Supplementary data:

The molecular genetic basis of age-related macular degeneration: an overview

Saritha Katta, Inderjeet Kaur and Subhabrata Chakrabarti

J. Genet. **88**, 425–449

Table 1. Distribution of epidemiological studies in AMD worldwide.

Population	Sub groups	Location	Study (Time frame)	Participants (N)	Age group (Yr)	All AMD	Early AMD	Intermediate AMD	Advanced AMD	Associated risk factor	*P* value	OR (95%CI)	Reference
American		Baltimore		2520	65–84								Bressler *et al.* 2008
	White		SEE#	1854 (Whites)		3.50%			3.50%	None	0.02		
	Black			666 (Black)		1.40%			1.40%				
American		Maryland	Baltimore Eye Survey (1995–1998)	5308	> 40					large drusen > 125microm	0.02	4.4(1.5–12.4)	Friedman *et al.* 1999
	White			2913		1.91%			1.91%				
	Black			2395		0.19%			0.19				
American		Wisconsin	Beaver Dam Eye Study (1987–1988)	4926	43–75	9.30%	1.90%	7.30%	0.10%	Age	< 0.001		Klein R *et al.* 1992
					< 75	57.20%	24%	26.60%	7.20%	Smoking	< 0.001		
Australian		Victoria	Visual Impairment Project #	5147	> 40	23.96%	15.10%	8.20%	0.66%	gender	< 0.001	N/A	van Newkirk *et al.* 2000
Australian		Sydney	BMES (1991–1993)	3654	> 49				0%				
					< 55				1.90%				
					> 85		13.30%		18.50%				
Hispanic		Arizona	Proyecto VER (1997–1999)	3178	50–59	0.10%	20%						Munoz *et al.* 2005
					> 80	4.30%	54%		0.58%				
Inuit		Denmark	Greenland Inuit Study (2000–2001)	695	> 60	9.50%							Anderson *et al.* 2008

Table 1 (*contd*)

Population	Sub groups	Location	Study (Time frame)	Participants (N)	Age group (Yr)	All AMD	Early AMD	Intermediate AMD	Advanced AMD	Associated risk factor	*P* value	OR (95%CI)	Reference
Icelandic		Iceland	Reykjavik eye study#	1020	> 50								Jonasson *et al.* 2003
Japanese		Japan	Funagata Study (2000–2002)	1625	> 35		3.50%		0.50%	Age		2.27 (1.1–4.67)	Kawasaki *et al.* 2008
										Smoking		5.03 (1.00–25.47)	
American		US	AREDS (1992–1998)	4757	55–80	43%		26.40%		Age		3.12	AREDS Report 3. 2000
										Smoking		1.61	
										Race		4.22	
										Hypertension		1.45	
										BMI		1.43	
										Hyperopia		2.31	
										Lens opacity		1.32	
South Indian		India	APEDS (1996–2000)	3723	> 40	1.82%				Smoking	0	1.65 (0.55–5.01)	Krishnaiah *et al.* 2005
										Age (> 70)	0	3.29 (1.57–7.01)	
										Prior Cataract Surgery	0	2.87 (1.57–5.26)	
South Indian		India	Aravind Comprehensive Eye Study#	4917	> 40	14.70%	2.70%		0.60%	Age		26.91 (2.92–248.13)	Nirmalan *et al.* 2004
North Indian		Indian	INDEYE Study (2002–2003)	1443	> 50	14.90%	34%	10.80%	1.40%				Gupta *et al.* 2007
Dutch		Netherlands	Rotterdam study (1990–1993)	4953	55–64								Klaver *et al.* 2001
					65–74	*0.75%							
					75–84	*3.07%							
					> 85	*8.8%							

*incidence
#time frame undefined

The following table as supplied by the author. Please use zoom facility to view the contents.

Table 2. Distribution of different *CFH* haplotypes in multiple populations worldwide.

