Whole exome sequence reveals genetic profiles of primary cardiomyopathy and genotype-phenotype association in Chinese population

Supplement materials:

Table S1. The filtered 111 variants from 77 cardiomyopathy patients.

Table S2. Association analysis of the L/LP variants and clinical symptoms in DCM patients.

Table S3. Primer pairs of PCR and sanger sequence.

Table S4. Cardiomyopathy related gene panel.

Table S1.The filtered 111 variants from 77 cardiomyopathy patients

Sample_ID	Disease	Gene	Chr	Start	End	Ref	Alt	Genotype	Rs Number	ACMG	ACMG Evidence
D1	DCM	AKAP9	chr7	91641872	91641872	C	T	het	rs750350575	VUS	PP3+BP1+PM2, VUS
S3	DCM	FBN1	chr15	48829887	48829887	G	C	het	rs774754863	LP	PS1+PM1+PP3+PM2, LP
S3	DCM	TTN	chr2	179485032	179485032	C	T	het	.	VUS	PP3+BP1+PM2, VUS
S3	DCM	DES	chr2	220283470	220283470	G	T	het	rs201190593	VUS	PM2+PP2+PP3, VUS
S3	DCM	LMNA	chr1	156105724	156105726	GGA	-	het	.	VUS	PM1+PM4+PM2, VUS
B1	DCM	MYBPC3	chr11	47365175	47365175	G	A	het	rs778161908	VUS	PM5+PP3, VUS
B1	DCM	TTN	chr2	179514619	179514620	GC	AA	het	rs727503630	VUS	PM2+BP1, VUS
B6	DCM	TNNT2	chr1	201333463	201333463	C	T	het	rs730881101	P	PM1+PP2+PM2+PM5+PP3, P
B7	DCM	TTN	chr2	179410333	179410333	C	-	het	.	LP	PVS1+PM2, LP
B8	DCM	RBM20	chr10	112590912	112590912	G	A	het	rs563762318	VUS	BS1+PP3, VUS
B9	DCM	BAG3	chr10	121431912	121431912	G	T	het	.	VUS	PM5+PP3+PM2, VUS
B10	DCM	TNNI3K	chr1	74808579	74808579	G	C	het	.	VUS	PP3+PM2, VUS
B12	DCM	PLEC	chr8	144994565	144994565	C	T	het	rs373523333	VUS	PP3, VUS

B13	DCM	TTN	chr2	179643673	179643673	A	G	het	.	VUS	PP3+PM2+BP1, VUS
B14	DCM	TTN	chr2	179421655	179421655	C	A	het	.	VUS	PP3+PM2+BP1, VUS
B15	DCM	DGKD	chr2	234343502	234343502	C	T	het	rs578036955	VUS	PP3, VUS
B15	DCM	TCAP	chr17	37822311	37822316	ACTTCG	CCTTCA	het	.	VUS	PS1+PM2, VUS
B16	DCM	TTN	chr2	179404839	179404839	T	C	het	rs762387276	VUS	PP3+BP1, VUS
B17	DCM	MYPN	chr10	69970135	69970135	T	A	het	rs199585352	VUS	BS1+PP3, VUS (LB)
B18	DCM	TTN	chr2	179408636	179408636	C	T	het	rs200540781	VUS	BP1+PP3, VUS
B18	DCM	PLEC	chr8	145008532	145008532	C	T	het	rs199866423	VUS	PP3, VUS (LB)
C1-2	DCM	MYPN	chr10	69970183	69970183	C	T	het	rs142354704	VUS	BS1, VUS
C1-3	DCM	GSK3B	chr3	119585439	119585439	G	A	het	rs768507443	VUS	PP3, VUS
C1-4	DCM	CDH2	chr18	25565705	25565705	T	C	het	rs763345128	VUS	PM2, VUS
C1-5	DCM	MYH7	chr14	23894494	23894494	C	T	het	rs141414377	P	PM1+PP2+PM2+PM5+PP3, P
C1-5	DCM	TTN	chr2	179399634	179399634	C	T	het	rs72629782	VUS	PP3+BP1, VUS
C1-6	DCM	DTNA	chr18	32418720	32418720	G	-	het	.	LP	PVS1+PM2, LP
C1-7	DCM	LMNA	chr1	156105885	156105885	G	A	het	rs61672878	P	PM1+PP2+PM2+PM5+PP3, P

C1-7	DCM	LMNA	chr1	156106994	156106994	C	T	het	rs57318642	P	PM1+PP2+PM2+PM5, P
C2-1	DCM	DMD	chrX	31198539	31198539	C	T	hom	.	VUS	PP3+PM2, VUS
C2-2	DCM	LMNA	chr1	156100452	156100452	T	C	het	.	LP	PP3+PM1+PM2, LP
C2-2	DCM	COL1A1	chr17	48262919	48262919	C	A	het	rs367952133	VUS	PM2+PP2, VUS
C2-2	DCM	TRPM4	chr19	49671243	49671243	C	T	het	rs760925558	VUS	PM2, VUS
C2-2	DCM	AFF4	chr5	132228794	132228794	C	G	het	rs201192755	VUS	PP3, VUS
C2-3	DCM	TNNT2	chr1	201330409	201330409	C	T	het	.	VUS	PP3+PM2, VUS
C2-4	DCM	TNNT2	chr1	201333449	201333449	T	C	het	.	VUS	PM2+PP3, VUS
C2-9	DCM	FBN2	chr5	127686693	127686693	G	C	het	rs778473524	VUS	PM2+PP3, VUS
C3-1	DCM	CHD7	chr8	61750797	61750797	G	A	het	rs750258756	VUS	PP2+PP3, VUS
C3-3	DCM	BRAF	chr7	140476716	140476716	T	C	het	rs745783052	LP	PP3+PM2+PP2, LP
C3-3	DCM	RBM20	chr10	112590912	112590912	G	A	het	rs563762318	VUS	PP3+BS1, VUS
C3-5	DCM	TNNT2	chr1	201333449	201333449	T	C	het	.	VUS	PM2+PP3, VUS
C3-8	DCM	TTN	chr2	179587536	179587536	G	A	het	rs397517500	VUS	PP3+BP1, VUS
C3-8	DCM	LAMA2	chr6	129419391	129419391	C	G	het	rs756699354	VUS	PP3, VUS

