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Supplementary webappendix

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WebAppendix

A genome-wide analysis of rare chromosomal deletions and duplications in attention-deficit hyperactivity disorder

Nigel M. Williams^{1,2*}, Irina Zaharieva^{1,2}, Andrew Martin^{1,2}, Kate Langley^{1,2}, Kiran Mantripragada^{1,2}, Ragnheidur Fossdal³, Hreinn Stefansson³, Kari Stefansson³, Pall Magnusson⁴, Olafur O. Gudmundsson⁴, Omar Gustafsson^{3,5}, Peter Holmans^{1,2}, Michael J. Owen^{1,2}, Michael O'Donovan^{1,2}, Anita Thapar^{1,2*}

1 MRC Centre in Neuropsychiatric Genetics and Genomics, Cardiff University School of Medicine

2 Department of Psychological Medicine and Neurology, Cardiff University School of Medicine

3 deCODE genetics, Sturlugata 8, 101 Reykjavik ,Iceland

4 Child and Adolescent Psychiatry, Landspítali University Hospital, Dalbraut 12 , IS-105, Iceland

5 Department of Psychiatry, Oslo University Hospital, Kirkevejen 166, 0407 Oslo, Norway.

Supplementary Table 2

Chromosome and start and end point of all 118 CNVs >500kb spanning or intersecting a known gene in HG18, together with the ID of genes disrupted. The 19 CNVs that do not span a gene are not presented

CNV coordinates	Genes
chr1:742429-1303878	SAMD11 NOC2L KLHL17 PLEKHN1 HES4 ISG15 AGRN C1orf159 TTLL10 TNFRSF18 TNFRSF4 SDF4 B3GALT6 FAM132A UBE2J2 SCNN1D CENTB5 PUSL1 CPSF3L GLTPD1 TAS1R3 DVL1 MXRA8 AURKAIP1 CCNL2
chr1:144943150-146293282	PRKAB2 FMO5 CHD1L BCL9 ACP6 GJA5 GJA8 GPR89B GPR89C NBPF11 LOC728912
chr1:159732044-160374743	FCGR2A HSPA6 FCGR3A FCGR2C FCGR3B FCGR2B FCRLA FCRLB

DUSP12
ATF6
OLFML2B
NOS1AP

chr1:235967993-237654135 RYR2
ZP4

chr1:245714416-246522135 OR2W5
OR2C3
C1orf150
OR2G2
OR2G3
OR13G1
OR6F1
OR1C1
OR14A16
OR11L1
TRIM58
OR2W3
OR2T8
OR2L13
OR2L8
OR2AK2
OR2L2
OR2L3
OR2M5
OR2M2
OR2M3
OR2M4
OR2T33
OR2T12

chr2:32487194-33232767 BIRC6
TTC27
LTBP1

chr2:32497032-33142223 BIRC6
TTC27
LTBP1

chr2:32497032-33181898 BIRC6
TTC27
LTBP1

chr2:45264692-45857588 SRBD1
PRKCE

chr2:100194339-100878975	LONRF2 CHST10 NMS LOC643896 PDCL3 NPAS2
chr2:117555176-118507870	DDX18 CCDC93
chr2:153846483-154565034	RPRM GALNT13
chr2:158090314-158967549	ACVR1C ACVR1 UPP2 CCDC148
chr2:205197898-205833558	PARD3B
chr3:209726-1685731	CHL1 CNTN6
chr3:840779-1402079	CNTN6
chr3:849785-1352410	CNTN6
chr3:1166638-2183832	CNTN6 CNTN4
chr3:2969491-4557680	CNTN4 IL5RA TRNT1 CRBN LRRN1 SETMAR SUMF1 ITPR1
chr3:3423534-4063576	LRRN1
chr3:3999657-4922174	SETMAR SUMF1 ITPR1

chr3:58472819-60222161	KCTD6 ACOX2 FAM107A FAM3D C3orf67 FHIT
chr3:75816778-77177489	ZNF717 ROBO2
chr3:96242194-98526743	EPHA6
chr3:155817656-156432544	MME
chr3:162419058-163993833	NMD3 C3orf57 OTOL1
chr3:174547344-176016137	NLGN1
chr3:194235339-194817685	HRASLS ATP13A5 ATP13A4 OPA1
chr3:194759366-195501036	ATP13A4 OPA1 HES1
chr4:23596065-26908642	DHX15 SOD3 CCDC149 LGI2 SEPSECS PI4K2B ZCCHC4 ANAPC4 SLC34A2 KIAA0746 RBPJ CCKAR TBC1D19 STIM2

chr4:26350218-27533657	TBC1D19 STIM2
chr4:64314017-65184017	SRD5A2L2
chr4:65791505-66506505	EPHA5
chr4:65846170-66521854	EPHA5
chr4:70889530-72068561	STATH HTN3 HTN1 C4orf40 ODAM C4orf7 CSN3 C4orf35 SMR3A SMR3B PROL1 MUC7 AMTN AMBN ENAM IGJ UTP3 RUFY3 GRSF1 MOBKL1A DCK
chr4:75931128-76957385	BTC DKFZP564O0823 RCHY1 THAP6 C4orf26 CDKL2 G3BP2 USO1
chr4:89231344-90428182	ABCG2 PPM1K HERC6 HERC5 PIGY HERC3 NAP1L5 FAM13A1 TIGD2