Population (Cases, Controls)	5'	Intron 1	Intron 1	Intron 1	exon 2	Intron 4	intron 6	Exon 7	exon9	intron 9	intron 9	intron 9	Intron 9	Intron 9	Exon 10	intron 10	intron 11	intron 11	Exon 13	Intron 14	Intron 15	Intron 15	Intron 15	Intron 15	Exon 16	Exon 18	Intron 18	Intron 18	Haplotype Frequencies			OR (95%CI)	p value	Reference
	rs3753394	rs7524776	rs529825	rs551397	rs800292	rs1329429	rs3766404	rs1061147	rs1061170	rs10801555	rs2019727	rs2019724	1048663	rs1831281	rs2274700	rs203674	rs6677604	rs10489456	rs3753396	rs1410996	rs380390	rs2284664	rs1329428	rs412852	2509 G>A	rs1065489	rs11582939	rs1280514	%Pooled	% Cases	%Controls			
American (549, 272)	C	x	C	x	G	x	T	x	C	x	x	x	x	x	x	T	x	x	A	x	x	x	x	x	x	G	x	x		0.5	0.29	2.46 (1.95 - 3.11)	<0.0001	Hageman et al. 2005
	C	x	T	x	A	x	T	x	T	x	x	x	x	x	x	G	x	x	A	x	x	x	x	x	x	G	x	x		0.12	0.21	0.54 (0.41 - 0.73)	0.00003	
	T	x	C	x	G	x	T	x	T	x	x	x	x	x	x	G	x	x	G	x	x	x	x	x	x	T	x	x				NS		
	C	x	C	x	G	x	C	x	T	x	x	x	x	x	x	G	x	x	A	x	x	x	x	x	x	G	x	x		0.06	0.13	0.48 (0.33 - 0.69)	0.00008	
	T	x	C	x	G	x	T	x	T	x	x	x	x	x	x	G	x	x	A	x	x	x	x	x	x	G	x	x				NS		
American (96, 50)	x	x	x	x	x	x	x	x	x	x	A	x	x	x	x	x	x	C	T	x	C	C	G	x	x	x	x	x	0.59			7.4 (3-19)		Klein et al. 2005
	x	x	x	x	x	x	x	x	x	x	A	x	x	x	x	x	x	C	T	x	G	C	G	x	x	x	x	x	0.0068					
	x	x	x	x	x	x	x	x	x	x	A	x	x	x	x	x	x	C	T	x	G	T	A	x	x	x	x	x	0.12					
	x	x	x	x	x	x	x	x	x	x	A	x	x	x	x	x	x	T	C	x	G	C	G	x	x	x	x	x	0.15					
	x	x	x	x	x	x	x	x	x	x	T	x	x	x	x	x	x	C	T	x	G	C	A	x	x	x	x	x	0.12					
	x	x	x	x	x	x	x	x	x	x	T	x	x	x	x	x	x	C	T	x	G	C	G	x	x	x	x	x	0.0071					
American (210, 183)	x	x	x	x	x	x	x	A	C	x	x	x	x	x	C	x	x	x	x	x	x	x	x	x	x	x	x	x	0.47	0.57	0.36	0.43 (0.29–0.64)	$<10^{-29}$	Francis et al. 2007
	x	x	x	x	x	x	x	C	T	x	x	x	x	x	T	x	x	x	x	x	x	x	x	x	x	x	x	x	0.33	0.2	0.48	3.88 (2.51–6.01)	$<10^{-14}$	
	x	x	x	x	x	x	x	C	T	x	x	x	x	x	C	x	x	x	x	x	x	x	x	x	x	x	x	x	0.2	0.23	0.16	0.62 (0.38–1.03)	0.28	
American (2201)	x	x	x	x	x	x	x	A	C	x	x	x	x	x	C	x	x	x	x	x	x	x	x	x	x	x	x	x	0.44	0.58	0.35	0.40 (0.30–0.52)	<0.0001	
	x	x	x	x	x	x	x	C	T	x	x	x	x	x	T	x	x	x	x	x	x	x	x	x	x	x	x	x	0.36	0.21	0.46	3.08 (2.30–4.13)	<0.0001	
	x	x	x	x	x	x	x	C	T	x	x	x	x	x	C	x	x	x	x	x	x	x	x	x	x	x	x	x	0.2	0.21	0.19	0.90 (0.64–1.27)	0.77	
American (584, 248)	C	x	A	x	A	x	T	x	T	x	x	x	x	x	x	T	x	x	A	x	x	x	x	x	x	G	x	x		0.125	0.204		0.021	Spencer et al. 2007
	C	x	G	x	G	x	C	x	T	x	x	x	x	x	x	T	x	x	A	x	x	x	x	x	x	G	x	x		0.054	0.084		0.018	
	C	x	G	x	G	x	T	x	C	x	x	x	x	x	x	G	x	x	A	x	x	x	x	x	x	G	x	x		0.512	0.376		0.041	
	T	x	G	x	G	x	T	x	T	x	x	x	x	x	x	T	x	x	G	x	x	x	x	x	x	T	x	x		0.138	0.135			
Indian (100,120)	C	x	x	x	G	x	T	x	C	x	x	x	x	x	x	x	x	x	A	x	x	x	x	x	x	G	x	x		0.308	0.154	NA	0.0003	Kaur *et al.* 2006
	C	x	x	x	A	x	T	x	T	x	x	x	x	x	x	x	x	x	A	x	x	x	x	x	x	G	x	x		0.076	0.148	NA	0.0238	
