Table S1. Articles meeting inclusion criteria for all disorders.

Disease Category	Study	Cases (N)	Controls (N)
ID			
	Bachmann-Gagescu et al. (2010)[1]	23,084[a]	7,700
	Cooper et al. (2011)[2]	15,767	8,329
	Girirajan et al. (2010)[3]	11,873	8,540
		9,254[b]	6,299[b]
	Hannes et al. (2009)[4]	1,027	2,014
	Kaminsky et al. (2011)[5]	15,749	10,118
	Mefford et al. (2008)[6]	5,218	2,063
	Mefford et al. (2009)[7]	1,010	2,493
	Rosenfeld et al. (2010)[8]	9,773	2,493
	Sharp et al. (2008)[9]	1,040	960
ASD			
	Glessner et al. (2009)[10]	859	1,409
		1,336[b]	1,110[b]
	Kumar et al. (2008)[11]	180	372
		532[c]	465[c]
	Marshall et al. (2008)[12]	427	500
			1,152[b]
	Moreno-De-Luca et al. (2010)[13]	15,749	4,519
		1,182[d]	47,929[b]
	Pinto et al. (2010)[14]	996	1,287
			3,677[b]
	Sanders et al. (2011)[15]	1,124[e]	872[e]
	Sato et al. (2012)[16]	1,614	15,122
	Weiss et al. (2008)[17]	512	434
		1,441	2,814
		299	18,834
Schizophrenia			
	Glessner et al. (2010)[18]	977	2,000
		758[b]	1,485[b]
	Grozeva et al. (2012)[19,f]	>5,000	>37,000
	Ingason et al. (2011a)[20]	7,582	41,370
	Ingason et al. (2011b)[21]	4,345	35,079
	International Schizophrenia Consortium (2008)[22]	3,391	3,181
	Kirov et al. (2009)[23]	471	2,792
	Lee et al. (2012)[24]	5,325	9,279
	Levinson et al. (2011)[25]	3,945	3,611
	McCarthy et al. (2009)[26]	1,906	3,971
		2,645[b]	2,420[b]
	Moreno-De-Luca et al. (2010)[13]	6,340	47,929
	Mulle et al. (2010)[27]	7,545	39,748
	Rujescu et al. (2009)[28]	2,977	33,746

	Stefansson et al. (2008)[29]	1,433 3,285[b]	33,250 7,951[b]
	Vacic et al. (2011)[30]	8,290	7,431
	Zhao et al. (2012)[31]	2,058	3,275
Epilepsy			
	de Kovel et al. (2010)[32]	1,234	3,022
	Dibbens et al. (2009)[33]	539	3,777
	Heinzen et al. (2010)[34]	3,812	1,299
	Helbig et al. (2009)[35]	1,223	3,699
	Mefford et al. (2010)[36]	517	2,493

Note: Eight autism spectrum disorder (ASD), 9 intellectual disability (ID), 15 schizophrenia, and 5 epilepsy articles were eligible. The number of cases and controls are reported.

[a]Patients referred for genetic testing due to developmental delay

[b]Replicated cohort sample

[c]Follow-up cohort

[d]Replicated cohort, ASD/neurocognitive impairment cohort

[e]872 matched probands, 872 matched sibling; 4,457 total participants

[f]10,259 control subjects from UK Wellcome Trust Case Control Consortium as well as controls from other published studies and compared with previously published schizophrenia articles

1. Bachmann-Gagescu R, Mefford HC, Cowan C, et al. Recurrent 200-kb deletions of 16p11.2 that include the SH2B1 gene are associated with developmental delay and obesity. Genetics in medicine : official journal of the American College of Medical Genetics. 2010;12(10):641-647.
2. Cooper GM, Coe BP, Girirajan S, et al. A copy number variation morbidity map of developmental delay. Nature genetics. Sep 2011;43(9):838-846.
3. Girirajan S, Rosenfeld JA, Cooper GM, et al. A recurrent 16p12.1 microdeletion supports a two-hit model for severe developmental delay. Nature genetics. Mar 2010;42(3):203-209.
4. Hannes FD, Sharp AJ, Mefford HC, et al. Recurrent reciprocal deletions and duplications of 16p13.11: the deletion is a risk factor for MR/MCA while the duplication may be a rare benign variant. Journal of medical genetics. Apr 2009;46(4):223-232.
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6. Mefford HC, Sharp AJ, Baker C, et al. Recurrent rearrangements of chromosome 1q21.1 and variable pediatric phenotypes. The New England journal of medicine. Oct 16 2008;359(16):1685-1699.
7. Mefford HC, Cooper GM, Zerr T, et al. A method for rapid, targeted CNV genotyping identifies rare variants associated with neurocognitive disease. Genome Res. Sep 2009;19(9):1579-1585.
8. Rosenfeld JA, Coppinger J, Bejjani BA, et al. Speech delays and behavioral problems are the predominant features in individuals with developmental delays and 16p11.2 microdeletions and microduplications. J Neurodev Disord. Mar 2010;2(1):26-38.
9. Sharp AJ, Mefford HC, Li K, et al. A recurrent 15q13.3 microdeletion syndrome associated with mental retardation and seizures. Nature genetics. Mar 2008;40(3):322-328.
10. Glessner JT, Wang K, Cai G, et al. Autism genome-wide copy number variation reveals ubiquitin and neuronal genes. Nature. May 28 2009;459(7246):569-573.
11. Kumar RA, KaraMohamed S, Sudi J, et al. Recurrent 16p11.2 microdeletions in autism. Hum

