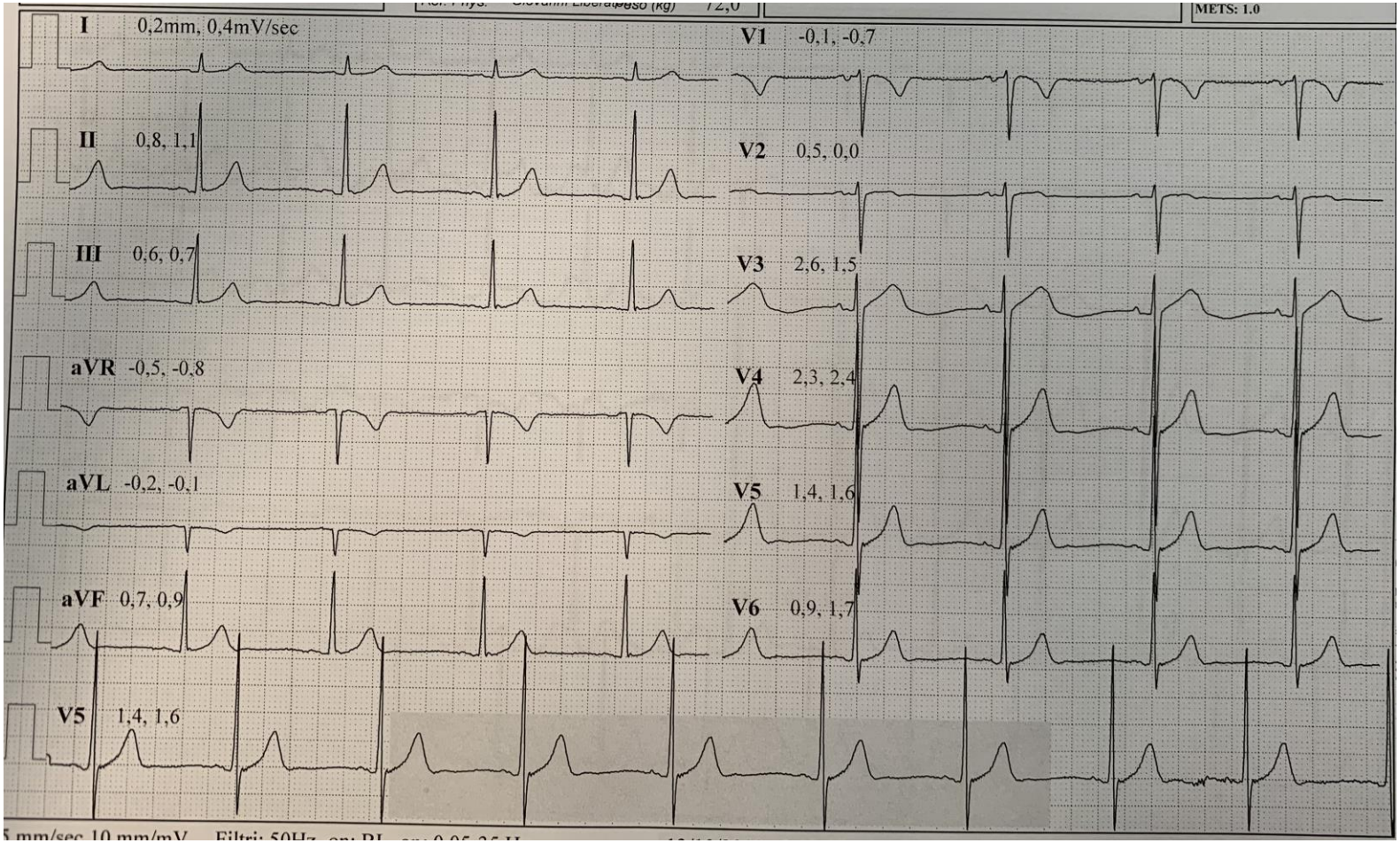

Numero referto: 0019037
Report Date:0
V-Tach (4 batt @153 BPM)

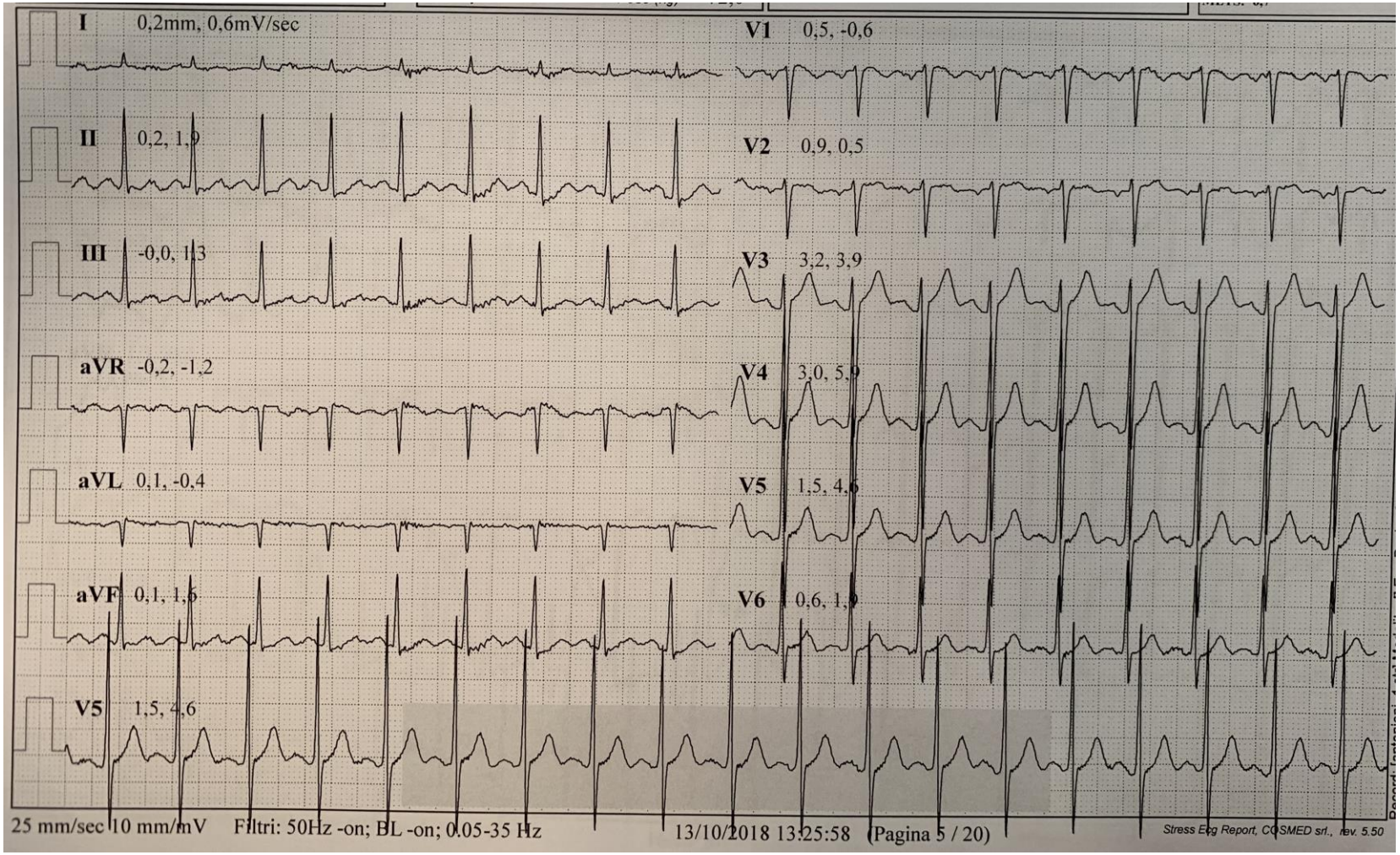


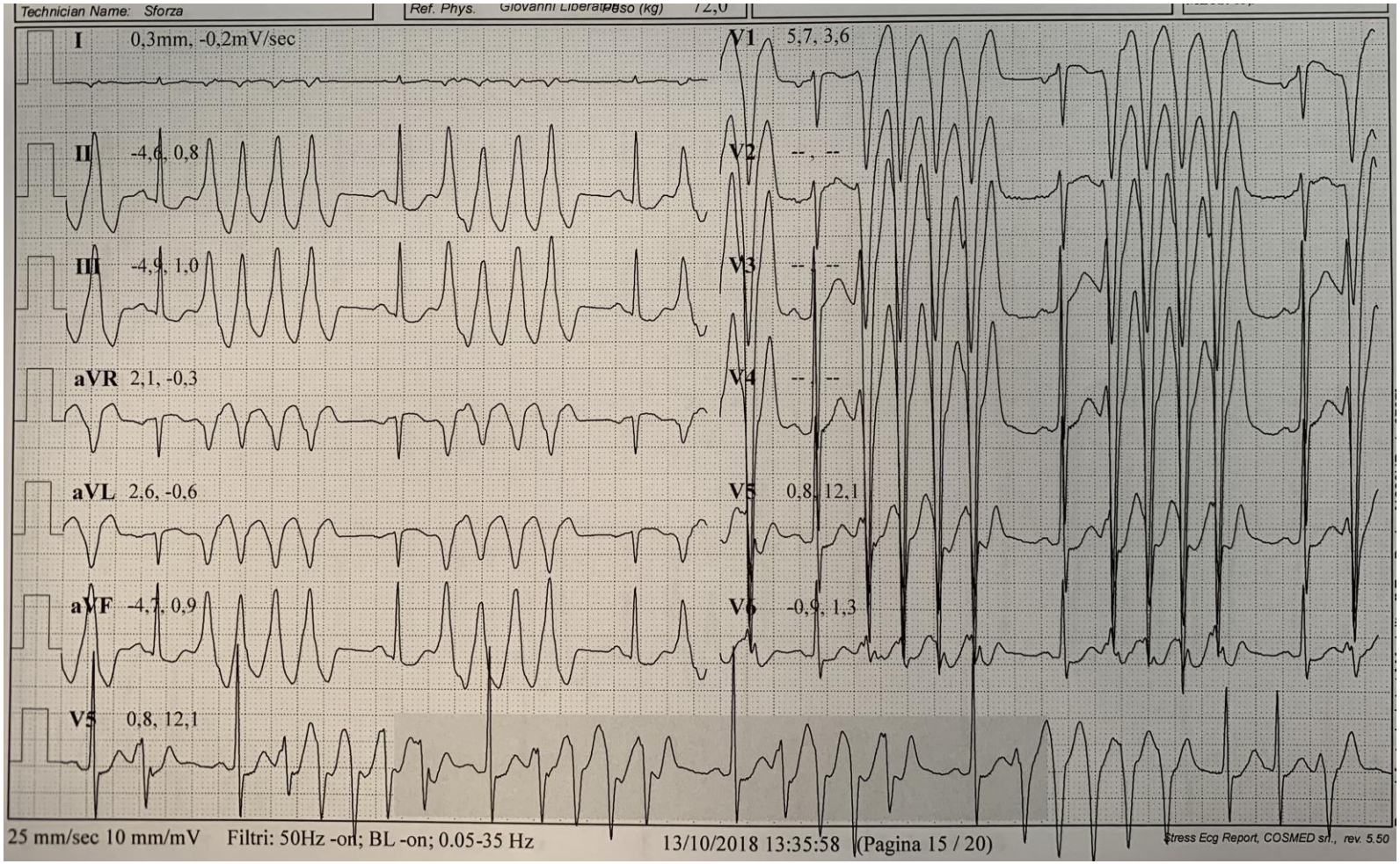


Massimo VE per Minuto : 63




I 0,2mm, 0,4mV/sec
II 0,8, 1,1
III 0,6, 0,7
aVR -0,5, -0,8
aVL -0,2, -0,1
aVF 0,7, 0,9
V5 1,4, 1,6
V1 -0,1, -0,7
V2 0,5, 0,0
V3 2,6, 1,5
V4 2,3, 2,4
V5 1,4, 1,6
V6 0,9, 1,7
METS: 1.0


I 0,2mm, 0,6mV/sec
II 0,2, 1,9
III -0,0, 1,3
aVR -0,2, -1,2
aVL 0,1, -0,4
aVF 0,1, 1,6
V5 1,5, 4,6
V1 0,5, -0,6
V2 0,9, 0,5
V3 3,2, 3,9
V4 3,0, 5,9
V5 1,5, 4,6
V6 0,6, 1,9
25 mm/sec 10 mm/mV Filtri: 50Hz -on; BL -on; 0.05-35 Hz
13/10/2018 13:25:58 (Pagina 5 / 20)
Stress Ecg Report, COSMED srl., rev. 5.50


Technician Name: Sforza
Ref. Phys. Giovanni Liberati
Peso (kg) 72,0
I 0,3mm, -0,2mV/sec
II -4,6, 0,8
III -4,9, 1,0
aVR 2,1, -0,3
aVL 2,6, -0,6
aVF -4,7, 0,9
V5 0,8, 12,1
V1 5,7, 3,6
V2 --, --
V3 -- , --
V4 -- , --
V5 0,8, 12,1
V6 -0,9, 1,3
25 mm/sec 10 mm/mV Filtri: 50Hz -on; BL -on; 0.05-35 Hz
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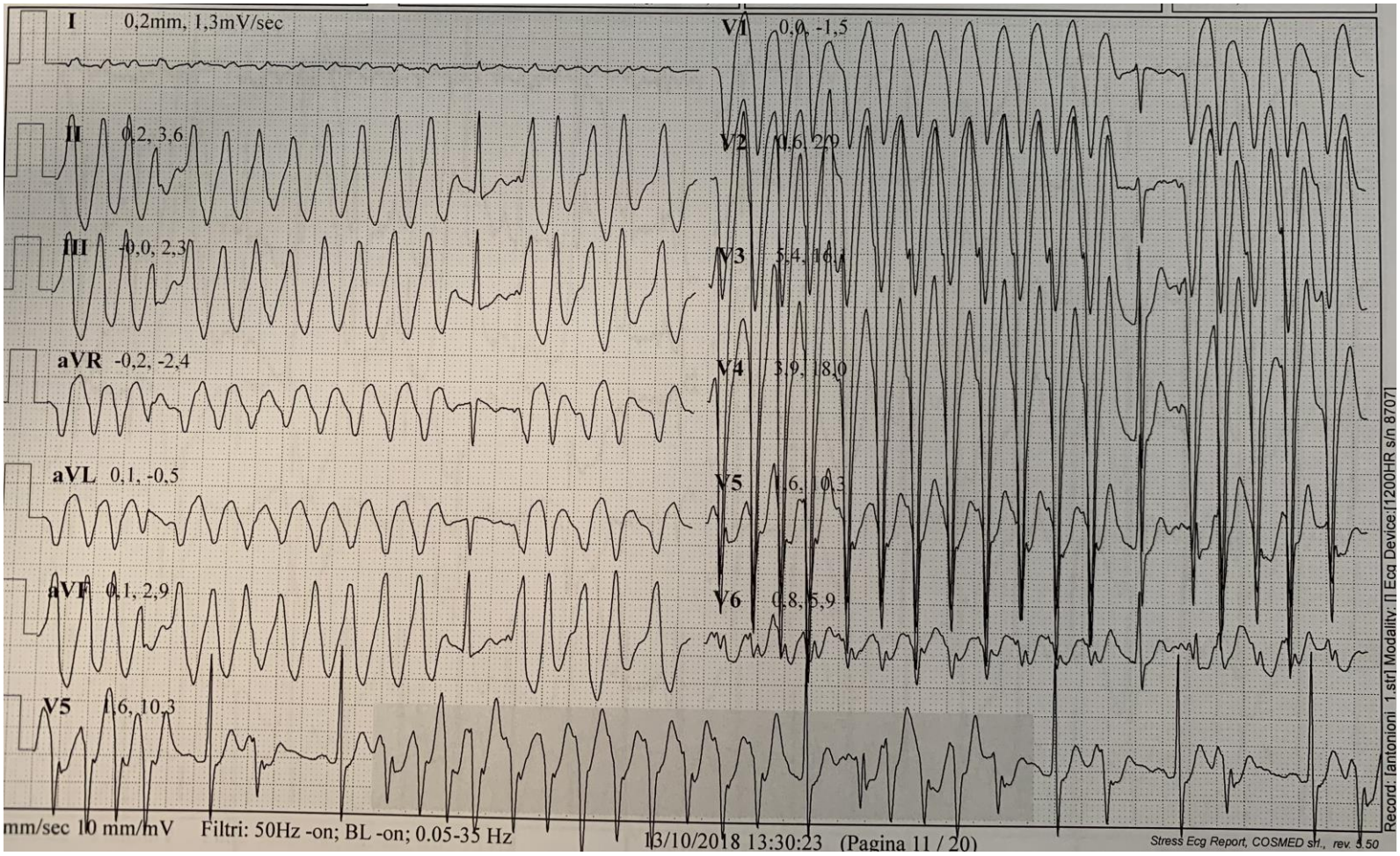
Stress Ecg Report, COSMED srl., rev. 5.50


