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Cerebellar ataxias: Insights from recent preclinical and clinical studies

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Cerebellar ataxia is a rare neurological disorder characterized by the inability to control voluntary movements, and can manifest among other as gait ataxia, intention tremor or dysmetria. Cerebellar ataxias are a genetically heterogeneous group of related disorders, with different modes of inheritance, presenting in a number of diverse clinical phenotypes. Episodic Ataxias (EA) are channelopathies caused by mutations in ion channels and neurotransmitter transporters, including KV1.1, CaV2.1 and SLC1A3. However, to date there is no confirmed treatment for cerebellar ataxias, and responder rates for those existing drugs are generally low. Novel preclinical model systems that incorporate the molecular genetics of EA and closely resemble the human condition have brought about a paradigm change that requires us to revise our strategies for drug discovery and development for cerebellar ataxias. In this talk, the latest experimental findings in basic, translational and clinical research, discuss new drug targets and outline novel strategies for drug discovery that focus on restoring synaptic transmission in the EA-afflicted brain will be presented.

Biography

Simon Kaja is an experienced neurobiologist with a long-standing research track record in academia and the biotech and pharmaceutical industry. He currently serves as Associate Director of Preclinical Research at the Vision Research Center at the University of Missouri - Kansas City, School of Medicine. He is the Director of Microscopy of the imaging core facility at the Vision Research Center and also holds an appointment to Assistant Professor of Ophthalmology. His NIH-funded research program focuses on human neurological and neurodegenerative diseases, visual disorders and inflammation. He obtained his BSc (Hons.) degree in Molecular Biology and Biochemistry from Durham University, UK and holds a PhD degree in Medicine/Neuroscience from Leiden University, The Netherlands. Prior to joining the faculty at the University of Missouri - Kansas City, he has performed Postdoctoral work at The University of British Columbia (Vancouver, B.C., Canada) and the University of North Texas Health Science Center (Fort Worth, TX). In addition to his academic work, he has worked and consulted for a number of pharmaceutical companies, incl. Novo Nordisk A/S, Bayer AG, Neuromed Pharmaceuticals Inc., and NeuroSearch A/S. He is CEO and Co-founder of K&P Scientific LLC, a scientific consulting company headquartered in Kansas City, MO.

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