Mol Genet. Feb 15 2008;17(4):628-638.
12. Marshall CR, Noor A, Vincent JB, et al. Structural variation of chromosomes in autism spectrum disorder. Am J Hum Genet. Feb 2008;82(2):477-488.
13. Moreno-De-Luca D, Mulle JG, Kaminsky EB, et al. Deletion 17q12 is a recurrent copy number variant that confers high risk of autism and schizophrenia. Am J Hum Genet. Nov 12 2010;87(5):618-630.
14. Pinto D, Pagnamenta AT, Klei L, et al. Functional impact of global rare copy number variation in autism spectrum disorders. Nature. Jul 15 2010;466(7304):368-372.
15. Sanders SJ, Hus V, Luo R, et al. Multiple recurrent de novo CNVs, including duplications of the 7q11. 23 Williams syndrome region, are strongly associated with autism. Neuron. 2011;70(5):863-885.
16. Sato D, Lionel AC, Leblond CS, et al. SHANK1 Deletions in Males with Autism Spectrum Disorder. Am J Hum Genet. May 4 2012;90(5):879-887.
17. Weiss LA, Shen Y, Korn JM, et al. Association between microdeletion and microduplication at 16p11.2 and autism. The New England journal of medicine. Feb 14 2008;358(7):667-675.
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26. McCarthy SE, Makarov V, Kirov G, et al. Microduplications of 16p11. 2 are associated with schizophrenia. Nature genetics. 2009;41(11):1223-1227.
27. Mulle JG, Dodd AF, McGrath JA, et al. Microdeletions of 3q29 confer high risk for schizophrenia. Am J Hum Genet. Aug 13 2010;87(2):229-236.
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spectrum of sporadic epilepsy syndromes. American Journal of Human Genetics. May 14 2010;86(5):707-718.
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36. Mefford HC, Muhle H, Ostertag P, et al. Genome-wide copy number variation in epilepsy: novel susceptibility loci in idiopathic generalized and focal epilepsies. PLoS Genetics. May 2010;6(5):e1000962.

Table S2. Distribution of copy number variants (CNVs) and genes among autism spectrum disorder (ASD), intellectual disability (ID), schizophrenia, and epilepsy.

CNV (strict)			n (%)
	ID (n=27)		
		ASD	6 (22.2)
		Schizophrenia	9 (33.0)
		Epilepsy	3 (11.0)
	ASD (n=13)		
		ID	6 (46.2)
		Schizophrenia	6 (46.2)
		Epilepsy	0 (0)
	Schizophrenia (n=21)		
		ID	9 (43.0)
		ASD	6 (28.6)
		Epilepsy	2 (9.5)
	Epilepsy (n=3)		
		ID	3 (100)
		ASD	0 (0)
		Schizophrenia	3 (100.0)
Genes (strict)			
	ID (n=1,139)		
		ASD	244 (21.4)
		Schizophrenia	386 (33.9)
		Epilepsy	111 (9.7)
	ASD (n=252)		
		ID	244 (96.8)
		Schizophrenia	212 (84.1)
		Epilepsy	0 (0)
	Schizophrenia (n=416)		
		ID	386 (92.8)
		ASD	212 (51.0)
		Epilepsy	111 (26.7)
	Epilepsy (n=111)		
		ID	111 (100)
		ASD	0 (0)
		Schizophrenia	111 (100)

Note: The number of CNVs/genes, and percentages for each disorder found within the other disorders are reported (i.e. the number of ASD CNVs/genes that are also found in ID CNVs/genes).

Table S3. List of significant *DSM* processes for each disorder (i.e. disorder-specific) from pathway analysis (i.e. significant processes from all genes identified from eligible autism spectrum disorder [ASD], intellectual disability [ID], schizophrenia, or epilepsy copy number variants [CNVs]).

