Supplementary Information

Genomic variants in the *FTO* gene are associated with sporadic amyotrophic lateral sclerosis in Greek patients

Konstantinos Mitropoulos[1,#], Eleni Merkouri Papadima[2,#,%], Georgia Xiromerisiou[3], Angeliki Balasopoulou[2], Kyriaki Charalampidou[2], Vasiliki Galani[2], Krystallia-Vassiliki Zafeiri[2], Efthymios Dardiotis[3], Styliani Ralli[3], Georgia Deretzi[4], Anne John[5], Kyriaki Kydonopoulou[6], Elpida Papadopoulou[6], Alba di Pardo[7], Fulya Akcimen[8], Annalisa Loizedda[9,10], Valerija Dobričić[11], Ivana Novaković[11,12], Vladimir S. Kostić[11], Clint Mizzi[2], Brock A. Peters[13,14], A. Nazli Basak[8], Sandro Orrù [9], Evangelos Kiskinis[7], David N. Cooper[15], Spyridon Gerou[6], Radoje Drmanac[13], Marina Bartsakoulia[2], Evangelia-Eirini Tsermpini[2], George M. Hadjigeorgiou[3], Bassam R. Ali[5], Theodora Katsila[2], George P. Patrinos[2,5,]*

[1] National and Kapodistrian University of Athens School of Medicine, Athens, Greece;

[2] Department of Pharmacy, University of Patras School of Health Sciences, Patras, Greece;

[3] School of Medicine, University of Thessaly, Larisa, Greece;

[4] Papageorgiou hospital, Thessaloniki, Greece;

[5] United Arab Emirates University, College of Medicine and Health Sciences, Department of Pathology, Al-Ain, UAE;

[6] ANALYSI Diagnostic Laboratories S.A., Thessaloniki, Greece;

[7] Departments of Neurology and Physiology, Northwestern University Feinberg School of Medicine, Chicago, IL, USA;

[8] Bogazici University, Suna and Inan Kirac Foundation, NDAL, Istanbul, Turkey;

[9] University of Cagliari, Department of Medical Sciences and Public Health, Cagliari, Italy;

[10] CNR IRGB, Cagliari, Italy;

[11] Institute of Neurology CCS, School of Medicine, University of Belgrade, Belgrade, Serbia;

[12] Faculty of Medicine, University of Belgrade, Institute of Human Genetics, Belgrade, Serbia;

[13] Complete Genomics Inc, Mountain View, CA, USA;

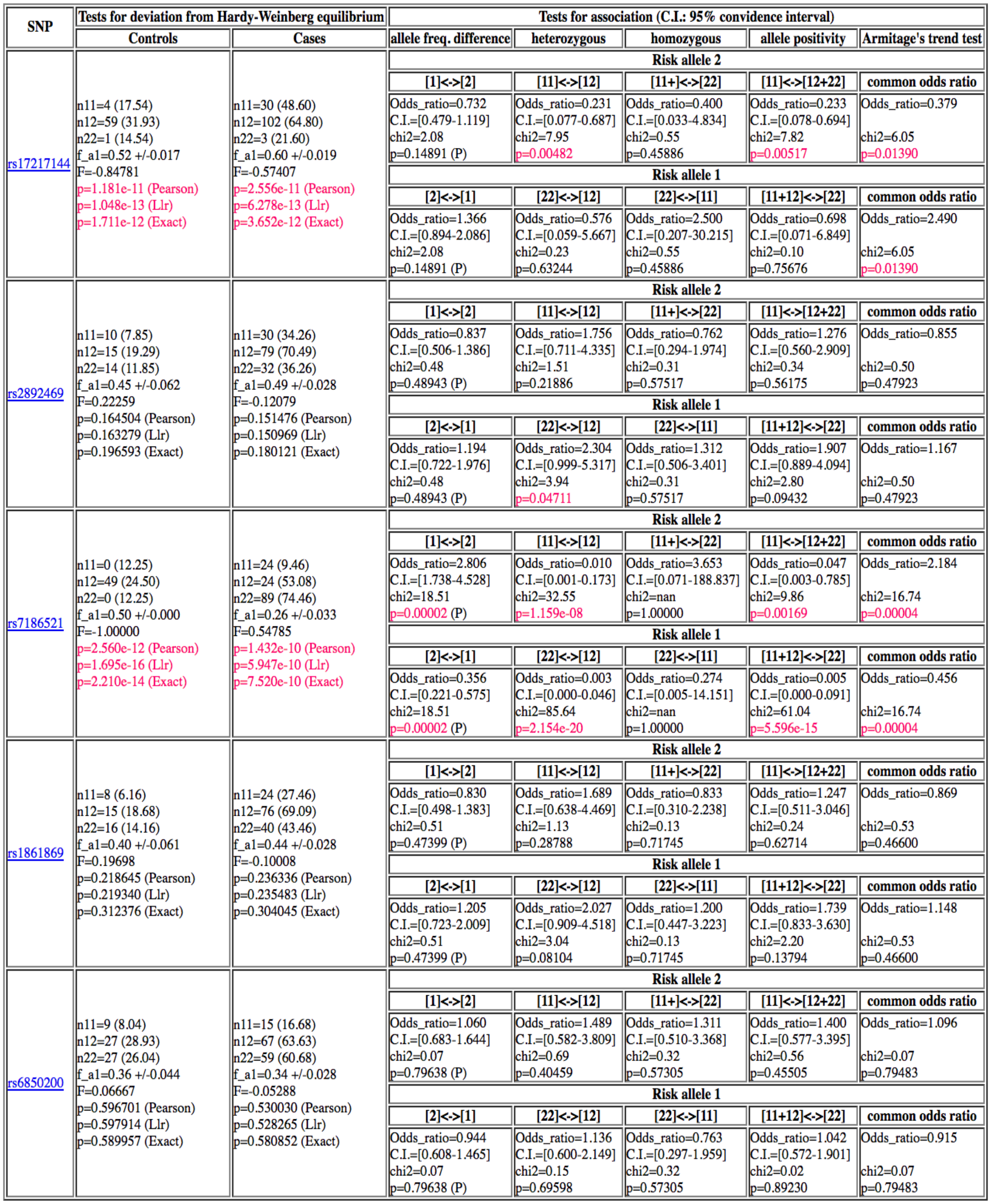
[14] BGI Shenzhen, Shenzhen, People's Republic of China;

