Congenital malformations can arise in isolation or within complex syndromes, and are the leading cause of infant mortality in the USA. One group of complex developmental syndromes is the RASopathies, caused by inappropriate activation of RAS signaling. RASopathy patients typically carry mutations in components of the core signaling pathway. However, there are patients with “RASopathy-like” features that lack specific molecular diagnoses. Identifying new disease associated variants can potentially improve patient care, while extending our understanding of normal development. We are using model organisms to explore how mutations that occur in RASopathies affect the function of the pathway in vivo, and using our systems to identify new genes involved in these disorders. We are also developing new tools, such as optogenetic approaches, to explore how RASopathy mutations impact development over time.

Noon, Monday, December 16, 2019
Auditorium, Life Sciences Building,
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