

Attitudes towards polygenic risk testing in individuals with glaucoma

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PURPOSE: Glaucoma is the leading cause of irreversible blindness worldwide however, vision loss from glaucoma can generally be prevented through early identification and timely implementation of treatment. Recently, polygenic risk scores (PRS) have shown promise in stratifying individual risk and prognostication for primary open-angle glaucoma to reduce disease burden. Integrating PRS testing into clinical practice is becoming an increasingly realistic prospect, however, little is known about the attitudes of patients towards such testing.

DESIGN: Cross-sectional, questionnaire-based study.

PARTICIPANTS: 2369 participants were invited to participate from the Australian and New Zealand Registry of Advanced Glaucoma (ANZRAG) , who fit the inclusion criteria of adults with a diagnosis of POAG, had not received genetic results that explain their condition, were not known to be deceased, resided in Australia and had agreed to receive correspondence.

METHODS: 1169 individuals (response rate 49%) with primary open-angle glaucoma completed the survey evaluating their attitudes towards polygenic risk testing for glaucoma.

MAIN OUTCOME MEASURES: Sociodemographic, health, perception, and emotional factors were examined to assess associations with interest in PRS testing. Interest in PRS testing was evaluated through assessing likelihood to take the test to predict personal risk of disease and disease severity, and whether the individual would recommend the test to family or non-family members.

RESULTS: Our results show strong interest in the test, with 69.4% of individuals (798 of 1150) indicating a keenness in testing prior to diagnosis, had it been available. In particular, interest was seen in those from an urban area (OR 1.70, 95%CI (1.15-2.49) , $p=0.007$) , those who perceived their risk of developing glaucoma as higher (OR 2.05, 95%CI (1.28-3.29) , $p=0.003$) , and those who were worried about developing glaucoma (OR 2.07, 95%CI (1.27-3.37) , $p=0.004$) . People who were interested in testing were more likely to change their eye health-seeking intentions and recommend testing to family and non-family members, as well as undergo testing for prognostication.

CONCLUSIONS: These findings will help to facilitate the clinical implementation of PRS testing for glaucoma to reduce irreversible vision loss.

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