



# One Mission: End AHC!



YOUR ALTERNATING HEMIPLEGIA OF CHILDHOOD FOUNDATION NEWSLETTER

April 2018

2000 Town Center ■ Suite 1900 ■ Southfield, Michigan 48075



## **A MESSAGE FROM THE PRESIDENT** **Big News to Share in this Newsletter**

It is with great excitement that I get to share with all of our supporters the great news of a new research publication! Because of the hard work of the last several years, **groundbreaking research has just been published.**

It is also very exciting to see families registering for **the AHC Family Meeting that is just two months away.** For families that have been to meetings in the past, there will be plenty of new material presented. For first-time families, there are a variety of opportunities for you to meet and socialize with AHC specialists.

I hope you read about the latest AHC research on page 2 and that you will be able to join us in New Jersey this June.

Lynn Egan  
President and Family Support

## **AHCF IN ACTION – FAMILY MEETING** **Make Your Plans Now to be in New Jersey this June**



The AHC Foundation has been **hosting a Family Meeting every other year for almost two decades.** We are excited to be hosting another fabulous meeting this year at the Renaissance Woodbridge Hotel in Iselin, New Jersey.

On June 21st, informal consultation **appointments with AHC specialists** will be scheduled in the morning. In the afternoon, a workshop will take place with Dr. Swoboda, Dr. Kevin Ess and several members of the AHCF Medical Advisory Board.

The conference meetings will take place June 22 – 24 with a variety of **scientific and daily presentations** as well as **fun social activities.** Meet or reunite with AHC families from around the world. This is the best opportunity you will have to **have your AHC questions answered** by those with the most AHC experience.



The latest information on how to register for the hotel and conference can be found on the meeting website: [ahcffamilymeeting.org](http://ahcffamilymeeting.org)

Everyone is encouraged to attend. **Come as a family.** Come as a couple and have some “parent time.” **Invite a family member** to join you and learn more about AHC. **Come by yourself.** We always have a group of parents that come by themselves since one parent needs to stay home with the kiddos.

Whether you will be coming as a family or a single parent, you are sure to **have fun, meet great families, and learn from the best AHC specialists** in the world. Come to New Jersey and join the AHC Community in June 2018.

**Join in on the fun with your friends or family at one of our upcoming events**

## AHC INFO EXCHANGE

### Major AHC Research Breakthrough Published Online in March

In the online version of *Neurobiology of Disease*, the ground breaking article, “**Direct Evidence of Impaired Neuronal Na/K-ATPase Pump Function in Alternating Hemiplegia of Childhood,**” was published on March 19, 2018.



The article is the **direct result of research funded by the AHC Foundation** and covers an incredible amount of work done by doctors **Kevin Ess, Al George, Christine Simmons, and Kathryn Swoboda.**

The article discussed how most AHC cases are explained by de novo heterozygous ATP1A3 mutation, but the fundamental **molecular and cellular consequences of these mutations in human neurons are not known.** In this study, they investigated the electrophysiological properties of neurons generated from AHC patient-specific **induced pluripotent stem cells (iPSCs)** to ascertain functional disturbances underlying this neurological disease.

#### What are iPSC cells?

iPSC are derived from skin or blood cells that have been reprogrammed back into an embryonic-like pluripotent state that enables the development of an unlimited source of any type of human cell needed for therapeutic purposes.

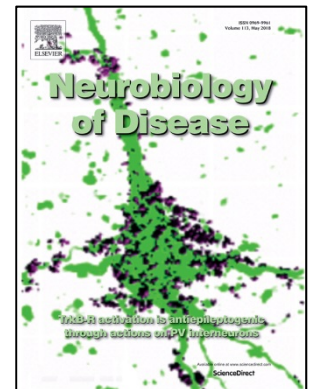
Fibroblasts derived from two subjects with AHC, a male and a female, both heterozygous for **the common ATP1A3 mutation G947R, were reprogrammed to iPSCs.** Neuronal differentiation of iPSCs was initiated by neurogenin-2 (NGN2) induction followed by co-culture with mouse glial cells to promote maturation of cortical excitatory neurons. Whole-cell current clamp recording demonstrated that, compared with control iPSC-derived neurons, neurons differentiated from AHC iPSCs exhibited a significantly lower level of ouabain-sensitive outward current (‘pump current’).

