Distrofia miotonica e cuore

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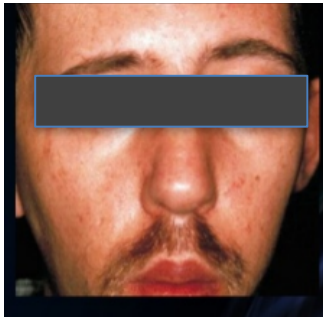
Myotonic Dystrophy



Autosomal dominant, multi-systemic disorder

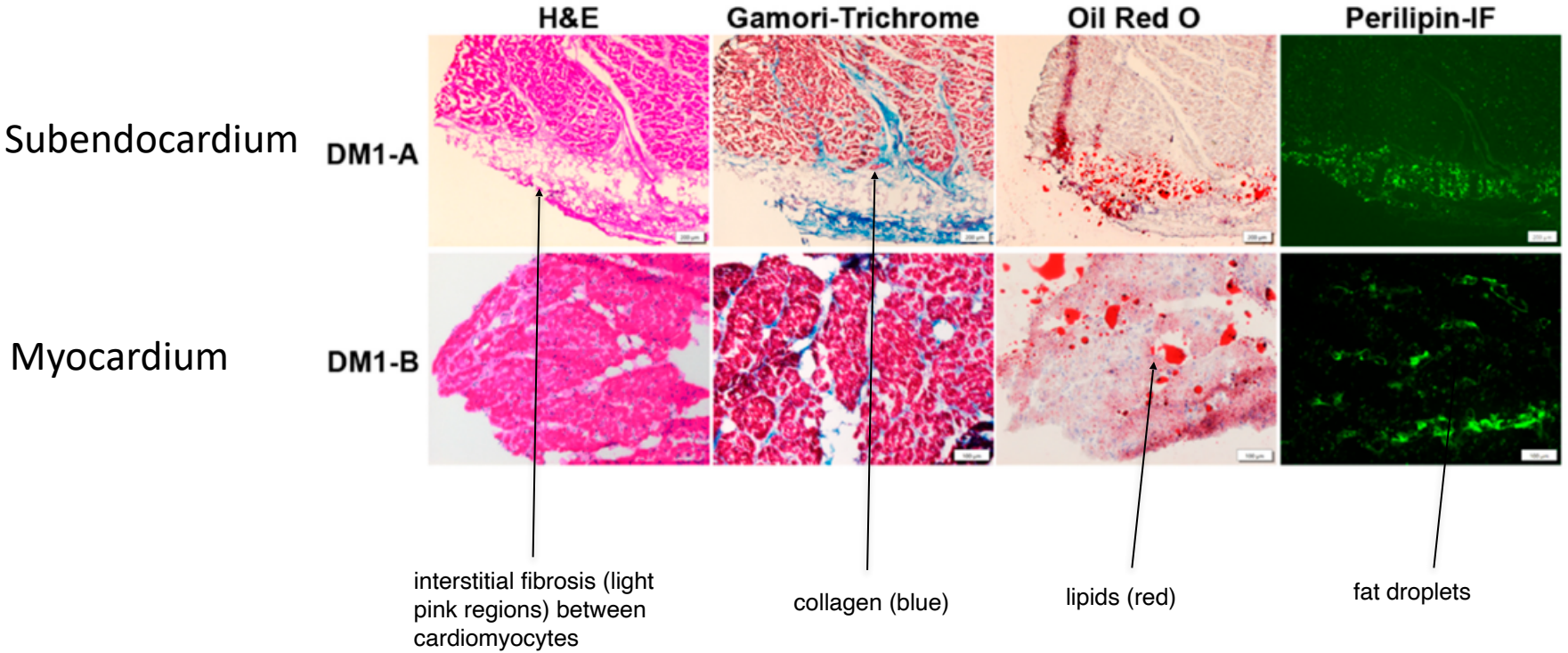
Expansion of a (CTG) trinucleotide repeat sequence within the DMPK gene

CTG repeat expansion leads to RNA toxicity and widespread splicing defects.

These splicing abnormalities affect multiple systems, including the:
respiratory, skeletal, cardiac, nervous, and endocrine systems

Miotonia (incapacità di rilassamento muscolare dopo contrazione)

Atrial and **ventricular** fatty infiltration and diffuse fibrotic changes in the myocardium and in the **conduction system** (SA node, AV node, bundles of His)



Form of DM1	CTG Repeat Range	Onset *	Key Cardiac Features	Skeletal Muscle and Other Systemic Features	Severity	Primary Challenges	References
Mild (Late-Onset) Form	50–150	Middle to late adulthood	Mild conduction abnormalities; occasional first-degree AV block; low incidence of structural abnormalities.	Cataracts, mild myotonia, minimal muscle weakness.	Least severe	Often undiagnosed due to subtle symptoms; may miss early intervention for cardiac monitoring.	[24,27,41–45]
Classical (Adult-Onset) Form	250–500	Late teens to early adulthood	Progressive conduction defects, including PR prolongation and bundle branch block; QTc (corrected QT) prolongation; moderate arrhythmia risk.	Progressive muscle weakness, severe myotonia, cataracts, insulin resistance.	Moderate to severe	Requires ongoing cardiac monitoring due to arrhythmia risk; symptomatic management of muscle issues.	[24,43–49]
Congenital Form	>1000	Birth or early infancy	Severe conduction delays; AV block, high risk of ventricular arrhythmias; QTc prolongation; structural abnormalities including fibrosis.	Severe hypotonia, respiratory distress, developmental delay, cognitive impairments, dysphagia.	Most severe	Immediate cardiac and respiratory support; early intervention needed for developmental support.	[24,33,45,49,50]
Juvenile Form	500–1000	Childhood to early adolescence	PR and QRS prolongation; moderate QTc prolongation; risk of atrial and ventricular arrhythmias.	Cognitive deficits, myotonia, gastrointestinal issues, mild developmental delay.	Severe	Progressive arrhythmia risk; requires multidisciplinary care to address systemic complications.	[24,45,49]

* Onset is defined as approximate age at which general DM1 symptoms first appear, regardless of the system affected. Abbreviations: Myotonic dystrophy type 1 (DM1), Atrioventricular block (AV block).

Journal of
Cardiovascular
Development and Disease

MDPI

Review

Comprehensive Cardiovascular Management of Myotonic Dystrophy Type 1 Patients: A Report from the Italian Neuro-Cardiology Network

Vincenzo Russo [1,*], Giovanni Antonini [2], Roberto Massa [3], Carlo Casali [4], Alfredo Mauriello [1,5], Anna Maria Martino [6], Roberto Marconi [7], Matteo Garibaldi [2], Pasquale Franciosa [8], Massimo Zecchin [9], Carlo Gaudio [8], Antonello D'Andrea [5] and Stefano Strano [8]

Cardiovascular Involvement in MD1

LV Systolic dysfunction

RV Systolic dysfunction

Diastolic dysfunction

Hypertrophy; MV prolapse, LV non-compaction

Myocardial fibrosis

Conduction System Disease

Atrial fibrillation

VT

SCD (VF; complete AV block, asystole)

Echocardiographic Features of Cardiac Involvement in Myotonic Dystrophy 1: Prevalence and Prognostic Value

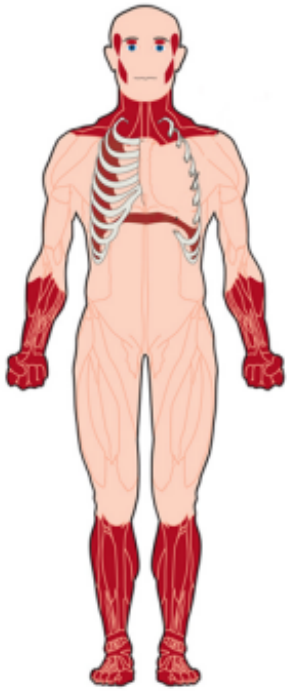
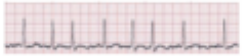
Vincenzo Russo [1,*], Antonio Capolongo [1], Roberta Bottino [1], Andreina Carbone [1], Alberto Palladino [2], Biagio Liccardo [1], Gerardo Nigro [1], Michał Marchel [3], Paolo Golino [1] and Antonello D'Andrea [4]

Table 1. Main clinical studies on echocardiographic features in DM1 patients.