C4-1	DCM	MYPN	chr10	69934389	69934389	G	T	het	rs181848049	VUS	PM2+BP1, VUS
C4-1	DCM	SCN5A	chr3	38647480	38647480	A	T	het	.	VUS	PP3+PM2, VUS
C4-2	DCM	DSC2	chr18	28671017	28671017	C	T	het	.	VUS	PP3+PM2, VUS
C4-2	DCM	DSC2	chr18	28671071	28671071	G	A	het	rs727504578	VUS	PP3+PM2+BP1, VUS
C5-3	DCM	TPM1	chr15	63354797	63354797	C	T	het	rs397516387	LP	PP3+PP5+PM1+PM2, LP
C5-4	DCM	JPH2	chr20	42788948	42788948	C	T	het	.	VUS	PP3+PM2, VUS
C5-5	DCM	MYH6	chr14	23857492	23857492	C	T	het	rs146172839	VUS	BS2+PP3, VUS
C5-7	DCM	MYH6	chr14	23869994	23869994	T	C	het	.	VUS	PP3+PM2, VUS
C5-10	DCM	LZTR1	chr22	21341857	21341857	A	G	het	rs200390894	VUS	PP2+PP3, VUS
C5-10	DCM	SOX17	chr8	55372117	55372118	CA	AT	het	.	VUS	PM2, VUS
C5-11	DCM	TTN	chr2	179641264	179641264	G	T	het	.	VUS	PM2+BP1, VUS
C5-12	DCM	MYBPC3	chr11	47364405	47364405	G	A	het	rs730880540	VUS	PM2+PP3, VUS
C5-14	DCM	LMNA	chr1	156100421	156100421	G	A	het	.	LP	PP3+PM1+PM2, LP
C5-15	DCM	CASZ1	chr1	10720540	10720540	C	T	het	.	VUS	PM2+BP1, VUS
C5-16	DCM	MYBPC3	chr11	47370005	47370005	C	A	het	rs768625454	VUS	PM2+PP3, VUS

C6-5	DCM	LMNA	chr1	156104627	156104627	C	-	het	.	LP	PVS1+PM2, LP
C6-11	DCM	JPH2	chr20	42788829	42788829	A	C	het	.	VUS	PM2+PP3, VUS
C6-16	DCM	GATA6	chr18	19751805	19751805	C	-	het	.	LP	PVS1+PM2, LP
C6-16	DCM	DSC2	chr18	28671017	28671017	C	T	het	.	VUS	PM2, VUS
C6-16	DCM	DSC2	chr18	28671071	28671071	G	A	het	rs727504578	VUS	PM2, VUS
C6-18	DCM	TTN	chr2	179421655	179421655	C	A	het	.	VUS	PP3+PM2+BP1, VUS
C6-18	DCM	MYPN	chr10	69926319	69926325	CAGGCCC	AAGGCCT	het	.	VUS	PM2, VUS
C2-5	HCM	CSRP3	chr11	19206571	19206571	G	A	het	rs376198883	VUS	PM2+PP3, VUS
C2-5	HCM	SCN5A	chr3	38674671	38674671	C	T	het	rs199473047	VUS	PP3, VUS
C2-7	HCM	MYBPC3	chr11	47359085	47359085	C	T	het	rs2856655	P	PM2+PM5+PP3+PM1+PP5, P
C2-7	HCM	LAMA2	chr6	129691055	129691055	C	T	het	rs773658276	VUS	PP3, VUS
C2-7	HCM	TTN	chr2	179430305	179430306	GG	AA	het	rs397517716	VUS	PM2+BP1, VUS
C2-8	HCM	COL1A1	chr17	48264049	48264049	C	T	het	rs148216434	VUS	PP2+PP3, VUS (LB)
C3-2	HCM	TTN	chr2	179451454	179451454	G	A	het	rs72646859	VUS	PP3, VUS
C3-4	HCM	TTN	chr2	179430305	179430306	GG	AA	het	rs397517716	VUS	PM2+BP1, VUS

C3-7	HCM	KLHL24	chr3	183382799	183382799	G	T	hom	.	VUS	PP3+PM2+BP1，VUS
S1	HCM	MYBPC3	chr11	47369414	47369414	C	-	het	.	LP	PVS1+PM2，LP
S5	HCM	MYH7	chr14	23896866	23896866	C	T	het	rs121913627	P	PP5+PM1+PP3+PM2，P
S6	HCM	MYBPC3	chr11	47364480	47364480	G	-	het	.	LP	PVS1+PM2，LP
C5-2	HCM	MYH7	chr14	23898214	23898214	G	A	het	rs121913625	P	PM1+PP2+PM2+PM5+PP3，P
C6-17	HCM	MYH7	chr14	23884861	23884861	G	A	het	rs121913650	P	PS4+PM2+PM5+PP3+PP2+PP5，P
B1	DCM	MYRF	chr11	61551814	61551814	A	G	het	.	LB	BP4+PM2，LB
B1	DCM	CHRM2	chr7	136700966	136700966	A	G	het	.	LB	BP1+BP4+PM2，LB
B3	DCM	JUP	chr17	39921271	39921271	G	A	het	rs200740462	LB	BP1+BP4，LB
B4	DCM	PLEC	chr8	144992912	144992912	C	T	het	rs369497741	B	BS1+BS2+BP6,B（BP1+BP4,LB）
B4	DCM	MYPN	chr10	69970183	69970183	C	T	het	rs142354704	LB	BS1+BP4，LB
B4	DCM	LDB3	chr10	88477741	88477741	T	G	het	rs566463138	LB	BS1+BP6+PP3，LB
B10	DCM	TTN	chr2	179594601	179594601	A	C	het	rs370812788	B	BS1+BS2+PP3，B
B15	DCM	PLEC	chr8	144997673	144997673	C	T	het	rs370942881	B	B
B16	DCM	TTN	chr2	179585717	179585717	C	T	het	rs367826445	LB	BS1+BP6+PP3，LB

C1-2	DCM	MYOT	chr5	137222565	137222565	T	A	het	rs78633961	LB	BS1+BP4, LB
C2-1	DCM	ACTN2	chr1	236918452	236918452	A	T	het	rs370862426	LB	BS1+BP6+PP3, LB
C2-2	DCM	DMD	chrX	31165612	31165612	C	T	hom	rs139547504	B	BS1+BS2+BP6,B
C2-4	DCM	UPF3B	chrX	118971949	118971949	C	T	hom	rs181034787	B	BS1+BS2+BP6,B
C3-1	DCM	TNNI3K	chr1	74901811	74901811	T	A	het	.	LB	BP4+PM2, LB
C3-3	DCM	TTN	chr2	179428511	179428511	A	G	het	.	LB	BP1+BP4+PM2, LB
C3-5	DCM	LDB3	chr10	88477741	88477741	T	G	het	rs566463138	LB	PP3+BS1+PP6, LB
C3-5	DCM	TCAP	chr17	37822319	37822319	G	A	het	rs780544264	LB	BP1+BP4+PM2, LB
C4-3	DCM	TTN	chr2	179404327	179404327	T	C	het	rs191054704	LB	PM2+BP4+BP1,LB
C4-3	DCM	TTN	chr2	179632870	179632870	T	A	het	rs727504501	LB	PM2+BP4+BP1,LB
C5-3	DCM	DSP	chr6	7556049	7556049	A	G	het	rs188516326	B	B
C5-6	DCM	MYPN	chr10	69970135	69970135	T	A	het	rs199585352	LB	BP4+BP6+PM2, LB
C5-6	DCM	SDHA	chr5	233687	233687	G	A	het	rs200526913	LB	BP4+PM2, LB
C5-7	DCM	PCSK9	chr1	55505558	55505558	-	CTG	het	.	LB	BP4+BP3+PM2, LB
C6-14	DCM	PKP2	chr12	33030850	33030850	C	T	het	rs200069860	B	BS1+BS2+PP3, B

