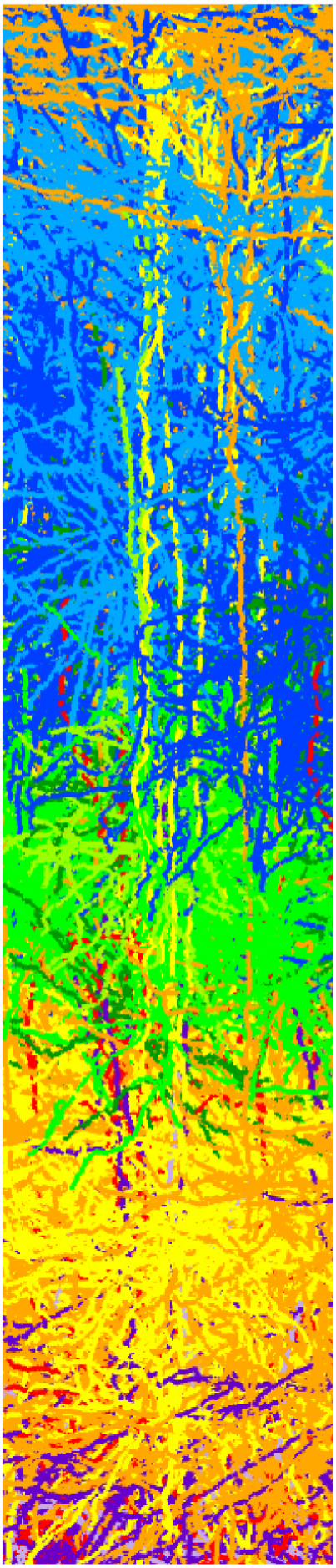
UNIVERSITÄT BONN

Neuroscience Lecture

Bonn Center of Neuroscience



Gain-of-Function mutations in calcium channel genes are an emerging cause of severe neurodevelopmental disorder

Philippe Lory, Prof. Dr.

'Canaux Ioniques - Excitabilité Neuronale et Canalopathies', Institut de Génomique Fonctionnelle, Departement Neurosciences, CNRS UMR5203 - INSERM U1191 - Université de Montpellier

The work of Philippe Lory's group is focused on studies on the properties of calcium channels activated by membrane depolarization (Cav) and further also on NALCN channels. He has contributed seminal publications particularly on the modulation of T-type calcium channels (Cav3) by endogenous ligands including lipids, zinc, calcium, phosphorylation and other factors. A major topic of his presentation will be on recent advances aiming to decipher the consequences of gain-of-function mutations in Cav3 (especially Cav3.1 / CACNA1G) channels. Cav3.1 is responsible for early-onset severe spinocerebellar ataxia-42 with neurodevelopmental deficits (SCA42ND), an ultrarare but clinically very severe autosomal dominant syndrome, and a wider spectrum of neurodevelopmental, cerebellar and epileptic conditions.

September 25, 2023, 04:00 – 05:15 p.m.

Epileptology, Seminar Room 266/83, Ground Floor

Find more talks

If you would like to meet with the speaker, please contact:

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