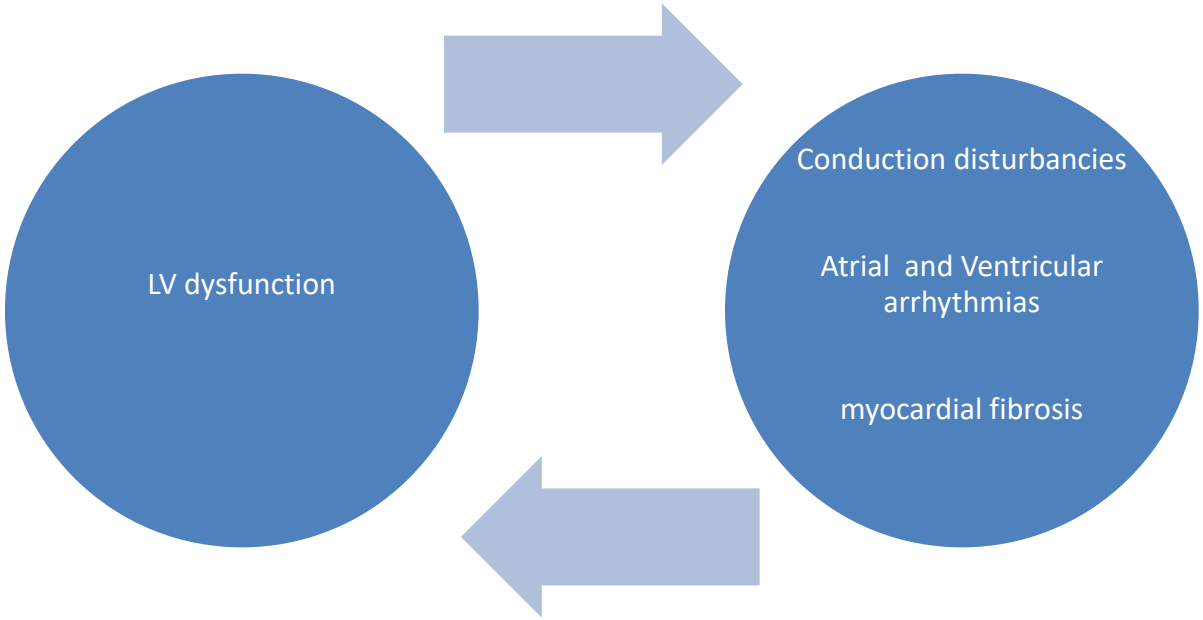
Study	Year	Study Design	DM1 Study Population (n)	DM1 Patients Age (Years)	Healthy Controls (n)	Echocardiographic Feature	Main Findings
Left Ventricular Systolic Dysfunction							
Russo [20]	2020	Systematic review	876	42.68	-	LVEF < 55%	The prevalence of LVEF < 55% was 13.8%.
Garcia [29]	2017	Prospective study	33	38.2 ± 12.9	33	LV GLS	DM1 patients exhibited significantly altered LV GLS, particularly at the apex (−20.0 ± 3.3 vs. −22.7 ± 3.1; $p < 0.001$), as compared with controls.
Garcia [13]	2017	Prospective study	46	40 [29–49]	-	LV GLS	LV GLS (cut-off value of −17.2%) predict cardiovascular events, regardless LVEF.
Guedes [36]	2017	Observational study	25	36.9 ± 16.0	25	LA and LV GLS	LA longitudinal strain is significantly decreased in patients with DM1 compared to controls
Petri [28]	2014	Cross-sectional study	129	44 (15)	-	GLS	The prevalence of abnormal GLS was 21.7% Abnormal GLS was above −15.9%; 60% had preserved LVEF> 50%.
Sousa [27]	2013	Case–control study	25	36.7 ± 12.5	13	GLS	DM1 patients showed a lower GLS than controls (−16.6 ± 3.6% vs. −18.7 ± 1.8%, $p = 0.022$). GLS correlates with PR interval duration.
Right Ventricular Function							
Lindqvist [46]	2010	Case–control study	36	45 ± 10	16	Right ventricular function by Doppler and RV strain	DM1 patients showed a prolonged IVCT and IRVT (both $p < 0.05$); shorter ET ($p < 0.05$); a higher right ventricular–right atrial pressure drop (23 ± 7 vs. 18 ± 2 mm Hg, $p < 0.05$); a reduction in RV free wall Sm ($p < 0.001$) and Am velocities ($p < 0.05$); a reduction in RV free wall systolic strain (−21.1 ± 8.6 vs. −31.2 ± 11%, $p < 0.001$).
Ozyigit [34]	2010	Case–control study	21	32.3 ± 12.3	21	Right ventricular function by Doppler	DM1 patients showed a reduction in peak velocity (cm/s) of Sm (12.38 ± 2.91 versus 14.40 ± 2.25 $p = 0.016$), Em (11.91 ± 3.54 versus 14.39 ± 3.87 $p = 0.037$); Tei index was significantly higher in DM1 patients compared with controls (0.27 ± 0.17, $p = 0.013$).

							with controls (0.27 ± 0.17, $p = 0.013$).
Left Ventricular Diastolic Dysfunction							
Fayssoil [16]	2014	Case–control study	26	45.1 (10.9)	13	Diastolic function	Increased left atrium diameter and increased mitral deceleration time compared with healthy controls; no differences were found regarding mean peak E/A mitral ratio, mean peak lateral early diastolic velocity and mean peak septal early diastolic velocity.
Wahbi [26]	2011	Case–control study	39	37.5 ± 12.1	39	LV GLS	Speckle tracking GLS was able to identify LV contractility abnormalities in DM1 patients with normal LVEF. DM1 patients showed a lower apical 4 chambers GLS compared to controls (−17.8 ± 2.5 vs. −19.2 ± 2.3 $p = 0.01$), which significantly correlated with PR interval.
Left atrial function							
Bhakta [48]	2003	Based on a prospective multicenter registry	382	42.2 ± 12.3 (17.9–77.8)	-	Structural cardiac abnormalities	Structural cardiac abnormalities determined with cardiac imaging included left ventricular hypertrophy (19.8%), left ventricular dilatation (18.6%), left ventricular systolic dysfunction (14.0%), mitral valve prolapse (13.7%), regional wall motion abnormality (11.2%) and left atrial dilatation (6.3%).
Fragola [15]	1997	Prospective study	42	37 ± 12	41	Left Ventricular Diastolic Function	The most common abnormalities were increased deceleration time (>224 ms), prolonged isovolumic relaxation time (>103 ms) and reduced rate of decline of flow velocity in early diastole (<2.1 m/s^2).

DM1: myotonic dystrophy 1; LVEF: left ventricular ejection fraction; LV GLS: left ventricular global longitudinal strain; LA: left atrial; RV: right ventricular; RVEF: right ventricular ejection fraction; (FAC: fractional area change).

DM1 PATIENTS	ECHOCARDIOGRAPHIC FINDINGS	PREVALENCE	PROGNOSTIC IMPACT
	**LEFT VENTRICULAR SYSTOLIC DYSFUNCTION**	• LVEF <55%. • ↓GLS (apical segment)	13.8% 28%	
	DIASTOLIC DYSFUNCTION	• ↑E peak velocity. • ↑Isovolumic relaxation time • ↓Deceleration time	29%	?
	RIGHT VENTRICULAR DYSFUNCTION	• ↓RV TDI velocities • ↑RV Tei index • ↓RV free wall strain	- - -	?
	LEFT ATRIAL FUNCTION	• Impaired longitudinal strain. • ↑inter-AEMD and intraleft-AEMD	- -	
	STRUCTURAL ABNORMALITIES	• Left ventricular hypertrophy • Left ventricular dilatation • Left ventricular non-compaction. • Mitral valve prolapse	19-29% 18.6% 35% 13-37%	?

Electro-mechanical correlation



McNally EM et al. J Am Heart Assoc. 2020;9:e014006

Petri et al. Journal of Cardiovascular Magnetic Resonance 2014, 16:59
http://jcmr-online.com/content/16/1/59

Journal of Cardiovascular Magnetic Resonance

RESEARCH Open Access

Myocardial fibrosis in patients with myotonic dystrophy type 1: a cardiovascular magnetic resonance study

Helle Petri[1*], Kiril Aleksov Ahtarovski[1], Niels Vejlstrup[1], John Vissing[2], Nanna Witting[2], Lars Køber[1] and Henning Bundgaard[3]

Abstract

Background: Myotonic dystrophy type 1 (DM1) is associated with increased cardiac morbidity and mortality. Therefore, assessment of cardiac involvement and risk stratification for sudden cardiac death is crucial. Nevertheless, optimal screening-procedures are not clearly defined. ECG, echocardiography and Holter-monitoring are useful but insufficient. Cardiovascular magnetic resonance (CMR) can provide additional information of which myocardial fibrosis may be relevant.
The purpose of this study was to describe the prevalence of myocardial fibrosis in patients with DM1 assessed by CMR, and the association between myocardial fibrosis and abnormal findings on ECG, Holter-monitoring and echocardiography.

Methods: We selected 30 unrelated patients with DM1: 18 patients (10 men, mean age 51 years) with, and 12 patients (7 men, mean age 41 years) without abnormal findings on ECG and Holter-monitoring.
Patients were evaluated with medical history, physical examination, ECG, Holter-monitoring, echocardiography and CMR.

Results: Myocardial fibrosis was found in 12/30 (40%, 9 men). The presence of myocardial fibrosis was associated with the following CMR-parameters: increased left ventricular mass (median (range) 55 g/m^2 (43–83) vs. 46 g/m^2 (36–64), p = 0.02), increased left atrial volume (median (range) 52 ml/m^2 (36–87) vs. 46 ml/m^2 (35–69), p = 0.04) and a trend toward lower LVEF (median (range) 63% (38–71) vs. 66% (60–80), p = 0.06). Overall, we found no association between the presence of myocardial fibrosis and abnormal findings on: ECG (p = 0.71), Holter-monitoring (p = 0.27) or echocardiographic measurements of left ventricular volumes, ejection fraction or global longitudinal strain (p = 0.18).