I 0,2mm, 1,3mV/sec
II 0,2, 3,6
III 0,0, 2,3
aVR -0,2, -2,4
aVL 0,1, -0,5
aVF 0,1, 2,9
V5 1,6, 10,3
V1 0,0, -1,5
V2 1,6, 2,9
V3 5,4, 11,1
V4 3,9, 8,0
V5 1,6, 10,3
V6 0,8, 5,9
mm/sec 10 mm/mV Filtri: 50Hz -on; BL -on; 0.05-35 Hz
13/10/2018 13:30:23 (Pagina 11 / 20)
Stress Ecg Report, COSMED srl., rev. 3.50
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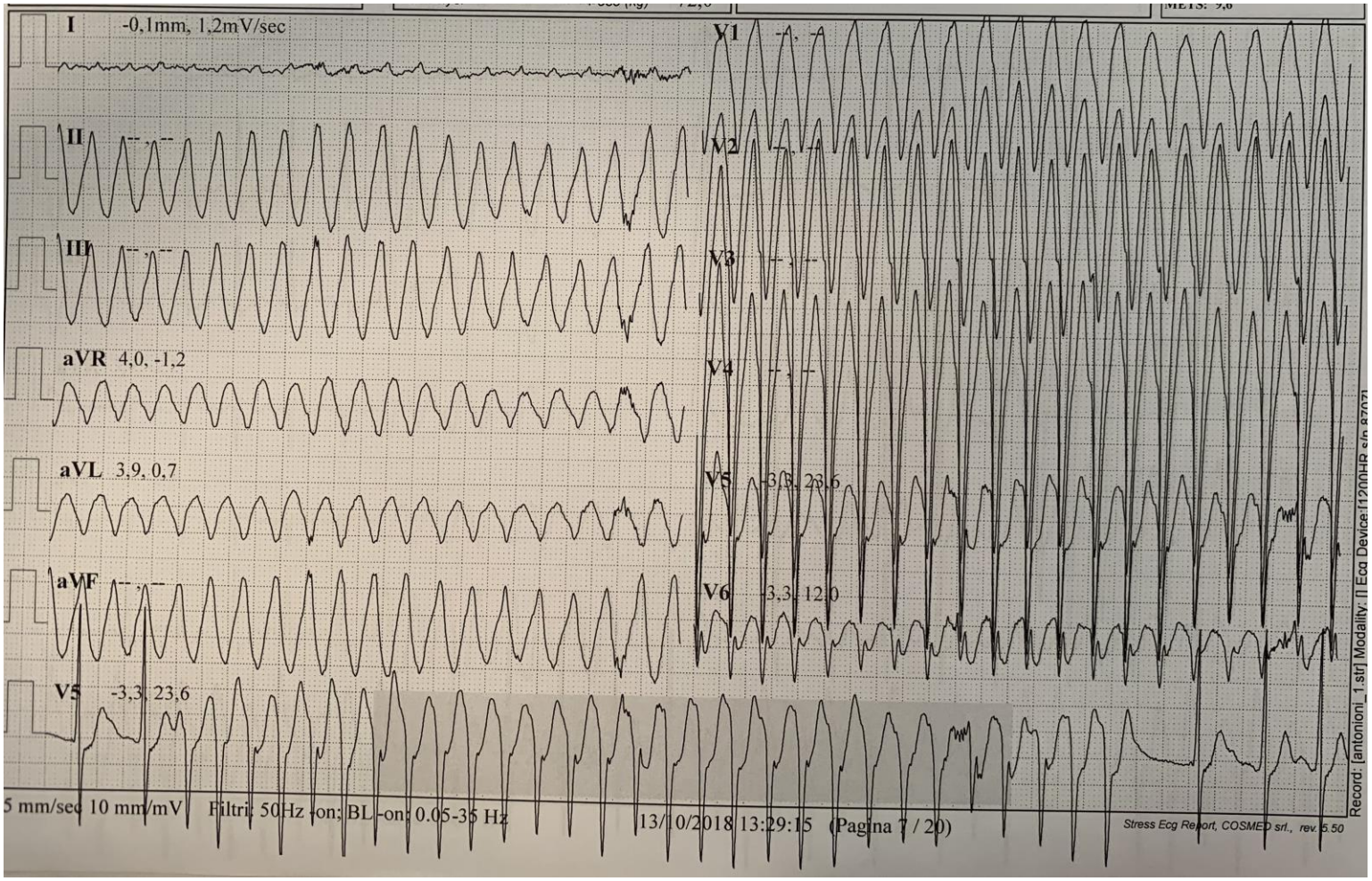
I -0,1mm, 1,2mV/sec
II -- , --
III -- , --
aVR 4,0, -1,2
aVL 3,9, 0,7
aVF -- , --
V5 -3,3, 23,6
V1 -1, -1
V2 -1, -1
V3 -- , --
V4 -- , --
V5 -3,3, 23,6
V6 -3,3, 12,0
5 mm/sec 10 mm/mV Filtri: 50Hz -on; BL -on; 0.05-35 Hz
13/10/2018 13:29:15 (Pagina 7 / 20)
Stress Ecg Report, COSMED srl., rev. 5.50
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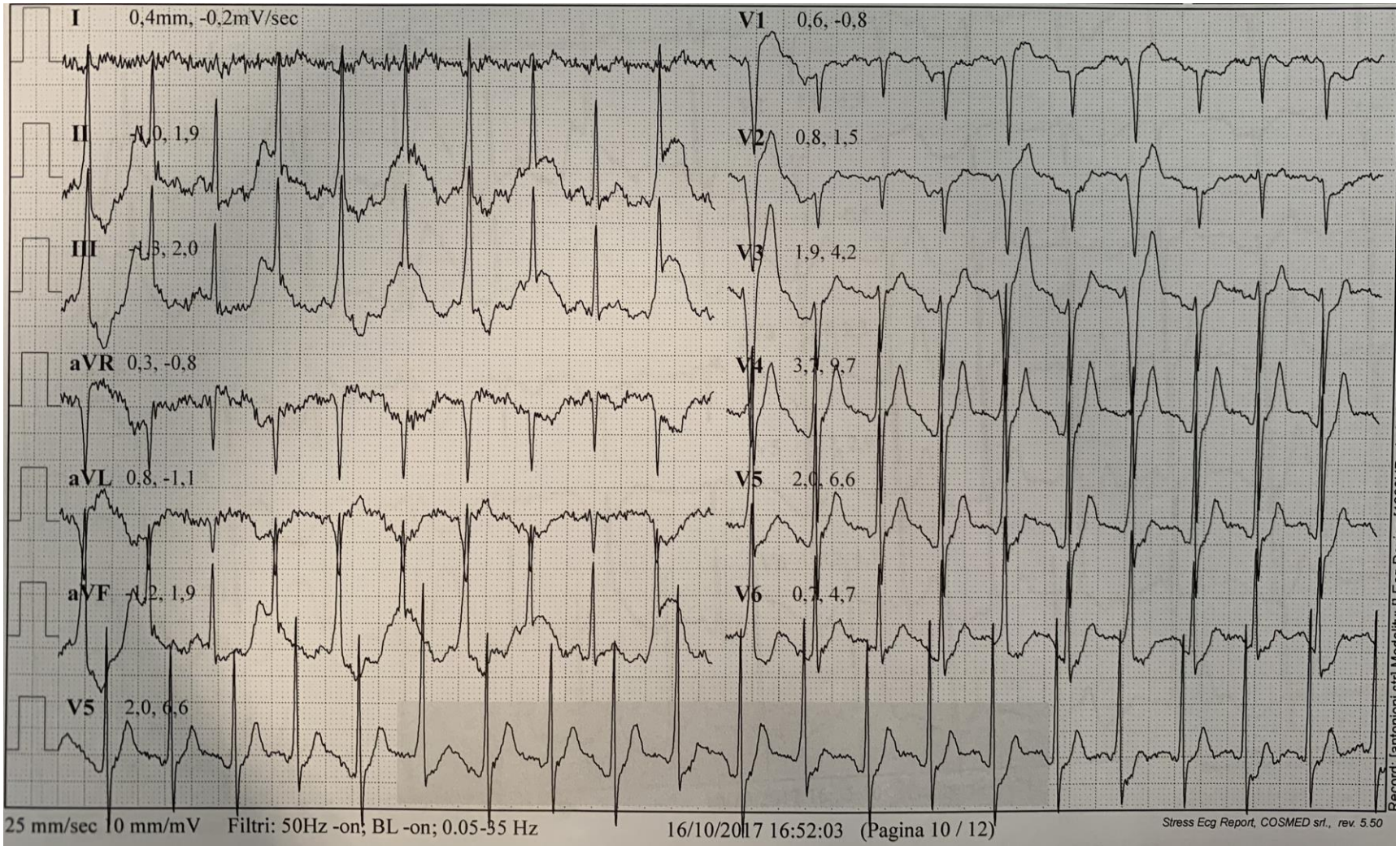

I 0,4mm, -0,2mV/sec
II 1,0, 1,9
III 1,3, 2,0
aVR 0,3, -0,8
aVL 0,8, -1,1
aVF 1,2, 1,9
V5 2,0, 6,6
V1 0,6, -0,8
V2 0,8, 1,5
V3 1,9, 4,2
V4 3,7, 9,7
V5 2,0, 6,6
V6 0,7, 4,7
25 mm/sec 10 mm/mV Filtri: 50Hz -on; BL -on; 0.05-35 Hz
16/10/2017 16:52:03 (Pagina 10 / 12)
Stress Ecg Report, COSMED srl., rev. 5.50

II. Curable and Mild Monomorphic Ventricular Extrasystoles with Paroxysms of Tachycardia

This type is entirely different from the first and, since Gallavardin's original descriptions, its particular features, its benign course, and its curability have become well established. It is characterized by an almost permanent extrasystolic irregularity interspersed with paroxysms of tachycardia of brief duration; in severe forms these paroxysms tend to become longer and longer until they coalesce into prolonged attacks which are interrupted by sinus beats only at long intervals. This trouble specially affects young subjects with healthy hearts and, as a rule, it is very resistant to therapy, including quinidine.

The paroxysms are strictly monomorphic from an electrical point of view; the frequent isolated extrasystoles have the same contour as the beats that constitute the paroxysms. It is a rare functional disorder which may cause vertigo with alternating pallor and flushing of the face such as is seen in Stokes-Adams syndrome; more rarely faintness occurs at the onset, and still more rarely, episodes of transient heart failure occur when the attacks are unusually prolonged (Gallavardin). Yet the essentially benign character of these paroxysms is proved by their persistence over long periods, for example two of our cases were followed over periods of 35 and 24 years: in both the disorder proved perfectly benign and in both it was completely cured in spite of its initial severity.

Case 1. A woman was regularly observed from 33 to 64 years of age (1920–51) with ventricular extrasystoles and paroxysms of tachycardia: these were intense for the first 20 years, but gradually improved and almost completely disappeared during the last 15 or 20 years.

This case was originally published by Louis Gallavardin; further information was supplied by Froment (1932) and by Léon Gallavardin (1946). The patient was re-examined in 1951 when she was perfectly well and completely free from her trouble.

From 1915 to 1932 the rhythm disorder was very persistent with innumerable ventricular extrasystoles. During bad periods there were prolonged paroxysms of tachycardia lasting 5 to 30 days, with asystolic manifestations; they were of such intensity that the severe ascites wrongly led to a diagnosis of tuberculous peritonitis, and in our absence the patient was submitted to operation.

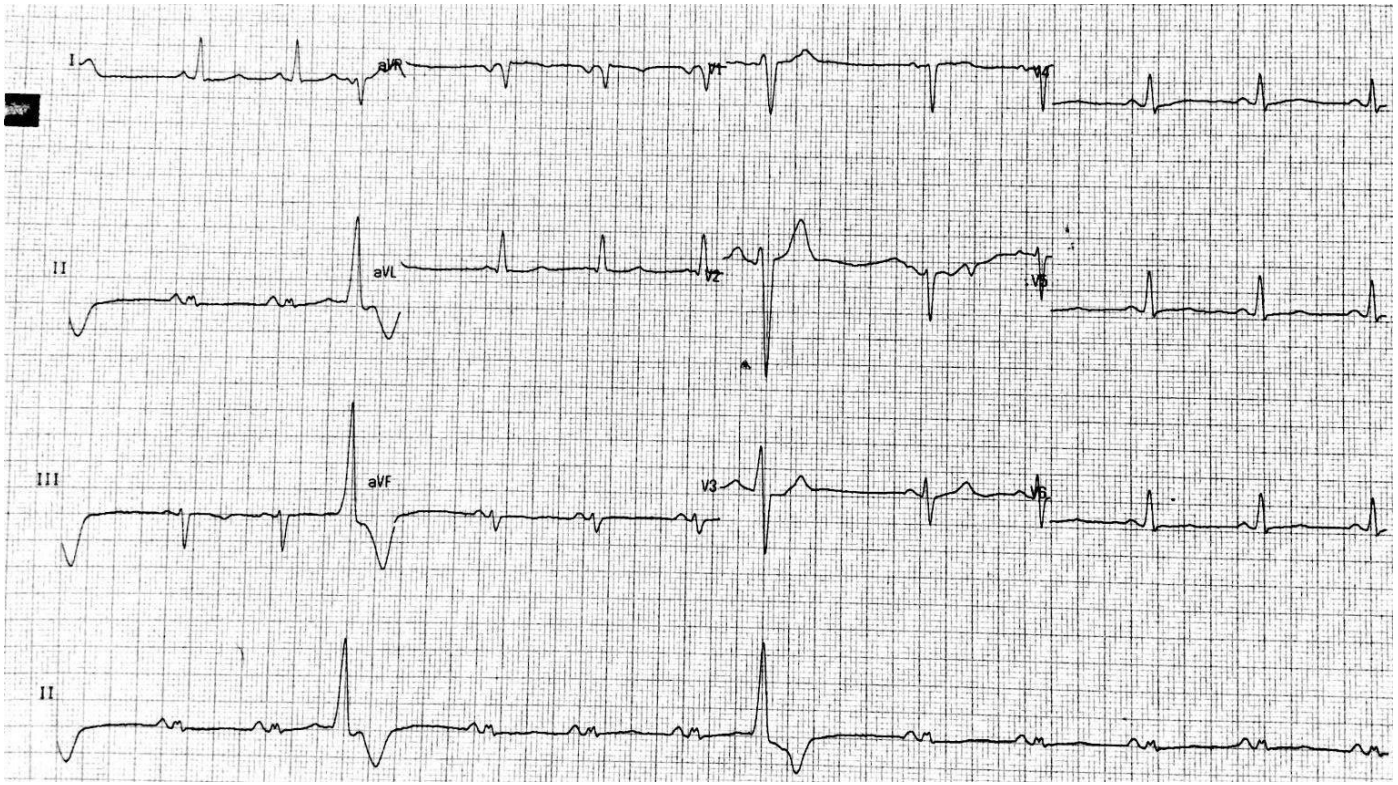
After 1932 the trouble diminished, the paroxysms became shorter and shorter, not exceeding ten seconds, and much less frequent. Since that time she has been working hard as a midwife in a hospital and in 1946 she only presented infrequent extrasystoles with occasional paroxysms. At present she is in excellent health and leads an active life for a 64-year-old woman.

The cardiovascular examination between attacks showed nothing abnormal; in 1946 her blood pressure was 130/80 mm., the size of the heart was normal, and so was the electrocardiogram except for infrequent ventricular extrasystoles similar to the more numerous ones presented before. In 1951, there was no longer any disturbance of rhythm or any sign of heart failure.