ASD	**# Genes**	***p* value**
GO:0044456~synapse part	11	0.000
GO:0045202~synapse	12	0.000
GO:0030054~cell junction	14	0.000
GO:0001505~regulation of neurotransmitter levels	6	0.000
GO:0016079~synaptic vesicle exocytosis	4	0.000
GO:0007268~synaptic transmission	10	0.000
GO:0019226~transmission of nerve impulse	10	0.001
GO:0006887~exocytosis	6	0.002
GO:0048489~synaptic vesicle transport	4	0.002
GO:0007269~neurotransmitter secretion	4	0.002
GO:0005230~extracellular ligand-gated ion channel activity	5	0.002
GO:0045211~postsynaptic membrane	6	0.003
GO:0022604~regulation of cell morphogenesis	6	0.003
GO:0042802~identical protein binding	13	0.003
GO:0006836~neurotransmitter transport	5	0.004
GO:0003001~generation of a signal involved in cell-cell signaling	5	0.004
GO:0032940~secretion by cell	7	0.005
ID		
GO:0043120~tumor necrosis factor binding	5	0.000
GO:0030036~actin cytoskeleton organization	18	0.000
GO:0030029~actin filament-based process	18	0.000
GO:0042493~response to drug	16	0.001
GO:0019717~synaptosome	10	0.001
GO:0005031~tumor necrosis factor receptor activity	4	0.001
GO:0045211~postsynaptic membrane	12	0.003
GO:0030054~cell junction	28	0.006
GO:0007010~cytoskeleton organization	23	0.006
GO:0045202~synapse	21	0.007
GO:0044456~synapse part	16	0.010
GO:0007265~Ras protein signal transduction	9	0.010
GO:0008092~cytoskeletal protein binding	25	0.013
GO:0017076~purine nucleotide binding	73	0.013
GO:0007492~endoderm development	4	0.013
GO:0007015~actin filament organization	7	0.016
GO:0004890~GABA-A receptor activity	4	0.017
GO:0005230~extracellular ligand-gated ion channel activity	7	0.018
GO:0006650~glycerophospholipid metabolic process	9	0.018
GO:0002673~regulation of acute inflammatory response	4	0.020
GO:0032555~purine ribonucleotide binding	69	0.020

GO:0032553~ribonucleotide binding	69	0.020
GO:0005501~retinoid binding	4	0.022
GO:0016338~calcium-independent cell-cell adhesion	4	0.023
GO:0050853~B cell receptor signaling pathway	3	0.025
GO:0034707~chloride channel complex	6	0.027
GO:0019840~isoprenoid binding	4	0.028
GO:0016247~channel regulator activity	6	0.028
GO:0016917~GABA receptor activity	4	0.032
GO:0022604~regulation of cell morphogenesis	9	0.032
GO:0022834~ligand-gated channel activity	9	0.032
GO:0015276~ligand-gated ion channel activity	9	0.032
GO:0042802~identical protein binding	28	0.034
GO:0050909~sensory perception of taste	5	0.035
GO:0000166~nucleotide binding	80	0.037
GO:0016879~ligase activity, forming carbon-nitrogen bonds	13	0.038
GO:0008624~induction of apoptosis by extracellular signals	8	0.039
GO:0001505~regulation of neurotransmitter levels	6	0.040
GO:0046474~glycerophospholipid biosynthetic process	6	0.042
GO:0051219~phosphoprotein binding	4	0.043
GO:0002260~lymphocyte homeostasis	4	0.043
GO:0007268~synaptic transmission	15	0.043
GO:0006887~exocytosis	8	0.044
GO:0051960~regulation of nervous system development	11	0.045
GO:0045309~protein phosphorylated amino acid binding	3	0.046
GO:0048365~Rac GTPase binding	3	0.046
GO:0015459~potassium channel regulator activity	4	0.047
GO:0007369~gastrulation	6	0.047
GO:0031988~membrane-bounded vesicle	26	0.048
GO:0030554~adenyl nucleotide binding	58	0.049
Schizophrenia		
GO:0032446~protein modification by small protein conjugation	9	0.000
GO:0016567~protein ubiquitination	8	0.000
GO:0070647~protein modification by small protein conjugation or removal	9	0.000
GO:0019787~small conjugating protein ligase activity	9	0.000
GO:0007268~synaptic transmission	12	0.000
GO:0016879~ligase activity, forming carbon-nitrogen bonds	10	0.001
GO:0007267~cell-cell signaling	17	0.001
GO:0004842~ubiquitin-protein ligase activity	8	0.001
GO:0016881~acid-amino acid ligase activity	9	0.001
GO:0019226~transmission of nerve impulse	12	0.001
GO:0006650~glycerophospholipid metabolic process	7	0.002
GO:0042493~response to drug	9	0.002
GO:0045202~synapse	12	0.003
GO:0008360~regulation of cell shape	5	0.003
GO:0016247~channel regulator activity	5	0.003
GO:0006644~phospholipid metabolic process	8	0.005
GO:0008654~phospholipid biosynthetic process	6	0.005

