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MPI-NAT SEMINAR SERIES

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De novo disease-causing mutations in the GABAB receptor signaling complex

My lab studies the neuronal functions of GABAB receptors, the G protein-coupled receptors for the neurotransmitter γ-aminobutyric acid (GABA). Over the past decade, our investigations have revealed that GABAB receptors interact with proteins that determine receptor localization and function. Pharmacological studies have long suggested that dysfunction in GABAB receptor signaling might contribute to various neurological and psychiatric conditions. However, it is only recently that potentially pathogenic variants have been identified in the genes for the GABAB receptor subunits GB1 and GB2, as well as in receptor-associated proteins such as AJAP1 and PIANP. We have analyzed gene variants linked to epileptic encephalopathy, Rett syndrome, developmental delay, and autism spectrum disorder, both in vitro and using mutant mouse models. Our studies provide insights into disease mechanisms and help to propose therapies.

Tuesday, 23.04.2024, 11:00

Host: Nils Brose

City Campus Lecture Hall

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