Utilizing Induced Pluripotent Stem Cells to Functionally Dissect Genetic Risk Factors for Autism

Due to the scarcity of available brain tissue, the subtle brain pathophysiology of autism spectrum disorders (ASDs), and the genetic and phenotypic heterogeneity of the disorders, it has been difficult to establish a biological basis for autism even in instances where a highly penetrant genetic cause has been found. The goal of this research is to identify the cell biological phenotypes that lead to ASDs in patients with known genetic variants, such as those causing rare genetic disorders, including RASopathies, or chromosomal microdeletion/microduplication, including 16p11.2. These experiments will contribute to our understanding of the underlying biological mechanism and could eventually lead to the development of assays for identifying drugs that could be used to treat the disease.

This project is a collaboration with Dr. Erik Ullian [1] in the Department of Ophtalmology and Physiology at UCSF.

UCSF Main Site

Links:
[1] http://vision.ucsf.edu/ullian/