

Supplementary Appendix

Left ventricular noncompaction and congenital heart disease increase risk of congestive heart failure

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LVNC study collaborates.

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Table S1. List of 182 analyzed genes of NGS.

Gene	Chromosome	NCBI Reference Sequence:	Sequence : (Start..End)	
<i>ABCC9</i>	12p12.1	NG_012819.1	NC_000012.11 (21950323..22094797, complement)	http://www.ncbi.nlm.nih.gov/gene/10060
<i>ACAD9</i>	3q21.3	NG_017064.1	NC_000003.12 (128879490..128913114)	https://www.ncbi.nlm.nih.gov/gene/28976
<i>ACADVL</i>	17p13.1	NG_007975.1	NC_000017.11 (7217125..7225267)	https://www.ncbi.nlm.nih.gov/gene/37
<i>ACTC1</i>	15q14	NG_007553.1	NC_000015.9 (35080297..35087927, complement)	http://www.ncbi.nlm.nih.gov/gene/70
<i>ACTN2</i>	1q42-q43	NG_009081.1	NC_000001.10 (236849754..236927931)	http://www.ncbi.nlm.nih.gov/gene/88
<i>ADAMTS1</i>	21q21.3	NC_000021.9	NC_000021.9 (26836287..26845409, complement)	https://www.ncbi.nlm.nih.gov/gene/9510
<i>ADAMTS9</i>	3p14.1	NC_000003.12	NC_000003.12 (64515654..64688000, complement)	https://www.ncbi.nlm.nih.gov/gene/56999
<i>ADCK3</i>	1q42.13	NG_012825.2	NC_000001.11 (226939339..226987545)	https://www.ncbi.nlm.nih.gov/gene/56997
<i>AKAP9</i>	7q21-q22	NG_011623.1	NC_000007.13 (91570181..91739987)	http://www.ncbi.nlm.nih.gov/gene/10142
<i>AMPD1</i>	1p13.2	NG_008012.1	NC_000001.11 (114673098..114695618, complement)	https://www.ncbi.nlm.nih.gov/gene/270
<i>ANK2</i>	4q25-q27	NG_009006.2	NC_000004.11 (113739239..114304896)	http://www.ncbi.nlm.nih.gov/gene/287
<i>ARFGEF2</i>	20q13.13	NG_011490.1	NC_000020.11 (48921721..49036693)	https://www.ncbi.nlm.nih.gov/gene/10564
<i>BAG3</i>	10q25.2-q26.2	NG_016125.1	NC_000010.10 (121410859..121437331)	http://www.ncbi.nlm.nih.gov/gene/9531
<i>BMP10</i>	2p13.3	NG_032117.1	NC_000002.12 (68860916..68871517, complement)	https://www.ncbi.nlm.nih.gov/gene/27302
<i>BMPR1A</i>	10q22.3	NG_009362.1	NC_000010.10 (88516396..88684945)	http://www.ncbi.nlm.nih.gov/gene/657
<i>BOLA3</i>	2p13.1	NG_031910.1	NC_000002.12 (74135401..74147912, complement)	https://www.ncbi.nlm.nih.gov/gene/388962
<i>BRAF</i>	7q34	NG_007873.3	NC_000007.14 (140719327..140924928, complement)	https://www.ncbi.nlm.nih.gov/gene/673
<i>C10orf2</i>	10q24.31	NG_012624.1	NC_000010.11 (100987527..100994403)	https://www.ncbi.nlm.nih.gov/gene/56652
<i>CACNA1C</i>	12p13.3	NG_008801.2	NC_000012.11 (2079952..2807115)	http://www.ncbi.nlm.nih.gov/gene/775
<i>CACNA2D1</i>	7q21.11	NC_000007.14	NC_000007.14 (81946444..82443806, complement)	https://www.ncbi.nlm.nih.gov/gene/781
<i>CACNB2</i>	10p12	NG_016195.1	NC_000010.10 (18429373..18830688)	http://www.ncbi.nlm.nih.gov/gene/783
<i>CALR3</i>	19p13.11	NG_031959.2	NC_000019.9 (16589767..16607015, complement)	http://www.ncbi.nlm.nih.gov/gene/125972
<i>CAPN3</i>	15q15.1	NG_008660.1	NC_000015.9 (42646545..42704515)	http://www.ncbi.nlm.nih.gov/gene/825
<i>CASQ2</i>	1p13.1	NG_008802.1	NC_000001.11 (115700003..115768805, complement)	https://www.ncbi.nlm.nih.gov/gene/845

