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Supplemental Data

Copy-Number Variation of the Glucose Transporter Gene *SLC2A3* and Congenital Heart Defects in the 22q11.2 Deletion Syndrome

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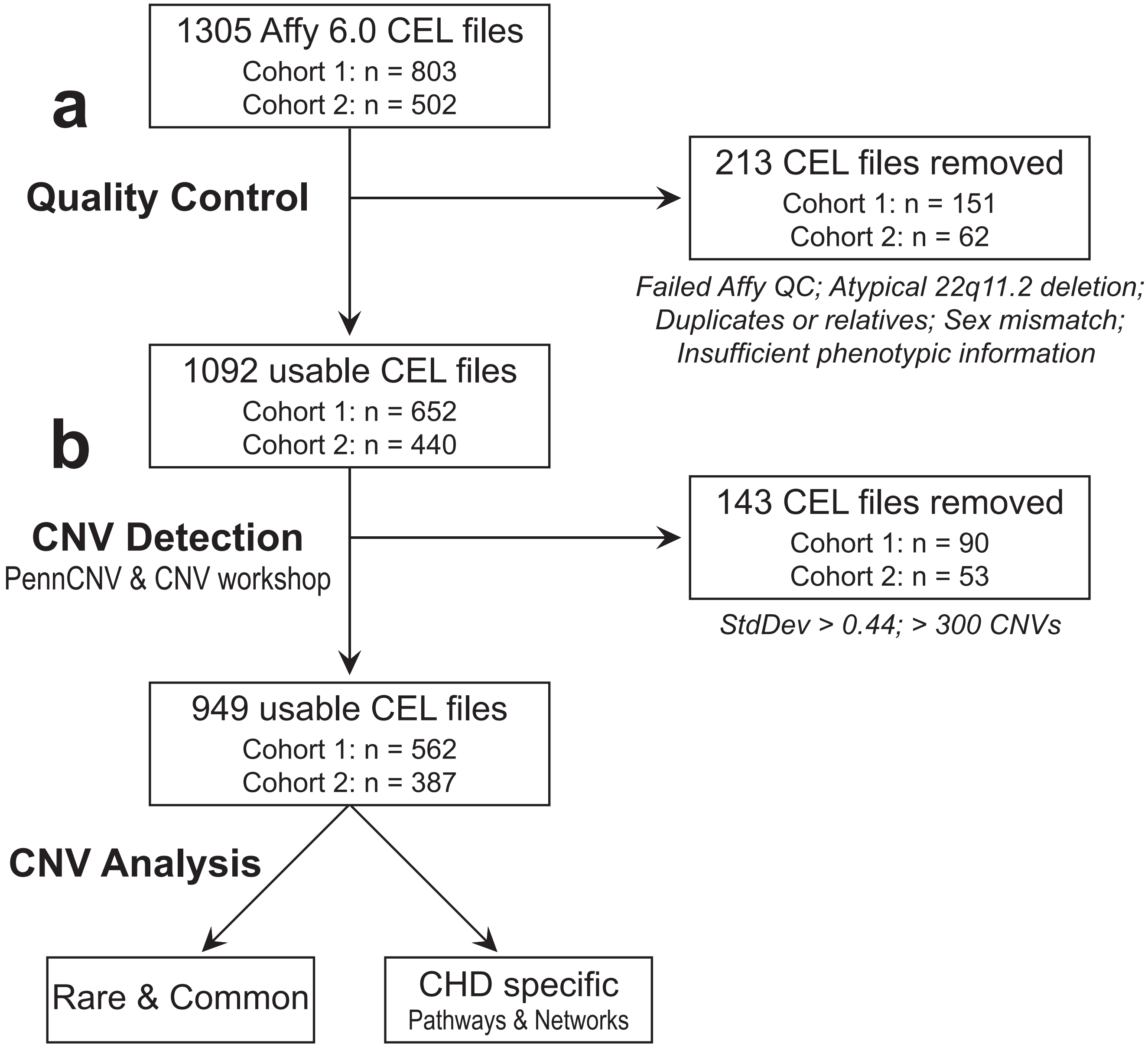
Figure S1:



1305 Affy 6.0 CEL files
Cohort 1: n = 803
Cohort 2: n = 502
a
Quality Control
213 CEL files removed
Cohort 1: n = 151
Cohort 2: n = 62
Failed Affy QC; Atypical 22q11.2 deletion; Duplicates or relatives; Sex mismatch; Insufficient phenotypic information
1092 usable CEL files
Cohort 1: n = 652
Cohort 2: n = 440
b
CNV Detection
PennCNV & CNV workshop
143 CEL files removed
Cohort 1: n = 90
Cohort 2: n = 53
StdDev > 0.44; > 300 CNVs
949 usable CEL files
Cohort 1: n = 562
Cohort 2: n = 387
CNV Analysis
Rare & Common
CHD specific
Pathways & Networks

Figure S1: CNV detection and analysis. Flow chart representing our CNV detection and analysis and the number of .CEL files from each cohort included at each step. (a) initial assessment of .cel file quality. (b) CNV detection and quality control.

