

Supplementary Table S3. Microdeletion burden analysis sample count.

Microdeletions Sets	N° Genes	Affymetrix GenomeWide 6.0		Illumina Omni Express Array		
		GGE Cases (%), N=1,366	Ctrl (%), N=5,234	RE Cases (%), N=281	AFE Cases (%), N=807	Ctrl (%), N=1,512
All microdeletions		100 (7.32%)	208 (3.97%)	9 (3.2%)	24 (2.97%)	26 (1.71%)
Overlapping CNV constrained genes	20,206	78 (5.71%)	129 (2.46%)	9 (3.2%)	15 (1.85%)	16 (1.05%)
Overlapping Neurodevelopmental genes	1,559	59 (4.31%)	52 (0.99%)	3 (1.06%)	8 (0.99%)	10 (0.66%)
Overlapping ASD-Related genes	1,683	44 (3.22%)	45 (0.85%)	7 (2.49%)	6 (0.74%)	9 (0.59%)
Overlapping developmental disorders genes	294	6 (0.43%)	2 (0.03%)	1 (0.35%)	2 (0.24%)	1 (0.06%)
Overlapping loss of function intolerant genes	2,506	46 (3.36%)	39 (0.74%)	6 (2.13%)	6 (0.74%)	8 (0.52%)
Overlapping hotspots loci genes	330	38 (2.78%)	20 (0.38%)	3 (1.06%)	2 (0.24%)	2 (0.13%)
Microdeletions Sets outside hotspots						
All microdeletions outside hotspots		63 (4.61%)	189 (3.61%)	7 (2.49%)	22 (2.72%)	25 (1.65%)
Overlapping CNV constrained genes outside hotspots		40 (2.92%)	110 (2.10%)	7 (2.49%)	13 (1.61%)	14 (0.92%)
Overlapping Neurodevelopmental genes outside hotspots		25 (1.83%)	34 (0.64%)	2 (0.71%)	6 (0.74%)	8 (0.52%)
Overlapping ASD-Related genes outside hotspots		10 (0.73%)	28 (0.53%)	4 (1.42%)	4 (0.49%)	8 (0.52%)
Overlapping developmental disorders genes outside hotspots		1 (0.07%)	0 (0.00%)	1 (0.35%)	1 (0.12%)	1 (0.06%)
Overlapping loss of function intolerant genes outside hotspots		12 (0.87%)	22 (0.42%)	4 (1.42%)	4 (0.49%)	7 (0.46%)
Control						
Overlapping random genes	4,696	19 (1.39%)	71 (1.35%)	3 (1.06%)	7 (0.86%)	7 (0.46%)