<i>CASZ1</i>	1p36.22	NC_000001.11	NC_000001.11 (10636604..10796676, complement)	https://www.ncbi.nlm.nih.gov/gene/54897
<i>CAV3</i>	3p25	NG_008797.2	NC_000003.11 (8775486..8788451)	http://www.ncbi.nlm.nih.gov/gene/859
<i>CDKN1C</i>	11p15.4	NG_008022.1	NC_000011.10 (2883218..2885804, complement)	https://www.ncbi.nlm.nih.gov/gene/1028
<i>COL4A1</i>	13q34	NG_011544.1	NC_000013.10 (110801310..110959496, complement)	http://www.ncbi.nlm.nih.gov/gene/1282
<i>COL7A1</i>	3p21.31	NG_007065.1	NC_000003.12 (48564073..48595302, complement)	https://www.ncbi.nlm.nih.gov/gene/1294
<i>CPT2</i>	1p32.3	NG_008035.1	NC_000001.11 (53196429..53214197)	https://www.ncbi.nlm.nih.gov/gene/1376
<i>CSRP3</i>	11p15.1	NG_011932.2	NC_000011.10 (19182030..19210571, complement)	https://www.ncbi.nlm.nih.gov/gene/8048
<i>CTNNA3</i>	10q21.3	NG_034072.1	NC_000010.11 (65912518..67696217, complement)	https://www.ncbi.nlm.nih.gov/gene/29119
<i>DAAM1</i>	14q23.1	NG_047127.1	NC_000014.9 (59188657..59371405)	https://www.ncbi.nlm.nih.gov/gene/23002
<i>DAAM2</i>	6p21.2	NC_000006.12	NC_000006.12 (39792366..39904877)	https://www.ncbi.nlm.nih.gov/gene/23500
<i>DES</i>	2q35	NG_008043.1	NC_000002.11 (220283099..220291461)	http://www.ncbi.nlm.nih.gov/gene/1674
<i>DMD</i>	Xp21.2	NG_012232.1	NC_000023.10 (31137345..33357726, complement)	http://www.ncbi.nlm.nih.gov/gene/1756
<i>DMPK</i>	19q13.32	NG_009784.1	NC_000019.10 (45769709..45782557, complement)	https://www.ncbi.nlm.nih.gov/gene/1760
<i>DNAJC19</i>	3q26.33	NG_022933.1	NC_000003.12 (180983709..180989774, complement)	https://www.ncbi.nlm.nih.gov/gene/131118
<i>DSC2</i>	18q12.1	NG_008208.1	NC_000018.9 (28645938..28682388, complement)	http://www.ncbi.nlm.nih.gov/gene/1824
<i>DSG2</i>	18q12.1	NG_007072.3	NC_000018.9 (29078027..29128814)	http://www.ncbi.nlm.nih.gov/gene/1829
<i>DSP</i>	6p24	NG_008803.1	NC_000006.11 (7541808..7586946)	http://www.ncbi.nlm.nih.gov/gene/1832
<i>DTNA</i>	18q12.1	NG_009201.1	NC_000018.10 (34493290..34891844)	https://www.ncbi.nlm.nih.gov/gene/1837
<i>DVL1</i>	1p36.33	NG_008048.1	NC_000001.11 (1335278..1349142, complement)	https://www.ncbi.nlm.nih.gov/gene/1855
<i>EED</i>	11q14.2	NG_029595.1	NC_000011.10 (86244384..86285420)	https://www.ncbi.nlm.nih.gov/gene/8726
<i>ELN</i>	7q11.23	NG_009261.1	NC_000007.13 (73442119..73484237)	http://www.ncbi.nlm.nih.gov/gene/2006
<i>EMD</i>	Xq28	NG_008677.1	NC_000023.10 (153607597..153609883)	http://www.ncbi.nlm.nih.gov/gene/2010
<i>ERBB2</i>	17q12	NG_007503.1	NC_000017.11 (39688084..39728662)	https://www.ncbi.nlm.nih.gov/gene/2064
<i>ERBB4</i>	2q34	NG_011805.1	NC_000002.12 (211375717..212538628, complement)	https://www.ncbi.nlm.nih.gov/gene/2066
<i>EZH2</i>	7q36.1	NG_032043.1	NC_000007.14 (148807372..148884349, complement)	https://www.ncbi.nlm.nih.gov/gene/2146
<i>FBN2</i>	5q23.3	NG_008750.1	NC_000005.10 (128257909..128538042, complement)	https://www.ncbi.nlm.nih.gov/gene/2201

