



Undergraduate Summer Internship in Systems Biology

Internship Project Description: Summer 2018

Project Title: Analyzing intraflagellar transport in the context of ciliopathy gene mutations

Supervisor Name: Peter Czarnecki

Lab PI Name: Jagesh Shah

Project Description: Primary cilia play a crucial role in tissue patterning and developmental signaling, by sensing, integrating and transducing mechanical and chemical stimuli. Gene mutations that affect ciliary constituents give rise to a spectrum of disorders (“ciliopathies”), among which there is polycystic kidney disease, the most frequent monogenic disease in man, but also a multitude of rare and severe complex organ malformation syndromes. Previous work in this lab has established a cell culture based model of gene knockouts and lentiviral addbacks for a number of ciliopathy genes, their individual domains and their mutant isoforms that recapitulate human disease. By analyzing ciliary transport parameters, perturbations of calcium and cAMP and responses to extracellular stimuli, we hope to better understand, how a set of ciliopathy proteins and their interactions regulate signaling in developmental processes and in disease.