genes

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Supplementary Materials

Clinical relevance of the systematic analysis of CNVs in the genetic study of cardiomyopathies.

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Table S1. List of the 95 CNVs pathogenic (P) or likely pathogenic (LP) according to the ACMG guidelines considered positive in the study.

Phenotype	Gene	Pathogenicity	Exons involved	zygosity	HGVS Names
DCM	*DMD*	P	48-51	hemi	NC_000023.10:g.31792079_31893492del NM_004006.2:c.6913_7542del NP_003997.1:p.Val2305_Lys2514del
DCM	*DMD*	LP	48-49	hemi	NC_000023.10:g.31849704_31911151delinsGGAATAGTAG-GAATAGTAGA NM_004006.2:c.6913-17661_7200+5131delinsTCTACTA-TTCCTACTATTCC NP_003997.1:p.Val2305_Lys2400del
DCM	*DMD*	P	45-48	hemi	NC_000023.10:g.31856965_32119775del NM_004006.2:c.6438+115262_7099-2025del NP_003997.1:p.Glu2147_Lys2366de
DCM	*DMD*	LP	2	hetero	NC_000023.10:g.33038257_33038318dup NM_004006.2:c.32_93dup NP_003997.1:p.Phe32Metfs*15
DCM	*DMD*	P	47-48	hemi	NC_000023.10:g.31893307_31947864del NM_004006.2:c.6763_7098del NP_003997.1:p.Leu2255_Lys2366del
DCM	*DMD*	P	3-9	hemi	NC_000023.10:g.32685760_32988401del NM_004006.2:c.93+49857_961-22489del NP_003997.1:p.Phe32_Gln320del
DCM	*DMD*	P	48-51	hetero	NC_000023.10:g.31792079_31893492del NM_004006.2:c.6913_7542del NP_003997.1:p.Val2305_Lys2514del
DCM	*DMD*	LP	19-43	hetero	NC_000023.10:g.32305646_32519959dup NM_004006.2:c.2293_6290dup NP_003997.1:p.?
DCM	*DMD*	P	49-51	hetero	NC_000023.10:g.31792079_31854938del NM_004006.2:c.7099_7542del NP_003997.1:p.Glu2367_Lys2514del

DCM	*DMD*	P	45	hetero	NC_000023.10:g.31986458_31986633del NM_004006.2:c.6439_6614del NP_003997.1:p.Glu2147Alafs*17
DCM	*DMD*	P	45-48	hemi	NC_000023.10:g.31893307_31986633del NM_004006.2:c.6439_7098del NP_003997.1:p.Glu2147_Lys2366del
DCM	*DMD*	LP	44-53	hetero	NC_000023.10:g.31697703_32235033del NM_004006.2:c.6438_7661del NP_003997.1:p.Lys2146_Ile2554delinsAsn
DCM	*DMD*	LP	56-59	hemi	NC_000023.10:g.31496223_31525570dup NM_004006.2:c.8218_8937dup
DCM	*DMD*	P	45-53	hetero	NC_000023.10:g.31641792_31947605del NM_004006.2:c.6912+112_8217+4002del NP_003997.1:p.Val2305_Gln2739del
DCM	*DMD*	P	20-24	hetero	NC_000006.11:g.7577967_7584099del NM_004415.3:c.2878-45_6604del NP_004406.2:p.Asp960Glnfs*16
DCM	*DMD*	P	46-47	hetero	NC_000023.10:g.31947715_31950346del NM_004006.2:c.6615_6912del NP_003997.1:p.Leu2206Phefs*16
DCM	*DMD*	P	45-55	hemi	NC_000023.10:g.31645790_31986631del NM_004006.2:c.6439_8217del NP_003997.1:p.Glu2147_Gln2739del
DCM	*DMD*	P	48-49	hemi	NC_000023.10:g.31845995_31904473del NM_004006.2:c.6913-10983_7201-7795del NP_003997.1:p.Val2305_Lys2400del
DCM	*DMD*	LP	48-51	hemi	NC_000023.10:g.31787841_31896426delinsCCTTTGAAC NM_004006.2:c.6913-2936_7542+4236delinsGTTCAAAGG NP_003997.1:p.?
DCM	*DMD*	LP	19-25	hemi	NC_000023.10:g.32474947_32524395dup NM_004006.2:c.2293-4436_3433-1998dup NP_003997.1:p.?
DCM	*DMD*	P	48-49	hemi	NC_000023.10:g.31854837_31893492del NM_004006.2:c.6913_7200del NP_003997.1:p.Val2305_Lys2400del
DCM	*DMD*	LP	35-44	hemi	NC_000023.10:g.32177150_32390103del NM_004006.2:c.4846-6787_6438+57883del NP_003997.1:p.Ala1616_Lys2146del
DCM	*DMD*	P	46-48	hetero	NC_000023.10:g.31869704_31954286del NM_004006.2:c.6615-3939_7099-14765del NP_003997.1:p.Leu2206Lysfs*4
DCM	*DMD*	P	52-53	hemi	NC_000023.10:g.31697494_31747867del NM_004006.2:c.7543_7872del NP_003997.1:p.Ala2515_Lys2624de
DCM	*DMD*	P	48-49	hemi	NC_000023.10:g.31854837_31893492del NM_004006.2:c.6913_7200del NP_003997.1:p.Val2305_Lys2400de

