Exome Sequencing to Detect Rare Variants Associated With General Cognitive Ability

Twin Research and Human Genetics

Exome Sequencing to Detect Rare Variants Associated With General Cognitive Ability: A Pilot Study

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Supplementary Table S1

Results From Gene Set Analysis for Significant Variants With Frequency <0.01

	Obesity controls			Depression controls		
	Gene	Number of variants	*p*-value	Gene	Number of variants	*p*-value
All variants				*RP11-414H17.5*	2	1.34^{-6}
				RP11-118B18.1	2	1.97^{-6}
				MESP2/SNORD113-9	2	5.51^{-6}
Nonsynonymous	***SYNGAP1***	**3**	**4.0^{-6}**	***SYNGAP1***	**2**	**1.23^{-6}**
				HOXD1/ HOXD-AS1	2	8.82^{-7}
				CECR6	2	6.22^{-7}
				AC022201.5	2	1.04^{-5}
				CYP26C1	2	6.99^{-6}
				ZNF703	2	5.75^{-7}
				NFKBIL1	3	2.77^{-5}
Synonymous				*C9orf66*	2	1.92^{-8}
				FAM110C	2	3.91^{-6}
				ID4/ RP1-167F1.2	2	9.91^{-6}
				TBC1	2	3.44^{-6}

Note: Significant genes containing a single variant are not shown. Bold indicates common results across case-control subgroup analyses.

Supplementary Table S2

Results From Gene Set Analysis for Significant Variants With Frequency <0.05

	Obesity controls			Depression controls		
	Gene	Number of variants	*p*-value	Gene	Number of variants	*p*-value
All variants	***RP11-673E1.4/ GYPB***	**14**	**2.74^{-12}**	***RP11-673E1.4/GYPB***	**11**	**9.94^{-7}**
	/GYPA	**/ 9**	**5.76^{-12}**	***/GYPA***	**/ 7**	**8.05^{-7}**
				RP11-414H17.5	2	1.16^{-6}
				RP11-118B18.1	2	1.08^{-6}
Nonsynonymous	***RP11-673E1.4/ GYPB***	**6**	**4.35^{-12}**	***RP11-673E1.4/ GYPB***	**5**	**2.85^{-7}**
	/GYPA	**/ 2**	**5.17^{-12}**	***/GYPA***	**/ 2**	**2.4^{-7}**
				CECR6	2	4.41^{-8}
				FAM136A/AC022201.5	2	4.62^{-6}
				ZNF703	2	7.40^{-7}
Synonymous				*SOX17*	2	2.61^{-7}
				FAM110C	2	4.16^{-6}
				ID4/RP1-167F1.2	2	9.35^{-6}

Note: Significant genes containing a single variant are not shown. Bold indicates common results across case-control subgroup analyses.

Supplementary Table S3

Significant Gene Ontology Pathways Enriched in the Varying Analyses Comprising the Depression Controls

	Gene ontology	*p*-value	FDR *p*-value	Enrichment values*	Genes in pathway
Synonymous					
SNVs<.01					
Molecular Function GO:0008376	acetylgalactosaminyl transferase activity	1.23E-5	.03	5.63 (6764,18,667,10)	*GALNT6* — udp-n-acetyl-alpha-d-galactosamine:polypeptide n-acetylgalactosaminyltransferase 6 (galnac-t6) *GALNT12* — udp-n-acetyl-alpha-d-galactosamine:polypeptide n-acetylgalactosaminyltransferase 12 (galnac-t12) *GALNT10* — udp-n-acetyl-alpha-d-galactosamine:polypeptide n-acetylgalactosaminyltransferase 10 (galnac-t10) *B4GALNT3* — beta-1,4-n-acetyl-galactosaminyl transferase 3 *GALNT18* — udp-n-acetyl-alpha-d-galactosamine:polypeptide n-acetylgalactosaminyltransferase 18 *GALNT3* — udp-n-acetyl-alpha-d-galactosamine:polypeptide n-acetylgalactosaminyltransferase 3 (galnac-t3) *GALNT2* — udp-n-acetyl-alpha-d-galactosamine:polypeptide n-acetylgalactosaminyltransferase 2 (galnac-t2) *CHPF* — chondroitin polymerizing factor *B3GALNT2* — beta-1,3-n-acetylgalactosaminyltransferase 2 *B4GALNT4* — beta-1,4-n-acetyl-galactosaminyl transferase 4

Note: *p*-value, FDR corrected *p*-value, enrichment values, and prominent genes in each pathway are listed.
SNV: Single nucleotide variants* Enrichment is defined as (b/n)/(B/N) [*N*: Total number of genes; B: Total number of genes associated with a specific GO term, n: Number of genes in the 'target set', b: Number of genes in the 'target set' associated with a specific GO term].