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ARSACS NEWS

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This month's newsletter features recent publications, news and upcoming conferences. The purpose of this publication is to disperse information about ARSACS so that links and associations can be made, enabling these to the discovery of treatments more quickly. To update all ARSACS stakeholders on the latest news.

RESEARCH HIGHLIGHT

"*Longitudinal Imaging Biomarkers Correlate with Progressive Motor Deficit in the Mouse Model of Charlevoix-Saguenay Ataxia*" by Dr. Francesca Maltecca and her team. In autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS) disease, severity and age of onset vary greatly, hindering to objectively measure and predict clinical progression. Thickening of the retinal nerve fiber layer is distinctive of ARSACS patients, as assessed by optical coherence tomography, whereas conventional brain magnetic resonance imaging findings include both supratentorial and infratentorial changes. Because longitudinal imaging studies in ARSACS patients are not available to define these changes as biomarkers of disease progression, we aimed to address this issue in the ARSACS mouse model.

Read the full article here: https://onlinelibrary.wiley.com/doi/10.1002/ana.27146

FUNDING OPPORTUNITY

Stay tuned for our upcoming funding opportunity. Again, this year, the Foundation will issue its call for proposals to fund 2025-2026 research projects on ARSACS. More details to come in February.

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UPCOMING CONFERENCES FOR ARSACS

⋙ Drs Nicolas Dupré and Elise Duchesne will be the hosts of a webinar organized by the National Ataxia Foundation. This informative webinar will be presented on **February 13, 2025** and will be covering clinical aspects of the disease and the ongoing research.

⋙ Sonia Gobeil (cofounder of the Ataxia of Charlevoix-Saguenay Foundation) and Dr. Benoit Gentil (researcher at McGill) will be presenting at the Regroupement Québecois des maladies orphelines on **February 14, 2025**. Their talk will address their collaborative project on gene therapy for ARSACS.

⋙ The Ataxia of Charlevoix-Saguenay Foundation and ARSACS research work will be well represented at the World Orphan Drug Congress. This well-attended congress which will be held in Boston from **April 22-24, 2025**, convenes top pharmaceutical and biotech companies, government and regulatory authorities, patient advocacy groups, payers, investors and solution providers. This congress provides a forum to discuss and advance orphan drug development and enhance access to life-saving therapies for individuals living with a rare disease.

Two ARSACS presentations are scheduled on the program:

- Sonia Gobeil and Betsy Trainor (member of the ARSACS board) along with Dr. Jeremy Schmahmann will present "*Fireside Chat: ARSACS (Ataxia of Charlevoix-Saguenay): Finding Solutions for Rare Disease Patients*".
- Alexandre Paré (McGill PhD student) will present "*Development of a gene therapy for Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay; challenges and success*".

In addition, Sonia will be having a booth exhibit at the congress. Come and meet with her.

WORLD OrphanDrug Congress USA 2025

For more information about the 2025 World Orphan Drug Congress and to register, please visit :
https://www.terrapinn.com/bookwodc_arsacs
50% OFF PROMO CODE : **ARSACS50**

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