ELECTRONIC SUPPLEMENTARY MATERIAL

A missense variant in *CST3* exerts a recessive effect on susceptibility to age-related macular degeneration resembling its association with Alzheimer's disease

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а ма		GG		Odds Ratio				Odds Ratio				
Study or Subgroup	Events	Total	Events	Total	Weight	M-H, Fixed, 95% CI Y	íear		M-H, F	ixed, 95% C	1	
Finckh et al. (USA) (2000)	19	22	257	418	8.3%	3.97 [1.16, 13.62] 2	2000				•	_
Crawford et al. (2000)	16	23	183	263	21.3%	1.00 [0.40, 2.52] 2	2000			+		
Finckh et al. (2000)	10	13	65	164	5.2%	5.08 [1.35, 19.15] 2	2000				-	
Roks et al. (2001)	6	12	60	133	11.8%	1.22 [0.37, 3.97] 2	2001			+•	-	
Monastero et al. (2005)	8	15	120	245	15.3%	1.19 [0.42, 3.38] 2	2005					
Beyer et al. (2005)	6	11	109	215	11.5%	1.17 [0.35, 3.94] 2	2005				-	
Cathcart et al. (2005)	17	23	106	204	13.3%	2.62 [0.99, 6.91] 2	2005					
Nacmias et al. (2006)	6	11	173	299	13.3%	0.87 [0.26, 2.93] 2	2006			•		
Total (95% CI)		130		1941	100.0%	1.73 [1.18, 2.54]				•		
Total events	88		1073									
Heterogeneity: Chi ² = 8.78, df = 7 (P = 0.27); l ² = 20%									1	!	1	
Test for overall effect: Z = 2.82 (P = 0.005) 0.05 0.2 1 5										20		

h												
0	AG		GG			Odds Ratio		Odds Ratio				
Study or Subgroup	Events	Total	Events	Total	Weight	M-H, Fixed, 95% CI Yea	r	M-H, F	ixed, 95% CI			
Finckh et al. (2000)	131	207	257	418	21.2%	1.08 [0.77, 1.52] 2000	D					
Finckh et al. (USA) (2000)	35	83	65	164	8.6%	1.11 [0.65, 1.90] 2000	D		- - -			
Crawford et al. (2000)	110	157	183	263	13.9%	1.02 [0.66, 1.57] 2000	D					
Roks et al. (2001)	30	69	60	133	7.9%	0.94 [0.52, 1.68] 2001	1	_	-			
Monastero et al. (2005)	63	123	120	245	13.3%	1.09 [0.71, 1.69] 2005	5					
Cathcart et al. (2005)	56	93	106	204	9.0%	1.40 [0.85, 2.30] 2008	5		+			
Beyer et al. (2005)	48	92	109	215	10.6%	1.06 [0.65, 1.73] 2005	5		- -			
Nacmias et al. (2006)	64	119	173	299	15.5%	0.85 [0.55, 1.30] 2006	6	-	•			
Total (95% CI)		943		1941	100.0%	1.06 [0.90, 1.24]			•			
Total events	537		1073									
Heterogeneity: Chi ² = 2.51, d	- 05		<u> </u>	+								
Test for overall effect: Z = 0.67 (P = 0.50) 0.05 0.2 1 5 20										20		

Fig. S1 Forest plots for the meta-analysis of *CST3* rs1064039 with respect to Alzheimer's disease in the Caucasian population using a fixed effects model. Area of the squares represents the weight of the study and horizontal bars represent 95% confidence interval of the OR. Applied to (**a**) "AA" genotype versus genotypes [data taken directly from Hua et al. (2012)] and (**b**) "AG" genotype versus "GG" genotype [data inferred from Hua et al. (2012)]



Fig. S2 Representative chromatograms of the three genotypes at rs1064039. Colour of peaks represents the different nucleotides (green=A, black=G, blue=C, red=T). The PCR products were generated using primers CST3RIIF and CST3LIIR and sequenced using CST3BIIR