CBI
Toulouse
Centre de Biologie Intégrative

CBI SEMINAR

11h00 A.M

FRIDAY
March 25 TH 2021

Stanislav Shvartsman

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“Quantitative biology of developmental abnormalities”

Summary:
Quantitative studies of development in model genetic organisms can shed light on the mechanisms underlying the origins and progression of developmental abnormalities in humans. I will illustrate this point by presenting our ongoing work on the RASopathies, a family of developmental disorders associated with deregulated signaling through the highly conserved RAS pathway. Using genome editing and imaging approaches in Drosophila, we directly tested the causal links between multiple mutations in the same signaling component and their effects on development. Our results point to a quantitative model whereby the emerging phenotypes have both deterministic and stochastic components, mediated by the interaction between mutations and time-dependent inductive signals. Furthermore, based on tracking large cohorts of mutant embryos through multiple stages of development, we proposed that most of the individuals studied in the clinic are escapers. An unanticipated outcome of this work on developmental diseases is a new set of optogenetic techniques uniquely suited for addressing fundamental questions about the dynamics of gene control by the RAS pathway. These tools allowed us to can probe signal-dependent transcriptional repression on a time-scale of minutes.

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