Premature ventricular beats characteristics				
Variable	Overall n = 249	LGE + n =28	LGE - n = 221	p
Ventricular arrhythmias morphology				
Monomorphic, n (%)	199 (79.9)	9 (32.1)	190 (86.0)	0.01
- RBBB with superior axis, n (%)	37 (18.5)	5 (55.5)	32 (16.8)	ns
- RBBB with inferior axis, n (%)	28 (14.0)	2 (22.2)	26 (13.7)	ns
- LBBB with superior or intermed axis, n (%)	38 (19.0)	2 (22.2)	36 (18.9)	ns
- LBBB with inferior axis, n (%)	87 (43.7)	0	87 (45.8)	0.001
- Fascicular, n (%)	9 (4.5)	0	9 (4.7)	ns
Polymorphic, n (%)	50 (20.0)	19 (65.5)	31 (14.0)	<.001
Ventricular arrhythmias complexity				
Repetitive, n (%)	178 (71.5)	27 (96.4)	151 (68.3)	ns
- monomorphic, n (%)	132 (74.2)	9 (33.3)	123 (81.5)	0.023
- polymorphic, n (%)	46 (25.8)	18 (66.7)	28 (18.5)	<.001
Isolated, n (%)	71 (28.5)	1 (3.6)	70 (31.7)	0.011
- monomorphic, n (%)	67 (93.0)	0	67 (94.3)	ns
- polymorphic, n (%)	4 (5.6)	1 (50.0)	4 (5.7)	0.012
Response to exercise testing				
Decrease/Suppression, n (%)	187 (75.1)	13 (46.4)	174 (78.7)	ns
Increase/Persistence, n (%)	62 (24.9)	15 (53.6)	47 (21.3)	0.008
- isolated, n (%)	22 (35.5)	3 (20.0)	19 (40.4)	ns
- repetitive, n (%)	40 (64.5)	12 (80.0)	28 (59.6)	ns
- repetitive and polymorphic, n (%)	15 (24.2)	8 (53.3)	7 (14.9)	0.027
24 h ECG monitoring				
PVBs, n	7126±9358	1687±3157	7747±9630	0.002
Couplets and/or triplets, n (%)	148 (59.4)	22 (78.6)	126 (57.0)	ns
Non-sustained VT, n (%)	44 (17.7)	6 (21.4)	38 (17.2)	ns

C.L., 51 y-o., female

- Palpitations and presyncope
- ECG evidence of PVBs



- ECG-Holter: NSVT
- Cardiac magnetic resonance:

INDICAZIONI

Note Presincopi in paziente con TVNS e cardiopatia dilatativa.

TECNICA

Qualita dello studio: - **buono**

Sequenza: - **SSFP** - **DYN FFE** - **IRTFE**

VALUTAZIONE FUNZIONALE VENTRICOLO SINISTRO

LVEDV ml	156	LVSV ml	62	PC litri/min	3,0	**Note**
LVES ml	94	Massa gr.		Diam.Telediag. mm	55	
LVEDVind ml/m2	93	Massa ind.		Diam.Telesist. mm		
LVESind ml/m2	56	LVEF %	39,0			

VALUTAZIONE FUNZIONALE VENTRICOLO DESTRO

RVEDV ml	121	RVSV ml	59	PC litri/min	2,9	**Note**
RVES ml	62	RVIT AC		RVEF %	49,0	
RVEDVind ml/m2	72	RVIT BIV				
RVESind ml/m2	43,0	RVOT BIV				

MORFOLOGIA CARDIACA

- **Ventricolo Sinistro:**

Dimensionie volumi aumentati con riduzione moderata della funzione sistolica globale in relazione ad ipocinesia globale con acinesia a carico della parete anteriore ed inferiore .

- **Ventricolo Destro**

Normali volumi endocavitari con funzione sistolica globale lievemente ridotta.

- **Valvola Aorta**

Diam. cm | Ar. Flusso cm2 | Area planimetrica cm2

- **Valvola Mitrale**

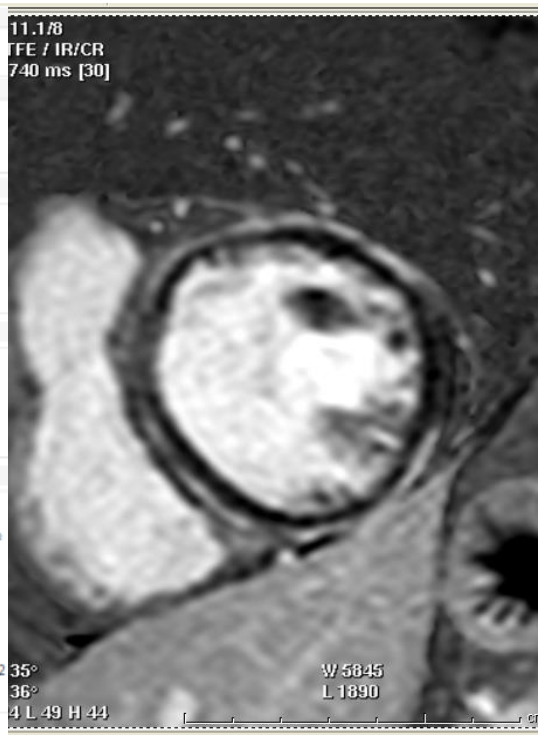
Diam. cm | Ar. flusso cm2 | Area planimetrica cm2

POTENZIAMENTO MIOCARDICO POST - CONTRASTO

Esteso potenziamento subepicardico pressocchè circonferenziale con area intramiocardica a carico del SIV .

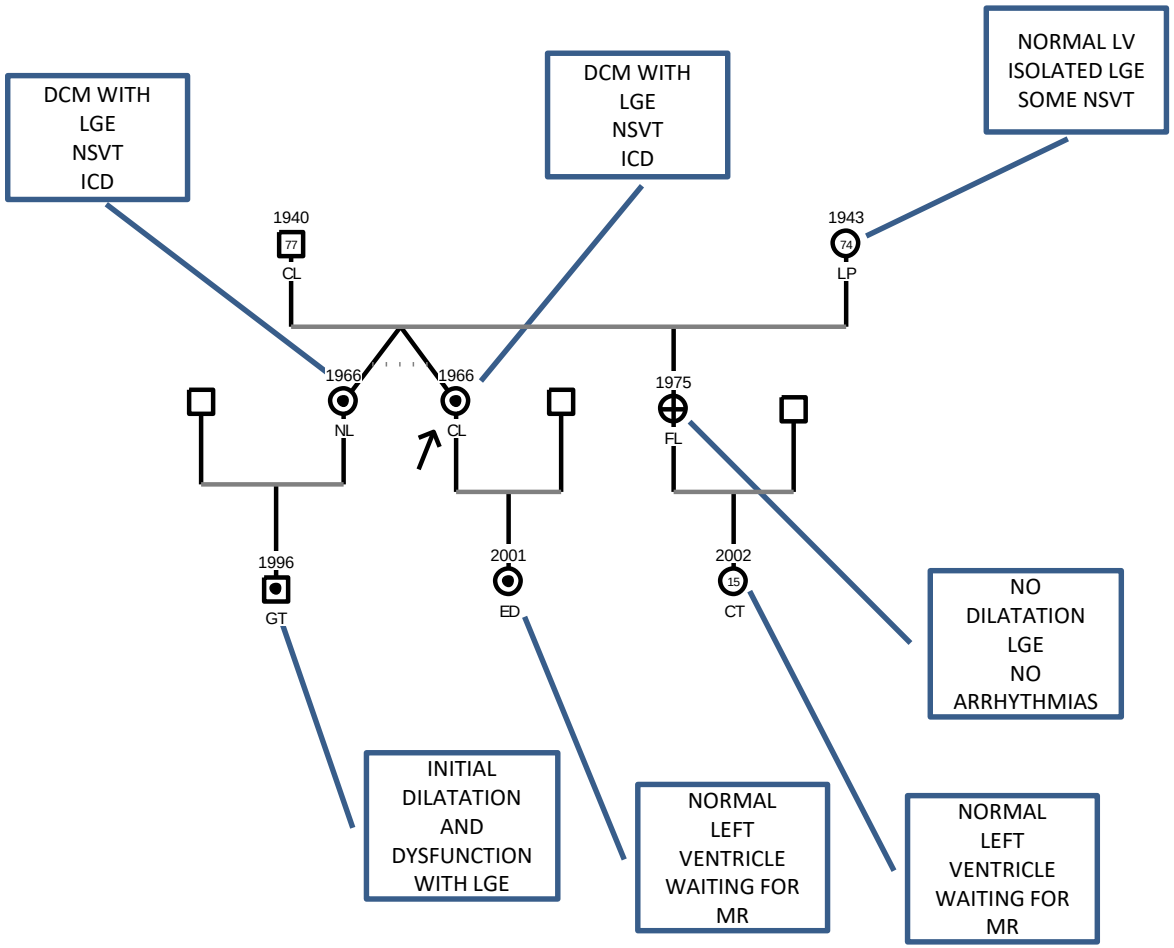
ALTRI REPERTI

Atrio destro 17 cm2.Atrio sinistro 19 cm2.Jet di rigurgito valvolare tricuspidale.Sdoppiamento e dislocazione destroconvessa del SIA.

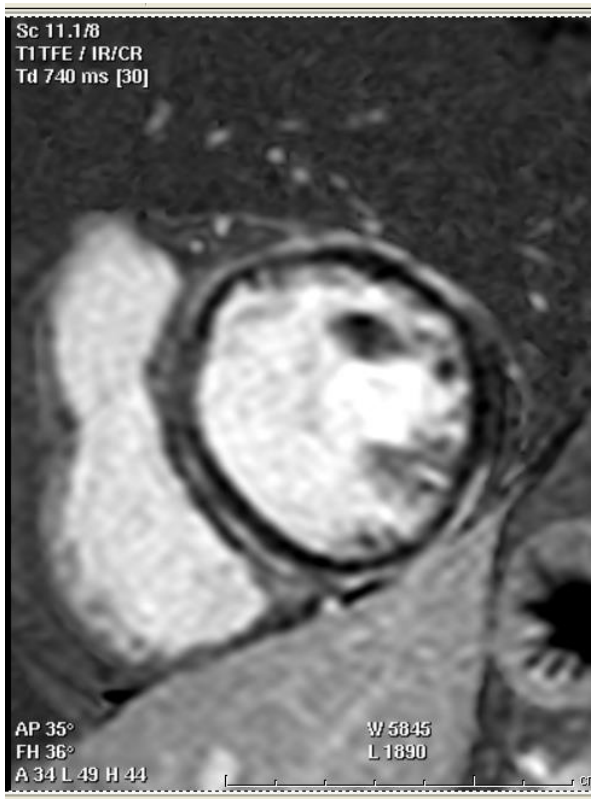


- EPS: presyncopal SVT inducibility
- ICD implantation
- Genetic testing:

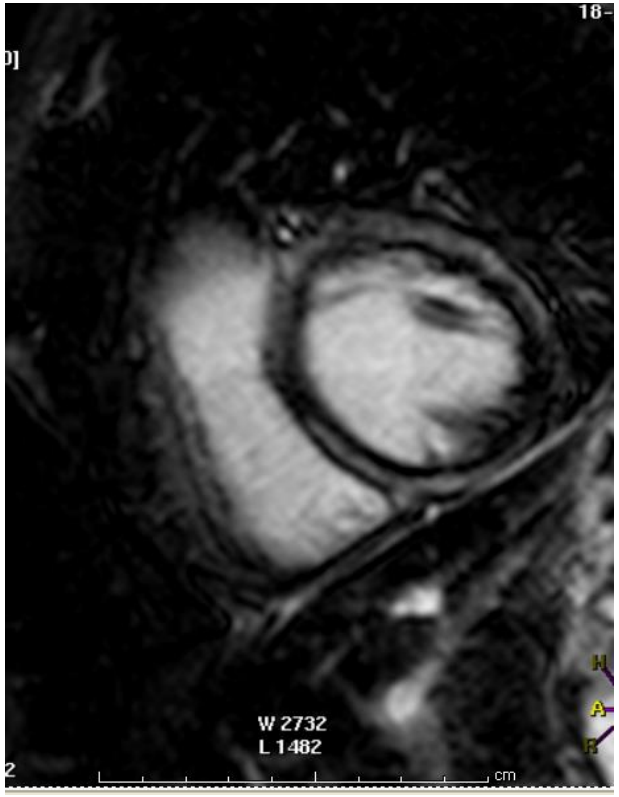
Gene	Variant	Result	Pathogenicity	Population frequency	Number of references
FLNC	NP_001449.3:p.Arg81Alafs*15 NM_001458.4:c.241delC NC_000007.13:g.128470932delC	Heterozygosis	Pathogenic	Mutation (not found in controls)	0
LDB3	NP_009009.1:p.Ile558Val NM_007078.2:c.1672A>G NC_000010.10:g.88476524A>G	Heterozygosis	Unknown clinical significance	Mutation (not found in controls)	6


DCM WITH
LGE
NSVT
ICD
DCM WITH
LGE
NSVT
ICD
NORMAL LV
ISOLATED LGE
SOME NSVT
1940
77
CL
1943
74
LP
1966
NL
1966
CL
1975
FL
1996
GT
2001
ED
2002
15
CT
NO
DILATATION
LGE
NO
ARRHYTHMIAS
INITIAL
DILATATION
AND
DYSFUNCTION
WITH LGE
NORMAL
LEFT
VENTRICLE
WAITING FOR
MR
NORMAL
LEFT
VENTRICLE
WAITING FOR
MR

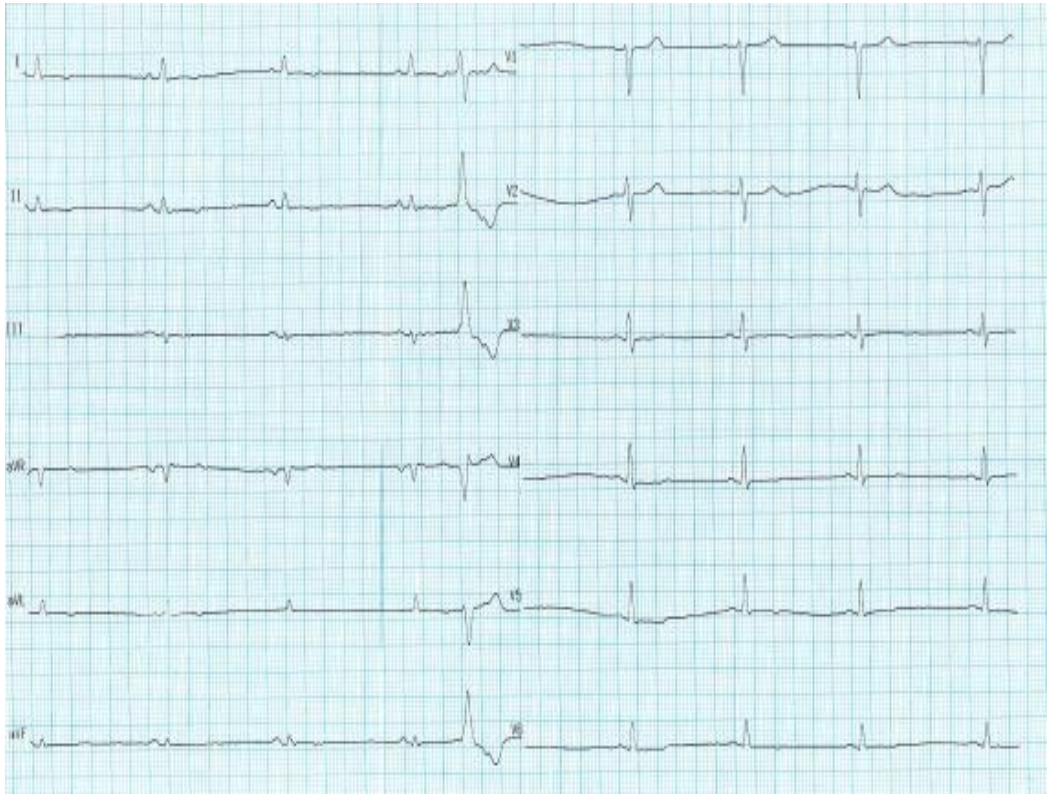
CL, index case,51 yo



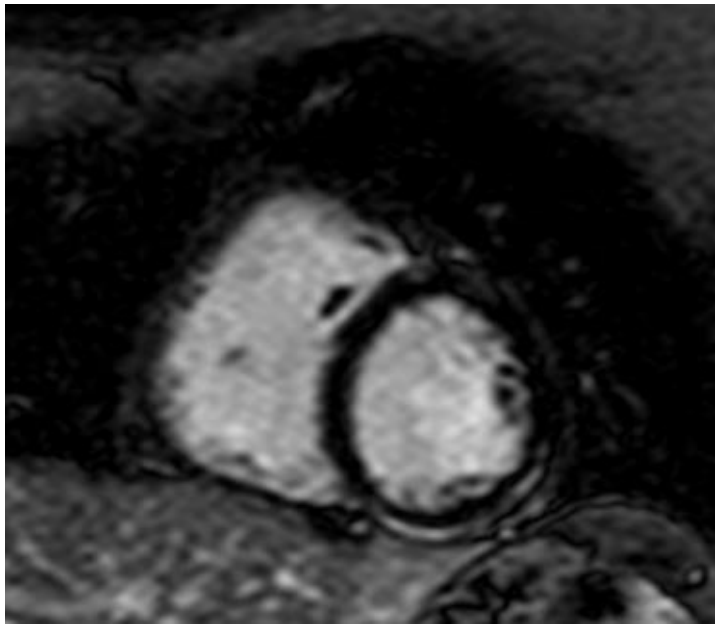
NL, twin sister, 51 yo



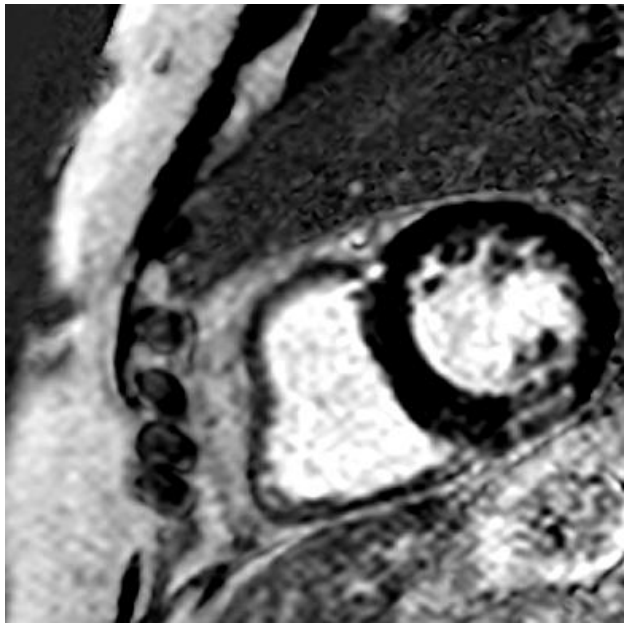
N.L.