C2-7	HCM	DSP	chr6	7556049	7556049	A	G	het	rs188516326	B	B
C2-7	HCM	TTN	chr2	179430305	179430305	G	A	het	rs185887755	LB	BS1+BP6+PP3，LB
C2-8	HCM	TBX1	chr22	19748523	19748525	CCG	-	het	.	LB	LB，BP4
C3-4	HCM	TTN	chr2	179430305	179430305	G	A	het	rs185887755	LB	BP4+BP1+PM2，LB
S1	HCM	MYH6	chr14	23856794	23856794	G	A	het	rs201827489	B	BS1+BS2，B
S1	HCM	FOXC1	chr6	1612017	1612017	-	CGG	het	rs398123612	B	B
S1	HCM	HCN4	chr15	73635946	73635946	G	A	het	rs370442588	LB	BS2+PP3，LB

Table S2. Association analysis of the L/LP variants and clinical symptoms in DCM patients [a].

Baseline characteristics	With P/LP(n=12)	Without P/LP(n=53)	P-Value [c]
Age at onset (years)[b]	32.36±13.97	30.00±18.55	0.69
Female sex, n (%) [c]	5(41.67%)	12(22.64%)	0.27
NYHA class III/IV, n (%)	9(75.00%)	43(81.13%)	0.69
Smoking history, n (%)	3(25.00%)	16(30.19%)	0.72
LVEDD, mm [b]	64.91±10.81	63.27±13.94	0.72
LVEF, n (%) [b]	28.55±8.36	29.63±10.73	0.76
RVD, mm [b]	35.09±6.36	34.38±7.12	0.76
IVSD, mm [b]	8.10±1.45	9.37±1.54	0.03
Ntpro-BNP>1000pg/ml, n (%)	6(50.00%)	12(22.64%)	0.07
Hyperuricemia [d]	6(50.00%)	33(62.26%)	0.52
Atrial fibrillation, n (%)	3(25.00%)	3(5.66%)	0.07
Left Bundle branch block, n (%)	1(8.33%)	10(18.87%)	0.67
History of radiofrequency ablation, n (%)	1(8.33%)	1(1.89%)	0.34
History of ICD, n (%)	3(25.00%)	5(9.62%)	0.16
History of heart transplantation, n (%)	2(16.67%)	5(9.62%)	0.62

[a] LVEDD, left ventricular end-diastolic dimension; LAD, left atrial dimension; LVEF, left ventricular ejection fraction; IVSD, Interventricular septum dimension.
[b] Values are given as means±SD.
[c] Calculated between subgroup using either unpaired t test for quantitative data, or Chi-square test or Fisher exact test for qualitative data.
[d] Defined as a level of fasting blood uric acid higher than 420 μmol/L in men and 360 μmol/L in women.

Table S3. Primer pairs of PCR and sanger sequence

Primer_ID	Forward	Reverse
D1-AKAP9	GCTACAGATGGAAGCCCAACGC	ACCTTCCTCTGTGGGTTCTATGAC
D2-FBN1	TGATCAGCAACCAGATGTGCC	TTGACAAGCTCCCGTGCGGATA
D2-TTN	GACAACCGTGTCTCATACAGAGTTGA	GTTAGCTTGCAATGATGGCCACT
D2-DES	GGTTTCGGCTCTAAGGGCTC	TGCTCCAGGAAGCGCACCTT
D2-LMNA	TCTCCTACACCGACCCACGT	ATGTCCAGAAGCTCCTGGTACT
D3-MYBPC3	CCCAGCCACAGCCACAGT	AGCGTCATGGTCAGCCAGT
D3-TTN	ACCCACAGAATTTGGTCCCACAT	AGTACCTTTAGGAGGCGGTGCTT
D4-TNNT2	ATGCAGGTTTCTGTACCTGCGAT	GCACCCGGCCAATATTGTCTCT
D5-TTN	GCTATTCCATCACCACCCGGCAT	CTGGTCACCTTCAGCCTGGTATCAT
D6-RBM20	GCCATCCTAACCCTGCGTGTCTAT	GGAGCTAGACTCAGAGGCAAGCA
D7-BAG3	AGCTGCCTCTGACTGCTCAT	CTCGATGCACCCTGGACAGAT
D8-TNNI3k	CAATTTCCAGGAATCACTCAGGCACT	ACAGGTGTAAGGGGGTATCTCCAT
D9-PLEC	TCTCCTCATCCAGGCAGCCT	CTACACAGGGCAGAGCGTCT
D10-TTN	GGTACAAAGATGGCAAGCGCAT	ACCTGATTCTGCTCACTGGCTCT
D11-TTN	CGTGACCTTCCAGATGGCAGATG	GCAAGTAATGGGCCCTACAACCT
D12-TCAP	AGGACACCCCCATCCAGCTT	TGGGCAAACTACAAAGCAGCCAT
D12-DGKD	CTTTGCCCTCTTGGTTTCGTTCT	TCAAGTAAAATGCCAGGCCACCT
D13-TTN	CAAGTGTAACACCACTCCAACCAAGAT	GATGACGCCACCTTGCCTTAC
D14-MYPN	CTGCAGCGTACATGCTGGACT	TGTTCTCATTCCCCCTCCACAGT
D15-TTN	CCTTGCGCTTGATGGTGTCTGT	AGGACAGGGACCAGGAGTATCTG
D16-PLEC	CCTGCCTATAGGGCCTGAGTG	TCCTAAGGCGAGTGGGAGCT
D17-MYPN	CAGTGGCAGTCGCTACGGAT	TGTTCTCATTCCCCCTCCACAGT
D18-CSK3B	GACTTGCTACTGTGAAGACTGATG	GAATCACCCAAGAGGCTGACAAT
D19-CDH2	AAGGGCCAGAAGGTTAGAAGCT	ATTGGGGTCTGGAGTTTCGCA
D20-MYH7	CTGGGGCTGCTGGAGGAAAT	CTGTGTTTGAAGATCTGCTGAGCTT
D20-TTN	ATCTTGGGCGTGGTGAGTTT	AGCTGACGGGCTTGACCAAAT
D21-DTNA	CTGGCATCCAGTGACCTTGCT	ACCCTTCCTTTACCTGCTTGGT
D22-LMNA1	CAGCCAAGGAGGCGAAGCTT	AGAGGACACTGCCAGCACCT
D22-LMNA2	AGCGCTGGGGTAAGTGTCCTTT	GCTCTAGAAAGGGGCCCTGAAT
D23-DMD	CACTTACCTTGTGGGACTAATGAAC	AGGCTGGCGTCAAACTTACC
D24-LMNA	CAAGGATGCCCTCTCCTGGTAATT	AGGTGAATGGCTCTGAAATCAGGT
D24-TRPM4	CGCTCCCATGTGTCCACAGTT	GTGACAATCCAGGCTCCTACACAG
D24-AFF4	AAAATAGGCGACCTCAGCCTT	ATGGCTTATGGCCTGCTGAATT

