

Table 1. Risk of AMD in relatives of AMD patients based on familial aggregation studies [24–26]

	Siblings	Offspring	Reference
Late AMD	25 (3.4–519)	7.4 (0.4–153)	Silvestri et al., 1994
Exudative AMD		3 (1.5–6.7)	Seddon et al., 1997
ARMD	4.8	6.6	Klaver et al., 1998
AMD	19.8	19.8	

ARMD = ■

Table 2. Loci associated with AMD in the main linkage analyses [31–37 + Kenealy et al. ■, missing]

Chromosome	Locus	Significant HLOD	–log10 (P)	p	Number of markers	Number of families	Number of affected individuals	References
1	1q24	2.41			393	158	490	Seddon et al., 2003
	1q31	3–2.07	2.89		572+46	1	10	Klein et al., 1998
					406	391	860	Weeks et al., 2001
					393	158	490	Seddon et al., 2003
					364	70	344	Majewski et al., 2003
					383+43	34	381	Iyengar et al., 2004
					773	113	331	Abecasis et al., 2004
2	2p25.3	1.96		0.001	773	113	331	Abecasis et al., 2004
	2p21		2.39	0.0024	383+43	34	381	Iyengar et al., 2004
	2q33	2.37	1.81		393	158	490	Seddon et al., 2003
3	3p13	2.19			364	70	344	Majewski et al., 2003
	3q26.1		2.09	0.0062	345+25	102	258	Schick et al., 2003
4	4p16		2.06	0.0065	383+43	34	381	Iyengar et al., 2004
	4q32	2.66			364	70	344	Majewski et al., 2003
5	5p14.1–p13.3	2.55		0.0003	773	113	331	Abecasis et al., 2004
	5q12–5q13			0.0382	345+25	102	258	Schick et al., 2003
	5q34		2.31	0.0021	383+43	34	381	Iyengar et al., 2004
6	6q13–6q14			0.0415	345+25	102	258	Schick et al., 2003
	6q23–6q24			0.0286	345+25	102	258	Schick et al., 2003
9	9p24		2.32	0.0032	383+43	34	381	Iyengar et al., 2004
	9p13	1.79	2.04		406	391	860	Weeks et al., 2000
	9q31		2.1	0.0038	383+43	34	381	Iyengar et al., 2004
	9q31.1		1.65		773	113	331	Abecasis et al., 2004
	9q31–q33	2.07			364	70	344	Majewski et al., 2003
10	10q26	1.52–3.06	3.06–1.90		406	391	860	Weeks et al., 2000
					364	70	344	Majewski et al., 2003
					393	158	490	Seddon et al., 2003
					15	70	133	Kenealy et al., 2004
					383+43	34	381	Iyengar et al., 2004
12	12q13		2.18	0.006	383+43	34	381	Iyengar et al., 2004
	12q22–12q23		2.2	0.0159	345+25	102	258	Schick et al., 2003
	12q23		2.7	0.0019	383+43	34	381	Iyengar et al., 2004
15	15q14		5	2×10^{-7}	383+43	34	381	Iyengar et al., 2004
	15q11.1–15q14			0.0477	345+25	102	258	Schick et al., 2003
	15q25–15q26			0.0444	345+25	102	258	Schick et al., 2003
16	16p12.1		1.92–2.07		345+25	102	258	Schick et al., 2003
					383+43	34	381	Iyengar et al., 2004
17	17q25	3.16			406	428	1,089	Weeks et al., 2000
18	18p11.3		2.48	0.0038	383+43	34	381	Iyengar et al., 2004
20	20q13		2.28	0.0035	383+43	34	381	Iyengar et al., 2004
22	22q12.1	2.03		0.001	773	113	331	Abecasis et al., 2004
	22q12–13		2		393	158	490	Seddon et al., 2003

Table 3. Genetic susceptibility factors associated with AMD in more than 2 case-control studies

Gene	SNP	OR	Locus	p	First reference
CFB	rs641153 ^a	0.32 (0.21–0.48)	6p21.3	6.10 ⁻⁹	Gold et al., 2006
	rs4151667 ^b	0.36 (0.23–0.56)		4.10 ⁻⁶	Gold et al., 2006
C2	rs9332739	0.36 (0.23–0.56)	6p21.3	4.10 ⁻⁶	Gold et al., 2006
	rs547154	0.44 (0.33–0.6)		8.10 ⁻⁸	Gold et al., 2006
CFH	rs1061170	2.8 (2.1–3.8)	1q32	1.6.10 ⁻¹³	Hageman et al., 2005
C3	rs2230199	2.6 (1.6–4.1)	19p13.3–13.2	5.10 ⁻⁵	Yates et al., 2007
ABCA4	rs1800553	5 (1.6–20)	1p22.1	0.0013	Allikmets et al., 2000
	rs1800555	2.8 (1.2–7.4)		0.014	Allikmets et al., 2000
Apo ε4	rs429358 and	0.43 (0.21–0.88)	19q13.2	0.002	Klaver et al., 1998,
	rs7412	0.34 (0.17–0.68)		<0.001	Souied et al., 1998
FBLN5	5 missense variations	–	14q32.1	0.006	Stone et al., 2004
VEGF	rs1413711	2.4 (1.1–5.3)	6p12	0.027	Churchill et al., 2006
TLR3	rs3775291	0.44 (0.23–0.84) ^c	4q35	0.005	Yang et al., 2008

FBLN5 = Fibulin 5; VEGF = vascular endothelial growth factor; TLR3 =toll-like receptor 3.

^a rs641153 is in strong complete LD with rs547154.

^b rs4151667 is in strong LD with 9332739.

^c In Utah case-control series.

Table 4. Polymorphisms of the CFH gene associated with AMD in different Asian populations

Reference	rs1061170 (Y402H)	rs1410996	rs2274700 (A473A)	rs551397 (ISV1- 36C>T)	rs3753394 (-257 T>C)	rs1329428 (IVS15)	rs800292 (I62V)	rs3753396 (Q672Q)
<i>Japanese</i>								
Okamoto et al. 2006	no	—	—	—	—	—	yes	yes
Gotoh et al., 2006	no	—	—	—	—	—	—	—
Uka et al., 2006	no	—	—	—	—	—	—	—
Fuse et al., 2006	no	—	—	—	—	—	no	—
Tanimoto et al., 2007	no	—	—	—	—	—	—	—
Mori et al., 2007	no	yes	yes	—	—	—	yes	—
Goto et al., 2009	—	—	—	—	—	—	yes	—
Hayashi et al., 2010	yes	—	—	—	—	—	yes	—
<i>Chinese</i>								
Lau et al., 2006	yes	—	—	—	—	—	—	—
Chen et al., 2006	no	—	—	—	yes	yes	yes	—
Ng et al., 2008	no	—	yes	yes	yes	yes	yes	yes
Lee et al., 2008*	no	—	no	—	yes	no	yes	—
Xu et al., 2008	no	—	—	—	—	—	—	—
Chu et al., 2008	yes	yes	—	—	—	—	—	—
Cui et al., 2010	—	yes	—	—	—	—	—	—
Liu et al., 2010	yes	no	no	—	yes	yes	yes	—
Dong et al., 2011	yes	—	—	—	yes	yes	—	—

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