

Supplementary Material

Identification of common variants influencing risk of the tauopathy Progressive Supranuclear Palsy

Günter U. Höglinger, Nadine M. Melhem, Dennis W. Dickson, Patrick M.A. Sleiman, Li-San Wang, Lambertus Klei, Rosa Rademakers, Rohan de Silva, Irene Litvan, David E. Riley, John C. van Swieten, Peter Heutink, Zbigniew K. Wszolek, Ryan J. Uitti, Jana Vandrovcova, Howard I. Hurtig, Rachel G. Gross, Walter Maetzler, Stefano Goldwurm, Eduardo Tolosa, Barbara Borroni, Pau Pastor, PSP Genetics Study Group[#], Laura B. Cantwell, Mi Ryung Han, Allissa Dillman, Marcel P. van der Brug, J Raphael Gibbs, Mark R. Cookson, Dena G. Hernandez, Andrew B. Singleton, Matthew J. Farrer, Chang-En Yu, Lawrence I. Golbe, Tamas Revesz, John Hardy, Andrew J. Lees, Bernie Devlin, Hakon Hakonarson, Ulrich Müller, Gerard D. Schellenberg

[#]PSP Genetics Study Group coauthors: Roger L. Albin, Elena Alonso, Angelo Antonini, Manuela Apfelbacher, Steven E. Arnold, Jesus Avila, Thomas G. Beach, Sherry Beecher, Daniela Berg, Thomas D. Bird, Nenad Bogdanovic, Agnita J.W. Boon, Yvette Bordelon, Alexis Brice, Herbert Budka, Margherita Canesi, Wang Zheng Chiu, Roberto Cilia, Carlo Colosimo, Peter P. De Deyn, Justo García de Yébenes, Laura Donker Kaat, Ranjan Duara, Alexandra Durr, Sebastiaan Engelborghs, Giovanni Fabbrini, NiCole A. Finch, Robyn Flook, Matthew P. Frosch, Carles Gaig, Douglas R. Galasko, Thomas Gasser, Marla Gearing, Evan T. Geller, Bernardino Ghetti, Neill R. Graff-Radford, Murray Grossman, Deborah A. Hall, Lili-Naz Hazrati, Matthias Höllerhage, Joseph Jankovic, Jorge L. Juncos, Anna Karydas, Hans A. Kretschmar, Isabelle Leber, Virginia M. Lee, Andrew P. Lieberman, Kelly E. Lyons, Claudio Mariani, Eliezer Masliah, Luke A. Massey, Catriona A. McLean, Nicoletta Meucci, Bruce L. Miller, Brit Mollenhauer, Jens C. Möller, Huw R. Morris, Chris Morris, Sean S. O'Sullivan, Wolfgang H. Oertel, Donatella Ottaviani, Alessandro Padovani, Rajesh Pahwa, Gianni Pezzoli, Stuart Pickering-Brown, Werner

Poewe, Alberto Rabano, Alex Rajput, Stephen G Reich, Gesine Respondek, Sigrun Roeber, Jonathan D. Rohrer, Owen A. Ross, Martin N. Rossor, Giorgio Sacilotto, William W. Seeley, Klaus Seppi, Laura Silveira-Moriyama, Salvatore Spina, Karin Srulijes, Peter St. George-Hyslop, Maria Stamelou, David G. Standaert, Silvana Tesei, Wallace W. Tourtellotte, Claudia Trenkwalder, Claire Troakes, John Q. Trojanowski, Juan C. Troncoso, Vivianna M. Van Deerlin, Jean Paul G. Vonsattel, Gregor K. Wenning, Charles L. White, Pia Winter, Chris Zarow, Anna L. Zecchinelli

Supplementary Table 1. PSP subjects with autopsy confirmation of diagnosis (Stage 1)

Source	Total	Number Genotyped	Number in Analysis
Ludwig-Maximilians-Universität and Brain Net Europe	21	21	21
University of Würzburg	2	2	2
Universitat de Barcelona Hospital Clínic and Banc de Teixits Neurològics	17	17	16
Universidad Autónoma de Madrid	5	5	4
Erasmus University Medical Center	23	23	23
London Neurodegenerative Diseases Brain Bank	8	8	8
University of Saskatchewan	37	37	35
University of Antwerp	2	2	2
Australian Brain Bank Network and Flinders University	9	9	9
University of Navarra	1	1	1
Mayo Clinic College of Medicine	588	538	514
University of Louisville	13	11	11
Reta Lila Weston Institute of Neurological Studies	144	142	141
Emory University	6	6	6
McLean Brain Bank	64	63	62
Indiana University School of Medicine	31	30	29
West Los Angeles Veterans Affairs Medical Center, University of California Los Angeles	14	14	14
Massachusetts General Institute for Neurodegenerative Disease Research	34	34	34
Columbia University, NY Brain Bank	22	22	21
Rancho Los Amigos Medical Center	3	0	0
Sun Health Research Institute	38	38	38
University of Michigan	23	22	20
University of Pennsylvania	51	50	50

Supplementary Table 1 (continued). PSP subjects with autopsy confirmation of diagnosis (Stage 1)

Source	Total	Number Genotyped	Number in Analysis
University of Washington	4	4	4
University of Southern California	2	1	1
University of California San Diego	10	10	10
University of Texas Southwestern Medical Center	14	14	12
Johns Hopkins University	27	26	26
Total	1213	1150	1114

Supplementary Table 2. Results from Stage 1, Stage 2, and joint analysis for PSP for subjects of all ancestries

