



IMPORTANT NEWS FROM OUR PRESIDENT – Josh Marszalek

Just three months ago, the foundation was busy putting the final touches on our plans for the AHC 2020 Family Meeting to take place in beautiful California. As soon as the COVID-19 pandemic took shape, the foundation's board held meetings to eventually postpone the meeting until 2021. What an amazing three months it has been for everyone.



These are incredibly difficult and uncertain times for our country and the world. I am happy to reassure you that the foundation is busy working on your behalf & our wonderful community.

- We secured our family meeting in 2021 without incurring financial penalties
- We created an Emergency Room protocol to introduce AHC to first responders
- We brought on a new board member with an incredible skill set
- We updated the AHC-related research bibliography to keep you informed
- We met with all of our researchers to ensure the viability of our funded projects
- We are here for you, as always, to answer questions & provide help. Good news indeed.

NEW TOOL TO HELP AHC PATIENTS *Introduction to AHC for ER Doctors & Staff*

The AHC Foundation has published an **Emergency Room Protocol**. The intent of this document is to educate doctors unfamiliar with AHC and with your child's specific medical needs.



Especially in this time of COVID-19 viral infections, where your child may need to be admitted for treatment, this document will accompany your child and be passed off to care givers for their reference.

Vetted by our Medical Advisory Board, we are confident this will serve as a diverse and valuable tool for the ER, the school nurse, and other professionals.

Published in two formats:

PDF format for easy reference and WORD (.doc) format for customization of the last two pages to your specific circumstances. Simply download, customize, and print.

<http://ahckids.org/er-protocol-published/>

Also available here within our other resources at <http://ahckids.org/patient-care/ahcf-411/>

The AHC Foundation volunteers continually work on behalf of all AHC families to improve the quality of life of the patient and all those around them.

AHCF
Serving the
International
AHC community
since 1993



AHCF
Newsletter

Sharing
information
for advocates,
caregivers,
professionals
and families.



Thank you
to all of our
kind and
**generous
donors**
who made
donations to
the
foundation.

You make
all things
possible
for the
foundation
through your
generosity
and support.

EXCITING NEW ADDITION TO THE BOARD
Kate Byrne is the Latest Director to Join the Board

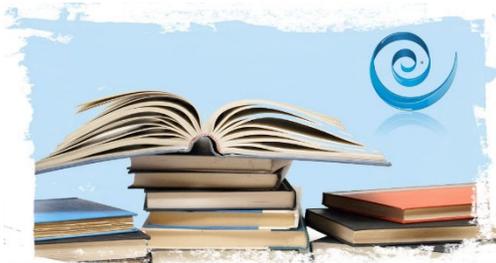
We are pleased to announce that Kate Byrne joined the AHCF Board of Directors this year.

Her son, Connor, was diagnosed with AHC in 2019 at 8 weeks old. Upon learning of Connor’s diagnosis, Kate and her husband Tom immediately decided to dedicate any time they could to increasing awareness and supporting fundraising efforts for the foundation. They live in Indiana with Connor and their young daughter Jane.



Kate works in Human Resources for the University of Kentucky and has a passion for promoting AHC awareness through social media and website development. She is also committed to working with resources in the Chicagoland area to plan fundraising opportunities and events increasing rare disease awareness. kate@ahckids.org

Please join us in welcoming Kate to the board.
 Read more about our enthusiastic board members here:
<http://ahckids.org/foundation/board-of-directors>



LOOKING FOR THE LATEST & GREATEST AHC-RELATED RESEARCH?
The AHC Foundation’s Research Bibliography is a Great Place to Start and is Now Updated & Online

Our Research Bibliography has been updated through 2019 and is available on our website!

The AHC Foundation is pleased to provide the AHC Community with a list of research articles published over the years on AHC-related topics. While this bibliography is in no way complete, it is a comprehensive record of academic and scientific publications relevant to our community. While all articles are not available in full text for free, sometimes the abstract will provide a quick look into what conclusions were drawn from the research.

Please use this resource to increase your understanding of our rare disease, and to empower yourself to better advocate for our kids.
<http://ahckids.org/resources/published-papers-directory/>



Our thanks and appreciation to Ryan Andrasco for designing our recent merchandise items AND to Kelly Andrasco for coordinating the selling of these items. Brother and mother to AHC patient, Kiley Andrasco, your volunteer efforts to help End AHC are greatly appreciated.

- AHCF 2020 Directors:**
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 - Lynn Egan
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 - Bill Gerber
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 - April Hawk
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 - Renee Hodes
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 - Meredith Schalick

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 - Dr. Zupanc

PUBLISHED RESEARCH SHEDS LIGHT ON AHC **Three New Papers Offer Important Insights**



New research relating to AHC is being published every month. It is with great excitement that we share with you a short abstract of three papers published in March and April. A link to the full text article is provided when available.

Neuronal Modeling of Alternating Hemiplegia of Childhood Reveals Transcriptional Compensation and Replicates a Trigger-induced Phenotype. On April 27th, in *Neurobiology of Disease*, a collaboration of researchers published this interesting article. Researchers you might recognize include Jon Snow, Kathy Swoboda, Al George, and Kevin Ess.