Table S1: 22q11.2 Deletion Sizes

	First Cohort [a]						Second Cohort [b]						Cohorts Combined [c]					
	Total		CHD		No CHD		Total		CHD		No CHD		Total		CHD		No CHD	
	(n = 562)		(n = 363)		(n = 199)		(n = 387)		(n = 240)		(n = 147)		(n = 949)		(n = 603)		(n = 346)	
	#	%	#	%	#	%	#	%	#	%	#	%	#	%	#	%	#	%
A-D	534	95.0	343	94.5	191	96.0	358	92.5	221	92.1	137	93.2	892	94.0	564	93.5	328	94.8
A-C	7	1.3	5	1.4	2	1.0	8	2.1	5	2.1	3	2.0	15	1.6	10	1.7	5	1.4
A-B	21	3.7	15	4.1	6	3.0	21	5.4	14	5.8	7	4.8	42	4.4	29	4.8	13	3.8

[a] The frequency of the 22q11.2 deletion sizes in first cohort subjects with CHDs, without CHDs and combined.

[b] The frequency of the 22q11.2 deletion sizes in second cohort subjects with CHDs, without CHDs and combined.

[c] Overall 22q11.2 deletion frequencies with the first and second cohorts combined.

Table S2: Validated CNVs (GRCh37/hg19 genome build)

Sample	Phenotype	Chr	Start (bp)	End (bp)	Size (bp)	Type	# of Probes	CNV Detection Programs	Inherited	Gene(s)
7462	CHD	1	228,807,528	228,957,150	149,622	Duplication	86	CNV Workshop & PennCNV	Not tested	*RHOU*
5471	CHD	2	27,263,122	27,648,118	384,996	Duplication	158	CNV Workshop & PennCNV	Not tested	*EMILIN1, TRIM54*
6504	CHD	2	32,627,998	33,331,779	703,781	Duplication	453	CNV Workshop & PennCNV	Not tested	*LTBP1*
5373	CHD	2	32,630,549	33,331,779	701,230	Duplication	452	CNV Workshop & PennCNV	Not tested	*LTBP1*
6719	CHD	2	113,926,885	114,236,566	309,681	Duplication	150	CNV Workshop & PennCNV	Not tested	*PAX8*
4160	CHD	3	7,615,567	8,156,424	540,857	Duplication	503	CNV Workshop & PennCNV	Not tested	*GRM7*
6501	CHD	5	179,612,667	179,883,302	270,635	Duplication	175	CNV Workshop & PennCNV	Not tested	*MAPK9*
5387	No CHD	9	8,748,942	8,809,841	60,899	Deletion	61	CNV Workshop & PennCNV	Not tested	*PTPRD*
4212	No CHD	9	9,798,243	9,822,054	23,811	Deletion	57	CNV Workshop & PennCNV	Yes	*PTPRD*
4251	No CHD	9	9,847,865	9,966,099	118,234	Deletion	144	CNV Workshop & PennCNV	Yes	*PTPRD*
3935	CHD	9	80,063,217	80,086,725	23,508	Deletion	23	CNV Workshop & PennCNV	Yes	*GNA14*
6516	CHD	9	119,411,347	119,659,873	248,526	Deletion	267	CNV Workshop & PennCNV	Not tested	*ASTN2;TRIM32*
7542	No CHD	10	53,383,564	53,404,882	21,318	Deletion	18	CNV Workshop & PennCNV	Yes	*PRKG1*
5456	No CHD	10	79,250,577	79,370,519	119,942	Deletion	98	CNV Workshop & PennCNV	Not tested	*KCNMA1*
3920	CHD	12	8,004,426	8,123,238	118,812	Duplication	59	CNV Workshop & PennCNV	Not tested	*SLC2A3*
3943	CHD	12	7,994,787	8,123,900	129,113	Duplication	25	CNV Workshop & PennCNV	Not tested	*SLC2A3*
4248	CHD	12	8,004,426	8,130,958	126,532	Duplication	61	CNV Workshop & PennCNV	Yes	*SLC2A3*
5355	No CHD	12	8,004,425	8,123,900	119,475	Duplication	60	CNV Workshop & PennCNV	Not tested	*SLC2A3*
5368	CHD	12	8,004,425	8,135,092	130,667	Duplication	64	CNV Workshop & PennCNV	Not tested	*SLC2A3*
5377	No CHD	12	8,004,425	8,123,900	119,475	Duplication	60	CNV Workshop & PennCNV	Not tested	*SLC2A3*
5393	CHD	12	8,004,425	8,123,900	119,475	Duplication	60	CNV Workshop & PennCNV	Not tested	*SLC2A3*
5646	CHD	12	8,004,425	8,123,900	119,475	Duplication	60	CNV Workshop & PennCNV	Not tested	*SLC2A3*
5647	CHD	12	8,004,425	8,123,900	119,475	Duplication	60	CNV Workshop & PennCNV	Not tested	*SLC2A3*
5650	CHD	12	8,004,425	8,123,900	119,475	Duplication	60	CNV Workshop & PennCNV	Not tested	*SLC2A3*
6498	CHD	12	7,971,035	8,143,483	172,448	Duplication	69	CNV Workshop & PennCNV	Yes	*SLC2A3*
6522	CHD	12	8,004,426	8,123,901	119,475	Duplication	59	CNV Workshop & PennCNV	Not tested	*SLC2A3*
6696	CHD	12	7,958,406	8,130,958	172,552	Duplication	72	CNV Workshop & PennCNV	Not tested	*SLC2A3*
6708	CHD	12	8,004,426	8,123,901	119,475	Deletion	60	CNV Workshop & PennCNV	Yes	*SLC2A3*
6709	CHD	12	7,962,762	8,123,901	161,139	Duplication	69	CNV Workshop & PennCNV	Yes	*SLC2A3*
6953	CHD	12	7,994,787	8,123,900	129,113	Duplication	61	CNV Workshop & PennCNV	Yes	*SLC2A3*
6960	CHD	12	7,994,787	8,123,900	129,113	Duplication	56	CNV Workshop & PennCNV	Yes	*SLC2A3*
6967	CHD	12	7,994,787	8,150,607	155,820	Duplication	60	CNV Workshop & PennCNV	Not tested	*SLC2A3*
7530	CHD	12	7,994,787	8,123,900	129,113	Duplication	59	CNV Workshop & PennCNV	Yes	*SLC2A3*
7591	CHD	12	8,006,507	8,134,669	128,162	Duplication	61	CNV Workshop & PennCNV	Not tested	*SLC2A3*
7657	CHD	12	8,016,749	8,090,715	73,966	Duplication	26	CNV Workshop & PennCNV	Not tested	*SLC2A3*

