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Supplemental Data

Genome-wide Transcriptome Profiling Reveals the Functional Impact of Rare De Novo and Recurrent CNVs in Autism Spectrum Disorders

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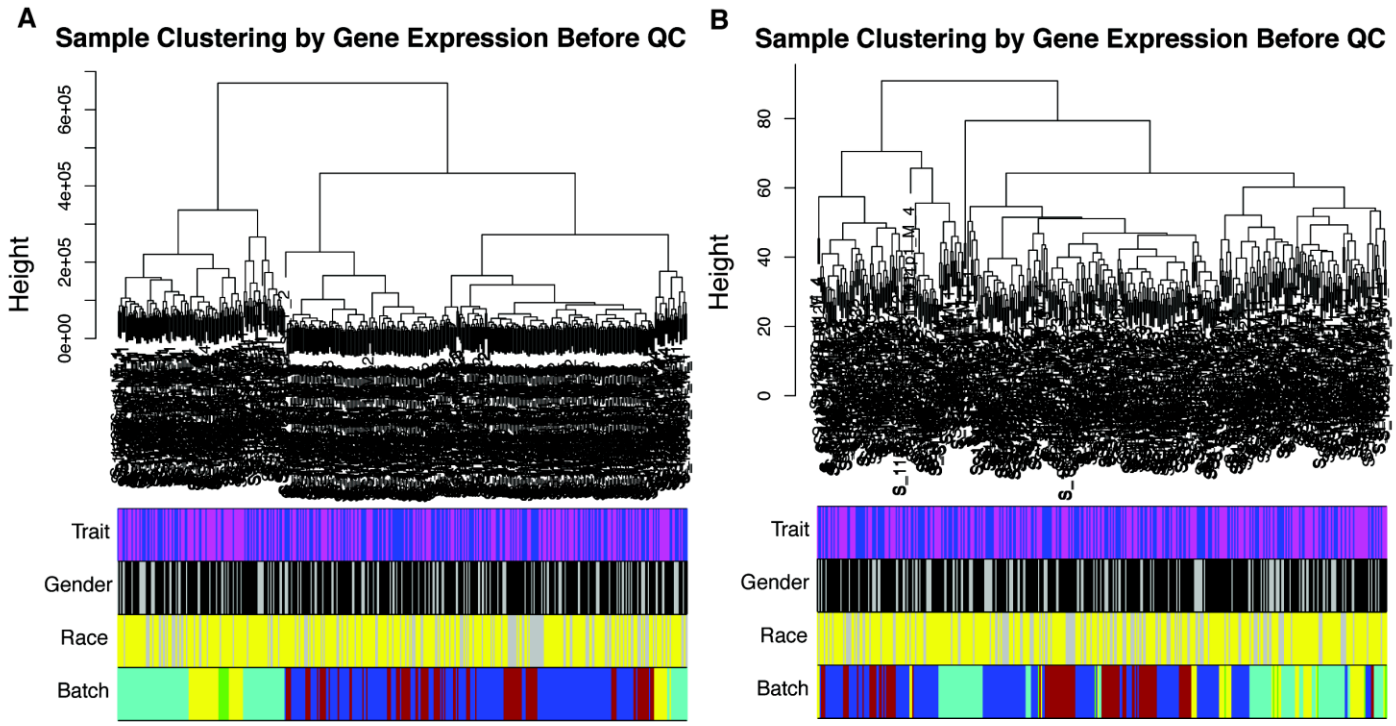


Figure S1. Data Preprocessing to Remove Outlier Chips and Correct for Batch Effects

(A) Hierarchical clustering of samples before data quality control (QC). Color bars show the trait (case: magenta; control: cyan), gender (male: black; female: grey), race (Caucasian: yellow; non-Caucasian: grey) and batch of each sample. Batch is defined based on the hybridization date.

(B) Hierarchical clustering after quality control including removing outlier chips, quantile normalization and combat for removing batch effects.

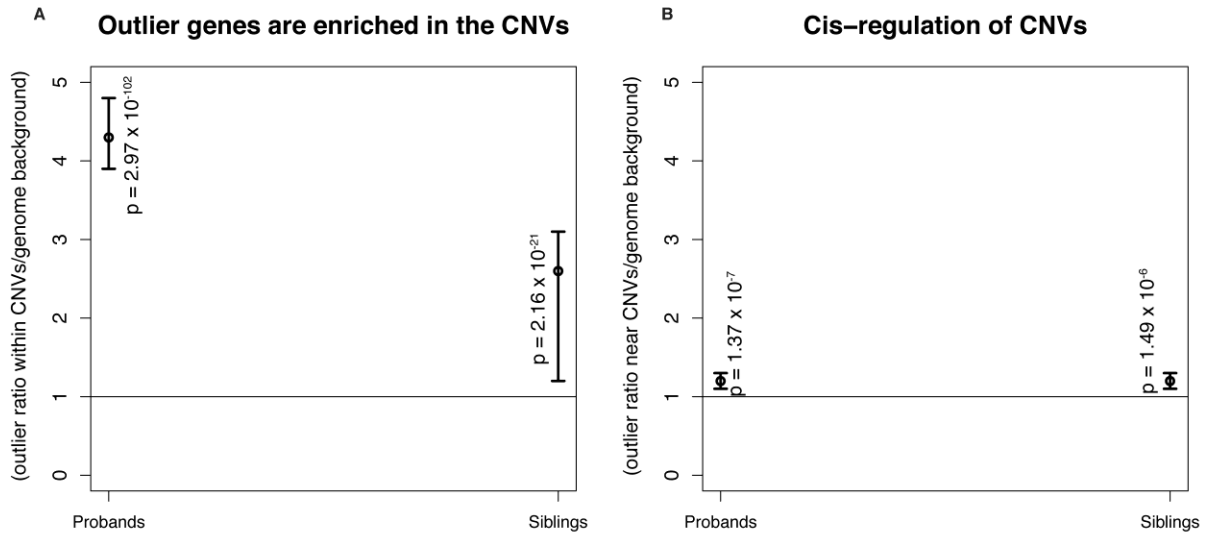


Figure S2. CNVs Affect the Expression of Genes within CNVs and up to 500 kb Surrounding Them

(A) Odds ratio (OR) of the percentage of dysregulated genes (2SD) within CNVs compared to the percentage of dysregulated genes out of 9,524 genes (11,150 expressed probes) across the genome (background). Bar height shows the 95% confidence interval (CI). The CNVs comprise all CNVs each individual has, including both rare and common CNVs.