Conclusion: Patients with DM1 had a high prevalence of myocardial fibrosis which was not predicted by ECG, Holter-monitoring or echocardiography. CMR add additional information to current standard cardiac assessment and may prove to be a clinically valuable tool for risk stratification in DM1.

Keywords: Myotonic dystrophy, Cardiovascular magnetic resonance, Myocardial fibrosis, Risk stratification, Sudden cardiac death

Cardona et al. Journal of Cardiovascular Magnetic Resonance (2019) 21:26
https://doi.org/10.1186/s12968-019-0535-6

Journal of Cardiovascular Magnetic Resonance

RESEARCH Open Access

Myocardial fibrosis by late gadolinium enhancement cardiovascular magnetic resonance in myotonic muscular dystrophy type 1: highly prevalent but not associated with surface conduction abnormality

Andrea Cardona[1,2], William D. Arnold[3], John T. Kissel[3], Subha V. Raman[1] and Karolina M. Zareba[1*]

Abstract

Background: Conduction disease and arrhythmias represent a major cause of mortality in myotonic muscular dystrophy type 1 (MMD1). Permanent pacemaker (PPM) implantation is the cornerstone of therapy to reduce cardiovascular mortality in MMD1. Cardiovascular magnetic resonance (CMR) studies demonstrate a high prevalence of myocardial fibrosis in MMD1, however the association between CMR myocardial fibrosis with late gadolinium enhancement (CMR-LGE) and surface conduction abnormality is not well established in MMD1.
We investigated whether myocardial fibrosis by CMR-LGE is associated with surface conduction abnormalities meeting criteria for PPM implantation according to current guidelines in a cohort of patients with genetically confirmed MMD1.

Methods: Patients with genetically confirmed MMD1 were retrospectively evaluated. 12-lead electrocardiography (ECG) performed within 6 months of CMR was necessary for inclusion. The severity and extent of MMD1 was quantified using a validated Muscular Impairment Rating Scale (MIRS). Based on current guidelines for device-based therapy of cardiac rhythm abnormalities, we defined surface conduction abnormality as the presence of ECG alterations meeting criteria for PPM implant (class I or II indications): PR interval > 200 ms (type I atrioventricular (AV) block) and/or mono or bifascicular block (QRS > 120 ms), or evidence of advanced AV block. Balanced steady-state free precession sequences (bSSFP) were used for assessment of left ventricular (LV) volumes and ejection fraction. MOdified Look-Locker Inversion Recovery (MOLLI) acquisition schemes were used to acquire T1 maps. Patients' charts were reviewed up to 12 months post-CMR for occurence of PPM implantation.

Results: Fifty-two patients (38% male, 41 ± 14 years) were included. Overall, 31 (60%) patients had a surface conduction abnormality and 22 (42%) demonstrated midwall myocardial fibrosis by CMR-LGE. After a median of 57 days from CMR exam, 15 patients (29%) underwent PPM implantation. Subjects with vs. without surface conduction abnormality had significantly longer disease length (15.5 vs. 7.8 years, $p = 0.015$) and higher disease severity on the MIRS scale ($p = 0.041$). High prevalence of myocardial fibrosis by CMR-LGE was detected in subjects with and without surface conduction abnormality with no significant difference between the two cohorts (42% vs. 43%, $p = 0.999$). By multivariate logistic regression analysis, disease length was the only independent variable associated with surface conduction abnormality (OR 1.071, 95%CI 1.003–1.144, $p = 0.040$); while CMR-LGE was not associated with conduction abnormality ($p = -0.009$, $p = 0.949$).

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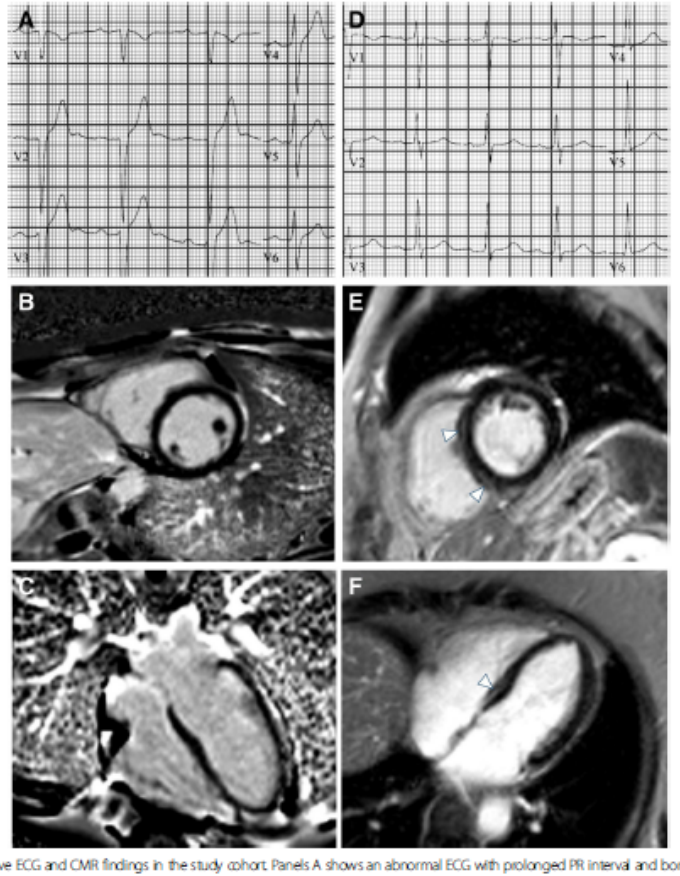

Fig. 1 Representative ECG and CMR findings in the study cohort. Panels A shows an abnormal ECG with prolonged PR interval and borderline QRS interval of a subject with no evidence of myocardial fibrosis by CMR-LGE (B, C). Panels D shows a normal ECG of a subjects with evidence of midwall fibrosis mostly evident in the interventricular septum (arrows in E, and F)

Contents lists available at ScienceDirect

Neuromuscular Disorders

journal homepage: www.elsevier.com/locate/nmd

Review

Prevalence and prognostic impact of cardiac resonance abnormalities in myotonic dystrophy patients

Vincenzo Russo [a,*], Alfredo Mauriello [b], Roberta Bottino [a], Antonio Giordano [d], Michal Marchel [e], Antonello D'Andrea [c], Théo Pezel [f,g,h], Gerardo Nigro [a,i], Santo Dellegrottaglie [i,t]

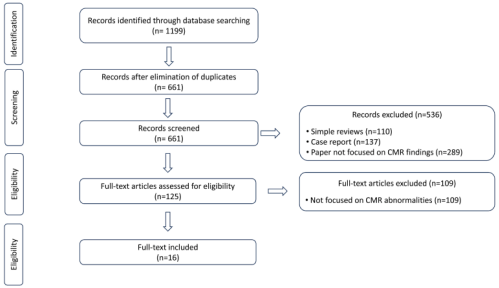

Fig. 1. Flow diagram of the study selection process for the systematic review.

CRM detected LGE and LVEF among study population.

	Patients (n)	Age (yrs)	AF (.%)	PR interval (ms)	First degree AVB (.%)	Second degree AVB (.%)	LVEF (.%)	LGE+ (.%)
Cardona et al. [22]	52	41± 14	–	199± 47	38	2	60± 6	42
Choudhary et al. [18]	40	45± 14	5	187± 29	35	–	58± 8	13
Chmielewski et al. [17]	57	43± 13	14	–	7	1,7	59± 6	18
Petri et al. [11]	30	47± 14	13	215 (130- 260)	33	3,3	63 (38–71)	40
Hermans et al. ((12)	80	45± 15	3	200 (136- 460)	38	–	53± 15	13
Kashyap et al. [21]	46	43.0 ± 9.5	–	–	39,1		56± 6	2
Leali et al. [8]	34	45 ± 12	12	–	35	3	58± 7	26

AF: atrial fibrillation; AVB: atrioventricular block; LGE: late gadolinium enhancement; LVEF: left ventricular ejection fraction.

Table 3
Main features of the studies with outcome and follow-up.