	GPRIN3
chr4:92505628-95897466	GRID2 ATOH1 SMARCAD1 PGDS PDLIM5 BMPR1B
chr4:92969417-93647358	GRID2
chr4:152041535-153449318	LRBA RPS3A SH3D19 ESSPL FAM160A1 PET112L
chr4:188474188-191164126	ZFP42 TRIML2 TRIML1 FRG1 TUBB4Q
chr5:9235717-9946560	SEMA5A TAS2R1
chr5:10912186-11505944	CTNND2
chr5:14525040-15234322	TRIO FAM105A FAM105B ANKH
chr5:45573056-46384240	HCN1
chr5:75209785-76193792	SV2C IQGAP2 F2RL2 F2R F2RL1 S100Z
chr5:141874445-142427181	FGF1 ARHGAP26

chr6:71032233-71665770	COL9A1 FAM135A C6orf57 SMAP1 B3GAT2
chr6:77644608-78365965	HTR1B
chr6:86816704-87733426	HTR1E
chr6:91445589-94638804	EPHA7
chr6:118705946-119293670	SLC35F1 C6orf204 PLN ASF1A MCM9
chr6:128863230-130509862	PTPRK LAMA2 ARHGAP18 C6orf191 L3MBTL3 SAMD3
chr6:168985464-169590571	THBS2 WDR27
chr7:14326377-15604412	DGKB TMEM195
chr7:62215205-63929057	ZNF680 ZNF107 ZNF138
chr7:69777660-71680977	AUTS2 WBSCR17 CALN1
chr7:105235446-105857403	ATXN7L1 FLJ23834 SYPL1 NAMPT

chr7:122788319-123545119	IQUB NDUFA5 ASB15 LMOD2 WASL HYAL4 SPAM1
chr8:12977952-14075129	DLC1 SGCZ
chr8:14879967-16226257	SGCZ TUSC3 MSR1
chr8:43177542-43910848	HGSNAT A26A1
chr8:85438801-88322287	RALYL LRRCC1 E2F5 C8orf59 CA13 CA13 CA3 CA2 REXO1L1 PSKH2 ATP6V0D2 SLC7A13 WWP1 FAM82B CPNE3 CNGB3 CNBD1
chr8:89227035-90315131	MMP16
chr8:107876892-108508538	ANGPT1
chr8:125965894-126563499	ZNF572 SQLE KIAA0196 NSMCE2 TRIB1

chr8:131230753-132355063	DDEF1 ADCY8
chr9:5647977-6819942	KIAA1432 ERMP1 MLANA KIAA2026 RANBP6 IL33 TPD52L3 UHRF2 GLDC JMJD2C
chr9:86604848-87529107	NTRK2 AGTPBP1
chr10:92079458-92641459	HTR7 RPP30
chr10:132024456-134872862	TCERG1L PPP2R2D BNIP3 JAKMIP3 DPYSL4 STK32C LRRC27 PWWP2B C10orf91 INPP5A NKX6-2 C10orf92 C10orf93 GPR123 KNDC1
chr11:5582423-7391991	TRIM6 TRIM6-TRIM34 TRIM34 TRIM5 TRIM22 OR56B1 OR52N4 OR52N5 OR52N1 OR52N2 OR52E6 OR52E8 OR52E4

OR56A3
OR52L1
OR56A4
OR56A1
OR56B4
OR52B2
OR52W1
C11orf42
FAM160A2
CNGA4
CCKBR
PRKCDBP
SMPD1
APBB1
HPX
TRIM3
ARFIP2
FXC1
C11orf47
DNHD1
KIAA0409
ILK
TAF10
TPP1
DCHS1
MRPL17
OR2AG2
OR2AG1
OR6A2
OR10A5
OR10A2
OR10A4
OR2D2
OR2D3
ZNF215
ZNF214
NLRP14
RBMXL2
SYT9

