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ARSACS NEWS

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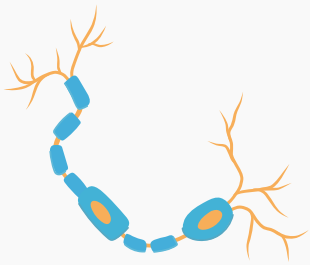
We are pleased to share the latest updates from the ARSACS Foundation. In this edition, you'll find a newly published scientific article on ARSACS, details about our upcoming 2025 *Diner des Producteurs* as well as information on our participation at the upcoming World Orphan Drug Congress (WODC) Europe in Amsterdam. As well, this edition features exclusive content such as the video presented at ASGCT and a touching interview with Maxine Monks, a mother sharing her perspective on living with ARSACS.

RESEARCH HIGHLIGHT



"*S100B Mitigates Cytoskeletal and Mitochondrial Alterations in a Glial Cell Model of Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay* ", by Dr. Federico Herrera and his team published in the journal *Molecular Neurobiology*.

Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay (ARSACS) is an early-onset neurological disorder caused by mutations in the SACS gene, resulting in the loss of sacsin function. Sacsin is a multidomain protein that plays key roles in chaperone regulation, protein quality control, and neurofilament dynamics. Sacsin deficiency leads to disruption of intermediate filament and mitochondrial networks. S100B, a multifunctional brain-enriched protein, exhibits protective neuroprotective functions that include chaperone activity and interactions with filament proteins and mitochondria. In this study, we used an established astroglial C6 cell model of ARSACS to investigate the potential compensatory effects of S100B on sacsin loss with respect to neurofilament integrity and mitochondrial morphological and functional hallmarks. Our results demonstrate that sacsin deletion induces S100B upregulation at both mRNA and protein levels, with the S100B protein colocalizing with perinuclear nestin aggregates and filamentous mitochondria networks. Genetic silencing and pharmacological inhibition of S100B exacerbate filament protein aggregation and mitochondrial defects, while supplementation with exogenous recombinant S100B improves ARSACS hallmarks, including decreased nestin aggregates. These findings provide evidence for functional compensation of sacsin loss by S100B in glial cells, and suggests a potential role for glial cells in ARSACS.

Read the full article here

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A MOTHER'S PERSPECTIVE: LIVING WITH ARSACS AN INTERVIEW WITH MAXINE MONKS

In this edition of our newsletter, we are pleased to highlight a personal story from the ARSACS community. We spoke with Maxine Monks, whose daughter Olivia lives with ARSACS, to learn more about their journey, their hopes and their connection to the Foundation.

Q: When was Olivia diagnosed with ARSACS?

Maxine: Olivia was diagnosed during the COVID pandemic at the age of 24 years old. It was a long process — she spent time in the hospital where they ran a full battery of neurological tests. For months, we received letters ruling out various conditions, one by one. Each time we waited anxiously for the next result. Finally, the last letter arrived, and it confirmed the ARSACS diagnosis. It was overwhelming to get that news, but in some ways, it was also a relief to finally have an answer after such a difficult and uncertain period.

Q: How did you come across the Ataxia Charlevoix-Saguenay Foundation?

Maxine: After Olivia's diagnosis, I naturally started searching online for any information I could find about ARSACS. That's how I came across Betsy, another mother whose daughter also has ARSACS. She was so kind and helpful, and I later found out that she's a board member of the ARSACS Foundation. Connecting with Betsy and the Foundation gave me not just information, but a real sense of support and community — knowing that other families understood exactly what we were going through made such a difference.

Q: You are organizing the ARSACS Gala in the UK this October. What are you hoping to achieve with this event?

Maxine: For me, it's not just about raising funds, but also about bringing together families, researchers, and everyone who cares about the ARSACS cause. I hope the event will raise awareness, inspire greater support for research, and provide a space where people affected by ARSACS can connect and share their experiences. Creating this sense of community is so important when living with a rare disease like ARSACS. Learn more about the event

Q: What are your hopes for the future when it comes to ARSACS research and development?

Maxine: Like any parent, I hope that treatments (or even a cure) will be developed to slow or stop the progression of this disease. But I also think there are things that can be done right now to improve quality of life for families like ours. For example, I'd love to see every newly diagnosed family assigned a specialized nurse or care coordinator to help navigate the system from day one, along with access to regular physiotherapy without the financial burden. Small improvements in support and care would make a big difference while we wait for scientific breakthroughs.

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UPCOMING ARSACS CHARITY EVENTS

DONATE TODAY!

The *Diner des Producteurs* is the Foundation's most important financing event of the year — a special occasion that helps us advance research and support families affected by ARSACS.

Together, we can make a meaningful impact.

How You Can Contribute:

- ✓ Buy tickets or a table
- ✓ Make a donation
- ✓ Donate a product or service for our auction
- ✓ Sponsor the event

Learn more and get involved

Make an online donation

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MAKE YOUR VOICE HEARD

The Schmahmann Laboratory (Massachusetts General Hospital Ataxia Center, Boston) is conducting a study to validate a new tool for assessing emotional aspects related to the Cerebellar Cognitive and Affective Syndrome (CCAS).

>>> Who can participate?

- English-speaking adults (18+) diagnosed with any cerebellar disorder (which includes ARSACS)

>>> What is involved?

- One-time online participation (~15 minutes)
- Complete 2 short forms and a questionnaire
- 100% remote and voluntary

Click here to participate in the study

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UPCOMING CONFERENCES FOR ARSACS

We are proud to announce our participation to the World Orphan Drug Congress (WODC) Europe, which will be held this October in Amsterdam. A booth will be displayed to engage with attendees and raise awareness about our mission among international stakeholders in the rare disease community. Also, we will have the privilege of hosting two sessions:

October 28th, 4:50 PM

A gene therapy approach for ARSACS: insights from preclinical models – presented by Drs. Francesca Maltecca and Daniele Di Ritis.

April 29th, 2:50 PM

ARSACS : Finding Solutions for Rare Disease Patients – Sonia Gobeil, Maxine Monks and Dr. Bart Van de Warrenburg.

For more information on the World Orphan Drug Congress (WODC) or to buy tickets, visit this link.

MISSED THESE CONFERENCES? WATCH THEM NOW!

Sonia Gobeil, about ARSACS — watch the RareCheck interview

Gene therapy project for ARSACS featured at ASGCT — watch the presentation

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