

Supplementary table 4. USF1 SNPs and haplotypes and their association with SP type and NFT prevalence (APOE4 carriership as a covariate) in under 65 year old.

	rs10908821				rs2073658				rs2774276				rs2516839				rs1556259				rs2774279			
	Aff. (%) CC*/G+	OR	CI	p	Aff. (%) CC*/T+	OR	CI	p	Aff. (%) CC*/G+	OR	CI	p	Aff. (%) TT*/C+	OR	CI	p	Aff. (%) AA*/G+	OR	CI	p	Aff. (%) CC*/T+	OR	CI	p
SP	26(13.3)/4(6.7)	0.46	0.15-1.41	NS	14(12.6)/17(11.6)	0.99	0.46-2.16	NS	17(11.6)/14(12.4)	1.02	0.47-2.22	NS	12(12.8)/18(11.0)	0.86	0.39-1.91	NS	23(12.4)/8(11.0)	0.96	0.40-2.30	NS	16(11.0)/14(12.7)	1.10	0.50-2.42	NS
Non neuritic	17(8.7)/2(3.3)	0.34	0.08-1.55	NS	10(9.0)/10(6.8)	0.82	0.32-2.07	NS	8(5.4)/12(10.6)	1.87	0.73-4.78	NS	6(6.4)/13(8.0)	1.19	0.43-3.27	NS	15(8.1)/5(6.8)	0.86	0.30-2.49	NS	10(6.9)/9(8.2)	1.13	0.44-2.91	NS
Neuritic	10(5.1)/1(1.7)	0.29	0.04-2.54	NS	3(2.7)/8(5.4)	2.44	0.61-9.79	NS	9(6.1)/2(1.8)	0.26	0.05-1.24	NS	7(7.4)/4(2.5)	0.31	0.08-1.12	NS	9(4.8)/2(2.7)	0.62	0.13-3.04	NS	7(4.8)/4(3.6)	0.66	0.18-2.40	NS
Diffuse	8(4.1)/1(1.7)	0.36	0.04-3.00	NS	6(5.4)/4(2.7)	0.56	0.15-2.07	NS	1(0.7)/9(8.0)	11.1	1.37-89.7	0.024	1(1.1)/8(4.9)	4.40	0.54-36.0	NS	8(4.3)/2(2.7)	0.66	0.13-3.22	NS	5(3.4)/4(3.6)	0.98	0.25-3.80	NS
Primitive	9(4.6)/1(1.7)	0.33	0.04-2.64	NS	4(3.6)/6(4.1)	1.19	0.32-4.35	NS	7(4.8)/3(2.7)	0.55	0.14-2.18	NS	5(5.3)/5(3.1)	0.55	0.16-1.97	NS	7(3.8)/3(4.1)	1.08	0.27-4.32	NS	5(3.4)/5(4.5)	1.28	0.36-4.56	NS
Classic	10(5.1)/1(1.7)	0.29	0.04-2.35	NS	3(2.7)/8(5.4)	2.42	0.60-9.72	NS	9(6.1)/2(1.8)	0.26	0.05-1.27	NS	7(7.4)/4(2.5)	0.31	0.09-1.14	NS	9(4.8)/2(2.7)	0.62	0.13-3.03	NS	7(4.8)/4(3.6)	0.66	0.18-2.39	NS
Burnt out	0(0.0)/0(0.0)	.	.	.	0(0.0)/0(0.0)	.	.	.	0(0.0)/0(0.0)	.	.	.	0(0.0)/0(0.0)	.	.	.	0(0.0)/0(0.0)	.	.	.	0(0.0)/0(0.0)	.	.	.
Cerad ≤1.5%	20(10.2)/3(5.0)	0.45	0.13-1.60	NS	11(9.9)/13(8.8)	0.95	0.40-2.24	NS	11(7.5)/13(11.5)	1.46	0.62-3.45	NS	8(8.5)/15(9.2)	1.06	0.43-2.64	NS	18(9.7)/6(8.2)	0.90	0.34-2.40	NS	14(9.7)/9(8.2)	0.83	0.34-2.02	NS
Cerad >1.5%	6(3.1)/1(1.7)	0.50	0.06-4.38	NS	3(2.7)/4(2.7)	1.17	0.25-5.52	NS	6(4.1)/1(0.9)	0.19	0.02-1.66	NS	4(4.3)/3(1.8)	0.42	0.09-2.01	NS	5(2.7)/2(2.7)	1.18	0.22-6.53	NS	2(1.4)/5(4.5)	3.06	0.56-16.6	NS
NFT	39(23.1)/5(10.4)	0.39	0.15-1.06	NS	17(17.9)/27(21.6)	1.30	0.66-2.56	NS	28(22.6)/17(17.3)	0.70	0.36-1.39	NS	16(21.3)/28(19.6)	0.91	0.46-1.82	NS	31(19.9)/13(20.0)	1.04	0.50-2.15	NS	25(20.2)/19(20.7)	1.03	0.53-2.01	NS
