

Appendix 5. Risk of AMD (subgroup) cases for *SLC2A1* SNP rs841853 in five replication populations

	No AMD	Early AMD	GA	CNV	CNV+ Mixed cases	Mixed AMD
The Rotterdam study						
	N=2217	N=894	N=72	N=64	N=103	N=39
<i>rs841853</i>	No. (%)	No. (%)	OR (95% CI)	No. (%)	OR (95% CI)	No. (%)
Genotype						
Noncarrier (AA)	1027 (46.3)	417 (46.6)	1	35 (48.6)	17 (26.6)	24 (61.5)
Heterozygous (Aa)	961 (43.3)	377 (42.2)	0.98 (0.83-1.15)	31 (43.1)	2.55 (1.43-4.58)	13 (33.3)
Homozygous (aa)	229 (10.3)	100 (11.2)	1.11 (0.85-1.44)	6 (8.3)	2.59 (1.13-5.94)	2 (5.1)
m.a.f(%)	32	32.3		29.9	43.8	21.8
The Franconian AMD study						
	N=591	N=83	N=159	N=415	N=538	N=123
<i>rs841853</i>	No(%)	No(%)	OR (95% CI)	No(%)	OR (95% CI)	No(%)
Genotype						
Noncarrier (AA)	265 (44.8)	41 (49.4)	1	75 (47.2)	199 (48.0)	59 (48.0)
Heterozygous (Aa)	260 (44.0)	36 (43.4)	0.89 (0.55-1.45)	70 (44.0)	0.88 (0.68-1.15)	48 (39.0)
Homozygous (aa)	66 (11.2)	6 (7.2)	0.59 (0.24-1.44)	14 (8.8)	0.89 (0.58-1.36)	16 (13.0)
m.a.f(%)	33.2	28.9		30.8	31.6	32.5
AREDS						
	N=215	N=253	N=167	N=325	N=504	N= 179
<i>rs841853</i>	No(%)	No(%)	OR (95% CI)	No(%)	OR (95% CI)	No(%)
Genotype						
Noncarrier (AA)	111 (51.6)	117 (46.2)	1	80 (47.9)	139 (42.8)	93 (52.0)
Heterozygous (Aa)	90 (41.9)	109 (43.1)	1.15 (0.78-1.68)	62 (37.1)	0.96 (0.62-1.47)	68 (38.0)
Homozygous (aa)	14 (6.5)	27 (10.7)	1.83 (0.91-3.67)	25 (15.0)	2.48 (1.21-5.06)	18 (10.0)
m.a.f(%)	27.4	32.2		35.1	32.9	
Columbia University						
	N=355	N=362	N=206		N=515	
<i>rs841853</i>	No(%)	No(%)	OR (95% CI)	No(%)	OR (95% CI)	No(%)
Genotype						
Noncarrier (AA)	158 (44.5)	162 (44.8)	1	107 (51.9)	N.P.	N.P.
Heterozygous (Aa)	170 (47.9)	159 (43.9)	0.91 (0.67-1.24)	76 (36.9)	0.66 (0.46-0.95)	0.80 (0.61-1.07)
Homozygous (aa)	27 (7.6)	41 (11.3)	1.48 (0.87-2.52)	23 (11.2)	1.26 (0.68-2.31)	1.15 (0.69-1.91)
m.a.f(%)	31.5	33.3		29.6	30.5	
The university of Iowa						
	N=130	N=101	N=28	N=175	N=187	N=12
<i>rs841853</i>	No(%)	No(%)	OR (95% CI)	No(%)	OR (95% CI)	No(%)
Genotype						
Noncarrier (AA)	67 (51.1)	53 (52.5)	1	13 (46.4)	82 (46.9)	5 (41.7)
Heterozygous (Aa)	49 (37.4)	36 (35.6)	0.93 (0.53-1.63)	14 (50.0)	1.47 (0.64-3.41)	6 (50.0)
Homozygous (aa)	14 (10.7)	12 (11.9)	1.08 (0.46-2.54)	1 (3.6)	0.37 (0.04-3.05)	1 (8.3)
m.a.f(%)	29.6	29.7		28.6	30.2	33.3

AMD = age- related macular degeneration; CI = confidence interval; OR = odds ratio; SNP = single nucleotide polymorphism; m.a.f = minor allele frequency; GA= geographic atrophy; CNV = choroidal neovascularization; Mixed AMD = combination of GA + CNV; N.P = not performed. "A" indicates common allele, "a" minor allele. Percentages not always 100% because of rounding. ORs are estimated with logistic regression analysis. Adjustment for age and gender only in the Rotterdam study.