Chr band	SNP Location (bp)	MAF ^a Cases	MAF Controls	Gene or nearby gene	Stage 1		Stage 2		Joint P	
					OR ^b /CI	P	OR/CI	P	OR/CI	P
SNPs Significant at P < 5 x 10⁻⁸										
1q25.3	rs1411478 179,229,155	0.50	0.42	<i>STX6</i>	0.72 0.65 - 0.80	3.3 x 10 ⁻¹⁰	0.85 0.77 - 0.94	1.5 x 10 ⁻³	0.79 0.73 - 0.84	3.5 x 10 ⁻¹¹
2p11.2	rs7571971 88,676,716	0.32	0.26	<i>EIF2AK3</i>	0.76 0.67 - 0.85	1.0 x 10 ⁻⁶	0.75 0.67 - 0.83	8.7 x 10 ⁻⁸	0.75 0.70 - 0.82	4.2 x 10 ⁻¹³
3p22.1	rs1768208 39,498,257	0.36	0.29	<i>MOBP</i>	0.71 0.63 - 0.79	6.3 x 10 ⁻¹⁰	0.74 0.67 - 0.82	1.3 x 10 ⁻⁸	0.73 0.67 - 0.78	5.3 x 10 ⁻¹⁷
17q21.31	rs8070723 41,436,651	0.05	0.23	<i>MAPT</i>	5.57 4.48 - 6.94	1.8 x 10 ⁻⁵³	4.74 3.92 - 5.74	4.8 x 10 ⁻⁶⁷	5.11 4.43 - 5.91	1.5 x 10 ⁻¹¹⁸
	rs242557 41,375,823	0.53	0.35	<i>MAPT</i>	0.48 0.43 - 0.54	3.1 x 10 ⁻³⁸	0.54 0.49 - 0.59	5.0 x 10 ⁻³⁵	0.51 0.48 - 0.55	2.7 x 10 ⁻⁷¹
	^d rs242557/ rs8070723	---	---	<i>MAPT</i>	0.66 0.59 - 0.74	7.5 x 10 ⁻¹²	0.74 0.67 - 0.83	6.3 x 10 ⁻⁸	0.70 0.65 - 0.76	8.5 x 10 ⁻¹⁸
^eSNPs with strongly suggestive evidence of association										
1q41	rs6687758 220,231,821	0.23	0.19	<i>None</i>	0.77 0.68 - 0.87	4.9 x 10 ⁻⁵	0.80 0.71 - 0.91	3.0 x 10 ⁻⁴	0.79 0.72 - 0.86	6.2 x 10 ⁻⁸
10q11.21	rs2142991 42,661,361	0.14	0.17	<i>BMS1</i>	1.35 1.16 - 1.56	6.1 x 10 ⁻⁵	1.26 1.10 - 1.44	1.2 x 10 ⁻³	1.30 1.18 - 1.44	3.2 x 10 ⁻⁷
12p12.1	rs11568563 21,348,951	0.08	0.05	<i>SLCO1A2</i>	0.66 0.54 - 0.81	7.3 x 10 ⁻⁵	0.70 0.58 - 0.85	2.4 x 10 ⁻⁴	0.68 0.59 - 0.78	7.0 x 10 ⁻⁸

Supplementary Table 2 (continued). Results from Stage 1, Stage 2, and joint analysis for PSP for subjects of all ancestries

Chr band	SNP Location (bp)	MAF ^a Cases	MAF Controls	Gene or nearby gene	Stage 1		Stage 2		Joint P	
					OR ^b /CI	P	OR/CI	P	OR/CI	P
SNPs where allele frequencies in young and old controls differ										
2p11.2	rs6547705 86,898,077	0.18	0.23	<i>CD8B</i>	1.27 1.12 - 1.45	3.1 x 10 ⁻⁴	1.29 1.14 - 1.46	4.4 x 10 ⁻⁵	1.28 1.17 - 1.40	5.2 x 10 ⁻⁸
	^c rs6547705/ rs7571971	---	---	<i>CD8B</i>	1.28 1.13 - 1.46	8.5 x 10 ⁻⁵	1.29 1.14 - 1.46	4.1 x 10 ⁻⁵	1.28 1.18 - 1.40	1.4 x 10⁻⁸
4q27	rs6852535 123,698,416	0.29	0.34	<i>IL2/IL21</i>	1.22 1.10 - 1.37	3.3 x 10 ⁻⁴	1.23 1.11 - 1.38	1.1 x 10 ⁻⁴	1.23 1.13 - 1.32	1.3 x 10 ⁻⁷
6p25.3	rs12203592 341,571	0.16	0.19	<i>IRF4</i>	1.33 1.16 - 1.53	5.5 x 10 ⁻⁵	1.63 1.42 - 1.87	4.7 x 10⁻¹²	1.48 1.34 - 1.63	6.2 x 10⁻¹⁵

^aMAF, minor allele frequency; ^bOR based on major allele, ^crs6547705 controlling for rs7571971; ^drs242557 controlling for rs8070723; ^e5.0 ≤ |Z_{joint}| ≤ 5.44 or 5.7 x 10⁻⁷ ≤ P < 5 x 10⁻⁸

Supplementary Table 3. Comparison of Minor Allele Frequencies (MAF) of SNPs with significant or suggestive evidence of association with PSP, based on European PSP cases and controls and older European controls from three publicly available datasets.

Chr band	SNP Location (bp)	Gene or nearby gene	PSP cases		PSP controls		dbGap ^a older controls				Weighted ^b MAF dbGap older controls			
			Stage 1	Stage 2	Stage 1	Stage 2	Health ABC ^c	NIA AD ^d	Cataract ^e	Overall	Health ABC	NIA AD	Cataract	Overall
SNPs Significant at P<5·10⁻⁸														
1q25.3	rs1411478 179,229,155	<i>STX6</i>	0.496	0.464	0.418	0.426	0.414	0.407	0.426	0.416	0.404	0.395	0.434	0.411
2p11.2	rs7571971 88,676,716	<i>EIF2AK3</i>	0.314	0.307	0.257	0.248	0.261	0.283	0.272	0.270	0.265	0.285	0.284	0.276
3p22.1	rs1768208 39,498,257	<i>MOBP</i>	0.357	0.353	0.286	0.287	0.285	0.282	0.275	0.281	0.286	0.275	0.275	0.280
17q21.31	rs8070723 41,436,651	<i>MAPT</i>	0.051	0.060	0.235	0.232	0.231	0.230	0.210	0.224	0.232	0.234	0.211	0.223
	rs242557 41,375,823		0.530	0.495	0.348	0.355	0.350	0.369	0.359	0.358	0.352	0.362	0.359	0.357
^fSNPs with strongly suggestive evidence of association														
1q41	rs6687758 220,231,821	<i>None</i>	0.229	0.227	0.189	0.191	0.211	0.200	0.196	0.204	0.211	0.200	0.194	0.203
10q11.21	rs2142991 42,661,361	<i>BMS1</i>	0.139	0.139	0.173	0.169	0.176	0.155	0.163	0.167	0.178	0.152	0.170	0.169
12p12.1	rs11568563 21,348,951	<i>SLCO1A2</i>	0.078	0.077	0.053	0.055	0.060	0.059	0.058	0.059	0.068	0.062	0.057	0.063
SNPs where allele frequencies differ between young and old controls														
2p11.2	rs6547705 ^g 86,898,077	<i>CD8B</i>	0.187	0.187	0.229	0.230	0.209	0.194	0.218	0.208	0.201	0.193	0.215	0.203
4q27	rs6852535 ^g 123,698,416	<i>IL2/IL21</i>	0.289	0.277	0.338	0.322	0.316	0.279	0.292	0.299	0.308	0.277	0.294	0.296
6p25.3	rs12203592 ^g 341,571	<i>IRF4</i>	0.164	0.131	0.200	0.198	0.200	0.164	0.120	0.166	0.206	0.176	0.125	0.172
6p25.3	rs2493013 ^h 461,660	<i>EXOC2</i>	0.228	0.251	0.203	0.209	0.202	0.200	0.233	0.211	0.202	0.205	0.231	0.212

