

Appendix 3. Risk of AMD (subgroup) cases for *SLC2A1* SNP rs3754219 in five replication populations

	No AMD		Early AMD		GA		CNV		CNV + Mixed cases		Mixed AMD	
The Rotterdam study												
<i>rs3754219</i>	N= 2221	N= 894	N=72		N=64		N=103		N=39			
	No. (%)	No. (%)	OR (95% CI)	No. (%)	OR (95% CI)	No. (%)	OR (95% CI)			No. (%)	OR (95% CI)	
Genotype												
Noncarrier (AA)	711 (32.0)	279 (31.2)	1	18 (25.0)	1	25 (39.1)	1	38 (36.9)	1	13 (33.3)	1	
Heterozygous (Aa)	1109 (49.9)	442 (49.4)	1.02 (0.85-1.22)	38 (52.8)	1.38 (0.78-2.46)	33 (51.6)	0.87 (0.51-1.48)	51 (49.5)	0.86 (0.56-1.32)	18 (46.2)	0.93 (0.45-1.92)	
Homozygous (aa)	401 (18.0)	173 (19.4)	1.07 (0.86-1.35)	16 (22.2)	1.56 (0.78-3.12)	6 (9.3)	0.41 (0.17-1.02)	14 (13.6)	0.65 (0.35-1.22)	8 (20.5)	1.04 (0.42-2.56)	
m.a.f(%)	43	44.1		48.6		35.1		38.3		43.6		
The Franconian AMD study												
<i>rs3754219</i>	N=607	N=86	N=162		N=420		N=546		N=126			
	No(%)	No(%)	OR (95% CI)	No(%)	OR (95% CI)	No(%)	OR (95% CI)	No(%)	OR (95% CI)	No(%)	OR (95% CI)	
Genotype												
Noncarrier (AA)	194 (32.0)	26 (30.2)	1	42 (25.9)	1	135 (32.1)	1	170 (31.1)	1	35 (28.8)	1	
Heterozygous (Aa)	298 (49.1)	42 (48.8)	1.05 (0.62-1.77)	82 (50.6)	1.27 (0.84-1.92)	192 (45.7)	0.93 (0.70-1.23)	256 (46.9)	0.98 (0.75-1.28)	64 (50.8)	1.19 (0.76-1.87)	
Homozygous (aa)	115 (18.9)	18 (20.9)	1.17 (0.61-2.22)	38 (23.5)	1.53 (0.93-2.51)	93 (22.1)	1.16 (0.82-1.65)	120 (22.0)	1.19 (0.86-1.65)	27 (21.4)	1.30 (0.75-2.26)	
m.a.f(%)	43.5	45.3		48.8		45		45.4		46.8		
AREDS												
<i>rs3754219</i>	N=212	N=250	N=167		N=322		N=500		N=178			
	No(%)	No(%)	OR (95% CI)	No(%)	OR (95% CI)	No(%)	OR (95% CI)	No(%)	OR (95% CI)	No. (%)	OR (95% CI)	
Genotype												
Noncarrier (AA)	56 (26.4)	84 (33.6)	1	54 (32.3)	1	98 (30.4)	1	151 (30.2)	1	53 (29.8)	1	
Heterozygous (Aa)	108 (50.9)	113 (45.2)	0.70 (0.45-1.07)	73 (43.7)	0.70 (0.43-1.13)	160 (49.7)	0.85 (0.56-1.27)	251 (50.2)	0.86 (0.59-1.26)	91 (51.1)	0.89 (0.56-1.42)	
Homozygous (aa)	48 (22.6)	53 (21.2)	0.74 (0.44-1.23)	40 (23.9)	0.86 (0.49-1.52)	64 (19.9)	0.76 (0.46-1.25)	98 (19.6)	0.76 (0.48-1.20)	34 (19.1)	0.75 (0.42-1.33)	
m.a.f(%)	48.1	43.8		45.8		44.7		44.7		44.7		
Columbia University												
<i>rs3754219</i>	N=365	N=361	N=208		N=522							
	No(%)	No(%)	OR (95% CI)	No(%)	OR (95% CI)	No(%)	OR (95% CI)	No(%)	OR (95% CI)	No(%)	OR (95% CI)	
Genotype												
Noncarrier (AA)	121 (33.2)	126 (34.9)	1	59 (28.4)	1	N.P.	167 (32.0)	1	N.P.			
Heterozygous (Aa)	187 (51.2)	173 (47.9)	0.89 (0.64-1.23)	103 (49.5)	1.13 (0.76-1.67)		248 (47.5)	0.96 (0.71-1.30)				
Homozygous (aa)	57 (15.6)	62 (17.2)	1.04 (0.67-1.62)	46 (22.1)	1.66 (1.01-2.72)		107 (20.5)	1.36 (0.91-2.02)				
m.a.f(%)	41.2	41.1		46.9			44.3					
The University of Iowa												
<i>rs3754219</i>	N=383	N=323	N=79		N=649		N=689		N=40			
	No(%)	No(%)	OR (95% CI)	No(%)	OR (95% CI)	No(%)	OR (95% CI)	No(%)	OR (95% CI)	No. (%)	OR (95% CI)	
Genotype												
Noncarrier (AA)	110 (28.7)	100 (30.9)	1	30 (37.9)	1	197 (30.3)	1	208 (30.2)	1	11 (27.5)	1	
Heterozygous (Aa)	194 (50.6)	156 (48.3)	0.88 (0.63-1.25)	34 (43.0)	0.64 (0.37-1.11)	318 (49.0)	0.92 (0.68-1.23)	342 (49.6)	0.93 (0.70-1.25)	24 (60.0)	1.24 (0.58-2.62)	
Homozygous (aa)	79 (20.6)	67 (20.7)	0.93 (0.61-1.42)	15 (19.0)	0.70 (0.35-1.38)	134 (20.6)	0.95 (0.66-1.36)	139 (20.2)	0.93 (0.65-1.33)	5 (12.5)	0.63 (0.21-1.89)	
m.a.f(%)	45.9	44.9		40.5		45.1		45.0		40.0		

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.