	C	x	x	x	G	x	T	x	T	x	x	x	x	x	x	x	x	x	A	x	x	x	x	x	x	G	x	x		0.064	0.119	NA	0.0583	
	C	x	x	x	A	x	C	x	T	x	x	x	x	x	x	x	x	x	A	x	x	x	x	x	x	G	x	x		0.048	0.113	NA	0.0161	
	T	x	x	x	G	x	T	x	T	x	x	x	x	x	x	x	x	x	A	x	x	x	x	x	x	G	x	x		0.053	0.092	NA	0.136	
	C	x	x	x	G	x	C	x	T	x	x	x	x	x	x	x	x	x	A	x	x	x	x	x	x	G	x	x		0.049	0.06	NA	0.6192	
	T	x	x	x	G	x	T	x	T	x	x	x	x	x	x	x	x	x	G	x	x	x	x	x	x	T	x	x		0.055	0.052	NA	0.8946	
	T	x	x	x	G	x	T	x	C	x	x	x	x	x	x	x	x	x	A	x	x	x	x	x	x	G	x	x		0.075	0.032	NA	0.06	
	T	x	x	x	A	x	T	x	T	x	x	x	x	x	x	x	x	x	A	x	x	x	x	x	x	G	x	x		0.035	0.024	NA	0.5313	
	T	x	x	x	G	x	T	x	T	x	x	x	x	x	x	x	x	x	A	x	x	x	x	x	x	T	x	x		0.048	0.047	NA	0.956	
	T	x	x	x	G	x	C	x	T	x	x	x	x	x	x	x	x	x	A	x	x	x	x	x	x	G	x	x		0.013	0.033	NA	0.1889	
	T	x	x	x	G	x	T	x	C	x	x	x	x	x	x	x	x	x	A	x	x	x	x	x	x	T	x	x		0.031	0.014	NA	0.239	
	C	x	x	x	G	x	T	x	C	x	x	x	x	x	x	x	x	x	G	x	x	x	x	x	x	T	x	x		0.03	0.005	NA	0.0549	
	C	x	x	x	A	x	T	x	C	x	x	x	x	x	x	x	x	x	A	x	x	x	x	x	x	G	x	x		0.019	0.012	NA	0.5852	
	C	x	x	x	A	x	C	x	C	x	x	x	x	x	x	x	x	x	A	x	x	x	x	x	x	G	x	x		0.006	0.023	NA	0.1475	
	T	x	x	x	A	x	T	x	C	x	x	x	x	x	x	x	x	x	A	x	x	x	x	x	x	G	x	x		0.014	0.01	NA	0.6566	
Japanese (96, 89)	x	x	x	x	G	x	T	x	T	x	x	x	x	x	x	x	x	x	G	x	x	x	x	x	x	x	x	x		0.38	0.38		1	Okamoto *et al.* 2006
	x	x	x	x	A	x	T	x	T	x	x	x	x	x	x	x	x	x	A	x	x	x	x	x	x	x	x	x		0.18	0.35	1.6 (0.26 - 0.67)	0.001	
	x	x	x	x	G	x	T	x	T	x	x	x	x	x	x	x	x	x	A	x	x	x	x	x	x	x	x	x		0.19	0.1	1.9 (1.12 - 3.69)	0.028	
	x	x	x	x	A	x	T	x	T	x	x	x	x	x	x	x	x	x	C	x	x	x	x	x	x	x	x	x		0.15	0.06	2.5 (1.42 - 6.38)	0.004	
	x	x	x	x	G	x	T	x	C	x	x	x	x	x	x	x	x	x	A	x	x	x	x	x	x	x	x	x		0.04	0.04		0.802	
Japanese (188, 139)	x	x	x	x	G	x	x	x	T	x	x	x	x	x	C	x	x	x	x	C	x	x	x	x	x	x	x	x		0.566	0.437	NA	1.12 x 10-3	Mori *et al.* 2007
	x	x	x	x	A	x	x	x	T	x	x	x	x	x	T	x	x	x	A	T	x	x	x	x	x	x	x	x		0.254	0.416	NA	1.28 x 10- 5	
	x	x	x	x	G	x	x	x	T	x	x	x	x	x	T	x	x	x	A	T	x	x	x	x	x	x	x	x		0.078	0.077	NA	0.959	
	x	x	x	x	G	x	x	x	C	x	x	x	x	x	C	x	x	x	A	C	x	x	x	x	x	x	x	x		0.087	0.047	NA	0.046	
Chinese (163,244)	T	x	x	x	G	C	x	x	T	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	0.56	N/A	N/A	1.68 (1.26 - 2.23)	0.0003	Chen *et al.* 2006
Chinese (163,155)	x	x	x	C	G	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x		0.752	0.613	1.91 (1.36-2.68)	0.0001	Ng *et al.* 2008
	x	x	x	T	A	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x		0.248	0.387	0.52 (0.37-0.74)	0.0001	
	x	x	x	x	x	x	x	x	x	x	x	x	x	x	A	x	x	x	A	x	x	x	T	x	x	x	x	x		0.264	0.387	0.57 (0.41-0.79)	0.0024	
	x	x	x	x	x	x	x	x	x	x	x	x	x	x	G	x	x	x	G	x	x	x	C	x	x	x	x	x		0.659	0.558	1.53 (1.11-2.11)	0.03	
	C	x	x	T	A	x	x	x	x	x	x	x	x	x	A	x	x	x	A	x	x	x	T	x	x	x	x	x		0.204	0.316	0.56 (0.39-0.80)	0.0052	
	T	x	x	C	G	x	x	x	x	x	x	x	x	x	G	x	x	x	G	x	x	x	C	x	x	x	x	x		0.624	0.505	1.63 (1.19-2.23)	0.0087	
