SUPPLEMENTAL MATERIALS

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Supplemental Table 1: List of Potentially Causative Genetic Variants

Gene	Location	Description	Conservation score**	Cardiac expression-LV*	Cardiac expression-LAA*	Mutation Taster prediction	SIFT score	Polyphen score	rsID	MAF ExAC	MAF EVS
NBAS	chr2-15319114:T->C	T2280A	-4.204	6.617	5.178	polymorphism	0.273	benign	rs140346039	0.157	0.100
FAM179A	chr2-29274799:G->A	R967H	3.643	3.352	4.186	disease causing	0.007	probably damaging	rs200413260	0.036	0.033
COL6A6	chr3-130293046:G->C	G1075A	2.335	0.297	2.337	polymorphism	0.068	probably damaging	rs149093613	0.131	0.294
PLOD3	chr7-100852231:G->A	P564L	5.062	13.487	18.047	disease causing	0.001	probably damaging	-	0.002	-
ST7	chr7-116770645:C->T	R184C	3.372	4.215	5.022	disease causing	0.000	benign	-	0.002	-
FLNC	chr7-128494628:G->A	V2297M	5.775	264.079	95.877	disease causing	0.001	probably damaging	-	-	-
PRPF40B	chr12-50024353:C->T	P10L	1.89	3.743	4.331	disease causing	0.101	NA	-	0.008	-
CCDC176	chr14-74495929:G->A	A110T	1.945	-	0.946	disease causing	0.139	possibly damaging	-	0.008	-
IRF2BPL	chr14-77493085:C->T	A351T	2.453	8.372	13.686	disease causing	0.139	probably damaging	rs369556741	-	0.008
TBXA2R	chr19-3594840:C->G	K406N	-0.508	1.152	1.014	polymorphism	0.000	NA	rs201087249	0.032	-
MYO18B	chr22-26224825:A->G	K957E	0.491	26.184	19.971	polymorphism	0.164	benign	rs200316291	0.149	0.162

