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2nd Channelopathy Meeting Tübingen

Genetic epilepsies and other neuronal ion channel disorders: Mechanisms and therapeutic perspectives



Program

Thursday, 5th October 2023

8:30-8:40 am	Introduction: Holger Lerche (Tübingen)
8:40-11:45 am	<p>Session 1: Sodium channelopathies Chair: Thomas Wuttke (Tübingen) & Moran Rubinstein (Tel Aviv)</p> <p>Andreas Brunklaus (Glasgow): Emerging phenotypes in sodium channelopathies - the role of functional prediction and therapeutic advances</p> <p>Evgenia Rusina (Nice): A homeostatic response boosted as therapeutic approach in <i>Scn1a^{+/−}</i> Dravet syndrome mice</p> <p>Nikolas Layer (Tübingen): Early electrophysiological and transcriptomic alterations drive epileptogenesis in Dravet syndrome</p> <p>Elena Gardella (Dianalund): SCN8A: Clinical complexity and natural history study</p> <p>Heinz Beck (Bonn): Targeting aberrant dendritic integration to treat cognitive comorbidities of epilepsy</p> <p>Sanjay Sisodiya (London): Genetic epilepsies: complexities to come - SCN1A as an example</p>
10:15 am (v)	Coffee Break
10:35-11:05 am	
11:05 am	Angelika Lampert (Aachen): Precision Therapy in sodium channel related neuropathic pain
11:25 am	JP Johnson (Xenon, Burnaby): A Selective Na _v 1.1 Potentiator Enhances Interneuron Excitability to Normalize Motor Performance in a Dravet Syndrome Mouse Model
11:45 am-1:25 pm	<p>Session 2: Potassium channelopathies Chair: Philip Ahring (Sydney) & Stephan Luxmann (Tübingen)</p> <p>Jennifer Kearney (Chicago): Modeling <i>Kcnb1</i>-associated developmental and epileptic encephalopathy in mice</p> <p>Peter Müller (Tübingen): Transcriptomic insights into epileptogenesis and compensatory changes in a <i>Kcna2</i> loss-of-function mouse model</p> <p>Konstantin Khodosevich (Copenhagen): Understanding mechanisms of epileptogenesis using single cell omics</p> <p>Annapurna Poduri (Boston): Modeling potassium channelopathies in zebrafish</p>
1:00-2:00 pm	Lunch Break
2:00-3:50 pm	<p>Session 3: HCN, calcium and ligand-gated channelopathies Chair: Dirk Isbrandt (Cologne/Bonn) & Ulrike Hedrich-Klimosch (Tübingen)</p> <p>Bina Santoro (New York): Molecule, cell, organism: multi-level analysis of a sick ion channel</p> <p>Christopher Reid (Melbourne): A small-molecule precision medicine for HCN1 DEE</p> <p>Stephan Marguet (Cologne): HCN/h channel modulation of excitation/inhibition balance in the MEC is crucial for cortico-hippocampal information flow and intrahippocampal dynamics</p> <p>Jörg Striessnig (Innsbruck): CACNA1D L-type calcium channelopathies in neurodevelopmental disorders: opportunities for drug repurposing</p> <p>Philip Ahring (Sydney)/Rikke S. Møller (Dianalund): Pathogenic GABRA3 variants lead to dominant or recessive X-linked disorders depending on functional outcomes</p>
3:50-5:00 pm	Poster Session (with coffee)
5:00-7:00 pm	<p>Session 4: Clinical trials and molecular therapeutic board (case reports: 5-10 min including discussion) Chair: Holger Lerche (Tübingen) & Michael Alber (Tübingen)</p>

5:00 pm (v)	Steven Petrou (Praxis Precision Medicines, Boston): First in patient data for PRAX-222, an ASO for SCN2A gain of function DEE
5:20 pm	Neetha Balaram (Kozhikode): Acetazolamide responsive early-onset absence epilepsy and ataxia in a toddler with a KCNA2 genetic variant; a case report
5:30 pm	Cristina Franco (Naples): tbd
5:40 pm	Hasnaa Elbendary (Cairo): Mutational Spectrum of Epileptic encephalopathy in a cohort of pediatric Egyptian population
5:50 pm	Sopio Gverdtsiteli (Dianalund): Neonatal developmental and epileptic encephalopathy with movement disorders and arthrogryposis (NDEEMA)
6:00 pm	Rikke Steensbjerre Møller (Dianalund): Treatment responsiveness in LOF and GOF GABA-A receptor related epilepsies
6:10 pm	Stephan Luxmann/Michael Alber (Tübingen): Drug repurposing in KCNT1 encephalopathies
6:20 pm	Robert Lauerer-Braun (Tübingen): DECADE-Deciphering the CACNA1E-related DEE: a natural history trial
8:00 pm	Dinner Restaurant „Liquid“

Friday, 6th October 2023

8:30-10:35 am	Session 5: Gene therapy and human models Chair: Maria-Patapia Zafeiriou & Niklas Schwarz
8:30 am	Yichen Qiu (London): On demand gene therapy for epilepsy
8:50 am	Moran Rubinstein (Tel Aviv): Viral-mediated expression of Na _v 1.1 ameliorates Dravet syndrome in mice post seizure onset
9:10 am	Nael Kasri (Nijmegen): Leveraging spontaneous activity in human stem cell derived neurons to model SCN1A-related epilepsies
9:30 am	Maria-Patapia Zafeiriou/Hendrik Rosewich (Göttingen/Tübingen): Modeling ATP1A3 related disorders in bioengineered neuronal organoids
10:00 am	Sarah Weckhuysen (Antwerp): KCNQ2-related epilepsies in iPSC-derived neuronal cultures
10:20 am	Filip Rosa (Tübingen): KCNQ2-DEE stem cell model shows a developmental delay along a prolonged culture time up to 6 months
10:35 am	Snezana Maljevic (Melbourne): Insights from in vitro disease models of SCN2A DEE
10:55-11:20 am	Coffee Break
11:20 am-12:50 pm	Session 6: Epigenetics, protein structural modeling & prediction tools Chair: Rikke Steensbjerre Møller & Christopher Reid
11:20 am	Katja Kobow (Erlangen): Epigenetics for diagnosis and mechanistic understanding of drug-resistant focal structural epilepsies
11:40 am	Alison Obergrussberger (Nanion, München): How Nanion's technologies are advancing channelopathy research
12:00 pm	Christian Bosselmann (Cleveland): Predicting the functional effects of ion channel variants: State of the art and future directions
12:15 pm	Tobias Brünger (Cologne): Predicting molecular function and pathogenicity in ion channel disorders
12:30 pm (v)	Giulia Rossetti (Jülich): Studying functional gating mechanisms in wild-type or mutant Na _v channels/ <i>In-silico</i> variant-specific drug screening
12:50-1:30 pm	Farewell Lunch with free discussions