First Author	Year Of Publication	Study Design	Study Population	Main Outcomes	Follow-Up
Hermans et al. [12]	2012	Cross sectional	80 DM1	Structural and functional abnormalities on CMR in DM1 patients	None
Petri et al. [11]	2014	Cross sectional	30 DM1	Myocardial fibrosis on CMR and association with electrocardiogram abnormalities	None
Nazarian et al. [13]	2010	Cross sectional	43 DM (35 DM1; 8 DM2)	Relationship between CMR signal-to-noise ratio variance and QRS duration on electrocardiogram	None
Alì et al. [16]	2020	Case-control	9 DM1 9 Controls	CMR myocardial strain and extracellular volume as subclinical biomarkers of cardiac involvement in DM1 patients	None
Chmielewski et al. [17]	2018	Case-control	57 DM1 15 Controls	Relationship between CMR abnormalities and electrocardiogram abnormalities in DM1 patients	None
Choudhary et al. [18]	2016	Case-control	41 DM1 41 Controls	Relationship between CMR abnormalities and clinical, electrical and genetic disturbances in DM1 patients	None
Luetkens et al. [19]	2008	Case-control	35 DM (13 DM1; 22 DM2) 31 Controls	Subclinical CMR abnormalities in asymptomatic DM patients	None
Schmacht et al. [14]	2023	Case-control	29 DM2 patients 17 Controls	Subclinical CMR abnormalities in DM2 patients with preserved ejection fraction	None
Turkbey et al. [15]	2003	Case-control	35 DM (24 DM1; 9 DM2) 13 Controls	CMR functional and post-contrast T1 time characteristics in DM patients.	1.9 (IQR 1.1 – 3.1) years
Cardona et al. [22]	2019	Retrospective observational	52 DM1	Association between CMR-LGE and electrocardiogram abnormalities in need of pacemaker in DM1 patients. CMR-LGE is an independent factor of needed of implantation of pacemaker.	1-year follow-up for major adverse cardiovascular events and death
Almogheer et al. [23]	2021	Retrospective observational	27 DM	CMR-LGE pattern, progression rate in neuromuscular diseases; Association between CMR-LGE, cardiac function and major adverse cardiovascular events in neuromuscular diseases.	4.1 years
Tanawuttiwat et al. [24]	2016	Retrospective observational	155 DM (136 DM1; 28 DM2)	Description of the prevalence and the longitudinal incidence of conduction abnormalities on electrocardiogram	5.5 years
Blaszczyk et al. [20]	2021	Prospective observational	27 DM2	Description of cardiac remodeling progression; myocardial tissue differentiation; CMR-global longitudinal strain; Association between CMR findings with arrhythmias and conduction system abnormalities	3.9 ± 0.3 years
Kashyap et al. [21]	2023	Prospective observational	46 DM1	Myocardial tissue differentiation; CMR-global longitudinal strain; Association between CMR findings with MACEs	1.7–6.3 years
Abati et al. [25]	2024	Retrospective observational	18 DM1 4 DM2	Association between CMR findings with demographic and clinical parameters, ECG-derived measures of cardiac conduction, and neuromuscular performance status	None
Leali et al. [8]	2023	Retrospective observational	34 DM1 68 Controls	Association between CMR findings with cardiac rhythm disorders and worsening outcome.	1.5–4.0 years

CMR: cardiac magnetic resonance; DM: myotonic muscular dystrophy; LGE: late gadolinium enhancement; MACEs: major cardiovascular events.

LGE (6 studies)

mid-myocardial and/or epicardial LGE (13 to 42%)

in the septum and within the basal inferolateral segment of the LV wall

ECV (5 studies)

All studies showed a significant increase in ECV values compared with reference limits

Association between CMR results and ECG abnormalities (5 studies)

-LGE does not correlate with conduction system defects

-LGE predictor of AF

-LGE extension significantly correlated with Lown class 4 ventricular escape beats

Neuromuscular Disorders 52 (2025) 105396

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MDPI

Review

Comprehensive Cardiovascular Management of Myotonic Dystrophy Type 1 Patients: A Report from the Italian Neuro-Cardiology Network

Vincenzo Russo [1,*], Giovanni Antonini [2], Roberto Massa [3], Carlo Casali [4], Alfredo Mauriello [1,5], Anna Maria Martino [6], Roberto Marconi [7], Matteo Garibaldi [2], Pasquale Franciosa [8], Massimo Zecchin [9], Carlo Gaudio [8], Antonello D'Andrea [5] and Stefano Strano [8]

Cardiovascular Involvement in MD1

LV Systolic dysfunction

RV Systolic dysfunction

Diastolic dysfunction

Hypertrophy; MV prolapse, LV non-compaction

Myocardial fibrosis

Conduction System Disease

Atrial fibrillation

VT

SCD (VF; complete AV block, asystole)

Conduction system disease

First-degree AV block, (28.2%– 45.0%)

Bundle branch block, (16.5%–19.9%)

Tachy-arrhythmias

Supraventricular arrhythmias (5.0%–12.5%)

Nonsustained VT (2.2%– 4.1%)

Sustained VT (1.0%–2.7%)

Heart Rhythm, Vol 19, No 10, October 2022

SCD

SCD: about 30 to 40% of cases

Ventricular tachycardia, complete AV block, asystole, electromechanical dissociation

Groh, W.J et al. N. Engl. J. Med. **2008**, 358, 2688–2697
Mathieu, J. Et al. Neurology **1999**, 52, 1658–1662
Wahbi, K et al. Eur. Heart J. **2017**, 38, 751–758

Genetics and SCD

Journal of Neurology
https://doi.org/10.1007/s00415-018-8773-3

ORIGINAL COMMUNICATION

A 34-year longitudinal study on long-term cardiac outcomes in DM1 patients with normal ECG at baseline at an Italian clinical centre

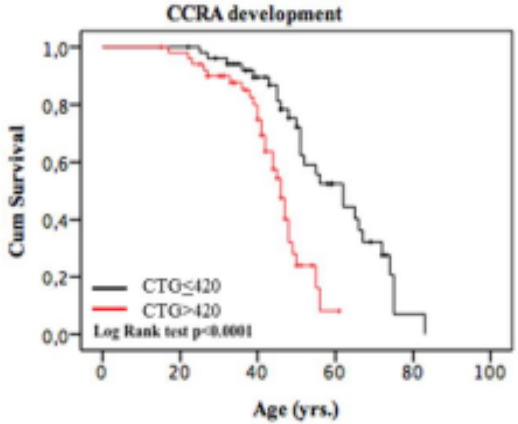
Elisabetta Bucci[1] · Marco Testa[2] · Loretta Licchelli[1] · Alessandra Frattari[2] · Nadia Attalla El Halabieh[2] · Erica Gabriele[2] · Giulia Pignatelli[2] · Tiziana De Santis[1] · Laura Fionda[1] · Fiammetta Vanoli[1] · Stefania Morino[1] · Matteo Garibaldi[1] · Antonella Di Pasquale[1] · Nicola Vanacore[3] · Annalisa Botta[4] · Giovanni Antonini[1]

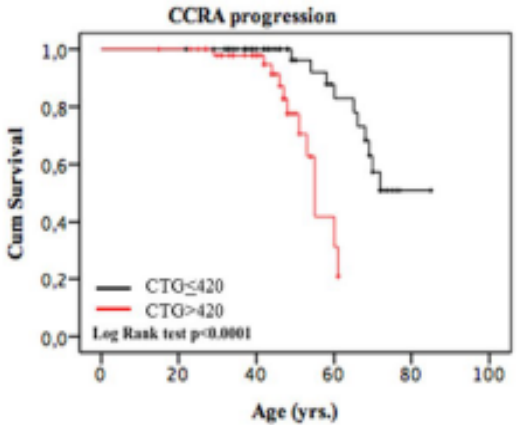
Received: 8 October 2017 / Revised: 22 January 2018 / Accepted: 29 January 2018

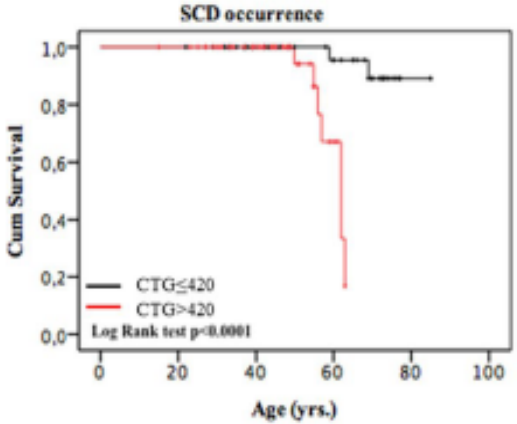
Abstract

Cardiac conduction and/or rhythm abnormalities (CCRA) are the most frequent and life-threatening complications in DM1. In order to determine prevalence, incidence, characteristics, age of onset and predictors of CCRA, CCRA progression and sudden cardiac death (SCD) in DM1, we collected ECG/24hECG-Holter data from a yearly updated 34-year database of a cohort of 103 DM1 patients without cardiac abnormalities at baseline, followed for at least 1 year. Fifty-five patients developed CCRA [39 developed conduction abnormalities (CCA) and 16 rhythm abnormalities (CRA)], which progressed in 22. Nine had SCD. Risk and incidence of CCRA amounted to 53.4 and 6.83% person-years (CCA: 37.9 and 4.8%; CRA 15.5 and 2%), respectively; risk and incidence of SCD amounted to 8.74 and 0.67% person-years, respectively. CTG expansion represented a predictor of CCRA incidence (HR 1.10, $p = 0.04$), CCRA progression (HR 1.28, $p = 0.001$) and SCD (HR 1.39, $p = 0.002$). MIRS progression during follow-up was associated with CCRA prevalence (OR 5.82, $p = 0.004$); older age and larger CTG expansion to SCD prevalence (OR 2.67, $p = 0.012$; OR 1.54, $p = 0.005$). Age of CCRA onset and CCRA progression was significantly lower in patients with larger CTG expansion and in those with MIRS progression. Age when SCD occurred was significantly lower in patients with larger CTG expansion. Amongst recorded cardiac abnormalities, both atrial flutter (OR 8.70; $p = 0.031$) and paroxysmal supraventricular tachycardia (OR 8.67; $p = 0.040$) were associated with SCD. Although all DM1patients may develop cardiac abnormalities at any time in their life, patients older than 30 years with larger CTG expansion and MIRS progression in particular should be carefully monitored via periodical ECG.