^aThe datasets used for the analyses described were obtained from Database for Genotypes and Phenotypes (dbGap) at <http://www.ncbi.nlm.nih.gov/gap>. A total of 3,816 controls from three studies were included. ^bAllele frequencies in dbGap controls weighted by ancestry representation of the PSP controls. ^cWhole Genome Association Study of Visceral Adiposity in the Health Aging and Body Composition (Health ABC) Study (Study accession number: phs000169.v1.p1); ^dNational Institute on Aging - Late Onset Alzheimer's Disease Family Study: Genome-Wide Association Study for Susceptibility Loci (Study accession number: phs000168.v1.p1), General Research Use and Non-Profit only consent groups; ^eA Genome-Wide Association Study on Cataract and HDL in the Personalized Medicine Research Project Cohort (Study accession number: phs000170.v1.p1); ^f $5.0 \leq |Z_{\text{joint}}| \leq 5.44$ or $5.7 \times 10^{-7} \leq P < 5 \times 10^{-8}$; ^grs12203592, rs6547705, and rs6852535 had significantly different allele frequencies between PSP controls and dbGap older controls; ^hrs2493013 approached threshold for strongly suggestive evidence ($Z_{\text{joint}} = -4.99$, $p = 6.0 \times 10^{-7}$) and had significantly different allele frequencies between PSP controls and Cataract controls.

Supplementary Table 4. Clinical and autopsy-documented PSP subjects used in Stage 2

Source	Total n (autopsied)	Genotyped n (autopsied)	Analysis n (autopsied)
University of Pennsylvania	25 (10)	25 (10)	23 (8)
Reta Lila Weston Institute of Neurological Studies	138	108	11
UCL Institute of Neurology	7	7	7
University of Brescia	64	58	58
Parkinson Institute, Istituti Clinici di Perfezionamento	78	76	75
Erasmus University Medical Center	199	184	183
Philipps-Universität	41	41	41
University of Toronto	11 (11)	11 (11)	6 (6)
Mayo Clinic College of Medicine	199 (105)	187 (94)	171 (89)
University of Louisville	226 (2)	210 (2)	195 (1)
Newcastle University	6 (6)	6 (6)	5 (5)
Cardiff University, Royal Gwent Hospital	20	19	19
University of Manchester	17	17	16
Hertie-Institute for Clinical Brain Research, and German Center for Neurodegenerative Diseases	92	92	92
Hôpital Pitié-Salpêtrière	55	44	28
Georg-August University and Paracelsus-Elena Hospital	19	17	12
Universitat de Barcelona Hospital Clínic and Banc de Teixits Neurològics	43	42	41

Supplementary Table 4 (continued). Clinical and autopsy-documented PSP subjects used in Stage 2

Source	Total n (autopsied)	Genotyped n (autopsied)	Analysis n (autopsied)
University of Navarra	66	57	28
Sapienza Università di Roma	9	9	8
University of California San Francisco	23 (8)	21 (8)	20 (8)
Innsbruck Medical University	13	13	12
Universidad Autónoma de Madrid Spain	3 (3)	0	0
Totals	1354 (142)	1244 (131)	1051 (117)

Supplementary Table 5. Results for SNPs significant even after controlling for most significant SNP in the region (European Ancestry)

Chr band	SNP Location (bp)	MAF Cases	MAF Controls	Gene or nearby gene	Stage 1		Stage 2		Joint P	
					OR / CI	P	OR / CI	P	OR/CI	P
2p11.2	rs6547705 ^a / rs7571971	0.19	0.23	<i>CD8B</i>	1.26 1.10 - 1.43	8.8 x 10 ⁻⁴	1.29 1.14 - 1.46	4.1 x 10 ⁻⁵	1.28 1.16 - 1.40	1.4 x 10 ⁻⁷
6p25.3	rs2493013 ^b 461,600	0.23	0.20	<i>EXOC2</i>	0.83 0.73 - 0.89	3.1 x 10 ⁻³	0.79 0.71 - 0.89	5.4 x 10 ⁻⁵	0.81 0.74 - 0.88	6.0 x 10 ⁻⁷
	rs2493013 ^c / rs12203592	----	----		0.85 0.76 - 0.97	3.1 x 10 ⁻²	0.83 0.74 - 0.93	1.0 x 10 ⁻³	0.84 0.77 - 0.91	5.3 x 10 ⁻⁵
17q21.3 ^d	rs11869096 41,300,889	0.57	0.43	<i>LOC100128977</i>	0.79 0.70 - 0.89	9.5 x 10 ⁻⁵	0.80 0.72 - 0.89	5.8 x 10 ⁻⁵	0.80 0.73 - 0.86	2.1 x 10 ⁻⁸
	rs11867549 41,369,064	0.39	0.24	<i>MAPT</i>	0.69 0.61 - 0.78	2.7 x 10 ⁻⁹	0.75 0.68 - 0.84	2.5 x 10 ⁻⁷	0.72 0.66 - 0.78	4.7 x 10 ⁻¹⁵
	rs242557 41,375,573	0.53	0.35		0.66 0.58 - 0.74	1.3 x 10 ⁻¹¹	0.74 0.67 - 0.83	6.3 x 10 ⁻⁸	0.70 0.65 - 0.76	9.5 x 10 ⁻¹⁸
	rs2435211 41,419,081	0.42	0.29		0.79 0.70 - 0.90	2.1 x 10 ⁻⁴	0.91 0.81 - 1.01	7.5 x 10 ⁻²	0.85 0.78 - 0.92	1.3 x 10 ⁻⁴
	rs6503454 41,474,116	0.25	0.15	<i>KIAA1267</i>	0.71 0.62 - 0.82	2.6 x 10 ⁻⁶	0.80 0.71 - 0.91	5.2 x 10 ⁻⁴	0.76 0.69 - 0.83	8.9 x 10 ⁻⁹
	rs7225002 41,544,850	0.32	0.41		0.75 0.66-0.86	4.8 x 10 ⁻⁵	0.83 0.73 - 0.93	1.8 x 10 ⁻³	0.79 0.72 - 0.86	4.2 x 10 ⁻⁷
	rs2016730 41,560,151	0.26	0.17		0.75 0.65 - 0.86	2.9 x 10 ⁻⁵	0.75 0.66 - 0.84	3.0 x 10 ⁻⁶	0.75 0.68 - 0.82	3.7 x 10 ⁻¹⁰
	rs4792831 41,562,443	0.26	0.17		0.75 0.65 - 0.86	4.2 x 10 ⁻⁵	0.80 0.70 - 0.90	2.6 x 10 ⁻⁴	0.78 0.71 - 0.85	4.6 x 10 ⁻⁸
	rs17692129 42,148,466	0.44	0.31	<i>NSF</i>	0.75 0.66 - 0.85	2.5 x 10 ⁻⁶	0.86 0.77 - 0.96	5.8 x 10 ⁻³	0.80 0.74 - 0.87	1.6 x 10 ⁻⁷
rs11650531 42,229,159	0.33	0.26	<i>WNT3</i>	0.79 0.70 - 0.89	1.4 x 10 ⁻⁴	0.90 0.80 - 1.00	5.9 x 10 ⁻²	0.84 0.78 - 0.92	6.8 x 10 ⁻⁵	