GO:0019637~organophosphate metabolic process	8	0.006
GO:0046474~glycerophospholipid biosynthetic process	5	0.006
GO:0030054~cell junction	14	0.006
GO:0005230~extracellular ligand-gated ion channel activity	5	0.007
GO:0044456~synapse part	9	0.007
GO:0030384~phosphoinositide metabolic process	5	0.008
GO:0046486~glycerolipid metabolic process	7	0.008
GO:0046489~phosphoinositide biosynthetic process	4	0.009
GO:0045017~glycerolipid biosynthetic process	5	0.011
GO:0044265~cellular macromolecule catabolic process	16	0.012
GO:0007610~behavior	12	0.013
GO:0022604~regulation of cell morphogenesis	6	0.014
GO:0008022~protein C-terminus binding	6	0.016
GO:0004890~GABA-A receptor activity	3	0.017
GO:0044432~endoplasmic reticulum part	10	0.018
GO:0007010~cytoskeleton organization	11	0.020
GO:0005124~scavenger receptor binding	2	0.021
GO:0009057~macromolecule catabolic process	16	0.021
GO:0019941~modification-dependent protein catabolic process	13	0.022
GO:0043632~modification-dependent macromolecule catabolic process	13	0.022
GO:0016917~GABA receptor activity	3	0.026
GO:0006512~ubiquitin cycle	3	0.027
GO:0051603~proteolysis involved in cellular protein catabolic process	13	0.029
GO:0015267~channel activity	10	0.029
GO:0031988~membrane-bounded vesicle	13	0.029
GO:0022803~passive transmembrane transporter activity	10	0.030
GO:0044257~cellular protein catabolic process	13	0.030
GO:0007611~learning or memory	5	0.032
GO:0005789~endoplasmic reticulum membrane	8	0.035
GO:0001505~regulation of neurotransmitter levels	4	0.036
GO:0030163~protein catabolic process	13	0.037
GO:0007015~actin filament organization	4	0.043
GO:0031982~vesicle	14	0.043
GO:0042175~nuclear envelope-endoplasmic reticulum network	8	0.044
GO:0022834~ligand-gated channel activity	5	0.044
GO:0015276~ligand-gated ion channel activity	5	0.044
Epilepsy		
GO:0007218~neuropeptide signaling pathway	3	0.008
GO:0055085~transmembrane transport	5	0.009
GO:0048585~negative regulation of response to stimulus	3	0.009
GO:0032101~regulation of response to external stimulus	3	0.023

Note: Significance was determined as $p<.05$.

Table S4. List of significant processes for strict distribution among all 4 disorders from pathway analysis.

ID-ASD-Schizophrenia (Figure 1D, #1)	**# Genes**	***p* value**
GO:0042802~identical protein binding	10	0.000
GO:0008360~regulation of cell shape	4	0.001
GO:0022604~regulation of cell morphogenesis	5	0.002
GO:0004890~GABA-A receptor activity	3	0.002
GO:0016917~GABA receptor activity	3	0.003
GO:0007268~synaptic transmission	6	0.006
GO:0031982~vesicle	9	0.006
GO:0042803~protein homodimerization activity	6	0.007
GO:0007267~cell-cell signaling	8	0.008
GO:0031988~membrane-bounded vesicle	8	0.009
GO:0042493~response to drug	5	0.009
GO:0019226~transmission of nerve impulse	6	0.011
GO:0031410~cytoplasmic vesicle	8	0.016
GO:0044456~synapse part	5	0.019
GO:0034707~chloride channel complex	3	0.022
GO:0016023~cytoplasmic membrane-bounded vesicle	7	0.026
GO:0001505~regulation of neurotransmitter levels	3	0.027
GO:0005230~extracellular ligand-gated ion channel activity	3	0.028
GO:0005254~chloride channel activity	3	0.028
GO:0007369~gastrulation	3	0.030
GO:0031404~chloride ion binding	3	0.032
GO:0005253~anion channel activity	3	0.032
GO:0007610~behavior	6	0.034
GO:0003001~generation of a signal involved in cell-cell signaling	3	0.042
GO:0043168~anion binding	3	0.044
GO:0046983~protein dimerization activity	6	0.045
GO:0030594~neurotransmitter receptor activity	3	0.046
ID-Schizophrenia-Epilepsy (Figure 1D, #2)		
GO:0007218~neuropeptide signaling pathway	3	0.006
GO:0048585~negative regulation of response to stimulus	3	0.007
GO:0032101~regulation of response to external stimulus	3	0.017
GO:0055085~transmembrane transport	4	0.032
GO:0031348~negative regulation of defense response	2	0.044
GO:0007613~memory	2	0.048
ID-ASD (Figure 1D, #3)		
GO:0030054~cell junction	4	0.014
GO:0016338~calcium-independent cell-cell adhesion	2	0.026
GO:0046982~protein heterodimerization activity	3	0.030
ID-Schizophrenia (Figure 1D, #4)		
GO:0005789~endoplasmic reticulum membrane	5	0.001
GO:0042175~nuclear envelope-endoplasmic reticulum network	5	0.001

