ONLINE DATA SUPPLEMENT

Characterization of a unique form of arrhythmic cardiomyopathy caused by recessive mutation in LEMD2

Nelly Abdelfatah[a*], PhD; Ruping Chen[b*], PhD; Henry J. Duff[a], MD; Colette M. Seifer[c], MB; Ilan Buffo[d], MD; Cathleen Huculak[e], MSc; Stephanie Clarke[f], MSc; Robin Clegg[g], MD; Davinder S. Jassal[c], MD; Paul MK Gordon[h], PhD; Carole Ober[i], PhD; Care4Rare Canada Consortium; Patrick Frosk[f,j], PhD, MD; Brenda Gerull[a,b,k], MD.

[a]Department of Cardiac Sciences, Libin Cardiovascular Institute of Alberta, Cumming School of Medicine, University of Calgary, Calgary, Alberta, Canada.

[b]Comprehensive Heart Failure Center, University Hospital Würzburg, Würzburg, Germany.

[c]Section of Cardiology, Department of Internal Medicine, Max Rady College of Medicine, Rady Faculty of Health Sciences, University of Manitoba, Winnipeg, MB, Canada.

[d]Variety Children's Heart Centre, University of Manitoba, Winnipeg, MB, Canada.

[e]Department of Medical Genetics, Alberta Health Services, Calgary, AB, Canada.

[f]Department of Biochemistry and Medical Genetics, Max Rady College of Medicine, Rady Faculty of Health Sciences, University of Manitoba, Winnipeg, MB, Canada

[g]Department of Pediatrics, University of Calgary, Calgary, AB, Canada.

[h]Cumming School of Medicine Centre for Health Genomics and Informatics, University of Calgary, Calgary, AB, Canada.

[i]Department of Human Genetics, The University of Chicago, Chicago, IL, USA.

[j]Department of Pediatrics and Child Health, Max Rady College of Medicine, Rady Faculty of Health Sciences, University of Manitoba, Winnipeg, MB, Canada.

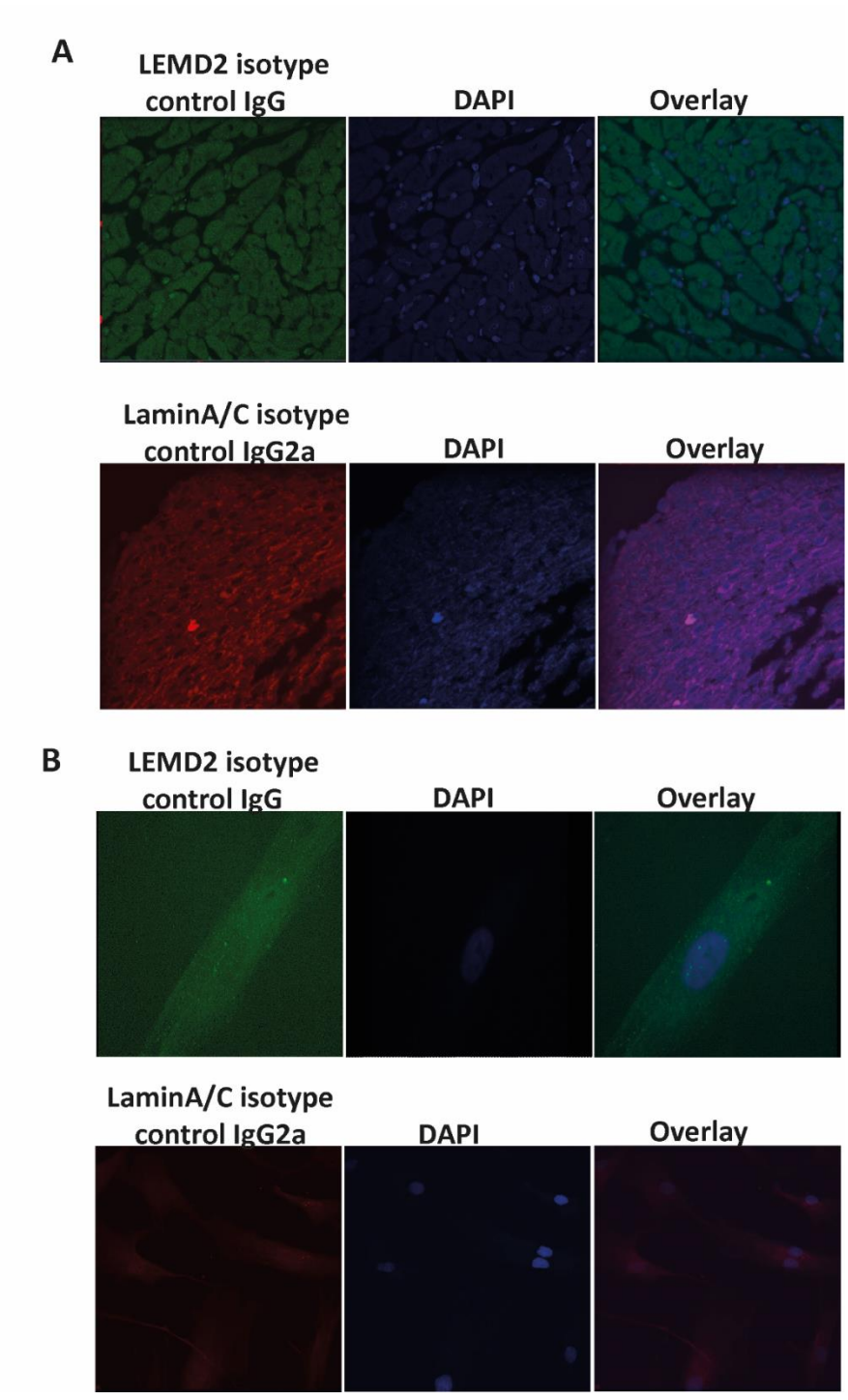
[k]Department of Internal Medicine I, University Hospital Würzburg, Würzburg, Germany.