^ars6547705 controlling for rs7571971, the most significant SNP in the 2p11.2 region; ^brs2493013, main effect only; ^crs2493013 controlling for rs12203592, the most significant SNP in the 6p25.3 region; ^dSNPs controlling for rs8070723; P<1·10⁻³ was used for significance of SNPs when controlling for other SNP

Supplementary Table 6. Results from Stage 1, Stage 2, and joint analysis for subjects of European ancestry: SNPs with suggestive evidence of association

Chr band	SNP Location (bp)	Gene or nearby gene	Stage 1				Stage 2				Joint P	
			MAF ^a Cases	MAF Controls	OR ^b /CI	P	MAF ^a Cases	MAF Controls	OR/CI	P	OR/CI	P
1q41	rs6687758 220,231,821	<i>None</i>	0.23	0.19	0.79 0.69 - 0.89	2.3 x 10 ⁻⁴	0.23	0.19	0.8 0.71-0.91	3.1 x 10 ⁻⁴	0.80 0.73-0.87	2.8 x 10 ⁻⁷
10q11.21	rs2142991 42,661,361	<i>BMS1</i>	0.14	0.17	1.35 1.16 - 1.56	8.6 x 10 ⁻⁵	0.14	0.17	1.26 1.10-1.44	1.2 x 10 ⁻³	1.30 1.17-1.44	4.9 x 10 ⁻⁷
12p12.1	rs11568563 21,348,951	<i>SLCO1A2</i>	0.08	0.05	0.67 0.55 - 0.83	2.1 x 10 ⁻⁴	0.08	0.06	0.7 0.58-0.85	2.4 x 10 ⁻⁴	0.69 0.60-0.79	1.9 x 10 ⁻⁷

^aMAF, minor allele frequency; ^bOR based on major allele

Abbreviations and gene symbols: *SLCO1A2*, solute carrier organic anion transporter family member 1a2; *BMS1*; BMS1-like, ribosome assembly protein; A summary of the function of each gene listed is in Supplementary Table 4.

Supplementary Table 8. Overlap between PSP GWAS results and those for AD and PD

GWAS	CHR	SNP	Gene	Nearby HapMap SNP with LD info	LD Region: r-squared>=0.3		PSP Stage 1		PSP Stage 2		PSP Stage 1+2		
					START	END	Top SNP	P	Top SNP	P	Top SNP	P	Gene
AD	1	rs3818361	CR1	-	205,720,018	205,873,612	none	-	none	-	none	-	-
	1	rs6701713	CR1	-	205,720,018	205,873,612	none	-	none	-	none	-	-
	2	rs7561528	BIN1	-	127,560,126	127,611,085	none	-	none	-	none	-	-
	2	rs744373	BIN1	-	127,556,004	127,611,085	none	-	none	-	none	-	-
	6	rs9349407	CD2AP	-	47,502,755	47,561,337	none	-	none	-	none	-	-
	7	rs11767557	EPHA1	-	142,809,229	142,819,261	none	-	none	-	none	-	-
	8	rs1532278	CLU	-	27,512,170	27,582,264	rs520769	0.00201	rs520769	0.0489	rs520769	0.000384	2Kbp upstream of SCARA3
	10	rs2588969	ARID5B	-	63,227,654	63,307,282	none	-	none	-	none	-	-
	11	rs610932	MS4A6A	-	59,583,253	59,856,488	none	-	none	-	none	-	-
	11	rs670139	MS4A4E	-	59,570,863	59,856,488	none	-	none	-	none	-	-
	11	rs4938933	MS4A4A	-	59,593,673	59,791,005	none	-	none	-	none	-	-
	11	rs561655	PICALM	-	85,312,382	85,548,832	none	-	rs713346	0.069	rs10792820	0.076	PICALM
	19	rs3764650	ABCA7	-	992,352	1,007,492	none	-	none	-	none	-	-
	19	rs3752246	ABCA7	-	997,520	1,032,617	none	-	none	-	none	-	-
19	rs4420638	APOE	-	50,084,094	50,114,786	rs2075650	0.0034	rs2075650	0.00121	rs2075650	1.25 x 10 ⁻⁵	TOMM40	
19	rs3865444	CD33	-	56,396,851	56,429,803	none	-	none	-	none	-	-	
PD	1	-	SYT11	rs822519	154,107,330	154,279,135	none	-	none	-	none	-	-
	2	rs6710823	ACMSD	rs12614226	135,154,655	135,329,387	none	-	none	-	none	-	-
	2	rs2102808	STK39	rs1850437	168,798,166	168,873,722	none	-	none	-	none	-	-
	3	rs11711441	MCCC1, LAMP3	rs11710342	184,211,987	184,344,424	none	-	none	-	none	-	-

Supplementary Table 8 (continued). Overlap between PSP GWAS results and those for AD and PD