DCM	*DMD*	P	45-48	hemi	NC_000023.10:g.31893307_31986633del NM_004006.2:c.6439_7098del NP_003997.1:p.Glu2147_Lys2366del
DCM	*DMD*	P	48-51	hemi	NC_000023.10:g.31792079_31893492del NM_004006.2:c.6913_7542del NP_003997.1:p.Val2305_Lys2514del
DCM	*DMD*	P	45-55	hemi	NC_000023.10:g.31645790_31986631del NM_004006.2:c.6439_8217del NP_003997.1:p.Glu2147_Gln2739del
DCM	*DMD*	P	45-53	hemi	NC_000023.10:g.31697494_31986633del NM_004006.2:c.6439_7872del NP_003997.1:p.Glu2147_Lys2624del
DCM	*DMD*	LP	67-74	hetero	NC_000023.10:g.31175526_31235032delins [NC_000011.9:g.2434 9496-24355552] NM_004006.2:c.9361+6132_10554-9891delins NP_003997.1:p.Leu3121Serfs*11
DCM	*DSP*	LP	1-2	hetero	NC_000006.11:g.7542149_7556053del NM_004415.3:c.1_273del NP_004406.2:p.0?
DCM	*FLNC*	LP	3-5	hetero	NC_000007.13:g.128476527_128477737del NM_001458.4:c.602-687_897del NP_001449.3:p.?
DCM	*FLNC*	LP	24-32	hetero	NC_000007.13:g.128486595_128490469del NM_001458.4:c.4127+77_5329del NP_001449.3:p.?
DCM	*LAMP2*	LP	all	hetero	NC_000023.10:g.119562339_119590624del NM_002294.2:c.65_*2839del NP_002285.1:p.?
DCM	*LAMP2*	LP	6-8	hetero	NC_000023.10:g.119575587_119580284del NM_002294.2:c.742_1093del NP_002285.1:p.Val248Leufs*15
DCM	*LAMP2*	LP	1-8	hetero	NC_000023.10:g.119573044_119636423delins174 NM_002294.2:c.-33399_1093+2541delins174 NP_002285.1:p.Met1Glyfs*11
DCM	*LMNA*	P	2	hetero	NC_000001.10:g.156099922_156102230del NM_170707.3:c.357-487_513+1665del NP_733821.1:p.Asn120Leufs*5
DCM	*LMNA*	LP	all	hetero	NC_000001.10:g.156052337_156109880del NM_170707.3:c.-32373_*983del NP_733821.1:p.0?
DCM	*LMNA*	P	2-12	hetero	NC_000001.10:g.156095595_156111755del NM_170707.3:c.357-4813_*2858del NP_733821.1:p.Arg119fs
DCM	*LMNA*	LP	5-10	hetero	NC_000001.10:156104900_156108240dup NM_170707.3:c.811-78_1699-39dup
DCM	*PKP2*	LP	all	hetero	NC_000012.11:g.32943680_33049780del NM_004572.3:c.-115_*1678del NP_004563.2:p.Met1_*882del