Primer_ID	Forward	Reverse
D24-COL1A1	CAGGCCATAGTGCCCTCTCT	TGGGATGGAGGGAGTTTACAGGA
D25-TNNT2	GGAGAAGGCCAAGGAGCTGT	GGTGGACATGGAACACTGCT
D26-TNNT2	ATGCAGGTTTCTGTACCTGCGAT	GCACCCGGCCAATATTGTCTCT
D27-FBN2	CAGGCGTGAGCCAGTAATCTTT	CTCCTACCTAGTTCACACCGCT
D28-CHD7	TGGATGAGGAGGATGAAGGGTCT	ATTCCCAATGCATCTTGTAAGCACT
D29-BRAF	GTGTGAGGGCTCCAGCTTGTAT	AAGGCTGCACTAGACAGAGACAT
D29-RBM20	CCATCCTAACCCTGCGTGTCTAT	CCAGACACCTGGCTCGGTTAT
D30-TNNT2	ATGCAGGTTTCTGTACCTGCGAT	GCACCCGGCCAATATTGTCTCT
D31-TTN	GAACCTCTGGAGGCAGCAGTT	ACTCTCCACTGTCATTGATGTTCAC
D31-LAMA2	GGTGTTCCAGATCGCGTATGTGATT	GGCCCAGTGCGGGGATAAATATT
D32-MYPN	ACCAACACCACCACCATTCACAT	GAATCCACAAAATGAGAGCCCTGTT
D32-SCN5A	ATCTTCCTGGGGTCCTTCTACCT	GCCCACAGGCACCAGCTTTA
D33-DSC2-1	CCTCCTGAGAGGAGAAAGATGTGTCA	TGGAAACTATAGACTCCCACAGCAGA
D33-DSC2-2	CCTCCTGAGAGGAGAAAGATGTGTCA	TGGAAACTATAGACTCCCACAGCAGA
D34-TPM1	GCCATTTCTCACAGAGGTGACTGA	AGCGTATCAATGTGGATGCCCTT
D35-JPH2	GGGACGTACCAAGGCCAGTT	CTGAGGTCGCTCTTAAGGAAGCT
D36-MYH6	CTCCAGTGTTGCCCAGTAGAGT	GAGCGCTCTACGTCCACCAT
D37-MYH6	GGTGAAAGTGGGCAACGAGTATGT	GTCCCAACTCACGTCGAAGATCT
D38-LZTR1	AAAAGAAAGGCAGCACAGGGAT	GCTGTACACAGCACGAGCACAAA
D39-TTN	GCTCCATGATGGAAAGCCACTT	CTCCCCTCAGGCAATTGGGAT
D40-MYBPC3	GCACGGAGCTCTTTGTGAAAGGT	AGATGCCCCCAACACCCAT
D41-LMNA	CAAGGATGCCCTCTCCTGGTAATT	AGGTGAATGGCTCTGAAATCAGGT
D42-CASZ1	ACACTGACCCCCTCCCTCTAT	CCGCTTGCTGAGGGTATCCT
D43-MYBPC3	CTCCATGCACACAGGTCTATCTGTT	GATGGGACGAGGCATCCTCCTT
D44-LMNA	TGGCCTCCCAGGAACTAAT	GAACCAATCGAGAGCAAGCA
D45-JPH2	CCAGTTCACCAACGGCAT	CGCTCTTAAGGAAGCTGACAC
D46-GATA4	GTCTACGTGCCCACCAC	GACAGCGAGCTGTACTGG
D46-DSC2-1	TCCTCCTGAGAGGAGAAAGATG	ACTATAGACTCCCACAGCAGAA
D46-DSC2-2	TCCTCCTGAGAGGAGAAAGATG	ACTATAGACTCCCACAGCAGAA
D47-TTN	GGCCAGCTTCACCAATGTTA	CCCAGCTAGTCCATAATTTCTTCC
D47-MYPN	TTCGTGTGCACTTCAACCT	TCACAGAAGCTCCTGGTTATTC
H11-MYH7	ATCCTCAGGACACCCAGATT	CAGGTCAGCATCCATCTTCTTC
H10-MYH7	AACCCTGCTCAATATGGGTCTCT	TGTCTGGTCCACAGCTGGCT

Primer_ID	Forward	Reverse
H9-MYBPC3	GCACGGAGCTCTTTGTGAAAGGT	AGATGCCCCCAACACCCAT
H8-MYH7	CCTGGGCAAATCCGCCAACTT	ACCTTGGCAGAATCCCTGCT
H7-MYBPC3	AAGGCTGGGAAGGTGAGATCCT	CTGCCTGGGAAGCTTGCTCT
H6-KLHL24	CAACAGCCGTGATGTCTGGATT	TGTGCTACACTGCCATGAATTCCA
H5-TTN	ATGGCGGTAGCAGAGTCCTG	AGGTCTTGGTCGGCCTATAACT
H4-TTN	GTTTTTCTTCTAGGTGAGCCGGAT	ATCTGCAGGCTGCTCTTCTTCT
H3-COL1A1	ACCTCAAGAGAAGGCTCACGAT	GGGAGCAGCCAGCACCATAT
H2-TTN	ATGGCGGTAGCAGAGTCCTG	AGGTCTTGGTCGGCCTATAACT
H2-MYBPC3	CTGGGTTCCAGACCAGAGCT	TACACCACGCCCTCGATCAT
H2-LAMA2	GAACACTATCACTGCAAGGCCAT	CTGGTCAGCAGCTCGTTCAT
H1-CSRP3-F	CGTGTCACTCCTCTGGATGCT	CTCCAGAATCACTCACCTTTGCAAT
H1-SCN5A-F	GCAACGTGAGGAGAGCCTGT	AGGTGGGTGGTAGTCACCTT

Table S4. Cardiomyopathy related gene panel.