[15] Cardiff University, Institute of Medical Genetics, Cardiff, UK

[#] These authors contributed equally to this work

[%] Present address: University of Cagliari, School of Medicine, Department of Biomedical Sciences, Cagliari, Italy

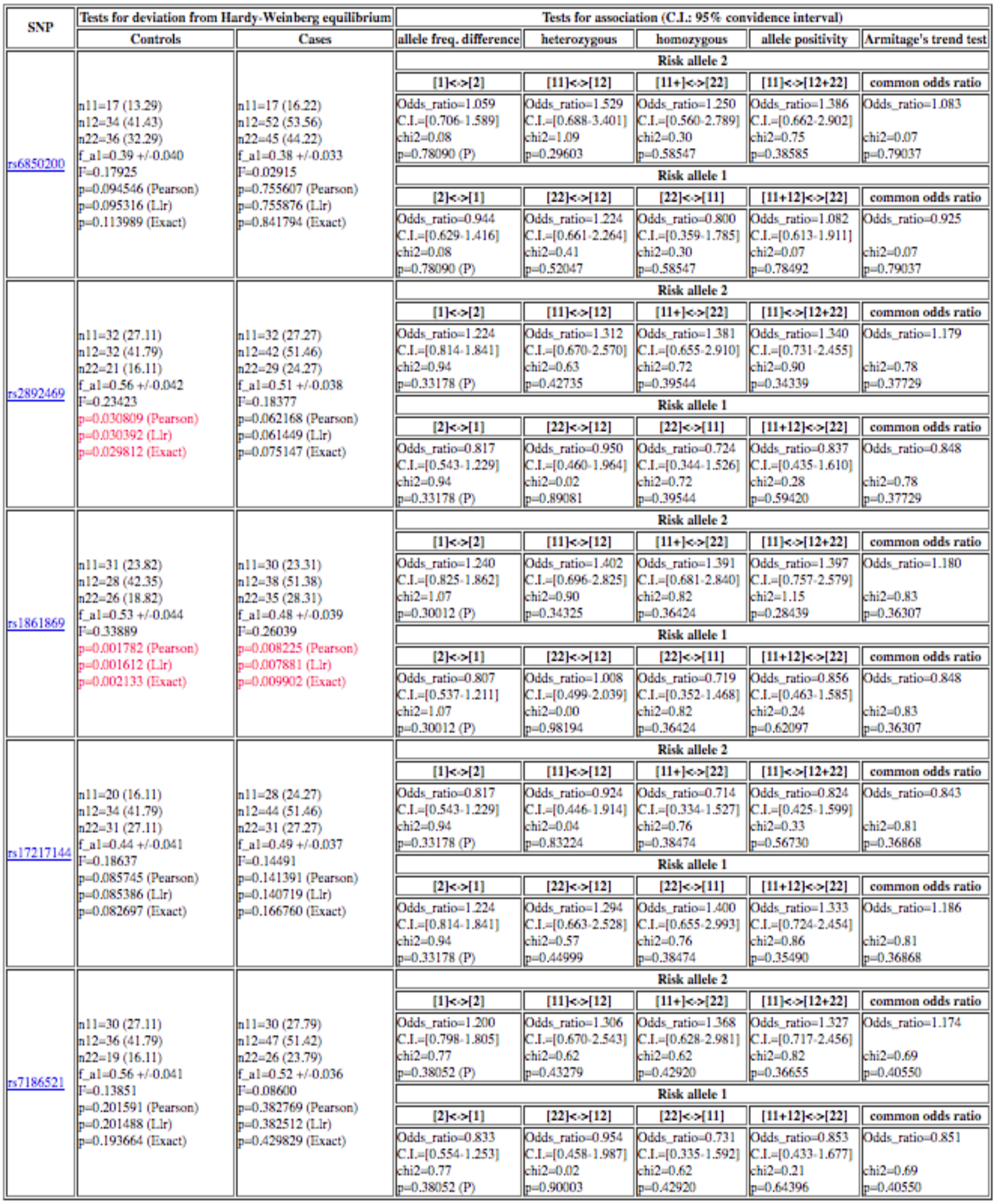
Supplementary Table 1. Statistical and Hardy-Weinberg analyses on the genotyping data of sALS patients and healthy individuals of Greek origin.