DCM	*PLN*	LP	all	hetero	NC_000006.11:g.(?_118869427)_(118880244_?)x4
DCM	*TBX5*	LP	6	hetero	NC_000012.11:g.114832548_114832700del NM_000192.3:c.511_663del NP_000183.2:p.Ile171_Lys221del
DCM	*TTN*	LP	154	hetero	NC_000002.11:g.179433230_179436528delinsTGCGA-TAACATTAAGGGTTTCAATAGCTTCACCAGCTGAGTTAGTCA-GTTTAACCACATAATGGCCAACATCTTCTCGGCAGGC NM_003319.4:c.47136_50434delinsGCCTGCCGAGAA-GATGTTGGCCATTATGTGGTTAAACTGACTAACTCA-GCTGGTGAAGCTATTGAAACCCTTAATGTTATCGCA NP_003310.4:p.Asp15712Glufs*14
DCM	*TTN*	LP	188-191	hetero	NC_000002.11:g.179391078_179393287dup NM_003319.4:c.79996_*661dup NP_003310.4:p.P26666_*26927dup.
			154-191		NC_000002.11:g.179391452_179424996del NM_003319.4:c.58668_*287del NP_003310.4:p.Tyr19556fs
DCM	*TXNRD2* *2	P	all*	hetero	NC_000022.10:g.19167732_21351637del 22q11.2del
DCM/LVNC	*TTN*	LP	154	hetero	NC_000002.11:g.179433230_179436528delins-GCCTGCCGAGAAGATGTTGGCCATTATGTGGTTAAACTGAC-TAACTCAGCTGGTGAAGCTATTGAAACCCTTAATGTTATCGCA NM_003319.4:c.47136_50434delinsTGCGATAACAT-TAAGGGTTTCAATAGCTTCACCAGCTGAGTTAGTCAGTTTAAC-CACATAATGGCCAACATCTTCTCGGCAGGC NP_003310.4:p.Cys15714Ilefs*19
DCM/LVNC	*TTN*	LP	154	hetero	NC_000002.11:g.179433230_179436528delins-GCCTGCCGAGAAGATGTTGGCCATTATGTGGTTAAACTGAC-TAACTCAGCTGGTGAAGCTATTGAAACCCTTAATGTTATCGCA NM_003319.4:c.47136_50434delinsTGCGATAACAT-TAAGGGTTTCAATAGCTTCACCAGCTGAGTTAGTCAGTTTAAC-CACATAATGGCCAACATCTTCTCGGCAGGC NP_003310.4:p.Cys15714Ilefs*19
DCM/LVNC	*DMD*	LP	48-51	hemi	NC_000023.10:g.31792079_31893492de NM_004006.2:c.6913_7542del NP_003997.1:p.Val2305_Lys2514del
DCM/LVNC	*DMD*	LP	26-42	hemi	NC_000023.10:g.32328203_32472953del NM_004006.2:c.3433_6117del NP_003997.1:p.Val1145_Lys2039del
LVNC	*DMD*	P	49	hetero	NC_000023.10:g.31845186_31878210del NM_004006.2:c.7098+15096_7201-6985del NP_003997.1:p.Glu2367_Lys2400del
LVNC	*DSP*	LP	12	hetero	NC_000006.11:g. 7569421_7569481dup NM_004415.3:c.1422_1482dup NP_004406.2:p.Val495Asnfs*4
LVNC	*PRDM16* *1	P	all	hetero	1p36.33-p36.32 del (4Mb)

