CBR Seminar Series

Date and Time of Talk: December 21st, 2017 at 4.00 PM
Venue: CNS seminar hall, IISc

Title of the talk:
From genotype to phenotype to function: Understanding the complex genetics of neurodevelopmental disorders

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Abstract:
The primary focus of my research is to understand the genomic basis of clinical heterogeneity associated with neurodevelopmental disorders such as autism, schizophrenia, and intellectual disability, with a special focus on pathogenic copy-number variants (CNVs), or duplications and deletions in the genome. Rare CNVs (with <0.1% population frequency) are a significant source of genetic defects in individuals with neurodevelopmental disorders. However, understanding the exact molecular mechanisms of disease due to these variants is complicated by extensive phenotypic heterogeneity. My research combines gene discovery and dissecting phenotypic heterogeneity using large-scale genomic studies with studying the molecular functions and mechanisms of CNV pathogenicity in model systems and developing methods for functional genomic analysis. In this talk, I will discuss observations on the genetic basis of phenotypic variability drawn from my lab’s research projects, including genome sequencing studies of 200 families, assessment of molecular function for 100 genes and about 750 genetic interactions using fly models, and development of methods for sequence data analysis and quantitative assays in model systems.