	Gene	Associated phenotypes	Inheritance	ClinVar	HGMD
1	AARS2	Leukoencephalopathy, progressive, with ovarian failure, Combined oxidative phosphorylation deficiency 8	AR	19	31
2	ABCC6	Pseudoxanthoma elasticum	AR	352	377
3	ABCC9	Atrial fibrillation, Cantu syndrome, Dilated cardiomyopathy (DCM)	AD	27	46
4	ACAD9	Acyl-CoA dehydrogenase family, deficiency	AR	26	61
5	ACADVL	Acyl-CoA dehydrogenase, very long chain, deficiency	AR	119	282
6	ACTA1	Myopathy	AD/AR	68	212
7	ACTC1	Left ventricular noncompaction, Hypertrophic cardiomyopathy (HCM), Cardiomyopathy, restrictive, Atrial septal defect, Dilated cardiomyopathy (DCM)	AD	23	63
8	ACTN2	Hypertrophic cardiomyopathy (HCM), Dilated cardiomyopathy (DCM)	AD	11	44

	Gene	Associated phenotypes	Inheritance	ClinVar	HGMD
9	AGK	Sengers syndrome, Cataract 38	AR	18	27
10	AGL	Glycogen storage disease	AR	142	245
11	ALMS1	Alstr 枚 m syndrome	AR	197	302
12	ALPK3	Pediatric cardiomyopathy	AR	12	6
13	ANO5	Gnathodiaphyseal dysplasia, LGMD2L and distal MMD3 muscular dystrophies	AD/AR	64	121
14	APOA1	Amyloidosis, systemic nonneuronopathic, Hypoalphalipoproteinemia	AD/AR	28	71
15	BAG3	Dilated cardiomyopathy (DCM), Myopathy, myofibrillar	AD	39	62
16	BRAF	LEOPARD syndrome, Noonan syndrome, Cardiofaciocutaneous syndrome	AD	134	65

	Gene	Associated phenotypes	Inheritance	ClinVar	HGMD
17	CALR3	Cardiomyopathy, familial hypertrophic, 19	AD		3
18	CAPN3	Muscular dystrophy, limb-girdle, Eosinophilic myositis	AD/AR	184	437
19	CASQ2	Ventricular tachycardia, catecholaminergic, polymorphic	AR	24	34
20	CASZ1	Dilated cardiomyopathy (DCM), Ventricular septal defect	AD	3	2
21	CBL	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia	AD	24	43
22	CDH2	Arrhythmogenic right ventricular cardiomyopathy (ARVC), Neurodevelopmental disorder	AD	1	6
23	CHRM2	Dilated cardiomyopathy (DCM)	AD/AR		1
24	COX15	Leigh syndrome, Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency	AR	7	5

	Gene	Associated phenotypes	Inheritance	ClinVar	HGMD
25	CPT2	Carnitine palmitoyltransferase II deficiency	AR	72	111
26	CRYAB	Cataract, myofibrillar myopathy and cardiomyopathy, Congenital cataract and cardiomyopathy, Dilated cardiomyopathy (DCM), Myopathy, myofibrillar, Cataract 16, multiple types, Myopathy, myofibrillar, fatal infantile hypertonic, alpha-B crystallin-related	AD	14	28
27	CSRP3	Hypertrophic cardiomyopathy (HCM), Dilated cardiomyopathy (DCM)	AD	4	30
28	CTNNA3	Arrhythmogenic right ventricular dysplasia	AD	7	46
29	DBH	Dopamine beta-hydroxylase deficiency	AR	10	11
30	DES	Dilated cardiomyopathy (DCM), Myopathy, myofibrillar, Scapuloperoneal syndrome, neurogenic, Kaeser type	AD/AR	64	124
31	DMD	Becker muscular dystrophy, Duchenne muscular dystrophy, Dilated cardiomyopathy (DCM)	XL	832	3915

	Gene	Associated phenotypes	Inheritance	ClinVar	HGMD
32	DNAJC19	3-methylglutaconic aciduria	AR	3	6
33	DOLK	Congenital disorder of glycosylation	AR	8	11
34	DPM3	Congenital disorder of glycosylation, Dilated cardiomyopathy (DCM), Limb-girdle muscular dystrophy	AR	3	2
35	DSC2	Arrhythmogenic right ventricular dysplasia with palmoplantar keratoderma and woolly hair, Arrhythmogenic right ventricular dysplasia	AD/AR	32	87
36	DSG2	Arrhythmogenic right ventricular dysplasia, Dilated cardiomyopathy (DCM)	AD	44	129
37	DSP	Cardiomyopathy, dilated, with wooly hair, keratoderma, and tooth agenesis, Arrhythmogenic right ventricular dysplasia, familial, Cardiomyopathy, dilated, with wooly hair and keratoderma, Keratosis palmoplantaris striata II, Epidermolysis bullosa, lethal acantholytic	AD/AR	177	296
38	DTNA	Left ventricular noncompaction 1	AD	3	7

	Gene	Associated phenotypes	Inheritance	ClinVar	HGMD
39	DYSF	Miyoshi muscular dystrophy, Muscular dystrophy, limb-girdle, Myopathy, distal, with anterior tibial onset	AR	244	529
40	EEF1A2	Epileptic encephalopathy, early infantile, Mental retardation	AD	17	12
41	ELAC2	Combined oxidative phosphorylation deficiency 17	AR	11	15
42	EMD	Emery-Dreifuss muscular dystrophy	XL	48	113
43	EPG5	Vici syndrome	AR	36	66
44	ETFA	Glutaric aciduria, Multiple acyl-CoA dehydrogenase deficiency	AR	8	29
45	ETFB	Glutaric aciduria, Multiple acyl-CoA dehydrogenase deficiency	AR	6	15
46	ETFDH	Glutaric aciduria, Multiple acyl-CoA dehydrogenase deficiency	AR	43	190

	Gene	Associated phenotypes	Inheritance	ClinVar	HGMD
47	FBXL4	Mitochondrial DNA depletion syndrome	AR	55	47
48	FBXO32	Dilated cardiomyopathy (DCM)	AD/AR		2
49	FHL1	Myopathy with postural muscle atrophy, Emery-Dreifuss muscular dystrophy, Reducing bod myopathy	XL	26	62
50	FHOD3	Cardiomyopathy, familial hypertrophic	AD		1
51	FKRP	Muscular dystrophy-dystroglycanopathy	AR	66	140
52	FKTN	Muscular dystrophy-dystroglycanopathy, Dilated cardiomyopathy (DCM), Muscular dystrophy-dystroglycanopathy (limb-girdle)	AD/AR	45	58
53	FLNC*	Myopathy	AD	54	109
54	FOXD4	Dilated cardiomyopathy (DCM)	AD		1