A CTG expansion







HR 1.1. p 0.04

HR 1.28. p 0.001

HR 1.39. p 0.002

ECG

The NEW ENGLAND JOURNAL of MEDICINE

ORIGINAL ARTICLE

Electrocardiographic Abnormalities and Sudden Death in Myotonic Dystrophy Type 1

William J. Groh, M.D., M.P.H., Miriam R. Groh, M.S., Chandan Saha, Ph.D., John C. Kincaid, M.D., Zachary Simmons, M.D., Emma Ciafaloni, M.D., Rahman Pourmand, M.D., Richard F. Otten, M.D., Deepak Bhakta, M.D., Girish V. Nair, M.D., M.S., Mohammad M. Marashdeh, M.D., Douglas P. Zipes, M.D., and Robert M. Pascuzzi, M.D.

N ENGL J MED 358;25 WWW.NEJM.ORG JUNE 19, 2008

Severe ECG abnormalities:

Rhythm other than sinus

PR> 240 ms or II-III° AVB

QRSd > 120 ms

Atrial tachyarrhythmias

BACKGROUND

Sudden death can occur as a consequence of cardiac-conduction abnormalities in the neuromuscular disease myotonic dystrophy type 1. The determinants of the risk of sudden death remain imprecise.

METHODS

We assessed whether the electrocardiogram (ECG) was useful in predicting sudden death in 406 adult patients with genetically confirmed myotonic dystrophy type 1. A patient was characterized as having a severe abnormality if the ECG had at least one of the following features: rhythm other than sinus, PR interval of 240 msec or more, QRS duration of 120 msec or more, or second-degree or third-degree atrioventricular block.

RESULTS

Patients with severe abnormalities according to the entry ECG were older than patients without severe abnormalities, had more severe skeletal-muscle impairment, and were more likely to have heart failure, left ventricular systolic dysfunction, or atrial tachyarrhythmia. Such patients were more likely to receive a pacemaker or an implantable cardioverter–defibrillator during the follow-up period. During a mean follow-up period of 5.7 years, 81 patients died; there were 27 sudden deaths, 32 deaths from progressive neuromuscular respiratory failure, 5 nonsudden deaths from cardiac causes, and 17 deaths from other causes. Among the 17 patients who died suddenly in whom postcollapse rhythm was evaluated, a ventricular tachyarrhythmia was observed in 9. A severe ECG abnormality (relative risk, 3.30; 95% confidence interval [CI], 1.24 to 8.78) and a clinical diagnosis of atrial tachyarrhythmia (relative risk, 5.18; 95% CI, 2.28 to 11.77) were independent risk factors for sudden death.

CONCLUSIONS

Patients with adult myotonic dystrophy type 1 are at high risk for arrhythmias and sudden death. A severe abnormality on the ECG and a diagnosis of an atrial tachyarrhythmia predict sudden death. (ClinicalTrials.gov number, NCT00622453.)

Table 3. The Relation between Patient Characteristics and the Risk of Death.*

Characteristic	Sudden Death		Death from Progressive Neuromuscular Respiratory Failure		Death from Any Cause	
	Relative Risk (95% CI)	P Value	Relative Risk (95% CI)	P Value	Relative Risk (95% CI)	P Value
Age†	1.16 (0.76–1.75)	0.50	1.81 (1.20–2.74)	0.005	1.46 (1.13–1.87)	0.003
Muscular-impairment score‡						
1 or 2			1.0		1.0	
3			1.28 (0.20–8.19)	0.80	1.13 (0.47–2.73)	0.79
4			2.85 (0.60–13.55)	0.19	1.47 (0.67–3.19)	0.33
5			13.07 (2.77–61.61)	0.001	4.54 (2.02–10.19)	<0.001
Heart failure			5.39 (1.46–19.82)	0.01	2.85 (1.20–6.74)	0.02
Atrial tachyarrhythmia	5.18 (2.28–11.77)	<0.001	2.41 (1.10–5.31)	0.03	2.59 (1.55–4.32)	<0.001
Pacemaker	1.35 (0.51–3.56)	0.54	1.80 (0.77–4.20)	0.17	1.46 (0.82–2.60)	0.19
Ventricular tachyarrhythmia					6.41 (2.30–17.89)	<0.001
Severe ECG abnormality§	3.30 (1.24–8.78)	0.02	1.33 (0.53–3.33)	0.55	1.58 (0.90–2.81)	0.11

Incidence and predictors of sudden death, major conduction defects and sustained ventricular tachyarrhythmias in 1388 patients with myotonic dystrophy type 1

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Aims To describe the incidence and identify predictors of sudden death (SD), major conduction defects and sustained ventricular tachyarrhythmias (VTA) in myotonic dystrophy type 1 (DM1).

Methods and results We retrospectively enrolled 1388 adults with DM1 referred to six French medical centres between January 2000 and October 2013. We confirmed their vital status, classified all deaths, and determined the incidence of major conduction defects requiring permanent pacing and sustained VTA. We searched for predictors of overall survival, SD, major conduction defects, and sustained VTA by Cox regression analysis. Over a median 10-year follow-up, 253 (18.2%) patients died, 39 (3.6%) suddenly. Analysis of the cardiac rhythm at the time of the 39 SD revealed sustained VTA in 9, asystole in 5, complete atrioventricular block in 1 and electromechanical dissociation in two patients. Non-cardiac causes were identified in the five patients with SD who underwent autopsies. Major conduction defects developed in 143 (19.3%) and sustained VTA in 26 (2.3%) patients. By Cox regression analysis, age, family history of SD and left bundle branch block were independent predictors of SD, while age, male sex, electrocardiographic conduction abnormalities, syncope, and atrial fibrillation were independent predictors of major conduction defects; non-sustained VTA was the only predictor of sustained VTA.

Conclusions SD was a frequent mode of death in DM1, with multiple mechanisms involved. Major conduction defects were by far more frequent than sustained VTA, whose only independent predictor was a personal history of non-sustained VTA. ClinicalTrials.gov no: NCT01136330.

Table 3 Multiple variable analysis of association between baseline characteristics and clinical outcomes

	Overall survival		Sudden death		Sustained ventricular tachyarrhythmia		Major conduction defect	
	Adjusted HR (95% CI)	P	Adjusted HR (95% CI)	P	Adjusted HR (95% CI)	P	Adjusted HR (95% CI)	P
Age, per year	1.07 (1.06–1.08)	<0.0001	1.05 (1.02–1.08)	0.0006	–	–	1.02 (1.01–1.04)	0.0005
Male sex	1.47 (1.12–1.93)	0.005	–	–	–	–	1.49 (1.05–2.11)	0.024
Family history of sudden death	0.99 (0.63–1.57)	0.98	2.82 (1.21–6.53)	0.016	–	–	1.07 (0.58–1.95)	0.84
History of								
Coronary artery disease	0.72 (0.40–1.30)	0.27	–	–	2.77 (0.64–11.9)	0.17	0.68 (0.30–1.52)	0.35
Atrial fibrillation	0.99 (0.69–1.42)	0.95	1.69 (0.76–3.77)	0.20	2.46 (0.89–6.77)	0.081	2.12 (1.41–3.17)	0.0003
Ventricular tachycardia								
Non-sustained	0.94 (0.45–1.96)	0.86	2.36 (0.69–8.08)	0.17	8.57 (2.59–28.3)	0.0004	1.86 (0.91–3.80)	0.090
Sustained	1.65 (0.51–5.33)	0.40	–	–	2.13 (0.36–12.7)	0.41	–	–
Major conduction defect	1.30 (0.65–2.62)	0.46	–	–	–	–	–	–
Syncope	1.60 (1.13–2.28)	0.008	–	–	–	–	2.39 (1.61–3.56)	<0.0001
Heart rate, per 10 bpm	1.17 (1.08–1.26)	<0.0001	1.16 (0.97–1.39)	0.099	–	–	1.10 (0.98–1.24)	0.095
1st degree atrioventricular block	1.73 (1.31–2.28)	0.0001	1.55 (0.79–3.04)	0.21	1.37 (0.54–3.48)	0.51	1.60 (1.12–2.30)	0.010
Bundle branch block				0.012	1.78 (0.57–5.55)	0.32	1.85 (1.18–2.91)	0.007
Left	1.40 (0.98–2.01)	0.063	2.57 (1.23–5.37)					0.010
Right	1.27 (0.84–1.93)	0.26	–	–	–	–	1.91 (1.17–3.11)	
Left ventricular ejection fraction <50%	1.06 (0.64–1.75)	0.82	0.93 (0.27–3.17)	0.90	1.92 (0.57–6.50)	0.30	1.59 (0.90–2.79)	0.11

CI = confidence interval; HR = hazard ratio.

ORIGINAL RESEARCH

Cardiac Conduction Disorders as Markers of Cardiac Events in Myotonic Dystrophy Type 1

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BACKGROUND: Myotonic dystrophy type 1 involves cardiac conduction disorders. Cardiac conduction disease can cause fatal arrhythmias or sudden death in patients with myotonic dystrophy type 1.