	Haplotype 2 (CCCTAT)				Haplotype 3 (CCCCGC)				Haplotype 4 (CCGCAC)				Haplotype 5 (CTCTAC)				Haplotype 7 (GCGCAC)							
	Aff. (%) H2*/H2+	OR	CI	p	Aff. (%) H3*/H3+	OR	CI	p	Aff. (%) H4*/H4+	OR	CI	p	Aff. (%) H5*/H5+	OR	CI	p	Aff. (%) H7*/H7+	OR	CI	p				
SP	18(11.7)/14(12.4)	0.99	0.46-2.15	NS	24(12.4)/8(10.8)	0.94	0.39-2.25	NS	23(11.0)/9(15.8)	1.44	0.60-3.41	NS	14(12.5)/18(11.6)	1.03	0.47-2.22	NS	27(13.2)/5(7.9)	0.54	0.19-1.50	NS				
Non neuritic	12(7.2)/9(8.0)	1.01	0.40-2.51	NS	16(7.8)/5(6.8)	0.87	0.30-2.49	NS	12(5.4)/9(15.8)	2.98	1.17-7.59	0.022	10(8.9)/11(6.6)	0.83	0.33-2.06	NS	18(8.3)/3(4.8)	0.50	0.14-1.76	NS				
Neuritic	8(4.8)/4(3.5)	0.62	0.18-2.19	NS	10(4.9)/2(2.7)	0.57	0.12-2.77	NS	11(5.0)/1(1.8)	0.34	0.04-2.76	NS	3(2.7)/9(5.4)	2.52	0.64-9.84	NS	11(5.1)/1(1.6)	0.26	0.03-2.07	NS				
Diffuse	6(3.6)/4(3.5)	0.87	0.24-3.22	NS	8(3.9)/2(2.7)	0.70	0.14-3.43	NS	3(1.4)/7(12.3)	9.21	2.26-37.5	0.002	6(5.4)/4(2.4)	0.52	0.14-1.91	NS	8(3.7)/2(3.2)	0.73	0.15-3.60	NS				
Primitive	6(3.6)/5(4.4)	1.14	0.34-3.88	NS	8(3.9)/3(4.1)	1.03	0.26-4.02	NS	9(4.1)/2(3.5)	0.90	0.19-4.34	NS	4(3.6)/7(4.2)	1.28	0.36-4.55	NS	10(4.6)/1(1.6)	0.30	0.04-2.42	NS				
Classic	8(4.8)/4(3.5)	0.62	0.18-2.19	NS	10(4.9)/2(2.7)	0.57	0.12-2.76	NS	11(5.0)/1(1.8)	0.35	0.04-2.81	NS	3(2.7)/9(5.4)	2.50	0.64-9.78	NS	11(5.1)/1(1.6)	0.26	0.03-2.08	NS				
Burnt out	0(0.0)/0(0.0)	.	.	.	0(0.0)/0(0.0)	.	.	.	0(0.0)/0(0.0)	.	.	.	0(0.0)/0(0.0)	.	.	.	0(0.0)/0(0.0)	.	.	.				
Cerad ≤1.5%	16(10.4)/9(8.0)	0.74	0.31-1.76	NS	19(9.8)/6(8.1)	0.87	0.33-2.32	NS	16(7.6)/9(15.8)	2.05	0.84-5.03	NS	11(9.8)/14(9.0)	1.00	0.43-2.33	NS	21(10.3)/4(6.3)	0.56	0.18-1.72	NS				
Cerad >1.5%	2(1.3)/5(4.4)	3.12	0.58-16.9	NS	5(2.6)/2(2.7)	1.20	0.22-6.62	NS	7(3.3)/0(0.0)	.	.	.	3(2.7)/4(2.6)	1.14	0.24-5.41	NS	6(2.9)/1(1.6)	0.47	0.05-4.10	NS				
NFT	29(20.9)/20(21.1)	1.00	0.53-1.91	NS	35(20.8)/14(21.2)	1.05	0.52-2.11	NS	36(19.9)/13(24.5)	1.27	0.61-2.63	NS	18(18.8)/31(22.5)	1.30	0.67-2.50	NS	43(23.5)/6(11.8)	0.43	0.17-1.09	NS				

.denotes the values were unable to be computed. *denotes the most common haplotype/homozygous genotype acting as the reference group in the analyses. Aff.(%) refers to number of individuals affected with SP/NFT.

USF1 – upstream transcription factor 1 gene, SNPs – single nucleotide polymorphisms, SP – senile plaques, NFT – neurofibrillary tangles, APOE4 – Apolipoprotein E ε4, OR – odds ratio, CI – confidence interval, p – p-value, NS – non significant p-value