	Gene	Associated phenotypes	Inheritance	ClinVar	HGMD
55	FOXRED1	Leigh syndrome, Mitochondrial complex I deficiency	AR	15	8
56	FXN	Friedreich ataxia	AR	13	63
57	GAA	Glycogen storage disease	AR	193	573
58	GATA4*	Tetralogy of Fallot, Atrioventricular septal defect, Testicular anomalies with or without congenital heart disease, Ventricular septal defect, Atrial septal defect	AD	37	140
59	GATA6	Heart defects, congenital, and other congenital anomalies, Atrial septal defect 9, atrioventricular septal defect 5, Persistent truncus arteriosus, Tetralogy of Fallot	AD	16	82
60	GATAD1	Dilated cardiomyopathy (DCM)	AR	31	1
61	GATC	Cardiomyopathy, fatal	AR	1	
62	GBE1	Glycogen storage disease	AR	36	70

	Gene	Associated phenotypes	Inheritance	ClinVar	HGMD
63	GFM1	Combined oxidative phosphorylation deficiency	AR	19	19
64	GLA	Fabry disease	XL	226	937
65	GLB1	GM1-gangliosidosis, Mucopolysaccharidosis (Morquio syndrome)	AR	90	220
66	GMPPB	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), Limb-girdle muscular dystrophy-dystroglycanopathy	AR	19	41
67	GSK3B	Hypertrophic cardiomyopathy, Dilated cardiomyopathy (DCM)		2	
68	GTPBP3	Combined oxidative phosphorylation deficiency 23	AR	14	15
69	GUSB	Mucopolysaccharidosis	AR	27	62
70	HADHA	Trifunctional protein deficiency, Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency	AR	65	71

	Gene	Associated phenotypes	Inheritance	ClinVar	HGMD
71	HAND1	Congenital heart defects, Dilated cardiomyopathy	AD		9
72	HCN4	Sick sinus syndrome, Brugada syndrome, Left ventricular non-compaction cardiomyopathy (LVNC)	AD	8	34
73	HFE	Hemochromatosis	AR/Digenic	11	56
74	HRAS	Costello syndrome, Congenital myopathy with excess of muscle spindles	AD	43	31
75	IDUA	Mucopolysaccharidosis	AR	105	282
76	ILK	Dilated cardiomyopathy (DCM)	AD		10
77	ISPD	Muscular dystrophy-dystroglycanopathy	AR	38	53
78	JPH2	Hypertrophic cardiomyopathy (HCM)	AD	3	13

	Gene	Associated phenotypes	Inheritance	ClinVar	HGMD
79	JUP	Arrhythmogenic right ventricular dysplasia, Naxos disease	AD/AR	8	46
80	KLHL24	Epidermolysis bullosa simplex, generalized, with scarring and hair loss, Dilated cardiomyopathy (DCM), Hypertrophic cardiomyopathy (HCM)	AD/AR	5	5
81	KRAS*	Noonan syndrome, Cardiofaciocutaneous syndrome	AD	63	35
82	LAMA2	Muscular dystrophy, congenital merosin-deficient	AR	199	301
83	LAMP2	Danon disease	XL	62	101
84	LARGE	Muscular dystrophy-dystroglycanopathy	AR	19	27
85	LDB3	Dilated cardiomyopathy (DCM), Myopathy, myofibrillar	AD	9	14
86	LEMD2	Cataract 46, juvenile onset, Arrhythmogenic right ventricular cardiomyopathy (ARVC), Dilated cardiomyopathy (DCM)	AR	1	1

	Gene	Associated phenotypes	Inheritance	ClinVar	HGMD
87	LMNA	Heart-hand syndrome, Slovenian, Limb-girdle muscular dystrophy, Muscular dystrophy, congenital, LMNA-related, Lipodystrophy (Dunnigan), Emery-Dreiffus muscular dystrophy, Malouf syndrome, Dilated cardiomyopathy (DCM), Mandibuloacral dysplasia type A, Progeria Hutchinson-Gilford type	AD/AR	250	564
88	LMOD2	Familial dilated cardiomyopathy	AR		
89	LRRC10	Dilated cardiomyopathy (DCM)	AD/AR		4
90	LZTR1	Schwannomatosis, Noonan syndrome	AD/AR	34	71
91	MAP2K1	Cardiofaciocutaneous syndrome	AD	45	23
92	MAP2K2	Cardiofaciocutaneous syndrome	AD	21	35
93	MAP3K8	Noonan syndrome	AD		1

	Gene	Associated phenotypes	Inheritance	ClinVar	HGMD
94	MIPEP	Combined oxidative phosphorylation deficiency 31	AR	5	8
95	MLYCD	Malonyl-CoA decarboxylase deficiency	AR	14	38
96	MT-ATP6	Neuropathy, ataxia, and retinitis pigmentosa, Leber hereditary optic neuropathy, Ataxia and polyneuropathy, adult-onset, Cardiomyopathy, infantile hypertrophic, Leigh syndrome, Striatonigral degeneration, infantile, mitochondrial	Mitochondrial	19	
97	MT-ATP8	Cardiomyopathy, apical hypertrophic, and neuropathy, Cardiomyopathy, infantile hypertrophic	Mitochondrial	4	
98	MT-CO1	Myoglobinuria, recurrent, Leber hereditary optic neuropathy, Sideroblastic anemia, Cytochrome C oxidase deficiency	Mitochondrial	17	
99	MT-CO2	Cytochrome c oxidase deficiency	Mitochondrial	8	
100	MT-CO3	Cytochrome c oxidase deficiency, Leber hereditary optic neuropathy	Mitochondrial	9	

	Gene	Associated phenotypes	Inheritance	ClinVar	HGMD
101	MT-CYB		Mitochondrial	69	
102	MT-ND1	Mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke-like episodes, Leber hereditary optic neuropathy, Leber optic atrophy and dystonia	Mitochondrial	21	
103	MT-ND2	Leber hereditary optic neuropathy, Mitochondrial complex I deficiency	Mitochondrial	6	
104	MT-ND3	Leber optic atrophy and dystonia, Mitochondrial complex I deficiency	Mitochondrial	7	
105	MT-ND4	Leber hereditary optic neuropathy, Leber optic atrophy and dystonia, Mitochondrial complex I deficiency	Mitochondrial	11	
106	MT-ND4L	Leber hereditary optic neuropathy	Mitochondrial	2	
107	MT-ND5	Myoclonic epilepsy with ragged red fibers, Mitochondrial myopathy, encephalopathy, Mitochondrial complex I deficiency	Mitochondrial	19	
108	MT-ND6	Mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke-like episodes, Oncocytoma, Leber hereditary optic neuropathy, Leber optic atrophy and dystonia, Mitochondrial complex I deficiency	Mitochondrial	16	