METHODS AND RESULTS: This study enrolled 506 patients with myotonic dystrophy type 1 (aged ≥15 years; >50 cytosine-thymine-guanine repeats) and was treated in 9 Japanese hospitals for neuromuscular diseases from January 2006 to August 2016. We investigated genetic and clinical backgrounds including health care, activities of daily living, dietary intake, cardiac involvement, and respiratory involvement during follow-up. The cause of death or the occurrence of composite cardiac events (ie, ventricular arrhythmias, advanced atrioventricular blocks, and device implantations) were evaluated as significant outcomes. During a median follow-up period of 87 months (Q1–Q3, 37–138 months), 71 patients expired. In the univariate analysis, pacemaker implantations (hazard ratio [HR], 4.35; 95% CI, 1.22–15.50) were associated with sudden death. In contrast, PQ interval ≥240 ms, QRS duration ≥120 ms, nutrition, or respiratory failure were not associated with the incidence of sudden death. The multivariable analysis revealed that a PQ interval ≥240 ms (HR, 2.79; 95% CI, 1.9–7.19, $P<0.05$) or QRS duration ≥120 ms (HR, 9.41; 95% CI, 2.62–33.77, $P < 0.01$) were independent factors associated with a higher occurrence of cardiac events than those observed with a PQ interval <240 ms or QRS duration <120 ms; these cardiac conduction parameters were not related to sudden death.

CONCLUSIONS: Cardiac conduction disorders are independent markers associated with cardiac events. Further investigation on the prediction of occurrence of sudden death is warranted.

J Am Heart Assoc. 2020;9:e015709

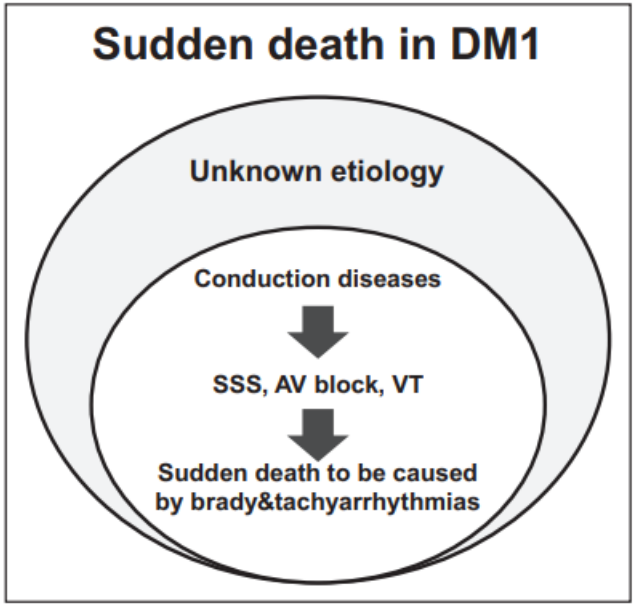

Figure 5. A schema associated with the etiology of sudden death in DM1.
AV, indicates atrioventricular; DM1, myotonic dystrophy type 1; SSS, sick sinus syndrome; and VT, ventricular fibrillation.

Electrophysiologic study

Insufficiency of electrocardiogram alone in predicting infrahisian abnormalities in patients with type 1 myotonic dystrophy

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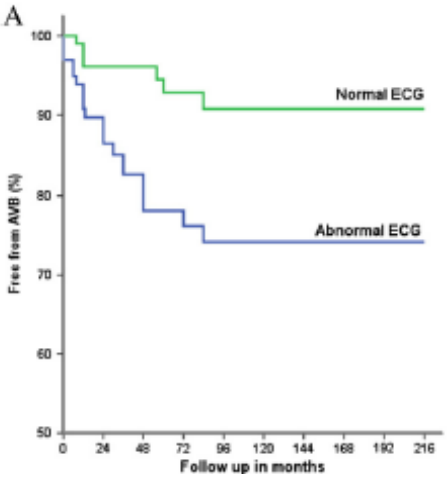
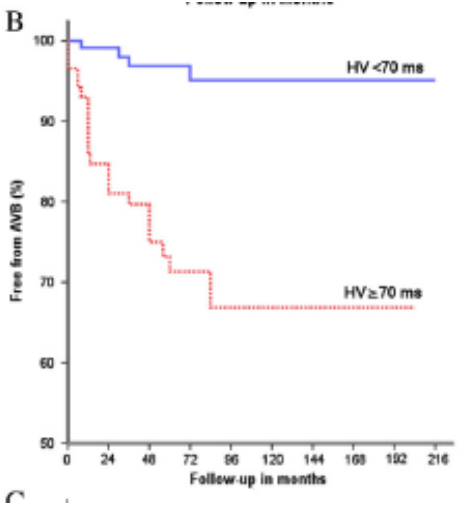
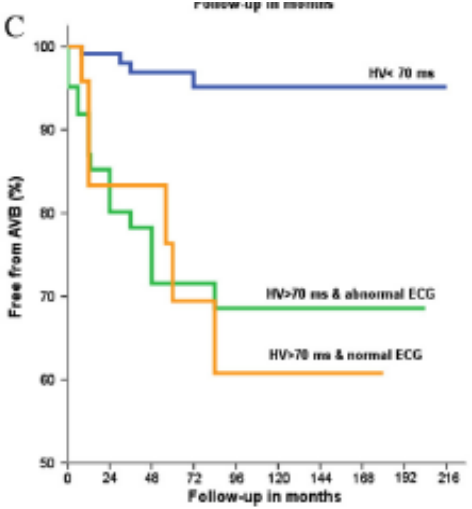
220 pts
mean age 41 ± 13 years
Median FU: 6 years







› Int J Cardiol. 2014 Apr 1;172(3):625-7

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Original Investigation

Electrocardiogram vs Electrophysiological Study and Major Conduction Delays in Myotonic Dystrophy Type 1

Nicolas Clementy, MD, PhD[1,2]; Fabien Labombarda, MD, PhD[3]; François Grolleau, MD, PhD[4,5,6]; et al

Author Affiliations

JAMA Cardiol
Published Online: September 24, 2025
2025;10;(11):1121-1129.
doi:10.1001/jamacardio.2025.3055

Design, Setting, and Participants This was a cohort analysis of retrospectively collected longitudinal data from the DM1 Heart Registry. The setting included cardiology and neurology departments of 6 French university hospitals. Study participants were selected from individuals enrolled in the DM1 Heart Registry between 2000 and 2020. The DM1 Heart Registry includes adults with genetically confirmed DM1. Included patients had a history of first EPS after 1999 and no personal history of advanced atrioventricular block or sustained ventricular tachycardia. Study data were analyzed from January to July 2025.

Results Of 1778 adults with genetically confirmed DM1 enrolled in the DM1 Heart Registry, a total of 706 patients (mean [SD] age, 42 [13] years; 359 male [51%]) were included in this study. At baseline, 273 patients (38%) had an HV interval greater than or equal to 70 milliseconds, and 232 (32%) met ECG criteria. Over a median (IQR) follow-up of 5.9 (2.3-9.7) years, 99 patients (14%) experienced an MBAE. In multivariable Cox and joint models incorporating baseline and time-varying values of PR and QRS durations, the HV interval was the only variable significantly associated with the incidence of MBAEs (hazard ratio [HR], 1.77; 95% CI, 1.46-2.16; $P < .001$ and HR, 1.78; 95% CI, 1.40-2.22; $P = .001$, respectively). Compared with ECG-based criteria, the EPS criterion proved to be a more reliable (HR, 2.89; 95% CI, 1.92-4.34 vs HR, 1.95; 95% CI, 1.31-2.89) and more sensitive (performance index [SE], 68.35% [6.24%] vs 34.76% [6.47%]) predictor of MBAE and accurately reclassified 28.8% of patients with an MBAE. Lowering the threshold to HV greater than or equal to 65 milliseconds further improved sensitivity (performance index [SE], 90.18% [3.85%]) and net reclassification improvement (33.7%; 95% CI, 19.6%-48.2%) for MBAE prediction.

major bradyarrhythmic events

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Electrophysiological Study With Prophylactic Pacing and Survival in Adults With Myotonic Dystrophy and Conduction System Disease

PR > 200 ms
QRS > 100 ms

JAMA. 2012;307(12):1292-1301

Context Up to one-third of patients with myotonic dystrophy type 1 die suddenly. Thus far, no intervention has effectively prevented sudden death.

Objective To determine whether an invasive strategy based on systematic electrophysiological studies and prophylactic permanent pacing is associated with longer survival in patients presenting with myotonic dystrophy type 1 and major infranodal conduction delays than a noninvasive strategy.

