

Appendix

Genomic Structural Variations Lead to Dysregulation of Important Coding and Non-Coding RNA Species in Dilated Cardiomyopathy

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Appendix Table 1: Top 20 associated genes from SNV-QTL analysis

Gene Name	ENSEMBL Gene ID	p-value	beta	FDR
<i>HLA-DRA</i>	ENSG00000204287	7.694270×10^{-22}	-4.136061	3.373082×10^{-14}
<i>AGPAT1</i>	ENSG00000204310	2.609624×10^{-19}	-1.908875	4.527451×10^{-12}
<i>HLA-DRB5</i>	ENSG00000198502	1.373991×10^{-18}	-2.906355	4.527451×10^{-12}
<i>HLA-DQB1</i>	ENSG00000179344	3.164864×10^{-18}	-2.615057	4.527451×10^{-12}
<i>HLA-DQA1</i>	ENSG00000196735	3.190135×10^{-18}	-2.288957	4.527451×10^{-12}
<i>RP11-166B2.1</i>	ENSG00000234719	3.799762×10^{-18}	-1.427571	4.527451×10^{-12}
<i>TMEM117</i>	ENSG00000139173	3.829671×10^{-18}	-4.598441	4.527451×10^{-12}
<i>TOM1L1</i>	ENSG00000141198	4.770673×10^{-18}	-4.177049	4.527451×10^{-12}
<i>ERAP2</i>	ENSG00000164308	4.830384×10^{-18}	-2.070406	4.527451×10^{-12}
<i>HLA-F</i>	ENSG00000204642	6.093213×10^{-18}	-2.091995	4.527451×10^{-12}
<i>LDHC</i>	ENSG00000166796	9.327095×10^{-18}	-2.120558	5.181929×10^{-12}
<i>ZC3H10</i>	ENSG00000135482	9.976414×10^{-18}	-3.800280	5.181929×10^{-12}
<i>AASDH</i>	ENSG00000157426	1.251584×10^{-17}	-4.302499	5.751366×10^{-12}
<i>AVIL</i>	ENSG00000135407	3.003591×10^{-17}	-3.268834	1.261246×10^{-11}
<i>PRRC2A</i>	ENSG00000204469	6.334865×10^{-17}	-2.218379	2.552513×10^{-11}
<i>FAM21B</i>	ENSG00000152726	7.860013×10^{-17}	-3.430117	3.155441×10^{-11}
<i>ZNF155</i>	ENSG00000204920	1.320437×10^{-16}	-3.225438	4.586884×10^{-11}
<i>DENND2C</i>	ENSG00000175984	3.613585×10^{-16}	-2.729215	1.124509×10^{-10}
<i>PDIK1L</i>	ENSG00000175087	3.693736×10^{-16}	-4.166681	1.124509×10^{-10}
<i>FABP2</i>	ENSG00000145384	6.789247×10^{-16}	2.4202388	2.030239×10^{-10}