	Gene	Associated phenotypes	Inheritance	ClinVar	HGMD
109	MT-RNR1	Deafness, mitochondrial	Mitochondrial	3	
110	MT-RNR2	Chloramphenicol toxicity/resistance	Mitochondrial	2	
111	MT-TA		Mitochondrial	4	
112	MT-TC	Mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke-like episodes	Mitochondrial	3	
113	MT-TD		Mitochondrial	1	
114	MT-TE	Diabetes-deafness syndrome, Mitochondrial myopathy, infantile, transient, Mitochondrial myopathy with diabetes	Mitochondrial	5	
115	MT-TF	Myoclonic epilepsy with ragged red fibers, Nephropathy, tubulointerstitial, Encephalopathy, mitochondrial, Epilepsy, mitochondrial, Myopathy, mitochondrial, Mitochondrial encephalomyopathy with lactic acidosis and stroke-like episodes	Mitochondrial	7	

	Gene	Associated phenotypes	Inheritance	ClinVar	HGMD
116	MT-TG		Mitochondrial	3	
117	MT-TH		Mitochondrial	4	
118	MT-TI		Mitochondrial	7	
119	MT-TK		Mitochondrial	5	
120	MT-TL1	Cytochrome c oxidase deficiency, Myoclonic epilepsy with ragged red fibers, Mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke-like episodes, Diabetes-deafness syndrome, Cyclic vomiting syndrome, SIDS, susceptibility to	Mitochondrial	14	
121	MT-TL2	Mitochondrial multisystemic disorder, Progressive external ophthalmoplegia	Mitochondrial	5	
122	MT-TM	Leigh syndrome, Mitochondrial multisystemic disorder	Mitochondrial	1	
123	MT-TN	Progressive external ophthalmoplegia, Mitochondrial multisystemic disorder	Mitochondrial	3	

	Gene	Associated phenotypes	Inheritance	ClinVar	HGMD
124	MT-TP		Mitochondrial	2	
125	MT-TQ	Mitochondrial multisystemic disorder	Mitochondrial	2	
126	MT-TR	Encephalopathy, mitochondrial	Mitochondrial	2	
127	MT-TS1	Myoclonic epilepsy with ragged red fibers, Mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke-like episodes	Mitochondrial	10	
128	MT-TS2	Mitochondrial multisystemic disorder	Mitochondrial	2	
129	MT-TT		Mitochondrial	5	
130	MT-TV	Hypertrophic cardiomyopathy (HCM), Leigh syndrome, Mitochondrial multisystemic disorder	Mitochondrial	3	
131	MT-TW	Leigh syndrome, Myopathy, mitochondrial	Mitochondrial	8	

	Gene	Associated phenotypes	Inheritance	ClinVar	HGMD
132	MT-TY	Mitochondrial multisystemic disorder	Mitochondrial	4	
133	MTO1#	Combined oxidative phosphorylation deficiency	AR	16	24
134	MYBPC3	Left ventricular noncompaction, Hypertrophic cardiomyopathy (HCM), Dilated cardiomyopathy (DCM)	AD	482	1048
135	MYBPHL	Dilated cardiomyopathy (DCM)	AD		3
136	MYH6	Hypertrophic cardiomyopathy (HCM), Dilated cardiomyopathy (DCM), Atrial septal defect 3	AD	14	123
137	MYH7	Hypertrophic cardiomyopathy (HCM), Myopathy, myosin storage, Myopathy, distal, Dilated cardiomyopathy (DCM)	AD	305	986
138	MYL2	Hypertrophic cardiomyopathy (HCM), Infantile type I muscle fibre disease and cardiomyopathy	AD	21	67
139	MYL3	Hypertrophic cardiomyopathy (HCM)	AD/AR	12	41

	Gene	Associated phenotypes	Inheritance	ClinVar	HGMD
140	MYL4	Atrial fibrillation, familial, 18	AD	2	2
141	MYOT	Myopathy, myofibrillar, Muscular dystrophy, limb-girdle, 1A, Myopathy, spheroid body	AD	6	16
142	MYPN	Hypertrophic cardiomyopathy (HCM), Cardiomyopathy, restrictive, Dilated cardiomyopathy (DCM), Nemaline myopathy 11, autosomal recessive	AD	6	44
143	MYRF	Congenital heart malformations, Congenital abnormalities of the kidney and urinary tract	AD	1	1
144	NDUFAF2	Mitochondrial complex I deficiency, Leigh syndrome	AR	9	8
145	NEXN	Hypertrophic cardiomyopathy (HCM), Dilated cardiomyopathy (DCM)	AD	6	43
146	NF1	Watson syndrome, Neurofibromatosis, Neurofibromatosis-Noonan syndrome	AD	1157	2901
147	NKX2-5	Conotruncal heart malformations, Hypothyroidism, congenital nongoitrous,, Atrial septal defect, Ventricular septal defect 3, Conotruncal heart malformations, variable, Tetralogy of Fallot	AD	45	108

	Gene	Associated phenotypes	Inheritance	ClinVar	HGMD
148	NONO	Mental retardation, X-linked, syndrome 34, Left ventricular non-compaction cardiomyopathy (LVNC)	XL	10	4
149	NRAP	Dilated cardiomyopathy (DCM)	AR	1	6
150	NRAS	Noonan syndrome	AD	31	14
151	PCCA	Propionic acidemia	AR	66	125
152	PCCB	Propionic acidemia	AR	68	115
153	PKP2	Arrhythmogenic right ventricular dysplasia	AD	150	289
154	PLEC	Muscular dystrophy, limb-girdle, Epidermolysis bullosa	AD/AR	36	103
155	PLEKHM2	Dilated cardiomyopathy (DCM), left ventricular noncompaction	AR	1	1

	Gene	Associated phenotypes	Inheritance	ClinVar	HGMD
156	PLN	Hypertrophic cardiomyopathy (HCM), Dilated cardiomyopathy (DCM)	AD/AR	8	30
157	PNPLA2	Neutral lipid storage disease with myopathy	AR	13	35
158	PPA2	Sudden cardiac failure, infantile	AR	8	8
159	PPCS	Dilated cardiomyopathy (DCM)	AR		4
160	PPP1CB	Noonan syndrome-like disorder with loose anagen hair 2	AD	8	11
161	PRDM16	Left ventricular noncompaction, Dilated cardiomyopathy (DCM)	AD	17	20
162	PRKAG2	Hypertrophic cardiomyopathy (HCM), Wolff-Parkinson-White syndrome, Glycogen storage disease of heart, lethal congenital	AD	19	57
163	PTPN11	Noonan syndrome, Metachondromatosis	AD	135	140

