

References - Genetics of AMD

1. Guymer R. The genetics of age-related macular degeneration. *Clin Exp Optom*. 2001; 84: 182-9.
2. Seddon JM, Francis PJ, George S, Schultz DW, Rosner B, Klein ML. Association of CFH Y402H and LOC387715 A69S with progression of age-related macular degeneration. *JAMA* 2007; 297 (16): 1793-800.
3. Smith W, Mitchell P. Family history and age-related maculopathy: the Blue Mountains Eye Study. *Aust N Z J Ophthalmol*. 1998; 26 (3): 203-6.
4. Luo L, Harmon J, Yang X, Chen H, Patel S, Mineau G, Yang Z, Constantine R, Buehler J, Kaminoh Y, Ma X, Wong TY, Zhang M, Zhang K. Familial aggregation of age-related macular degeneration in the Utah population. *Vision Res* 2008; 48 (3): 494-500.
5. Fisher SA, Abecasis GR, Yashar BM, Zareparsis S, Swaroop A, Iyengar SK, Klein BE, Klein R, Lee KE, Majewski J, Schultz DW, Klein ML, Seddon JM, Santangelo SL, Weeks DE, Conley YP, Mah TS, Schmidt S, Haines JL, Pericak-Vance MA, Gorin MB, Schulz HL, Pardi F, Lewis CM, Weber BH. Meta-analysis of genome scans of age-related macular degeneration. *Hum Mol Genet* 2005; 14 (15): 2257-64.
6. Klein ML, Schultz DW, Edwards A, Matise TC, Rust K, Berselli CB, Trzupke K, Weleber RG, Ott J, Wirtz MK, Acott TS. Age-related macular degeneration. Clinical features in a large family and linkage to chromosome 1q. *Arch Ophthalmol* 1998; 116 (8): 1082-8.
7. Majewski J, Schultz DW, Weleber RG, Schain MB, Edwards AO, Matise TC, Acott TS, Ott J, Klein ML. Age-related macular degeneration--a genome scan in extended families. *Am J Hum Genet* 2003; 73 (3): 540-50.
8. Donoso LA, Kim D, Frost A, Callahan A, Hageman G. The role of inflammation in the pathogenesis of age-related macular degeneration. *Surv Ophthalmol* 2006; 51 (2): 137-52.
9. Tuo J, Bojanowski CM, Chan CC. Genetic factors of age-related macular degeneration. *Prog Retin Eye Res* 2004; 23 (2): 229-49.
10. Johnson PT, Betts KE, Radeke MJ, Hageman GS, Anderson DH, Johnson LV. Individuals homozygous for the age-related macular degeneration risk-conferring variant of complement factor H have elevated levels of CRP in the choroid. *Proc Natl Acad Sci U S A* 2006; 103 (46): 17456-61.
11. Edwards AO, Ritter R, Abel KJ, Manning A, Panhuysen C, Farrer LA. Complement factor H polymorphism and age-related macular degeneration. *Science* 2005; 308 (5720): 362-4.
12. Hageman GS, Anderson DH, Johnson LV, Hancox LS, Taiber AJ, Hardisty LI, Hageman JL, Stockman HA, Borchardt JD, Gehrs KM, Smith RJ, Silvestri G, Russell SR, Klaver CC, Barbazetto I, Chang S, Yannuzzi LA, Barile GR, Merriam JC, Smith RT, Olsh AK, Bergeron J, Zernant J, Merriam JE, Gold B, Dean M, Allikmets R. A common haplotype in the complement regulatory gene factor H (HF1/CFH) predisposes individuals to age-related macular degeneration. *Proc Natl Acad Sci U S A* 2005; 102 (20): 7227-32.
13. Haines JL, Hauser MA, Schmidt S, Scott WK, Olson LM, Gallins P, Spencer KL, Kwan SY, Noureddine M, Gilbert JR, Schnetz-Boutaud N, Agarwal A, Postel EA, Pericak-Vance MA. Complement factor H variant increases the risk of age-related macular degeneration. *Science* 2005; 308 (5720): 419-21.
14. Klein RJ, Zeiss C, Chew EY, Tsai JY, Sackler RS, Haynes C, Henning AK, SanGiovanni JP, Mane SM, Mayne ST, Bracken MB, Ferris FL, Ott J, Barnstable C, Hoh J. Complement factor H polymorphism in age-related macular degeneration. *Science* 2005; 308 (5720): 385-9.
15. Tuo J, Ning B, Bojanowski CM, Lin ZN, Ross RJ, Reed GF, Shen D, Jiao X, Zhou M, Chew EY, Kadlubar FF, Chan CC. Synergic effect of polymorphisms in ERCC6 5' flanking region and complement factor H on age-related macular degeneration predisposition. *Proc Natl Acad Sci U S A* 2006; 103 (24): 9256-61.
16. Chen LJ, Liu DT, Tam PO, Chan WM, Liu K, Chong KK, Lam DS, Pang CP. Association of complement factor H polymorphisms with exudative age-related macular degeneration. *Mol Vis* 2006; 12: 1536-42.
17. Okamoto H, Umeda S, Obazawa M, Minami M, Noda T, Mizota A, Honda M, Tanaka M, Koyama R, Takagi I, Sakamoto Y, Saito Y, Miyake Y, Iwata T. Complement factor H polymorphisms in Japanese population with age-related macular degeneration. *Mol Vis* 2006; 12: 156-8.
18. Zareparsis S, Branham KE, Li M, Shah S, Klein RJ, Ott J, Hoh J, Abecasis GR, Swaroop A. Strong association of the Y402H variant in complement factor H at 1q32 with susceptibility to age-related

