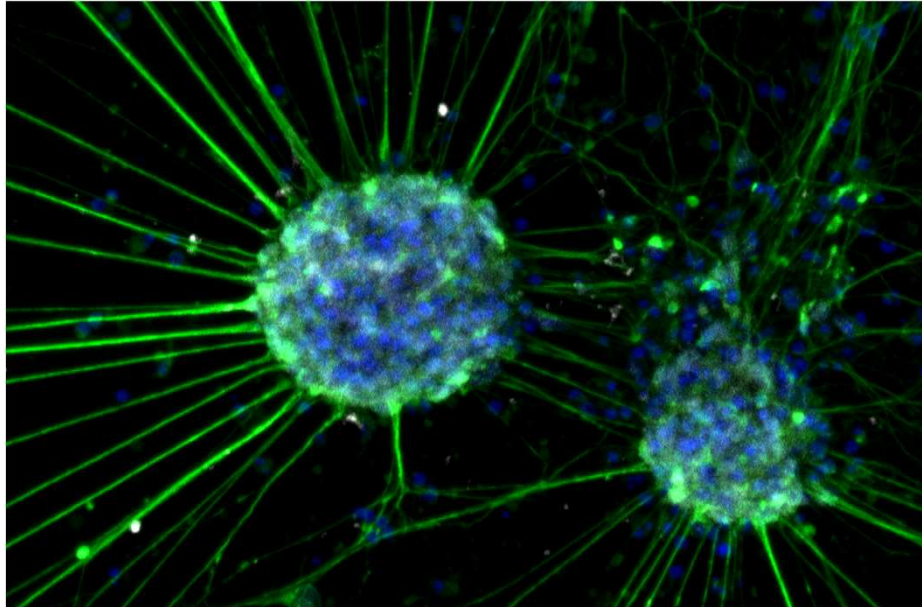


Symposium International – 6^e édition

Ataxie Récessive Spastique de Charlevoix-Saguenay (ARSACS)

De Charlevoix à ailleurs dans le monde...



Le jeudi 4 novembre 2021

Conférenciers:

Jeremy D. Schmahmann (États-Unis)
Paola Giunti (Royaume-Uni)
Francesca Maltecca (Italie)
Anne McKinney (Canada)
Thomas Schwarz (États-Unis)
Matthis Synofzik (Allemagne)
Alanna Watt (Canada)
Justin Wolter (États-Unis)

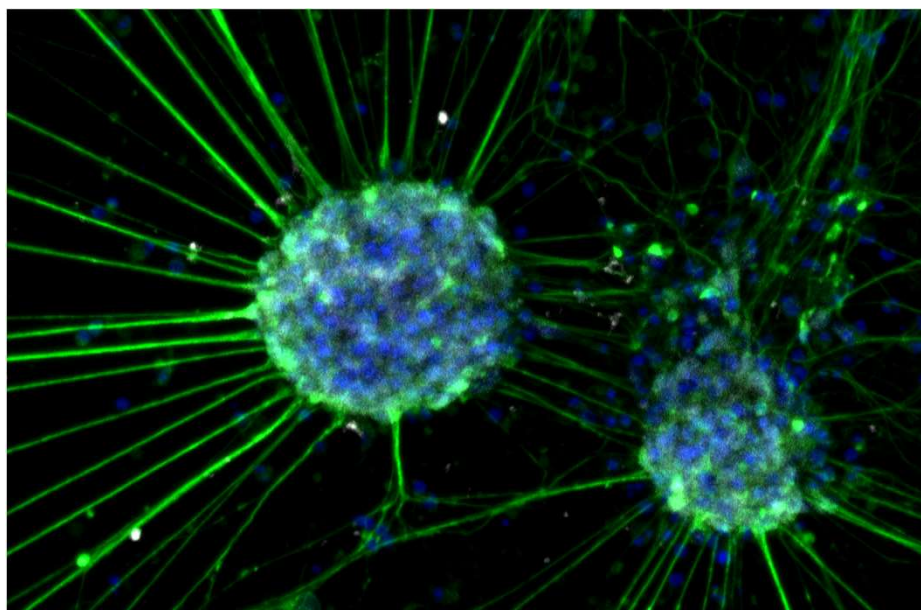
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<https://ulaval.zoom.us/meeting/register/u5csc-uvrj0oHNadcp8jebR-fuNXR9Bvo257>

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6th International Symposium on Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay (ARSACS)

From Charlevoix to the World...