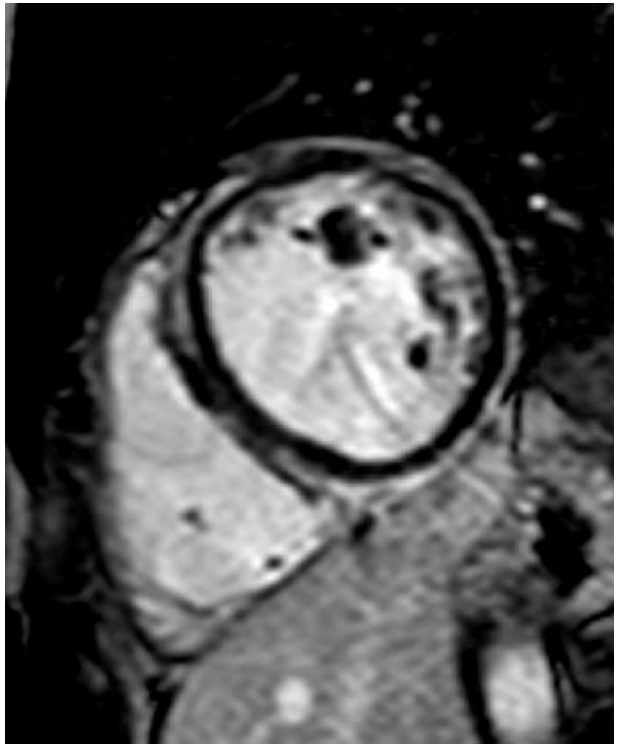
FL, sister, 42 yo

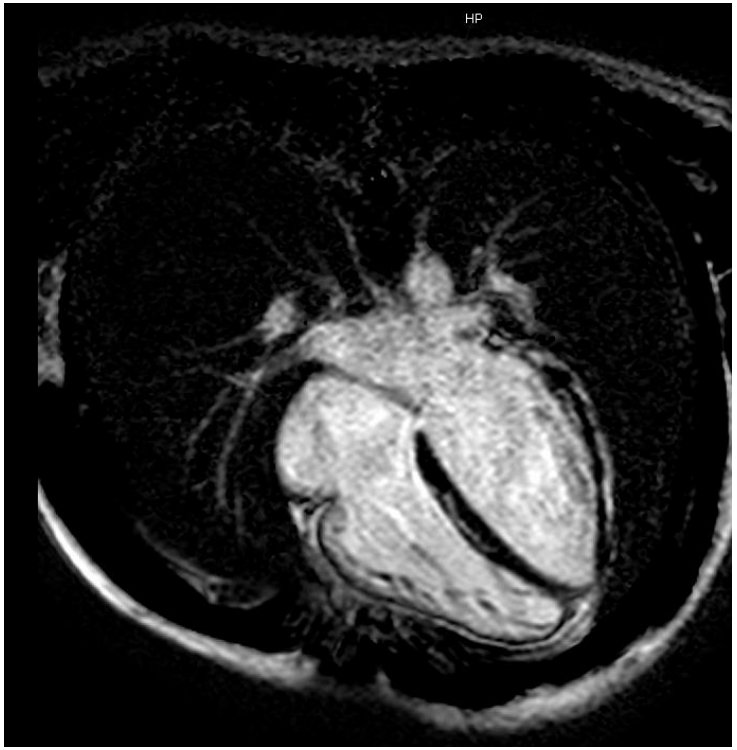


LP, mother, 74 yo

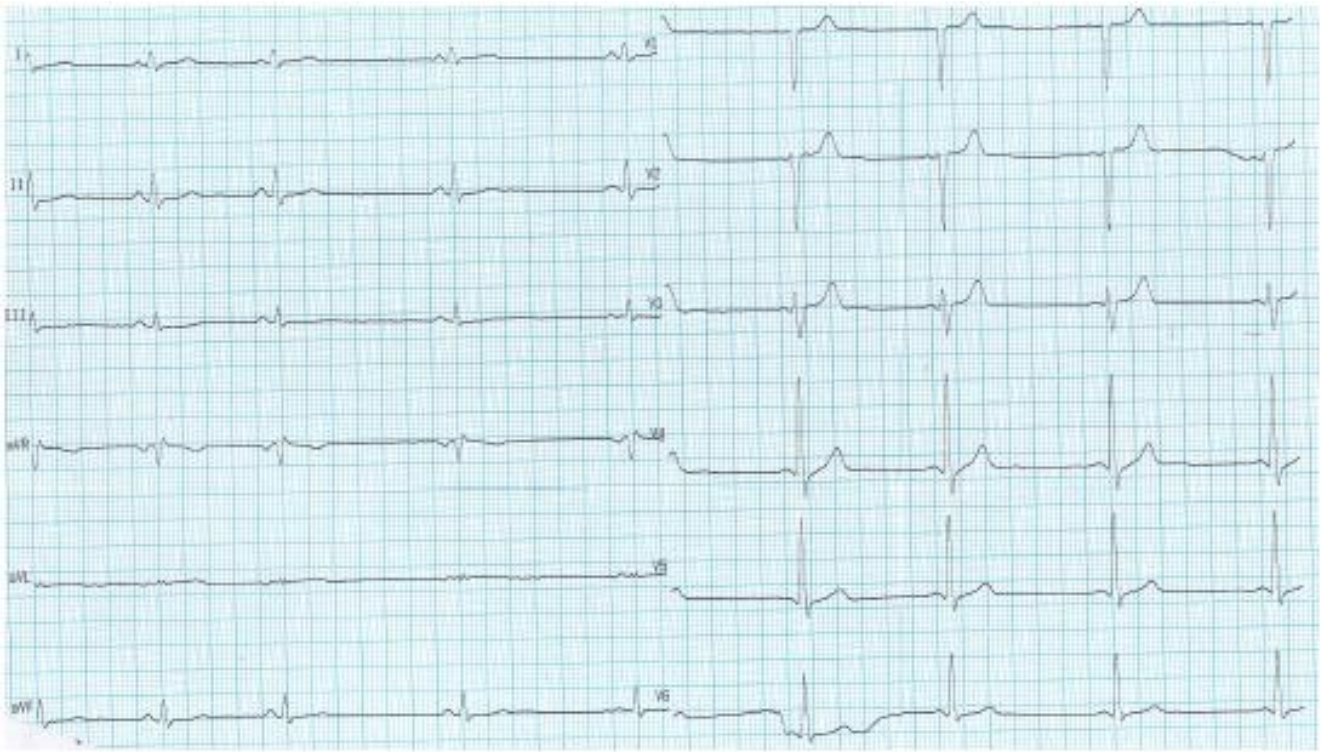


GT, nephiew

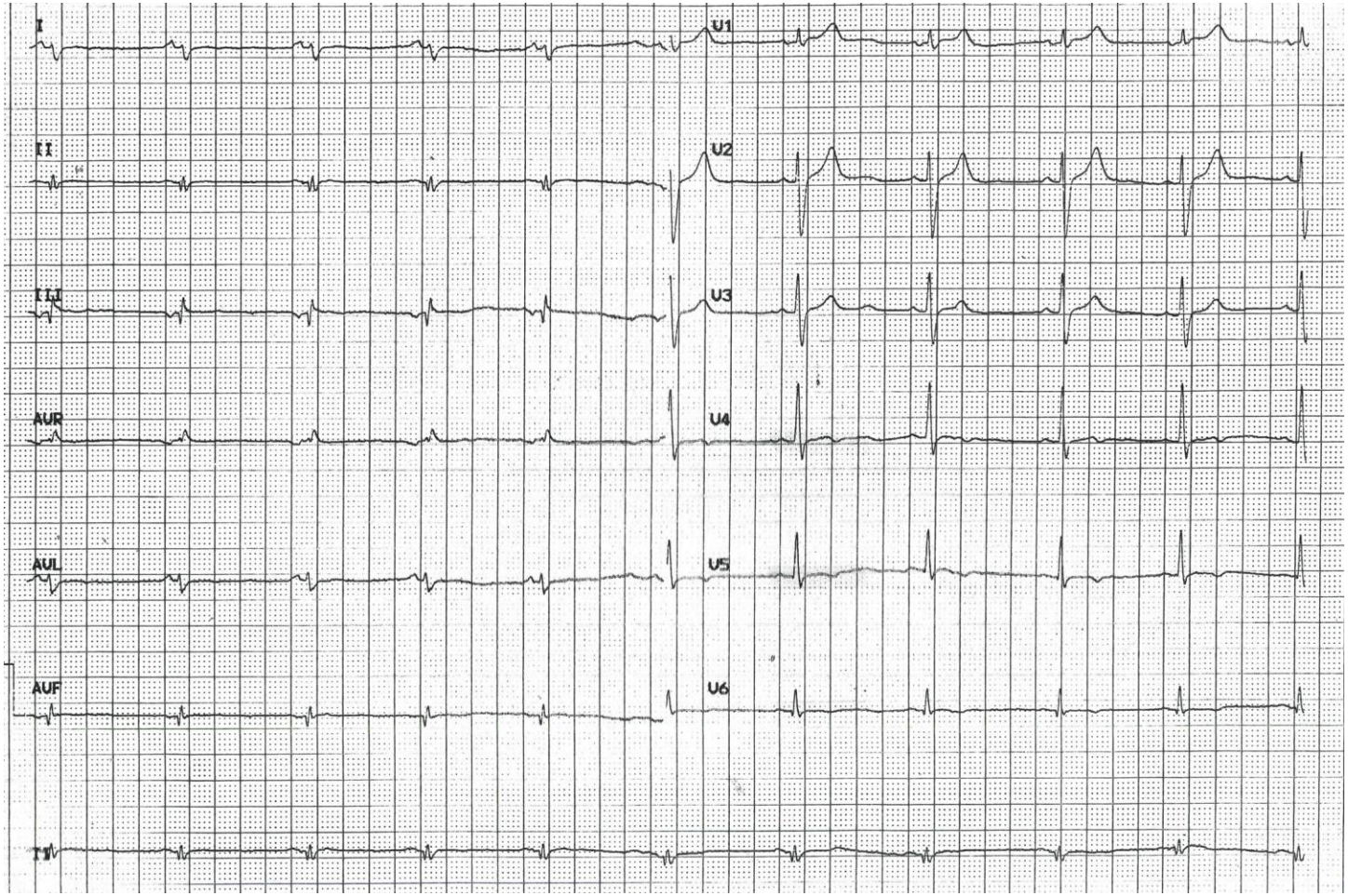


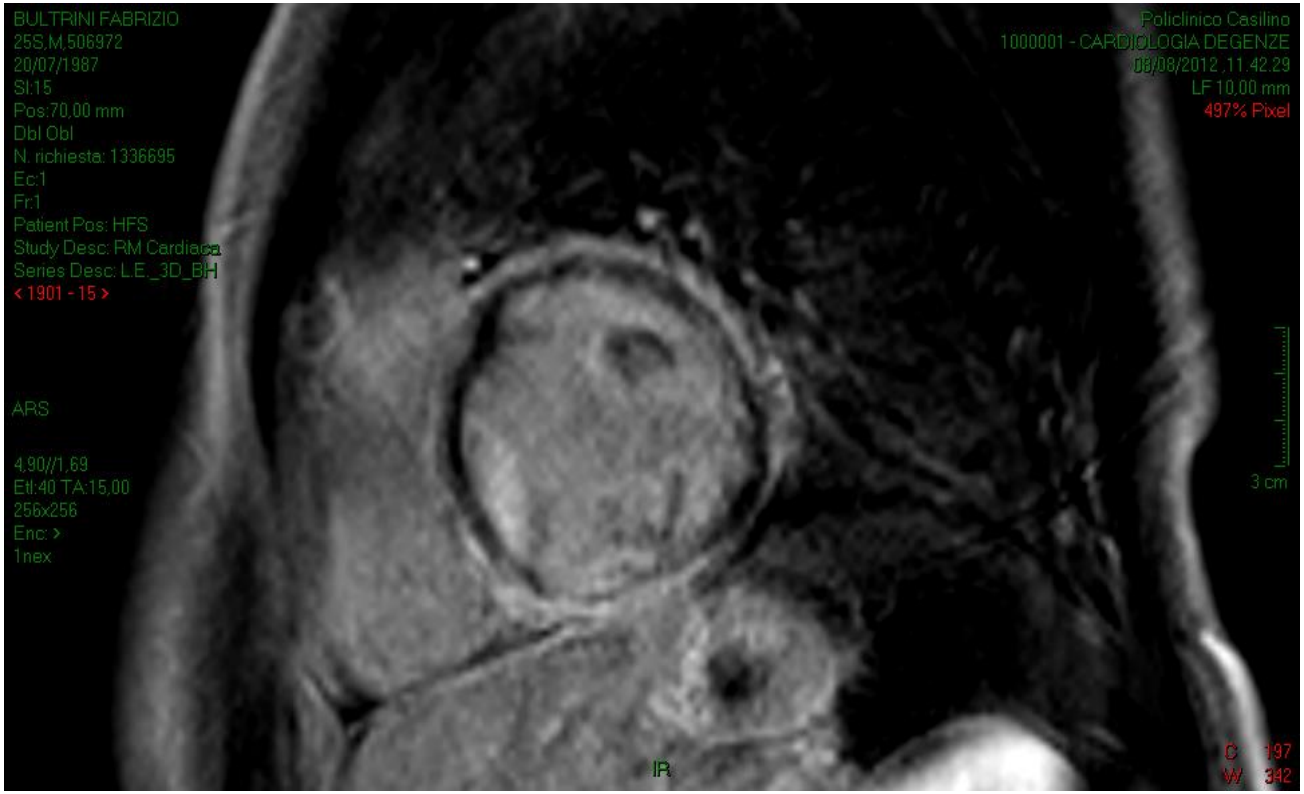


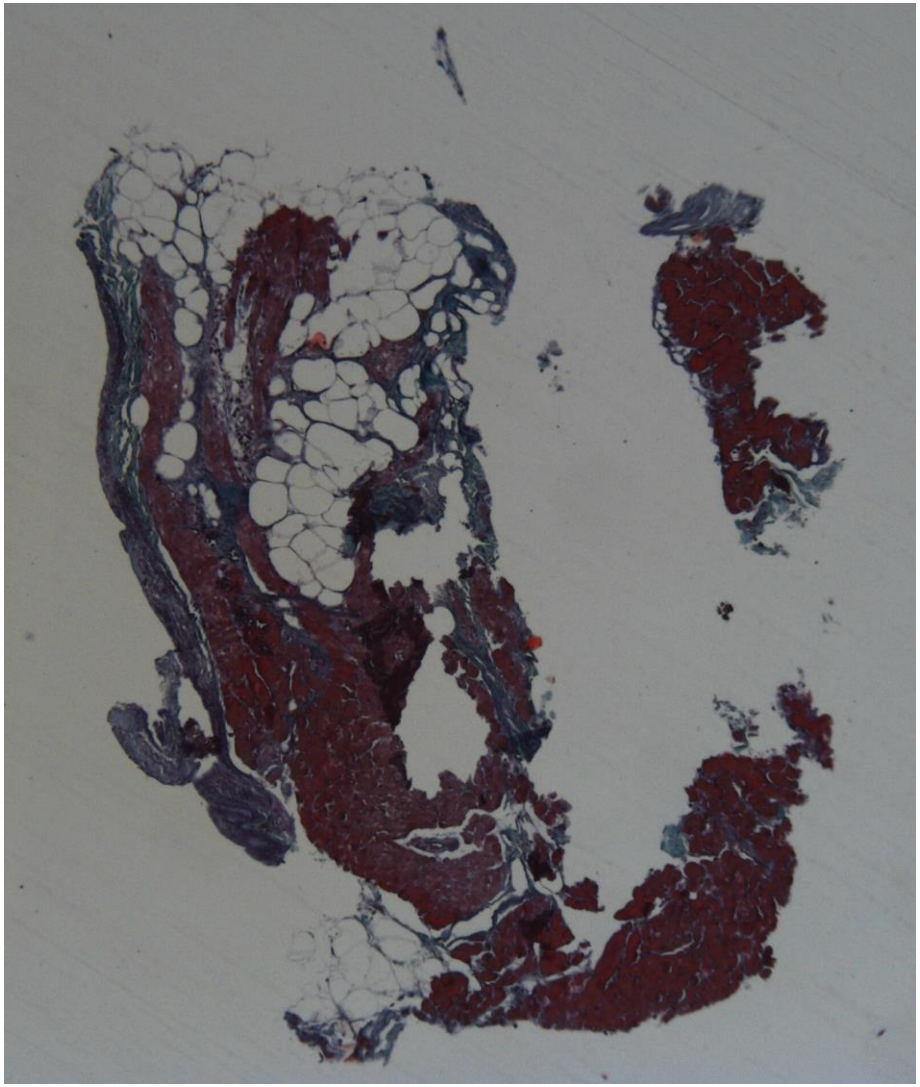
- GT, nephiew

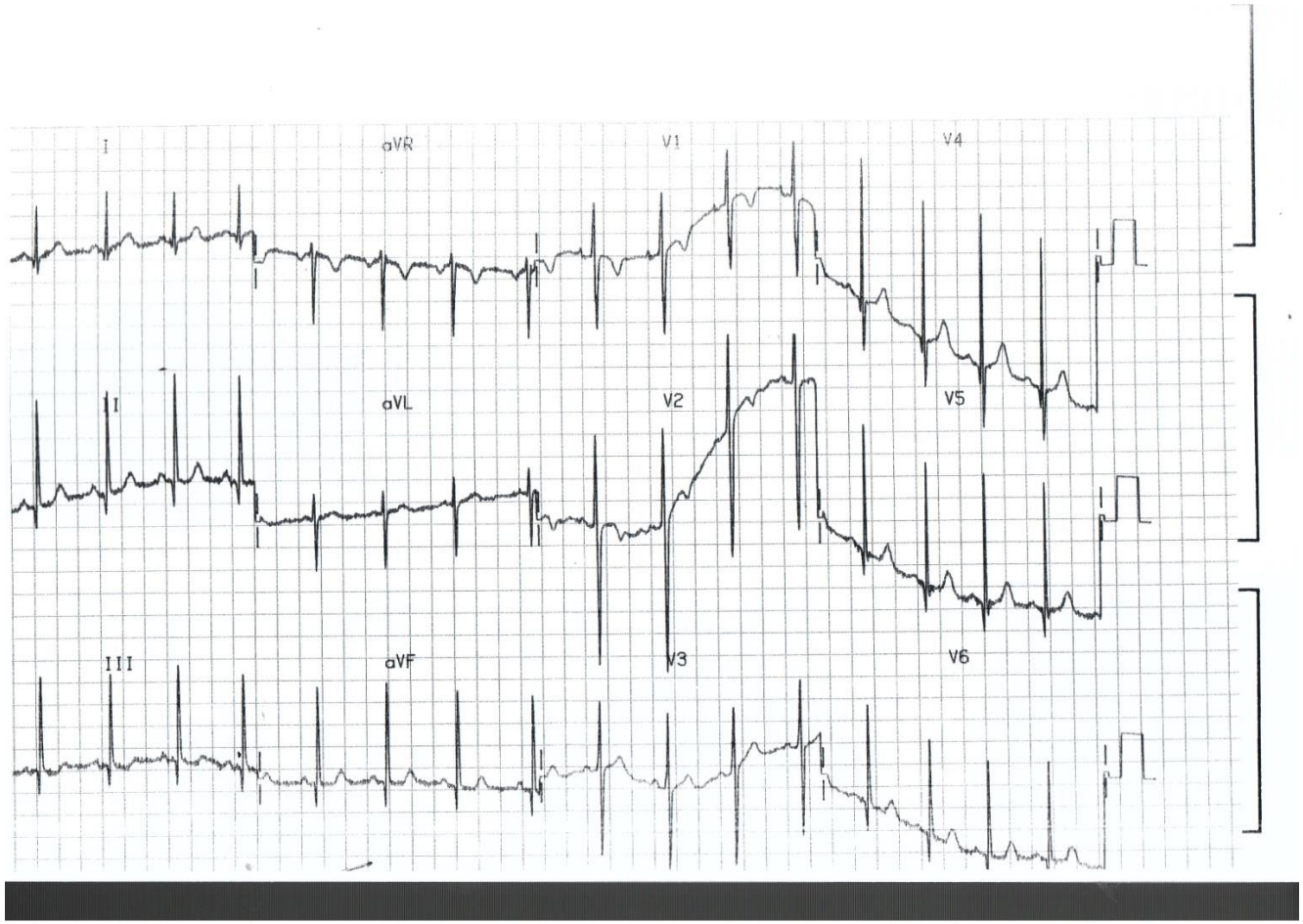


BUL. #1




BULTRINI FABRIZIO
25S,M,506972
20/07/1987
SI:15
Pos:70,00 mm
Dbl Obl
N. richiesta: 1336695
Ec:1
Fr:1
Patient Pos: HFS
Study Desc: RM Cardiaca
Series Desc: L.E._3D_BH
< 1901 - 15 >
ARS
4,90//1,69
Etl:40 TA:15,00
256x256
Enc: >
1nex
Policlinico Casilino
1000001 - CARDIOLOGIA DEGENZE
08/08/2012 ,11.42.29
LF 10,00 mm
497% Pixel
3 cm
IR
C 197
W 342

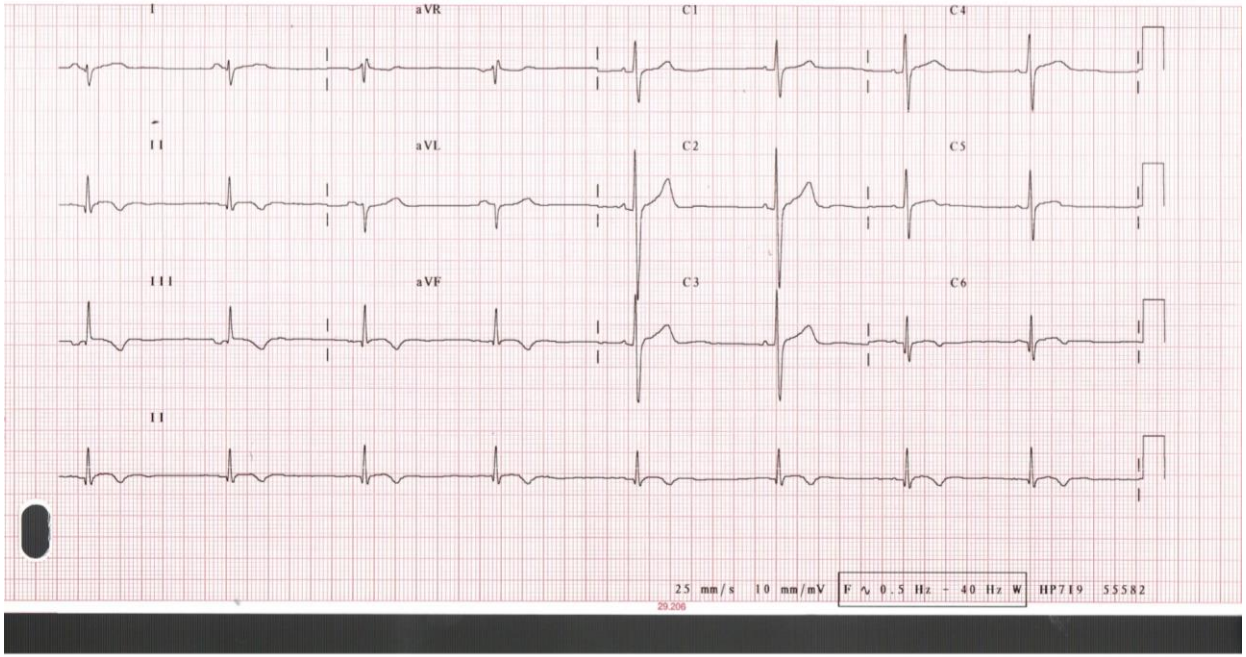



I
aVR
V1
V4
II
aVL
V2
V5
III
aVF
V3
V6

Freq.	47
PR	164
QRSD	92
QT	403
QTc	356

--Asse--

P	-12
QRS	120
T	-59