(B) Odds ratio of the percentage of dysregulated genes in the 500 kb surrounding region of probands and siblings compared to the ratio of dysregulated genes in the genome background. The OR is significant for both probands and siblings for genes within CNVs, as well as genes within 500 kb nearby (p value by Fisher's exact test).

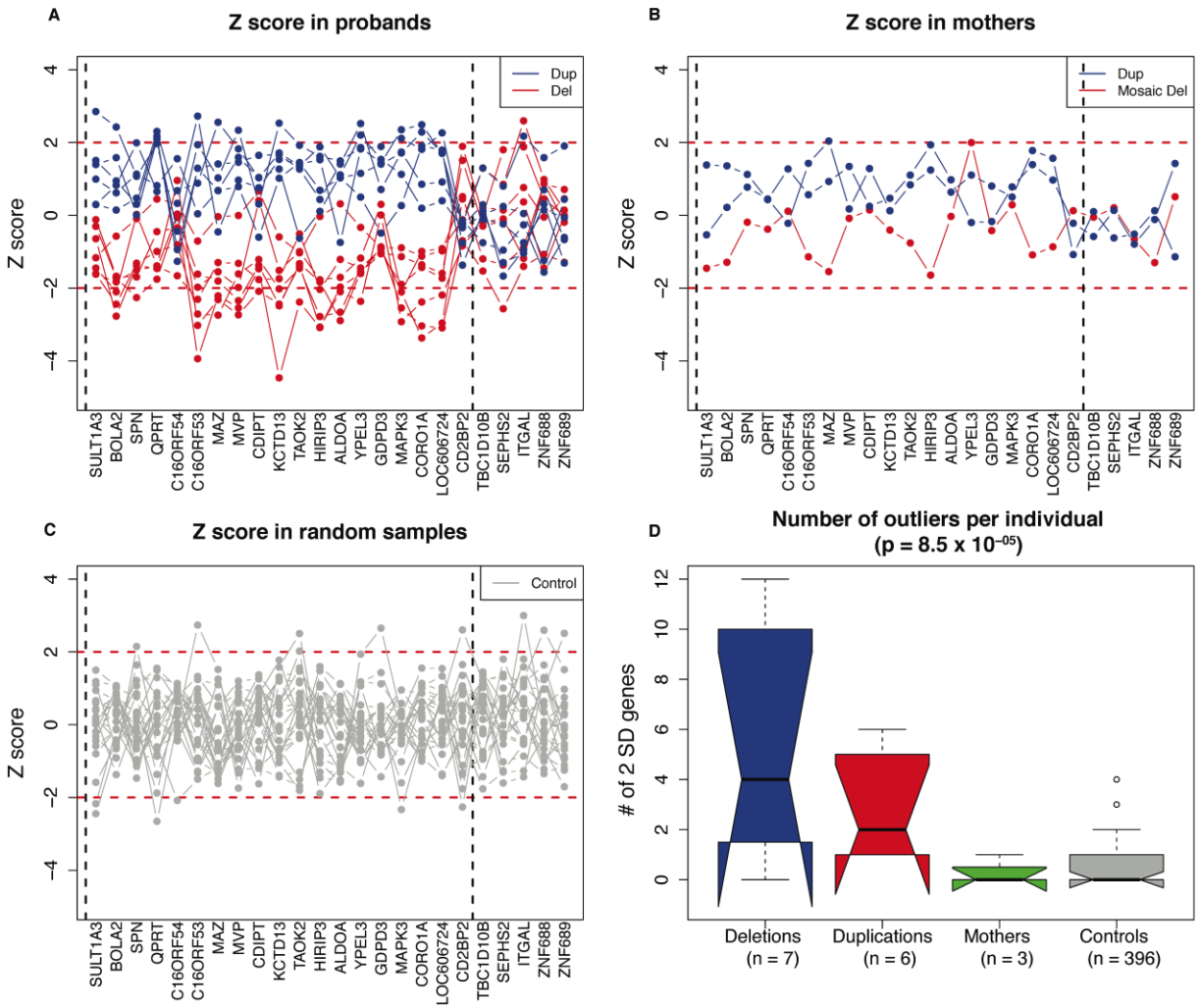


Figure S3. Dysregulation of Genes within 16p11.2 and the Closely Surrounding Region in Probands, Carriers, and Controls

(A) *Z* scores of 18 expressed genes within 16p11.2 and 6 expressed genes residing 500 kb upstream or downstream in probands (7 deletions: red; and 6 duplications: blue). Genes on x-axis are aligned based on their location on chromosome. The 16p11.2 boundaries are shown with vertical dashed lines. 2 SD is used as the cutoff to define outlier genes (horizontal dashed lines).

(B) *Z* scores of the same 24 genes in 3 mothers who carry the 16p11.2 events, but are unaffected (2 duplications: blue; and 1 mosaic deletion: red).

(C) *Z* scores of the same 24 genes in 20 randomly picked individuals (either probands or siblings) without known 16p11.2 events.

(D) The boxplot shows the number of outlier genes within 16p11.2 region per individual in different sample groups (p value = 8.5×10^{-5}, Kruskal-Wallis test).

