Universität Basel

Research Project

Selective mRNA Translation Control in Rodent Models Carrying Mutations in Genetic Autism Risk Factors

Third-party funded project

Project title Selective mRNA Translation Control in Rodent Models Carrying Mutations in Genetic Autism Risk Factors
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With an estimate incidence of 1 in 100 children, autism spectrum disorders represent an enormous burden on the population. These developmental disorders develop in the first years of life and to date no mechanism-based treatments are available to the patients. One of the most fundamental challenges in developing treatments for autism-spectrum disorders is the heterogeneity of the condition. More than one hundred genetic mutations confer high risk for autism, with each individual mutation accounting for only a small fraction of autism cases. Subsets of risk genes can be grouped into functionally-related pathways, most prominently synaptic proteins, translational regulation, and chromatin modifications. Recent work highlighted an unexpected convergence in pathophysiology between gene products contributing to seemingly distant cellular functions. Thus, findings from model organisms suggest that mutations in autism-associated synaptic components precipitate alterations in translational regulation which resemble dysfunctions emerging from direct genetic alterations in the mRNA translation machinery. Early work conceptualized translational de-regulation as representing “too high” or “too low” levels of translation. However, based on more recent evidence it is now hypothesized that alterations in translation machinery and cell signaling result in a selective translational de-regulation of specific mRNAs which are fundamental drivers of the pathophysiology of the disorders.

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