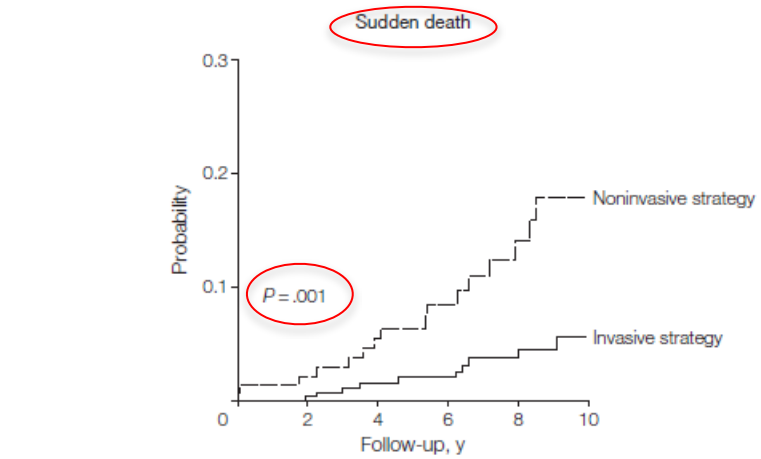
Design, Setting, and Patients A retrospective study, the DM1 Heart Registry included 914 consecutive patients older than 18 years with genetically confirmed myotonic dystrophy type 1 who were admitted to the Neurological Unit of the Myology Institute of Pitié-Salpêtrière Hospital, a teaching medical center in Paris, France, between January 2000 and December 2009.

Interventions Among 486 patients whose electrocardiogram showed a PR interval greater than 200 milliseconds, a QRS duration greater than 100 milliseconds, or both, the outcome of 341 (70.2%) who underwent an invasive strategy was compared with 145 (29.8%) who underwent a noninvasive strategy. A propensity score risk adjustment and propensity-based matching analysis was used to account for selection biases.

Main Outcome Measures Rates of overall survival (main outcome measure) and sudden death, respiratory death, and other deaths (secondary outcome measures).

Results Over a median follow-up of 7.4 years (range, 0-9.9 years), 50 patients died in the invasive strategy group and 30 died in the noninvasive strategy group (hazard ratio [HR], 0.74 [95 CI, 0.47-1.16]; *P*=.19), corresponding to an overall 9-year survival of 74.4% (95% CI, 69.2%-79.9%). Regardless of the technique used to adjust for between-group differences in baseline characteristics, the invasive strategy was associated with a longer survival, with adjusted HRs ranging from 0.47 (95% CI, 0.26-0.84; *P*=.01) for a covariate-adjusted analysis of propensity-matched data to 0.61 (95% CI, 0.38-0.99; *P*=.047) for an analysis adjusted for propensity score quintiles. The survival difference was largely attributable to a lower incidence of sudden death, which occurred in 10 patients in the invasive strategy group and in 16 patients in the noninvasive strategy group, with HRs ranging from 0.24 (95% CI, 0.10-0.56; *P*=.001) for an analysis adjusted for propensity score quintiles and covariates to 0.28 (95% CI, 0.13-0.61; *P*=.001) for an unadjusted analysis of propensity-matched data.

Conclusion Among patients with myotonic dystrophy type 1, an invasive strategy was associated with a higher rate of 9-year survival than a noninvasive strategy.



No. at risk					
Invasive strategy	341	270	205	165	106
Noninvasive strategy	145	124	98	67	43

ESC European Society of Cardiology
Europace (2022) 24, 1148–1155
https://doi.org/10.1093/europace/euab282

Arrhythmic CArdiac DEath in MYotonic dystrophy type 1 patients (ACADEMY 1) study: the predictive role of programmed ventricular stimulation

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Aims Myotonic dystrophy type 1 (DM1) predisposes to the development of life-threatening arrhythmias and sudden cardiac death. Our study aimed to evaluate the prognostic value of programmed ventricular stimulation (PVS) in DM1 patients with conduction system disease.

Methods and results Arrhythmic CArdiac DEath in MYotonic dystrophy type 1 patients (ACADEMY 1) is a double-arm non-randomized interventional prospective study. Myotonic dystrophy type 1 patients with permanent cardiac pacing indication were eligible for the inclusion. The study population underwent to pacemaker (PM) or implantable cardioverter-defibrillator (ICD) implantation according to the inducibility of ventricular tachyarrhythmias at PVS. Primary endpoint of the study was a composite of appropriate ICD therapy and cardiac arrhythmic death. The secondary study endpoint was all-cause mortality. Seventy-two adult-onset DM1 patients (51 ± 12 years; 39 male) were enrolled in the study. A ventricular tachyarrhythmia was induced in 25 patients (34.7%) at PVS (PVS+) who underwent dual chambers ICD implantation. The remaining 47 patients (65.3%) without inducible ventricular tachyarrhythmia (PVS−) were treated with dual-chamber PM. During an average observation period of 44.7 ± 10.2 months, nine patients (12.5%) met the primary endpoint, four in the ICD group (16%) and five (10.6%) in the PM group. Thirteen patients died (18.5%), 2 in the ICD group (8%) and 11 in PM group (23.4%). The Kaplan–Meier analysis did not show a significantly different risk of both primary and secondary endpoint event rates between the two groups.

Conclusions The inducibility of ventricular tachyarrhythmias has shown a limited value in the arrhythmic risk stratification among DM1 patients.

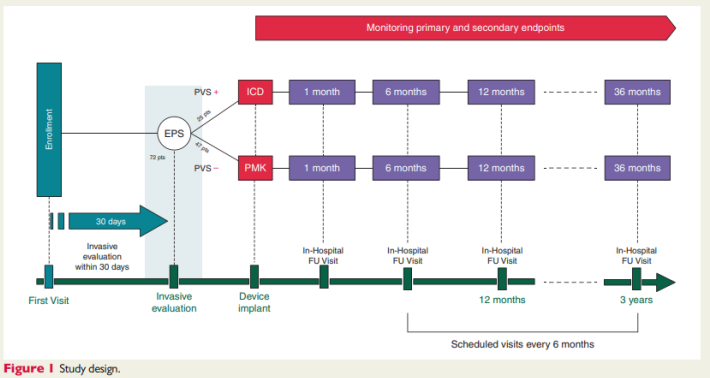
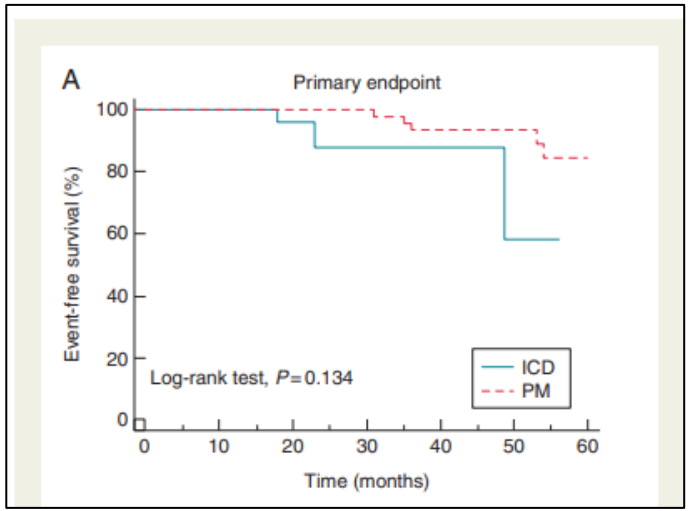


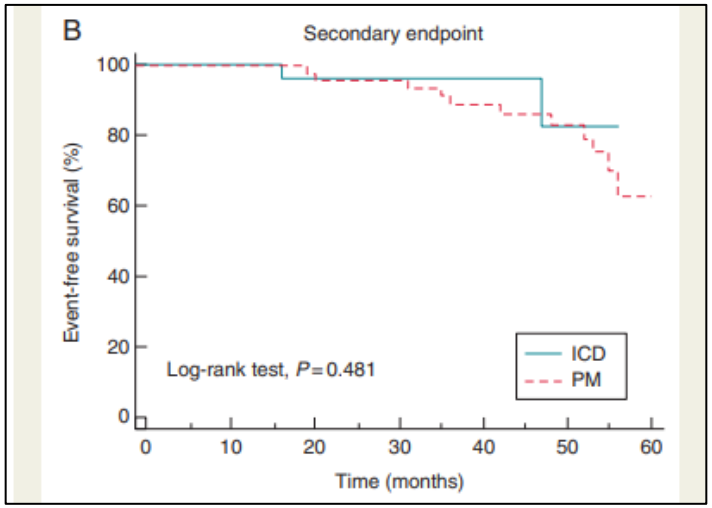
Figure 1 Study design.