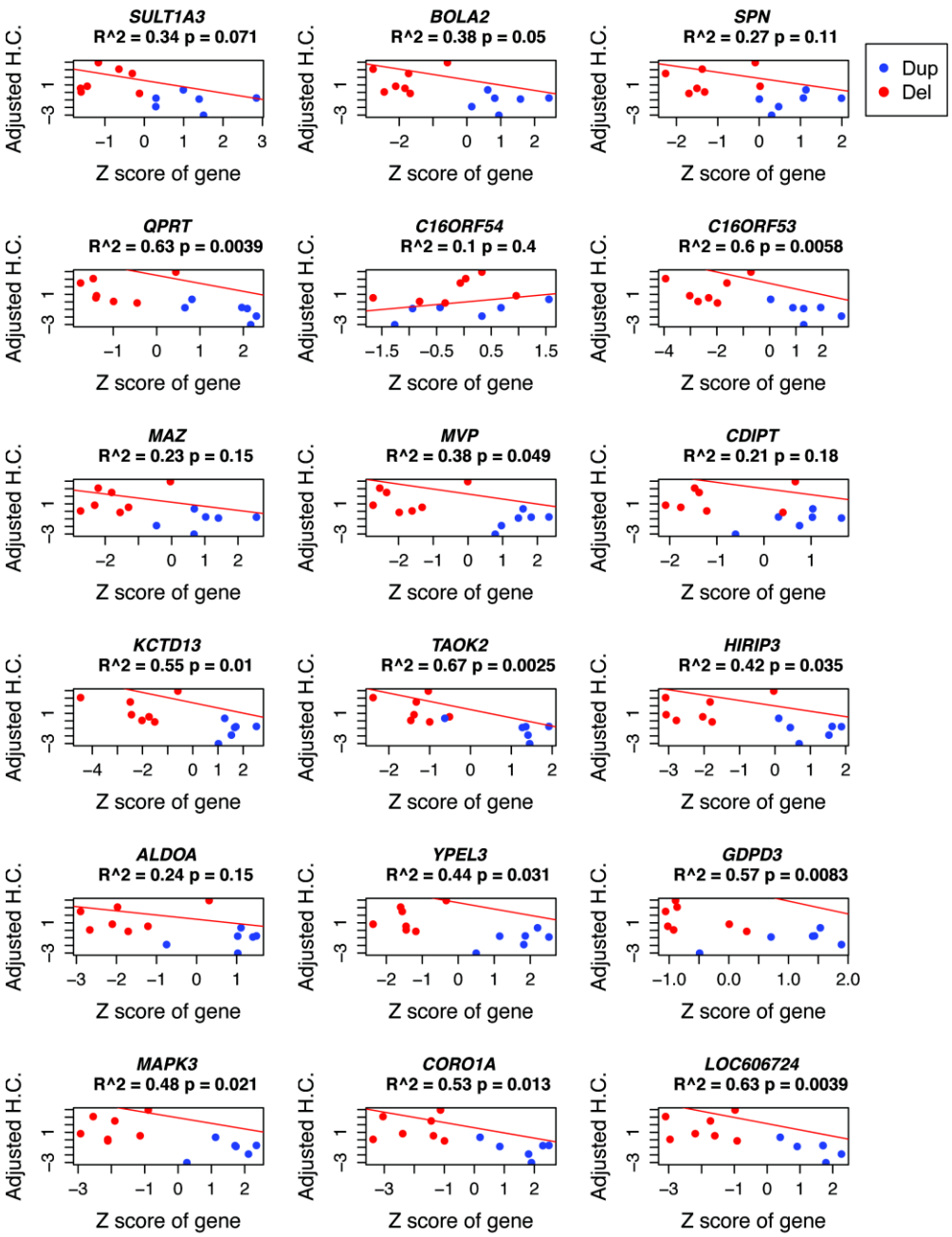


Figure S4. Correlation of Head Circumference and Gene Expression within 16p11.2

The Z scores of 18 expressed genes within 16p11.2 region (*x* axis) and adjusted head circumference (HC; *y* axis) are shown. A multivariate linear regression model is fitted (variables used are standardized expression value (*Z* score), age and gender; Material and Methods). R-square of the linear regression model and p-value of the correlation between standardized expression value and HC is shown.

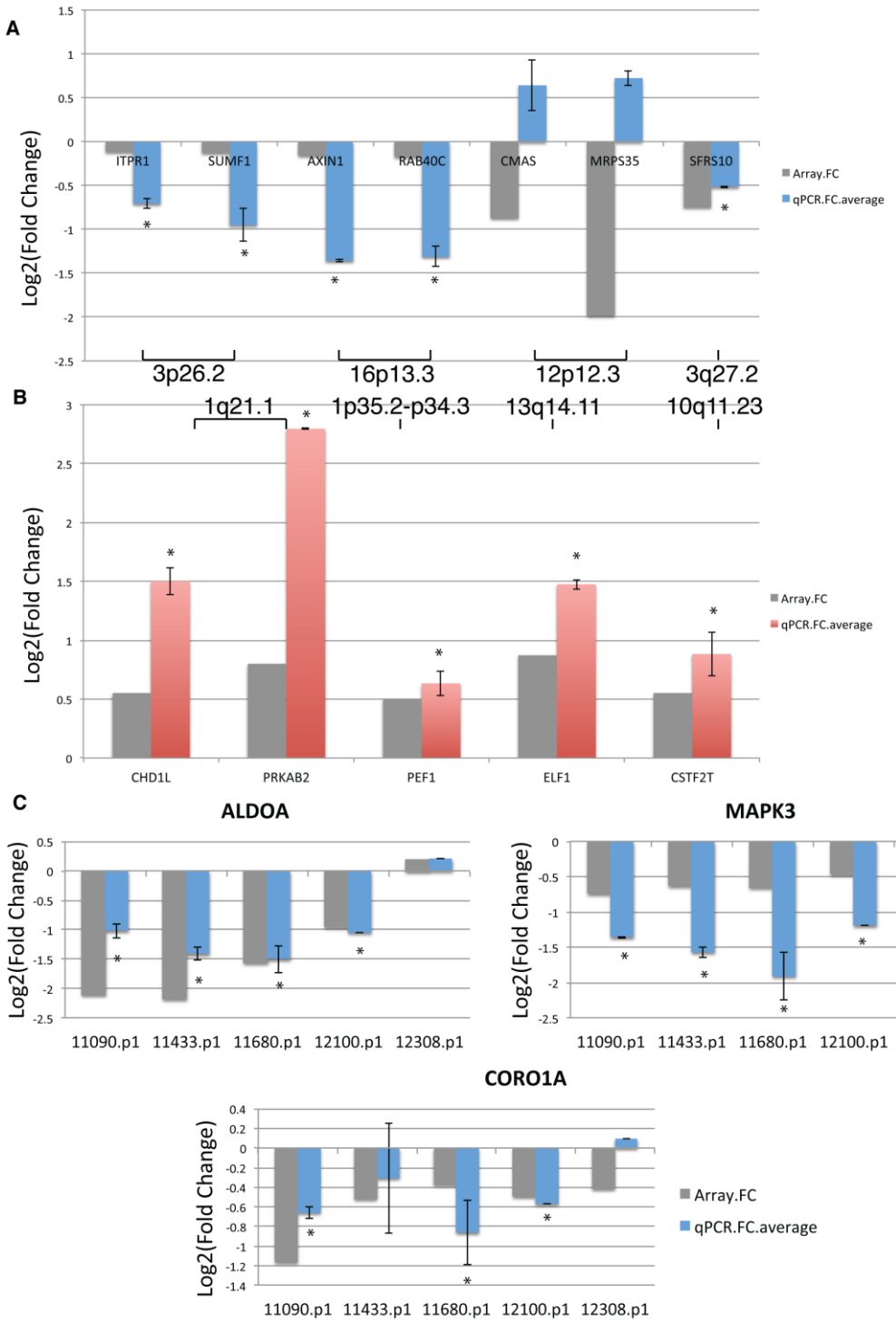
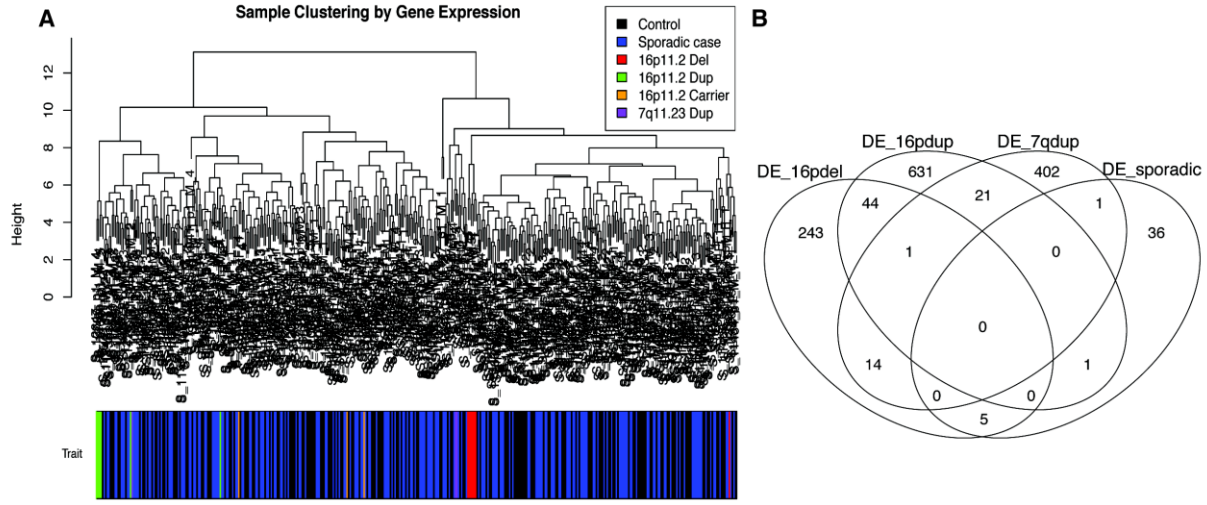