<i>FGF16</i>	Xq21.1	NG_034050.1	NC_000023.11 (77447675..77456522)	https://www.ncbi.nlm.nih.gov/gene/8823
<i>FGF9</i>	13q12.11	NG_016272.1	NC_000013.11 (21671076..21704501)	https://www.ncbi.nlm.nih.gov/gene/2254
<i>FGFR1</i>	8p11.23	NG_007729.1	NC_000008.11 (38411138..38468834, complement)	https://www.ncbi.nlm.nih.gov/gene/2260
<i>FGFR2</i>	10q26.13	NG_012449.2	NC_000010.11 (121478330..121598458, complement)	https://www.ncbi.nlm.nih.gov/gene/2263
<i>FKBP1A</i>	20p13	NC_000020.11	NC_000020.11 (1368977..1393172, complement)	https://www.ncbi.nlm.nih.gov/gene/2280
<i>FKBP1B</i>	2p23.3	NC_000002.12	NC_000002.12 (24033205..24067743)	https://www.ncbi.nlm.nih.gov/gene/2281
<i>FKTN</i>	9q31.2	NG_008754.1	NC_000009.12 (105558117..105655950)	https://www.ncbi.nlm.nih.gov/gene/2218
<i>FLNA</i>	Xq28	NG_011506.1	NC_000023.11 (154348532..154374638, complement)	https://www.ncbi.nlm.nih.gov/gene/2316
<i>FXN</i>	9q21.11	NG_008845.2	NC_000009.12 (69035563..69079077)	https://www.ncbi.nlm.nih.gov/gene/2395
<i>GAA</i>	17q25.2-q25.3	NG_009822.1	NC_000017.10 (78075339..78093680)	http://www.ncbi.nlm.nih.gov/gene/2548
<i>GATA4</i>	8p23.1-p22	NG_008177.1	NC_000008.10 (11534433..11617510)	http://www.ncbi.nlm.nih.gov/gene/2626
<i>GBE1</i>	3p12.2	NG_011810.1	NC_000003.12 (81489699..81761799, complement)	https://www.ncbi.nlm.nih.gov/gene/2632
<i>GFRA1</i>	10q25.3	NG_050620.1	NC_000010.11 (116056925..116273645, complement)	https://www.ncbi.nlm.nih.gov/gene/2674
<i>GFRA2</i>	8p21.3	NG_029215.1	NC_000008.11 (21690403..21789296, complement)	https://www.ncbi.nlm.nih.gov/gene/2675
<i>GLA</i>	Xq22	NG_007119.1	NC_000023.10 (100652779..100663001, complement)	http://www.ncbi.nlm.nih.gov/gene/2717
<i>GPD1L</i>	3p22.3	NG_023375.1	NC_000003.11 (32148003..32210207)	http://www.ncbi.nlm.nih.gov/gene/23171
<i>HADHA</i>	2p23.3	NG_007121.1	NC_000002.12 (26190635..26244726, complement)	https://www.ncbi.nlm.nih.gov/gene/3030
<i>HAS2</i>	8q24.13	NC_000008.11	NC_000008.11 (121613031..121641390, complement)	https://www.ncbi.nlm.nih.gov/gene/3037
<i>HBB</i>	11p15.4	NG_059281.1	NC_000011.10 (5225466..5227071, complement)	https://www.ncbi.nlm.nih.gov/gene/3043
<i>HCCS</i>	Xp22.2	NG_016460.1	NC_000023.11 (11111286..11123086)	https://www.ncbi.nlm.nih.gov/gene/3052
<i>HCN4</i>	15q24.1	NG_009063.1	NC_000015.9 (73612200..73661605, complement)	http://www.ncbi.nlm.nih.gov/gene/10021
<i>HEY2</i>	6q22.31	NC_000006.12	NC_000006.12 (125747639..125762243)	https://www.ncbi.nlm.nih.gov/gene/23493
<i>HMGCL</i>	1p36.11	NG_013061.1	NC_000001.11 (23801877..23825459, complement)	https://www.ncbi.nlm.nih.gov/gene/3155
<i>ITGA7</i>	12q13.2	NG_012343.1	NC_000012.12 (55684568..55716037, complement)	https://www.ncbi.nlm.nih.gov/gene/3679
<i>JARID2</i>	6p22.3	NC_000006.12	NC_000006.12 (15245975..15522042)	https://www.ncbi.nlm.nih.gov/gene/3720
<i>JUP</i>	17q21	NG_009090.2	NC_000017.10 (39910859..39942964, complement)	http://www.ncbi.nlm.nih.gov/gene/3728