GWAS	CHR	SNP	Gene	Nearby HapMap SNP with LD info	LD Region: $r^2 \geq 0.3$		PSP Stage 1		PSP Stage 2		PSP Stage 1+2		
					START	END	Top SNP	P	Top SNP	P	Top SNP	P	Gene
PD	4	-	GAK	rs6822424	804,080	950,081	none	-	none	-	none	-	-
	4	rs11724635	BST1	-	15,205,551	15,354,852	none	-	none	-	none	-	-
	4	rs356219	SNCA	-	90,825,620	91,038,809	none	-	none	-	none	-	-
	6	-	HLA-DRB5	rs35847514	32,395,097	32,765,894	none	-	none	-	none	-	-
	12	rs1491942	LRRK2	-	38,707,384	38,931,863	none	-	none	-	none	-	-
	12	rs12817488	CCDC62, HIP1R	rs7957643	121,779,189	121,951,396	none	-	none	-	none	-	-
	17	rs2942168	MAPT	-	40,872,185	41,268,567	rs7215239	3.31×10^{-51}	rs393152	8.28×10^{-71}	rs393152	1.41×10^{-120}	MAPT

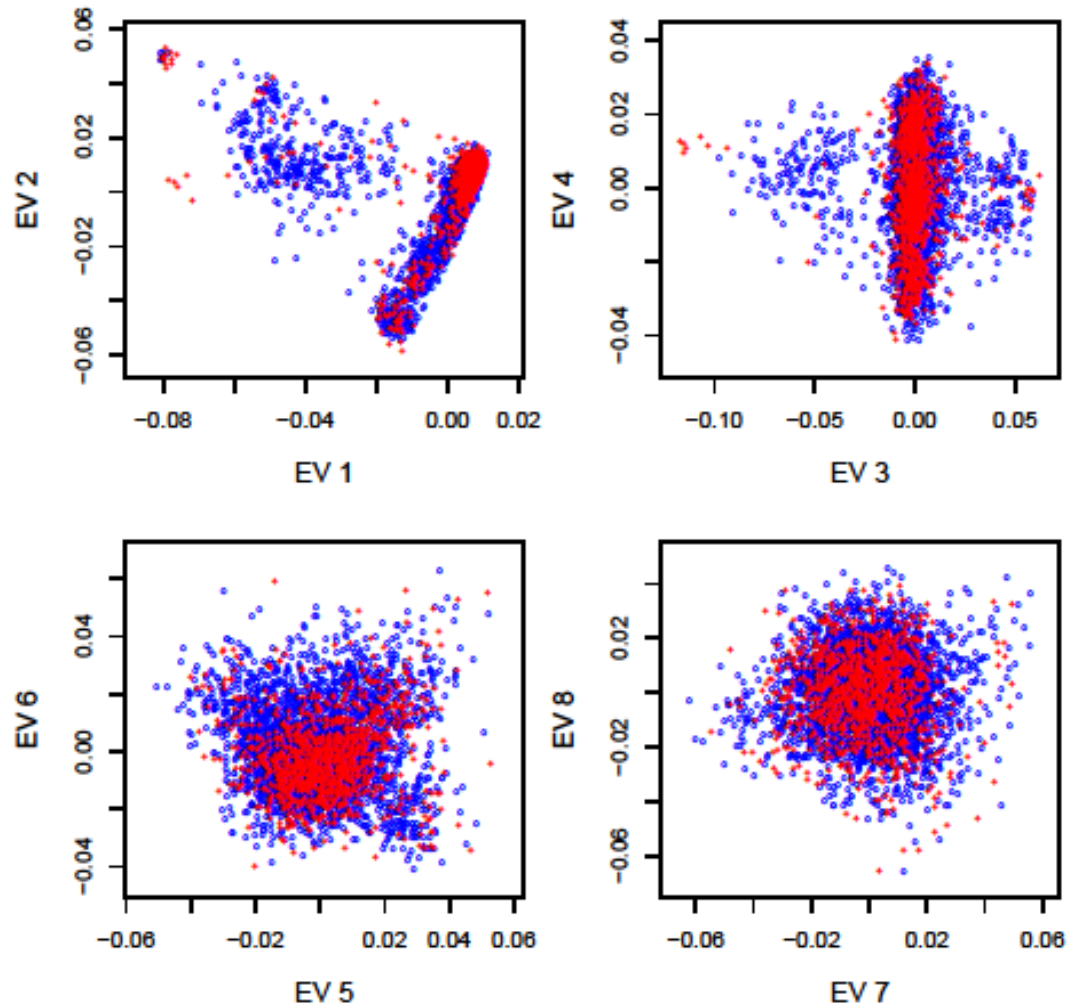
Overlap between GWAS signals PSP and for Parkinson's disease¹ and AD²⁻⁶. SNPs used were from published reports (for PD, Table 2 from the International Parkinson Disease Consortium¹; for AD, Table 1 from Hollingworth et al.⁶). For each AD or PD signal, LD regions surrounding the peak SNP were determined using the HapMap Release 27 CEU panel (http://hapmap.ncbi.nlm.nih.gov/downloads/ld_data/2009-04_rel27/) by finding the farthest flanking SNPs with $r^2 \geq 0.3$; if no LD information for the top SNP is available, the nearest SNP from HapMap is used. SNPs within the LD regions for either PD or AD loci, with the best p-values ($p < 0.1$) in the PSP dataset are listed. None indicates that there were no SNPs from the PSP GWAS with $P < 0.01$. At the CLU locus, while rs520769 exhibits the smallest p-value in the region, it is not correlated with rs1532278 ($r^2 = 0.06$), which is associated with risk for PD; another SNP genotyped in the PSP sample, rs1113600, is highly correlated ($r^2 = 0.96$) but its alleles show no association with PSP risk. The SNPs in the MAPT locus are all highly correlated ($r^2 \geq 0.94$). Correlation structure for the APOE locus is described in the body of the manuscript. All physical positions are from the Human Reference Genome Release 36.