*Expression levels from GTEx Portal expressed in RPKM

**Conservation Score is based on PhyloP Vertebrate basewise conservation (phyloP46wayAll)

Supplemental Table 2: Rare variants shared in individuals II-2, II-5, and II-7

Gene	Chr	Position	Ref_allele	Alt_allele	ExAC freq	Function	Polyphen-2 class
PICK1:NM_012407	22	38469017	A	C	8.13E-06	Missense:GAT=>GCT:D234A	deleterious
IRF2BPL:NM_024496	14	77493085	C	T	8.14E-06	Missense:GCC=>ACC:A351T	deleterious
CISH:NM_013324	3	50645169	G	A	1.63E-05	Missense:CGC=>TGC:R233C	neutral
AP1AR:NM_001128426	4	113187833	G	A	1.63E-05	Missense:GAC=>AAC:D169N	deleterious
PLOD3:NM_001084	7	100852231	G	A	1.63E-05	Missense:CCG=>CTG:P564L	deleterious
MUC16:NM_024690	19	9057031	T	C	1.63E-05	Missense:ATG=>GTG:M10139V	neutral
CCDC176:NM_025057	14	74495929	G	A	1.68E-05	Missense:GCC=>ACC:A110T	deleterious
ST7:NM_021908	7	116770645	C	T	2.44E-05	Missense:CGT=>TGT:R184C	neutral
GPR179:NM_001004334	17	36483591	A	G	2.45E-05	Missense:CTA=>CCA:L1954P	neutral
MUC16:NM_024690	19	9071624	A	T	2.45E-05	Missense:AGT=>AGA:S5274R	deleterious
ZZEF1:NM_015113	17	3926025	G	A	3.25E-05	Missense:ACG=>ATG:T2397M	deleterious
EXOC3L2:NM_138568	19	45735079	G	C	4.07E-05	Missense:CCC=>CGC:P11R	deleterious
PRPF40B:NM_001031698	12	50024353	C	T	4.08E-05	Missense:CCC=>CTC:P10L	NA
OR7D4:NM_001005191	19	9324990	G	A	5.69E-05	Missense:CCG=>CTG:P175L	deleterious
CRYZ:NM_001130043	1	75188918	C	T	7.32E-05	Missense:GTG=>ATG:V50M	deleterious
CD163:NM_004244	12	7640504	C	G	7.32E-05	Missense:GGA=>CGA:G534R	deleterious
LARP4B:NM_015155	10	909779	C	T	8.13E-05	Missense:GCA=>ACA:A112T	deleterious
FBXO10:NM_012166	9	37512704	C	T	8.17E-05	Missense:CGC=>CAC:R904H	neutral
HEATR5A:NM_015473	14	31856380	G	C	1.39E-04	Missense:CTT=>GTT:L379V	neutral
RCN3:NM_020650	19	50045849	C	T	1.47E-04	Missense:GCG=>GTG:A240V	deleterious
DOCK8:NM_203447	9	328143	C	T	1.71E-04	Missense:CCG=>CTG:P339L	deleterious
FAM179A:NM_199280	2	29274799	G	A	2.13E-04	Missense:CGC=>CAC:R967H	deleterious
TBXA2R:NM_201636	19	3594840	C	G	2.52E-04	Missense:AAG=>AAC:K406N	NA
COL22A1:NM_152888	8	139662020	T	A	2.93E-04	Exonic_Splice.Missense:GAT=>GTT:D1112V	deleterious
TIMP1:NM_003254	X	47444408	G	T	3.09E-04	Missense:ATG=>ATT:M65I	neutral
CCDC110:NM_001145411	4	186379567	T	C	4.07E-04	Missense:CAT=>CGT:H688R	neutral
ANKRD13A:NM_033121	12	110475337	A	G	6.02E-04	Missense:CAG=>CGG:Q584R	neutral
PKHD1:NM_170724	6	51824808	T	A	7.73E-04	Missense:CAG=>CTG:Q1923L	neutral
TRPV4:NM_001177431	12	110224603	C	T	7.73E-04	Missense:GTA=>ATA:V716I	deleterious
ZNF521:NM_015461	18	22806966	C	T	9.11E-04	Missense:GGG=>AGG:G306R	neutral
PTAFR:NM_001164721	1	28477220	C	T	Not observed	Missense:GTG=>ATG:V105M	deleterious
FLNC:NM_001458	7	128494628	G	A	Not observed	Missense:GTG=>ATG:V2297M	deleterious
CPA2:NM_001869	7	129919465	G	T	Not observed	Missense:GGG=>GTG:G317V	deleterious
UGGT2:NM_020121	13	96515959	C	T	Not observed	Missense:GAA=>AAA:E1190K	neutral

Supplemental Table 3: Primers for Site-directed Mutagenesis

Primer ID	Primer sequences
FLNCpart_1F	gctggccgggcgcccctgcaGGTGGCTGTGCTGGGCCC
FLNCpart_1R	tgggcaggcgCTTCAGCAGGCAGGGCTCC
FLNCpart_2F	cctgctgaagCGCCTGCCCAACCGGCAC
FLNCpart_2R	tgaaaggggcTGCCGGGCA**T**GTGCTGGC
FLNCpart_3F	**a**tgcccggcaGCCCCTTTCAGTTCACTGTG
FLNCpart_3R	agatgagtttctgctcgagcggccGCGTACGCGTAGGGACCT

Base substitutions are indicated in red. Lowercase letters indicate 5’ or 3’ overhang utilized in Gibson Assembly.

Supplemental Table 4: Potential off-target candidates for CRISPR-Cas9 mediated mutagenesis