Mechanisms underlying patient episodes including environmental triggers remain poorly understood, and there are no empirically proven treatments for AHC. In this study, we generated patient-specific induced pluripotent stem cells (iPSCs) and isogenic controls for the E815K ATP1A3 mutation that causes the most phenotypically severe form of AHC.

Induction of cellular stress by elevated temperature revealed a hyperactivity phenotype following heat stress in ATP1A3+/E815K neurons compared to control lines. Treatment with flunarizine, a drug commonly used to prevent AHC episodes, did not impact this stress-triggered phenotype. These findings support the use of iPSC-derived neuronal cultures for studying complex neurodevelopmental conditions such as AHC and provide a platform for mechanistic discovery in a human disease model.

To read a full text copy of this article, go to:

<https://www.sciencedirect.com/science/article/pii/S096999612030156X?via%3Dihub>

Alternating Hemiplegia of Childhood: Understanding the Genotype-Phenotype Relationship of ATP1A3 Variations. On March 30th, in the *Application of Clinical Genetics*, a group of 4 researchers from Italy collaborated on this paper. Their objective was to summarize all clinical, genetic and molecular aspects of AHC considering ATP1A3 as its primary disease-causing determinant.

Most of the patients with ATP1A3 pathogenic variants fall into one of these phenotypes, which underlie a nearly non-overlapping subset of causative mutations. Nevertheless, some individuals show atypical features or combine features of two or more of these major phenotypes. On the other hand, some pathogenic variants have been reported to cause different phenotypes, even in the same family. As a result, it has been proposed to consider ATP1A3-related disorders as a clinical continuum rather than distinct entities, with an age-dependent pattern of emergence and progression of different signs and symptoms. **To read the full text of this article, go to:**

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC7125306/pdf/tacg-13-71.pdf>

Movement Disorders in Patients with Alternating Hemiplegia: "Soft" and "Stiff" at the Same Time. On March 31st, in the journal *Neurology*, a large coalition of researchers worked to assess nonparoxysmal movement disorders in ATP1A3 mutation-positive patients with alternating hemiplegia of childhood (AHC).

Twenty-eight patients underwent neurologic examination with a focus on movement phenomenology. Ten patients were diagnosed with chorea, 16 with dystonia, 4 with myoclonus, and 2 with ataxia. Nine patients had more than one movement disorder and 8 patients had none. The degree of movement disorder was moderate to severe in 12/28 patients. Patients with dystonia or chorea, or both, had earlier disease onset and more severe neurologic impairment, but this did not correlate with genotype. All patients presented with hypotonia, which was characterized as moderate or severe. **An abstract of this article can be found at:**

<https://n.neurology.org/content/94/13/e1378.long>

IOWA FAMILY SHARES AHC STORY WITH LOCAL COMMUNITY **Spreading the Word About AHC Earlier this Month with the Henderson's**



Stephen Henderson, an AHC dad from IOWA, had an opportunity to share the story of his family with their church via YouTube on May 6th.

His church has a broad reach in his area, and has been hosting live-streaming Wednesday night Bible studies. He was invited to share Estella's journey live on YouTube.

We thank Stephen, and his family, for sharing their personal AHC journey and increasing awareness about our community.

DO YOU WANT TO BE HEARD ABOUT COVID-19 and AHC? **NIH-Supported Research Survey Wants to Hear from You**

For the millions of people living with a rare disease, the novel coronavirus disease COVID-19 presents challenges, from potential reduced access to needed medical care to possible heightened anxiety and stress.



A new online survey launched by the National Institutes of Health-supported Rare Diseases Clinical Research Network (RDCRN) **aims to find out how the COVID-19 pandemic is impacting individuals with rare diseases, their families and their caregivers.**

Results will help the rare disease research community shed light on the needs of people with rare diseases during the COVID-19 pandemic and other potential health crises, in addition to informing future research efforts.

For more information on the COVID-19 survey, including how to participate, go to <https://www.rarediseasesnetwork.org/COVIDsurvey>.

COVID-19, Epilepsy Management, and Safety **With over 50% of AHC Patients with Epilepsy-This is Helpful Info**



An article was published in *Neurology* last month with the objective of providing information on the impact of the COVID-19 pandemic on people with epilepsy and provide consensus recommendations on how to provide the best possible care for people with epilepsy while avoiding visits to urgent care facilities and hospitalizations during the Novel Coronavirus pandemic.

Leading neurologists published two key suggestions on caring for people with epilepsy during the COVID-19 pandemic. First, as much care as possible should be administered at home, including the strategic use of rescue therapies, to keep people out of health care facilities, where they are more likely to encounter COVID-19. Second, to minimize the risk of increased seizure severity or frequency, physicians should ensure that their patients adhere to treatment plans and have a regular supply of medication.

The complete article can be found at the following link:

<https://n.neurology.org/content/neurology/early/2020/04/22/WNL.0000000000009632.full.pdf>