LVNC	*PRDM16*	LP	2-17	hetero	NC_000001.10:g.3102689_3350375del NM_022114.3:c.38_3831del NP_071397.3:p.*12fs
LVNC	*RYR2*	LP	3	hetero	NC_000001.10:g.237494234_237503798del NM_001035.2:c.223_273+9514del NP_001026.2:p.Val75_Gly91del
LVNC	*RYR2*	LP	4	hetero	NC_000001.10:g.237519265_237519285del NM_001035.2:c.274_294del NP_001026.2:p.Gln92_Trp98del
LVNC	*TBX20*	LP	7-8	hetero	NC_000007.13:g.35242042_35244194del NM_001077653.2:c.891_1344del NP_001071121.1:p.Arg297(?)
LVNC/HCM	*MYBPC3*	LP	21	hetero	NC_000011.9:g.47361014_47362361del NM_000256.3:c.1927+193_2068-59del NP_000247.2:p.Glu643Glyfs*3
HCM	*FHL1*	LP	all	hetero	NC_000023.10:g.135278961_135292184del NM_001159700.1:c.-26-9605_843del NP_001153172.1:p.0?
HCM	*FHOD3*	P	15	hetero	NC_000018.9:g.34260087_34262764delinsGTTCTGCCCA-TAGTCTTTGCTCCCAGG NM_001281740.2:c.1836-1312_1970+1231delinsTTCTGCC-CATAGTCTTTGCTCCCAGG NP_001268669.1:p.Arg612_Phe656del
HCM	*FHOD3*	P	15-16	hetero	NC_000018.9:g.34259872_34271130del NM_001281740.2:c.1836-1527_2022-2042del NP_001268669.1: p.Ser613_Arg674del
HCM	*FHOD3*	P	15-16	hetero	NC_000018.9:g.34258856_34269358delinsT-GTAAAAAAAAAATAAAAGATATAAAGATCCAAATGGAAATA-TATATAGAACAG-GAAAATTAAAATAGAAAATTTGTCAGATGGGCCAAATAGAATTGCTATGA NM_001281740.2:c.1836-2543_2021+2217delinsTGTAAAAAAAAAATAAAAGATA-TAAAGATCCAAATGGAAATATATATAGAACAG-GAAAATTAAAATAGAAAATTTGTCAGATGGGCCAAATAGAATTGCTATGAG NP_001268669.1:p.Ser613_Arg674del
HCM	*LAMP2*	LP	7	hetero	NC_000023.10:g.119576455_119576518del NM_002294.2:c.865_928del NP_002285.1:p.Lys289Phefs*36
HCM	*MYBPC3*	LP	4-5	hetero	NC_000011.9:g.47371423_47372009del NM_000256.3:c.406+45_557del NP_000247.2:p.Ser137Leufs*13
HCM	*MYBPC3*	LP	1-5	hetero	NC_000011.9:g.47370237_47375203del NM_000256.3:c.-999_655-139del NP_000247.2:p.0?
HCM	*MYBPC3*	LP	4-7	hetero	NC_000011.9:g.47369431_47371766del NM_000256.3:c.407-103_798del NP_000247.2:p.?

HCM	*MYBPC3* *3	LP	all	hetero	NC_000011.9:g.34196603_49219155del
HCM	*MYBPC3*	LP	1-17	hetero	NC_000011.9:g.47364046_47375171del NM_000256.3:c.-973_1624+83del NP_000247.2:p.0?
HCM	*MYBPC3*	LP	18	hetero	NC_000011.9:g.47363206_47363745del NM_000256.3:c.1625-38_1790+336del NP_000247.2:p.Glu542Glyfs*5
HCM	*MYBPC3*	P	23-26	hetero	NC_000011.9:g.47357175_47360721del NM_000256.3:c.2148+154_2737+253del NP_000247.2:p.Leu717Alafs*11
HCM	*MYBPC3*	P	23-26	hetero	NC_000011.9:g.47357175_47360721del NM_000256.3:c.2148+154_2737+253del NP_000247.2:p.Leu717Alafs*11
HCM	*MYBPC3*	LP	27-7	hetero	NC_000011.9:g.47356553_47357279delinsC NM_000256.3:c.2737+149_2905+40delinsG NP_000247.2:p?
HCM	*MYBPC3*	LP	27	hetero	NC_000011.9:g.47356553_47357279delinsC NM_000256.3:c.2737+149_2905+40delinsG NP_000247.2:p.?
HCM	*MYH7* *MYH6*	P	*MYH7* all *MYH6* 1-5	hetero	NC_000014.8:g.23874432_23901847del
HCM	*PKP2*	LP	13-14	hetero	NC_000012.11:g.32945358_32945665del NM_004572.3:c.2490_2646del NP_004563.2:p.Ala830fs
HCM	*PKP2*	P	8	hetero	NC_000012.11:g.32976981_32977098del NM_004572.3:c.1689_1806del NP_004563.2:p.Asn564Profs*53
HCM	*PLN*	LP	all	hetero	NC_000006.11:g.118869442_118880243del NM_002667.3:c.-211_159del NP_002658.1:p.0?
RCM	*DES*	LP	1-4	hetero	NC_000002.11:g.220267400_220285223dupinv NM_001927.3:c.-15785_742dupinv
ARVC	*DMD*	LP	25-30	hemi	NC_000023.10:g.32423738_32481666del NM_004006.2:c.3322_4233+6131del NP_003997.1:p.Val1108_Gln1411del
ARVC	*DMD*	LP	40-55	hemi	NC_000023.10:g.31645790_32361403dup NM_004006.2:c.5587_8217dup NP_003997.1:p.?
ARVC	*DSP*	P	21-24	hetero	NC_000006.11:g.7577966_7584094del NM_004415.2:c.2878-46_6599del NP_004406.2:p.Asp960Alafs*21
ARVC	*FLNC*	LP	all	hetero	NC_000007.13:g.128470694_128498579del NM_001458.4:c.3_*2del NP_001449.3:p.0?
ARVC	*FLNC*	P	FLNC 3-48	hetero	NC_000007.13:g.128476955_128510390del NM_001458.4:c.602-259_*11813del NP_001449.3:p.?