FDR = FDR adjusted p-value

Appendix Table 2: Structural variant eQTL loci

Ensembl ID	GeneName	SV-type	Chr	SV-start	SV-end	min_pval_SNV_eqtl_FDR
ENSG00000070808	CAMK2A	INVDEL	5	150319082	150319613	0.0553727407460288
ENSG00000079482	OPHN1	DEL	X	66412759	66413050	1
ENSG00000085224	ATRX	DEL	X	77121388	77123852	1
ENSG00000086712	TXLNG	DEL	X	16805893	16806310	1
ENSG00000099984	GSTT2	DEL	22	24274003	24311292	0.00193727130356245
ENSG00000101407	TTI1	DEL	20	36311365	36311762	1
ENSG00000105889	STEAP1B	DEL	7	21538064	21540560	0.0761151020089635
ENSG00000108349	CASC3	DEL	17	38385648	38387943	0.0315762689410888
ENSG00000117226	GBP3	DEL	1	89475931	89478608	2.81397292529639e-07
ENSG00000117601	SERPINC1	DEL	1	173872947	173886516	0.0856173998178337
ENSG00000121053	EPX	DEL	17	57238676	57241299	0.0843994588157762
ENSG00000133433	GSTT2B	DEL	22	24274003	24311292	0.00171206712674254
ENSG00000134970	TMED7	DEL	5	115531957	115610698	0.00136487415285159
ENSG00000139344	AMDHD1	DEL	12	96340185	96342949	0.545898731833455
ENSG00000141433	ADCYAP1	DEL	18	828483	828959	1
ENSG00000142528	ZNF473	DUP	19	50522237	50592537	4.1891645830034e-07
ENSG00000144671	SLC22A14	DUP	3	37751532	37753602	1
ENSG00000145782	ATG12	DEL	5	115531957	115610698	0.00111788679229252
ENSG00000155957	TMBIM4	DEL	12	66527500	66529870	3.88067681384311e-05
ENSG00000155959	VBP1	DEL	X	154485907	154486947	1
ENSG00000155959	VBP1	DEL	X	154918462	154921721	1
ENSG00000155962	CLIC2	DEL	X	154485907	154486947	1
ENSG00000155962	CLIC2	DEL	X	154918462	154921721	1
ENSG00000163596	ICA1L	DEL	2	203272279	203279933	0.00525158829466544
ENSG00000167768	KRT1	DEL	12	53086343	53087692	0.103227347214968
ENSG00000169914	OTUD3	DEL	1	19720443	19732042	0.00141681597663234
ENSG00000169981	ZNF35	INVDUP	3	43835086	43836002	1
ENSG00000171084	FAM86JP	DEL	3	125672294	125676372	0.0123409136583227
ENSG00000175220	ARHGAP1	DUP	11	46742757	46743621	0.37734820366397
ENSG00000178381	ZFAND2A	DEL	7	1185026	1187649	0.491554349678595
ENSG00000180438	TPRXL	DEL	3	12904009	12914616	0.406584397191934
ENSG00000181541	MAB21L2	DUP	4	151292346	151293914	0.601220303762998
ENSG00000184674	GSTT1	DEL	22	24274003	24311292	8.32851122310166e-06
ENSG00000185168	LINC00482	DEL	17	79143178	79148174	0.166525112807724
ENSG00000188002	RP11-43F13.1	DEL	5	1627101	1628833	0.0378748913640489
ENSG00000196646	ZNF136	DEL	19	11260612	11272111	0.00164511985536848
ENSG00000198028	ZNF560	DEL	19	9274503	9284355	1
ENSG00000204136	GGTA1P	DUP	9	124226892	124227825	0.00533577717742678