GO:0044432~endoplasmic reticulum part	5	0.002
GO:0046474~glycerophospholipid biosynthetic process	3	0.003
GO:0045017~glycerolipid biosynthetic process	3	0.004
GO:0008654~phospholipid biosynthetic process	3	0.007
GO:0008610~lipid biosynthetic process	4	0.007
GO:0006650~glycerophospholipid metabolic process	3	0.009
GO:0046486~glycerolipid metabolic process	3	0.017
GO:0006644~phospholipid metabolic process	3	0.023
GO:0019637~organophosphate metabolic process	3	0.026
GO:0005922~connexon complex	2	0.036
GO:0012505~endomembrane system	5	0.036
GO:0006879~cellular iron ion homeostasis	2	0.038
GO:0005911~cell-cell junction	3	0.038
GO:0006506~GPI anchor biosynthetic process	2	0.042
GO:0006505~GPI anchor metabolic process	2	0.043
GO:0055072~iron ion homeostasis	2	0.044
GO:0005921~gap junction	2	0.047
ASD-Schizophrenia (Figure 1D, #5)		
No results		
ASD-only (Figure 1D, #6)		
GO:0044456~synapse part	3	0.004
GO:0045202~synapse	3	0.007
GO:0030054~cell junction	3	0.015
GO:0014069~postsynaptic density	2	0.027
GO:0045664~regulation of neuron differentiation	2	0.039
GO:0050804~regulation of synaptic transmission	2	0.040
GO:0051969~regulation of transmission of nerve impulse	2	0.043
GO:0031644~regulation of neurological system process	2	0.044
GO:0050767~regulation of neurogenesis	2	0.048
ID-only (Figure 1D, #7)		
GO:0005031~tumor necrosis factor receptor activity	4	0.000
GO:0043120~tumor necrosis factor binding	4	0.001
GO:0050909~sensory perception of taste	5	0.009
GO:0007264~small GTPase mediated signal transduction	13	0.013
GO:0017076~purine nucleotide binding	52	0.013
GO:0007265~Ras protein signal transduction	7	0.014
GO:0032553~ribonucleotide binding	50	0.014
GO:0032555~purine ribonucleotide binding	50	0.014
GO:0051219~phosphoprotein binding	4	0.015
GO:0043065~positive regulation of apoptosis	16	0.016
GO:0043068~positive regulation of programmed cell death	16	0.017
GO:0010942~positive regulation of cell death	16	0.018
GO:0045309~protein phosphorylated amino acid binding	3	0.022
GO:0016481~negative regulation of transcription	16	0.028
GO:0030036~actin cytoskeleton organization	10	0.028
GO:0019717~synaptosome	6	0.029

GO:0016597~amino acid binding	5	0.030
GO:0042981~regulation of apoptosis	24	0.030
GO:0043067~regulation of programmed cell death	24	0.033
GO:0010941~regulation of cell death	24	0.034
GO:0006855~multidrug transport	2	0.037
GO:0019001~guanyl nucleotide binding	14	0.039
GO:0032561~guanyl ribonucleotide binding	14	0.039
GO:0005353~fructose transmembrane transporter activity	2	0.039
GO:0030029~actin filament-based process	10	0.039
GO:0046578~regulation of Ras protein signal transduction	9	0.046
GO:0015149~hexose transmembrane transporter activity	3	0.048
GO:0007242~intracellular signaling cascade	33	0.049
Schizophrenia-only (Figure 1D, #8)		
GO:0016247~channel regulator activity	2	0.031

Note: Strict criteria classification was defined as copy number variants (CNVs) that were significant for a given disorder. See Figure 1D for distribution groupings. ASD = autism spectrum disorder; ID = intellectual disability.

Table S5. List of significant processes for broad distribution among all 4 disorders from pathway analysis.

ID-ASD-Schizophrenia-Epilepsy (Figure 1J, #1)	**# Genes**	***p* value**
GO:0008360~regulation of cell shape	5	0.000
GO:0022604~regulation of cell morphogenesis	6	0.001
GO:0030054~cell junction	10	0.001
GO:0042802~identical protein binding	10	0.002
GO:0015267~channel activity	8	0.002
GO:0022803~passive transmembrane transporter activity	8	0.002
GO:0004890~GABA-A receptor activity	3	0.003
GO:0005230~extracellular ligand-gated ion channel activity	4	0.004
GO:0034702~ion channel complex	6	0.004
GO:0007268~synaptic transmission	7	0.004
GO:0016917~GABA receptor activity	3	0.005
GO:0044459~plasma membrane part	21	0.005
GO:0030594~neurotransmitter receptor activity	4	0.008
GO:0044456~synapse part	6	0.008
GO:0019226~transmission of nerve impulse	7	0.009
GO:0045202~synapse	7	0.009
GO:0042165~neurotransmitter binding	4	0.010
GO:0042803~protein homodimerization activity	6	0.014
GO:0031348~negative regulation of defense response	3	0.014
GO:0015276~ligand-gated ion channel activity	4	0.017
GO:0022834~ligand-gated channel activity	4	0.017
GO:0031982~vesicle	9	0.020
GO:0007010~cytoskeleton organization	7	0.024
GO:0031988~membrane-bounded vesicle	8	0.025
GO:0005216~ion channel activity	6	0.025
GO:0022838~substrate specific channel activity	6	0.028
GO:0031224~intrinsic to membrane	37	0.029
GO:0016021~integral to membrane	36	0.029
GO:0045211~postsynaptic membrane	4	0.031
GO:0034707~chloride channel complex	3	0.032
GO:0007610~behavior	7	0.032
GO:0007267~cell-cell signaling	8	0.033
GO:0005254~chloride channel activity	3	0.039
GO:0000226~microtubule cytoskeleton organization	4	0.039
GO:0007017~microtubule-based process	5	0.040
GO:0006811~ion transport	9	0.041
GO:0031404~chloride ion binding	3	0.044
GO:0031410~cytoplasmic vesicle	8	0.044
GO:0022836~gated channel activity	5	0.044
GO:0005253~anion channel activity	3	0.045
GO:0000287~magnesium ion binding	6	0.045
GO:0001505~regulation of neurotransmitter levels	3	0.046
ID-ASD-Epilepsy (Figure 1J, #2)		
GO:0005031~tumor necrosis factor receptor activity	4	0.000
GO:0043120~tumor necrosis factor binding	4	0.000
GO:0050909~sensory perception of taste	5	0.006