Figure S5. Confirmation of the Outlier Genes by qRT-PCR

(A) Eight down-regulated genes in 4 probands tested by qRT-PCR (Material and Methods). Seventy-five percent of them are validated, showing at least 1.3-fold change (*). The CNV harboring each gene is shown.

(B) Five up-regulated genes in 4 probands are validated by qRT-PCR. One hundred percent of them are validated (* highlights genes with at least 1.3-fold change by qRT-PCR).

(C) Three genes down-regulated in 16p11.2 deletions are validated in 5 probands. Results represent the log 2 fold change of each gene on microarray and qRT-PCR (1.3-fold change: *).



C

	brain DEX (444)	15q11-13dup (80)	FMR1-FM (120)	Seno et al (33)	Hu et al (45)
sporadic(43)	*CEBPD*	NA	NA	NA	*HMBOX1*
16pdel(307)	*D4S234E, HERC6,ITPR1, PREPL,PRKCE, PTK2B,TNNT2*	NA	ZFP36	NA	NA
16pdup(698)	*RTPRT,SLC29A1*	*RIMS3,NR3C1*	*RIMS3,NR3C1*	NA	NA
7q11.23dup(439)	*LCMT1,PRKCB1,PRKCE*	*CYF1P1,RIMS3, CHST12,PRKCB1*	*PSEN2,GNG2, MBTPS1,RIMS3, CHST12, PRKCB1*	*LEF1,MFGE8, PLD1*	*SRD5A1*

Figure S6. Differential Expression Analysis in the Simons Simplex Collection (SSC)

(A) Sample clustering analysis for all sporadic cases (blue), controls (black),16p11.2 deletions (red), 16p11.2 duplications (blue), 16p11.2 carriers (orange) and 7q11.23 duplications.

(B) Venn diagram of the overlap of DEX genes ($p < 0.05$) identified in different groups (DEX, differentially expressed genes).

(C) DEX overlap with autism brain[1], recurrent events: 15q11-13dup, FMR1-FM [2] and LCLs [3; 4].