10357	No CHD	12	7,994,787	8,123,900	129,113	Duplication	60	CNV Workshop & PennCNV	Not tested	*SLC2A3*
10553	CHD	12	7,970,721	8,130,958	160,237	Duplication	65	CNV Workshop & PennCNV	Yes	*SLC2A3*
10638	CHD	12	7,994,787	8,137,722	142,935	Duplication	65	CNV Workshop & PennCNV	Not tested	*SLC2A3*
10717	CHD	12	8,004,425	8,135,092	130,667	Duplication	63	CNV Workshop & PennCNV	Yes	*SLC2A3*
10904	CHD	12	7,994,787	8,123,900	129,113	Duplication	60	CNV Workshop & PennCNV	Not tested	*SLC2A3*
11025	CHD	12	8,004,425	8,123,900	119,475	Duplication	59	PennCNV	Not tested	*SLC2A3*
11159	CHD	12	7,994,787	8,137,722	142,935	Duplication	65	CNV Workshop & PennCNV	Yes	*SLC2A3*
11223	CHD	12	7,994,787	8,123,900	129,113	Duplication	60	CNV Workshop & PennCNV	Not tested	*SLC2A3*
11258	CHD	12	7,994,787	8,123,900	129,113	Duplication	60	CNV Workshop & PennCNV	Yes	*SLC2A3*
11292	CHD	12	7,994,787	8,123,900	129,113	Duplication	60	CNV Workshop & PennCNV	Not tested	*SLC2A3*
11316	No CHD	12	8,004,425	8,123,900	119,475	Duplication	59	PennCNV	Yes	*SLC2A3*
11322	CHD	12	8,004,425	8,123,900	119,475	Duplication	59	CNV Workshop & PennCNV	Not tested	*SLC2A3*
11323	CHD	12	8,004,425	8,123,900	119,475	Duplication	59	CNV Workshop & PennCNV	Not tested	*SLC2A3*
11327	CHD	12	7,994,787	8,123,900	129,113	Duplication	60	CNV Workshop & PennCNV	Not tested	*SLC2A3*
11336	CHD	12	8,004,425	8,123,900	119,475	Duplication	59	PennCNV	Not tested	*SLC2A3*
11342	No CHD	12	7,994,787	8,123,900	129,113	Duplication	60	CNV Workshop & PennCNV	Yes	*SLC2A3*
11960	CHD	12	7,994,787	8,123,900	129,113	Duplication	60	CNV Workshop & PennCNV	Yes	*SLC2A3*
11964	CHD	12	7,994,787	8,123,900	129,113	Duplication	60	CNV Workshop & PennCNV	Yes	*SLC2A3*
11971	No CHD	12	7,994,787	8,123,900	129,113	Duplication	60	PennCNV	Yes	*SLC2A3*
12000	CHD	12	8,007,799	8,123,900	116,101	Duplication	57	CNV Workshop & PennCNV	Not tested	*SLC2A3*
5402	CHD	13	110,593,018	111,185,674	592,656	Duplication	570	CNV Workshop & PennCNV	Not tested	*COL4A1*
6502	CHD	14	64,731,581	65,007,547	275,966	Duplication	184	CNV Workshop & PennCNV	Not tested	*MTHFD1*
5496	No CHD	14	73,018,692	73,188,701	170,009	Duplication	125	CNV Workshop & PennCNV	Not tested	*DPF3*
5491	CHD	15	22,673,400	23,478,050	804,650	Duplication	322	CNV Workshop & PennCNV	Not tested	*15q11.2*
6948	CHD	15	22,681,251	23,487,547	806,296	Duplication	323	CNV Workshop & PennCNV	Yes	*15q11.2*
6696	CHD	15	22,751,681	23,282,812	531,131	Deletion	308	CNV Workshop & PennCNV	Yes	*15q11.2*
4194	CHD	16	14,835,068	16,396,512	1,561,444	Deletion	679	CNV Workshop & PennCNV	Yes	*MYH11*
6501	CHD	16	82,714,047	82,765,607	51,560	Deletion	47	CNV Workshop & PennCNV	Not tested	*CDH13*
6700	CHD	16	83,308,001	83,353,031	45,030	Duplication	59	CNV Workshop & PennCNV	Yes	*CDH13*
3935	CHD	17	34,816,256	36,295,000	1,478,744	Duplication	996	CNV Workshop & PennCNV	Yes	17q12
5482	CHD	17	72,629,123	73,413,449	784,326	Duplication	382	CNV Workshop & PennCNV	Not tested	*GRB2*
4178	CHD	18	23,634,475	28,940,034	5,305,559	Deletion	3360	CNV Workshop & PennCNV	Not tested	18q11.2
4165	CHD	21	18,660,957	19,220,122	559,165	Duplication	374	CNV Workshop & PennCNV	Not tested	*CXADR*
6963	CHD	21	18,736,782	19,124,700	387,918	Duplication	295	CNV Workshop & PennCNV	Not tested	*CXADR*
5442	CHD	21	35,722,995	35,903,830	180,835	Duplication	109	CNV Workshop & PennCNV	Yes	*RCAN1*