<i>KCNE1</i>	21q22.12	NG_009091.1	NC_000021.8 (35790910..35884573, complement)	http://www.ncbi.nlm.nih.gov/gene/3753
<i>KCNE2</i>	21q22.12	NG_008804.1	NC_000021.8 (35736323..35743440)	http://www.ncbi.nlm.nih.gov/gene/9992
<i>KCNE3</i>	11q13.4	NG_011833.1	NC_000011.9 (74165886..74178600, complement)	http://www.ncbi.nlm.nih.gov/gene/10008
<i>KCNH2</i>	7q36.1	NG_008916.1	NC_000007.13 (150642044..150675402, complement)	http://www.ncbi.nlm.nih.gov/gene/3757
<i>KCNJ2</i>	17q24.3	NG_008798.1	NC_000017.10 (68164757..68176189)	http://www.ncbi.nlm.nih.gov/gene/3759
<i>KCNQ1</i>	11p15.5	NG_008935.1	NC_000011.9 (2466221..2870340)	http://www.ncbi.nlm.nih.gov/gene/3784
<i>KRAS</i>	12p12.1	NG_007524.1	NC_000012.11 (25358180..25403870, complement)	http://www.ncbi.nlm.nih.gov/gene/3845
<i>LAMP2</i>	Xq24	NG_007995.1	NC_000023.10 (119560003..119603204, complement)	http://www.ncbi.nlm.nih.gov/gene/3920
<i>LDB3</i>	10q22.3-q23.2	NG_008876.1	NC_000010.10 (88426542..88495829)	http://www.ncbi.nlm.nih.gov/gene/11155
<i>LMNA</i>	1q22	NG_008692.2	NC_000001.10 (156052369..156109880)	http://www.ncbi.nlm.nih.gov/gene/4000
<i>LMX1B</i>	9q33.3	NG_017039.1	NC_000009.12 (126614443..126701032)	https://www.ncbi.nlm.nih.gov/gene/4010
<i>LRPPRC</i>	2p21	NG_008247.1	NC_000002.12 (43886224..43996005, complement)	https://www.ncbi.nlm.nih.gov/gene/10128
<i>MADD</i>	11p11.2	NG_029462.1	NC_000011.10 (47269376..47330031)	https://www.ncbi.nlm.nih.gov/gene/8567
<i>MBL2</i>	10q21.1	NG_008196.1	NC_000010.11 (52764977..52772847, complement)	https://www.ncbi.nlm.nih.gov/gene/4153
<i>MED1</i>	17q12	NG_046996.1	NC_000017.11 (39404285..39451281, complement)	https://www.ncbi.nlm.nih.gov/gene/5469
<i>MEST</i>	7q32.2	NG_009226.1	NC_000007.14 (130486175..130506297)	https://www.ncbi.nlm.nih.gov/gene/4232
<i>MIB1</i>	18q11.2	NG_033272.2	NC_000018.10 (21740793..21870957)	https://www.ncbi.nlm.nih.gov/gene/57534
<i>MIPEP</i>	13q12.12	NG_052977.1	NC_000013.11 (23730189..23889448, complement)	https://www.ncbi.nlm.nih.gov/gene/4285
<i>MLYCD</i>	16q23.3	NG_009079.1	NC_000016.10 (83899125..83916182)	https://www.ncbi.nlm.nih.gov/gene/23417
<i>MMACHC</i>	1p34.1	NG_013378.1	NC_000001.11 (45500184..45511266)	https://www.ncbi.nlm.nih.gov/gene/25974
<i>MRPS22</i>	3q23	NG_012174.1	NC_000003.12 (139344014..139357129)	https://www.ncbi.nlm.nih.gov/gene/56945
<i>MTO1</i>	6q13	NG_032856.1	NC_000006.12 (73461731..73501456)	https://www.ncbi.nlm.nih.gov/gene/25821
<i>MYBPC3</i>	11p11.2	NG_007667.1	NC_000011.9 (47352957..47374253, complement)	http://www.ncbi.nlm.nih.gov/gene/4607
<i>MYCN</i>	2p24.3	NG_007457.1	NC_000002.12 (15940438..15947007)	https://www.ncbi.nlm.nih.gov/gene/4613
<i>MYH11</i>	16p13.11	NG_009299.1	NC_000016.9 (15796992..15950887, complement)	http://www.ncbi.nlm.nih.gov/gene/4629
<i>MYH6</i>	14q12	NG_023444.1	NC_000014.8 (23849942..23878836, complement)	http://www.ncbi.nlm.nih.gov/gene/4624
<i>MYH7</i>	14q12	NG_007884.1	NC_000014.8 (23881947..23904870, complement)	http://www.ncbi.nlm.nih.gov/gene/4625
<i>MYH7B</i>	20q11.22	NG_016984.2	NC_000020.11 (34955835..35002437)	https://www.ncbi.nlm.nih.gov/gene/57644