Table S1. Sample Inclusion Information

11345.s1	11868.p1	11511.p1	11393.p1	13128.p1	11066.p1	11540.p1	11030.s1	12261.p1	11428.p1	11337.s1*
11046.p1	11868.s1	11511.s1	11393.s1	12958.p1	11066.s1	11540.s1	11510.p1	12261.s1	11428.s1	11244.p1*
11046.s1	11300.p1	11425.p1	12295.s1	13195.p1	11276.s1	11274.p1	11510.s1	11220.p1	11004.p1	11244.s1*
11581.p1	11300.s1	11425.s1	12420.p1	12044.s1	11276.p1	11274.s1	11587.p1	11220.s1	11004.s1	11102.p1*
11581.s1	11014.p1	11329.p1	12420.s1	12435.p1	11156.p1	11098.p1	11587.s1	11316.p1	11551.p1	11102.s1*
11479.p1	11014.s1	11329.s1	12523.p1	12581.p1	11156.s1	11098.s1	11063.p1	11316.s1	11551.s1	11443.p1*
11479.s1	11473.p1	11519.p1	12523.s1	11121.p1	12073.p1	11000.p1	11063.s1	11029.p1	11085.s1	11443.s1*
11364.p1	11473.s1	11519.s1	11334.p1	11121.s1	12073.s1	11000.s1	11424.p1	11029.s1	11285.p1	11461.p1*
11364.s1	11348.p1	11080.p1	11135.p1	11557.s1	12032.p1	11839.p1	11424.s1	11413.p1	11285.s1	11461.s1*
11962.p1	11348.s1	11059.p1	11356.p1	11429.p1	12451.p1	11839.s1	11578.p1	11413.s1	11089.p1	11533.p1*
11962.s1	11275.p1	11059.s1	11356.s1	11429.s1	12351.s1	11247.p1	11578.s1	11254.p1	11089.s1	11533.s1*
11390.p1	11275.s1	11879.p1	11411.p1	11291.p1	11524.p1	11247.s1	11333.p1	11254.s1	11696.p1	11426.p1*
11475.p1	11523.p1	11879.s1	11411.s1	11291.s1	11524.s1	11091.p1	11333.s1	11577.p1	11216.p1	11426.s1*
11475.s1	11523.s1	11723.p1	11267.p1	11376.p1	11489.p1	11091.s1	11490.p1	11577.s1	11216.s1	11457.p1*
11353.p1	11189.p1	11723.s1	11267.s1	11376.s1	11489.s1	11208.p1	11490.s1	11303.p1	11387.p1	11457.s1*
11353.s1	11189.s1	11629.mo	11028.p1	11550.s1	11458.p1	11208.s1	11327.p1	11303.s1	12297.p1	11579.p1*
11520.p1	11537.p1	11629.p1	11028.s1	11417.p1	11458.s1	11731.p1	11327.s1	11083.p1	12297.s1	11579.s1*
11520.s1	11537.s1	11168.p1	11546.p1	11417.s1	11610.p1	11731.s1	11406.p1	11083.s1	11480.p1	11442.p1*
11484.p1	11178.p1	11168.s1	11546.s1	11328.p1	11610.s1	12184.p1	11406.s1	11152.p1	11480.s1	11442.s1*
11484.s1	11178.s1	11071.p1	11410.p1	11328.s1	11345.p1	12184.s1	11412.p1	11152.s1	11718.p1	11418.p1*
11193.p1	11445.p1	11071.s1	11410.s1	11634.p1	11948.p1	12308.p1	11412.s1	11242.p1	11718.s1	11418.s1*
11193.s1	11445.s1	11113.p1	11415.p1	11634.s1	11948.s1	12417.mo	11435.p1	11242.s1	11090.p1	11076.p1*
11857.p1	11469.p1	11113.s1	11415.s1	11765.p1	11990.p1	12417.p1	11057.p1	11197.p1	12044.p1	11076.s1*
11857.s1	11469.s1	11554.p1	11809.p1	11765.s1	11990.s1	11006.p1	11057.s1	11197.s1	11342.p1*	11293.p1*
11399.p1	11450.p1	11554.s1	11998.p1	11680.p1	11979.p1	11006.s1	11452.p1	11129.p1	11342.s1*	11293.s1*
11399.s1	11450.s1	11379.p1	11998.s1	12346.s1	11979.s1	11509.p1	11452.s1	11284.p1	11186.p1*	11062.p1*
12096.p1	11466.p1	11379.s1	12083.p1	12647.p1	12014.p1	11509.s1	11301.p1	11284.s1	11186.s1*	11062.s1*
12096.s1	11466.s1	11383.p1	12083.s1	12399.p1	12014.s1	11207.p1	11301.s1	11325.p1	11338.p1*	11007.p1*
11824.s1	11420.p1	11383.s1	12279.p1	12399.s1	12048.p1	11207.s1	11555.mo	11325.s1	11338.s1*	11007.s1*
11459.p1	11420.s1	11219.p1	12279.s1	11501.s1	12048.s1	11474.p1	11555.p1	11689.p1	11271.p1*	11335.p1*
11459.s1	11455.p1	11219.s1	12117.s1	11501.p1	12100.p1	11474.s1	11555.s1	11625.p1	11271.s1*	11335.s1*
11502.p1	11455.s1	11201.s1	12219.p1	11146.p1	12299.s1	11114.p1	11154.p1	11625.s1	11260.p1*	11191.p1*
11502.s1	11053.p1	11075.p1	12235.p1	11146.s1	12457.s1	11114.s1	11154.s1	12015.p1	11260.s1*	11191.s1*
11947.p1	11053.s1	11075.s1	12239.p1	11532.p1	11378.p1	11073.p1	11736.p1	12015.s1	11138.p1*	11482.p1*
11947.s1	11180.p1	11323.p1	12241.p1	11495.s1	11378.s1	11073.s1	11736.s1	12339.p1	11138.s1*	11482.s1*
12385.p1	11180.s1	11323.s1	12241.s1	11495.p1	11032.p1	11233.p1	11831.p1	12327.p1	11149.p1*	11005.p1*
12385.s1	11499.p1	11177.p1	12224.p1	11407.p1	11032.s1	11233.s1	11831.s1	12327.s1	11149.s1*	11005.s1*
12383.p1	11499.s1	11177.s1	12685.p1	11407.s1	11041.p1	11382.p1	12078.p1	12603.p1	11192.p1*	11427.p1*
12343.p1	11572.p1	11433.p1	12512.p1	11265.s1	11563.p1	11382.s1	12078.s1	12603.s1	11192.s1*	11427.s1*
12594.p1	11572.s1	11343.p1	12984.p1	11265.p1	11563.s1	11030.p1	12007.p1	12736.p1	11337.p1*	11337.s1*

* Samples can be included, while these families did not meet SSC Inclusion Criteria.

Table S2. GO Enrichment of Outlier Genes in Probands and Siblings

DAVID GO Enrichment of Outlier Genes in Probands

Term[a]	p Value[b]
protein modification by small protein conjugation or removal	0.00191
cytosol	0.00219
cartilage development	0.00241
protein folding	0.00283
ubiquitin ligase complex	0.00491
regulation of synaptic transmission	0.00498
regulation of transmission of nerve impulse	0.00498
regulation of neurological system process	0.00510
endochondral bone morphogenesis	0.00530
regulation of cellular amine metabolic process	0.00639
protein modification by small protein conjugation	0.00733
endoplasmic reticulum	0.00892
soluble fraction	0.00925
cell-cell adherens junction	0.00937
ligase activity, forming carbon-nitrogen bonds	0.00995
bone morphogenesis	0.01123
cell-cell junction	0.01237
spliceosomal snRNP biogenesis	0.01262
acid-amino acid ligase activity	0.01457
actin polymerization or depolymerization	0.01573

DAVID GO Enrichment of Outlier Genes in Siblings

Term	p Value
anatomical structure homeostasis	5.24×10^{-5}
hydrogen peroxide metabolic process	0.00489
tissue homeostasis	0.00663
response to inorganic substance	0.00663
protein amino acid dephosphorylation	0.01210
dephosphorylation	0.01410
phosphatase activity	0.01438
protein amino acid deacetylation	0.01515
Wnt receptor signaling pathway	0.01573
phosphoprotein phosphatase activity	0.01650
response to UV	0.01653
multicellular organismal homeostasis	0.01653
response to hydrogen peroxide	0.01764
protein tyrosine phosphatase activity	0.01796
cellular response to oxidative stress	0.01815
proton-transporting ATP synthase complex, coupling factor F(o)	0.01909
ATP biosynthetic process	0.02031
proton-transporting ATP synthase complex	0.02170
purine nucleoside triphosphate biosynthetic process	0.02183
histone methylation	0.02390

[a] Top 15 terms with p value smaller than 0.05 are listed.
[b] Uncorrected p value reported by DAVID GO.