Table S3: Gene Ontology - Common CNVs

A: All CNV types (17 statistically significant terms)

GO ID	GO Term Description	GO Term Type	CHD (#)	No CHD (#)	p Value	padjust_BH	FDR
GO:0005996	monosaccharide metabolic process	biological process	50	7	2.65E-05	0.032536	0.0540
GO:0005975	carbohydrate metabolic process	biological process	78	17	2.83E-05	0.032536	0.0275
GO:0044723	single-organism carbohydrate metabolic process	biological process	75	17	6.80E-05	0.039545	0.0507
GO:0007275	multicellular organismal development	biological process	45	7	0.000149	0.039545	0.0488
GO:0005355	glucose transmembrane transporter activity	molecular function	43	7	0.000292	0.039545	0.0166
GO:0006766	vitamin metabolic process	biological process	43	7	0.000292	0.039545	0.0166
GO:0006767	water-soluble vitamin metabolic process	biological process	43	7	0.000292	0.039545	0.0166
GO:0008643	carbohydrate transport	biological process	43	7	0.000292	0.039545	0.0166
GO:0008645	hexose transport	biological process	43	7	0.000292	0.039545	0.0166
GO:0015144	carbohydrate transmembrane transporter activity	molecular function	43	7	0.000292	0.039545	0.0166
GO:0015145	monosaccharide transmembrane transporter activity	molecular function	43	7	0.000292	0.039545	0.0166
GO:0015149	hexose transmembrane transporter activity	molecular function	43	7	0.000292	0.039545	0.0166
GO:0015749	monosaccharide transport	biological process	43	7	0.000292	0.039545	0.0166
GO:0015758	glucose transport	biological process	43	7	0.000292	0.039545	0.0166
GO:0019852	L-ascorbic acid metabolic process	biological process	43	7	0.000292	0.039545	0.0166
GO:0051119	sugar transmembrane transporter activity	molecular function	43	7	0.000292	0.039545	0.0166
GO:1901476	carbohydrate transporter activity	molecular function	43	7	0.000292	0.039545	0.0166

B: Duplications (28 statistically significant terms)

GO ID	GO Term Description	GO Term Type	CHD (#)	No CHD (#)	p Value	padjust_BH	FDR
GO:0044723	single-organism carbohydrate metabolic process	biological process	41	3	4.13E-06	0.003262	0
GO:0005975	carbohydrate metabolic process	biological process	44	4	5.68E-06	0.003262	0
GO:0005996	monosaccharide metabolic process	biological process	40	3	6.21E-06	0.003262	0
GO:0055085	transmembrane transport	biological process	36	3	3.11E-05	0.004497	0.0005
GO:0044281	small molecule metabolic process	biological process	46	6	3.55E-05	0.004497	0.0004
GO:0005355	glucose transmembrane transporter activity	molecular function	34	3	6.85E-05	0.004497	0.0016
GO:0006766	vitamin metabolic process	biological process	34	3	6.85E-05	0.004497	0.0016
GO:0006767	water-soluble vitamin metabolic process	biological process	34	3	6.85E-05	0.004497	0.0016
GO:0007275	multicellular organismal development	biological process	34	3	6.85E-05	0.004497	0.0016
GO:0007276	gamete generation	biological process	34	3	6.85E-05	0.004497	0.0016
GO:0007283	spermatogenesis	biological process	34	3	6.85E-05	0.004497	0.0016
GO:0008643	carbohydrate transport	biological process	34	3	6.85E-05	0.004497	0.0016
GO:0008645	hexose transport	biological process	34	3	6.85E-05	0.004497	0.0016
GO:0015144	carbohydrate transmembrane transporter activity	molecular function	34	3	6.85E-05	0.004497	0.0016

GO:0015145	monosaccharide transmembrane transporter activity	molecular function	34	3	6.85E-05	0.004497	0.0016
GO:0015149	hexose transmembrane transporter activity	molecular function	34	3	6.85E-05	0.004497	0.0016
GO:0015749	monosaccharide transport	biological process	34	3	6.85E-05	0.004497	0.0016
GO:0015758	glucose transport	biological process	34	3	6.85E-05	0.004497	0.0016
GO:0019752	carboxylic acid metabolic process	biological process	34	3	6.85E-05	0.004497	0.0016
GO:0019852	L-ascorbic acid metabolic process	biological process	34	3	6.85E-05	0.004497	0.0016
GO:0030154	cell differentiation	biological process	34	3	6.85E-05	0.004497	0.0016
GO:0048232	male gamete generation	biological process	34	3	6.85E-05	0.004497	0.0016
GO:0051119	sugar transmembrane transporter activity	molecular function	34	3	6.85E-05	0.004497	0.0016
GO:1901476	carbohydrate transporter activity	molecular function	34	3	6.85E-05	0.004497	0.0016
GO:0006082	organic acid metabolic process	biological process	36	4	0.000123	0.007459	0.0020
GO:0043436	oxoacid metabolic process	biological process	36	4	0.000123	0.007459	0.0020
GO:0048869	cellular developmental process	biological process	35	4	0.000178	0.010416	0.0025
GO:0044710	single-organism metabolic process	biological process	80	22	0.000483	0.027198	0.0071

