

# Third International Symposium on ARSACS

(Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay)

Montreal Neurological Institute, Montreal, Canada, Wednesday 12<sup>th</sup> and Thursday 13<sup>th</sup>, 2014

**Sponsors: Ataxia Charlevoix-Saguenay Foundation, The Genetics Institute of the Canadian Institutes for Health Research (CIHR), The CIHR New Emerging Team on ARSACS and the Montreal Neurological Institute (The Neuro)**



**Purpose:** This International symposium brings together researchers, clinicians and drug developers to discuss the most recent advances in our understanding of ARSACS in the larger context of mitochondrial dysfunction in neurodegeneration. The clinical discussions will be focused on developing an international network of centers for future drug trials and a critical evaluation of a proposed clinical severity ARSACS scale that could be used internationally. Discussion on the uncovering of the pathophysiology of ARSACS will aim at accelerating the discovery of treatments for ARSACS.

There will be a 30 minute presentation by all speakers, followed by 10 minutes for Q and A \*

## **Wednesday 12<sup>th</sup> of November, Jeanne Timmins Amphitheater, Montreal Neurological Institute**

8:30 - 09:00 Welcome and Introduction by Guy Rouleau, Sonia Gobeil, Jean Groleau & Bernard Brais

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**09:00 - 12:10 Session I: Clinical features and genetics of ARSACS**  
(Chairs: Bernard Brais, McGill University & Cynthia Gagnon, Sherbrooke University, Canada)

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09:00 - 09:10 Bernard Brais, introduction

09:10 - 09:40 **Cynthia Gagnon**, Sherbrooke University, Saguenay, Canada: Still learning from the Quebec ARSACS cohort

09:40 - 10:30 **Sascha Vermeer**, Groningen, Netherlands: ARSACS and beyond

10:30 - 10:50 Coffee Break

10:50-11:30 **Paola Giunti**, University College, London, UK: The UK ARSACS cohort

11:30 - 12:10 **Cynthia Gagnon & Bernard Brais**: Open discussion on ARSACS severity scale and future steps in increasing international collaboration

12:10 - 13:10 Lunch

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**13:10 - 13:30 Session II: Possible overlap in the pathophysiology of ARSACS and SCA28**  
(Chairs: Heather Durham & Alanna Watt, McGill University, Montreal, Canada)

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13:10 - 13:20 Heather Durham & Alanna Watt, introduction

13:20 - 14:00 **Roxanne Larivière**, McGill University, Montreal, Canada: Sacs knockout mice present pathophysiological defects underlying ARSACS

14:00 - 14:40 **Elena Rugarli**, Cologne, Germany: Cell-autonomous roles of AFG3L2 in neurodegeneration

- 14:40-15:00 Coffee Break
- 15:00 - 15:40 **Francesca Maltecca**, Università Vita-Salute San Raffaele Milan, Italy: Ineffective mitochondrial calcium buffering exacerbates glutamate stimulation in Purkinje neurons of the spinocerebellar ataxia type-28 (SCA28) preclinical model
- 15:40 - 16:20 **Filippo Santorelli**, University of Pisa, Italy: Sacsin in Italy: From A(rsacs) to Z(ebrafish)
- 17:00 - 18:30 "Café Scientifique", Scientific forum and cocktail organized by the Genetics Institute of the CIHR, McCord Museum
- 19:30 - 22:00: Dinner for speakers

**Thursday 13<sup>th</sup> of November, de Grandpré Communication Centre, Montreal Neurological Institute**

**08:50 - 12:00 Session III: Insights from the Structure/Function of Sacsin; Links to mitochondria?**  
(Chairs: Kalle Gehring & Anne McKinney, McGill University, Canada)

- 08:50 - 09:00 Kalle Gehring & Anne McKinney, introduction
- 09:00 - 09:40 **Kalle Gehring & Jason Young**, McGill University, Montreal, Canada: Sacsin's structure and chaperone function
- 09:40 - 10:20 **Heather Durham & Benoit Gentil**, McGill University, Canada: Cytoskeletal disorganization and mitochondrial transport defect caused by loss of sacsin
- 10:20 - 10:40 Coffee Break
- 10:40 - 11:20 **Stefan Strack**, University of Iowa Carver College of Medicine, Iowa City, USA: Regulation of mitochondrial fission in neurons
- 11:20 - 12:00 **Paul Chapple**, Barts and the London School of Medicine, London, UK: Mitochondrial association of dynamin-related protein 1 is impaired in cellular models of ARSACS
- 12:00 - 13:00 Lunch

**13:00 - 16:30 Session IV: Mitochondrial dysfunction and dynamics in disease**  
(Chairs: Peter McPherson & Heidi McBride, McGill University, Canada)

- 13:00 - 13:10 Peter McPherson & Heidi McBride, introduction
- 13:10 - 13:50 **Richard Youle**, NIH, Bethesda, USA: Mechanisms of mitochondrial quality control
- 13:50 - 14:30 **Eric Shoubridge**, McGill University, Montreal, Canada: Mitochondrial dysfunction in disease: deciphering cause and effect
- 14:30 - 14:50 Coffee Break
- 14:50 - 15:30 **Thomas Schwarz**, Boston, USA: Turnover of Damaged Mitochondria in Neuronal Axons
- 15:30 - 16:00 **Bernard Brais, Cynthia Gagnon, Peter McPherson & Heidi McBride**, McGill University, Montreal, Canada: Wrap up and discussion of future directions in ARSACS research. How to accelerate our path from Bench to Bedside?
- 16:00 End of symposium