	Gene	Associated phenotypes	Inheritance	ClinVar	HGMD
164	QRSL1	Mitochondrial multisystemic disorder	AR	4	2
165	RAF1	LEOPARD syndrome, Noonan syndrome, Dilated cardiomyopathy (DCM)	AD	45	53
166	RASA2	Noonan syndrome	AD	1	3
167	RBCK1	Polyglucosan body myopathy	AR	11	14
168	RBM20	Dilated cardiomyopathy (DCM)	AD	19	47
169	RIT1	Noonan syndrome	AD	23	26
170	RMND1	Combined oxidative phosphorylation deficiency	AR	17	15
171	RRAS	Noonan-syndrome like phenotype	AD/AR		2

	Gene	Associated phenotypes	Inheritance	ClinVar	HGMD
172	RYR2	Ventricular tachycardia, catecholaminergic polymorphic, Arrhythmogenic right ventricular dysplasia	AD	124	372
173	SCN5A	Heart block, nonprogressive, Heart block, progressive, Long QT syndrome, Ventricular fibrillation, Atrial fibrillation, Sick sinus syndrome, Brugada syndrome, Dilated cardiomyopathy (DCM)	AD/AR/Digenic	234	899
174	SCNN1B	Liddle syndrome, Pseudohypoaldosteronism, Bronchiectasis with or without elevated sweat chloride	AD/AR	19	47
175	SCNN1G	Liddle syndrome, Pseudohypoaldosteronism, Bronchiectasis with or without elevated sweat chloride	AD/AR	8	20
176	SCO1	Mitochondrial complex IV deficiency	AR	6	5
177	SCO2	Leigh syndrome, Hypertrophic cardiomyopathy (HCM), Cardioencephalomyopathy, fatal infantile, due to cytochrome c oxidase deficiency, Myopia	AR	42	37
178	SDHA	Leigh syndrome/Mitochondrial respiratory chain complex II deficiency, Gastrointestinal stromal tumor, Paragangliomas, Dilated cardiomyopathy (DCM), Cardiomyopathy, dilated, 1GG	AD/AR	54	87

	Gene	Associated phenotypes	Inheritance	ClinVar	HGMD
179	SELENON	Muscular dystrophy, rigid spine, Myopathy, congenital, with fiber-disproportion	AR	38	63
180	SGCA	Muscular dystrophy, limb-girdle	AR	60	100
181	SGCB	Muscular dystrophy, limb-girdle	AR	37	64
182	SGCD	Muscular dystrophy, limb-girdle, Dilated cardiomyopathy (DCM)	AR	21	27
183	SGCG	Muscular dystrophy, limb-girdle	AR	33	63
184	SHOC2	Noonan-like syndrome with loose anagen hair	AD	2	4
185	SLC22A5	Carnitine deficiency, systemic primary	AR	98	151
186	SLC25A20	Carnitine-acylcarnitine translocase deficiency	AR	15	42

	Gene	Associated phenotypes	Inheritance	ClinVar	HGMD
187	SLC25A4	Progressive external ophthalmoplegia with mitochondrial DNA deletions, Mitochondrial DNA depletion syndrome	AD/AR	12	14
188	SMCHD1	Facioscapulohumeral muscular dystrophy, Facioscapulohumeral muscular dystrophy, type 2	AD	51	79
189	SOS1	Noonan syndrome	AD	44	71
190	SOS2	Noonan syndrome 9	AD	4	6
191	SPEG	Centronuclear myopathy 5	AR	5	11
192	SPRED1	Legius syndrome	AD	38	71
193	TAB2	Congenital heart defects, multiple types, 2	AD	13	31
194	TAZ	3-Methylglutaconic aciduria, (Barth syndrome)	XL	45	158

	Gene	Associated phenotypes	Inheritance	ClinVar	HGMD
195	TBX20	Atrial septal defect 4	AD	4	28
196	TBX5	Holt-Oram syndrome	AD	61	127
197	TCAP	Muscular dystrophy, limb-girdle, Hypertrophic cardiomyopathy (HCM), Dilated cardiomyopathy (DCM)	AD/AR	12	28
198	TGFB3	Loeys-Dietz syndrome (Reinhoff syndrome), Arrhythmogenic right ventricular dysplasia	AD	19	26
199	TMEM43	Arrhythmogenic right ventricular dysplasia, Emery-Dreifuss muscular dystrophy	AD	4	24
200	TMEM70	Mitochondrial complex V (ATP synthase) deficiency	AR	12	18
201	TNNC1	Hypertrophic cardiomyopathy (HCM), Dilated cardiomyopathy (DCM)	AD	9	24
202	TNNI3	Hypertrophic cardiomyopathy (HCM), Cardiomyopathy, restrictive, Dilated cardiomyopathy (DCM)	AD/AR	56	129

	Gene	Associated phenotypes	Inheritance	ClinVar	HGMD
203	TNNI3K	Cardiac conduction disease with or without dilated cardiomyopathy	AD	1	3
204	TNNT2	Left ventricular noncompaction, Hypertrophic cardiomyopathy (HCM), Cardiomyopathy, restrictive, Dilated cardiomyopathy (DCM)	AD	61	148
205	TOR1AIP1	Muscular dystrophy with progressive weakness, distal contractures and rigid spine	AD/AR	3	5
206	TPM1	Hypertrophic cardiomyopathy (HCM), Dilated cardiomyopathy (DCM)	AD	34	98
207	TRIM32	Bardet-Biedl syndrome, Muscular dystrophy, limb-girdle	AR	13	16
208	TSFM	Combined oxidative phosphorylation deficiency	AR	6	6
209	TTN	Dilated cardiomyopathy (DCM), Tibial muscular dystrophy, Limb-girdle muscular dystrophy, Hereditary myopathy with early respiratory failure, Myopathy, early-onset, with fatal cardiomyopathy (Salih myopathy), Muscular dystrophy, limb-girdle, type 2J	AD	818	327

	Gene	Associated phenotypes	Inheritance	ClinVar	HGMD
210	TTR	Dystransthyretinemic hyperthyroxinemia, Amyloidosis, hereditary, transthyretin-related	AD	52	148
211	VCL	Hypertrophic cardiomyopathy (HCM), Dilated cardiomyopathy (DCM)	AD	8	30
212	VCP	Amyotrophic lateral sclerosis, Inclusion body myopathy with early-onset Paget disease, Charcot-Marie-Tooth disease	AD	17	61
213	VPS13A	Choreoacanthocytosis	AR	19	115
214	XK	McLeod syndrome	XL	10	41