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ARSACS NEWS

EDITOR : ATAXIA OF CHARLEVOIX-SAGUENAY FOUNDATION

SGOBEIL@ARSACS.COM
WWW.ARSACS.COM

This newsletter features the latest updates on ARSACS, including a recent scientific publication and our funding opportunity for ARSACS research projects. We are also pleased to share that the Foundation will be participating in the World Orphan Drug Congress (WODC), where we will continue our efforts to raise awareness and foster collaborations in the field of rare diseases.

RESEARCH HIGHLIGHT

"ARSACS: Clinical Features, Pathophysiology and iPS-Derived Models", co-authored by Dr. Nicolas Dupré, a member of our board of directors.

Autosomal-recessive spastic ataxia of Charlevoix-Saguenay (ARSACS) is an early-onset neurodegenerative disease caused by mutations in the SACS gene. The first two mutations were identified in French Canadian populations 20 years ago. The disease is now known as one of the most frequent recessive ataxias worldwide. Prominent features include cerebellar ataxia, pyramidal spasticity, and neuropathy. Neuropathological findings revealed cerebellar atrophy of the superior cerebellar vermis and the anterior vermis associated with Purkinje cell death, pyramidal degeneration, cortical atrophy, loss of motor neurons, and demyelinating neuropathy. No effective therapy is available for ARSACS patients but, in the last two decades, there have been significant advances in our understanding of the disease. New approaches in ARSACS, such as the reprogramming of induced pluripotent stem cells derived from patients, open exciting perspectives of discoveries. Several research questions are now emerging. Here, we review the clinical features of ARSACS as well as the cerebellar aspects of the disease, with an emphasis on recent fields of investigation. Read the full article here:

https://arsacs.com/wp-content/uploads/2025/01/arsacs-clinical-features-pathophysiology-and-i-ps-derived-models.pdf

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VOLUNTEER COMITEE

In November 2024, Betsy Trainor launched an official United States volunteer committee to support the mission of the ARSACS Foundation. Betsy sits on the Board of Directors of the Foundation and has a daughter in her mid-twenties with ARSACS. The goal of the committee is to give more visibility to ARSACS in the United States and to unify fundraising efforts. During the first meeting, the committee discussed potential fundraising ideas for 2025. They are working on universal template letters that can be used to seek scientific support for ARSACS research, for fundraising events, and to gather donations for the annual gala that takes place annual in Montreal. The committee plans to meet monthly and is open to anyone who is interested in joining. Current members consist of ARSACS patients, ARSACS caregivers and extended family members. You do not need to live in the United States to join, as the committee hopes its discussions will be useful and beneficial to anyone interested in moving ARSACS research efforts forward.

For more information about the volunteer committee, contact Betsy Trainor at elizabethann1966@gmail.com.

FUNDING OPPORTUNITY

The call for proposals has been launched! The Ataxia Charlevoix-Saguenay Foundation is inviting researchers to apply for funding to support 2025-2026 research projects on ARSACS. This annual funding opportunity aims to advance scientific discoveries and potential treatments for the disease.

Apply now : https://arsacs.com/research/

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MISSED THESE CONFERENCES? WATCH THEM NOW!

https://www.youtube.com/watch?v=6Vpjssf6idU

https://www.youtube.com/watch?v=t90M3V7E24s

https://www.youtube.com/watch?v=MwOxSuueGKw&t=1s

*Presented in French only, with subtitles available in multiple languages.

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UPCOMING CONFERENCES FOR ARSACS

The World Orphan Drug Congress (WODC) 2025 is fast approaching, and the Ataxia of Charlevoix-Saguenay Foundation will be there!

From **April 22-24** in **Boston**, we will be participating in discussions and presentations focused on ARSACS research and rare disease advocacy:

>>> **April 22nd, 4:00 PM**
Development of a Gene Therapy for Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay; Challenges and Success – Presented by Alexandre Paré (McGill PhD student).

>>> **April 23rd, 2:30 PM**
Fireside Chat: ARSACS (Ataxia of Charlevoix-Saguenay): Finding Solutions for Rare Disease Patients – Presented by Sonia Gobeil (co-founder of the Foundation) , Betsy Trainor (ARSACS board member), and Dr. Jeremy Schmahmann (Harvard Medical School).

If you're a patient advocate, healthcare professional, or part of a patient group organization, you may qualify for a **complimentary pass** to attend. Don't miss this opportunity to connect with experts, engage in critical conversations, and support the rare disease community.

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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