sequence	score	mismatches	UCSC gene	locus
TGCCGTGGACAGCACGTGCCCAG	4.3	2MMs [2:9]		chr15:-51566300
TGCCGTGGCCAGCACCTGCCCAG	2.1	2MMs [2:16]		chr22:+19695074
TACAAGGGCCAGCACGTGCCTGG	1.4	3MMs [4:5:6]	NM_001456	chrX:-153580645
GACAGTGGCCAGCACGTGTCCAG	1.1	3MMs [1:4:19]		chr2:-144584224
AACCGTTGCCAGCACGTGCAGAG	1.1	3MMs [1:7:20]		chr8:+1532724
AACTGAGTCCAGCACGTGCCTAG	0.8	4MMs [1:4:6:8]		chr12:+117147314
TCCCGTGGCCTGCACGTGCGGAG	0.8	3MMs [2:11:20]		chr19:-40769138
TGCTGTGCCCTGCACGTGCCCGG	0.8	4MMs [2:4:8:11]	NM_001003841	chr5:+1222957
TCCCATGGCTAGCACGTGCTGGG	0.6	4MMs [2:5:10:20]		chr2:-107256808
GACCATGACCAGCACGAGCCCAG	0.6	4MMs [1:5:8:17]		chr13:-31309391
CCCCGGAGCCAGCACGTGCCCGG	0.6	4MMs [1:2:6:7]		chr22:+49023130
TGCCGCCACCAGCACGTGCCCGG	0.6	4MMs [2:6:7:8]		chr9:-34957777
TGCCATGACCAGCACGTGGCTGG	0.5	4MMs [2:5:8:19]		chr15:-88759708
TACCATACCCACCACGTGCCTGG	0.5	4MMs [5:7:8:12]		chr10:-23725961
CACCCTGGGCACCACGTGCCGAG	0.4	4MMs [1:5:9:12]	NM_033081	chr20:+61512541
CACTGAGGCCAGCACGTGCTGAG	0.4	4MMs [1:4:6:20]		chr2:-237638064
GACTGTGCCCAGCAGGTGCCTGG	0.4	4MMs [1:4:8:15]	NM_001018071	chr10:+49457281
TGCCCTGTCCAGCAGGTGCCTGG	0.4	4MMs [2:5:8:15]		chr4:+1163687
TAACAGGGCCAGCACGTGCTGAG	0.4	4MMs [3:5:6:20]		chr8:-144637771
TGATGTGGCCAGCAAGTGCCTGG	0.4	4MMs [2:3:4:15]		chr9:-139121628
CAGCGTGGCCTTCACGTGCCTGG	0.4	4MMs [1:3:11:12]		chr6:+21488275
TACCATGGCAGCCACGTGCCCGG	0.3	4MMs [5:10:11:12]		chr11:-12221998
TTCCGGAGCCTGCACGTGCCCGG	0.3	4MMs [2:6:7:11]		chr1:-22909646
TTCCCTGGCCTGCACGTGGCCAG	0.3	4MMs [2:5:11:19]		chr20:+61320478
TCACGTGGCCACTACGTGCCAAG	0.3	4MMs [2:3:12:13]		chr13:+36449100
GACCATGGCCAGAACGTGCTGAG	0.3	4MMs [1:5:13:20]		chr11:+134442994
GTCAGTGGCCAGCACCTGCCCAG	0.3	4MMs [1:2:4:16]	NM_175872	chr19:+35451280
CCACGTGGCCAGCACCTGCCAAG	0.3	4MMs [1:2:3:16]		chr22:-47763978
TCCTGTGTCCAGCACCTGCCCAG	0.3	4MMs [2:4:8:16]		chr3:+50305354
TACCTTGGCCAGCAGGTGACAGG	0.3	3MMs [5:15:19]		chr2:+47550555
TACCCAGGTCAACACGTGCCAGG	0.3	4MMs [5:6:9:12]		chr22:+29913489
CACCTTGGCAAGCACCTGCCGAG	0.3	4MMs [1:5:10:16]		chr1:-22515754
GCCCGTGGCTAGCACCTGCCCGG	0.3	4MMs [1:2:10:16]		chr16:+87598783
TGCCTTGGACAGCAGGTGCCTAG	0.3	4MMs [2:5:9:15]		chr17:+1404983
TGCCGAGCCCAGCAAGTGCCAGG	0.2	4MMs [2:6:8:15]		chr20:+62129853
TACCCTGGCCACCACCTGCCCAG	0.2	3MMs [5:12:16]		chr2:+102244068
TACAGGGCCCAGCAGGTGCCCAG	0.2	4MMs [4:6:8:15]		chr1:+916943
AGCCGTGGCCAGCACGTGAAGGG	0.2	4MMs [1:2:19:20]		chrX:+153045568
CACCGTGGCCAGCACGTAACAGG	0.2	3MMs [1:18:19]		chr2:-6038494
TACTGTCTCCAGCACGTTCCGAG	0.2	4MMs [4:7:8:18]		chr2:+237045432
GGCCGTGGCCACCAAGTGCCAAG	0.2	4MMs [1:2:12:15]	NM_139209	chr3:+141497508
GACAGTGGCCAACAAGTGCCAAG	0.2	4MMs [1:4:12:15]		chr13:-51517829
TGCAGTGGCCAACAGGTGCCTGG	0.2	4MMs [2:4:12:15]		chr1:+1092411
TGCCCTGGCCATCAAGTGCCTGG	0.2	4MMs [2:5:12:15]		chr8:+41710326
TCACGTGGCCAGCACGGGACGGG	0.2	4MMs [2:3:17:19]		chr4:-3255729
TACCCTGGGCTGCACGAGCCAGG	0.2	4MMs [5:9:11:17]		chr7:-32258079
TATGGTGGCCATCAAGTGCCAGG	0.2	4MMs [3:4:12:15]	NM_001456	chrX:-153587608
CACCGTGGCCGCCACGTGCTGGG	0.2	4MMs [1:11:12:20]		chr4:-7521263
TCCCGTTGCCAGGACGTGCGAGG	0.2	4MMs [2:7:13:20]		chr20:+47003220