	C	x	x	T	A	x	x	x	x	x	x	x	x	x	A	x	x	x	A	x	x	x	T	x	x	G	x	x		0.182	0.3	0.52 (0.36-0.76)	0.0012	
	T	x	x	C	G	x	x	x	x	x	x	x	x	x	G	x	x	x	G	x	x	x	C	x	x	G	x	x		0.056	0.017	3.57 (1.31-9.72)	0.052	
	T	x	x	C	G	x	x	x	x	x	x	x	x	x	G	x	x	x	G	x	x	x	C	x	x	T	x	x		0.568	0.487	1.38 (1.01-1.89)	0.177	
	x	x	x	x	A	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	G	x	x		0.208	0.336	0.52 (0.37-0.75)	0.0011	
	x	x	x	x	G	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	T	x	x		0.612	0.507	1.55 (1.13-2.12)	0.03	
	x	x	x	x	A	x	x	x	T	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x		0.248	0.387	0.52 (0.37-0.74)	0.0004	
	x	x	x	x	G	x	x	x	T	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x		0.693	0.584	1.61 (1.16-2.23)	0.014	
	x	x	x	x	G	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	C	x	x	x		0.751	0.609	1.94 (1.38-2.72)	0.0004	
	x	x	x	x	A	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	C	x	x	x		0.243	0.365	0.56 (0.40-0.79)	0.0015	
American (780, 265)	C	x	G	x	G	x	T	x	C	x	x	T	x	G	G	G	G	x	A	x	x	x	x	x	x	G	x	x		0.55	0.4	NA	<0.001	Spencer *et al.* 2008
	C	x	A	x	A	x	T	x	T	x	x	C	x	A	A	T	G	x	A	x	x	x	x	x	x	G	x	x		0.12	0.21	NA	<0.001	
	T	x	G	x	G	x	T	x	T	x	x	C	x	G	G	T	G	x	G	x	x	x	x	x	x	T	x	x		0.13	0.13	NA	0.7	
	C	x	G	x	G	x	C	x	T	x	x	C	x	G	A	T	A	x	A	x	x	x	x	x	x	G	x	x		0.05	0.07	NA	0.1	
	T	x	G	x	G	x	T	x	C	x	x	T	x	G	G	G	G	x	A	x	x	x	x	x	x	G	x	x						
	T	x	G	x	G	x	T	x	T	x	x	C	x	G	G	G	G	x	A	x	x	x	x	x	x	G	x	x						
	T	x	G	x	G	x	T	x	T	x	x	C	x	G	A	T	A	x	A	x	x	x	x	x	x	G	x	x						
																																Haplotype trend regression		
American (544, 268)	x	x	x	x	x	x	x	x	x	x	x	x	G	x	C	x	x	x	x	x	x	x	x	C	x	x	C	A		0.5797	0.3786	NA	<0.0001	Li M *et al.* 2006
	x	x	x	x	x	x	x	x	x	x	x	x	G	x	T	x	x	x	x	x	x	x	x	T	x	x	C	A		0.1403	0.4391	NA	<0.0001	
	x	x	x	x	x	x	x	x	x	x	x	x	A	x	C	x	x	x	x	x	x	x	x	T	x	x	T	A		0.0856	0.0633	NA	<0.0001	
	x	x	x	x	x	x	x	x	x	x	x	x	A	x	C	x	x	x	x	x	x	x	x	T	x	x	T	G		0.0318	0.1062	NA	<0.0001	
	x	x	x	x	x	x	x	x	x	x	x	x	G	x	C	x	x	x	x	x	x	x	x	T	x	x	C	A		0.0393	0.0028	NA		
	x	x	x	x	x	x	x	x	x	x	x	x	G	x	C	x	x	x	x	x	x	x	x	C	x	x	T	A		0.0309	0.0016	NA		
	x	x	x	x	x	x	x	x	x	x	x	x	A	x	C	x	x	x	x	x	x	x	x	T	x	x	C	A		0.0298	0.001	NA	<0.0001	
	x	x	x	x	x	x	x	x	x	x	x	x	G	x	T	x	x	x	x	x	x	x	x	C	x	x	C	A		0.0254	0.0009	NA		
British (173,170)	x	T	C	x	G	A	x	A	C	A	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x		0.55	0.38	1.99	8.47 x 10–6	Hughes *et al. 2006*
	x	T	C	x	G	C	x	C	T	G	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x		0.33	0.39	0.8	0.15	
	x	C	T	x	A	C	x	C	T	G	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x		0.08	0.14	0.54	0.012	
	x	T	T	x	A	C	x	C	T	G	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x	x		0.02	0.08	0.29	0.0007	