Thursday, November 4th, 2021

Guest Speakers:



Jeremy D. Schmahmann (USA)
Paola Giunti (United Kingdom)
Francesca Maltecca (Italy)
Anne McKinney (Canada)
Thomas Schwarz (USA)
Matthis Synofzik (Germany)
Alanna Watt (Canada)
Justin Wolter (USA)

<https://ulaval.zoom.us/meeting/register/u5csc-uvrj0oHNadcp8jebR-fuNXR9Bvo257>

Contact info: francois.gros-louis@fmed.ulaval.ca



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**CENTRE DE
RECHERCHE**



Agenda

8:00 EDT Words of Welcome

Dr. François Gros-Louis (Université Laval, Canada)

8:10 Opening remarks

Chair: Dr. Nicolas Dupré (Université Laval, Canada)

Provisory title: *The cerebellum in ARSACS*

Keynote Speaker – Dr. Jeremy D. Schmahmann (Harvard Medical School & Mass General Hospital, USA)

9:20 Break (10 mins)

9:30 – 10:30 **Modelling ARSACS in the lab – Insight into Disease Mechanisms Session**

Chair: Dr. Benoit Gentil (McGill University, Canada)

9:30 - 9:50 *Dissecting and targeting the molecular pathogenesis of ARSACS*

Dr. Francesca Maltecca (University Vita-Salute San Raffaele, Italy)

9:50 - 10:10 *Does SARM1 participate in the neurodegeneration of sarsin^{-/-} mice?*

Dr. Thomas Schwarz (Boston Children's Hospital, USA)

10:10 - 10:30 *Mechanisms of resilience: why do some cells die, and others survive in ARSACS?*

Drs. Anne McKinney & Alanna Watt (McGill University, Canada)

10:30 – 11:00 Discussion period

11:00 Break (10 mins)

11:10 – 12:10 **Biomarker and Disease Trajectories Session**

Chair: Dr. Jean-Denis Brisson (CIUSSS du Saguenay-Lac-Saint-Jean, Canada)

11:10 - 11:30 *Molecular and Cellular Deficiencies in ARSACS*

Dr. Justin Wolter (University of North Carolina, USA)

11:30 - 11:50 *Establish multi-center trial-readiness for ARSACS on a global scale: study protocols and clinical, patient-reported, imaging, digital-motor, and imaging outcomes.*

Dr. Matthis Synofzik (University of Tübingen, Germany)

11:50 – 12:10 *Provisory title: Retinal architecture in ARSACS*

Dr. Paola Giunti (University College London, UK)

12:10 - 12:40 Discussion period

12:40 Closing remarks



Jeremy D. Schmahmann, MD

Dr Schmahmann is Professor of Neurology at Harvard Medical School, and Neurologist at the Massachusetts General Hospital where he is Founding Director of the Ataxia Center (1994), Director of the Laboratory for Neuroanatomy and Cerebellar Neurobiology, and a member of the Cognitive Behavioral Neurology Unit. Dr. Schmahmann received his medical degree with distinction at the University of Cape Town, winning the Nestle Prize (pediatrics) and Wilfrid Exner Bauman Prize (best student). Dr. Schmahmann completed residency in the Neurological Unit of the Boston City Hospital, and Anatomy and Neurobiology Fellowship in the Boston University School of Medicine. He joined the MGH faculty in 1989. He is a Fellow of the American Academy of Neurology, the American Neurological Association, and the American Neuropsychiatric Association.

Dr. Schmahmann won the American Academy of Neurology's Norman Geschwind Prize (2000) for pioneering work on the role of the cerebellum in cognition and emotion, and description of the cerebellar cognitive affective syndrome (now, Schmahmann's syndrome).