Supplemental Table 5: Exome Sequencing Coverage Data

	II-2	III-2	IV-3	II-5	II-7
Total number of reads	53,806,354	108,395,860	100,257,666	98,948,094	91,120,572
Mapped reads	53,352,790	107,926,237	99,732,477	98,471,905	90,848,583
Average depth of coverage	48X	144X	135X	131X	123X
% of targeted regions with at least 20X coverage	70.09%	98.51%	98.31%	97.89%	97.98%

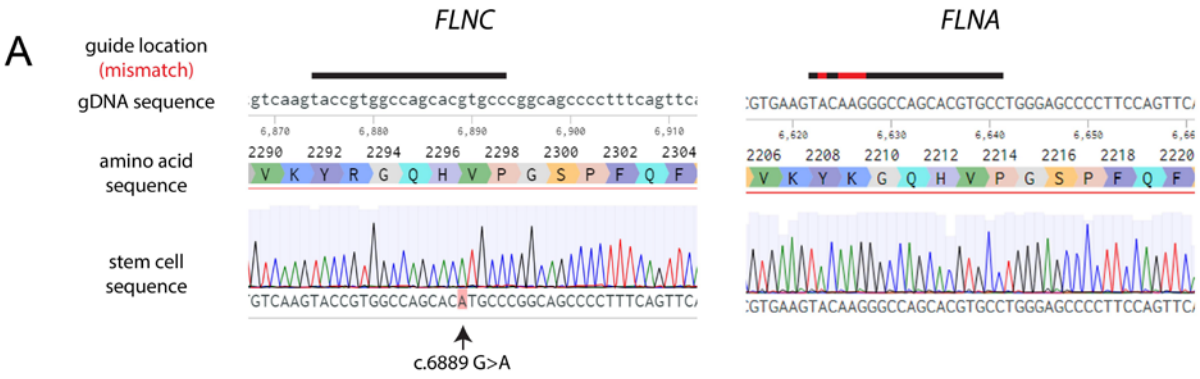
Supplemental Table 6: Number of Candidate Variants Identified by Exome Sequencing

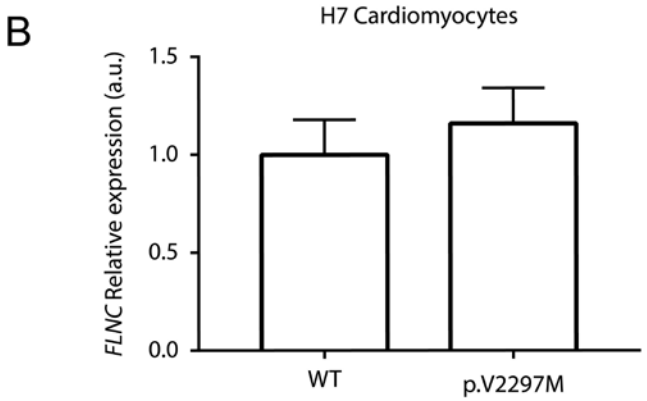
	II-2	III-2	IV-3	II-5	II-7
Total variants	19611	20849	21029	20665	20676
Variants following filtration based on population databases	18371	20110	20339	20019	19977
Missense variants	9409	9821	9859	9671	9704
Nonsense variants	86	67	76	68	70
Stopgain variants	28	32	33	36	33
Splice site variants	64	52	52	57	58

