



One Mission: End AHC!



YOUR ALTERNATING HEMIPLEGIA OF CHILDHOOD FOUNDATION NEWSLETTER

June/July 2017

2000 Town Center ■ Suite 1900 ■ Southfield, Michigan 48075

AHCF in ACTION **Board of Directors Hard at Work**

The AHCF Board of Directors gathered in Chicago this month for the Annual Board of Directors Meeting. It is the first time we met in person in a year without a family meeting.



We met as a group to discuss the financial operation of the foundation, funding research, and initiating new projects. We also met in small group strategy sessions focused on three topics, connecting with our community, finding new ways to keep the foundation moving forward, and methods for serving our community.

Many of the discussions led to exciting new opportunities for the foundation in the near future and leading up to the next family meeting in New Jersey. Be sure to keep your eyes peeled for announcements in the weeks and months ahead. There will be many reasons to join the AHC Foundation on our **One Mission: End AHC!**

Lynn Egan, AHCF President



AHCF in ACTION **AHCF Webinar Coming in August**

The next AHCF webinar will take place on **August 29, 2017 from 6:30 pm to 8:00 pm (CDT)**. Dr. Terry Sanger will present a talk on dystonia with a question and answer period for parents to **learn what dystonia is and how to deal with it.**

Dr. Sanger is an Electrical Engineer and Child Neurologist specializing in movement disorders of children. His NIH funded research includes work on **understanding motor learning in children**, work on the use of kinematic measurements of children to design assistive communication interfaces, and multiscale modeling of large-scale neural systems for control, with particular application to **understanding the development of spasticity and dystonia**. He is the director of the Child Movement Disorders Clinic at Children's Hospital of Los Angeles. His laboratory has made important recent discoveries on the role of long-latency stretch reflexes in the genesis of childhood secondary dystonia.

Among his many awards are: Dystonia Medical Research Foundation Millennium award, Pfizer scholars in pediatric research award, United Cerebral Palsy Leaves of Hope award, Hume scholars award, Stanford Biodesign fellowship teaching award, American Academy of Cerebral Palsy and Developmental Medicine Goldenson award.

The AHC Foundation is thrilled to offer this opportunity to our community. It is through the hard work of **everyone in the AHC community** that will help us reach our **One Mission: End AHC! Watch for call-in directions coming in August.**

Funding Research & Supporting Families.
The AHCF-serving the AHC community since 1993.



AHCF in ACTION

Three Projects Funded to Help END AHC

Vanderbilt/Northwestern Phase “6” - The AHCF is pleased to announce an award of \$132,535.00 to Northwestern University, plus an award of \$115,135.00 to Vanderbilt University School of Medicine.

An award of \$132,535.00 was made to the Department of Pharmacology, Northwestern University, Feinberg School of Medicine with Dr. Alfred George as primary investigator, plus an award of \$115,135.00 to the Division of Pediatric Neurology, Department of Medicine, Vanderbilt University School of Medicine with Dr. Kevin Ess as primary investigator. They will be working together on the project **“Molecular Physiology and Pharmacology of ATP1A3 Mutations in AHC”** known as **“phase six”**. The period of support is July 1, 2017 to June 30, 2018 and totals \$247,670.00.

This project will continue **investigating the cellular and molecular defects in AHC by studying human neurons generated** from patient-derived stem cells coupled with pharmacological strategies to correct the defect. **The team will also use gene editing to correct ATP1A3 mutations in patient-derived stem cells** and create a ‘knock-out’ stem cell line useful for investigating AHC causing mechanisms.

The investigators have made great progress identifying a central electrophysiological deficit in AHC neurons carrying the G947R mutation and are now turning attention to studying neurons with other common and less common mutations. In addition, the team is testing drugs and compounds for their ability to rescue the primary cellular defect as **a first step toward identifying new potential treatments for AHC.**

Antibody Acceleration Project: AHCF also has funded an acceleration project in the amount of \$62,237 awarded to Dr. Kevin Ess and his team at Vanderbilt to generate and validate highly specific



antibodies targeted against the alpha 3 and alpha 2 subunits of the Na/K ATPase. Dr. Ess says **“Such tools will greatly accelerate our research** as we will be able to be accurately assessing levels of these important proteins. We expect them to not only work for our human stem cell based models of AHC, but given the extremely high sequence homology, to also be useful for mouse models of AHC/RDP/CAPOS as well. We expect to generate unlimited and eventually very inexpensive antibodies. **As we will share these tools with other scientists throughout the world, this should be an excellent investment that should continue to pay dividends far into the future.”**



Mouse Models: In addition to the above grants, AHCF is at the halfway point of a grant which **implements two different mouse models of AHC caused by distinct ATP1A3 mutations (D801Y, G947R)** which will enable basic investigations of this disease.