Francesca Maltecca, PhD

Dre Maltecca obtained her Master in Biotechnology at the University of Milan in 2003 and her PhD in Molecular Medicine at University Vita-Salute San Raffaele in Milan in 2008. She has established her independent laboratory at Ospedale San Raffaele in Milan in 2017.

The major research interest of her group is neurodegeneration associated to mitochondrial dysfunction, with a focus on spastic-ataxias. Over the years, she has matured expertise in this field by working on *m*-AAA-associated diseases under the supervision of G. Casari, and through experiences as visiting scientist at Skejby Hospital, Aarhus (Denmark) under the supervision of P. Bross, and at University of Padua (Italy) under the supervision of R. Rizzuto.

Her work highlighted for the first time the intimate connection between mitochondria and calcium in Purkinje neurons. Alteration of this pathway is now clearly emerging as a crucial mediator of the pathogenesis of many cerebellar ataxias. Her studies focus on dissecting the mechanism of genetic forms of cerebellar ataxia that have in common defects in mitochondrial quality control, dynamics and metabolism. She was awarded in 2012 and 2013 with the Young Investigator Award for the SCAs Research, from National Ataxia Foundation, US



Thomas Schwarz, PhD

Dr. Schwarz received an A.B. in Biochemistry and Molecular Biology and a Ph.D. in Neurobiology from Harvard. He performed postdoctoral work at UCSF in the laboratory of Lily and Yuh-Nung Jan where he was part of a team that cloned the first gene for a K⁺ channel. He was on the faculty of Stanford Medical School for 10 years, where he focused on the molecular biology and electrophysiology of channels and on the genetic analysis of synaptic transmission. In 2000 he relocated to the FM Kirby Neurobiology Center at Boston Children's Hospital where he is Professor of Neurology with a joint appointment in the Dept. of Neurobiology at Harvard Medical School.

The Schwarz lab studies the cell biology of neurons and seeks to understand how nerve cells work, acquire their shape, and make their connections. We do so with the expectation that understanding how the cell functions can give us insight into larger properties of circuits. Much as the shape and style of a building is constrained and influenced by the properties of the building materials, so too is the architecture and circuitry of the brain dependent on how these cellular components operate. The research interests of the Schwarz Lab include 1) the means by which axonal transport allows the cell body of a neuron to communicate with and support its axons and dendrites; 2) the transport and turnover of neuronal mitochondria in response to mitochondrial damage, 3) the development and structural plasticity of synapses. Projects move back and forth between *Drosophila melanogaster*, mice, rats, and human cell lines as the scientific question demands.



Anne McKinney, PhD

Anne McKinney is a Professor in the Department of Pharmacology and Therapeutics and the Associate Dean in Medicine (Academic Affairs) at McGill University. She obtained her Ph.D., at the University of Ulster, UK in 1992. She then spent the next 5 years in the Department of Neurophysiology at the Brain Research Institute, University of Zurich as a postdoctoral fellow. In 1998 she obtained her own group at the Brain Research Institute. She joined the Department of Pharmacology and Therapeutics of McGill University in 2005 and became a Full Professor in 2015.

She is internationally recognized for her work on the cellular and molecular mechanisms that underlie synapse plasticity and maintenance in healthy brain and in neurodevelopment diseases and neurodegenerative disease. The McKinney lab is using a combination of techniques including, 4-dimensional confocal laser scanning microscopy, serial electron microscopy, transgenic animals, behaviour and electrophysiological techniques to find novel therapeutic targets. She has won awards for her research including the Pfizer prize in Neuroscience. Dr. McKinney serves as a member of the scientific advisory board of the National Research Council Canada's Medical Devices Research Centre as well as being an external advisory board member for University of Texas at Austin NSF grant NeuroNex technology hubs



Alanna Watt, PhD

Dre Alanna Watt is an Associate Professor in the department of Biology at McGill University, whose research plan is centered on the role of the cerebellum in health and disease. She obtained her PhD at Brandeis University in Massachusetts, USA, in 2003. She then spent the next 5 years at University College London (UCL) in London, UK as a postdoctoral fellow. She obtained a Dorothy Hodgkin Research Fellowship from the Royal Society (UK) in 2008, and was recruited to McGill University in 2011, receiving tenure in 2017.