<i>MYL2</i>	12q24.11	NG_007554.1	NC_000012.11 (111348623..111358404, complement)	http://www.ncbi.nlm.nih.gov/gene/4633
<i>MYL3</i>	3p21.3-p21.2	NG_007555.2	NC_000003.11 (46899357..46904973, complement)	http://www.ncbi.nlm.nih.gov/gene/4634
<i>MYLK</i>	3q21	NG_029111.1	NC_000003.11 (123331143..123603149, complement)	http://www.ncbi.nlm.nih.gov/gene/4638
<i>MYOZ2</i>	4q26-q27	NG_029747.1	NC_000004.11 (120056939..120108944)	http://www.ncbi.nlm.nih.gov/gene/51778
<i>NEXN</i>	1p31.1	NG_016625.1	NC_000001.11 (7788515..77948643)	https://www.ncbi.nlm.nih.gov/gene/91624
<i>NFATC1</i>	18q23	NG_029226.1	NC_000018.10 (79395772..79529323)	https://www.ncbi.nlm.nih.gov/gene/4772
<i>NKX2-5</i>	5q34	NG_013340.1	NC_000005.9 (172659107..172662315, complement)	http://www.ncbi.nlm.nih.gov/gene/1482
<i>NNT</i>	5p12	NG_032869.1	NC_000005.10 (43601092..43705566)	https://www.ncbi.nlm.nih.gov/gene/23530
<i>NR0B1</i>	Xp21.2	NG_009814.1	NC_000023.11 (30304422..30309378, complement)	https://www.ncbi.nlm.nih.gov/gene/190
<i>NRAS</i>	1p13.2	NG_007572.1	NC_000001.10 (115247085..115259515, complement)	http://www.ncbi.nlm.nih.gov/gene/4893
<i>NRG1</i>	8p12	NG_012005.2	NC_000008.11 (31639222..32771716)	https://www.ncbi.nlm.nih.gov/gene/3084
<i>NSD1</i>	5q35.3	NG_009821.1	NC_000005.10 (177131835..177300213)	https://www.ncbi.nlm.nih.gov/gene/64324
<i>NUMB</i>	14q24.2-q24.3	NG_029061.2	NC_000014.9 (73275210..73458580, complement)	https://www.ncbi.nlm.nih.gov/gene/8650
<i>NUMBL</i>	19q13.2	NC_000019.10	NC_000019.10 (40665905..40690658, complement)	https://www.ncbi.nlm.nih.gov/gene/9253
<i>PKP2</i>	12p11	NG_009000.1	NC_000012.11 (32943680..33049780, complement)	http://www.ncbi.nlm.nih.gov/gene/5318
<i>PLEC</i>	8q24.3	NG_012492.1	NC_000008.11 (143915147..143976800, complement)	https://www.ncbi.nlm.nih.gov/gene/5339
<i>PLEKHM2</i>	1p36.21	NG_053033.1	NC_000001.11 (15681506..15734769)	https://www.ncbi.nlm.nih.gov/gene/23207
<i>PLN</i>	6q22.1	NG_009082.1	NC_000006.11 (118869442..118881587)	http://www.ncbi.nlm.nih.gov/gene/5350
<i>PMP22</i>	17p12	NG_007949.1	NC_000017.11 (15229777..15265373, complement)	https://www.ncbi.nlm.nih.gov/gene/5376
<i>POLG</i>	15q26.1	NG_008218.2	NC_000015.10 (89316305..89334795, complement)	https://www.ncbi.nlm.nih.gov/gene/5428
<i>PRDM16</i>	1p36.32	NG_029576.1	NC_000001.11 (3068227..3438621)	https://www.ncbi.nlm.nih.gov/gene/63976
<i>PRKAG2</i>	7q36.1	NG_007486.1	NC_000007.13 (151253200..151574316, complement)	http://www.ncbi.nlm.nih.gov/gene/51422
<i>PTGS2</i>	1q31.1	NG_028206.2	NC_000001.11 (186671812..186680427, complement)	https://www.ncbi.nlm.nih.gov/gene/5743
<i>PTK2</i>	8q24.3	NG_029467.1	NC_000008.11 (140658382..141002079, complement)	https://www.ncbi.nlm.nih.gov/gene/5747