Table 3. Association studies in *CFH*-related genes haplotypes in different Caucasian populations.

Gene	Population	SNP	Cases	Controls	*P* Value	Odds Ratio (95%CI)	Reference
CFHR1	British	rs408519	173	170	2.26×10^{-6}	0.48(0.35–0.65)	Hughes *et al*. 2006
	American	rs4230	224	133	0.07	0.73(0.53–1.02)	Edwards *et al*. 2005
	American	rs2133138	1238	934	1.08×10^{-19}	n/a	Maller *et al*. 2006
CFHR4	American	rs12404243	495	185	n/a	n/a	Haines *et al*. 2005
	American	rs7417769	224	130	0.01	1.59(1.13–2.23)	Edwards *et al*. 2005
	American	rs10922147	1238	934	9.3×10^{-22}	n/a	Maller *et al*. 2006
	American	rs1853883	222	133	5×10^{-6}	2.04(1.5–2.78)	Edwards *et al*. 2005
	American		1238	934	2.8×10^{-46}	n/a	Maller *et al*. 2006
	American	rs1971579	213	124	0.03	1.48(1–2.12)	Edwards *et al*. 2005
	American		1238	934	6.37×10^{-16}	n/a	Maller *et al*. 2006
CFHR2	American	rs4915318	224	133	0	1.77(1.22–2.56)	Edwards *et al*. 2005
		rs3790414	224	132	0	1.77(1.22–2.58)	
	American	rs9427934	1238	934	2.82×10^{-17}	n/a	Maller *et al*. 2006
	American	rs7531555	216	129	0.01	0.58(0.40–0.85)	Edwards *et al*. 2005
	American	rs7548145	1238	934	1.84×10^{-11}	n/a	Maller *et al*. 2006
CFHR3	American	rs385390	1238	934	0.05	n/a	Maller *et al*. 2006
		rs445207			4.46×10^{-6}	n/a	
CFHR5	American	rs6428379	220	132	3.8×10^{-5}	1.9(1.4–2.59)	Edwards *et al*. 2005
	American		495	185	0	n/a	Haines *et al*. 2005
	American	rs6669207	1238	934	1.16×10^{-5}	n/a	Maller *et al*. 2006
		rs6667243			9.61×10^{-43}	n/a	
		rs3748557			2.09×10^{-6}	n/a	
		rs10922153			4.57×10^{-33}	n/a	

NA, data not avaiable.