SNP	Tests for deviation from Hardy-Weinberg equilibrium		Tests for association (C.I.: 95% convidence interval)				
	Controls	Cases	allele freq. difference	heterozygous	homozygous	allele positivity	Armitage's trend test
rs17217144	n11=4 (17.54) n12=59 (31.93) n22=1 (14.54) f_a1=0.52 +/-0.017 F=-0.84781 p=1.181e-11 (Pearson) p=1.048e-13 (Llr) p=1.711e-12 (Exact)	n11=30 (48.60) n12=102 (64.80) n22=3 (21.60) f_a1=0.60 +/-0.019 F=-0.57407 p=2.556e-11 (Pearson) p=6.278e-13 (Llr) p=3.652e-12 (Exact)	**Risk allele 2**				
			[1]<->[2]	**[11]<->[12]**	**[11+]<->[22]**	**[11]<->[12+22]**	**common odds ratio**
			Odds_ratio=0.732 C.I.=[0.479-1.119] chi2=2.08 p=0.14891 (P)	Odds_ratio=0.231 C.I.=[0.077-0.687] chi2=7.95 p=0.00482	Odds_ratio=0.400 C.I.=[0.033-4.834] chi2=0.55 p=0.45886	Odds_ratio=0.233 C.I.=[0.078-0.694] chi2=7.82 p=0.00517	Odds_ratio=0.379 chi2=6.05 p=0.01390
			Risk allele 1				
			[2]<->[1]	**[22]<->[12]**	**[22]<->[11]**	**[11+12]<->[22]**	**common odds ratio**
			Odds_ratio=1.366 C.I.=[0.894-2.086] chi2=2.08 p=0.14891 (P)	Odds_ratio=0.576 C.I.=[0.059-5.667] chi2=0.23 p=0.63244	Odds_ratio=2.500 C.I.=[0.207-30.215] chi2=0.55 p=0.45886	Odds_ratio=0.698 C.I.=[0.071-6.849] chi2=0.10 p=0.75676	Odds_ratio=2.490 chi2=6.05 p=0.01390
rs2892469	n11=10 (7.85) n12=15 (19.29) n22=14 (11.85) f_a1=0.45 +/-0.062 F=0.22259 p=0.164504 (Pearson) p=0.163279 (Llr) p=0.196593 (Exact)	n11=30 (34.26) n12=79 (70.49) n22=32 (36.26) f_a1=0.49 +/-0.028 F=-0.12079 p=0.151476 (Pearson) p=0.150969 (Llr) p=0.180121 (Exact)	**Risk allele 2**				
			[1]<->[2]	**[11]<->[12]**	**[11+]<->[22]**	**[11]<->[12+22]**	**common odds ratio**
			Odds_ratio=0.837 C.I.=[0.506-1.386] chi2=0.48 p=0.48943 (P)	Odds_ratio=1.756 C.I.=[0.711-4.335] chi2=1.51 p=0.21886	Odds_ratio=0.762 C.I.=[0.294-1.974] chi2=0.31 p=0.57517	Odds_ratio=1.276 C.I.=[0.560-2.909] chi2=0.34 p=0.56175	Odds_ratio=0.855 chi2=0.50 p=0.47923
			Risk allele 1				
			[2]<->[1]	**[22]<->[12]**	**[22]<->[11]**	**[11+12]<->[22]**	**common odds ratio**
			Odds_ratio=1.194 C.I.=[0.722-1.976] chi2=0.48 p=0.48943 (P)	Odds_ratio=2.304 C.I.=[0.999-5.317] chi2=3.94 p=0.04711	Odds_ratio=1.312 C.I.=[0.506-3.401] chi2=0.31 p=0.57517	Odds_ratio=1.907 C.I.=[0.889-4.094] chi2=2.80 p=0.09432	Odds_ratio=1.167 chi2=0.50 p=0.47923
rs7186521	n11=0 (12.25) n12=49 (24.50) n22=0 (12.25) f_a1=0.50 +/-0.000 F=-1.00000 p=2.560e-12 (Pearson) p=1.695e-16 (Llr) p=2.210e-14 (Exact)	n11=24 (9.46) n12=24 (53.08) n22=89 (74.46) f_a1=0.26 +/-0.033 F=0.54785 p=1.432e-10 (Pearson) p=5.947e-10 (Llr) p=7.520e-10 (Exact)	**Risk allele 2**				
			[1]<->[2]	**[11]<->[12]**	**[11+]<->[22]**	**[11]<->[12+22]**	**common odds ratio**
			Odds_ratio=2.806 C.I.=[1.738-4.528] chi2=18.51 p=0.00002 (P)	Odds_ratio=0.010 C.I.=[0.001-0.173] chi2=32.55 p=1.159e-08	Odds_ratio=3.653 C.I.=[0.071-188.837] chi2=nan p=1.00000	Odds_ratio=0.047 C.I.=[0.003-0.785] chi2=9.86 p=0.00169	Odds_ratio=2.184 chi2=16.74 p=0.00004
			Risk allele 1				
			[2]<->[1]	**[22]<->[12]**	**[22]<->[11]**	**[11+12]<->[22]**	**common odds ratio**
			Odds_ratio=0.356 C.I.=[0.221-0.575] chi2=18.51 p=0.00002 (P)	Odds_ratio=0.003 C.I.=[0.000-0.046] chi2=85.64 p=2.154e-20	Odds_ratio=0.274 C.I.=[0.005-14.151] chi2=nan p=1.00000	Odds_ratio=0.005 C.I.=[0.000-0.091] chi2=61.04 p=5.596e-15	Odds_ratio=0.456 chi2=16.74 p=0.00004
rs1861869	n11=8 (6.16) n12=15 (18.68) n22=16 (14.16) f_a1=0.40 +/-0.061 F=0.19698 p=0.218645 (Pearson) p=0.219340 (Llr) p=0.312376 (Exact)	n11=24 (27.46) n12=76 (69.09) n22=40 (43.46) f_a1=0.44 +/-0.028 F=-0.10008 p=0.236336 (Pearson) p=0.235483 (Llr) p=0.304045 (Exact)	**Risk allele 2**				
			[1]<->[2]	**[11]<->[12]**	**[11+]<->[22]**	**[11]<->[12+22]**	**common odds ratio**
			Odds_ratio=0.830 C.I.=[0.498-1.383] chi2=0.51 p=0.47399 (P)	Odds_ratio=1.689 C.I.=[0.638-4.469] chi2=1.13 p=0.28788	Odds_ratio=0.833 C.I.=[0.310-2.238] chi2=0.13 p=0.71745	Odds_ratio=1.247 C.I.=[0.511-3.046] chi2=0.24 p=0.62714	Odds_ratio=0.869 chi2=0.53 p=0.46600
			Risk allele 1				
			[2]<->[1]	**[22]<->[12]**	**[22]<->[11]**	**[11+12]<->[22]**	**common odds ratio**
			Odds_ratio=1.205 C.I.=[0.723-2.009] chi2=0.51 p=0.47399 (P)	Odds_ratio=2.027 C.I.=[0.909-4.518] chi2=3.04 p=0.08104	Odds_ratio=1.200 C.I.=[0.447-3.223] chi2=0.13 p=0.71745	Odds_ratio=1.739 C.I.=[0.833-3.630] chi2=2.20 p=0.13794	Odds_ratio=1.148 chi2=0.53 p=0.46600
rs6850200	n11=9 (8.04) n12=27 (28.93) n22=27 (26.04) f_a1=0.36 +/-0.044 F=0.06667 p=0.596701 (Pearson) p=0.597914 (Llr) p=0.589957 (Exact)	n11=15 (16.68) n12=67 (63.63) n22=59 (60.68) f_a1=0.34 +/-0.028 F=-0.05288 p=0.530030 (Pearson) p=0.528265 (Llr) p=0.580852 (Exact)	**Risk allele 2**				
			[1]<->[2]	**[11]<->[12]**	**[11+]<->[22]**	**[11]<->[12+22]**	**common odds ratio**
			Odds_ratio=1.060 C.I.=[0.683-1.644] chi2=0.07 p=0.79638 (P)	Odds_ratio=1.489 C.I.=[0.582-3.809] chi2=0.69 p=0.40459	Odds_ratio=1.311 C.I.=[0.510-3.368] chi2=0.32 p=0.57305	Odds_ratio=1.400 C.I.=[0.577-3.395] chi2=0.56 p=0.45505	Odds_ratio=1.096 chi2=0.07 p=0.79483
			Risk allele 1				
			[2]<->[1]	**[22]<->[12]**	**[22]<->[11]**	**[11+12]<->[22]**	**common odds ratio**
			Odds_ratio=0.944 C.I.=[0.608-1.465] chi2=0.07 p=0.79638 (P)	Odds_ratio=1.136 C.I.=[0.600-2.149] chi2=0.15 p=0.69598	Odds_ratio=0.763 C.I.=[0.297-1.959] chi2=0.32 p=0.57305	Odds_ratio=1.042 C.I.=[0.572-1.901] chi2=0.02 p=0.89230	Odds_ratio=0.915 chi2=0.07 p=0.79483

