



# Genetics, Bioinformatics, & Systems Biology Colloquium

presents

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University of Pennsylvania



**LIVE**  
**IN-PERSON**  
with Pizza

\*Live stream via Zoom available



**Thursday**  
**Feb 1, 2024**



**12PM**



**Leichtag**  
**Auditorium**



**Zoom**

## Genetic and Epigenetic Mechanisms of Neural Development and Disorders

Our lab studies genetic and epigenetic mechanisms of brain disorders that affect children with the ultimate goal to improve diagnosis and treatment strategies. Using forward genetic approaches followed by functional studies in model organisms and human pluripotent stem cells, we recently identified gain and loss of function variants in EZH1 as the basis of overlapping neurodevelopmental syndromes. While, EZH1 is the underexplored catalytic member of the Polycomb Repressive Complex 2 (PRC2), its paralogue, EZH2, has well established roles in the epigenetic transmission of transcriptionally repressed programs that guarantee multicellular organism development and cellular homeostasis. In this talk, I will first show our recent work unlocking non redundant functions of EZH1 on PRC2-mediated epigenetic regulation of nervous system development and the critical periods affected by EZH1 mutations. I will also discuss our efforts in exploring epigenetic treatments for neurodevelopmental disorders. Increasing evidence implicate genetic variants and environmental effects on epigenetic regulation as the cause of syndromic and complex neurodevelopmental disorders. Interestingly this is reminiscent of human cancers, for which a large repertoire of epigenetic drugs has been developed in the past decades. Our overarching goal is to explore the potential of these compounds for treating neurodevelopmental disorders leveraging insights gained from testing EZH1/2 inhibitors on phenotypes caused by EZH1 gain of function mutations. Here I will also describe our approach to design novel epigenetic drugs using artificial intelligence and machine learning approaches.

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