Her work focuses on the cerebellum in health and disease, with a focus on neurodegenerative disorders such as ataxia. She has published in several leading journals, including Nature Neuroscience, Neuron, Nature Methods, and Cell Reports. She is a member of the College of Reviewers for CIHR, serving on the Cellular and Molecular Neuroscience (NSB) grant evaluation panel since 2019. She is an editor for three journals (Scientific Reports, Frontiers in Synaptic Neuroscience and Open Biology). She will be the Chair of the upcoming Gordon Research Conference on the Cerebellum (in 2025, postponed due to Covid-19).



Justin Wolter, PhD

Dr. Wolter is a distinguished Postdoctoral Fellow of gene therapy at University of North Carolina at Chapel Hill School of Medicine, USA. He received his BS in Psychology at Utah State University, and earned his PhD at Arizona State University under the mentorship of Marco Mangone, where he studied the connection between evolution and targeting principles of microRNAs.

He works in the labs of Drs Mark Zylka and Jason Stein at the University of North Carolina, where he studies molecular and genetic mechanisms in neurodevelopmental disorders, including Angelman Syndrome, and autism. In collaboration with Dr. Paul Chapple, Dr. Wolter has been leading a team seeking to identify the causal molecular deficiencies in ARSACS.



Matthias Synofzik, MD

Prof Synofzik is head of the research unit “Translational Genomics of neurodegenerative diseases” and senior consultant neurologist at the Hertie Institute for Clinical Brain Research & Center of Neurology, University of Tübingen, Germany. Since 2013 he is co-chair of the Ataxia outpatient clinics at the Center for Neurology, and leads the outpatients clinics for Frontotemporal Dementias and for Amyotrophic Lateral Sclerosis. He holds graduate qualifications in philosophy and in medicine and postgraduate qualifications in neurology.

Matthias has received over €4,0 million in fellowships/scholarships or grants, leading several worldwide EJP-RD consortia on recessive ataxias like “PREPARE” and „PROSPAX” as well as co-lead of the global trial-readiness platform “ATAXIA GLOBAL INITIATIVE”. He coordinates the international multicenter prospective longitudinal database on recessive ataxias (“ARCA registry”), which brings together >25 world-leading ataxia centers. His expertise spans clinical research of disease and treatments in hereditary ataxias, frontotemporal dementias, ALS and orphan neurological diseases as well as the underlying molecular genetics and neurobiology. Using latest next-generation genomic sequencing techniques, he has been leading or involved in the identification of >10 novel ataxia genes in the last 7 years. In parallel, he has unravelled a wide range of both molecular (e.g. blood-based molecules) and digital-motor biomarkers (e.g. wearable sensors) directly facilitating trial-readiness of hereditary ataxias.



Paola Giunti, MD PhD

Prof Giunti is a Professorial Research Associate at the Department of Clinical and Movement Neurosciences, University College London Queen Square Institute of Neurology, UK. She studied Medicine specializing in Neurology and also gained her PhD at the University of Rome, La Sapienza, in Italy. During 1992-1995 she was a Research Fellow at the Institute of Neurology UCL, Queen Square in Department of Clinical Neurology, before returning to the Rome. In 2004 she was successful in gaining a Fellowship from Ataxia UK and was made a Senior Clinical Associate again in the Department of Clinical Neurology at Queen Square. She established an Ataxia Centre at the NHNN in 2005, a robust model of translational clinical service. This has been accredited as the first centre of clinical excellence in the UK by Ataxia UK, the charity of ataxia patients.

She has a longstanding interest in the pathogenesis of neurological disorders, in particular the inherited cerebellar ataxias and other movement disorders. At UCL, she has been researching the mechanisms that underlie neuronal degeneration in the central and peripheral nervous systems. Her contribution has been instrumental in the identification of genes causing different autosomal dominant cerebellar disorders. Paola's research has also expanded the clinical spectrum of several rare inherited disorders