(Pearson): Pearson's goodness-of-fit chi-square (degrees of freedom=1); (Llr): Log likelihood ratio chi-square (degrees of freedom=1); (Exact): Fisher's Exact test PA, PB: two-tailed p-values; if P, then PA=PB

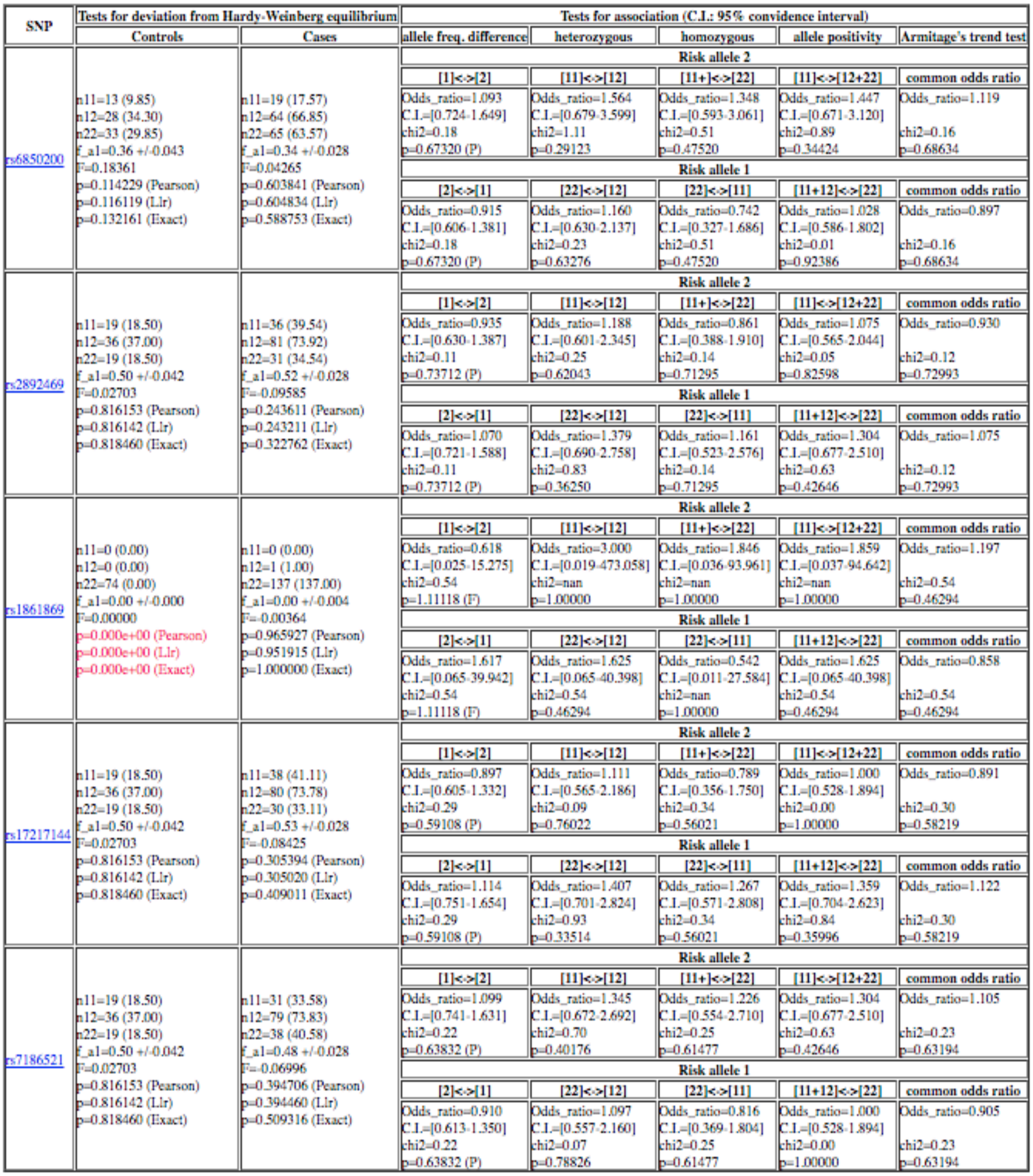
Supplementary Table 2. Statistical and Hardy-Weinberg analyses on the genotyping data of sALS patients and healthy individuals of Sardinian origin.



