CLSA Approved Project

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Project Title
Enabling precision management of ophthalmic disease through discovery of underlying biological pathways and clinical prediction tool development

Project Summary
Discovering the influence of genetic variation on phenotypes (i.e., traits or disease susceptibility) requires a cohort of individuals with both genetic information and accurate phenotype values, but accurately phenotyping large cohorts is a substantial challenge. Recent advances in deep learning enable the development of models to predict phenotypes from high-dimensional quantitative input data like medical imaging. In our proposed study, we will evaluate the generalizability of validated models trained on retinal fundus images to predict the severity and progression of glaucoma, age-related macular degeneration, and diabetic retinopathy in the CLSA dataset and discover genetic variants associated with these diseases. We will also explore the use of an alternative quantitative data modality, spirometry, for prediction and genetic discovery of chronic obstructive pulmonary disease.

Keywords
Machine learning, Glaucoma, Age-related macular degeneration, Diabetic retinopathy, Spirometry