



BIOINFORMATICS 2016 FALL SEMINAR SERIES

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<http://bioinformatics.udel.edu/Seminars/Current>

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3:30pm
DBI Room 102

**Integrated approach to decipher regulatory networks in
eye and craniofacial morphogenesis and
their associated defects**

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ABSTRACT: Identification of ocular disease associated genes has impacted therapeutic interventions and extended our understanding of eye development and homeostasis. Disease gene discovery in the eye, however, remains a formidable challenge. Indeed, the majority of the 26 genes associated with non-syndromic human pediatric cataract were identified over a period of 25 years. The advent of genomics presents new opportunities to design systems-based integrative approaches to facilitate disease gene identification and to assemble gene regulatory networks (GRN) underlying ocular development and homeostasis. Toward this goal, we have developed a web-based publically available bioinformatics resource tool termed *iSyTE* (integrated Systems Tool for Eye gene discovery, <http://bioinformatics.udel.edu/Research/iSyTE>). *iSyTE* is based on innovative processing and presentation of whole genome expression datasets for specific eye tissues, and conversion of the wealth of molecular functional data in the literature into an interactive resource to derive and visualize “evidence-based” GRNs. This integrated approach allows *iSyTE* to effectively predict genes that are relevant to the biology of specific ocular tissues. As a proof of principal, *iSyTE* has greatly expedited gene discovery in the lens, leading to the identification of several new cataract genes (*Tdrd7*, *Pvrl3*, *Sep15*, *Mafg/k*, *Celf1*), and has contributed to the understanding of many other important regulatory pathways (e.g. *Sip1*, *CBP*, *p300*, *Prox1*, etc.). To understand their function in eye development, we have characterized targeted gene deletion mouse mutants for each of these factors and are now focusing on deriving GRNs that control eye development. Finally, we are extending the *iSyTE* approach to identify high-priority candidate genes in other areas such as craniofacial development.