These mouse models will be used to **test the effectiveness of various approved drugs** and experimental compounds to alleviate symptoms of the disease; and 2) to **identify genetic factors** other than the primary ATP1A3 mutation that influences severity of symptoms (hemiplegia, dystonia) as well as the propensity for epilepsy.

The findings will contribute to the discovery of better pharmacological treatments for AHC and reveal new disease mechanisms that could be exploited for novel therapeutic strategies.

AHCF in ACTION

AHCF Starts Grant Program for Families

The AHCF is excited to announce our newly created Family Grant program.

This program was established by the AHCF Board as a way to alleviate some of the financial challenges faced by those with AHC and their families.



Eligibility:

Grant consideration is limited to individuals diagnosed with Alternating Hemiplegia of Childhood and their immediate family

Grant Criteria:

Grants can be awarded either as reimbursement for an approved purchase or paid directly to an individual. Depending on the situation, the AHCF may require an explanation of attempts to use other available resources.

Grants can be used for but not limited to:

Transportation for a medical appointment to see a specialist not available locally
 Transportation for medical testing not available locally
 Transportation, or other expenses, to the AHCF Family Meeting that creates a hardship to attend
 Medical equipment deemed necessary but not covered by insurance or local organizations

Grants will not be awarded for the following:

Transportation to other medical conferences
 Medical equipment that can be covered by private insurance or Medicaid
 Vacations
 Service dogs

Applications for the AHC Family Grant can be found on our website.

If you have questions, please contact Shannon@ahckids.org



AHC INFO EXCHANGE

Important AHC Research Just Published

A group of Italian researchers are publishing an article next month in the *Journal of the Neurological Sciences* titled, "ATP1A3 Mutant Patient with Alternating Hemiplegia of Childhood and Brain Spectroscopic Abnormalities."

The article will look at three issues:

- Long standing follow-up of this AHC patient with cerebellar atrophy.
- ATP1A3 gene mutation was associated with metabolic energy failure.
- Brain spectroscopy and muscle biopsy were used to characterize metabolic pathways.

A free copy of the article is not available. To purchase a copy, go to:

<http://www.sciencedirect.com/science/article/pii/S0022510X17303374>

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AHC ANSWERS

AHCF Medical Advisory Board Answers Your Questions



This month's question was posed to AHC Expert Dr. Terry Sanger and is about ocular dystonia.

Question: "A lot of discussion has been happening lately on our Facebook Support group about abnormal ocular movements. Some parents have described their kids eyes as "stuck", is this dystonia? What can you tell me about ocular dystonia? Is it part of an AHC episode? Is there any way to relieve it?"

Answer: It is possible to have unusual ocular episodes where the eyeball appears to be stuck up in one's head (oculogyric crisis); **emotional stress, fatigue and some medications may bring on an attack.** It also can be described sometimes as a fixed stare. It is the most common dystonic ocular occurrence and is frequently found in Parkinsonism, but since AHC has such a wide variety of presentations it is likely that this is part of an AHC episode.

Another unusual dystonic ocular occurrence is **Blepharospasm** - Blepharospasm means "eyelid". Spasm is an "uncontrolled muscle contraction". Blepharospasm can be applied to any abnormal blinking or eyelid tic or twitch resulting from any cause, it is both a cranial and a focal dystonia. It is described as abnormal involuntary sustained muscle contractions and spasms. Patients with blepharospasm have normal eyes. The visual disturbance is due solely to the forced closure of the eyelids.

Although not a typical AHC symptom, it is sometimes brought on by **bright lights, wind, pollution, smoke, emotional stress and fatigue.** It usually occurs in middle aged or older adults but again since AHC is so unpredictable it is not out of the realm of possibilities that is caused by AHC. If it is a long term problem, most patients can control it by medication and sensory adaptations.

To learn more about dystonia and ask all your questions; **please join us on August 29th** for a webinar on dystonia by Dr. Terry Sanger from UCLA; a renown dystonia specialist.



The AHC Foundation is incredibly fortunate to have **12 internationally recognized AHC experts** serving on our Medical Advisory Board (MAB) **from four different countries.** In this column, your questions are answered by one or more members of the MAB.

To have your question answered, contact sharon@ahckids.org



SAVE THE DATE
AHC FOUNDATION
2018 FAMILY MEETING
June 22nd – June 24th, 2018
Renaissance Marriott, Woodbridge, NJ