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Genetic analysis localizes a novel locus on chromosome 4q for the glaucoma endophenotype, cup-to-disc ratio: The Jiri Eye Study

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**Genetic analysis localizes a novel locus on chromosome 4q for the glaucoma endophenotype, cup-to-disc ratio: The Jiri Eye Study**

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### PURPOSE

- Endophenotypes are heritable, quantitative traits genetically correlated to disease risk.
- They can offer a better understanding of underlying disease mechanisms, and provide increased statistical power to localize and identify disease-related genes.
- Intraocular pressure (IOP), vertical cup-to-disc ratio (VCDR) and central corneal thickness (CCT) are three well-established primary open angle glaucoma (POAG) endophenotypes, which are influenced by genetic factors.
- The aim of this study was to investigate the genetics of these known POAG endophenotypes in a population from Nepal.

### METHODS

- This is a family-based study design utilizing data from the Jirel people, a small genetically isolated population from the Jiri region of eastern Nepal.¹
- Quantitative measures of IOP, CCT, and VCDR were obtained by Goldmann applanation tonometry (AT900, Haag Streit International, Switzerland), optical coherence tomography (Topcon, Singapore), and slit-lamp biomicroscopy (BD900, Haag Streit International), respectively.
- POAG diagnoses were in accordance with ISGEO criteria.²
- A variance components approach was used to estimate trait additive genetic heritability ($h^2$).
- Genome-wide genotypes (Illumina 660W-Quad) were used to conduct classical linkage mapping to localize QTLs.
- A measured genotype approach was employed to test SNV associations within the QTLs.
- All genetic analyses were performed in SOLAR.³

### RESULTS

- All 1,800 individuals (55.2% female) examined in this study belong to a single extended pedigree.
- The mean (SD) age at exam is 42.4 (16.6) years.
- There are 35 (1.95%) POAG cases, of which, 10 (28.6%) were high-tension glaucoma (HTG) and 25 (71.4%) were normal-tension glaucoma (NTG).
- The distribution of POAG endophenotypes, which are significantly heritable, are presented in Table 1.

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### REFERENCES


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### DISCLOSURES

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