Enhancing Glaucoma Detection: Integrating Deep Learning with Color Fundus Photos and Polygenic Risk Score

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**Purpose**
Early detection of glaucoma is critical in preventing irreversible vision loss. This study evaluates the efficacy of combining deep learning (DL) analysis of color fundus photos (CFPs) with a polygenic risk score (PRS) in improving glaucoma detection.

**Methods**
We developed DL models for glaucoma classification using over 4,000 glaucomatous and 100,000 normal CFPs, employing the ConvNeXt architecture pre-trained on ImageNet 1K. For the PRS, we utilized cross-ancestry meta-analysis GWAS summary statistics, excluding the UK Biobank (UKB) cohort, and applied the C+T method to derive a weighted PRS for primary open-angle glaucoma (POAG). Our DL models and PRS were evaluated on a subset of 270 POAG cases and 18,642 healthy controls from the UKB, all with available CFPs and genetic data. The performance was assessed using logistic regression and the area under the receiver operating characteristic curve (AUC).

**Results**
Both the DL score from CFPs and the PRS showed significant associations with POAG, with p-values of $8.76 \times 10^{-153}$ and $9.78 \times 10^{-8}$, respectively, after adjusting for age, sex, and the top four principal components of genetic ancestry. The AUC was 0.963 (95% CI: 0.952 - 0.974). Utilizing the Youden index to determine the optimal cutpoint, we achieved a sensitivity of 0.856 and a specificity of 0.945.

**Conclusions**
The study demonstrates that the DL score from CFPs, combined with PRS, significantly enhances the detection of glaucoma. This integrated approach of DL and genetic risk scoring presents a promising tool for glaucoma screening, potentially facilitating earlier intervention and better patient outcomes.

**Layman Abstract (optional): Provide a 50-200 word description of your work that non-scientists can understand. Describe the big picture and the implications of your findings, not the study itself and the associated details.**
Glaucoma is a serious eye condition that can cause blindness if not caught early. Imagine having a powerful computer program that can look at pictures of the eye and help doctors spot glaucoma sooner. We've trained a computer system to

recognize signs of glaucoma by showing it over a hundred thousand of eye photos—some healthy, some not. We also added a special score that helps predict someone's risk of glaucoma based on their genes. When we tested our system, it was very good at picking out the individuals with glaucoma. This means that in the future, our approach could help doctors find glaucoma early, when it's easier to treat. This could be a big step forward in preventing vision loss from this sneaky eye disease.