*contributed equally to this work

Address for correspondence

Dr. Brenda Gerull, Comprehensive Heart Failure Center and Department of Internal Medicine I, University Hospital Würzburg, Am Schwarzenberg 15, 97078 Würzburg, Germany, Phone: ++49-931-201-46457, Fax: ++49-931-201-646457, email: gerull_b@ukw.de

Online Figures and Legends



Supplement Figure 1: Isotype controls for LEMD2 and Lamin A/C antibodies. Heart tissue (A) and fibroblast cells (B) were stained with anti-rabbit IgG isotype control and IgG2a anti-mouse isotype control.

Supplement Tables and Table Legends

Supplemental Table 1: Primary and secondary antibodies

Antibody	Company	Catalogue Number	Application	Dilution
RB anti LEMD2	Novus Biologicals	NBP1-85175	IHC, WB	1:50, 1:1000
MS anti Lamin A/C	Cell signaling	4777	IHC, WB	1:100, 1:1000
RB anti-eGFP	Abnova	Ab111258	WB	1:1000
RB anti-Annexin V	Abcam	Ab14196	WB	1:1000
RB anti-Cleaved Caspase-3 (Asp	Cell signaling	9661	WB	1:1000
RB anti-Aurora B	Thermo Fisher Scientific	PA5-14075	WB	1:1000
RB anti -GAPDH	Abcam	Ab181602	WB	1:3000
RB IgG-Alexa 488	Invitrogen	A11034	IHC	1:100
MS IgG- Alexa 555	Invitrogen	A21422	IHC	1:100
RB IgG HRP	GE Healthcare	NA934V	WB	1:3000
MS IgG HRP	GE Healthcare	NXA931	WB	1:5000

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Supplemental Table 2: Haplotypes of affected family members

Gene	Variant	hg 38 Position	rs#	Global MAF	Family 600								Family 290			
					II-18		III-4		II-2		II-3		III-12		III-13	
GMNN	c.357+39C>T	chr6:24774931	rs2307308	0.0112/56	T	T	T	T	T	T	T	T	T	T	T	C
HIST1H4E	c.*28T>A	chr6:26204552	rs41266809	0.0026/13	A	A	A	A	A	A	A	A	A	A	A	T
HIST1H2BN	c.*10C>T	chr6:27838662	rs112586220	0.0028/14	T	T	T	T	T	T	T	T	T	T	T	C
ITPR3	g.33650433G>A	chr6:33620365	rs78238190	0.0064/32	A	A	A	A	A	A	A	A	A	A	A	A
LEMD2	c.38T>G	chr6:33771202	-	-	G	G	G	G	G	G	G	G	G	G	G	G
RNF8	c.*1189G>A	ch6:37353972	rs34150698	0.0292/146	A	A	A	A	A	A	A	A	-	-	-	-
CUL7	c.4762C>A	ch6:43037617	rs147493246	0.0057/28	A	A	A	A	-	-	-	-	C	C	A	C
PTK7	c.19G>A	chr6:43076268	rs139636574	0.0056/28	A	A	A	A	-	-	-	-	G	G	A	G

Supplemental Table 2: Haplotypes of six affected family members across an 18.3Mb region at chr6p between chr6:24,774,931-chr6:43,076,268.

Shared haplotypes are denoted by the red letters. The region of homozygosity is narrowed down to a 6.8 Mb by an affected member of family 290 III-13. c.38T>G variant in LEMD2 gene was the only variant segregates with the phenotype in the two families. rs# referred to the SNP number. MAF refereed to minor allele frequency and the global MAF for the variants were taken from SNP database Available from: http://www.ncbi.nlm.nih.gov/SNP