

Appendix 4. SNPs identified in the Complement Factor H gene re-sequencing in Mexicans, 48 Age-Macular Degeneration patients and 48 healthy controls.

Localization	dbSNP ID	Nucleotide change	Codon change	Functional effect	Allele frequency (%)		OR (CI 0.95)	p-value*	p-value†
					Cases (n=48)	Controls (n=48)			
intron 1	rs551397	C/T	----	----	22.3	37.5	0.45 (0.22-0.93)	3.17E-02	NS
exon 2	rs800292	G/A	GTA/ATA	Val62Ile	22.3	37.5	0.45 (0.22-0.93)	3.17E-02	NS
intron 2	---	ins TT	----	----	30.0	40.0	NS	NS	NS
exon 5	novel	C/T	GAC/GAT	Asp194Asp	0	1.06	NS	NS	NS
exon 7	rs1061147	C/A	GCC/GCA	Ala307Ala	30.2	8.0	9.21 (2.96-28.59)	1.22E-04	2.81E-03
intron 7	rs482934	T/G	----	----	30.7	5.3	12.93 (3.64- 45.87)	7.43E-05	1.71E-03
intron 8	rs12029785	A/G	----	----	30.7	7.5	8.69 (2.89- 26.11)	1.16E-04	2.68E-03
exon 9	rs1061170	T/C	TAT/CAT	Tyr402His	30.0	9.2	6.94 (2.35- 20.42)	4.33E-04	9.96E-03
intron 10	rs4658046	T/C	----	----	26.1	10.9	5.19 (1.92- 14.02)	1.14E-03	2.62E-02
exon 11	rs2274700	G/A	GCG/GCA	Ala473Ala	35.1	48.9	0.47 (0.23-0.94)	3.49E-02	NS
exon 14	rs3753396	A/G	CAA/CAG	Gln672Gln	37.2	37.2	NS	NS	NS
intron 16	rs375046	A/C	----	----	32.2	10.9	6.88 (2.49- 18.98)	1.92E-04	4.43E-03

intron 16	rs435628	T/G	----	----	0	2.2	NS	NS	NS
exon 17	novel	G/A	CAG/CAA	Gln816Gln	2.2	0	NS	NS	NS
exon 18	rs35292876	C/T	CAC/CAT	His878His	2.2	1.2	NS	NS	NS
exon 18	rs515299	G/T	AGT/ATT	Ser890Ile	0	1.2	NS	NS	NS
exon 19	rs1065489	G/T	GAG/GAT	Glu936Asp	37.0	37.2	NS	NS	NS
intron 19	rs16840522	T/C	----	----	12.0	13.9	NS	NS	NS
intron 20	novel	C/G	----	----	0	2	NS	NS	NS
exon 21	rs55679475	T/C	TAT/CAT	Tyr1058His	22.2	13.8	NS	NS	NS
exon 21	rs55771831	G/C	GTG/CTG	Val1060Leu	22.2	13.8	NS	NS	NS
exon 21	rs62625015	C/G	CAA/GAA	Gln1076Glu	22.2	12.8	2.84 (1.01- 7.98)	4.65E-02	NS
exon 21	rs76835795	G/A	ACG/ACA	Thr1097Thr	22.2	12.8	2.84 (1.01- 7.98)	4.65E-02	NS

*Association analyses were done using logistic regression assuming an additive model and adjusting by age and sex as covariates.

†p-value adjusted for multiple testing by Bonferroni correction.

OR= odds ratio; C.I.= confidence interval; NS= no significant