ENSG00000204209	<i>DAXX</i>	DEL	6	33010647	33019605	0.0636799248377916
ENSG00000204619	<i>PPP1R11</i>	DEL	6	29685464	29688071	3.0108282921388e-09
ENSG00000204650	<i>CRHR1-IT1</i>	DEL	17	44355134	44359562	1.6948442579842e-05
ENSG00000205488	<i>CALML3-AS1</i>	DEL	10	5914906	5915914	0.400524998563513
ENSG00000218537	<i>AP000350.4</i>	DEL	22	24274003	24311292	0.000556224185176398
ENSG00000221947	<i>XKR9</i>	DEL	8	71583568	71584637	0.00653807100311564
ENSG00000224216	<i>RP13-228J13.1</i>	DEL	X	154485907	154486947	1
ENSG00000224216	<i>RP13-228J13.1</i>	DEL	X	154918462	154921721	1
ENSG00000228701	<i>TNKS2-AS1</i>	DEL	10	93633247	93634556	0.000327478049455422
ENSG00000229043	<i>AC091729.9</i>	DEL	7	1185026	1187649	0.00047986557356117
ENSG00000230439	<i>RP11-488P3.1</i>	DEL	1	94288305	94291249	0.00739322710144155
ENSG00000231482	<i>AC141930.2</i>	DEL	2	936820	937135	1
ENSG00000234176	<i>HSPA8P1</i>	DEL	X	121138848	121139304	1
ENSG00000235573	<i>RP3-412A9.12</i>	DUP	22	32001252	32011923	0.636091195076709
ENSG00000236814	<i>RP11-446E9.1</i>	DUP	8	56979007	56979416	0.140735211657874
ENSG00000236829	<i>Z97634.3</i>	DEL	16	1290107	1290372	0.0466672753124635
ENSG00000238244	<i>GABARAPL3</i>	DEL	15	91219157	91222800	0.00089687726402956
ENSG00000239335	<i>LLPH-AS1</i>	DEL	12	66527500	66529870	0.00364890859550425
ENSG00000239445	<i>ST3GAL6-AS1</i>	DEL	3	98410646	98414778	0.764433089226613
ENSG00000250007	<i>RP11-814P5.1</i>	DEL	15	34717592	34863734	0.103939772677261
ENSG00000253327	<i>RAD21-AS1</i>	DEL	8	118910889	118928996	0.0981580849218429
ENSG00000253696	<i>KBTBD11-OT1</i>	DEL	8	1305165	1307074	0.696148042545162
ENSG00000253981	<i>ALG1L13P</i>	DEL	8	9054963	9062745	0.109212870336974
ENSG00000256278	<i>RP11-182J1.5</i>	DEL	15	85475174	85481029	0.0660845877278208
ENSG00000259244	<i>RP11-182J1.12</i>	DEL	15	84604908	84606926	0.300033773454947
ENSG00000259592	<i>PRELID1P4</i>	DEL	15	34709459	34709717	0.182331373843536
ENSG00000260646	<i>LA16c-385E7.1</i>	DUP	16	2186937	2192343	0.151239964949153
ENSG00000260646	<i>LA16c-385E7.1</i>	DEL	16	1290107	1290372	0.151239964949153
ENSG00000260923	<i>AC137934.1</i>	DEL	16	89967737	89969116	0.143014942049194
ENSG00000261770	<i>CTC-459F4.1</i>	DUP	19	27799110	27800050	0.0688959984596499
ENSG00000262902	<i>RP11-750B16.1</i>	DEL	17	52158671	52167998	0.104255652967692
ENSG00000264672	<i>SEPT4-AS1</i>	DEL	17	57238676	57241299	0.0188921070318906
ENSG00000266235	<i>MIR3176</i>	DEL	16	1290107	1290372	0.160588716208391
ENSG00000267179	<i>ZNF763</i>	DEL	19	11260612	11272111	0.0397989700457253
ENSG00000269102	<i>CTD-2525I3.5</i>	DEL	19	53272779	53277566	0.00778597861927192
ENSG00000272070	<i>AC005618.6</i>	DEL	5	140096982	140104134	0.43813848511727
ENSG00000273295	<i>AP000350.5</i>	DEL	22	24274003	24311292	4.89377115355646e-09

DEL = large deletion, DUP = large duplication, INVDEL= inversion and a deletion in one locus (complex SV), INV DUP= inversion and a duplication in one locus (complex SV),

Appendix Table 3: Differential Expression caused by SVs for functional gene sets

List	Size	Available	Intersect with SV-eQTL	Determi- nation
ALL HEART EXPRESSED GENES	20712	20712	16449	0.075
GO_MUSCLE_CELL_DEVELOPMENT	88	88	88	0.087
GO_MUSCLE_ORGAN_DEVELOPMENT	182	181	181	0.104
KEGG_ARRHYTHMOGENIC_RIGHT- VENTRICULAR_CARDIOMYOPATHY	54	53	53	0.099
KEGG_CARDIAC_MUSCLE_CONTRACTION	51	51	51	0.098
KEGG_DILATED_CARDIOMYOPATHY	64	64	64	0.101
KEGG_HYPERTROPHIC_CARDIOMYOPATHY_ HCM	60	60	60	0.096