Supplementary Table 9. PSP gene abbreviations and functions

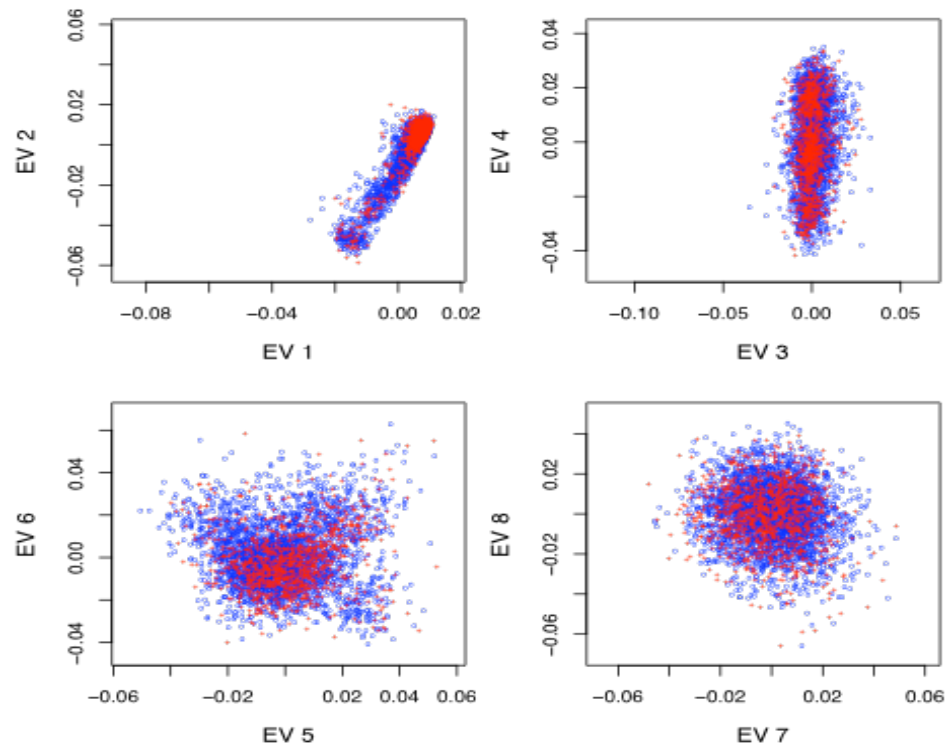
Gene		Function
Abbreviation	Name	
<i>BMS1</i>		Ribosome assembly protein
<i>EIF2AK3</i>	Eukaryotic translation initiation factor 2-alpha kinase 3	Inhibits translation initiation upon accumulation of mis-folded proteins in the endoplasmic reticulum (ER-stress)
<i>MAPT</i>	Microtubule associated protein tau	Stabilizes microtubules
<i>MOBP</i>	Myelin-Associated Oligodendrocytic Basic Protein	Abundant myelin constituent expressed exclusively by oligodendrocytes
<i>SLC01A2</i>	Solute carrier organic anion transporter family member 1a2	Sodium-independent transporter expressed in the apical blood-brain barrier
<i>STX6</i>	Syntaxin 6	Vesicle membrane fusion, Golgi-endosomes

Supplementary Table 10. Results of loci significant at $P < 5 \cdot 10^{-8}$ at Stage 1, Stage 2, or joint analysis for PSP after controlling for H1/H2 inversion among subjects of European ancestry

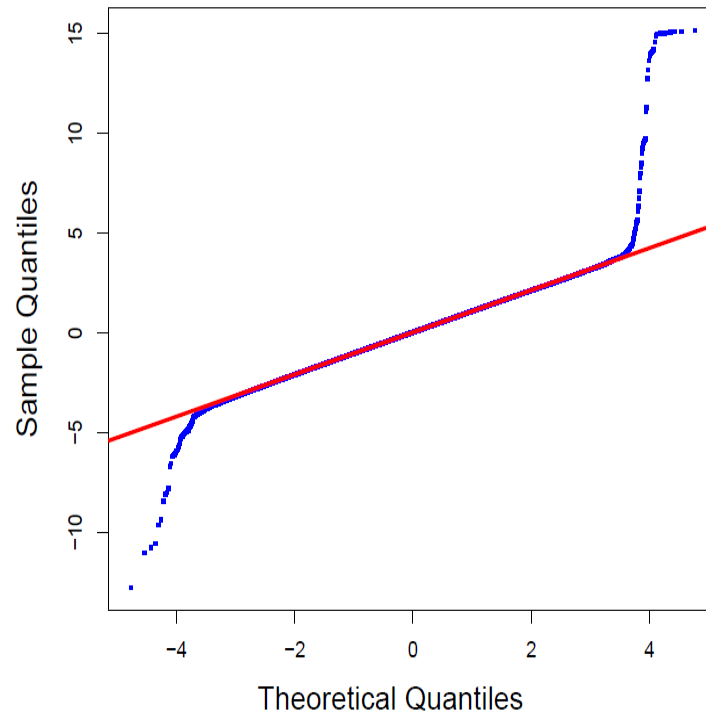
Chr band	SNP Location (bp)	Gene or nearby gene	Stage 1		Stage 2		Joint P	
			OR/CI	P	OR/CI	P	OR/CI	P
1q25.3	rs1411478 179,229,155	<i>STX6</i>	0.77 0.68 - 0.86	3.1×10^{-6}	0.85 0.77 - 0.94	2.2×10^{-3}	0.81 0.75 - 0.87	5.8×10^{-8}
2p11.2	rs7571971 88,676,716	<i>EIF2AK3</i>	0.75 0.67 - 0.86	1.1×10^{-5}	0.76 0.68 - 0.85	1.5×10^{-6}	0.76 0.69 - 0.82	7.0×10^{-11}
3p22.1	rs1768208 39,498,257	<i>MOBP</i>	0.71 0.63 - 0.80	2.9×10^{-8}	0.72 0.65 - 0.81	4.1×10^{-9}	0.72 0.66 - 0.78	6.2×10^{-16}
6p25.3	rs12203592 341,571	<i>IRF4</i>	1.34 1.15 - 1.57	1.6×10^{-4}	1.64 1.42-1.89	1.4×10^{-11}	1.49 1.33 - 1.65	6.1×10^{-14}