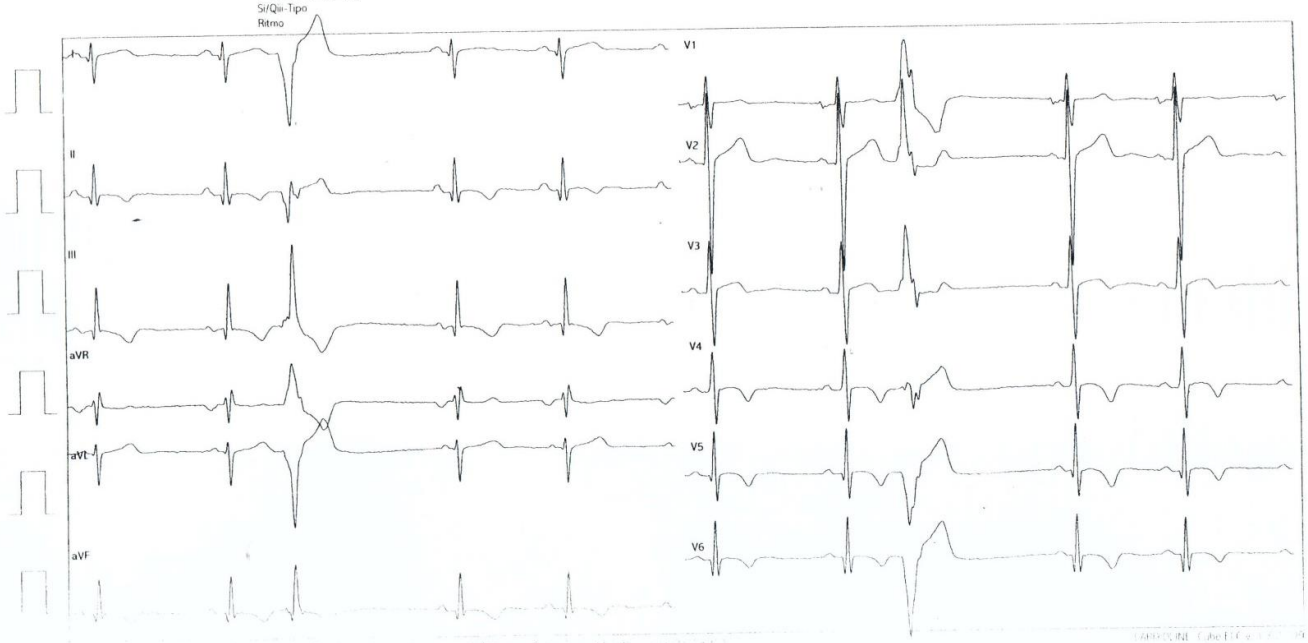


Data di nascita 30/04/1987
Altezza ____ Peso ____
Sesso: M
PAS/PAD ___/___ mmHg

P 116 ms P-Q 154 ms Qrs 96 ms
Q-T 412 ms Q-T/RR 0.433 Q-T Ref: 108 %
Hr: 63 /Min.
P 0.15 Axis 50 Deg
Qrs 1.15 Axis 119 Deg
T 0.28 Axis -72 Deg
Si/Qii-Tipo
Ritmo

Ritmo Sinusale
Con 1 Extrasistoli Ventricolari E Pausa Compensatoria
Interpretazione Qrs-T:
Probabile Ipertrofia Ventricolare Destra
Ritardo Destro

Firmato il: 19/01/2010
da: DOTT.



11/4/2013: autopsia

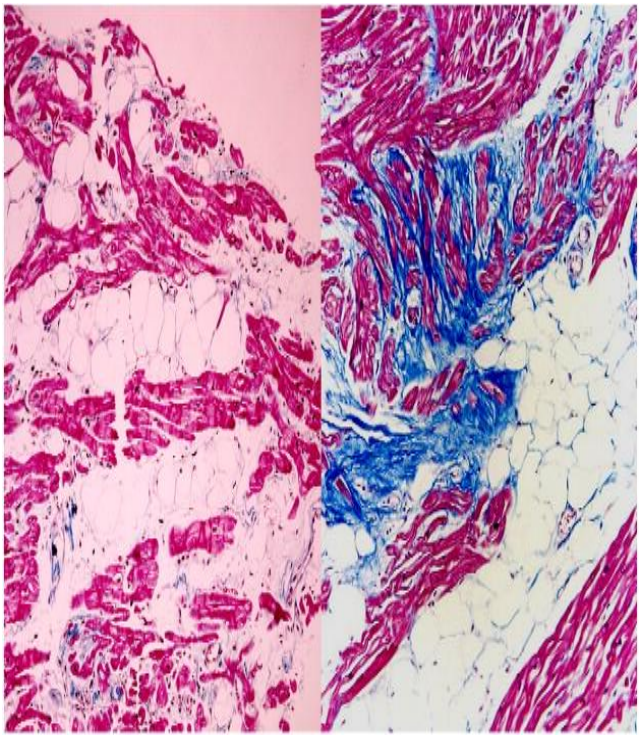
Ventricolo sinistro dilatato ed ipertrofico

dilatazione ventricolo destro

Sostituzione fibroadiposa biventricolare

RV patchy transmurale

LV subendocardica



LEFT POSTERIOR FASCICULAR BLOCK

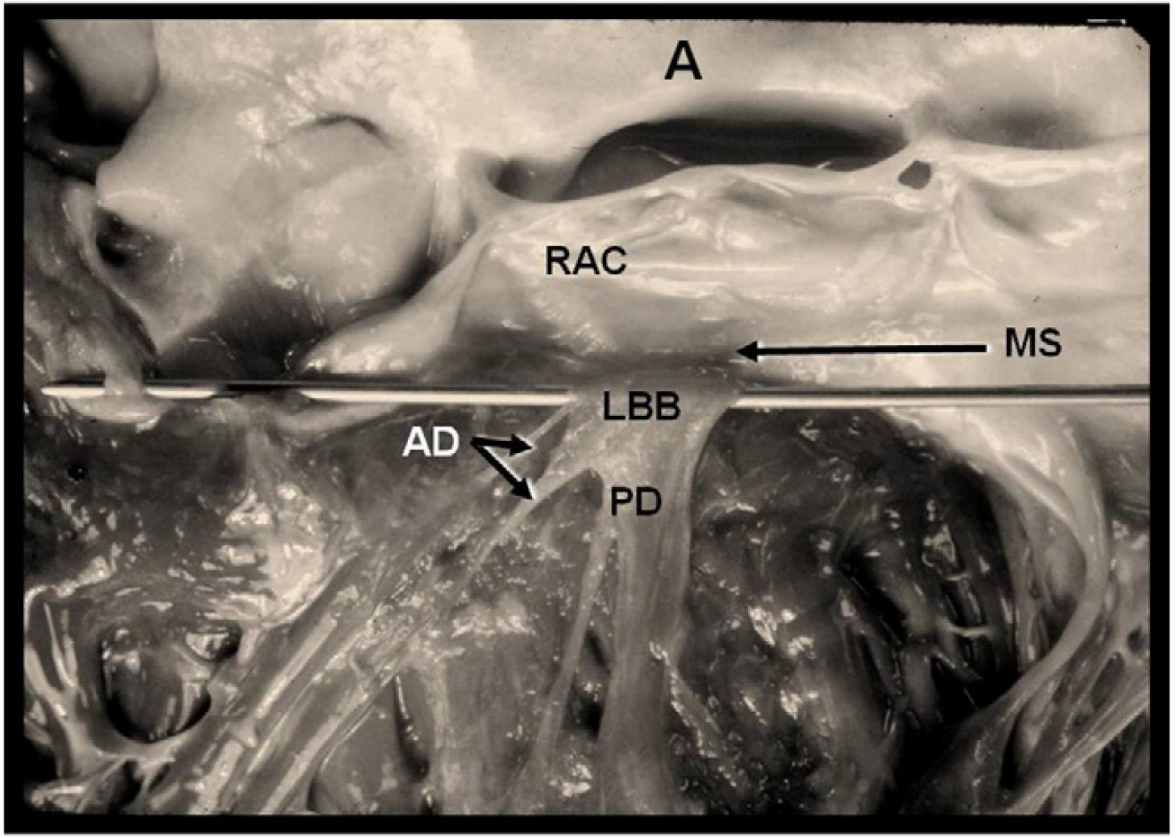
The clinical presentation of conduction disturbances in order of decreasing incidence is LAH, RBBB, LBBB, and last, LPH.