C: Deletions (none are significant - top 5 are shown)

GO ID	GO Term Description	GO Term Type	CHD (#)	No CHD (#)	p Value	padjust_BH	FDR
GO:0031424	keratinization	biological process	274	136	0.038366	0.921843	1
GO:0044707	single-multicellular organism process	biological process	346	178	0.044478	0.921843	1
GO:0016021	integral component of membrane	cellular component	277	141	0.069179	0.921843	1
GO:0044430	cytoskeletal part	cellular component	86	39	0.112198	0.921843	1
GO:0044446	intracellular organelle part	cellular component	164	81	0.113582	0.921843	1

p Value = two-tailed Fisher's exact test
padjust_BH = B-H adjusted p Value
FDR = false discovery rate

Table S4: Mammalian Phenotype - Common CNVs

A: All CNV types (none are significant - top 5 are shown)

MP ID	MP Term Description	CHD (#)	No CHD (#)	p Value	padjust_BH	FDR
MP:0011091	complete prenatal lethality	47	8	0.000205	0.086782	0.1470
MP:0001488	increased startle reflex	43	7	0.000292	0.086782	0.0273
MP:0004264	abnormal extraembryonic tissue physiology	43	7	0.000292	0.086782	0.0273
MP:0004265	abnormal placental transport	43	7	0.000292	0.086782	0.0273
MP:0010038	abnormal placenta physiology	43	7	0.000292	0.086782	0.0273

B: Duplications (14 statistically significant terms)

MP ID	MP Term Description	CHD (#)	No CHD (#)	p Value	padjust_BH	FDR
MP:0001488	increased startle reflex	34	3	0.000068	0.011375	0.0013
MP:0003088	abnormal prepulse inhibition	34	3	0.000068	0.011375	0.0013
MP:0004264	abnormal extraembryonic tissue physiology	34	3	0.000068	0.011375	0.0013
MP:0004265	abnormal placental transport	34	3	0.000068	0.011375	0.0013
MP:0009142	decreased prepulse inhibition	34	3	0.000068	0.011375	0.0013
MP:0010038	abnormal placenta physiology	34	3	0.000068	0.011375	0.0013
MP:0011095	complete embryonic lethality between implantation and placentation	34	3	0.000068	0.011375	0.0013
MP:0011091	complete prenatal lethality	37	4	0.000085	0.012442	0.0011
MP:0009850	embryonic lethality between implantation and placentation	36	4	0.000123	0.014403	0.0013
MP:0011090	partial perinatal lethality	36	4	0.000123	0.014403	0.0013
MP:0001363	increased anxiety-related response	34	4	0.000258	0.02158	0.0016
MP:0001486	abnormal startle reflex	34	4	0.000258	0.02158	0.0016
MP:0001961	abnormal reflex	34	4	0.000258	0.02158	0.0016
MP:0004994	abnormal brain wave pattern	34	4	0.000258	0.02158	0.0016

C: Deletions (none are significant - top 5 are shown)

MP ID	MP Term Description	CHD (#)	No CHD (#)	p Value	padjust_BH	FDR
MP:0000278	abnormal myocardial fiber morphology	101	46	0.092162	0.967473	1
MP:0004087	abnormal muscle fiber morphology	101	46	0.092162	0.967473	1
MP:0004937	dilated heart	101	46	0.092162	0.967473	1
MP:0002972	abnormal cardiac muscle contractility	107	50	0.109976	0.967473	1
MP:0000274	enlarged heart	107	50	0.109976	0.967473	1

p Value = two-tailed Fisher's exact test
padjust_BH = B-H adjusted p Value
FDR = false discovery rate