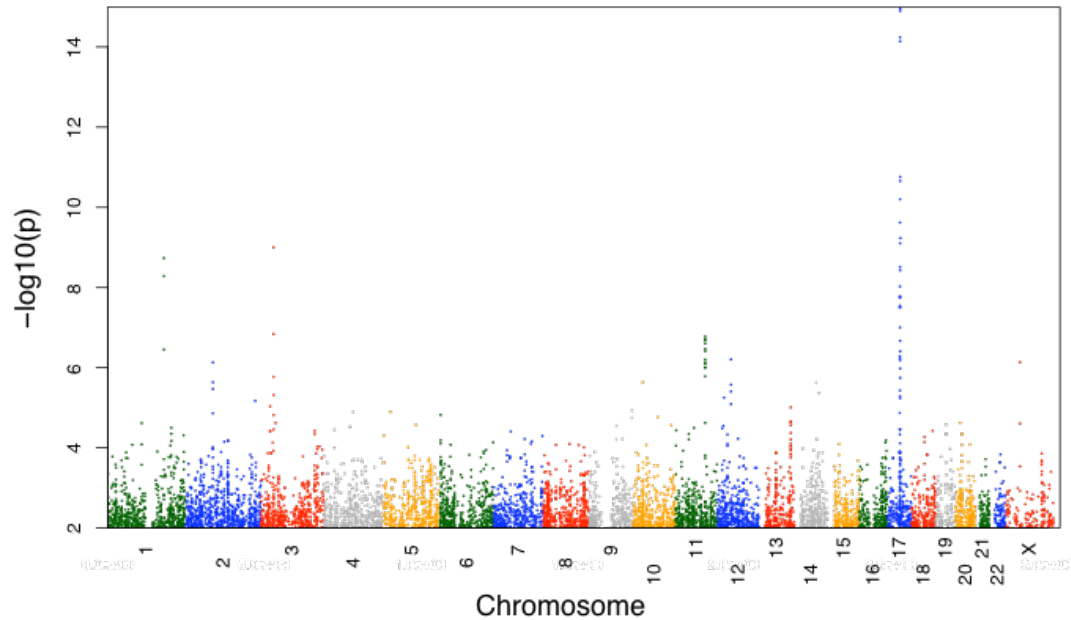
Supplementary Figure 1. Results from genetic ancestry analysis. SpectralGem was used to convert multilocus genotypes to ancestry dimension, expressed as eigenvectors. The decomposition resulted in an 8 dimensional space, of which only certain combinations of dimensions are shown here. Red symbols are cases, blue symbols are controls.



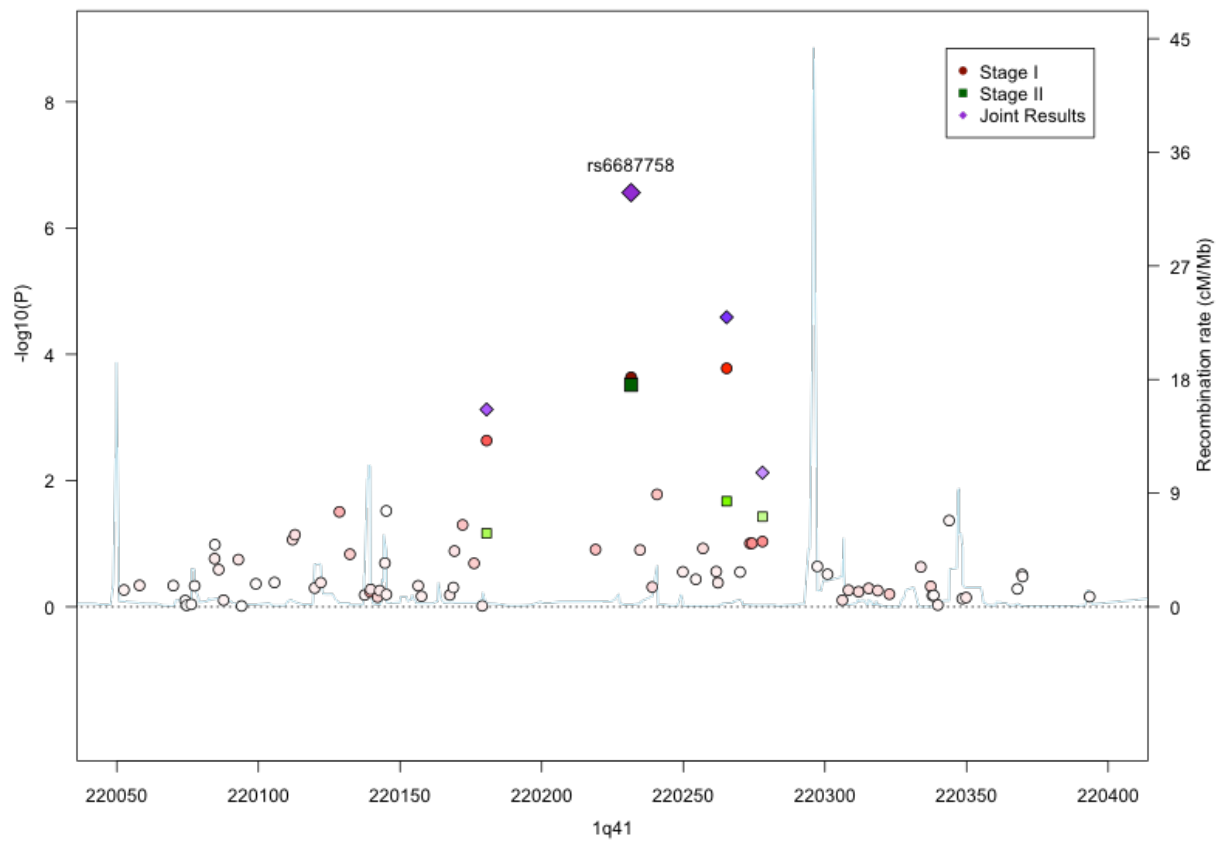
Supplementary Figure 2. Results from genetic ancestry analysis of subjects of European ancestry only. Red symbols are cases, blue symbols are controls.