chr11:49964834-50669978 OR4C12

chr11:67841909-68957312 LRP5
SAPS3
GAL
MTL5
CPT1A
MRPL21
IGHMBP2
MRGPRD
MRGPRF
TPCN2
MYEOV

chr11:133553353-134382876 NCAPD3
VPS26B
THYN1
ACAD8
GLB1L3
GLB1L2
B3GAT1

chr12:7888157-10044293 SLC2A14
SLC2A3
FOXJ2
C3AR1
NECAP1
CLEC4A
ZNF705A
FAM90A1
CLEC6A
CLEC4D
CLEC4E
AICDA
MFAP5
FAM80B
A2ML1
PHC1
M6PR
KLRG1
A2ML1
PZP
KLRB1
CLEC2D
CLECL1
CD69
KLRF1
CLEC2B
CLEC12A
CLEC1B

chr12:33415349-34496692 SYT10
ALG10

chr12:33415349-34711193 SYT10
ALG10

chr12:42798121-43455177 TMEM117
NEL2

chr12:84576944-85337498 RASSF9
NTS

MGAT4C

chr13:64959554-66299023 PCDH9

chr14:104032680-106358708 C14orf180
TMEM179
INF2
C14orf151
ADSSL1
SIVA1
AKT1
KIAA0284
PLD4
AHNAK2
C14orf79
CDCA4
GPR132
JAG2
NUDT14
BRF1
BTBD6
PACS2
MTA1
CRIP2
CRIP1
C14orf80
TMEM121

chr15:21016722-26002360 MKRN3
MAGEL2
NDN
C15orf2
SNRPN
SNURF
UBE3A
ATP10A
GABRB3
GABRA5
GABRG3
OCA2

chr15:27000239-28103304 APBA2
KIAA0574
NDNL2
TJP1

chr15:27013177-28153539 APBA2
KIAA0574
NDNL2
TJP1

chr15:29734334-30300525	OTUD7A CHRNA7
chr15:29734334-30302218	OTUD7A CHRNA7
chr15:29734334-30400921	OTUD7A CHRNA7
chr15:29807358-30319628	CHRNA7
chr15:30688712-32505886	ARHGAP11A SCG5 GREM1 FMN1 RYR3 AVEN CHRM5 C15orf24 PGBD4 C15orf29 TMEM85 SLC12A6 NOLA3 C15orf55 LPCAT4 GOLGA8A
chr15:30713368-32658588	ARHGAP11A SCG5 GREM1 FMN1 RYR3 AVEN CHRM5 C15orf24 PGBD4 C15orf29 TMEM85 SLC12A6 NOLA3 C15orf55 LPCAT4 GOLGA8A GOLGA8B
chr15:55108011-55905365	TCF12

CGNL1
Gcom1
GRINL1A

chr16:15032942-16143932 PDXDC1
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RRN3
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KIAA0430
NDE1
MYH11
C16orf63
ABCC1
ABCC6

chr16:15156431-18072544 MPV17L
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KIAA0430
NDE1
MYH11
C16orf63
ABCC1
ABCC6
NOMO3
LOC339047
XYLT1

chr16:15387380-16133472 MPV17L
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MYH11
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ABCC1
ABCC6

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KIAA0430
NDE1
MYH11

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ABCC6
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LOC339047
XYLT1

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C16orf63
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ABCC6

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POLR3E
CDR2
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chr16:29342178-31155425
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SULT1A4
SULT1A3
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MAZ
PRRT2
C16orf53
MVP
CDIPT
SEZ6L2
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KCTD13
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FAM57B
ALDOA
PPP4C
TBX6
YPEL3

GDPD3
MAPK3
CORO1A
BOLA2B
BOLA2
GIYD1
GIYD2
SULT1A4
SULT1A3
CD2BP2
TBC1D10B
MYLPF
ZNF553
ZNF771
XTP3TPA
SEPHS2
ITGAL
ZNF768
ZNF747
ZNF764
ZNF688
ZNF785
ZNF689
PRR14
FBRS
SRCAP
PHKG2
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FBXL19
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SETD1A
HSD3B7
STX1B
STX4
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ZNF646
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VKORC1
BCKDK
MYST1
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FUS
PYCARD
TRIM72
PYDC1

chr16:29554843-30271572

SPN
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C16orf54

MAZ
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C16orf53
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CDIPT
SEZ6L2
ASPHD1
KCTD13
TMEM219
TAOK2
HIRIP3
CCDC95
DOC2A
FLJ25404
FAM57B
ALDOA
PPP4C
TBX6
YPEL3
GDPD3
MAPK3
CORO1A
BOLA2B
BOLA2
GIYD1
GIYD2
SULT1A4
SULT1A3
CD2BP2
TBC1D10B

chr16:29563594-30367556

SPN
QPRT
C16orf54
MAZ
PRRT2
C16orf53
MVP
CDIPT
SEZ6L2
ASPHD1
KCTD13
TMEM219
TAOK2
HIRIP3
CCDC95
DOC2A
FLJ25404
FAM57B
ALDOA
PPP4C
TBX6
YPEL3
GDPD3