Table S5: qPCR primers used in CNV validation

Primer Name	Primer Sequence 5' -> 3'
RPPH1F (control)	GCGGACGGAAGCTCATCAG
RPPH1R (control)	TCAGACCTTCCCAAGGGACAT
ASTN2e10F	ACTCACCCGCAGAACATGAAG
ASTN2e10F	TGCCCCTGCCCTACGAT
ASTN2e12F	CCTGTGGAGAGGAGCAGAGAGA
ASTN2e12R	GCCCTGTAGTCCTCAGCAGTCT
ASTN2e16F	CCTGAAATGAGCTGGTCATCTG
ASTN2e16R	CATCACCTACCTCTCAGGTTTGC
ASTN2i9F	AGGAGGAGCCCAGGATTAGTG
ASTN2i9R	AAGAACATCTCACATTTTGGGAGAA
ASTNi16F	TGTTCAAAACAACAGCACAAGGT
ASTNi16R	GATGATGCCTTCTGCCTAAGCT
CDH13e1F	TCCCACGGAAAATATGCTCAGT
CDH13e1R	CCGGCGGCGTTTTCA
CDH13i1aF	GTGGGCACCACTTGAGACAGT
CDH13i1aR	GGTTTACCGGCCAGACACA
CDH13i1bF	ACGTGAGCCCACCTGTGAA
CDH13i1bR	AGCTTCCTGGTGCTGGACAT
CDH13e2F	GAAAGTTAGCAGGGCAAACACA
CDH13e2R	CAGCACCTGAGAAAACCAAAGC
CDH13e5F	CACTGGAAAGGGAGTGGATCA
CDH13e5R	CGCTCCCTGTGTTCTCATTGA
CDH13i5aF	AAAAGAAGAAAGGCAGGATAATCAAT
CDH13i5aR	CCATCCAAATGCAAGGTCATAA
CDH13i5bF	GGGATCCCAGCGAGCATAC
CDH13i5bR	GGTGGTTGGAGAGGACTTATTCC
CDH13e6F	TGCCTCTGGAAGTCATTGTGAT
CDH13e6R	GGGCCTTCCCGAAAGATC
COL4A1+205kbF	AGGGCCACAGAGACTTGGAA
COL4A1+205kbR	TGTCCTGACCCCATCCTATCA
COL4A1+212kbF	CATCACCTGGAAAAAAGGTCTGA
COL4A1+212kbR	GCCTGGGCTCACTCTCTTCA
COL4A1-222kbF	CCCTGGTGGTGAGGGAAAC
COL4A1-222kbR	AGTGTGGCTGATCAGACATTGG
COL4A1-227kbF	GAGGCCGAGCCCCTACA
COL4A1-227kbR	TTGACCGGCACACTCGTACA
COL4A1e49F	CAGCCAGTACGAGTAGTCATTTCG
COL4A1e49R	GCCCTTCCTGTTCTGCAATATT
COL4A1e50F	TGGCGTCTCCCCAGACA
COL4A1e50R	GGTGGTCCTCGCTGTGGAT
COL4A1i49F	CAAAAGCAGCAGCTTCAAGTACA
COL4A1i49R	ACACATCGAAGCTCAGCATGA
CXADR-275kbF	GTGGATTGAGCTACCCTGCAA
CXADR-275kbR	CCAGACACCTCGGCTAGTCTTT
CXADR-148kbF	GGCTTAAAATGGTGGCTTTATCC
CXADR-148kbR	GAATAGAATGAAGGAAATTAATGGTTAAGG
CXADRe6F	GCTTCAAATAAAGCTGGACTAATTGC
CXADRe6R	TGATAAGACCAATGAGCGCTAGAG
CXADR+274kbF	TCTGACCCCCAGGACCTCTA
CXADR+274kbR	TGCCTTATGCTTTTCCAATTAAATC
CXADR+292kbF	GATGCTCTTTCCCCTTTGCTT
CXADR+292kbR	GGTAAATCTCTTAGATGAGCCTGTTTG
DEFB-347kbF	CTCCTCTGCTCGTGCCTTTC
DEFB-347kbR	GGTGTTCAGGGTCCTGCTCTAG
DEFB-335kbF	AGCCACCATCACTGTCAATCC
DEFB-335kbR	CCCCAGAGCCCAAATTCTC
DEFBe1F	TGACGGCCGAGGTGAGA
DEFBe1R	CCCCGTCGGTATAAGGATGA
DEFB+6kbF	TGCCTTGGTGGCCTCTAGTG
DEFB+6kbR	AACACGCTTGCAGCTCTGACT
DEFB+38kbF	CCCATGTACTCCCTGCAGTATTT
DEFB+38kbR	TTCCGCTCGTCCGTCAA
GNA14i2aF	GAGCATCCTCCCAGGGAAAG
GNA14i2aR	CCCCTTAAATTTTGTGCCCTAGA
GNA14i2bF	GGAGAATGCTCACGGGTACAG
GNA14i2bR	CCCCACCCAGAGGTTCAGT
GNA14i2cF	CATTCATCAGGCATGCGTGTA
GNA14i2cR	CTCGAATAATGCCTGGCAAAG
GNA14i2dF	CGAGCACTAAGACCTCCTCGTT
GNA14i2dR	TCCTGTCTGGTGTACCTGCACTT
GRB2-19kbF	CTCTCCTGAGCATACCTGTATTGC
GRB2-19kbR	GGTCGTAATCTACAAAGCGTTTTG
GRB2-231F	TCCCCCGTCTAATCACTTTATTTT
GRB2-231R	GCCCCTACCCATCCAACTG
GRB2e3F	GAAGCCGTCTTTTCCATTAAGC
GRB2e3R	CAGGTTTTGAACGAAGAATGTGAT
GRB2+673kbF	GGGCTGACCATGCTACAGAGA
GRB2+673kbR	GGGAGGCACCAAAGCAGAA
GRB2+695kbF	CTGCTAGAATTGCCCTTTCCA
GRB2+695kbR	GCGCAGGTCCCTCTGAAAA
GRM7i7F	GCAAAGATCAGAAAGGATGATTCA
GRM7i7R	CTTGCCAGGAGCAAAACAGAT
GRM7e8F	GCCTTCATTCCAATTTTTTTTGG
GRM7e8R	TGATGTGCATTTTCACTTACCTTTTC
GRM7e14F	CCAAAGGGAGTGTTGAAACTCAA
GRM7e14R	GGCTCTCAGTGGTCCATTCTAAA
GRM7+370kbF	TCAAATCTGGCTTCCAGGAACT
GRM7+370kbR	GAGGAAGATCTAGCCAATGGAAGA
GRM7+373kbF	TGGAGGAGGGAGAAACCATTT
GRM7+373kbR	GAGAATCACCTCCTTTGGATGTCT
KCNMA1e1F	AGAGGCCCCCGAAGAAAGT
KCNMA1e1R	GCATGTGGTGGGCTTTCC
KCNMA1i1aF	GGGTGTCATTAGGGTCTGTGGAT
KCNMA1i1aR	GGGTATTTGAACCGGGTGATT
KCNMA1i1bF	GACAGGCATGGTGCTTCGA
KCNMA1i1bR	GCGAGGCTTCTCCAAGTGTT
KCNMA1i1cF	CAGGAAGAGCCACCAAGAGTTT
KCNMA1i1cR	AAGGTGCCTGTGGGCATTAG
LTBP1e2F	CAGCGATCTTGCTTTGTTTCAG
LTBP1e2R	GGCCTTACTCCAGCCATGAC
LTBP1e4F	GACCCAGGAATACGTGCTCAA
LTBP1e4R	GTGGACTGCTCTCCTGAAATCC
LTBP1e3F	CCATGTCAGAATGGAGGGATGT
LTBP1e3R	TGGTCCCTGGTTTACACACACA
LTBP1i3F	CCCTCGGCTTTGAACCAA
LTBP1I3R	AAGGATTCCAGCAACTTGAAGGT
LTBP1+550kbF	TGCAGCCTGCCTCATCTG
LTBP1+550kbR	CAACAAACGAGGCATACACTACAAG
MAPK9+56kbF	GGGCGTCAGGTATGCAAACT
MAPK9+56kbR	TTCCTCAGGGTTCCTTTCAGTATT
MAPK9-41kbF	CAGGAGGAATCCACAGTGTGTCT
MAPK9-41kbR	CGCCACCCCAACCCTATT
MAPK9e4F	GCTCCATGTGAATAACCTGACATAA
MAPK9e4R	CCTTTTCTCCCTCAGGTATTTGG

