

Ethnic specific association of the *CAVI/CAV2* locus with primary open-angle glaucoma

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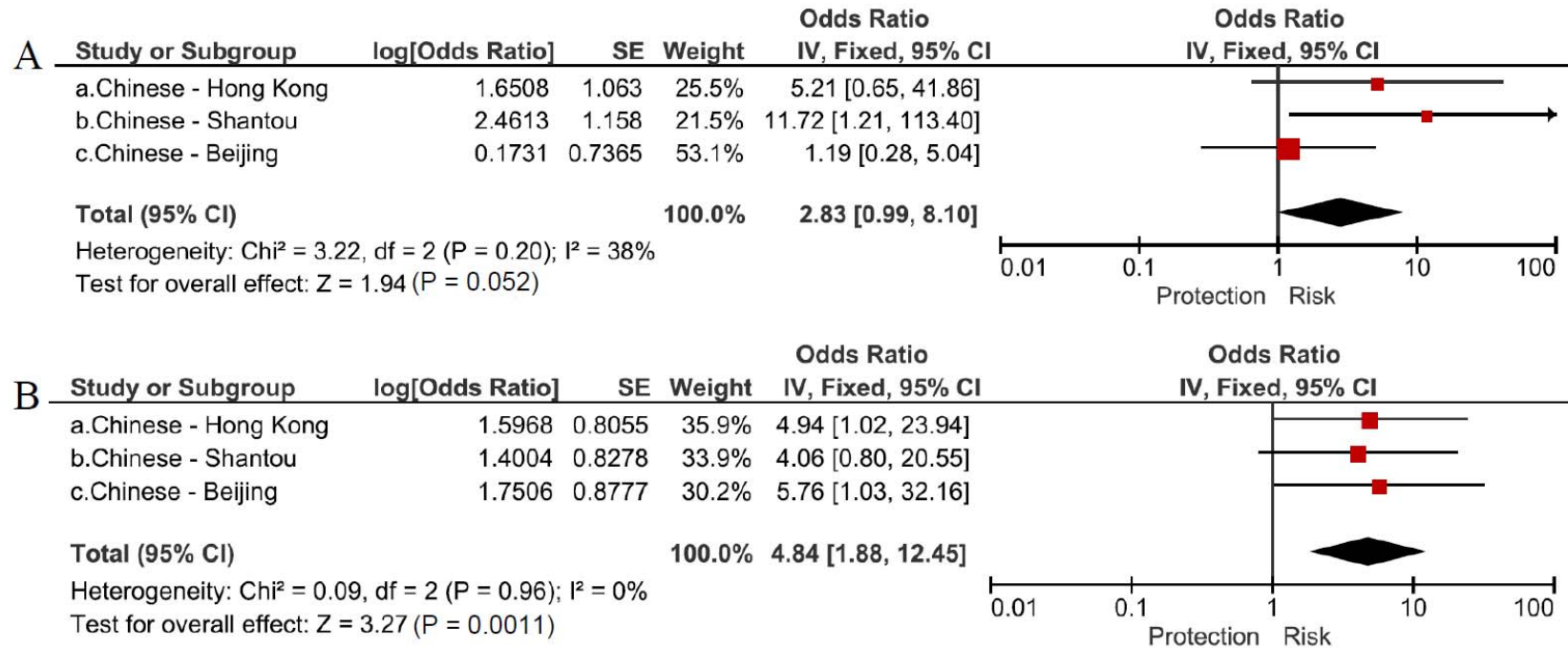
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Supplementary Fig S1. Association of rs4236601 with primary open-angle glaucoma in male and female subjects

A. Meta-analysis performed in Chinese male subjects; B. Meta-analysis performed in Chinese female subjects

Supplementary Table S1. Quality control results

No.	SNP	Chr: position ^a	Genotype call rate in all samples	Test for HWE in controls (P)
Chinese - Hong Kong			n=881	
1	rs1052990	7: 116148370	0.980	0.35
2	rs6466579	7: 116151155	0.994	0.87
3	rs7801950	7: 116154783	0.995	0.17
4	rs4236601	7: 116162729	0.988	1.00
5	rs3779512	7: 116171063	0.992	0.061
6	rs3807989	7: 116186241	0.995	0.78
7	rs3801994	7: 116190469	0.994	0.45
8	rs1049337	7: 116200587	0.989	1.00
Chinese – Shantou			n=854	
1	rs4236601	7: 116162729	0.984	1.00
2	rs3801994	7: 116190469	0.999	0.19
Chinese – Beijing			n=370	
1	rs4236601	7: 116162729	1.00	1.00
2	rs3801994	7: 116190469	1.00	0.64
Japanese – Osaka			n=308	
1	rs1052990	7: 116148370	0.996	0.85
2	rs4236601	7: 116162729	1.00	Monomorphic
3	rs3801994	7: 116190469	1.00	0.87

^a The position was aligned to GRCh37/hg19.