macular degeneration. *Am J Hum Genet* 2005; 77 (1): 149-53.

19. Thakkinstian A, Han P, McEvoy M, Smith W, Hoh J, Magnusson K, Zhang K, Attia J. Systematic review and meta-analysis of the association between complement factor H Y402H polymorphisms and age-related macular degeneration. *Hum Mol Genet* 2006;15 (18): 2784-90.

20. Hageman GS, Hancox LS, Taiber AJ, Gehrs KM, Anderson DH, Johnson LV, Radeke MJ, Kavanagh D, Richards A, Atkinson J, Meri S, Bergeron J, Zernant J, Merriam J, Gold B, Allikmets R, Dean M; AMD Clinical Study Group. Extended haplotypes in the complement factor H (CFH) and CFH-related (CFHR) family of genes protect against age-related macular degeneration: characterization, ethnic distribution and evolutionary implications. *Ann Med* 2006; 38 (8): 592-604.

21. Gold B, Merriam JE, Zernant J, Hancox LS, Taiber AJ, Gehrs K, Cramer K, Neel J, Bergeron J, Barile GR, Smith RT; AMD Genetics Clinical Study Group, Hageman GS, Dean M, Allikmets R. Variation in factor B (BF) and complement component 2 (C2) genes is associated with age-related macular degeneration. *Nat Genet* 2006; 38 (4): 458-62.

22. Hageman GS, Luthert PJ, Victor Chong NH, Johnson LV, Anderson DH, Mullins RF. An integrated hypothesis that considers drusen as biomarkers of immune-mediated processes at the RPE-Bruch's membrane interface in aging and age-related macular degeneration. *Prog Retin Eye Res* 2001; 20 (6): 705-32.

23. Johnson LV, Leitner WP, Staples MK, Anderson DH. Complement activation and inflammatory processes in Drusen formation and age related macular degeneration. *Exp Eye Res* 2001; 73 (6): 887-96.

24. Nozaki M, Raisler BJ, Sakurai E, Sarma JV, Barnum SR, Lambris JD, Chen Y, Zhang K, Ambati BK, Baffi JZ, Ambati J. Drusen complement components C3a and C5a promote choroidal neovascularization. *Proc Natl Acad Sci U S A* 2006; 103 (7): 2328-33.

25. Despret DD, van Duijn CM, Oostra BA, Uitterlinden AG, Hofman A, Wright AF, ten Brink JB, Bakker A, de Jong PT, Vingerling JR, Bergen AA, Klaver CC. Complement component C3 and risk of age-related macular degeneration. *Ophthalmology* 2009; 116 (3):474-480.e2.

26. Weeks DE, Conley YP, Tsai HJ, Mah TS, Schmidt S, Postel EA, Agarwal A, Haines JL, Pericak-Vance MA, Rosenfeld PJ, Paul TO, Eller AW, Morse LS, Dailey JP, Ferrell RE, Gorin MB. Age-related maculopathy: a genomewide scan with continued evidence of susceptibility loci within the 1q31, 10q26, and 17q25 regions. *Am J Hum Genet* 2004; 75 (2): 174-89.

27. Jakobsdottir J, Conley YP, Weeks DE, Mah TS, Ferrell RE, Gorin MB. Susceptibility genes for age-related maculopathy on chromosome 10q26. *Am J Hum Genet* 2005; 77 (3): 389-407.

28. Allikmets R, Dean M. Bringing age-related macular degeneration into focus. *Nat Genet* 2008; 40 (7): 820-1.

29. Conley YP, Jakobsdottir J, Mah T, Weeks DE, Klein R, Kuller L, Ferrell RE, Gorin MB. CFH, ELOVL4, PLEKHA1 and LOC387715 genes and susceptibility to age-related maculopathy: AREDS and CHS cohorts and meta-analyses. *Hum Mol Genet* 2006; 15 (21): 3206-18.