This rank depends not only on the intrinsic, anatomic, genetically determined differences among branches and fascicles but also on the manner in which the intraventricular conduction system is exposed to the various pathological processes of the surrounding cardiac structures.

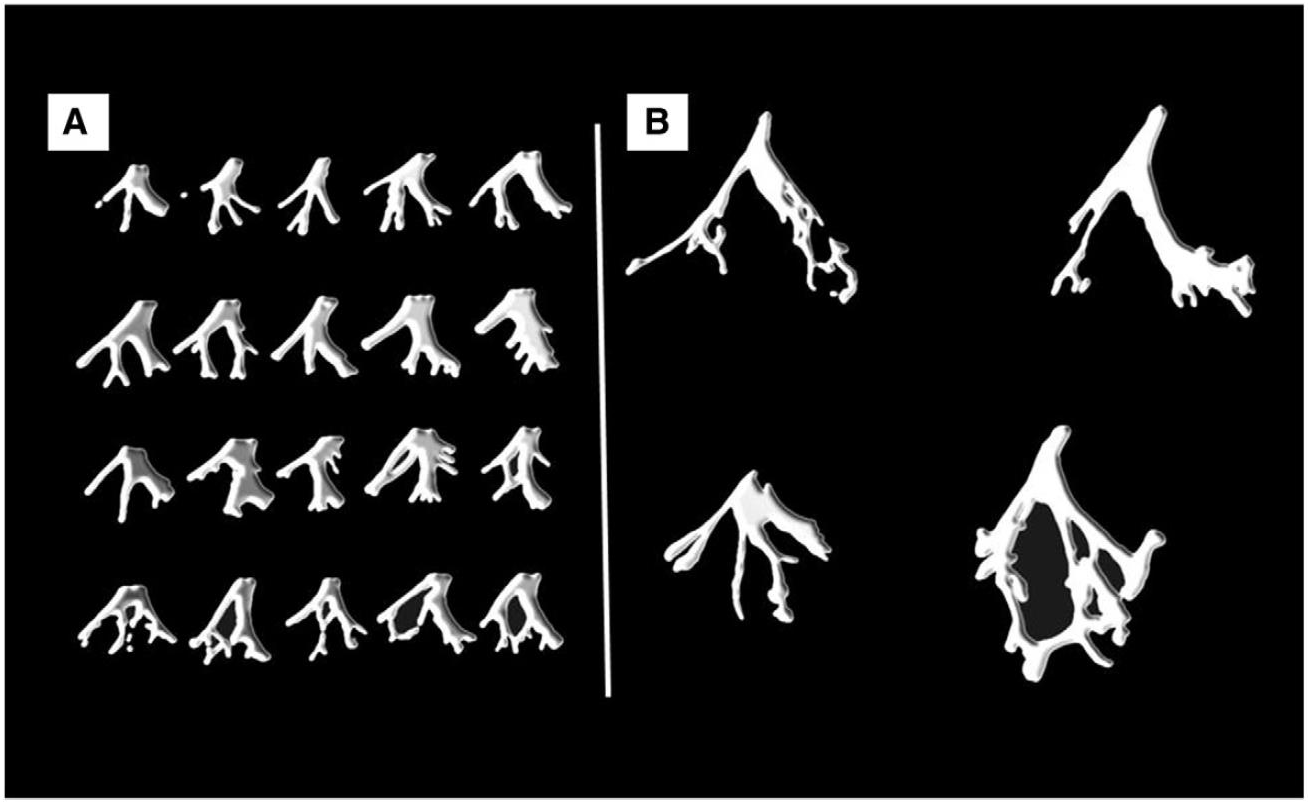
In fact, the posterior division is the least vulnerable segment of the whole system because it is short and wide, it is located in the inflow tract of the left ventricle, which is a less turbulent region than the outflow tract, it has double blood supply (from the anterior and posterior descending coronary arteries), and it is not related to structures that are so potentially dangerous.

Isolated LPFB is very rare finding (0.1% of all intraventricular conduction defects).

Little data exists regarding the prevalence of LPFB. Haataja et al., in Finland, based on the Health 2000 Survey conducted in 2000/2001 studied 6354 individuals aged 30.. In thislarge population only 8 patients had LPFB

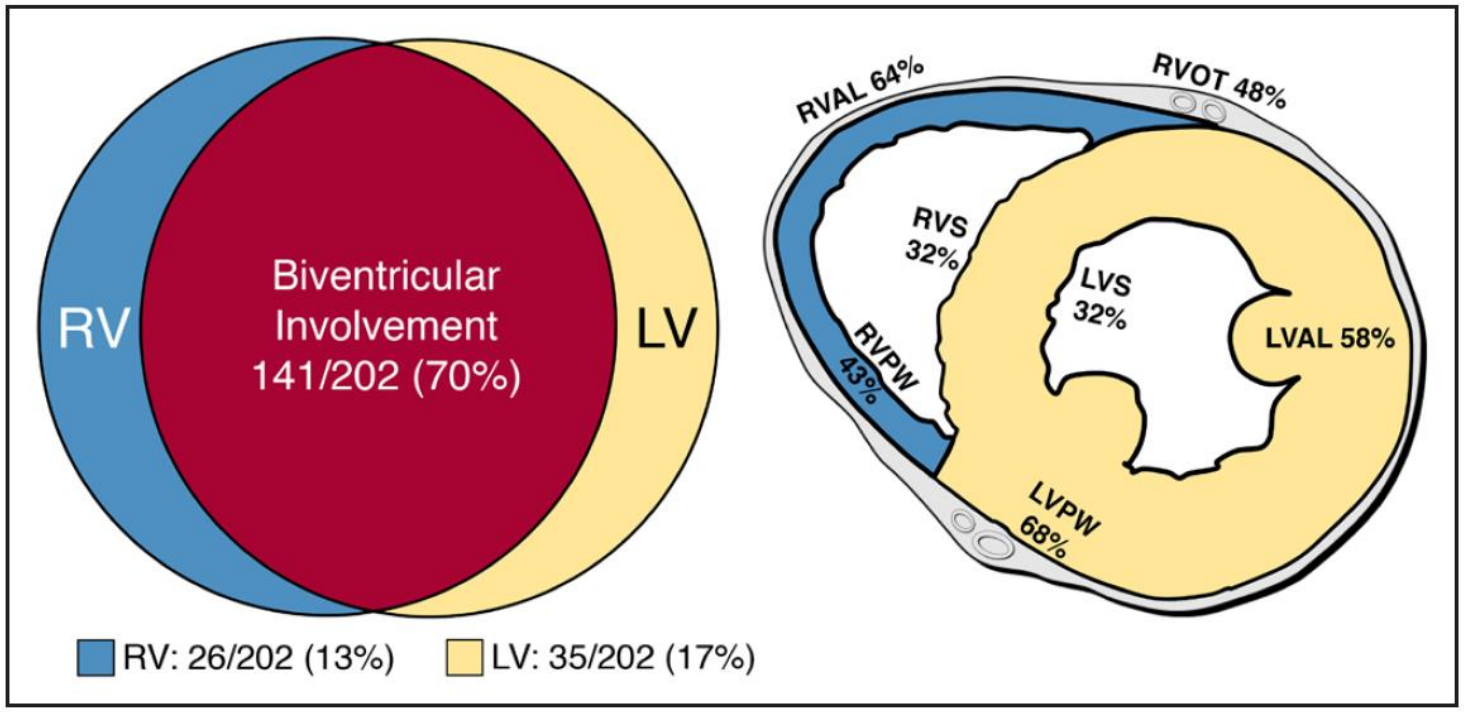


Human heart. The LBB (LBB) emerges in the subaortic region. The membranous septum (MS) is almost absent and the aortic valve lies directly over the LBB,which gives off the anterior division (AD) and posterior division (PD) from its very beginning. The membranous septum is strikingly small or practically absent in this case. The distance between the branching portion of the bundle of His from the aortic valve depends on the size of the MS. The larger the MS, the lesser the possibility that the aortic valve pathology involves this crucial part of the conducting system. A: aorta; RAC right aortic cusp.

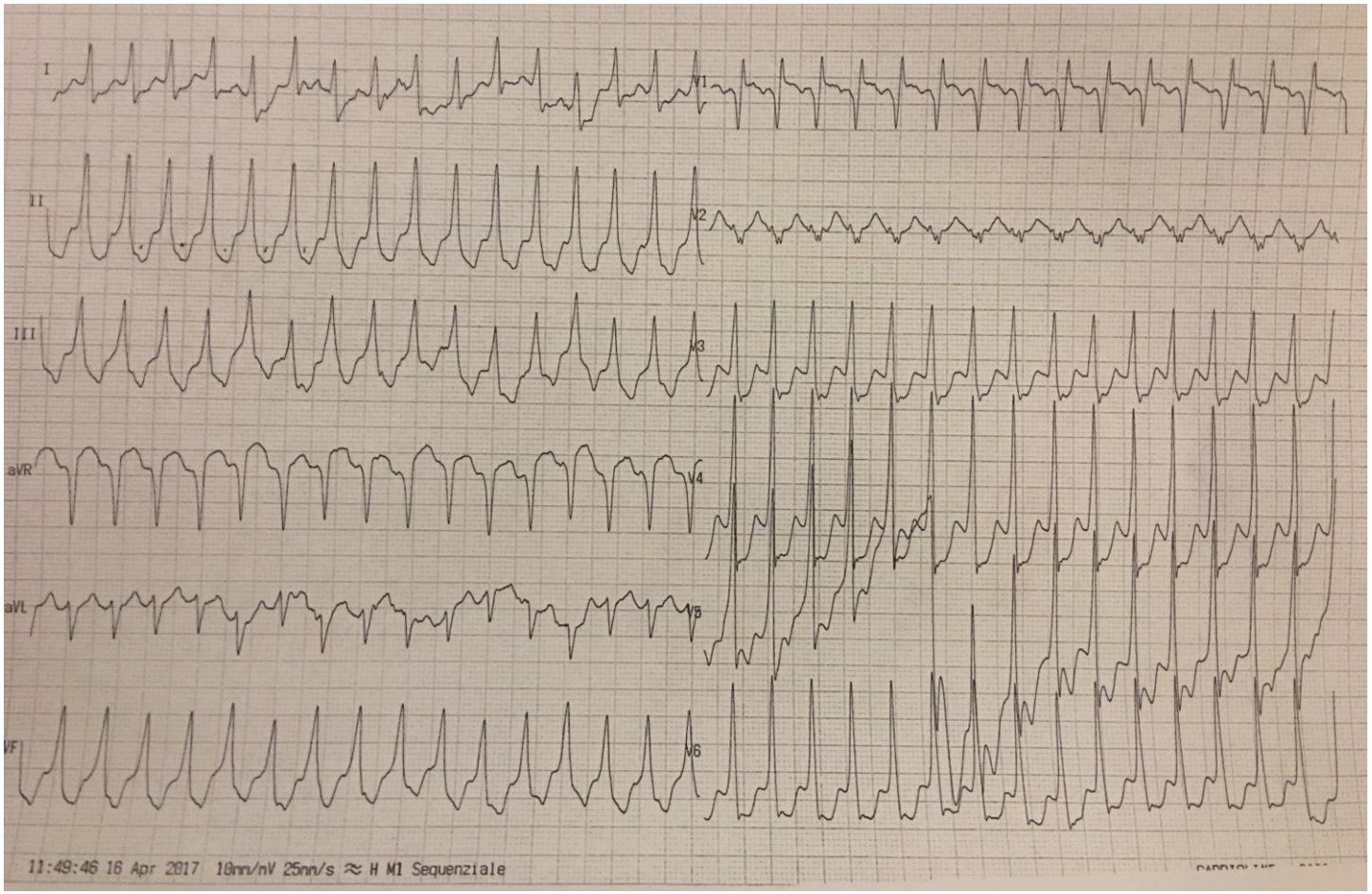


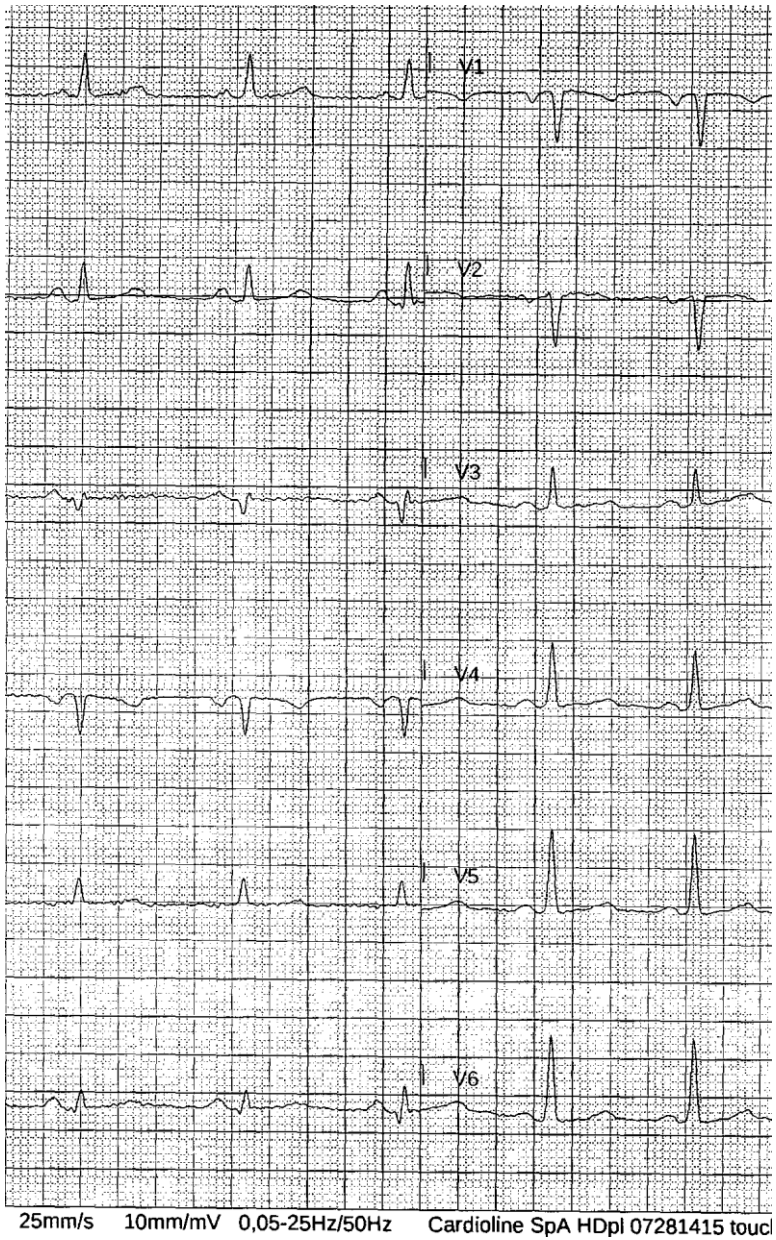
A. Fig. 2 from Demoulin JC, Kulbertus HE [24]. Diagrammatic sketches of the LBB conduction system in twenty human normal hearts. These sketches depict the anatomy from serial histologic sections of left septal myocardium. The LBB and its subdivisions were identified by their subendocardial location and histological features typical of the conducting fibers. B: Figure 28, Chapter 2 from RosenbaumMB et al. [13]. Four human LBB systems dissected and separated from the heart considered the main prototypes observed in our material. In every case, the main LBB is short and its divisions longer depicting a wider posterior division as compared with the anterior one. The bottom left LBB shows what can be considered a medial left septal fascicle arising at the bifurcation of the LBB, although it actually emerges from a wide posterior division, a pattern that can also be observed in the examples of Fig. 14 A in most cases.