Chr: chromosome; HWE: Hardy-Weinberg equilibrium; SNP: single-nucleotide polymorphism.

Supplementary Table S2. Association of rs4236601 with primary open-angle glaucoma conditioned on the other single nucleotide polymorphisms

No.	Adjusted SNP	MA	POAG vs. controls	
			P	OR (95% CI)
1	rs1052990	G	0.021	4.43 (1.25-15.66)
2	rs6466579	T	0.014	4.80 (1.38-16.76)
3	rs7801950	T	0.010	5.13 (1.47-17.86)
4	rs4236601	A	n.a.	n.a.
5	rs3779512	T	0.0080	5.62 (1.57-20.16)
6	rs3807989	A	0.016	4.62 (1.33-16.13)
7	rs3801994	A	0.0088	5.31 (1.52-18.50)
8	rs1049337	C	0.011	5.08 (1.46-17.72)

CI: confidence interval; MA: minor allele; n.a.: not applicable; OR: odds ratio; POAG: primary open-angle glaucoma; SNP: single-nucleotide polymorphism

Supplementary Table S3. Association of rs3809916 with primary open-angle glaucoma conditioned on the other single nucleotide polymorphisms

No.	Adjusted SNP	MA	POAG vs. controls	
			P	OR (95% CI)
1	rs1052990	G	0.10	1.31 (0.95-1.79)
2	rs6466579	T	0.035	1.36 (1.02-1.80)
3	rs7801950	T	0.091	1.31 (0.96-1.80)
4	rs4236601	A	0.038	1.22 (1.00-1.61)
5	rs3779512	T	0.055	1.29 (0.99-1.67)
6	rs3807989	A	0.159	1.32 (0.90-1.94)
7	rs3801994	A	n.a.	n.a.
8	rs1049337	C	0.038	1.31 (1.02-1.70)

CI: confidence interval; MA: minor allele; n.a.: not applicable; OR: odds ratio; POAG: primary open-angle glaucoma; SNP: single-nucleotide polymorphism

Supplementary Table S4. Selection of SNPs to be tested in the *CAVI/CAV2* locus

No.	SNP	NCBI36/hg18	Distance to a gene (bp)	MA	MAF in CHB	Reason for selection	Reference
1	rs1052990	c.*2195T>G, downstream of <i>CAV2</i>	2,677	G	0.173	POAG GWAS	1
2	rs6466579	g.115938641T>C, downstream of <i>CAV2</i>	2,810	T	0.278	Tag SNP	-
3	rs7801950	g.115942269C>T, downstream of <i>CAV2</i>	6,438	T	0.189	Tag SNP	-
4	rs4236601	g.115949965G>A, upstream of <i>CAVI</i>	2,110	A	0.017	POAG GWAS	1
5	rs3779512	<i>CAVI</i> c.102+4320T>G	Intron 2	T	0.100	Tag SNP	-
6	rs3807989	<i>CAVI</i> c.103-12759A>G	Intron 2	A	0.289	Tag SNP; ECG P-R interval & atrial fibrillation	2
7	rs3801994	<i>CAVI</i> c.103-8531G>A	Intron 2	A	0.189	Tag SNP	-
8	rs1049337	<i>CAVI</i> c.*1246C>T	1,246	T	0.477	Tag SNP	-

CAVI: caveolin 1; *CAV2*: caveolin 2; CHB: Chinese Han Beijing; ECG: electrocardiogram; MA: minor allele; MAF: minor allele frequency; NCBI: National Center for Biotechnology Information; SNP: single nucleotide polymorphism; GWAS: genome-wide association study

Reference

- 1 Thorleifsson, G. *et al.* Common variants near CAV1 and CAV2 are associated with primary open-angle glaucoma. *Nat Genet* **42**, 906-909, (2010).
- 2 Holm, H. *et al.* Several common variants modulate heart rate, PR interval and QRS duration. *Nat Genet* **42**, 117-122, (2010).