YOU ARE INVITED TO ATTEND
A SPECIAL SEMINAR ON
INVESTIGATING THE GENETIC BASIS OF INHERITED OCULAR DYSTROPHIES

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FRIDAY, APRIL 22, 12:45 P.M.
LUNCH WILL BE SERVED AT 12:00 P.M. IN THE LOBBY

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Investigating the genetic basis of inherited ocular dystrophies

Inherited diseases, although extremely unfortunate, provide geneticists with a valuable resource to identify proteins and pathways essential for the normal physiological functioning of the tissue of interest. Inherited ocular dystrophies are the leading cause of blindness worldwide. In an ongoing effort to elucidate the genetic factors associated with inherited ocular dystrophies, we have ascertained >500 inbred familial cases, each with multiple affected individuals. In particular, we have ascertained cases of congenital cataracts, primary congenital glaucoma, stationary night blindness, and Retinitis pigmentosa, among others.

The genomic DNA of members from these familial cases are subjected to a systematic investigation that begins with genome-wide linkage analysis to localize the disease phenotype to a particular locus on the human genome, followed by DNA sequencing of candidate genes residing within linkage interval. These efforts have led to the identification of multiple novel disease loci and genes harboring causal mutations, including a gene encoding for a protein associated with autophagy, FYCO1, and genes coding for the transcription factors, TCF8 and FOXE3. My lab is currently investigating the functional significance of these causal alleles through in vitro and in vivo systems, including animal models, to understand their role in the development and normal functioning of the eye.