<i>PTPN11</i>	12q24	NG_007459.1	NC_000012.11 (112856536..112947717)	http://www.ncbi.nlm.nih.gov/gene/5781
<i>RAD54L2</i>	3p21.2	NC_000003.12	NC_000003.12 (51538683..51668660)	https://www.ncbi.nlm.nih.gov/gene/23132
<i>RAF1</i>	3p25	NG_007467.1	NC_000003.11 (12625100..12705700, complement)	http://www.ncbi.nlm.nih.gov/gene/5894
<i>RANGRF</i>	17p13.1	NG_028189.1	NC_000017.11 (8288497..8290092)	https://www.ncbi.nlm.nih.gov/gene/29098
<i>RBM20</i>	10q25.2	NG_021177.1	NC_000010.11 (110641933..110839471)	https://www.ncbi.nlm.nih.gov/gene/282996
<i>RIT1</i>	1q22	NG_033885.1	NC_000001.11 (155897808..155911402, complement)	https://www.ncbi.nlm.nih.gov/gene/6016
<i>RPS6KA3</i>	Xp22.12	NG_007488.1	NC_000023.11 (20149911..20267514, complement)	https://www.ncbi.nlm.nih.gov/gene/6197
<i>RPS7</i>	2p25	NG_011744.1	NC_000002.11 (3622853..3628509)	http://www.ncbi.nlm.nih.gov/gene/6201
<i>RYR1</i>	19q13.2	NG_008866.1	NC_000019.10 (38433700..38587564)	https://www.ncbi.nlm.nih.gov/gene/6261
<i>RYR2</i>	1q43	NG_008799.2	NC_000001.10 (237205510..237997288)	http://www.ncbi.nlm.nih.gov/gene/6262
<i>SCN1B</i>	9q13.1	NG_013359.1	NC_000019.9 (35521555..35531353)	http://www.ncbi.nlm.nih.gov/gene/6324
<i>SCN3B</i>	11q23.3	NG_016283.1	NC_000011.9 (123499895..123525315, complement)	http://www.ncbi.nlm.nih.gov/gene/55800
<i>SCN4B</i>	11q23.3	NG_011710.1	NC_000011.9 (118004092..118023630, complement)	http://www.ncbi.nlm.nih.gov/gene/6330
<i>SCN5A</i>	3p21	NG_008934.1	NC_000003.11 (38589553..38691164, complement)	http://www.ncbi.nlm.nih.gov/gene/6331
<i>SCO2</i>	22q13.33	NG_016235.1	NC_000022.11 (50523568..50526439, complement)	https://www.ncbi.nlm.nih.gov/gene/9997
<i>SCRIB</i>	8q24.3	NG_030583.1	NC_000008.11 (143790920..143815379, complement)	https://www.ncbi.nlm.nih.gov/gene/23513
<i>SDHA</i>	5p15.33	NG_012339.1	NC_000005.10 (218223..264816)	https://www.ncbi.nlm.nih.gov/gene/6389
<i>SDHAF1</i>	19q13.12	NG_016869.1	NC_000019.10 (35995188..35996318)	https://www.ncbi.nlm.nih.gov/gene/644096
<i>SDHB</i>	1p36.13	NG_012340.1	NC_000001.11 (17018722..17054170, complement)	https://www.ncbi.nlm.nih.gov/gene/6390
<i>SDS</i>	12q24.13	NC_000012.12	NC_000012.12 (113392445..113403887, complement)	https://www.ncbi.nlm.nih.gov/gene/10993
<i>SGCD</i>	5q33-q34	NG_008693.2	NC_000005.9 (155462147..156194799)	http://www.ncbi.nlm.nih.gov/gene/6444
<i>SLC22A5</i>	5q31.1	NG_008982.2	NC_000005.10 (132369704..132395614)	https://www.ncbi.nlm.nih.gov/gene/6584
<i>SLC25A20</i>	3p21.31	NG_008171.1	NC_000003.12 (48856923..48898993, complement)	https://www.ncbi.nlm.nih.gov/gene/788
<i>SLC25A4</i>	4q35	NG_013001.1	NC_000004.11 (186064417..186071538)	http://www.ncbi.nlm.nih.gov/gene/291
<i>SLC25A4</i>	4q35.1	NG_013001.1	NC_000004.12 (185143263..185150384)	https://www.ncbi.nlm.nih.gov/gene/291
<i>SLC25A5</i>	Xq24	NG_013262.1	NC_000023.11 (119468400..119471396)	https://www.ncbi.nlm.nih.gov/gene/292
<i>SLC52A2</i>	8q24.3	NG_032872.1	NC_000008.11 (144358547..144361286)	https://www.ncbi.nlm.nih.gov/gene/79581
<i>SMAD3</i>	15q22.33	NG_011990.1	NC_000015.9 (67358036..67487533)	http://www.ncbi.nlm.nih.gov/gene/4088