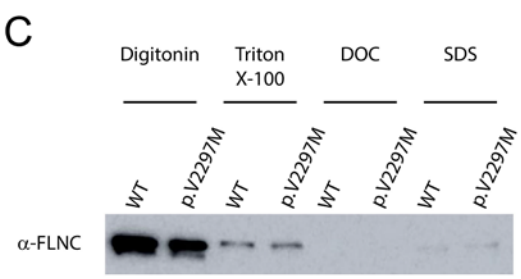
Supplemental Table 7: References for Known Filamin C mutations, illustrated in *Figure 1D*

Description	Primary Phenotypes	MAF Frequency ExAC	Citations
Truncation: p.R81*	**LDAC**	---	[1]
Truncation: p.T83*	**LDAC**	---	[1]
Missense: p.F106L	**DCM**	**0.00003390**	[2]
Truncation: p.E108*	**HCM**	**---**	[3]
Missense: p.V123A	**HCM/AF**	**---**	[3,4]
Missense: p.A193T	**Proximal/Distal Myopathy**	**---**	[5,6]
Truncation: p.L194*	**DCM**	**---**	[1]
Truncation: p.G201*	**DCM**	**---**	[1]
Missense: p.M251T	**Distal Myopathy**	**---**	[5,6]
Missense: p.N290K	**HCM**	**---**	[3]
Truncation: p.Q572*	**LDAC**	**---**	[1]
Truncation: p.L737*	**DCM**	**---**	[1]
Frameshift: p.K899_V904*del* // V899_C900*ins*	**MFM**	**---**	[7]
In frame deletion: p.V920	**MFM**	**---**	[8]
Truncation: p.P963*	**DCM**	**0.00000830**	[1]
Truncation: p.F1135*	**DCM**	**---**	[1]
Missense: p.Y1216R	**MFM**	**---**	[9]
Truncation: p.N1369*	**DCM**	**---**	[1]
Missense: p.A1539T	**HCM/AF**	**---**	[4]
Missense p.S1624L	**RCM**	**---**	[10]
Truncation: p.F1720*	**Distal Myopathy**	**---**	[11]
Truncation: p.G1800*	**LDAC**	**---**	[1]
Missense: p.R2133H	**HCM**	**---**	[3]
Truncation: p.G2070*	**DCM**	**---**	[1]
Truncation: p.S2077*	**DCM**	**---**	[1]
Truncation: p.P2081*	**DCM**	**---**	[1]
Missense: p.G2151S	**HCM**	**---**	[3]
Missense: p.I2160F	**RCM**	**---**	[10]
Missense: p.H2315N	**HCM**	**---**	[3]
Truncation: p.R2326*	**DCM**	**0.0000090**	[1]
Missense: p.T2419M	**MFM**	**---**	[12]
Missense: p.A2430V	**HCM**	**0.0002243**	[3]
Truncation: D2703*	**DCM**	**---**	[1]
Truncation: p.W2710X	**MFM**	**---**	[13]

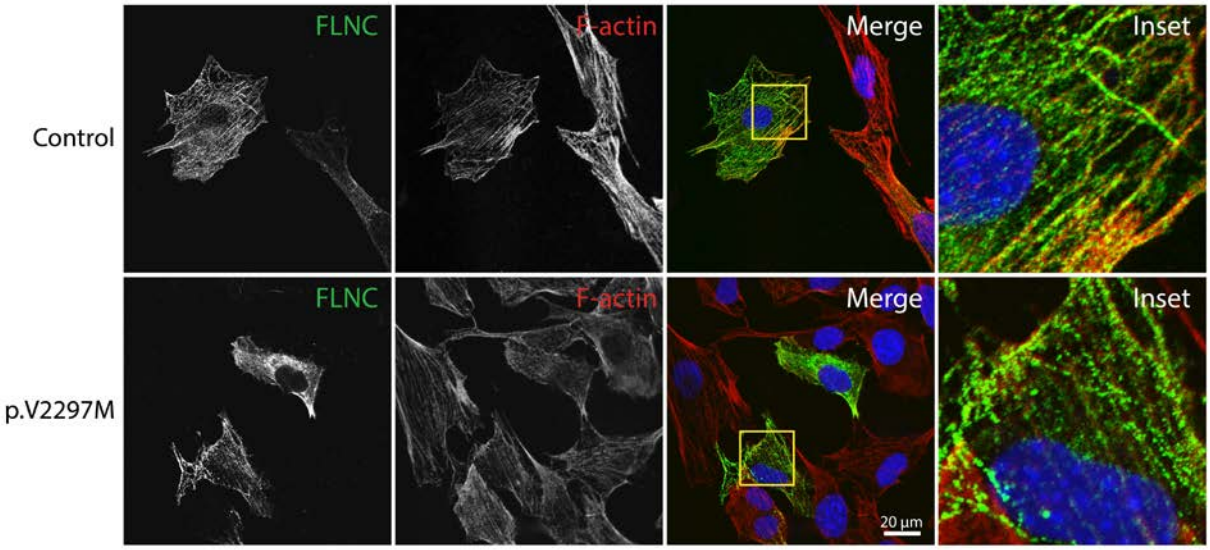
Supplemental Figure 1: Analysis of p.V2297M stem cell-derived cardiomyocytes. A: Chromatograms demonstrating successful mutation of FLNC but not FLNA in embryonic stem cells. B: qRT-PCR analysis of FLNC expression in 12 day post differentiation cardiomyocytes. C: Analaysis of prevalence of FLNC in subcellular fractions by Western blot.