ARVC	*PKP2*	LP	8	hetero	NC_000012.11:g.32976092_32978401del NM_004572.3:c.1689-1305_1807-527del NP_004563.2:p.Asn564Profs*53
ARVC	*PKP2*	LP	4	hetero	NC_000012.11:g.33019241_33026724del NM_004572.3:c.1034+4057_1170+2621del NP_004563.2:p.Asn346Leufs*14
ARVC	*PKP2*	LP	8	hetero	NC_000012.11:g.32976092_32978401del NM_004572.3:c.1689-1305_1807-527del NP_004563.2:p.Asn564Profs*53
ARVC	*PKP2*	P	8	hetero	NC_000012.11:g.32976092_32978401del NM_004572.3:c.1689-1305_1807-527del NP_004563.2:p.Asn564Profs*53
ARVC	*PKP2*	P	8	hetero	NC_000012.11:g.32976092_32978401del NM_004572.3:c.1689-1305_1807-527del NP_004563.2:p.Asn564Profs*53
ARVC	*PKP2*	LP	10	hetero	NC_000012.11:g.32974274_32974845del NM_004572.3:c.1972-380_2145+18del NP_004563.2:p.Gln658_Pro715del
ARVC	*PKP2*	LP	8-10	hetero	NC_000012.11:g.32974290_32977096dup NM_004572.3:c.1689_2145dup
ARVC	*PKP2*	LP	5-14	hetero	NC_000012.11:g.32924017_33011214delinsCATTAT NM_004572.3:c.1171-7307_*21341delinsATAATG NP_004563.2:p.?
ARVC	*PKP2*	LP	13-14	hetero	NC_000012.11:g.32942317_32945702del NM_004572.3:c.2490-35_*3043del NP_004563.2:p.Ala830fs
ARVC	*PKP2*	LP	13-14	hetero	NC_000012.11:g.32945358_32945665del NM_004572.3:c.2490_2646del NP_004563.2:p.Ala830fs
ARVC	*PKP2*	LP	8-10	hetero	NC_000012.11:g.32974290_32977096dup NM_004572.3:c.1689_2145dup
ARVC	*PKP2*	P	8	hetero	NC_000012.11:g.32976092_32978401del NM_004572.3:c.1689-1305_1807-527del NP_004563.2:p.Asn564Profs*53

*1 This deletion affects 112 genes (79 coding). The following are particularly relevant: *GNB1, ISG15, AGRN, TNFRSF4, B3GALT6 DVL1, ATAD3A, TMEM240, GABRD SKI, PEX10, PRDM16, SMIM1* and *CEP104*

2 This deletions affects 13 genes: CLTCL1, HIRA, CDC45, CLDN5, GP1BB, TBX1, TNXRD2, DGCR8, ZNF74, KLHL22, MED15, SNAP29* and *LZTR1*

*3 This deletion affects 158 genes (*MYBPC3* included), 85 are coding: *ABTB2, ACCS, ACCSL, ACP2, AGBL2, ALKBH3, ALX4, AMBRA1, API5, APIP, ARFGAP2, ARHGAP1, ATG13, C11orf49, C11orf74, C11orf94, C11orf96, C1QTNF4, CAT, CD44, CD82, CELF1, CHRM4, CHST1, CKAP5, COMMD9, CREB3L1, CRY2, DDB2, DGKZ, EHF, ELF5, EXT2, F2, FAM180B, FJX1, FNBP4, FOLH1, HARBI1, HSD17B12, KBTBD4, LARGE2, LDLRAD3, LRP4, LRRC4C, MADD, MAPK8IP1, MDK, MTCH2, MYBPC3, NDUFS3, NR1H3, NUP160, OR4A47, OR4B1, OR4C3, OR4S1, OR4X1, OR4X2, PACSIN3, PAMR1, PDHX, PEX16, PHF21A, PRDM11, PRR5L, PSMC3, PTPMT1, PTPRJ, RAG1, RAG2, RAPSN, SLC1A2, SLC35C1, SLC39A13, SPI1, SYT13, TP53I11, TRAF6, TRIM44, TRIM49B, TRIM64C, TSPAN18, TTC17, ZNF408*