Rare Copy Number Variation in Treatment-Resistant Major Depressive Disorder

Supplemental Information

Table S1. Burden of deletions and duplications in individuals with treatment-resistant major depressive disorder, by study cohort

Deletions	**i2b2 cohort**					**STAR*D cohort**				
		CNV burden (number)		**CNV burden (gene count)**			**CNV burden (number)**		**CNV burden (gene count)**	
	total	**P**	**case/control ratio**	**P**	**case/control ratio**	**total**	**P**	**case/control ratio**	**P**	**case/control ratio**
all	261	0.981	0.756	0.941	0.617	286	0.853	0.851	0.989	0.473
frequency										
1	131	0.896	0.778	0.956	0.440	106	0.620	0.865	0.919	0.372
2-6	81	0.920	0.753	0.982	0.415	139	0.811	0.885	0.900	0.644
size (kb)										
100-200	159	0.997	0.610	0.997	0.403	200	0.634	0.939	0.694	0.841
200-500	85	0.528	1.011	0.920	0.494	65	0.878	0.716	0.977	0.353
500+	17	0.503	1.115	0.412	1.187	21	0.935	0.516	0.961	0.124
Duplications										
		CNV burden (number)		**CNV burden (gene count)**			**CNV burden (number)**		**CNV burden (gene count)**	
	total	**P**	**case/control ratio**	**P**	**case/control ratio**	**total**	**P**	**case/control ratio**	**P**	**case/control ratio**
all	390	0.419	1.040	**0.048**	**1.501**	390	0.209	1.108	0.319	1.099
frequency										
1	175	0.191	1.195	0.113	1.525	150	0.320	1.095	0.620	0.883
2-6	140	0.682	0.941	0.157	1.433	176	0.460	1.049	0.548	1.001
size (kb)										
100-200	219	0.592	0.973	**0.049**	**1.672**	171	0.630	0.957	0.598	0.959
200-500	126	0.291	1.120	0.294	1.188	139	0.433	1.049	0.669	0.894
500+	45	0.370	1.164	0.150	1.644	80	**0.022**	**1.620**	0.174	1.464

Table S2. Results in loci known to be associated with schizophrenia or autism

Region	Type	p(control)	STAR*D case:control	i2b2 case:control	Observed	Expected
1q21.1	del	0.020%	0:0	0:1	1	0.25
1q21.1	dup	0.030%	0:1	0:1	2	0.38
3q29	del	0.002%	0:0	0:0	0	0.03
7q11.23	del	0.000%	0:0	0:0	0	0.00
7q11.23	dup	0.000%	0:0	0:0	0	0.00
7q36.3	dup	0.060%	1:0	0:0	1	0.76
15q11.2	del	0.270%	2:2	2:0	6	3.41
15q11.2-13.1	dup	0.009%	0:0	0:0	0	0.11
15q13.3	del	0.020%	0:0	0:0	0	0.25
16p13.11	dup	0.130%	1:1	1:0	3	1.64
16p11.2	del	0.040%	0:0	0:0	0	0.51
16p11.2	dup	0.030%	0:1	0:0	1	0.38
17p12	del	0.020%	0:0	0:1	1	0.25
17q12	del	0.006%	0:0	0:2	2	0.08
22q11.21	del	0.000%	0:0	0:0	0	0.00
22q11.2	dup	0.080%	1:0	1:0	2	1.01
			5:5	4:5	19	9.06

Prevalence's drawn from Malhotra and Sebat:

Malhotra D, Sebat J (2012): CNVs: harbingers of a rare variant revolution in psychiatric genetics. *Cell.* 148:1223-41.