Primer Name	**Primer Sequence 5' -> 3'**
MAPK9-161kbF	TTGACGGCACTAGAAAGCAAAG
MAPK9-161kbR	GCGCGGGCTTCCAAGT
MAPK9-180kbF	GAGTCGGTGGTGAAGCACAA
MAPK9-180kbR	CATGAGAGAGGAAGGCCTGAGA
MTHFD1e1F	GGGTTGGGTTGTCCTGCTT
MTHFD1e1R	CGATGGACACCACCAATATCG
MTHFD1e9F	GTTTTGATTTCTCCCCCACTTG
MTHFD1e9R	CACCCACAACTTTTCTCCCATT
MTHFD1e27F	TGCATGTCTGTTTACTTTAGTGACGTT
MTHFD1e27R	TGCAACACCAAGATGGCAAA
MTHFD1+85kbF	CAATTTATCAGTGCTGGTGTCACA
MTHFD1+85kbR	CATGAAACACCCTTAAATGCTCAT
MYH11e1F	CCCAGCCTTCCCCAACTC
MYH11e1R	TCACAATGTCGTTGGCTTTTCT
MYH11e42F	ATAACTCTACGTCCTCCAGACCTTCT
MYH11e42R	AACCCACAGGCGAGGAAAC
MYH11e2F	TGGTCCCCTGTGGAATAAGG
MYH11e2R	CTTCCGGGCTTGGTTGTG
PAX8e13F	CGGGAAAGAAAGGCGAGTTT
PAX8e13R	GGCTTGGGCTGTGCTTTG
PAX8e2F	CAGCCCTCCATGGCCTAAG
PAX8e2R	CAAACTCCTACCTGACCCTGACA
PAX8e1F	CTGGGCCCGGTGTCTCT
PAX8e1R	GGTGATGCCGGGTGGAT
PAX8-50kbF	TGCAAATCATACCCGCTTGA
PAX8-50kbR	AATGCTCATTAGCCCTGATTGG
PRKG1i3aF	CCTGGGAGGGTGGAGTATTACA
PRKG1i3aR	CACAAAGGAAAACACCACACACA
PRKG1i3bF	CCTATGCGGGAGTTTGAACTTG
PRKG1i3bR	AAAAGAGCCCTGAACACTAAAAATTT
PRKG1i3cF	AAATGGCTGTCACAATGGTACAA
PRKG1i3cR	CGTTTCCAGACAGCGTTCAG
PRKG1i3dF	GCTGCCAGTGGGTAAAAAGG
PRKG1i3dR	AATCACGGAAGTCACAAAGCATT
PTPRDe11F	CAGCGAGTCTGTCCGATCTG
PTPRDe11R	CCCGCTTTCAGGCTCTGA
PTPRDi10aF	TGCCCATTTCCTTGGCTTT
PTPRDi10aR	AGGAGGGAGAAGGCCCTACA
PTPRDi10bF	AATGGTGTGAGAGGTGAACAACA
PTPRDi10bR	TTGTGGTGGAGGGAGCAAAT
PTPRDi10cF	TCTCATTTGCGTTCCTGAAAAA
PTPRDi10cR	GCTGACCGTTTCTTTGTACATGAA
RCAN1e1F	GCATCCTGTTTGGACAGCAA
RCAN1e1R	CCTGTCTGCCTGCAAGCAT
RCAN1e4F	TCGCTGCGTGCAATTCAT
RCAN1e4R	CTGAGTGACCCTGCGATTATTTT
RCAN1-58kbF	GGATTTTTCCTGGTGATTGGAA
RCAN1-58kbR	TGCTGAGTTCACACAGTTGCTTAA
RCAN1-170kbF	GGTGGTGGCTGGCTCTGT
RCAN1-170kbR	TGTCCTTCAATTGGACTTTTTGC
RCAN1-163kbF	GAACCAGGCACAGAGAAAGTAGAAG
RCAN1-163kbR	GGGCATTGAGAATCTCTGGTAATT
RCAN1i2F	TGGGCTCAGGAGTGTATTTCTCT
RCAN1i2R	TCCCTGTGGCCAAGCAA
RHOU+70kbF	CTGGGAAAGTGGACGTTGGT
RHOU+70kbR	AGCAAGTATGAAACCCCTCCTAAA
RHOU+80kbF	AGCAGCCTTTGCCTATGCA