1°Endp: appropr AICD therapy + arrhythmic death

2°Endp: all-cause mortality



1°Endp:
appropr AICD therapy + arrhythmic death



2°Endp:
all-cause mortality

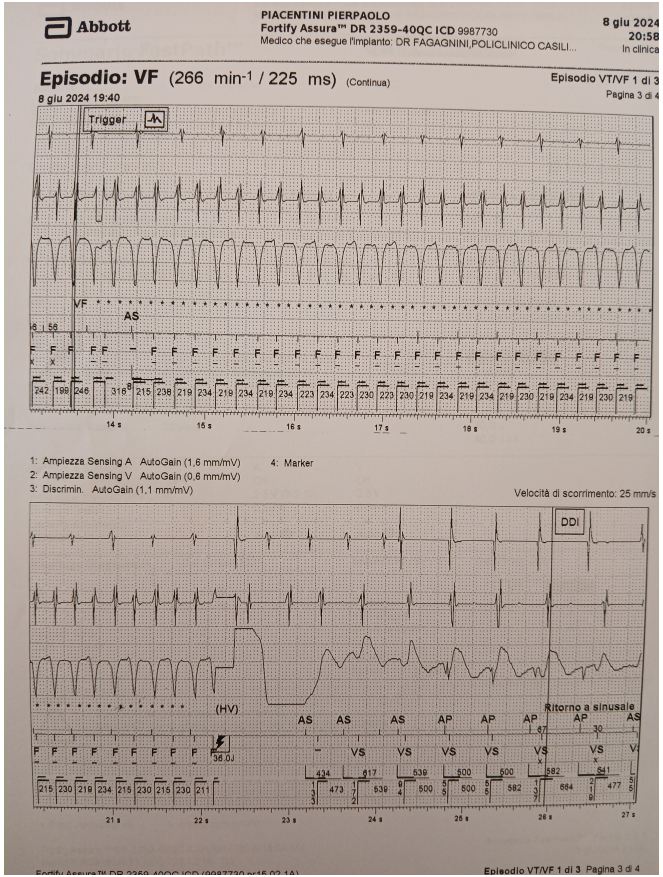
PP,

Man,

EPS +. AICD

LV scar (midwall, lateral wall)

8/6/ 2024. Three **appropriate AICD shocks** (4 years post implant)



ESC European Society of Cardiology
European Heart Journal (2022) 00, 1–130
https://doi.org/10.1093/eurheartj/ehac262

ESC GUIDELINES

2022 ESC Guidelines for the management of patients with ventricular arrhythmias and the prevention of sudden cardiac death

Developed by the task force for the management of patients with ventricular arrhythmias and the prevention of sudden cardiac death of the European Society of Cardiology (ESC)

Endorsed by the Association for European Paediatric and Congenital Cardiology (AEPC)

Factors favouring ICD implantation:

LBBB/AF/NSVT/palpitations

LV dysfunction, significant LGE in CMR

CTGexpansion, family history of SD,

2022 HRS expert consensus statement on evaluation and management of arrhythmic risk in neuromuscular disorders

William J. Groh, MD, MPH, FHRS (Chair),[1] Deepak Bhakta, MD, MBA, FHRS, FACC, FAHA, FACP, CCDS (Vice-Chair),[2] Gordon F. Tomaselli, MD, FHRS (Vice-Chair),[3] Ryan G. Aleong, MD, FHRS,[4] Ricardo Alkmim Teixeira, MD, PhD,[5*] Anthony Amato, MD,[6+] Samuel J. Asirvatham, MD, FHRS,[7] Yong-Mei Cha, MD, FHRS,[7] Domenico Corrado, MD, PhD, FESC,[8‡] Denis Duboc, MD, PhD,[9] Zachary D. Goldberger, MD, FHRS,[10] Minoru Horie, MD, PhD,[11§] Joseph E. Hornyak, MD, PhD,[12¶] John Lynn Jefferies, MD, MPH, FACC, FAHA, FHFSA,[13#] Stefan Kääb, MD, PhD,[14‡] Jonathan M. Kalman, MBBS, PhD, FHRS,[15] Naomi J. Kertesz, MD, FHRS, CEPS-P,[16**] Neal K. Lakdawala, MD,[6++] Pier D. Lambiase, BCH, BM, MBChB, PhD, FHRS,[17‡] Steven A. Lubitz, MD, MPH,[18‡‡] Hugh J. McMillan, MD, MSc,[19§§] Elizabeth M. McNally, MD, PhD,[20#] Margherita Milone, MD, PhD,[7+] Narayanan Namboodiri, MBBS, MD,[21¶¶] Saman Nazarian, MD, PhD, FHRS,[22] Kristen K. Patton, MD, FHRS,[23] Vincenzo Russo, MD, PhD,[24‡] Frederic Sacher, MD, PhD,[25‡] Pasquale Santangeli, MD, PhD,[22] Win-Kuang Shen, MD, FHRS,[26] Dario C. Sobral Filho, MD, PhD,[27##] Bruce S. Stambler, MD, FHRS,[28] Claudia Stöllberger, MD,[29] Karim Wahbi, MD, PhD,[30] Xander H.T. Wehrens, MD, PhD, FHRS,[31] Menachem Mendel Weiner, MD,[32***] Matthew T. Wheeler, MD, PhD,[33§] Katja Zeppenfeld, MD, PhD[34‡]

tions for ventricular arrhythmias, sudden cardiac death, and use of implantable lefibrillators in myotonic dystrophy types 1 and 2

udies that support recommendations are summarized in Appendix 3

COR	LOE	Recommendations	References
1	B-NR	1. In patients with DM1 or DM2 in whom ICD therapy is planned, an ICD system with permanent pacing capability is recommended.	104, 106, 110, 121, 132
1	B-NR	2. In patients with DM1 or DM2 who are survivors of spontaneously occurring hemodynamically significant sustained VT or VF, ICD therapy is indicated if concordant with the patient's goals of care and clinical status.	96, 104, 132, 133
1	B-NR	3. In patients with DM1 or DM2 and an LVEF ≤35% despite guideline-directed medical therapy, ICD therapy is indicated if concordant with the patient's goals of care and clinical status.	97, 132
1	B-NR	4. In patients with DM1 or DM2 in whom clinically relevant ventricular arrhythmias are induced during electrophysiological study, ICD therapy is recommended if concordant with the patient's goals of care and clinical status.	110, 124-126, 132, 133
2b	B-NR	5. In patients with DM1 or DM2 in whom permanent pacemaker implantation is indicated, ICD therapy may be considered if concordant with the patient's goals of care and clinical status.	106, 121, 132

While induction of ventricular arrhythmias including bundle branch reentrant ventricular tachycardia (BBRVT) has been observed in DM1 patients, the predictive value of programmed ventricular stimulation has not been thoroughly evaluated in patients with DM1 but may be helpful in patients with symptoms of unexplained syncope, presyncope, or palpitations. Stimulation protocols favoring induction of BBRVT have been described and may be utilized in this setting.[26, 124, 125, 128, 129] Unlike with other substrates, no data exist regarding appropriateness of ICD implantation in DM1 and DM2 patients who are found to have inducible ventricular arrhythmias, although inducibility in carefully selected patients suggests the presence of substrate prone to developing future VT or VF. Further studies are required to better clarify this issue.[124, 125, 128, 129]

CTGexpansion, family history of SD,

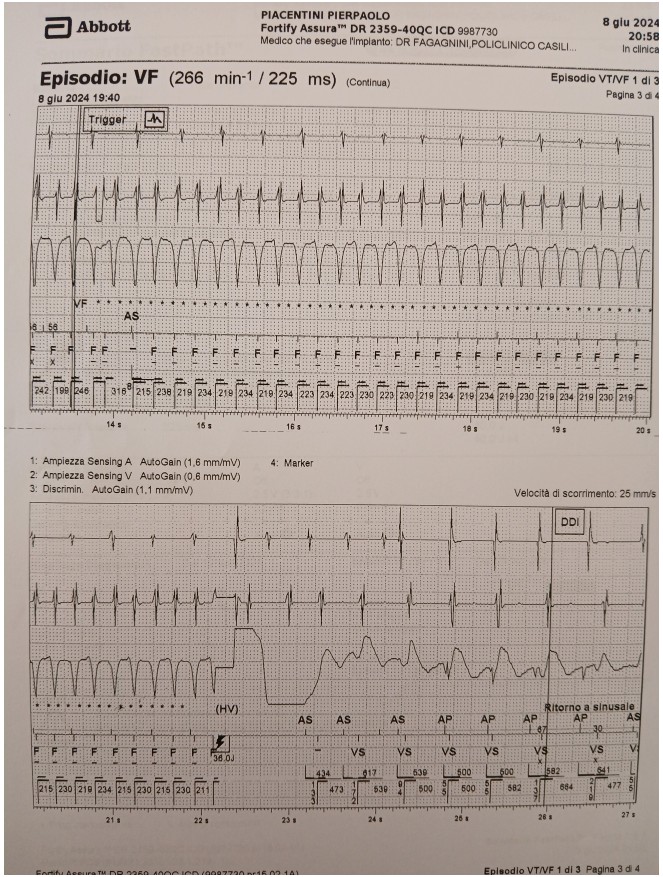
PP,

Man,

EPS +. AICD

LV scar (midwall, lateral wall)

8/6/ 2024. Three **appropriate AICD shocks** (4 years post implant)

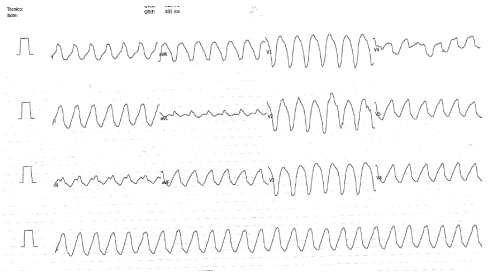


CF,

Woman,

EPS +. AICD

Normal cMRI (T1 mapping not done)



8/3/ 2016 (1 year post implant) **SVT (11 sec) interrupted by ATP.**

2-11-2020. **Six appropriate AICD shocks.**
VT ablation

SCD Risk stratification: open questions

1) Utility of EPS for prediction of VT/VF (MD1 patients with /without LGE)

2) Adequate timing for EPS repetition.

3) Utility of CCD progression velocity

4) Utility of LRI implantation

Thank you

proximal myotonic myopathy, is caused by a CCTG expansion in the zinc finger 9 (ZNF9) gene and has a prevalence of 1:20,000