Supplemental Figure 2: Localization of WT and mutant FLNC following overexpression in C2C12 myoblasts. Two dimensional reconstructions of confocal z-stacks visualizing the localization of overexpressed, FLAG-tagged FLNC (green) and filamentous actin (red) in undifferentiated C2C12 myoblasts. Merged image includes DAPI (blue) for labeling of nuclei.



Supplemental References

1. Ortiz-Genga MFMFMF, Cuenca SS, Dal Ferro M, Zorio E, Salgado-Aranda R, Climent V, Padrón-Barthe L, Duro-Aguado I, Jiménez-Jáimez J, Hidalgo-Olivares VMVMVM, García-Campo E, Lanzillo C, Suárez-Mier MP, Yonath H, Marcos-Alonso S, Ochoa JP, Santomé JL, García-Giustiniani D, Rodríguez-Garrido JL, Domínguez F, Merlo M, Palomino JJJ, Peña ML, Trujillo JP, Martín-Vila A, Stolfo D, Molina P, Lara-Pezzi E, Calvo-Iglesias FE, Nof E, Calò L, Barriales-Villa R, Gimeno-Blanes JR, Arad M, García-Pavía P, Monserrat L, Padr?n-Barthe L, Duro-Aguado I, Jim?nez-J?imez J, Hidalgo-Olivares VMVMVM, Garc?a-Campo E, Lanzillo C, Su?rez-Mier MP, Yonath H, Marcos-Alonso S, Ochoa JP, Santom? JL, Garc?a-Giustiniani D, Rodr?guez-Garrido JL, Dom?nguez F, Merlo M, Palomino JJJ, Pe?a ML, Trujillo JP, Mart?n-Vila A, Stolfo D, Molina P, Lara-Pezzi E, Calvo-Iglesias FE, Nof E, Cal? L, Barriales-Villa R, Gimeno-Blanes JR, Arad M, Garc?a-Pav?a P, Monserrat L. Truncating FLNC Mutations Are Associated With High-Risk Dilated and Arrhythmogenic Cardiomyopathies. *J Am Coll Cardiol*. 2016;68:2440–2451.

2. Reinstein E, Gutierrez-Fernandez A, Tzur S, Bormans C, Marcu S, Tayeb-Fligelman E, Vinkler C, Raas-Rothschild A, Irge D, Landau M, Shohat M, Puente XS, Behar DM, Lopez-Otın C. Congenital dilated cardiomyopathy caused by biallelic mutations in Filamin C. *Eur J Hum Genet*. 2016;24:1792–1796.

3. Valdés-Mas R, Gutiérrez-Fernández A, Gómez J, Coto E, Astudillo A, Puente DA, Reguero JR, Álvarez V, Morís C, León D, Martín M, Puente XS, López-Otín C. Mutations in filamin C cause a new form of familial hypertrophic cardiomyopathy. *Nat Commun*. 2014;5:5326.

4. Gómez J, Lorca R, Reguero JR, Morís C, Martín M, Tranche S, Alonso B, Iglesias S, Alvarez V, Díaz-Molina B, Avanzas P, Coto E. Screening of the Filamin C Gene in a Large Cohort of Hypertrophic Cardiomyopathy Patients. *Circ Cardiovasc Genet*. 2017;10:e001584.