30. Rivera A, Fisher SA, Fritsche LG, Keilhauer CN, Lichtner P, Meitinger T, Weber BH. Hypothetical LOC387715 is a second major susceptibility gene for age-related macular degeneration, contributing independently of complement factor H to disease risk. *Hum Mol Genet* 2005; 14 (21): 3227-36.

31. Ross RJ, Bojanowski CM, Wang JJ, Chew EY, Rochtchina E, Ferris FL 3rd, Mitchell P, Chan CC, Tuo J. The LOC387715 polymorphism and age-related macular degeneration: replication in three case-control samples. *Invest Ophthalmol Vis Sci* 2007; 48 (3): 1128-32.

32. Kanda A, Chen W, Othman M, Branham KE, Brooks M, Khanna R, He S, Lyons R, Abecasis GR, Swaroop A. A variant of mitochondrial protein LOC387715/ARMS2, not HTRA1, is strongly associated with age-related macular degeneration. *Proc Natl Acad Sci U S A*. 2007; 104 (41): 16227-32.

33. Wang JJ, Ross RJ, Tuo J, Burlutsky G, Tan AG, Chan CC, Favaloro EJ, Williams A, Mitchell P. The LOC387715 polymorphism, inflammatory markers, smoking, and age-related macular degeneration. A population-based case-control study. *Ophthalmology* 2008; 115 (4): 693-9.

34. Yang Z, Camp NJ, Sun H, Tong Z, Gibbs D, Cameron DJ, Chen H, Zhao Y, Pearson E, Li X, Chien J, Dewan A, Harmon J, Bernstein PS, Shridhar V, Zabriskie NA, Hoh J, Howes K, Zhang K. A variant of the HTRA1 gene increases susceptibility to age-related macular degeneration. *Science* 2006; 314 (5801): 992-3.

