SUPPLEMENTARY TABLES

Table 1. Case and control definition.

| AMD Cases (n = 134) | Hospital-based controls (n = 134) | | |
|--|---|--|--|
| 65 years or older. | 65 years or older. | | |
| Any gender. | Any gender. | | |
| With or without a family history of AMD. | No family history of AMD. | | |
| AMD CARMS grades 4 or 5†. | No retinal changes suggestive of advanced | | |
| No history of vitreoretinal procedures. | AMD by fundoscopy. | | |
| No other concurrent retinal diseases. | Drusen less than 65 µm in diameter by | | |
| | fundoscopy. # | | |

The presence of a few small hard drusen less than 65 μm in diameter is common and is no longer considered to be a risk factor for the development of age related maculopathy^{[31]}

| Characteristic | Cases | Controls | p† | |
|------------------------|----------------|----------------|---------|--|
| | (n = 134)* | (n = 134)* | - | |
| Age (years), mean ± SD | 76.7 ± 7.6 | 71.9 ± 8.1 | <0.0001 | |
| Age (years), n (%) | | | <0.0001 | |
| [50 - 60) | 0 (0.0) | 7 (5.3) | | |
| [60 – 65) | 7 (5.2) | 16 (12.0) | | |
| [65 – 70) | 18 (13.4) | 28 (21.1) | | |
| [70 – 75) | 28 (20.9) | 32 (24.1) | | |
| [75 – 80) | 30 (22.4) | 29 (21.8) | | |
| ≥80 | 51 (38.1) | 21 (15.8) | | |
| Sex, n (%) | | | 0.098 | |
| Male | 42 (31.3) | 55 (41.0) | | |
| Female | 92 (68.7) | 79 (59.0) | | |
| Type 2 diabetes, n (%) | 32 (24.4) | 39 (30.5) | 0.276 | |
| Hypertension, n (%) | 73 (54.5) | 61 (47.3) | 0.244 | |
| Smoking history, n (%) | | | 0.741 | |
| Never | 106 (79.7) | 96 (81.4) | | |
| Former or current | 27 (20.3) | 22 (18.6) | | |

Table 2. Description of the sample by case/control status (n = 268).

Stratified characteristics by case/control status. Statistically significant differences are shown in bold and were computed with Student's t-Test (Continuous variables) or χ^2 test (categorical variable). We excluded one subject from this analysis because of age (<50 years old).

* Numbers may not sum to totals due to missing data, and column percentages may not sum to 100% due to rounding.

+ P-value for Student's t-test (continuous variable) or χ^2 test (categorical variable).

^ One subject was excluded because of being younger than 50 years old.

In bold significant characteristics at the 0.05 level.

| Characteristic [^] | N* (% with AMD) | OR (95% CI) | p* | |
|-----------------------------|-----------------|-------------------|--------|--|
| rs970476 | | | | |
| G/G | 41 (30.6) | 1.00 | | |
| G/T | 63 (47.0) | 1.02 (0.59, 1.79) | NS | |
| Γ/Τ | 30 (22.4) | 1.06 (0.54, 2.07) | NS | |
| rs931798 | | | | |
| G/G | 51 (38.1) | 1.00 | | |
| G/A | 70 (52.2) | 1.74 (1.04, 2.90) | 0.034 | |
| A/A | 13 (9.7) | 1.22 (0.52, 2.83) | NS | |
| rs140617 | | | | |
| A/A | 97 (72.4) | 1.00 | | |
| G/A | 31 (23.1) | 0.80 (0.46, 1.40) | NS | |
| G/G | 6 (4.5) | 1.42 (0.40, 5.78) | NS | |
| rs140616 | | | | |
| T/T | 34 (25.4) | 1.00 | | |
| T/C | 70 (52.2) | 1.01 (0.56, 1.82) | NS | |
| C/C | 30 (22.4) | 0.86 (0.43, 1.70) | NS | |
| Age(years) | _ | 1.08 (1.05, 1.12) | <0.000 | |
| Sex | | | | |
| Female | 92 (68.7) | 1.00 | | |
| Male | 42 (31.3) | 0.66 (0.40, 1.08) | NS | |
| Type 2 diabetes | | | | |
| No | 99 (75.6) | 1.00 | | |
| Yes | 32 (24.4) | 0.73 (0.42, 1.28) | NS | |
| Hypertension | | | | |
| No | 61 (45.5) | 1.00 | | |
| Yes | 73 (54.5) | 1.33 (0.82, 2.17) | NS | |
| Smoking history | | | | |
| Never | 106 (79.7) | 1.00 | | |
| Former or current | 27 (20.3) | 1.11 (0.60, 2.08) | NS | |

Table 3. Unadjusted associations between study variables and age-related macular degeneration (n = 268).

Bivariate associations between baseline characteristics and AMD diagnosis (0–No, 1–Yes AMD). For genetic data, we assumed a genotypic mode of inheritance. Such models follow: $log(OR_{AMD}) \sim (AA_{00})$

 $SNP\begin{pmatrix}AA_{00}\\Aa_{01}\\aa_{10}\end{pmatrix}$ + \in where: AA is the most frequent allele in our population, taken as reference. We considered

statistically significant predictors of odds of disease those whose p-value < 0.05.

