

DISTINGUISHED GUEST SEMINAR SERIES



Institute
of Experimental
Medicine, CAS

12th June 2023 / 10:00

Turquoise seminar room of the IEM CAS

Dear colleagues, it is our pleasure to invite you to this special guest seminar

Synaptic dysfunction underlying neurodevelopmental disorders

Katherine W. Roche

The National Institutes of Health (NIH) / Receptor Biology Section

Human genetics studies have revealed that mutations in synaptic genes play a central role in the etiology of neurodevelopmental disorders such as autism spectrum disorder (ASD) and intellectual disability (ID). The Roche lab has focused on rare variants found in NMDA receptor genes, GRIN2A and GRIN2B, and find that several variants result in disruptions in binding to scaffolding proteins and deficits in NMDA receptor trafficking. The Roche lab also studies adhesion molecules, which are among the synaptic proteins most strongly associated with ASD/ID are the postsynaptic neuroligins. Her group has identified a sensitive region in neuroligin 4X that harbors a cluster of ASD/ID associated variants, which confer a trafficking deficit. In both cases, the result is a decrease in spine and synapse number.