SNP	Tests for deviation from Hardy-Weinberg equilibrium		Tests for association (C.I.: 95% convidence interval)				
	Controls	Cases	allele freq. difference	heterozygous	homozygous	allele positivity	Armitage's trend test
rs6850200	n11=17 (13.29) n12=34 (41.43) n22=36 (32.29) f_a1=0.39 +/-0.040 F=0.17925 p=0.094546 (Pearson) p=0.095316 (Llr) p=0.113989 (Exact)	n11=17 (16.22) n12=52 (53.56) n22=45 (44.22) f_a1=0.38 +/-0.033 F=0.02915 p=0.755607 (Pearson) p=0.755876 (Llr) p=0.841794 (Exact)	Risk allele 2				
			[1]<->[2]	[11]<->[12]	[11+]<->[22]	[11]<->[12+22]	common odds ratio
			Odds_ratio=1.059 C.I.=[0.706-1.589] chi2=0.08 p=0.78090 (P)	Odds_ratio=1.529 C.I.=[0.688-3.401] chi2=1.09 p=0.29603	Odds_ratio=1.250 C.I.=[0.560-2.789] chi2=0.30 p=0.58547	Odds_ratio=1.386 C.I.=[0.662-2.902] chi2=0.75 p=0.38585	Odds_ratio=1.083 chi2=0.07 p=0.79037
			Risk allele 1				
			[2]<->[1]	[22]<->[12]	[22]<->[11]	[11+12]<->[22]	common odds ratio
			Odds_ratio=0.944 C.I.=[0.629-1.416] chi2=0.08 p=0.78090 (P)	Odds_ratio=1.224 C.I.=[0.661-2.264] chi2=0.41 p=0.52047	Odds_ratio=0.800 C.I.=[0.359-1.785] chi2=0.30 p=0.58547	Odds_ratio=1.082 C.I.=[0.613-1.911] chi2=0.07 p=0.78492	Odds_ratio=0.925 chi2=0.07 p=0.79037
rs2892469	n11=32 (27.11) n12=32 (41.79) n22=21 (16.11) f_a1=0.56 +/-0.042 F=0.23423 p=0.030809 (Pearson) p=0.030392 (Llr) p=0.029812 (Exact)	n11=32 (27.27) n12=42 (51.46) n22=29 (24.27) f_a1=0.51 +/-0.038 F=0.18377 p=0.062168 (Pearson) p=0.061449 (Llr) p=0.075147 (Exact)	Risk allele 2				
			[1]<->[2]	[11]<->[12]	[11+]<->[22]	[11]<->[12+22]	common odds ratio
			Odds_ratio=1.224 C.I.=[0.814-1.841] chi2=0.94 p=0.33178 (P)	Odds_ratio=1.312 C.I.=[0.670-2.570] chi2=0.63 p=0.42735	Odds_ratio=1.381 C.I.=[0.655-2.910] chi2=0.72 p=0.39544	Odds_ratio=1.340 C.I.=[0.731-2.455] chi2=0.90 p=0.34339	Odds_ratio=1.179 chi2=0.78 p=0.37729
			Risk allele 1				
			[2]<->[1]	[22]<->[12]	[22]<->[11]	[11+12]<->[22]	common odds ratio
			Odds_ratio=0.817 C.I.=[0.543-1.229] chi2=0.94 p=0.33178 (P)	Odds_ratio=0.950 C.I.=[0.460-1.964] chi2=0.02 p=0.89081	Odds_ratio=0.724 C.I.=[0.344-1.526] chi2=0.72 p=0.39544	Odds_ratio=0.837 C.I.=[0.435-1.610] chi2=0.28 p=0.59420	Odds_ratio=0.848 chi2=0.78 p=0.37729
rs1861869	n11=31 (23.82) n12=28 (42.35) n22=26 (18.82) f_a1=0.53 +/-0.044 F=0.33889 p=0.001782 (Pearson) p=0.001612 (Llr) p=0.002133 (Exact)	n11=30 (23.31) n12=38 (51.38) n22=35 (28.31) f_a1=0.48 +/-0.039 F=0.26039 p=0.008225 (Pearson) p=0.007881 (Llr) p=0.009902 (Exact)	Risk allele 2				
			[1]<->[2]	[11]<->[12]	[11+]<->[22]	[11]<->[12+22]	common odds ratio
			Odds_ratio=1.240 C.I.=[0.825-1.862] chi2=1.07 p=0.30012 (P)	Odds_ratio=1.402 C.I.=[0.696-2.825] chi2=0.90 p=0.34325	Odds_ratio=1.391 C.I.=[0.681-2.840] chi2=0.82 p=0.36424	Odds_ratio=1.397 C.I.=[0.757-2.579] chi2=1.15 p=0.28439	Odds_ratio=1.180 chi2=0.83 p=0.36307