<i>SMAD7</i>	18q21.1	NG_023330.1	NC_000018.10 (48919853..48950711, complement)	https://www.ncbi.nlm.nih.gov/gene/4092
<i>SMARCA4</i>	19p13.2	NG_011556.2	NC_000019.10 (10960922..11062282)	https://www.ncbi.nlm.nih.gov/gene/6597
<i>SNTA1</i>	20q11.2	NG_011622.1	NC_000020.10 (31995763..32031698, complement)	http://www.ncbi.nlm.nih.gov/gene/6640
<i>SOS1</i>	2p21	NG_007530.1	NC_000002.11 (39208690..39347686, complement)	http://www.ncbi.nlm.nih.gov/gene/6654
<i>SQSTM1</i>	5q35.3	NG_011342.1	NC_000005.10 (179806388..179838078)	https://www.ncbi.nlm.nih.gov/gene/8878
<i>STARD3</i>	17q11-q12		NC_000017.10 (37793333..37820454)	http://www.ncbi.nlm.nih.gov/gene/10948
<i>TAZ</i>	Xq28	NG_009634.1	NC_000023.10 (153639877..153650065)	http://www.ncbi.nlm.nih.gov/gene/6901
<i>TBX20</i>	7p14.2	NG_015805.1	NC_000007.14 (35199936..35254100, complement)	https://www.ncbi.nlm.nih.gov/gene/57057
<i>TBX5</i>	12q24.1	NG_007373.1	NC_000012.11 (114791735..114846247, complement)	http://www.ncbi.nlm.nih.gov/gene/6910
<i>TCAP</i>	17q12	NG_008892.1	NC_000017.11 (39665346..39666554)	https://www.ncbi.nlm.nih.gov/gene/8557
<i>TGFB3</i>	14q24.3	NG_011715.1	NC_000014.9 (75958061..75983011, complement)	https://www.ncbi.nlm.nih.gov/gene/7043
<i>TGFBR1</i>	9q22	NG_007461.1	NC_000009.11 (101867412..101916474)	http://www.ncbi.nlm.nih.gov/gene/7046
<i>TGFBR2</i>	3p22	NG_007490.1	NC_000003.11 (30647994..30735634)	http://www.ncbi.nlm.nih.gov/gene/7048
<i>TMEM43</i>	3p25.1	NG_008975.1	NC_000003.11 (14166440..14185180)	http://www.ncbi.nlm.nih.gov/gene/79188
<i>TMEM70</i>	8q21.11	NG_016618.1	NC_000008.11 (73976142..73982783)	https://www.ncbi.nlm.nih.gov/gene/54968
<i>TNNC1</i>	3p21.1	NG_008963.1	NC_000003.11 (52485107..52488057, complement)	http://www.ncbi.nlm.nih.gov/gene/7134
<i>TNNI3</i>	19q13.4	NG_007866.2	NC_000019.9 (55663135..55669100, complement)	http://www.ncbi.nlm.nih.gov/gene/7137
<i>TNNT2</i>	1q32	NG_007556.1	NC_000001.10 (201328136..201346836, complement)	http://www.ncbi.nlm.nih.gov/gene/7139
<i>TPM1</i>	15q22.1	NG_007557.1	NC_000015.9 (63334838..63364114)	http://www.ncbi.nlm.nih.gov/gene/7168
<i>TTN</i>	2q31.2	NG_011618.3	NC_000002.12 (178525989..178807423, complement)	https://www.ncbi.nlm.nih.gov/gene/7273
<i>TTR</i>	18q12.1	NG_009490.1	NC_000018.10 (31591767..31599024)	https://www.ncbi.nlm.nih.gov/gene/7276
<i>VANGL2</i>	1q23.2	NG_023420.1	NC_000001.11 (160400574..160428678)	https://www.ncbi.nlm.nih.gov/gene/57216
<i>VCL</i>	10q22.2	NG_008868.1	NC_000010.10 (75757836..75879918)	http://www.ncbi.nlm.nih.gov/gene/7414
<i>YAP1</i>	11q22.1	NG_029530.1	NC_000011.10 (102109957..102233423)	https://www.ncbi.nlm.nih.gov/gene/10413
<i>YWHAE</i>	17p13.3	NG_009233.1	NC_000017.11 (1344539..1400262, complement)	https://www.ncbi.nlm.nih.gov/gene/7531
<i>ZNF25</i>	10p11.21	NC_000010.11	NC_000010.11 (37949572..37976655, complement)	https://www.ncbi.nlm.nih.gov/gene/219749

Table S2. Silico predictive algorithms used in the study.

