

ABSTRACT
A DEEP LEARNING AND CAUSAL MODELING SYSTEM FOR
RETINAL DISEASE DIAGNOSIS

Shiyu Tian, B.A., M.A.

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Vision-threatening retinal diseases, such as diabetic retinopathy (DR) and diabetic macular edema (DME), are major causes of preventable blindness worldwide. Early screening and follow-up are essential, but advanced imaging like Optical Coherence Tomography (OCT) is expensive and not always available outside large clinics. In contrast, color fundus photography is inexpensive and common, yet it only shows a surface view and can miss subtle changes inside the retina. These practical limits make it hard to deliver timely, high-quality eye care in many settings.

As AI is increasingly used in healthcare, retinal disease diagnosis has benefited as well. This work combines deep learning with causal modeling to improve retinal disease diagnosis. The system learns from both fundus photos and OCT scans, expands access by generating OCT-like views from fundus images when OCT is unavailable, and converts images into clear, quantitative biomarkers by segmenting retinal layers and fluid. These biomarkers are then analyzed with simple, well-stated causal assumptions to separate signals likely driven by disease from those influenced by confounders, which helps the model stay reliable, interpretable, and fair across settings.

This dissertation focuses on developing a unified deep learning and causal modeling system for retinal disease diagnosis. The system integrates screening models for fundus and OCT, a fundus-to-OCT translation module to recover cross-sectional structure from widely available photographs, and a biomarker pipeline with layer and fluid segmentation followed by causal analysis to estimate the effect of structure on disease. We evaluate the framework on public datasets and a lab cohort under appropriate approvals, showing strong image quality for the synthesized OCT and stable diagnostic performance across settings.