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Genome-wide analysis identifies common CNVs associated with primary open angle glaucoma

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Background

Copy number variation (CNV) is one of the major factors contributing to genomic diversity and diseases. Glaucoma is a major neurodegenerative disease causing irreversible vision loss across the globe. We wanted to analyze the impact of common CNVs in a genome-wide scale in patients of primary open angle glaucoma (POAG) collected from the West Bengal, India.

Method

Genome-wide data was generated on 364 POAG cases and 365 controls on Illumina 660W-Quad arrays and CNVs were called using PennCNV. Copy number variant regions (CNVRs) were analyzed for association. A publicly available dataset of POAG cohort of 866 cases and 495 controls from Caucasian origin (GLAUGEN study) was used as a validation cohort. Representative CNVs were validated using real-time PCR.

Results

We analyzed genome-wide CNV from 1928 samples. After association analysis we found 308 significantly associated ($p<0.05$) CNVRs in the Indian data. These POAG associated CNVRs were enriched in nervous system development. 113 CNVRs (37%) were significantly associated with the Caucasian data set. These contain 5 genes previously reported in eye diseases, namely, *IDUA*, *FOXE3*, *NDUF7*, *PRPF6* and *WNT3*. We also found 6 associated CNVRs in previously known glaucoma loci.

Conclusion

We have shown that common CNVRs are significantly associated in both datasets irrespective of the population background. We have also identified candidate genes/regions which are uniquely present in POAG cases and absent in controls. Our data might provide new insights into role of CNV in pathogenesis of POAG.

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