Category	Basis	Name	Website	Prediction Threshold
Missense prediction	Evolutionary conservation	FATHMM	http://fathmm.biocompute.org.uk	<-1.5 Damaging >-1.5 Tolerated
		SIFT	http://sift.jcvi.org	<0.05 Deleterious >0.05 Tolerated
Missense prediction	Protein structure/function and evolutionary conservation	Align GVGD	http://agvgd.iarc.fr/agvgd_input.php	≧ C15 Probably Damaging
		Mutation Taster	http://www.mutationtaster.org	Disease causing
		Polyphen-2	http://genetics.bwh.harvard.edu/pph2	0.85 to 1 Probably Damage 0.15 to 0.85 Possibly Damage
Missense and insertion/deletions prediction	Alignment and measurement of similarity between variant sequence and protein sequence homolog	PROVEAN	http://provean.jcvi.org/index.php	<-2.5 Deleterious >-2.5 Neutral
Missense and insertion/deletions prediction	Contrasts annotations of fixed/nearly fixed derived alleles in humans with simulated variants	CADD	http://cadd.gs.washington.edu	≧ 20 1% most deleterious ≧ 30 0.1% most deleterious

Reference

1. Richards S, Aziz N, Bale S, Bick D, Das S, Gastier-Foster J, Grody WW, Hegde M, Lyon E, Spector E, Voelkerding K, Rehm HL. Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the association for molecular autopsy. Genet Med 2015;17:405–423.

Table S3. List of patients with extracardiac anomaly.

Disease	Number of patients
Motor development delay	18
Mental retardation	15
Leigh encephalopathy	2
Odd looking face	2
Blepharophimosis, micrognathia	1
Corpus callosum hypoplasia	1
Corpus callosum defect, primary vitreous hyperplasia remnant	1
Deafness, umbilical hernia, high arched palate, muscular hypertonia	1
Eye corneal opacity, cornea rear deficit, small eyeballs	1
Forehead protruding	1
Forehead protruding, micrognathia	1
Forehead protruding, high arched palate	1
Glycogen disease	1
High arched palate, arthrogryposis, muscular hypotonia	1
Macrocephaly	1
Microcephaly	1
Mitochondrial disease	1
Muscular hypotonia, epilepsy	1
Labia majora hypertrophy, corneal opacity	1
Nephrotic syndrome, macrocephaly	1
Polycystic kidney	1
Proptosis, low set ear, high arched palate	1
Smooth encephalopathy	1
Slanted eyebrows, slanted palpebral fissure	1
Symphalangism, pyloric stenosis	1

Table S4. Gene collapsing test of rare variants.

Rank	Gene	Qualifying Cases	Frequency Qualifying Cases (n=53)	Qualifying Controls Cases	Frequency Qualifying Controls (n=4456)	Fisher's Exact Test p-value
1	<i>MYH7</i>	8	0.1509	0	0	2.104×10 ⁻¹⁶
2	<i>TPM1</i>	2	0.0377	0	0	1.356×10 ⁻⁴
3	<i>ACTC1</i>	1	0.0189	0	0	0.0119
3	<i>ANK2</i>	1	0.0189	2	0.0004	0.0349
3	<i>COL4A1</i>	1	0.0189	2	0.0004	0.0349
3	<i>DAAM1</i>	1	0.0189	0	0	0.0119
3	<i>DSG2</i>	1	0.0189	0	0	0.0119
3	<i>DSP</i>	1	0.0189	0	0	0.0119
3	<i>FGF16</i>	1	0.0189	0	0	0.0119
3	<i>FGFR2</i>	1	0.0189	0	0	0.0119
3	<i>HCN4</i>	1	0.0189	0	0	0.0119
3	<i>JUP</i>	1	0.0189	0	0	0.0119
3	<i>MYBPC3</i>	1	0.0189	0	0	0.0119
3	<i>MYH6</i>	1	0.0189	0	0	0.0119
3	<i>MYL2</i>	1	0.0189	0	0	0.0119
3	<i>PKP2</i>	1	0.0189	0	0	0.0119
3	<i>PRDM16</i>	1	0.0189	0	0	0.0119
3	<i>RYR2</i>	1	0.0189	0	0	0.0119