			Risk allele 1				
			[2]<->[1]	[22]<->[12]	[22]<->[11]	[11+12]<->[22]	common odds ratio
			Odds_ratio=0.807 C.I.=[0.537-1.211] chi2=1.07 p=0.30012 (P)	Odds_ratio=1.008 C.I.=[0.499-2.039] chi2=0.00 p=0.98194	Odds_ratio=0.719 C.I.=[0.352-1.468] chi2=0.82 p=0.36424	Odds_ratio=0.856 C.I.=[0.463-1.585] chi2=0.24 p=0.62097	Odds_ratio=0.848 chi2=0.83 p=0.36307
rs17217144	n11=20 (16.11) n12=34 (41.79) n22=31 (27.11) f_a1=0.44 +/-0.041 F=0.18637 p=0.085745 (Pearson) p=0.085386 (Llr) p=0.082697 (Exact)	n11=28 (24.27) n12=44 (51.46) n22=31 (27.27) f_a1=0.49 +/-0.037 F=0.14491 p=0.141391 (Pearson) p=0.140719 (Llr) p=0.166760 (Exact)	Risk allele 2				
			[1]<->[2]	[11]<->[12]	[11+]<->[22]	[11]<->[12+22]	common odds ratio
			Odds_ratio=0.817 C.I.=[0.543-1.229] chi2=0.94 p=0.33178 (P)	Odds_ratio=0.924 C.I.=[0.446-1.914] chi2=0.04 p=0.83224	Odds_ratio=0.714 C.I.=[0.334-1.527] chi2=0.76 p=0.38474	Odds_ratio=0.824 C.I.=[0.425-1.599] chi2=0.33 p=0.56730	Odds_ratio=0.843 chi2=0.81 p=0.36868
			Risk allele 1				
			[2]<->[1]	[22]<->[12]	[22]<->[11]	[11+12]<->[22]	common odds ratio
			Odds_ratio=1.224 C.I.=[0.814-1.841] chi2=0.94 p=0.33178 (P)	Odds_ratio=1.294 C.I.=[0.663-2.528] chi2=0.57 p=0.44999	Odds_ratio=1.400 C.I.=[0.655-2.993] chi2=0.76 p=0.38474	Odds_ratio=1.333 C.I.=[0.724-2.454] chi2=0.86 p=0.35490	Odds_ratio=1.186 chi2=0.81 p=0.36868
rs7186521	n11=30 (27.11) n12=36 (41.79) n22=19 (16.11) f_a1=0.56 +/-0.041 F=0.13851 p=0.201591 (Pearson) p=0.201488 (Llr) p=0.193664 (Exact)	n11=30 (27.79) n12=47 (51.42) n22=26 (23.79) f_a1=0.52 +/-0.036 F=0.08600 p=0.382769 (Pearson) p=0.382512 (Llr) p=0.429829 (Exact)	Risk allele 2				
			[1]<->[2]	[11]<->[12]	[11+]<->[22]	[11]<->[12+22]	common odds ratio
			Odds_ratio=1.200 C.I.=[0.798-1.805] chi2=0.77 p=0.38052 (P)	Odds_ratio=1.306 C.I.=[0.670-2.543] chi2=0.62 p=0.43279	Odds_ratio=1.368 C.I.=[0.628-2.981] chi2=0.62 p=0.42920	Odds_ratio=1.327 C.I.=[0.717-2.456] chi2=0.82 p=0.36655	Odds_ratio=1.174 chi2=0.69 p=0.40550
			Risk allele 1				
			[2]<->[1]	[22]<->[12]	[22]<->[11]	[11+12]<->[22]	common odds ratio
			Odds_ratio=0.833 C.I.=[0.554-1.253] chi2=0.77 p=0.38052 (P)	Odds_ratio=0.954 C.I.=[0.458-1.987] chi2=0.02 p=0.90003	Odds_ratio=0.731 C.I.=[0.335-1.592] chi2=0.62 p=0.42920	Odds_ratio=0.853 C.I.=[0.433-1.677] chi2=0.21 p=0.64396	Odds_ratio=0.851 chi2=0.69 p=0.40550

(Pearson): Pearson's goodness-of-fit chi-square (degrees of freedom=1); (Llr): Log likelihood ratio chi-square (degrees of freedom=1); (Exact): Fisher's Exact test PA, PB: two-tailed p-values; if P, then PA=PB

Supplementary Table 3. Statistical and Hardy-Weinberg analyses on the genotyping data of sALS patients and healthy individuals of Turkish origin.