5. van den Bogaart FJA, Claeys KG, Kley RA, Kusters B, Schrading S, Kamsteeg EJ, Voermans NC. Widening the spectrum of filamin-C myopathy: Predominantly proximal myopathy due to the p.A193T mutation in the actin-binding domain of FLNC. *Neuromuscul Disord*. 2017;27:73–77.

6. Duff RM, Tay V, Hackman P, Ravenscroft G, McLean C, Kennedy P, Steinbach A, Schöffler W, van der Ven PFM, Fürst DO, Song J, Djinović-Carugo K, Penttilä S, Raheem O, Reardon K, Malandrini A, Gambelli S, Villanova M, Nowak KJ, Williams DR, Landers JE, Brown RH, Udd B, Laing NG. Mutations in the N-terminal actin-binding domain of filamin C cause a distal myopathy. *Am J Hum Genet*. 2011;88:729–40.

7. Luan X, Hong D, Zhang W, Wang Z, Yuan Y. A novel heterozygous deletion-insertion mutation (2695-2712 del/GTTTGT ins) in exon 18 of the filamin C gene causes filaminopathy in a large Chinese family. *Neuromuscul Disord*. 2010;20:390–396.

8. Shatunov A, Olivé M, Odgerel Z, Stadelmann-Nessler C, Irlbacher K, van Landeghem F, Bayarsaikhan M, Lee H-S, Goudeau B, Chinnery PF, Straub V, Hilton-Jones D, Damian MS, Kaminska A, Vicart P, Bushby K, Dalakas MC, Sambuughin N, Ferrer I, Goebel HH, Goldfarb LG. In-frame deletion in the seventh immunoglobulin-like repeat of filamin C in a family with myofibrillar myopathy. *Eur J Hum Genet*. 2009;17:656–63.

9. Avila-Smirnow D, Gueneau L, Batonnet-Pichon S, Delort F, B?cane H-M, Claeys K, Beuvin M, Goudeau B, Jais J-P, Nelson I, Richard P, Ben Yaou R, Romero NB, Wahbi K, Mathis S, Voit T, Furst D, van der Ven P, Gil R, Vicart P, Fardeau M, Bonne G, Behin A, Bécane H-M, Claeys K, Beuvin M, Goudeau B, Jais J-P, Nelson I, Richard P, Ben Yaou R, Romero NB, Wahbi K, Mathis S, Voit T, Furst D, van der Ven P, Gil R, Vicart P, Fardeau M, Bonne G, Behin A. Cardiac arrhythmia and late-onset muscle weakness caused by a myofibrillar myopathy with unusual histopathological features due to a novel missense mutation in FLNC. *Rev Neurol (Paris)*. 2016;172:594–606.

10. Brodehl A, Ferrier RA, Hamilton SJ, Greenway SC, Brundler MA, Yu W, Gibson WT, Mckinnon ML, Mcgillivray B, Alvarez N, Giuffre M, Schwartzentruber J, Gerull B, Boycott K, Friedman J, Michaud J, Bernier F, Brudno M, Fernandez B, Knoppers B, Samuels M, Scherer S. Mutations in FLNC are Associated with Familial Restrictive Cardiomyopathy. *Hum Mutat*. 2016;37:269–279.

11. Guergueltcheva V, Peeters K, Baets J, Ceuterick-de Groote C, Martin JJ, Suls A, De Vriendt E, Mihaylova V, Chamova T, Almeida-Souza L, Ydens E, Tzekov C, Hadjidekov G, Gospodinova M, Storm K, Reyniers E, Bichev S, van der Ven PFM, Fürst DO, Mitev V, Lochmüller H, Timmerman V, Tournev I, De Jonghe P, Jordanova A. Distal myopathy with upper limb predominance caused by filamin C haploinsufficiency. *Neurology*. 2011;77:2105–14.

12. Tasca G, Odgerel Z, Monforte M, Aurino S, Clarke NF, Waddell LB, Udd B, Ricci E, Goldfarb LG. Novel FLNC mutation in a patient with myofibrillar myopathy in combination with late-onset cerebellar ataxia. *Muscle and Nerve*. 2012;46:275–282.

13. Vorgerd M, van der Ven PFMM, Bruchertseifer V, Löwe T, Kley RA, Schröder R, Lochmüller H, Himmel M, Koehler K, Fürst DO, Huebner A. A mutation in the dimerization domain of filamin c causes a novel type of autosomal dominant myofibrillar myopathy. *Am J Hum Genet*. 2005;77:297–304.