GO:0007265~Ras protein signal transduction	7	0.009
GO:0050853~B cell receptor signaling pathway	3	0.009
GO:0043065~positive regulation of apoptosis	15	0.014
GO:0007264~small GTPase mediated signal transduction	12	0.014
GO:0043068~positive regulation of programmed cell death	15	0.015
GO:0010552~positive regulation of specific transcription from RNA polymerase II promoter	5	0.015
GO:0010942~positive regulation of cell death	15	0.016
GO:0045309~protein phosphorylated amino acid binding	3	0.018
GO:0019717~synaptosome	6	0.019
GO:0017076~purine nucleotide binding	47	0.020
GO:0016597~amino acid binding	5	0.022
GO:0032555~purine ribonucleotide binding	45	0.023
GO:0032553~ribonucleotide binding	45	0.023
GO:0005829~cytosol	35	0.025
GO:0046578~regulation of Ras protein signal transduction	9	0.026
GO:0051056~regulation of small GTPase mediated signal transduction	10	0.027
GO:0006855~multidrug transport	2	0.033
GO:0005353~fructose transmembrane transporter activity	2	0.035
GO:0050778~positive regulation of immune response	7	0.036
GO:0015149~hexose transmembrane transporter activity	3	0.040
GO:0008624~induction of apoptosis by extracellular signals	6	0.041
GO:0005089~Rho guanyl-nucleotide exchange factor activity	5	0.043
GO:0015145~monosaccharide transmembrane transporter activity	3	0.044
GO:0006887~exocytosis	6	0.045
GO:0032582~negative regulation of gene-specific transcription	4	0.047
GO:0016481~negative regulation of transcription	14	0.047
ID-Schizophrenia-Epilepsy (Figure 1J, #3)		
GO:0005789~endoplasmic reticulum membrane	5	0.001
GO:0042175~nuclear envelope-endoplasmic reticulum network	5	0.002
GO:0046474~glycerophospholipid biosynthetic process	3	0.003
GO:0044432~endoplasmic reticulum part	5	0.004
GO:0045017~glycerolipid biosynthetic process	3	0.004
ID-ASD (Figure 1J, #4)		
GO:0009881~photoreceptor activity	2	0.011
GO:0018298~protein-chromophore linkage	2	0.012
GO:0007602~phototransduction	2	0.024
GO:0009583~detection of light stimulus	2	0.028
GO:0009582~detection of abiotic stimulus	2	0.047
ID-Epilepsy (Figure 1J, #5)		
GO:0042995~cell projection	5	0.045
ASD-Schizophrenia (Figure 1J, #6)		
No results		
ASD-only (Figure 1J, #7)		

No results		
ID-only (Figure 1J, #8)		
GO:0000781~chromosome, telomeric region	2	0.025
Schizophrenia-only (Figure 1J, #9)		
No results		

Note: Broad criteria classification was defined as copy number variants (CNVs) that were significant for a given disorder yet also exhibit the phenotype of another disorder of cognitive development. See Figure 1J for distribution groupings. ASD = autism spectrum disorder; ID = intellectual disability.

Table S6. List of significant processes for copy number variant (CNV) categories from pathway analysis.