SNP	Tests for deviation from Hardy-Weinberg equilibrium		Tests for association (C.I.: 95% convidence interval)				
	Controls	Cases	allele freq. difference	heterozygous	homozygous	allele positivity	Armitage's trend test
rs6850200	n11=13 (9.85) n12=28 (34.30) n22=33 (29.85) f_a1=0.36 +/-0.043 F=0.18361 p=0.114229 (Pearson) p=0.116119 (Llr) p=0.132161 (Exact)	n11=19 (17.57) n12=64 (66.85) n22=65 (63.57) f_a1=0.34 +/-0.028 F=0.04265 p=0.603841 (Pearson) p=0.604834 (Llr) p=0.588753 (Exact)	**Risk allele 2**				
			[1]<->[2]	**[11]<->[12]**	**[11+]<->[22]**	**[11]<->[12+22]**	**common odds ratio**
			Odds_ratio=1.093 C.I.=[0.724-1.649] chi2=0.18 p=0.67320 (P)	Odds_ratio=1.564 C.I.=[0.679-3.599] chi2=1.11 p=0.29123	Odds_ratio=1.348 C.I.=[0.593-3.061] chi2=0.51 p=0.47520	Odds_ratio=1.447 C.I.=[0.671-3.120] chi2=0.89 p=0.34424	Odds_ratio=1.119 chi2=0.16 p=0.68634
			Risk allele 1				
			[2]<->[1]	**[22]<->[12]**	**[22]<->[11]**	**[11+12]<->[22]**	**common odds ratio**
			Odds_ratio=0.915 C.I.=[0.606-1.381] chi2=0.18 p=0.67320 (P)	Odds_ratio=1.160 C.I.=[0.630-2.137] chi2=0.23 p=0.63276	Odds_ratio=0.742 C.I.=[0.327-1.686] chi2=0.51 p=0.47520	Odds_ratio=1.028 C.I.=[0.586-1.802] chi2=0.01 p=0.92386	Odds_ratio=0.897 chi2=0.16 p=0.68634
rs2892469	n11=19 (18.50) n12=36 (37.00) n22=19 (18.50) f_a1=0.50 +/-0.042 F=0.02703 p=0.816153 (Pearson) p=0.816142 (Llr) p=0.818460 (Exact)	n11=36 (39.54) n12=81 (73.92) n22=31 (34.54) f_a1=0.52 +/-0.028 F=-0.09585 p=0.243611 (Pearson) p=0.243211 (Llr) p=0.322762 (Exact)	**Risk allele 2**				
			[1]<->[2]	**[11]<->[12]**	**[11+]<->[22]**	**[11]<->[12+22]**	**common odds ratio**
			Odds_ratio=0.935 C.I.=[0.630-1.387] chi2=0.11 p=0.73712 (P)	Odds_ratio=1.188 C.I.=[0.601-2.345] chi2=0.25 p=0.62043	Odds_ratio=0.861 C.I.=[0.388-1.910] chi2=0.14 p=0.71295	Odds_ratio=1.075 C.I.=[0.565-2.044] chi2=0.05 p=0.82598	Odds_ratio=0.930 chi2=0.12 p=0.72993
			Risk allele 1				
			[2]<->[1]	**[22]<->[12]**	**[22]<->[11]**	**[11+12]<->[22]**	**common odds ratio**
			Odds_ratio=1.070 C.I.=[0.721-1.588] chi2=0.11 p=0.73712 (P)	Odds_ratio=1.379 C.I.=[0.690-2.758] chi2=0.83 p=0.36250	Odds_ratio=1.161 C.I.=[0.523-2.576] chi2=0.14 p=0.71295	Odds_ratio=1.304 C.I.=[0.677-2.510] chi2=0.63 p=0.42646	Odds_ratio=1.075 chi2=0.12 p=0.72993
rs1861869	n11=0 (0.00) n12=0 (0.00) n22=74 (0.00) f_a1=0.00 +/-0.000 F=0.00000 p=0.000e+00 (Pearson) p=0.000e+00 (Llr) p=0.000e+00 (Exact)	n11=0 (0.00) n12=1 (1.00) n22=137 (137.00) f_a1=0.00 +/-0.004 F=-0.00364 p=0.965927 (Pearson) p=0.951915 (Llr) p=1.000000 (Exact)	**Risk allele 2**				
			[1]<->[2]	**[11]<->[12]**	**[11+]<->[22]**	**[11]<->[12+22]**	**common odds ratio**
			Odds_ratio=0.618 C.I.=[0.025-15.275] chi2=0.54 p=1.11118 (F)	Odds_ratio=3.000 C.I.=[0.019-473.058] chi2=nan p=1.00000	Odds_ratio=1.846 C.I.=[0.036-93.961] chi2=nan p=1.00000	Odds_ratio=1.859 C.I.=[0.037-94.642] chi2=nan p=1.00000	Odds_ratio=1.197 chi2=0.54 p=0.46294
			Risk allele 1				
			[2]<->[1]	**[22]<->[12]**	**[22]<->[11]**	**[11+12]<->[22]**	**common odds ratio**
			Odds_ratio=1.617 C.I.=[0.065-39.942] chi2=0.54 p=1.11118 (F)	Odds_ratio=1.625 C.I.=[0.065-40.398] chi2=0.54 p=0.46294	Odds_ratio=0.542 C.I.=[0.011-27.584] chi2=nan p=1.00000	Odds_ratio=1.625 C.I.=[0.065-40.398] chi2=0.54 p=0.46294	Odds_ratio=0.858 chi2=0.54 p=0.46294
rs17217144	n11=19 (18.50) n12=36 (37.00) n22=19 (18.50) f_a1=0.50 +/-0.042 F=0.02703 p=0.816153 (Pearson) p=0.816142 (Llr) p=0.818460 (Exact)	n11=38 (41.11) n12=80 (73.78) n22=30 (33.11) f_a1=0.53 +/-0.028 F=-0.08425 p=0.305394 (Pearson) p=0.305020 (Llr) p=0.409011 (Exact)	**Risk allele 2**				
			[1]<->[2]	**[11]<->[12]**	**[11+]<->[22]**	**[11]<->[12+22]**	**common odds ratio**
			Odds_ratio=0.897 C.I.=[0.605-1.332] chi2=0.29 p=0.59108 (P)	Odds_ratio=1.111 C.I.=[0.565-2.186] chi2=0.09 p=0.76022	Odds_ratio=0.789 C.I.=[0.356-1.750] chi2=0.34 p=0.56021	Odds_ratio=1.000 C.I.=[0.528-1.894] chi2=0.00 p=1.00000	Odds_ratio=0.891 chi2=0.30 p=0.58219
			Risk allele 1				
			[2]<->[1]	**[22]<->[12]**	**[22]<->[11]**	**[11+12]<->[22]**	**common odds ratio**
			Odds_ratio=1.114 C.I.=[0.751-1.654] chi2=0.29 p=0.59108 (P)	Odds_ratio=1.407 C.I.=[0.701-2.824] chi2=0.93 p=0.33514	Odds_ratio=1.267 C.I.=[0.571-2.808] chi2=0.34 p=0.56021	Odds_ratio=1.359 C.I.=[0.704-2.623] chi2=0.84 p=0.35996	Odds_ratio=1.122 chi2=0.30 p=0.58219
rs7186521	n11=19 (18.50) n12=36 (37.00) n22=19 (18.50) f_a1=0.50 +/-0.042 F=0.02703 p=0.816153 (Pearson) p=0.816142 (Llr) p=0.818460 (Exact)	n11=31 (33.58) n12=79 (73.83) n22=38 (40.58) f_a1=0.48 +/-0.028 F=-0.06996 p=0.394706 (Pearson) p=0.394460 (Llr) p=0.509316 (Exact)	**Risk allele 2**				
			[1]<->[2]	**[11]<->[12]**	**[11+]<->[22]**	**[11]<->[12+22]**	**common odds ratio**
			Odds_ratio=1.099 C.I.=[0.741-1.631] chi2=0.22 p=0.63832 (P)	Odds_ratio=1.345 C.I.=[0.672-2.692] chi2=0.70 p=0.40176	Odds_ratio=1.226 C.I.=[0.554-2.710] chi2=0.25 p=0.61477	Odds_ratio=1.304 C.I.=[0.677-2.510] chi2=0.63 p=0.42646	Odds_ratio=1.105 chi2=0.23 p=0.63194
			Risk allele 1				
			[2]<->[1]	**[22]<->[12]**	**[22]<->[11]**	**[11+12]<->[22]**	**common odds ratio**
			Odds_ratio=0.910 C.I.=[0.613-1.350] chi2=0.22 p=0.63832 (P)	Odds_ratio=1.097 C.I.=[0.557-2.160] chi2=0.07 p=0.78826	Odds_ratio=0.816 C.I.=[0.369-1.804] chi2=0.25 p=0.61477	Odds_ratio=1.000 C.I.=[0.528-1.894] chi2=0.00 p=1.00000	Odds_ratio=0.905 chi2=0.23 p=0.63194