MAPK3
CORO1A
BOLA2B
BOLA2
GIYD1
GIYD2
SULT1A4
SULT1A3
CD2BP2
TBC1D10B
MYLPF
ZNF553
ZNF771
XTP3TPA
SEPHS2

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CDH13

chr17:31964475-33182480 MYO19
PIGW
GGNBP2
MGC4172
MRM1
LHX1
AATF
ACACA
C17orf78
TADA2L
DUSP14
AP1GBP1
DDX52
HNF1B

chr17:31979521-33205550 GGNBP2
MGC4172
MRM1
LHX1
AATF
ACACA
C17orf78
TADA2L
DUSP14
AP1GBP1
DDX52
HNF1B

chr18:23102007-24621659 CDH2

chr19:7054542-7871339 INSR

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C19orf45
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XAB2
PCP2
STXBP2
RETN
C19orf59
TRAPPC5
FCER2
CLEC4G
CD209
CLEC4M
EVI5L
LRRC8E
MAP2K7

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FLJ21438
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CYP4F22
CYP4F8
CYP4F3
CYP4F12
OR10H2
OR10H3
OR10H5
OR10H1
CYP4F2
CYP4F11
OR10H4
TPM4
RAB8A

chr19:21575835-22346694
ZNF100
ZNF43
ZNF208
ZNF257
ZNF676
LOC342994

chr20:3036010-3839896
UBOX5
FASTKD5
ProSAPiP1
C20orf116
ITPA
SLC4A11
C20orf194
ATRN

	GFRA4 ADAM33 SIGLEC1 HSPA12B C20orf27 SPEF1 CENPB CDC25B C20orf29 VISA PANK2
chr20:12795614-14137779	SPTLC3 C20orf82 TASP1 ESF1 C20orf7 SEL1L2 MACROD2
chr20:14574845-15159056	MACROD2
chr21:31669392-32494130	TIAM1 SOD1 SFRS15 HUNK
chr21:39655437-40231693	WRB LCA5L SH3BGR B3GALT5 IGSF5 PCP4
chr22:15634399-16265697	XKR3 GAB4 IL17RA CECR6 CECR5 CECR1
chr22:17257787-18686993	DGCR6 PRODH DGCR2 DGCR14 TSSK2 GSC2 SLC25A1 CLTCL1

HIRA
MRPL40
C22orf39
UFD1L
CDC45L
CLDN5
GP1BB
TBX1
GNB1L
C22orf29
TXNRD2
COMT
ARVCF
C22orf25
DGCR8
HTF9C
RANBP1
ZDHHC8
RTN4R
DGCR6L

chr22:22020325-23326630
IGLL1
LOC51233
RGL4
ZNF70
VPREB3
C22orf15
CHCHD10
MMP11
SMARCB1
DERL3
SLC2A11
MIF
GSTT2B
GSTT2
DDTL
DDTL
GSTT2
GSTT1
CABIN1
SUSD2
GGT5
SPECC1L
ADORA2A
UPB1
C22orf13
SNRPD3
GGT1
C22orf36

chr22:44041512-44638874
UPK3A
FAM118A
SMC1B

RIBC2
FBLN1
ATXN10

Supplementary Table 3

Global burden of rare CNVs larger than 200kb. RATE; the average number of CNVs per person. CNV(n); the number of CNVs observed. P values are empirical and one-sided.* IQ data were unavailable for 14 ADHD cases

		Burden of all CNVs				Burden of Deletions Only				Burden of Duplications Only			
		ADHD	Controls	Case/Control ratio	P	ADHD	Controls	Case/Control ratio	P	ADHD	Controls	Case/Control ratio	P
ADHD/Controls (n=366/1047) Size > 200kb	CNV(n)	131	387			37	145			94	242		
	RATE	0.36	0.37	0.97	0.62	0.10	0.14	0.73	0.94	0.26	0.23	1.11	0.23
	AVGKB	710.8	456.3		0.00011	630.7	393.6		0.0053	770.5	499.7		0.0049
ADHD with IQ≥70/Controls (n=319/1047) Size > 200kb	CNV(n)	108	387			27	145			81	242		
	RATE	0.34	0.37	0.92	0.78	0.08	0.14	0.61	0.99	0.25	0.23	1.10	0.27
	AVGKB	698.8	456.3		0.00011	603.7	393.6		0.018	747.8	499.7		0.00110
ADHD with IQ<70/Controls (n=33/1047) Size > 200kb	N	18	387			7	145			11	242		
	RATE	0.55	0.37	1.48	0.10	0.21	0.14	1.53	0.20	0.33	0.23	1.44	0.18
	AVGKB	750.3	456.3		0.0378	669.1	393.6		0.10	856.7	499.7		0.04