A	# Genes	*p* value
GO:0042493~response to drug	12	0.001
GO:0008092~cytoskeletal protein binding	19	0.002
GO:0017076~purine nucleotide binding	48	0.004
GO:0006650~glycerophospholipid metabolic process	8	0.004
GO:0000166~nucleotide binding	54	0.004
GO:0030036~actin cytoskeleton organization	11	0.005
GO:0046474~glycerophospholipid biosynthetic process	6	0.006
GO:0031090~organelle membrane	31	0.006
GO:0005789~endoplasmic reticulum membrane	12	0.006
GO:0015631~tubulin binding	7	0.006
GO:0022604~regulation of cell morphogenesis	8	0.007
GO:0007010~cytoskeleton organization	16	0.007
GO:0012505~endomembrane system	24	0.007
GO:0030054~cell junction	18	0.007
GO:0008654~phospholipid biosynthetic process	7	0.007
GO:0030029~actin filament-based process	11	0.008
GO:0042175~nuclear envelope-endoplasmic reticulum network	12	0.009
GO:0050853~B cell receptor signaling pathway	3	0.009
GO:0045017~glycerolipid biosynthetic process	6	0.011
GO:0002260~lymphocyte homeostasis	4	0.011
GO:0032555~purine ribonucleotide binding	44	0.012
GO:0032553~ribonucleotide binding	44	0.012
GO:0008360~regulation of cell shape	5	0.013
GO:0006887~exocytosis	7	0.013
GO:0006644~phospholipid metabolic process	9	0.015
GO:0044432~endoplasmic reticulum part	13	0.015
GO:0051028~mRNA transport	6	0.015
GO:0015893~drug transport	3	0.019
GO:0046486~glycerolipid metabolic process	8	0.020
GO:0019637~organophosphate metabolic process	9	0.020
GO:0042802~identical protein binding	19	0.021
GO:0004707~MAP kinase activity	3	0.022
GO:0001776~leukocyte homeostasis	4	0.022
GO:0050658~RNA transport	6	0.024
GO:0051236~establishment of RNA localization	6	0.024
GO:0050657~nucleic acid transport	6	0.024
GO:0032446~protein modification by small protein conjugation	7	0.024
GO:0016079~synaptic vesicle exocytosis	3	0.025
GO:0006403~RNA localization	6	0.027
GO:0046489~phosphoinositide biosynthetic process	4	0.029
GO:0030554~adenyl nucleotide binding	37	0.030
GO:0007265~Ras protein signal transduction	6	0.032
GO:0005230~extracellular ligand-gated ion channel activity	5	0.032
GO:0008017~microtubule binding	5	0.032
GO:0007015~actin filament organization	5	0.033
GO:0005124~scavenger receptor binding	2	0.033
GO:0006855~multidrug transport	2	0.033

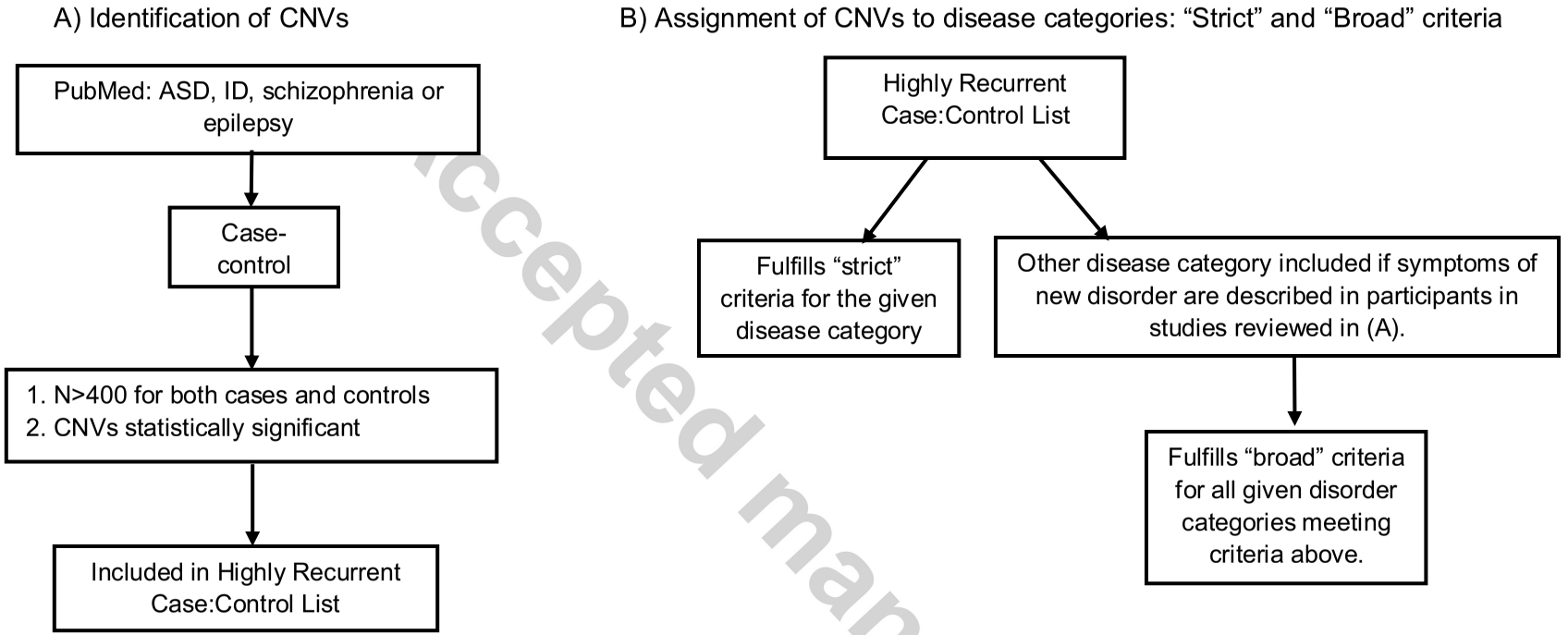
GO:0030384~phosphoinositide metabolic process	5	0.034
GO:0031967~organelle envelope	18	0.035
GO:0031975~envelope	18	0.036
GO:0001883~purine nucleoside binding	37	0.036
GO:0004890~GABA-A receptor activity	3	0.039
GO:0001882~nucleoside binding	37	0.039
GO:0045202~synapse	12	0.040
GO:0015931~nucleobase, nucleoside, nucleotide and nucleic acid transport	6	0.042
GO:0007267~cell-cell signaling	17	0.046
GO:0043029~T cell homeostasis	3	0.048
GO:0007163~establishment or maintenance of cell polarity	4	0.049
B		
GO:0050804~regulation of synaptic transmission	4	0.001
GO:0051969~regulation of transmission of nerve impulse	4	0.001
GO:0031644~regulation of neurological system process	4	0.001
GO:0044057~regulation of system process	4	0.009
GO:0044456~synapse part	4	0.009
GO:0009881~photoreceptor activity	2	0.019
GO:0018298~protein-chromophore linkage	2	0.022
GO:0050767~regulation of neurogenesis	3	0.022
GO:0045211~postsynaptic membrane	3	0.024
GO:0045202~synapse	4	0.025
GO:0048729~tissue morphogenesis	3	0.026
GO:0046928~regulation of neurotransmitter secretion	2	0.029
GO:0051960~regulation of nervous system development	3	0.029
GO:0060284~regulation of cell development	3	0.033
GO:0006512~ubiquitin cycle	2	0.033
GO:0043169~cation binding	11	0.034
GO:0051588~regulation of neurotransmitter transport	2	0.036
GO:0035295~tube development	3	0.038
GO:0043167~ion binding	11	0.038
GO:0007416~synaptogenesis	2	0.044
GO:0007602~phototransduction	2	0.045
GO:0060341~regulation of cellular localization	3	0.047
C		
GO:0005031~tumor necrosis factor receptor activity	4	0.000
GO:0043120~tumor necrosis factor binding	4	0.000
GO:0050909~sensory perception of taste	5	0.004
GO:0019717~synaptosome	6	0.011
GO:0016481~negative regulation of transcription	14	0.015
GO:0043065~positive regulation of apoptosis	13	0.021
GO:0042981~regulation of apoptosis	20	0.022
GO:0043068~positive regulation of programmed cell death	13	0.022
GO:0010942~positive regulation of cell death	13	0.023
GO:0043067~regulation of programmed cell death	20	0.024
GO:0010941~regulation of cell death	20	0.025
GO:0010629~negative regulation of gene expression	14	0.030