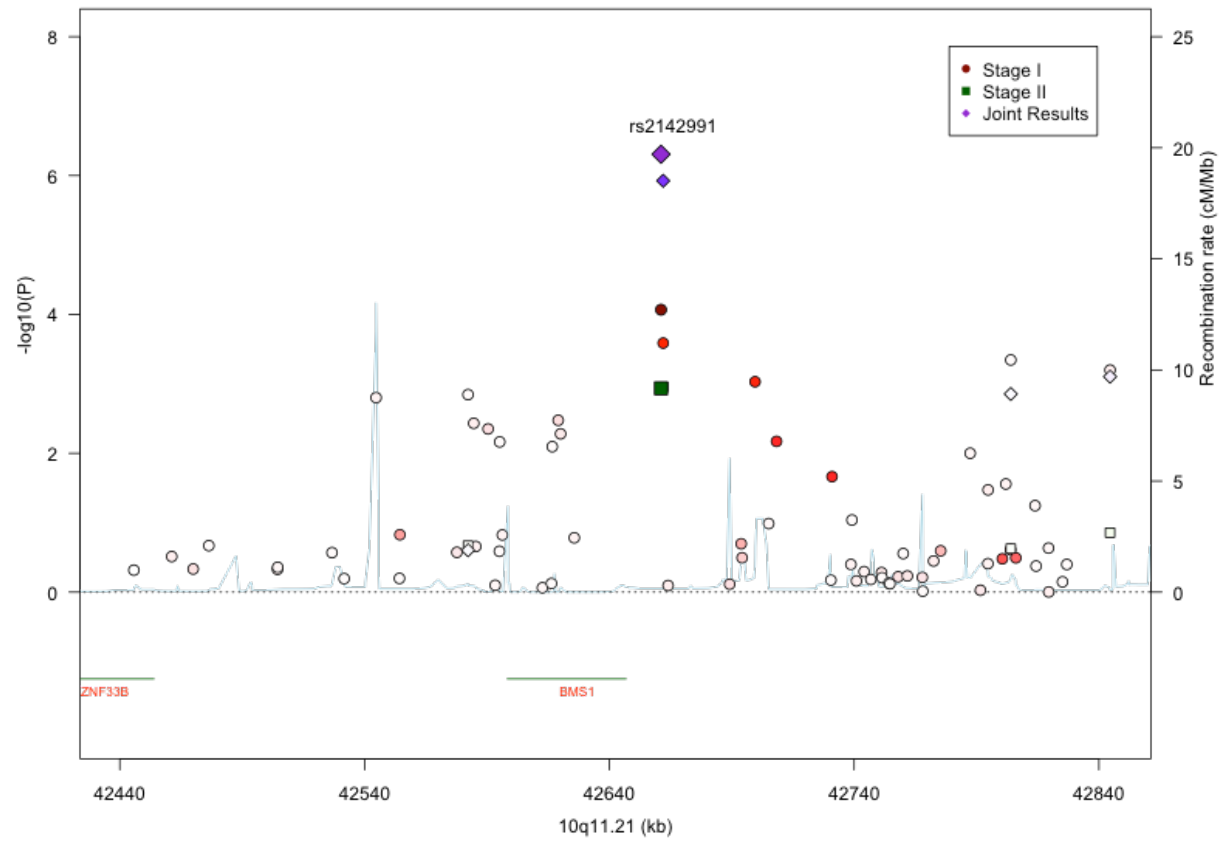
Supplementary Figure 3. Quantile-Quantile or Q-Q plot of association test statistics for subjects of European ancestry. Genomic inflation factor $\lambda=1.11$. If there were no difference between the observed distribution and that expected under the null hypothesis of no association, all points would fall on the line ($X=Y$).



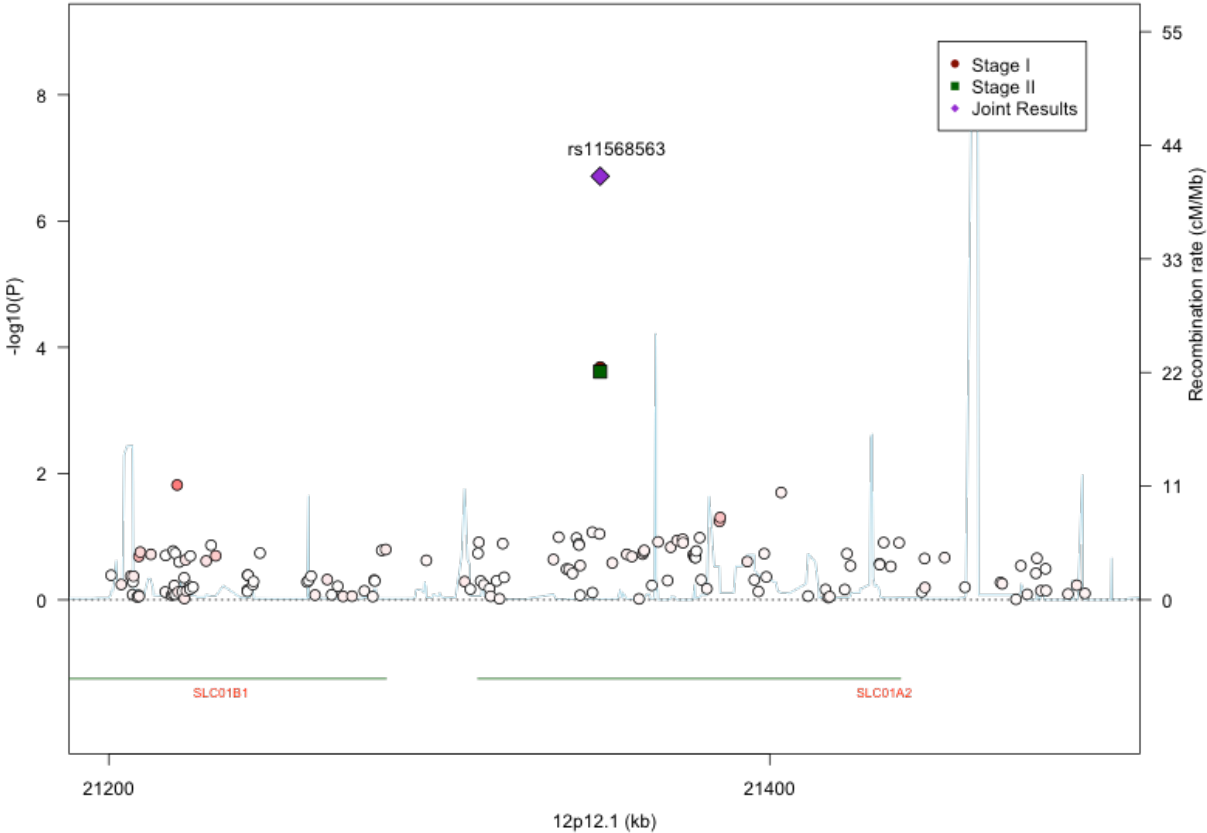
Supplementary Figure 4. Manhattan plot showing results for PSP data for subjects of European ancestry. Note that the vertical axis is truncated at 15, corresponding to $p\text{-value} = 10^{-15}$, even though P-values for some SNPs in the *MAPT* region of 17p fall well below that value. See Table 2 and Fig. 2, main text, for detailed results from the *MAPT* region.



Supplementary Figure 5A.



Supplementary Figure 5B.



Supplementary Figure 5C.

Supplementary Figure 5A-C. Manhattan plots for individual loci with strongly suggestive evidence of association in PSP among subjects of European ancestry. Recombination rate is derived from Hapmap3 data. Linkage disequilibrium (LD), encoded by intensity of the colors, is the pairwise LD of the most highly associated SNP at Stage 1 with each of the SNPs in the region.

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