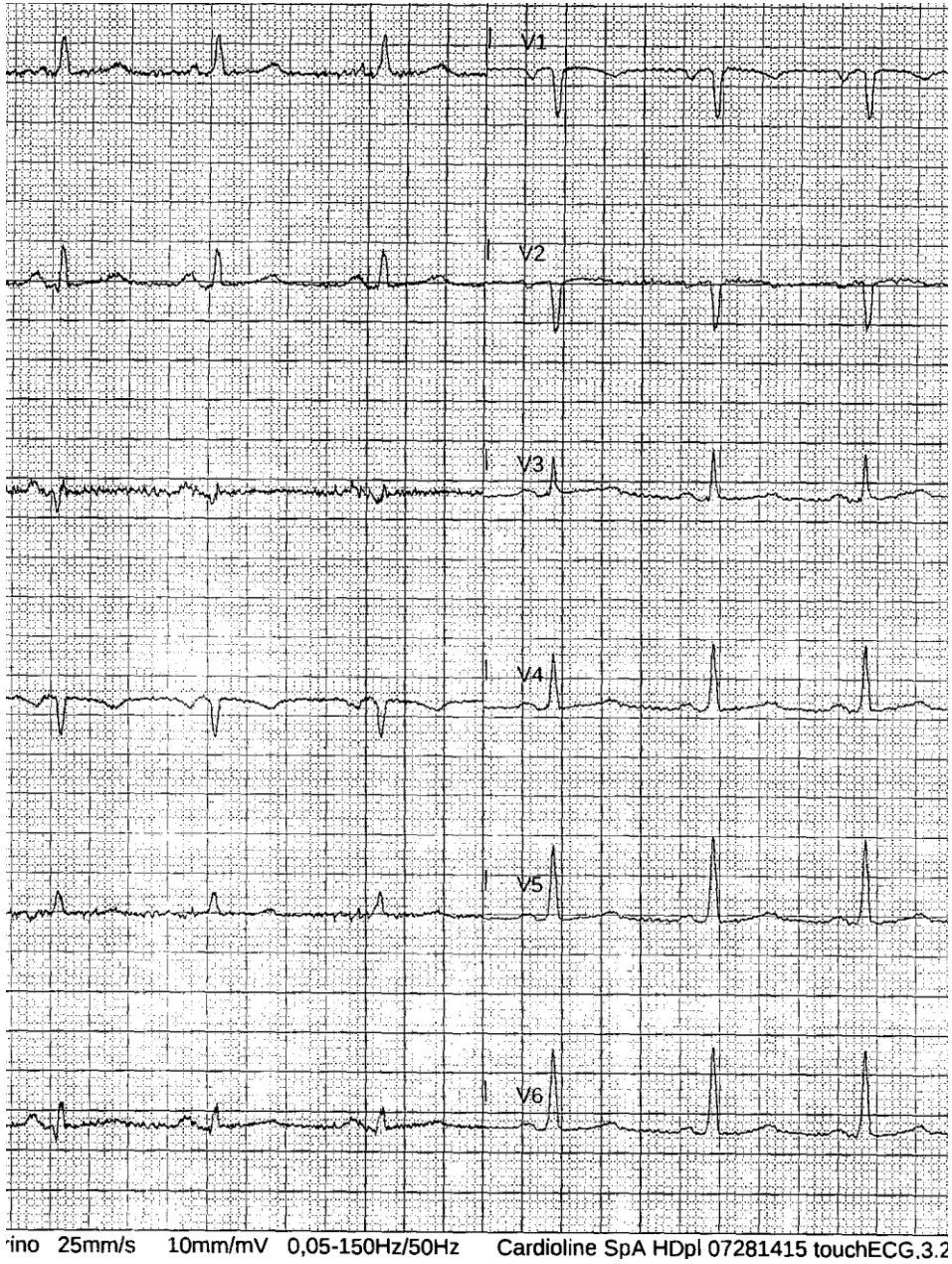
Distribution and location of disease involvement in arrhythmogenic cardiomyopathy

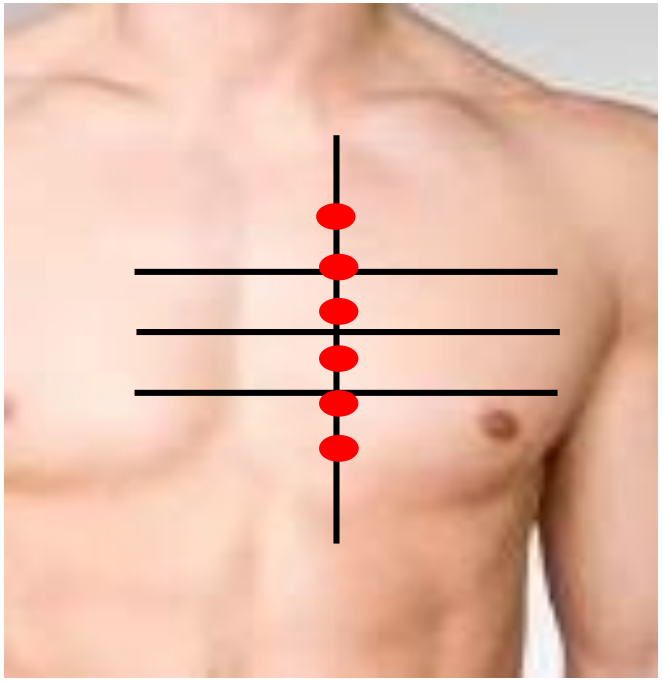


Miles et al Circulation 2019

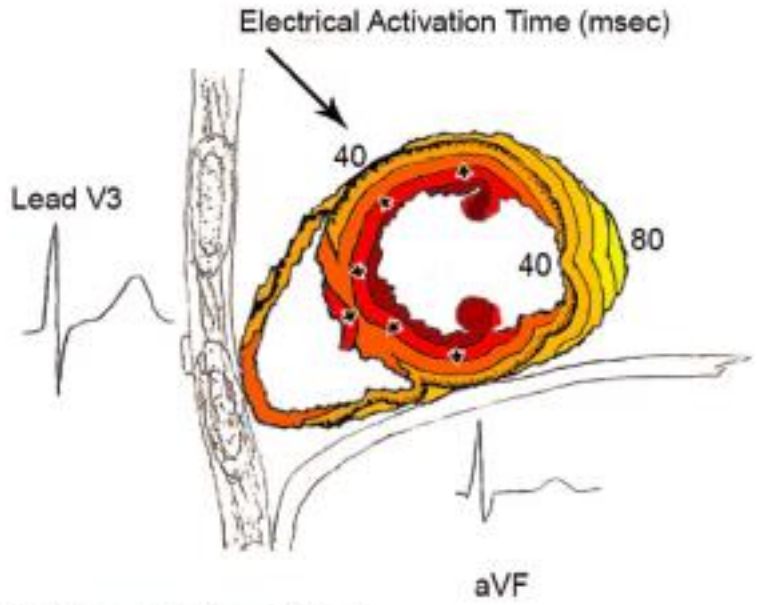

I
II
III
aVR
aVL
VF
V1
V2
V3
V4
V5
V6
11:49:46 16 Apr 2017 10mm/mV 25mm/s ≈ H M1 Sequenziale


V1
V2
V3
V4
V5
V6
25mm/s 10mm/mV 0,05-25Hz/50Hz Cardioline SpA HDpl 07281415 touc

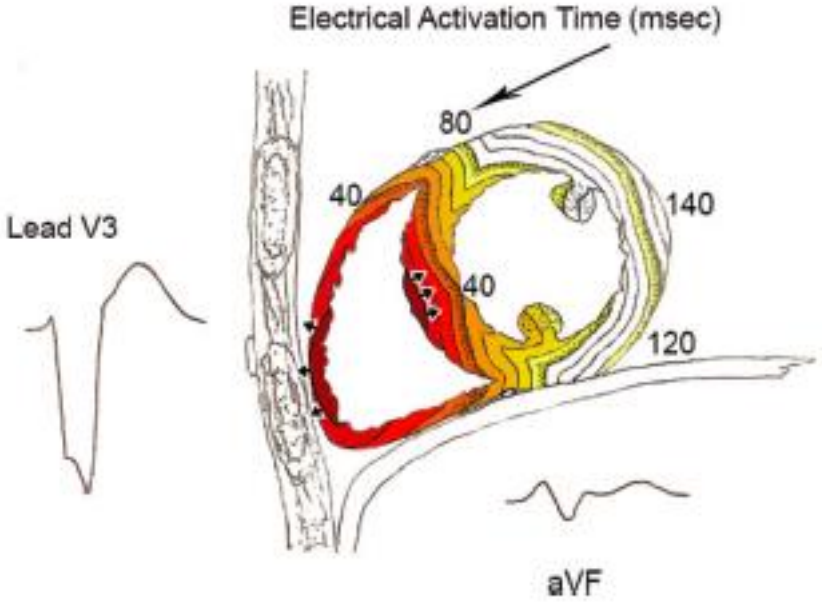

V1
V2
V3
V4
V5
V6
ino 25mm/s 10mm/mV 0,05-150Hz/50Hz Cardioline SpA HDpl 07281415 touchECG,3.2