Table S3. Known Autism Candidate Genes as Outlier Genes in Probands

Gene[a]	# of Proband (Up/Down)[b]	# of Sibling (Up/Down)[b]	Description	Location
PPP1R3F	3/1	0/0	protein phosphatase 1, regulatory (inhibitor) subunit 3F	Xp11.23
ADA	2/0	0/0	adenosine deaminase	20q12-q13.11
GLO1	2/0	0/0	glyoxalase I	6p21.3-p21.1
AUTS2	1/0	0/0	autism susceptibility candidate 2	7q11.22
CYFIP1	1/1	0/0	cytoplasmic FMR1 interacting protein 1	15q11.2
HRAS	1/0	0/0	v-Ha-ras Harvey rat sarcoma viral oncogene homolog	11p15.5
SLC1A1	1/0	0/0	solute carrier family 1 (neuronal/epithelial high affinity glutamate transporter, system Xag), member 1	9p24
UBE2H	1/0	0/0	ubiquitin-conjugating enzyme E2H (UBC8 homolog, yeast)	7q32
CASC4	0/1	0/0	cancer susceptibility candidate 4	15q15.3
CNTN4	0/2	0/0	contactin 4	3p26-p25
DPP6	0/2	0/0	dipeptidyl-peptidase 6	7q36.2
EGR2	0/1	0/0	early growth response 2 (Krox-20 homolog, Drosophila)	10q21.1
GPC6	0/1	0/0	glypican 6	13q32
JMJD1C	0/1	0/0	jumonji domain containing 1C	10q21.2
MEF2C	0/1	0/0	myocyte enhancer factor 2C	5q14
OXTR	0/1	0/0	oxytocin receptor	3p25
PCDH9	0/2	0/0	protocadherin 9	13q14.3-q21.1
RAI1	0/1	0/0	retinoic acid induced 1	17p11.2
RB1CC1	0/1	0/0	RB1-inducible coiled-coil 1	8q11
RIMS3	0/2	0/0	regulating synaptic membrane exocytosis 3	1pter-p22.2
SH3KBP1	0/1	0/0	SH3-domain kinase binding protein 1	Xp22.1-p21.3
SLC9A9	0/1	0/0	solute carrier family 9 (sodium/hydrogen exchanger), member 9	3q24
ST7	0/2	0/0	suppression of tumorigenicity 7	7q31.1-q31.3
UBE3A	1/0	0/0	ubiquitin protein ligase E3A (human papilloma virus E6-associated protein, Angel man syndrome)	15q11-q13

[a] Autism candidate gene list is downloaded from SFARI gene database: https://gene.sfari.org/.
[b] This column reports the number of probands and siblings who have the gene to be dysregulated (3 SD).

Table S4. Individuals with Outlier Genes (3 SD)

Probe_Id	Symbol	IDs with this Gene 3 SD Downregulated	IDs with this Gene 3 SD Upregulated
ILMN_1806408	ACADVL	11334.p1,11177.p1	0
ILMN_2038777	ACTB	11809.p1,12014.p1	0
ILMN_2053178	ACTG1	11809.p1,11267.p1	11382.s1
ILMN_1803686	ADA	11689.p1,11450.p1	0
ILMN_1814526	ADD3	11334.p1,12603.p1	11192.p1
ILMN_2342841	AFTIPHILIN	11348.p1,11337.p1	0
ILMN_1740752	AGPAT6	11207.p1,11523.p1	0
ILMN_1741148	ALDOA	11090.p1,11433.p1	0
ILMN_1712298	ANKRD46	11135.p1,11947.p1	11333.s1,11329.p1
ILMN_2402798	AP2M1	11475.p1,11192.p1	0
ILMN_1722491	APRT	11857.p1,11192.p1	11390.p1
ILMN_1726410	APRT	11857.p1,11192.p1	0
ILMN_1718610	ARHGAP17	11177.p1,11523.p1	0
ILMN_1811592	ARHGAP21	12512.p1,11610.p1,11406.p1	0
ILMN_1787879	ARL2	11625.p1,11519.p1	0
ILMN_1669113	ATF5	11041.p1,11540.p1	0
ILMN_2415189	ATP1A1	11177.p1,11482.p1	0
ILMN_1766185	AXIN1	11435.p1,11073.p1	0
ILMN_2342271	BCL11A	11041.p1,11879.p1	11519.s1,11625.p1,11519.p1,11480.s1,11177.s1,11947.p1
ILMN_1773780	C16ORF24	11435.p1,11475.p1	0
ILMN_2194828	C16ORF53	12451.p1,11540.p1	0
ILMN_1812441	C17ORF63	12032.p1,11114.p1	0
ILMN_1698233	C21ORF6	11046.p1,11461.p1	11276.p1,11146.s1
ILMN_1777318	C9ORF64	11180.p1,11348.p1	12014.p1,12523.p1
ILMN_1774196	C9ORF74	11219.p1,11425.p1,11192.p1	0
ILMN_1672807	CA5B	12014.p1,11415.p1	0
ILMN_1755504	CALCOCO2	11219.p1,11469.p1	0
ILMN_2388155	CASP3	11041.p1,11424.p1	0
ILMN_1715569	CCDC53	11532.p1,11007.p1,11046.p1	11947.s1
ILMN_1736567	CD74	12014.p1,12523.p1	0
ILMN_2379644	CD74	11274.p1,11424.p1	11276.s1
ILMN_2230683	CDCA7L	11041.p1,11083.p1	0
ILMN_1741459	CDK10	11333.p1,12241.p1	12299.s1
ILMN_1802615	CDK6	12512.p1,11337.p1	11177.p1
ILMN_1779401	CHP	11475.p1,11537.p1	0
ILMN_1705442	CMTM3	11274.p1,11587.p1	0
ILMN_1742432	COBRA1	12339.p1,11149.p1	0
ILMN_1730084	COMT	12239.p1,11879.p1	11947.s1,11177.p1
ILMN_1656920	CRIP1	11435.p1,11007.p1	11587.p1
ILMN_1690122	CRKL	12239.p1,11177.p1	11029.s1
ILMN_1812353	CSPP1	11461.p1,11490.p1	11537.s1
ILMN_1739576	CYB5R2	11220.p1,11537.p1	0
ILMN_1661599	DDIT4	11135.p1,11177.p1	0
ILMN_2145423	DET1	12523.p1,11475.p1	11495.p1
ILMN_1738124	DKFZP686I1569	11511.p1,11425.p1,11947.p1	0
ILMN_2380967	DNASE1L1	11868.p1,11879.p1	11178.s1
ILMN_1726990	DOM3Z	12327.p1,11461.p1	11482.s1
ILMN_2304624	EIF4H	11177.p1,11275.p1	11129.p1
ILMN_1710756	ENO1	11135.p1,11219.p1	0
ILMN_1663379	FBXL15	12327.p1,11410.p1,11461.p1	0
ILMN_1754489	FBXL20	12235.p1,11177.p1	11482.s1,11029.s1
ILMN_1737005	FLJ12886	11177.p1,11466.p1	0
ILMN_1708900	FLJ20422	11083.p1,11168.p1	0
ILMN_1730631	FLJ21945	11625.p1,11519.p1,11947.p1	0
ILMN_2344455	G3BP1	12297.p1,11348.p1	0
ILMN_1806754	GLDC	11041.p1,11857.p1	0