GO:0015149~hexose transmembrane transporter activity	3	0.030
GO:0005353~fructose transmembrane transporter activity	2	0.030
GO:0032582~negative regulation of gene-specific transcription	4	0.031
GO:0015145~monosaccharide transmembrane transporter activity	3	0.033
GO:0045934~negative regulation of nucleobase, nucleoside, nucleotide and nucleic acid metabolic process	14	0.033
GO:0016202~regulation of striated muscle tissue development	4	0.035
GO:0048546~digestive tract morphogenesis	3	0.036
GO:0055123~digestive system development	3	0.036
GO:0043193~positive regulation of gene-specific transcription	5	0.036
GO:0051172~negative regulation of nitrogen compound metabolic process	14	0.036
GO:0048634~regulation of muscle development	4	0.037
GO:0005501~retinoid binding	3	0.040
GO:0008276~protein methyltransferase activity	4	0.040
GO:0032583~regulation of gene-specific transcription	6	0.044
GO:0030036~actin cytoskeleton organization	8	0.044
GO:0005549~odorant binding	2	0.045
GO:0019840~isoprenoid binding	3	0.047
GO:0010552~positive regulation of specific transcription from RNA polymerase II promoter	4	0.048
GO:0048742~regulation of skeletal muscle fiber development	3	0.049

Figure S1. Flow diagram detailing the method for significant copy number variants (CNVs) identification from eligible genome-wide association studies (GWAS) (1A). Note: Inclusion criteria includes (1) case-control methodology, (2) participant N>400 in both cases and controls, and (3) statistically significant CNVs in cases. Figure S2, available online, shows the process for assigning CNVs to disease categories (i.e. autism spectrum disorder [ASD], intellectual disability [ID], schizophrenia, and epilepsy) based on strict and broad criteria.

Figure S2. Graphic illustrations of prototypical copy number variant (CNV) categories (e.g. A, B, and C). Note: This figure is used to demonstrate how CNVs were assigned to one of the three categories. ASD = autism spectrum disorder; ID = intellectual disability.

Supplemental Figure 1



Supplemental Figure 2