

Evaluation of Genetic Association of the INK4 Locus with Primary Open Angle Glaucoma in East Indian Population

Mansi Vishal^{1,2}, Anchal Sharma^{2,3}, Lalit Kaurani², Subhadip Chakraborty¹, Jharna Ray⁴,
Abhijit Sen⁵, Arijit Mukhopadhyay^{2,3}, Kunal Ray^{1,3}

¹Molecular and Human Genetics Division, CSIR-Indian Institute of Chemical Biology, Kolkata, India; ²Genomics & Molecular Medicine, CSIR-Institute of Genomics & Integrative Biology, Delhi, India; ³Academy of Scientific and Innovative Research (AcSIR), New Delhi, India; ⁴S.N. Pradhan Centre for Neurosciences, University of Calcutta, Kolkata, India; ⁵Dristi Pradip, Kolkata, India

Supplementary Table S1: Studies done on the association of 9p21 region SNPs with POAG and its subtypes

Population ancestry	Top associated SNP from 9p21 region (minor allele)	p- value/OR	Type of study	POAG subtype	Reference
African	rs10120688 (A), rs10965245 (A)	0.002/1.21, 0.0005/0.73	Case-control	POAG, NTG	<i>Liu et al, IOVS 2013 [1]</i>
European	rs2157719 (G)	1.86E ⁻¹⁸ /0.69	Meta-analysis	POAG	<i>Wiggs et al, PLoS One 2013 [2]</i>
European	rs1063192 (G)	2.2*10 ⁻⁵ /0.74	Case-control	POAG	<i>Dimasi et al, Am J Ophthalmol 2012 [3]</i>
Japanese	rs523096 (A)	1.59*10 ⁻⁵ /1.99	GWAS	NTG	<i>Takamoto et al, PLoS One 2012 [4]</i>
African	rs1063192 (C)	0.0008/0.39	Case-control	POAG	<i>Cao D et al, PLoS One 2012 [5]</i>
European	rs10120688	1.2x10 ⁻⁵ /NA	GWAS	POAG	<i>Gibson et al, MolVis 2012 [6]</i>
Japanese	rs1063192 (C)	0.0023/NA	Case-control	NTG	<i>Mabuchi et al, Ophthalmology 2012 [7]</i>
European	rs10120688 (A)	0.005/1.25	Case-control	Advanced POAG, HTG, NTG	<i>Burdon et al, Ophthalmology 2012 [8]</i>
Japanese	rs523096 (A)	3.8*10 ⁻⁹ /1.86	GWAS	POAG, NPG, HPG	<i>Nakano et al, PLoS One 2012 [9]</i>
Japanese	rs1063192 (C)	5.68*10 ⁻⁵ /0.79	GWAS	POAG	<i>Osman et al, Hum Mol Genet 2012 [10]</i>
European	rs1063192 (G)	1.41*10 ⁻⁸ /0.76	Meta-analysis	POAG	<i>Ramdas et al, Hum Mol Genet 2011 [11]</i>
European	rs1063192 (G)	0.0045/0.73	Case-control	VCDR, POAG	<i>Fan et al, IOVS 2011 [12]</i>
European	rs4977756 (A)	4.7*10 ⁻⁹ /1.5	GWAS	POAG	<i>Burdon et al, Nat Genet 2011 [13]</i>
European	rs1063192 (G)	6.15*10 ⁻¹¹ /NA	GWAS	VCDR	<i>Ramdas et al, PLoS Genet 2010 [14]</i>

Abbreviations: POAG- Primary Open Angle Glaucoma, NTG- Normal Tension Glaucoma, HTG- High Tension Glaucoma, HPG- High Pressure Glaucoma, NPG- Normal Pressure Glaucoma, VCDR- Vertical Cup-disc ratio, NA-Not available.

Note: Abbreviations used under 'POAG subtype' column are same as described in the original study. Numbers in parenthesis used in the last column denote the reference numbers.

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Supplementary Table S2: Haplotype association using 20 tag SNPs in 9p21 region

Haplotype	Frequency in cases (n=700)	Frequency in controls (n=708)	p-value
AAAGAGCAGAAGGAAAAACG*	0.005	0.017	0.0381
CAGGAAAAGAAGAAAAAGCA	0.024	0.043	0.0597
CAGGAAAAGAGGACAAAACG	0.335	0.288	0.0614
AAAGAGCAGAAGGAAAAGCA	0.008	0.0200	0.0633
AAAGAGCAGAAGACAAAACG	0.016	0.006	0.0783
CAGGAAAAGAGGACAAAACA	0.016	0.023	0.3866
CAGAAAAAGAAGAAAAAGCA	0.078	0.067	0.4198
CAGGAAAGGAAGGCAGAGCA	0.011	0.016	0.4555
AAGGCGAAGAAGGCGAAAAA	0.06	0.070	0.4694
AAGGCAAAGGGGACAACACG	0.013	0.010	0.5949
CGGGAAAGGAAGGCAGAGCA	0.171	0.161	0.6284
CGGGAAAGGAAGGCAGAGCG	0.011	0.010	0.8504
CAGAAAAAGAAGAAAAACG	0.085	0.085	0.9707

* Haplotype with significant p-value