ILMN_1702177	GLO1	12523.p1,11192.p1	0
ILMN_1656145	GOT1	12297.p1,11475.p1	0
ILMN_1662846	GPR160	11041.p1,11879.p1	0
ILMN_1711289	GYS1	12219.p1,11495.p1	12044.s1,12224.p1
ILMN_1678037	HIRIP3	12451.p1,11540.p1	11177.p1
ILMN_2157441	HLA-DRA	11532.p1,11495.p1	0
ILMN_1678290	HMG20A	12032.p1,11466.p1	0
ILMN_2409220	HMMR	12297.p1,11274.p1	0
ILMN_1697342	HOXC11	11572.p1,11208.p1	11610.p1,11461.p1
ILMN_1799387	INOC1	11501.p1,11572.p1	0
ILMN_2392080	IQWD1	11348.p1,11337.p1	12096.p1
ILMN_1745826	KATNAL2	11457.p1,11442.p1	0
ILMN_2153280	KIAA0090	11041.p1,12261.p1	11177.p1
ILMN_1668469	KIAA0922	11177.p1,11073.p1	0
ILMN_1679232	KIDINS220	12048.p1,11412.p1	11029.s1
ILMN_1741204	KLHDC2	12399.p1,11301.p1,11285.p1	0
ILMN_1804451	LEO1	12523.p1,11469.p1	0
ILMN_1805796	LOC114984	11180.p1,11461.p1	11129.p1,12014.p1,11300.s1
ILMN_1727553	LOC63920	11625.p1,11519.p1,11947.p1	0
ILMN_2070355	LOC644096	12297.p1,11475.p1	0
ILMN_1679685	LOC650040	11102.p1,11335.p1	0
ILMN_2053546	LOC653314	11333.p1,11075.p1	0
ILMN_2131756	LRRC40	11459.p1,11348.p1	0
ILMN_1807825	LY86	11219.p1,11469.p1	0
ILMN_1775522	MAGED1	11511.p1,11425.p1	0
ILMN_2205032	MAGEE1	11041.p1,11303.p1	0
ILMN_1709114	MAP3K7IP1	12235.p1,11177.p1	11537.p1
ILMN_1723625	MAP4K2	11276.p1,11442.p1	11824.s1
ILMN_1753639	MTAP	11041.p1,11276.p1	11063.s1
ILMN_1714438	MUTYH	11540.p1,11178.p1	0
ILMN_2087702	MYH9	11469.p1,11046.p1	11059.p1
ILMN_1777528	NCBP1	11466.p1,11178.p1	0
ILMN_2370091	NGFRAP1	11177.p1,11442.p1	0
ILMN_1724194	NPEPL1	11267.p1,11177.p1	12299.s1
ILMN_2336982	NPTN	11114.p1,11233.p1	12297.p1,11154.p1,11443.s1
ILMN_1728224	OGFR	12096.p1,11267.p1	0
ILMN_1732024	OR2A5	12224.p1,11461.p1	0
ILMN_1810100	PBX3	11475.p1,11007.p1	0
ILMN_1728684	PELP1	11433.p1,11387.p1	0
ILMN_2075051	PGS1	11177.p1,11014.p1	0
ILMN_1653220	PITPNM1	11177.p1,11379.p1	0
ILMN_2093343	PLAC8	12603.p1,11442.p1	0
ILMN_2361427	PMS2L3	11041.p1,11177.p1	12420.p1
ILMN_1659058	PPP1R10	11625.p1,11947.p1	0
ILMN_1739622	PPP1R12A	11059.p1,11348.p1	0
ILMN_1784822	PPP1R3F	12014.p1,11427.p1,11947.p1	11634.p1
ILMN_1728305	PUM2	11080.p1,11625.p1	0
ILMN_1664030	RAB1B	11046.p1,11180.p1	0
ILMN_1741957	RABEPK	11696.p1,11736.p1	11177.p1
ILMN_2221006	RAD21	12279.p1,11625.p1	0
ILMN_1755023	RAD50	12984.p1,13128.p1	11410.s1
ILMN_1801262	RAD51L1	12399.p1,11329.p1	11041.p1
ILMN_1662198	RANGAP1	12297.p1,11519.p1	0
ILMN_1751886	REC8L1	11333.p1,11424.p1,11458.p1	0
ILMN_1732336	RFC2	11178.p1,11461.p1	11537.s1
ILMN_1784584	RINT-1	11177.p1,11338.p1	0
ILMN_2339748	RNF13	12032.p1,11007.p1	11519.s1,11625.p1,11519.p1
ILMN_1655165	RNF138	11868.p1,11073.p1,11348.p1	0
ILMN_2160388	RPL24	12014.p1,12523.p1	0

