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ARSACS NEWS

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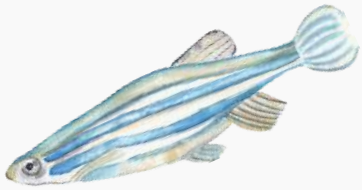
We are excited to share the latest updates from the Ataxia of Charlevoix-Saguenay Foundation. In this edition, you will find a newly published scientific article on ARSACS, details about the research projects funded by the Foundation for 2025–2026, and an invitation to participate in a Gene Therapy Survey for people with genetic ataxias. We also highlight upcoming webinars and conferences where you can connect with experts and learn more about ongoing research and therapeutic developments.

RESEARCH HIGHLIGHT



''*Long-term benefits of TUDCA supplement in ARSACS zebrafish model* '', by Dr. Valentina Naef and her team, published in *Scientific Reports*, a journal from the *Nature Publishing Group*, in July 2025.

Autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS) is an early-onset neurodevelopmental and neurodegenerative disorder characterized by ataxia, spasticity, and peripheral neuropathy. However, several studies have highlighted that some patients also experience cognitive, emotional and social deficits, suggesting a more complex clinical picture that extends beyond motor symptoms. Building on these findings, this study aimed to: (i) investigate locomotor, social and cognitive deficits in adult sacs-/- zebrafish versus wild-type (WT) controls through behavioural tests; (ii) identify molecular patterns associated with the adult disease phenotype using transcriptomic and proteomic analyses of sacs-/- and WT brains; (iii) evaluate the eơectiveness of long-term treatment with tauroursodeoxycholic acid (TUDCA) on behavioural outcomes and omics profiles in the zebrafish sacs-/- model. Our findings indicate impairments in cognitive, social, and emotional behaviors in aged sacs-/- zebrafish, which resemble some deficits observed in human patients. Transcriptomic and proteomic analyses of adult brains identified alterations in genes related to circadian rhythms and neuroin‫ammation. Notably, disruptions in sleep and circadian rhythms are frequently reported in individuals with cerebellar ataxia and may contribute to cognitive and behavioral changes. Long-term treatment with TUDCA, a neuroprotective molecule, was associated with partial improvements in social and cognitive behaviors and modifications in omics profiles in the zebrafish model. These results support the potential of further exploring TUDCA in future preclinical and clinical studies, while also emphasizing the need for additional investigations to better understand its mechanisms of action.

Read the full article here

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2025-2026 RESEARCH PROJECTS FUNDED BY THE FOUNDATION

The Ataxia of Charlevoix-Saguenay Foundation is proud to announce the funding of **15 innovative research projects for 2025–2026**. A total investment of **$1,288,097** will be dedicated to advancing the understanding of ARSACS mechanisms and supporting the development of new therapeutic avenues. This year, we wish to highlight the special financial contribution of the Richardson Trust Fund which contributed to the funding of three of these research projects.

"*Each year, we take another step forward toward a better understanding of ARSACS. These projects will bring together researchers from diverse scientific backgrounds to accelerate the discovery of promising treatments. By joining forces, we can transform hope into concrete solutions,*" said Jean Groleau, President and Co-Founder of the Ataxia of Charlevoix-Saguenay Foundation.

The funded projects span a wide range of disciplines, from fundamental research to emerging clinical approaches. This diversity ensures that ataxia is studied from multiple perspectives, increasing the chances of meaningful breakthroughs that will improve quality of life for patients.

For a complete description of the funded projects, please visit our website or contact Sonia Gobeil at ataxia@arsacs.com.

Congratulations to the 2025–2026 award-winning researchers and teams!

GENE THERAPY SURVEY FOR PEOPLE WITH GENETIC ATAXIAS

You are invited to participate in an **online survey**, about gene therapy for genetic ataxias (which includes ARSACS). The purpose of opinions about gene therapy research from people diagnosed with a genetic ataxia.

Gene therapy is a potential treatment for genetic ataxias, and as such the views of people with the condition are invaluable in the design of future research and clinical trials. The results of this research will provide information to the ataxia research community about opinions on gene therapy, which will help when designing future treatment trials.

People diagnosed with a genetic ataxia, or their family members, are eligible to complete the survey. Minors are required to complete the survey with a parent or guardian. We expect the survey will take roughly 30 minutes to complete, and the data collected will be anonymous.

If you have any questions about this research, contact the Foundation at ataxia@arsacs.com.

To access the survey, click here

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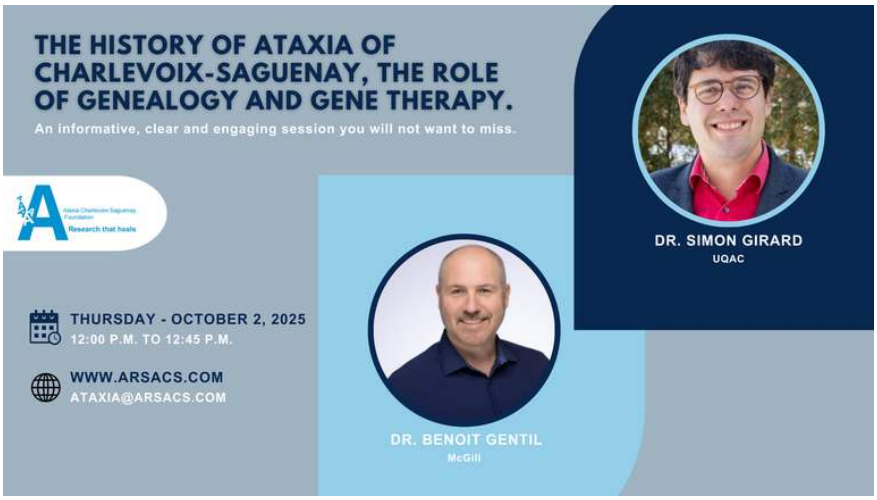
UPCOMING CONFERENCES FOR ARSACS

In collaboration with the Ataxia of Charlevoix-Saguenay Foundation, you are cordially invited to join the webinar presented by Drs. Benoit Gentil and Simon Girard.

They will discuss the identification of mutations around the world, how the ataxia of Charlevoix-Saguenay was identified, the prevalence of the SACS mutations and how genealogical data can support the development of gene therapy treatments. A Q&A session will also be included. In short, an informative, clear, and engaging session you will not want to miss.



Click here to join the LIVE Webinar at the scheduled date and time.

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, 4:10 PM

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

October 29th, 2:30 PM

ARSACS : Finding Solutions for Rare Disease Patients – presented by Sonia Gobeil, Drs. Bart Van de Warrenburg and Francesca Maltecca

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

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