* Numbers may not sum to total due to missing data.

+ p-value for β significance

NS: not significant at the 0.05 level.

[^]We took the most common allele for each case and set it as reference. Effects displayed first as those of the intercept for each model.

~ One subject was excluded because of being younger than 50 years old.

In bold significant predictors at the 0.05 level.

| Characteristic* | Geographic atrophy OR (95% CI) | p ⁺ | Neovascular OR (95% CI) | P ⁺ | |
|-------------------|-----------------------------------|-----------------------|----------------------------|-----------------------|--|
| rs931798^ | | | | | |
| G/G | 1.00 | | 1.00 | | |
| G/A | 1.82 (1.03, 3.21) | 0.038 | 1.41 (0.67, 2.98) | NS | |
| A/A | 1.27 (0.50, 3.22) | NS | 1.13 (0.33, 3.86) | NS | |
| Age(years) | 1.08 (1.04, 1.12) | <0.0001 | 1.09 (1.04, 1.14) | < 0.0001 | |
| Sex | | | | | |
| Female | 1.00 | | 1.00 | | |
| Male | 0.70 (0.40, 1.21) | NS | 0.54 (0.25, 1.18) | NS | |
| Type 2 diabetes | | | | | |
| No | 1.00 | | 1.00 | | |
| Yes | 0.65 (0.35, 1.22) | NS | 0.86(0.39, 1.92) | NS | |
| Hypertension | | | | | |
| No | 1.00 | | 1.00 | | |
| Yes | 1.39 (0.81, 2.38) | NS | 1.23 (0.61, 2.5 | NS | |
| Smoking history | | | | | |
| Never | 1.00 | | 1.00 | | |
| Former or current | 1.32 (0.67, 2.56) | NS | 0.77 (0.29, 2.04) | NS | |

Table 4. Unadjusted associations between study variables and age-related macular degeneration phenotype (n = 268).

Bivariate associations between baseline characteristics and AMD phenotype (either 1–GA, 0–else; or 1–NV, 0–else). For genetic data, we assumed a genotypic mode of inheritance. Such models follow: $log(OR_{AMD \ phenotype}) \sim SNP\begin{pmatrix}AA_{00}\\Aa_{01}\\aa_{10}\end{pmatrix} + \epsilon$ where: AA is the most frequent allele in our population, taken as reference. We considered statistically significant predictors of odds of disease those whose p-value < 0.05.

+ p-value for β significance

NS: not significant at the 0.05 level.

*Controls or non-diseased phenotype are set as reference for all multinomial logistic regression models.

^We took the most common allele and set it as reference.

In bold significant predictors at the 0.05 level.

| # | SNP 1 | SNP 2 | SNP 3 | SNP 4 | Pooled HF | Control HF | Case HF | OR (95% CI) | p* |
|---|----------|----------|----------|----------|--------------|---------------|------------|-------------------|-------|
| 1 | G | Α | Т | Т | 0.022 | 0.039 | 0.006 | 0.14 (0.02, 0.93) | 0.011 |
| 2 | А | G | Т | Т | 0.012 | 0.019 | 0.005 | 0.21 (0.02, 2.04) | 0.216 |
| 3 | G | А | С | Т | 0.099 | 0.107 | 0.089 | 0.82 (0.44, 1.51) | 0.428 |
| 4 | А | А | Т | G | 0.017 | 0.022 | 0.012 | 0.62 (0.16, 2.41) | 0.530 |
| 5 | G | А | Т | G | 0.022 | 0.022 | 0.021 | 1.00 (0.27, 3.75) | 0.754 |
| 6 | G | А | С | G | 0.378 | 0.384 | 0.375 | 1.00 (NA, NA) | 0.896 |
| 7 | G | G | Т | G | 0.114 | 0.114 | 0.115 | 0.97 (0.52, 1.81) | 0.956 |
| 8 | G | G | Т | Т | 0.025 | 0.023 | 0.027 | 1.84 (0.42, 7.98) | 0.857 |
| 9 | А | А | Т | Т | 0.293 | 0.257 | 0.329 | 1.31 (0.84, 2.02) | 0.084 |

Table 5. Unadjusted haplotypes of four SNPs with AMD (n = 268).

Haplotype configurations of SNPs in the SGCD gene. We show their frequency (HF) in the full sample (Pooled HF) and stratified by case/control status. Also, bivariate associations between a haplotype configuration (#) and AMD diagnosis using logistic regression modeling.

SNP1: rs931798, SNP2: rs140617, SNP3: rs140616, SNP4: rs970476.

Four single-nucleotide polymorphism (SNP) haplotype configuration.

+ p-value for haplotype $\chi 2$ test evaluated at the 0.05 level.

HF: Haplotype frequency among cases, controls, and full study sample (pooled).

NA: Not able to calculate by this method.

In bold significant haplotypes at the 0.05 level.