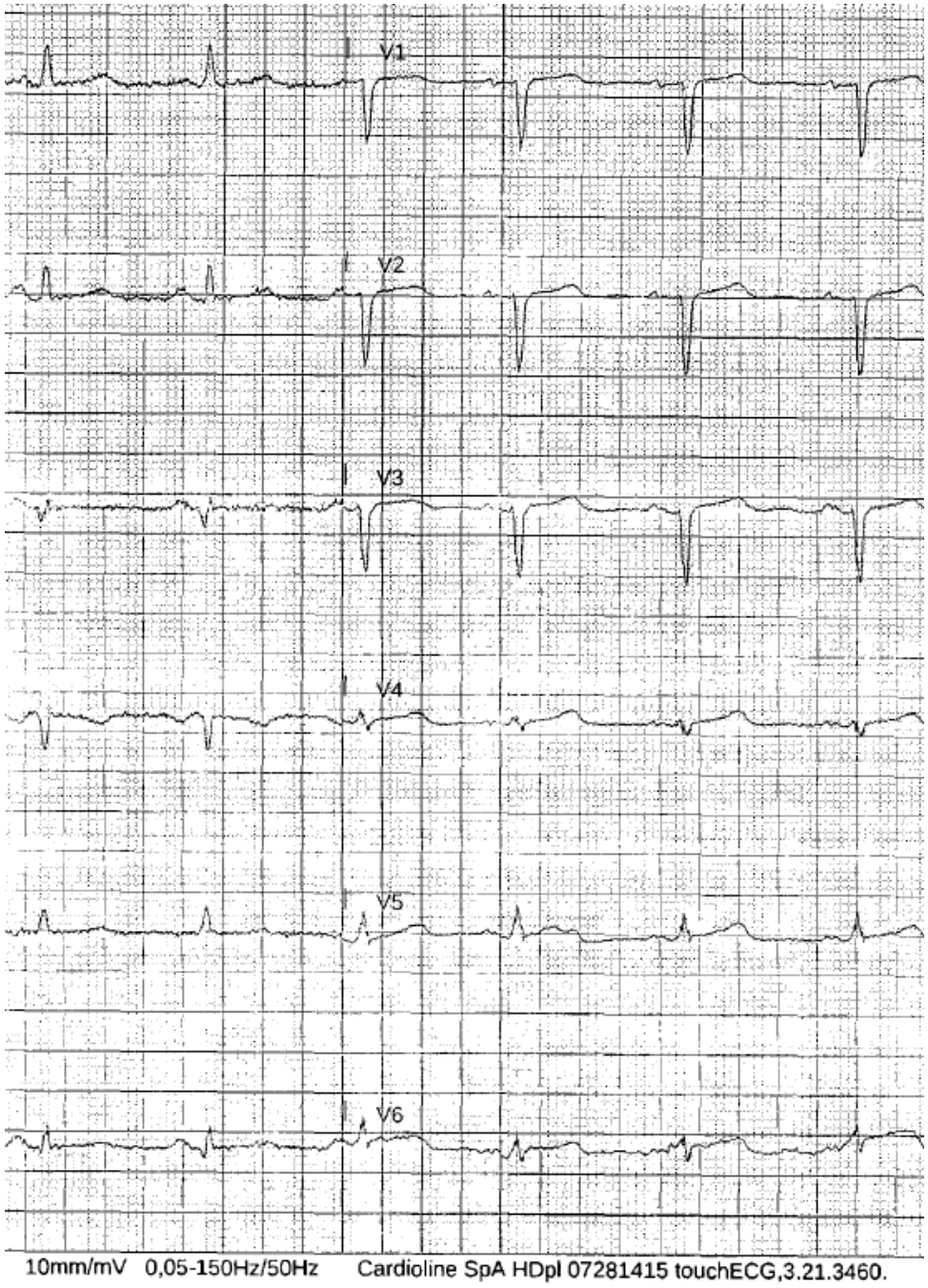


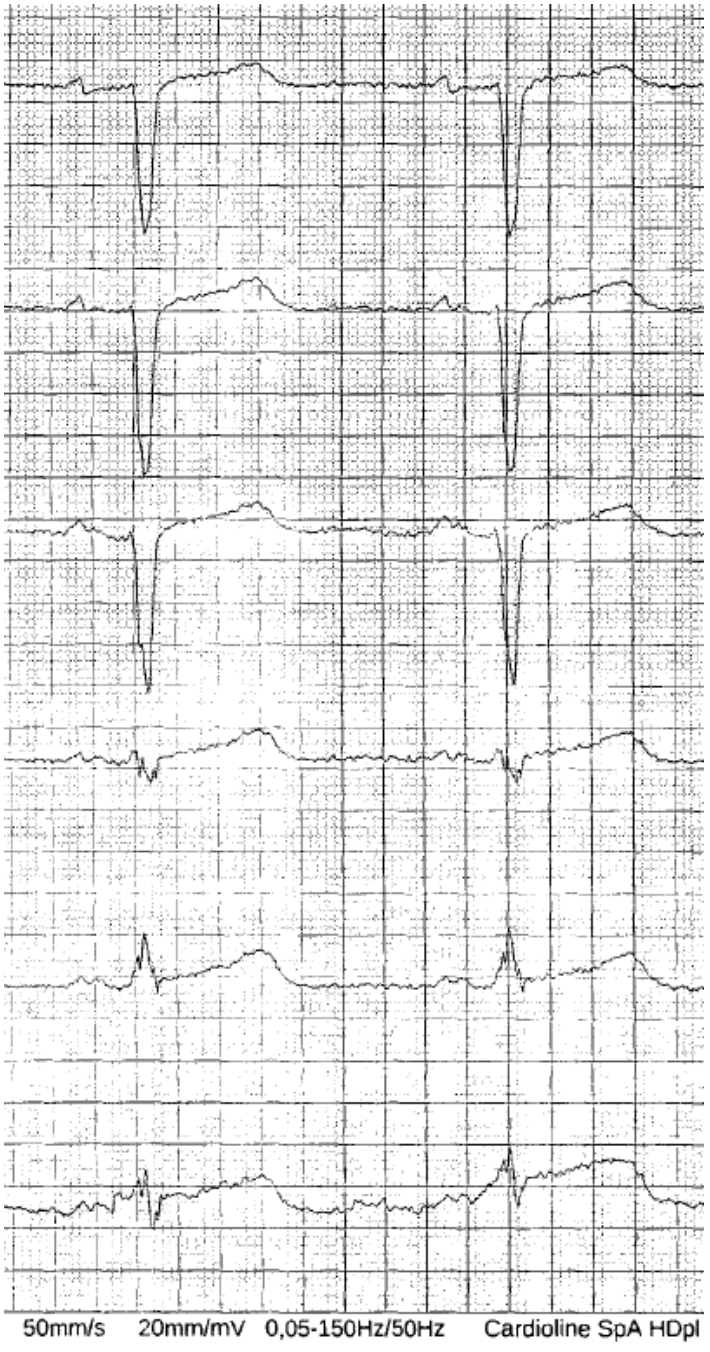
A **Normal Conduction**

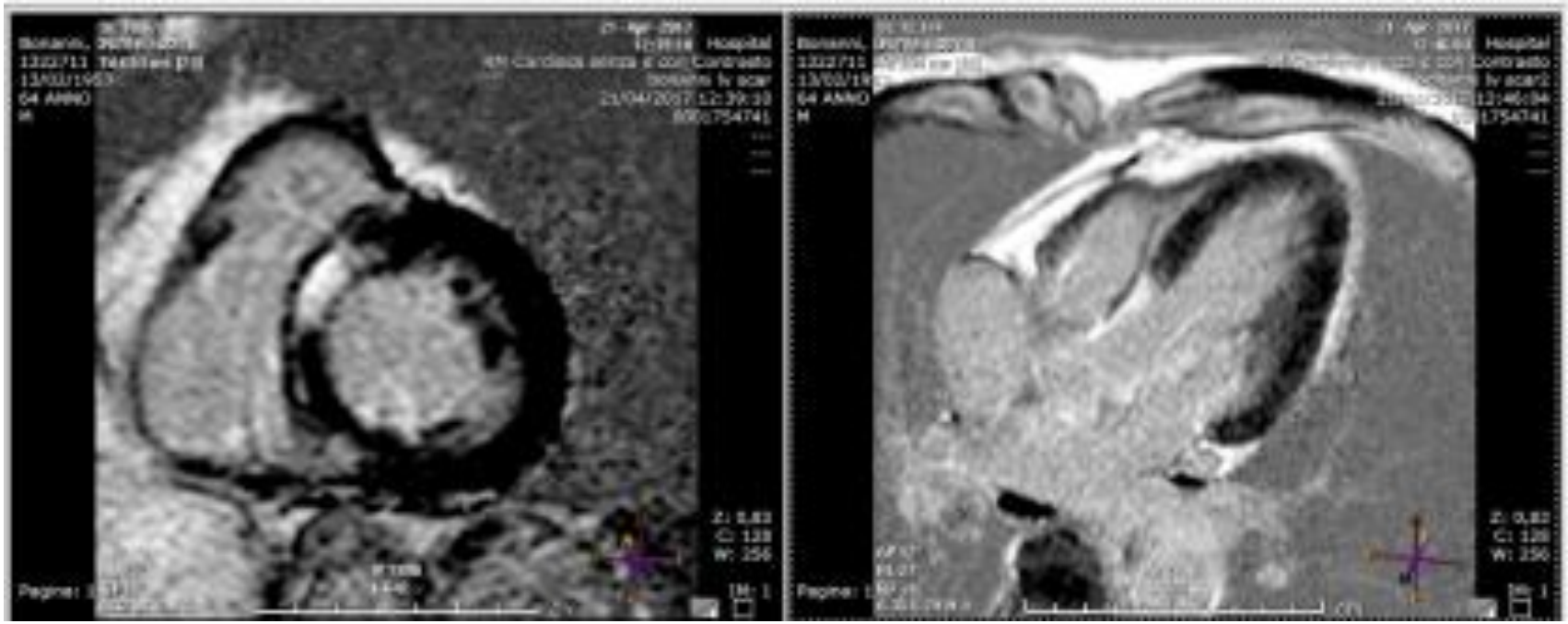


B **Left Bundle Branch Block**




V1
V2
V3
V4
V5
V6
10mm/mV 0,05-150Hz/50Hz Cardioline SpA HDpl 07281415 touchECG,3.21.3460.


50mm/s 20mm/mV 0,05-150Hz/50Hz Cardioline SpA HDpl



Gene: LMNA (Encoding the protein: Prelamin-A/C)
NP_733821.1:p.Arg189Trp/NC_000001.10:g.156104245C>T

Heterozygous carrier: Mutation occurs in only one copy of the gene.

Next Generation Sequencing stats: Depth of coverage: 221. Quality of the variant (0-255): 255.

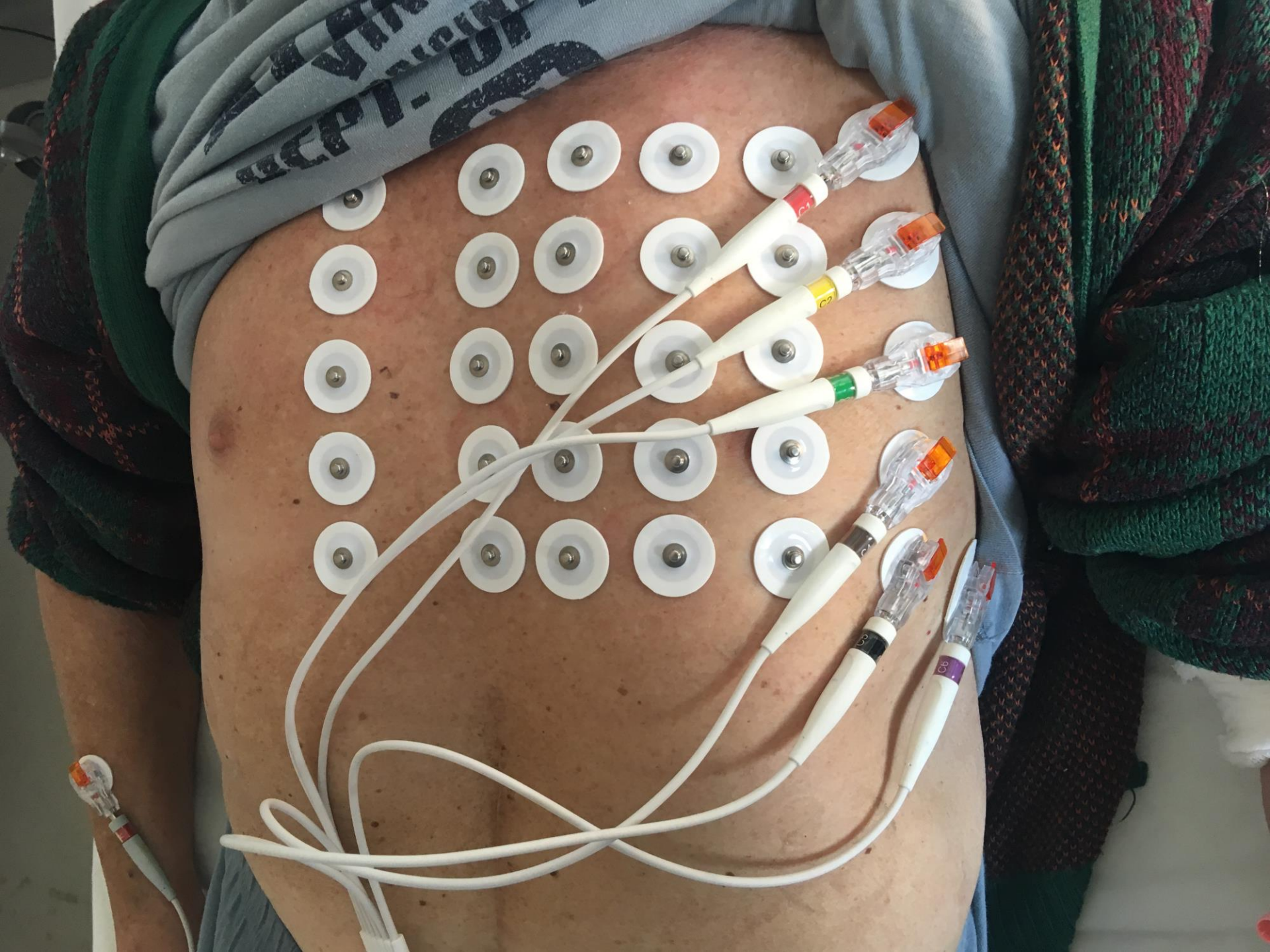
Mutation nomenclature: Nucleotide code: NM_170707.3:c.565C>T, NC_000001.10:g.156104245C>T. Amino acid code: NP_733821.1:p.Arg189Trp. dbSNP ID: rs267607626. Alternative names at the protein level: NP_733821.1:p.R189W. Located in: exon 3.

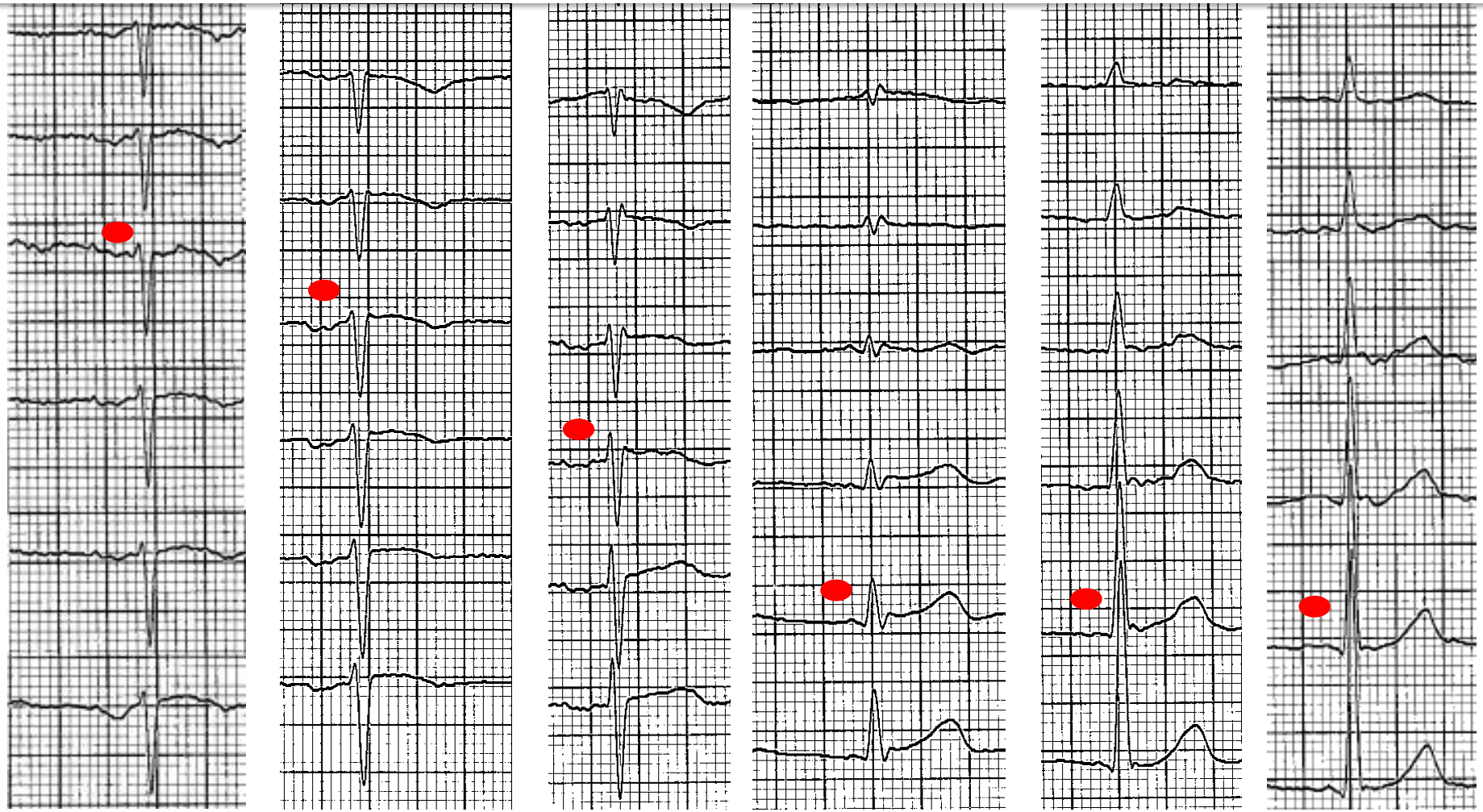
Pathogenicity: very likely to be pathogenic or disease-causing (++).

Population frequency: rare variant (found in <1% of controls).

Number of articles/communications that cite it: 10. Number of described families: 7. Number of families with additional

Major phenotypes	Carriers (families)	Non-carriers	Without genetic study	Total
No phenotypic study	5 (3)	0	4	9
Dilated cardiomyopathy	5 (5)	0	0	5
Not affected or healthy	3 (1)	2	4	9
Sudden death	2 (1)	0	1	3
Dilated cardiomyopathy + Left ventricular non-compaction/hypertrabeculation	1 (1)	0	0	1
Dilated cardiomyopathy + Muscular Dystrophy, Limb-Girdle + Peripheral Neuropathy	1 (1)	0	0	1
Left ventricular non-compaction/hypertrabeculation	0 (0)	0	1	1





1 2 3 4 5 6

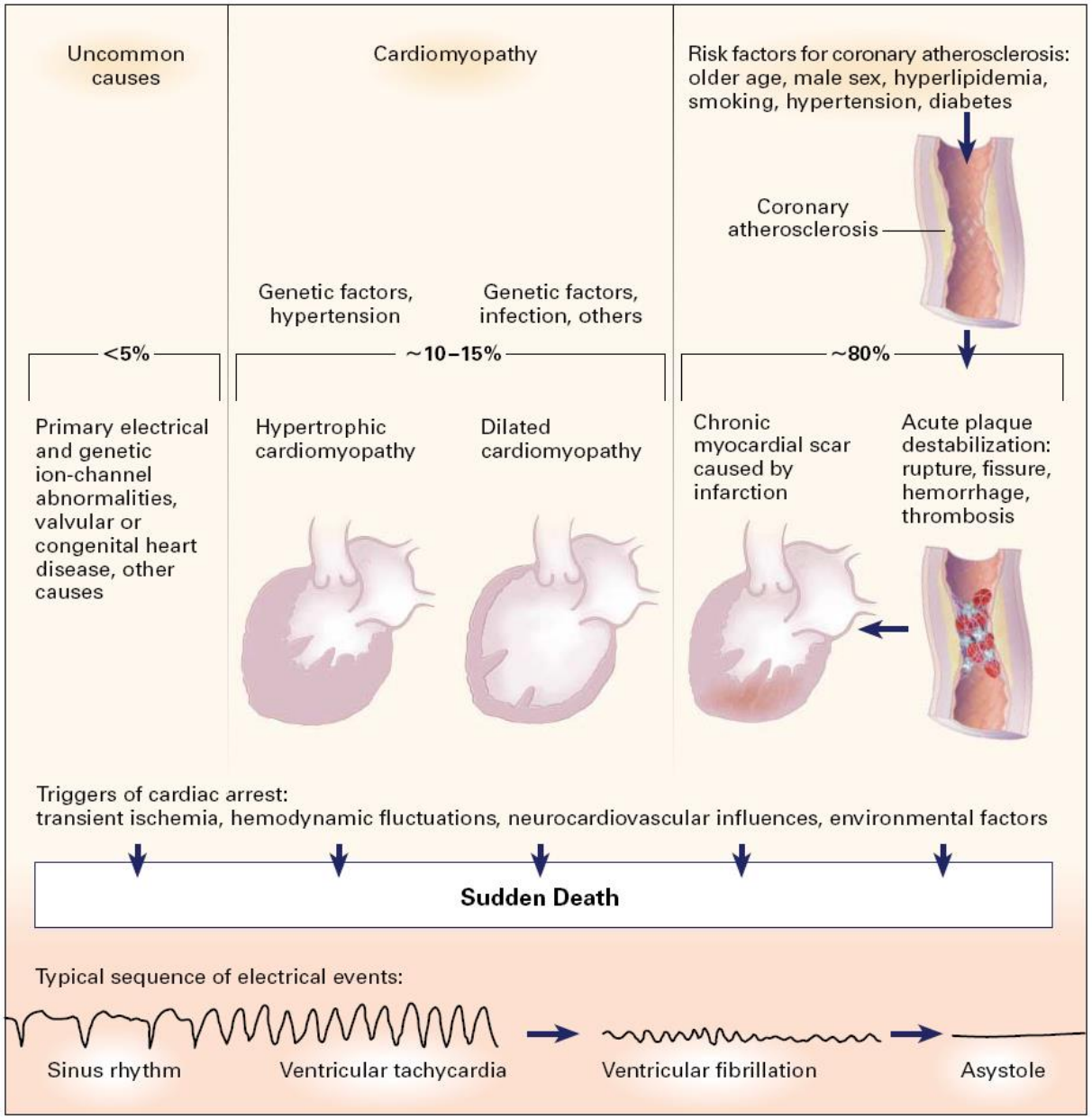
A community of trigger and driver mechanisms

CATALYSTS

neurovegetative system/ drugs
electrolyte imbalance /ischemia

TRIGGER

SUBSTRATE



Pathophysiology and Epidemiology of Sudden Death from Cardiac Causes.