ILMN_1707810	RPS5	12014.p1,12523.p1	0
ILMN_1806294	RPS6KA3	12014.p1,11192.p1	0
ILMN_1720889	SC4MOL	11333.p1,12184.p1	0
ILMN_2312498	SEMG1	11461.p1,11275.p1	0
ILMN_1788778	11-Sep	12297.p1,11337.p1	0
ILMN_1682404	SETMAR	13128.p1,11046.p1	11177.p1
ILMN_2059452	SLC12A2	11348.p1,11443.p1	11839.p1
ILMN_1749521	SLC35E3	11723.p1,11947.p1	0
ILMN_2124471	SLC36A1	11178.p1,11271.p1	0
ILMN_1791702	SMARCA2	11285.p1,11443.p1	11333.s1
ILMN_1732053	SNRP70	11466.p1,11178.p1	11197.s1,11265.s1
ILMN_1709772	SNX5	12523.p1,11519.p1	0
ILMN_1690920	SP100	11178.p1,11329.p1	11947.s1,11177.p1
ILMN_1756501	ST6GAL1	11387.p1,11879.p1	11519.s1,11519.p1
ILMN_1651692	STK10	11090.p1,11379.p1	0
ILMN_1655163	STK24	11301.p1,11219.p1	12343.p1,11254.s1,11466.s1
ILMN_1693726	TBC1D10A	11083.p1,11180.p1	0
ILMN_1743352	TBCC	12032.p1,11947.p1	0
ILMN_1656798	TIMM17A	11335.p1,11348.p1	0
ILMN_1711566	TIMP1	11387.p1,11177.p1,11442.p1	11443.s1
ILMN_2192316	TOP1	12297.p1,11947.p1	0
ILMN_1796063	TRIM44	12603.p1,11301.p1	0
ILMN_1806778	UBE2E1	11219.p1,11348.p1	11947.s1
ILMN_2395932	UNC45A	12512.p1,11177.p1	11466.s1
ILMN_1697906	WBP4	11625.p1,11947.p1	11333.s1
ILMN_2104106	XPR1	11519.p1,11947.p1	0
ILMN_1665205	ZFP260	11625.p1,11947.p1	11857.p1
ILMN_2358382	ZFYVE1	11502.p1,11406.p1,11180.p1	0
ILMN_1719202	ZNF174	11071.p1,11466.p1	0
ILMN_2117904	ZNF22	11276.p1,11301.p1	11461.s1
ILMN_1701875	ZYX	11610.p1,11177.p1	11578.s1

Table S5. CNVs with Expression Dysregulation

Submitted as a separate Excel file.

Table S6. Primer Information

Primers for Validating Expression Change

Gene	Sequence	Note
GAPDH_L	TCATCAGCAATGCCTCCTGCAC	Forward
GAPDH_R	GGTGGCAGTGATGGCATGGAC	Reverse
ITPR1_L	AGCTCCCGAGTGTCCTAAAG	Forward
ITPR1_R	GCAGCAGCATCATTTGAAAG	Reverse
SUMF1_L	AGGAACGAGGACCTTGAATG	Forward
SUMF1_R	CATGATTCAAAGCATCGGATA	Reverse
AXIN1_L	CACGTGTGCTGGGATCTACT	Forward
AXIN1_R	CAAGCTGTGTTGAAGGCACT	Reverse
RAB40C_L	GACTTTGAGGACCTGGATGG	Forward
RAB40C_R	AAAGAACGGTTCGGAGAGAA	Reverse
MRPS35_L	TCATCAGAAAGAAATATCCTGGAA	Forward
MRPS35_R	GGATTCTTTGTACTGAGAAATGGA	Reverse
SFRS10_L	AAACCGGGTGCTTCAAAGT	Forward
SFRS10_R	TGGTAAGCAAAGGACCTGAA	Reverse
GOT1_L	CTGAAGGAGCCAAAGTGTGA	Forward
GOT1_R	GGACACAACCATGCAGAAAG	Reverse
CHD1L_L	GCAAGATTTGTTGGCCTTG	Forward
CHD1L_R	CTCTTCTAGGGCTGCCATCT	Reverse
PRKAB2_L	ATTTCTTGTGACCCAGCCTT	Forward
PRKAB2_R	CGCTAAGGACCATCACACTG	Reverse
PEF1_L	GATGCCAGTGGTGAGTGTTC	Forward
PEF1_R	AAGCCACTGGTCCCATAGAC	Reverse
ELF1_L	TGCAGAGAAATAAGTGACCCA	Forward
ELF1_R	CTTAGCAACACAAGTTTACTAATGGA	Reverse
CSTF2T_L	CTGGCTTTCTTATACAGATGGTGT	Forward
CSTF2T_R	CTGGGCCTTGATTATTCCTG	Reverse
CTNND1_L	TTTGGACGTGACCAGGATAA	Forward
CTNND1_R	CCACAGGGTTCCGGTAATAA	Reverse
ADARB1_L	CGAGTACCAAGCCACAAGAA	Forward
ADARB1_R	CTGACTCCATTAGCGTTCCA	Reverse
ALODA_L	TATGTGACCGAGAAGGTGCT	Forward
ALODA_R	GCCTTCCAGGTAGATGTGGT	Reverse
MAPK3_L	CAGTTCTGGAATGGAAGGGT	Forward

MAPK3_R	TTCCTTCAGGGAAACTAGGG	Reverse
CORO1A_L	CCATGTTCAGTTCCAAGGAG	Forward
CORO1A_R	AGCTTGTAGAACCTGGCGAT	Reverse
TMLHE_L	GCTCAGCATCGTGCTACAAC	Forward
TMLHE_R	ACCATCTGGCCAAGTGAAA	Reverse
COX6A1_L	CTCGCATGTGGAAGACTCTC	Forward
COX6A1_R	AACGGCTTGGTCCTGATG	Reverse
TRIAP1_L	TTGCAGTGAACACCATTTCA	Forward
TRIAP1_R	GTTGAGAGCTGGCAATAGCA	Reverse
SEC23B_L1	CAGTCAGGCTCGATTCCTTT	Forward
SEC23B_R1	GTTAGGATGGGTGCTCCAGT	Reverse
TIMP1_L1	TACTTCCACAGGTCCCACAA	Forward
TIMP1_R1	GGAAACACTGTGCATTCCTC	Reverse

Primers for Validating Copy-Number Change

Gene	Sequence	Note
12q24.31_F	GTGCCTTAGTGCAAGTTCTTCAT	Forward
12q24.31_R	GAATTGAGACGTAATCCCAAGTG	Reverse
20p11.23_F	TTTAGGTTTGATGTGTGTGCATC	Forward
20p11.23_R	TGAAGAAGGCTACAGAGAACAGG	Reverse
Xp11.23_F	GACAGCAATGAAATGCAGGTAG	Forward
Xp11.23_R	TTCATAAAGGTGAGGGTCGAGT	Reverse
Xq28_F	TGTTGGAGGTGTTGGAAATAATC	Forward
Xq28_R	AACCTCATCAACAGTTTCCTTGA	Reverse

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