Primer Name	**Primer Sequence 5' -> 3'**
RHOU+80kbR	GCTGATTCTGGGAACCAGTTATG
RHOU-62kbF	AGGTAGCAGCAGCCCGATT
RHOU-62kbR	AGTAAAGAATAAGCTGTGGCAGTTCA
RHOU-82kbF	AGGCCTTTTGGTGCTTACCA
RHOU-82kbR	TCTGCCTCCTCTCTCTATTCAGTCA
RHOUe2F	CGTGAGACTCCAACTCTGTGACA
RHOUe2R	CCCAGCACTGAGCTGTAACG
SLC2A14-5'F	CATGGCCTTCAGAAGAAGTTAAGAT
SLC2A14-5'R	ACACTGGTTGCTGTAGCAGAAACTA
SLC2A14i7F	AGCATCATCTGTCCAGTCTTCATC
SLC2A14i7R	GCCTAATGTCCAGAGTGGGAAT
SLC2A3e9F	GGCCACAATAAACCAGGGAAT
SLC2A3e9R	GCTATCTTGGTCTTTGTAGCCTTCTT
SLC2A3e1F	TTTTCAGCCAACAAAACCTTCA
SLC2A3e1R	TCCTGAGGACGTGGAGAAAACT
SLC2A3-3'F	CACAACAGAGGCAAGGGTACATAT
SLC2A3-3'R	AAGAAGGAGCAAATGCCAAGTT
TRIM54+102kbF	CCCGTCCTCCAGTGTTTTCA
TRIM54+102kbR	GGGTTTGCGGAGTCTGAGAGT
TRIM54+126kbF	TCTCAGGTGAATCAACGGAATG
TRIM54+126kbR	CCTCTGTATCCATGGCCAGGTA
TRIM54-198kbF	TCTCACAGGTCCTCAAGGTGAA
TRIM54-198kbR	TCACTTCTCCCTCCAGAATCAAA
TRIM54-242kbF	TGAGGGTTGTGGGTCTAAGCTT
TRIM54-242kbR	TGAGTATGCACGTGAGTAGCTGTGT
TRIM54-255kbF	GCCATGTGTCCCTCAACTGA
TRIM54-255kbR	GGTGCATGGGAGGTGGAA
TRIM54e4F	TTCCTAGGACCCTTCATGCTTAAG
TRIM54e4R	GGAGTGCAGCGGCCTAGAG
15q11.2aF	GTGTGCTGGAGAAAGGATGCT
15q11.2aR	CCATCAGGCAGGCAAATAATC
15q11.2bF	CCTGGCTGTGGAAGGTGACT
15q11.2bR	TGTCAGTGACCAAGCCATCTG
15q11.2cF	TCCGCCATGTAAATTACGAGTTC
15q11.2cR	GATGTGGCCCCCGACTCT
15q11.2dF	GCAACAAGGCCTGCAGGTAT
15q11.2dR	TGTGTCTCCAAATGGGCAGTT
17q12aF	CATGGCGTCGCTCAAATGTA
17q12aR	TGGGCTTCTCCAAGCAGATC
17q12bF	CCCCCTGCAGCAGCAA
17q12bR	GAGCTAAATTTCCGCAGCAATAA
17q12cR	CCCATCTTCCAGCCCTGAA
17q12cR	TCCTCGCACTCGCCACTAG
17q12dF	GGGCTTGGTTGGTGGGTAA
17q12dR	GAGGGCCAGGGATCATCAC
18q11aF	GGTTGTCCCTGACTCATTGTCA
18q11aR	ACCATTCTGTGCCATCATCACA
18q11.2bF	CACATGCGCTCCACAAGTG
18q11.2bR	GCAGACACACACAAATTCAAATATTACA
18q11cF	CCGCTGTGACTGCTTTCCA
18q11cR	AGAACATCATGGTGGCTTTCAA
18q11dF	TGGATAGGCTCATCAGGATCAA
18q11dR	TGCAAACCCACCATGTCATC
18q11eF	CCATGTGCCTGCTGCTCTT
18q11eR	CCTGCCAAAGCCCCTTTAG
18q11fF	GCAGAATTCGGAACTGAGAAGAC
18q11fR	TTGCACGCAAATCCTGTGA