35. Dewan A, Liu M, Hartman S, Zhang SS, Liu DT, Zhao C, Tam PO, Chan WM, Lam DS, Snyder M,

- Barnstable C, Pang CP, Hoh J. HTRA1 promoter polymorphism in wet age-related macular degeneration. *Science* 2006; 314 (5801): 989-92.
36. Oka C, Tsujimoto R, Kajikawa M, Koshiha-Takeuchi K, Ina J, Yano M, Tsuchiya A, Ueta Y, Soma A, Kanda H, Matsumoto M, Kawaichi M. HtrA1 serine protease inhibits signaling mediated by Tgfbeta family proteins. *Development* 2004; 131 (5): 1041-53.
37. Launay S, Maubert E, Lebeurrier N, Tennstaedt A, Campioni M, Docagne F, Gabriel C, Dauphinot L, Potier MC, Ehrmann M, Baldi A, Vivien D. HtrA1-dependent proteolysis of TGF-beta controls both neuronal maturation and developmental survival. *Cell Death Differ* 2008; 15 (9): 1408-16.
38. Cameron DJ, Yang Z, Gibbs D, Chen H, Kaminoh Y, Jorgensen A, Zeng J, Luo L, Brinton E, Brinton G, Brand JM, Bernstein PS, Zabriskie NA, Tang S, Constantine R, Tong Z, Zhang K. HTRA1 variant confers similar risks to geographic atrophy and neovascular age-related macular degeneration. *Cell Cycle* 2007; 6 (9): 1122-5.
39. Mori K, Horie-Inoue K, Kohda M, Kawasaki I, Gehlbach PL, Awata T, Yoneya S, Okazaki Y, Inoue S. Association of the HTRA1 gene variant with age-related macular degeneration in the Japanese population. *J Hum Genet* 2007; 52(7): 636-41.
40. Tam PO, Ng TK, Liu DT, Chan WM, Chiang SW, Chen LJ, DeWan A, Hoh J, Lam DS, Pang CP. HTRA1 variants in exudative age-related macular degeneration and interactions with smoking and CFH. *Invest Ophthalmol Vis Sci* 2008; 49 (6): 2357-65.
41. Deangelis MM, Ji F, Adams S, Morrison MA, Harring AJ, Sweeney MO, Capone A Jr, Miller JW, Dryja TP, Ott J, Kim IK. Alleles in the HtrA serine peptidase 1 gene alter the risk of neovascular age-related macular degeneration. *Ophthalmology* 2008; 115 (7): 1209-1215.e7.
42. Fritsche LG, Loenhardt T, Janssen A, Fisher SA, Rivera A, Keilhauer CN, Weber BH. Age-related macular degeneration is associated with an unstable ARMS2 (LOC387715) mRNA. *Nat Genet* 2008; 40 (7): 892-6.
43. Barreau C, Paillard L, Osborne HB. AU-rich elements and associated factors: are there unifying principles? *Nucleic Acids Res* 2006; 33 (22): 7138-50.
44. Garneau NL, Wilusz J, Wilusz CJ. The highways and byways of mRNA decay. *Nat Rev Mol Cell Biol* 2007; 8 (2): 113-26.
45. Khabar KS. The AU-rich transcriptome: more than interferons and cytokines, and its role in disease. *J Interferon Cytokine Res* 2005; 25 (1): 1-10.
46. Ding X, Patel M, Chan CC. Molecular pathology of age-related macular degeneration. *Prog Retin Eye Res* 2009; 28 (1): 1-18.
47. Schmidt S, Klaver C, Saunders A, Postel E, De La PM, Agarwal A, Small K, Udar N, Ong J, Chalukya M, Nesburn A, Kenney C, Domurath R, Hogan M, Mah T et al. A pooled case-control study of the apolipoprotein E (APOE) gene in age-related maculopathy. *Ophthalmic Genet* 2002; 23 (4): 209-223.
48. Baird PN, Richardson AJ, Robman LD, Dimitrov PN, Tikellis G, McCarty CA, Guymer RH. Apolipoprotein (APOE) gene is associated with progression of age-related macular degeneration (AMD). *Hum Mutat* 2006; 27 (4): 337-342.
49. Kovacs KA, Pamer Z, Kovacs A, Fekete S, Miseta A, Kovacs B, Kovacs GL. Association of apolipoprotein E polymorphism with age-related macular degeneration and Alzheimer's disease in south-western Hungary. *Ideggyogy Sz* 2007; 60 (3-4): 169-172.
50. Souied EH, Benlian P, Amouyel P, Feingold J, Lagarde JP, Munnich A, Kaplan J, Coscas G, Soubrane G. The epsilon4 allele of the apolipoprotein E gene as a potential protective factor for exudative age-related macular degeneration. *Am J Ophthalmol* 1998; 125 (3): 353-359.
51. Zarepari S, Reddick AC, Branham KE, Moore KB, Jessup L, Thoms S, Smith-Wheelock M, Yashar BM, Swaroop A. Association of apolipoprotein E alleles with susceptibility to age-related macular degeneration in a large cohort from a single center. *Invest Ophthalmol Vis Sci* 2004; 45 (5): 1306-1310.
52. Kaur I, Hussain A, Hussain N, Das T, Pathangay A, Mathai A, Hussain A, Nutheti R, Nirmalan PK, Chakrabarti S. Analysis of CFH, TLR4, and APOE polymorphism in India suggests the Tyr402His variant of CFH to be a global marker for age-related macular degeneration. *Invest Ophthalmol Vis Sci* 2006; 47 (9): 3729-3735.
53. Schmidt S, Haines JL, Postel EA, Agarwal A, Kwan SY, Gilbert JR, Pericak-Vance MA, Scott WK. Joint effects of smoking history and APOE genotypes in age-related macular degeneration. *Mol Vis* 2005; 11:

941-949.

54. Schultz DW, Klein ML, Humpert A, Majewski J, Schain M, Weleber RG, Ott J, Acott TS. Lack of an association of apolipoprotein E gene polymorphisms with familial age-related macular degeneration. *Arch Ophthalmol* 2003; 121 (5): 679-683.
55. Wong TY, Shankar A, Klein R, Bray MS, Couper DJ, Klein BE, Sharrett AR, Folsom AR. Apolipoprotein E gene and early age-related maculopathy: the Atherosclerosis Risk in Communities Study. *Ophthalmology* 2006; 113 (2): 255-259.
56. Klein ML, Francis PJ, Rosner B, Reynolds R, Hamon SC, Schultz DW, Ott J, Seddon JM. CFH and LOC387715/ARMS2 genotypes and treatment with antioxidants and zinc for age-related macular degeneration. *Ophthalmology* 2008; 115 (6): 1019-1025.
57. Brantley MA, Jr., Fang AM, King JM, Tewari A, Kymes SM, Shiels A. Association of complement factor H and LOC387715 genotypes with response of exudative age-related macular degeneration to intravitreal bevacizumab. *Ophthalmology* 2007; 114 (12): 2168-2173.
58. Goverdhan SV, Hannan S, Newsom RB, Luff AJ, Griffiths H, Lotery AJ. An analysis of the CFH Y402H genotype in AMD patients and controls from the UK, and response to PDT treatment. *Eye (Lond)* 2008; 22 (6): 849-854.
59. Brantley MA, Jr., Edelstein SL, King JM, Plotzke MR, Apte RS, Kymes SM, Shiels A. Association of complement factor H and LOC387715 genotypes with response of exudative age-related macular degeneration to photodynamic therapy. *Eye (Lond)* 2009; 23 (3): 626-631.

[View PDF](#)