(Pearson): Pearson's goodness-of-fit chi-square (degrees of freedom=1); (Llr): Log likelihood ratio chi-square (degrees of freedom=1); (Exact): Fisher's Exact test PA, PB: two-tailed p-values; if P, then PA=PB

Supplementary Table 4. *C9orf72* genotyping of Greek ALS patients (second patient cohort)

Sample ID	***C9orf72* repeats**
1	6/9
2	2/5
3	2/7
4	5/10
5	2/2
6	7/10
7	2/8
8	2/5
9	2/2
10	2/3
11	5/6
12	8/10
13	Inconclusive
14	2/2
15	2/8
16	2/5
17	2/2
18	2/2
19	2/2
20	2/5
21	2/8
22	2/5
23	2/2
24	2/4
25	Inconclusive
26	**2/exp**
27	2/7
28	2/2
29	6/11
30	6/11
31	No amplification

Numbers given in the table (column *C9orf72* results) represent the numbers of *C9orf72* hexanucleotide repeats for a given patient. For example, 2/6 means that the patient has 2 repeats on one allele, and 6 repeats on the other. 2/2 means that the patient in question is homozygous for allele 2. For patient 26, 2/exp means that there are 2 repeats on one allele and an expansion on the other. The size of the expansion cannot be defined because we use repeat-primed PCR.