This finding correlated with significantly depolarized potassium equilibrium potential and depolarized resting membrane potential in AHC neurons compared with control neurons.

In this cellular model, they also observed a **lower evoked action potential firing frequency when neurons were held at their resting potential.** However, evoked action potential firing frequencies were not different between AHC and control neurons when the membrane potential was clamped to -80 mV.

Impaired neuronal excitability could be explained by lower voltage-gated sodium channel availability at the depolarized membrane potential observed in AHC neurons.

**These findings provide direct evidence of impaired neuronal Na/K-ATPase ion transport activity in human AHC neurons and demonstrate the potential impact of this genetic defect on cellular excitability.**



**Come Hear About this Research in Person at the New Jersey Family Meeting.**

Through research, education and family support, we have **ONE MISSION: END AHC!**

## *AHCF IN ACTION – GETTING TO THE FAMILY MEETING*

### 9 Tips to Help Kids Avoid Travel Meltdowns

**Traveling with kids can be tough no matter what.** Learn how to make three different types of travel kits packed with tools that can calm a child who is overwhelmed. Also, explore tips to help you anticipate your child's needs and avoid vacation meltdowns.



**1-Bring a self-regulation survival pack.** If your child has visual sensitivities or is sensitive to sounds, textures or smells, you might have already found tools that help him. Fill a small backpack with the items he might need (such as sunglasses, earplugs and fidgets). Keep it within his reach in the car or on the plane, so he can grab what he needs even if your focus is elsewhere.

**2-Pack familiar toiletries.** While it's nice that hotels and family provide shampoo, soap, toothpastes and even towels, your child may be overwhelmed by the new textures, smells or tastes. Pack the toiletries he's used to and the towels you know he'll use. It's a simple way to avoid a sensory meltdown and to make your child feel more at home.



**3-Practice your trip.** For older kids, this may mean looking over the route, routines and schedule for your trip. Younger kids may benefit from a pretend practice run. Make your home into an "airport" or "museum," and walk through what might happen there.

**4-Stop for frequent breaks during car trips.** Enough said.

**5-Give yourself extra time.** Schedule some extra time to get to the airport or your destination. It can reduce the chances that your child will feel overloaded by the stress of hurrying through a new situation. It may also prevent an anxiety-related meltdown.

**6-Look for quiet corners during airport waits.** For some kids, the noise of airport announcements, people rushing to catch flights and planes taking off may be too much. Try taking a walk around the terminal to look for a quiet corner. Many airports also have activity centers, which your child might enjoy if they're not too crowded.

**7-Be strategic in your boarding options.** Many airlines make accommodations for families that need extra support. Call ahead to see what can be done to help. Some airlines offer pre-boarding, but if your child might do better getting on the plane after everybody, ask if that's an option.



**8-Bring familiar foods with you.** Be prepared if your child is sensitive to tastes and food textures or tends to melt down when he gets hungry. Bring along foods you know he likes instead of asking him to adapt to new ones. You may have to arrange to have a fridge in your hotel room.

**9-Follow the same routine as you do at home.** Help your child go to sleep and wake up at the same time he does at home. Follow the same rules. If your child gets cranky at night, you might want to finish the day's driving before dinnertime. If mornings are tough, you might want to avoid an early departure.

## **AHC COMMUNITY CONNECTIONS**

### **Sad News for Our Global Community**

The AHC Community mourns the loss of another amazing AHC patient.

It is with great sadness to inform you that Garret Hafetepe passed on Friday April 6. Garret was 33 years old and lived in Texas with his Dad.

Please join us in expressing our condolences to Garret's family.



## **AHCF IN ACTION – GETTING TO THE FAMILY MEETING**

### **The AHC Foundation Has a Way to Help for Those with Financial Need**



**The AHCF Family Grant** is available to families needing financial assistance to attend to the AHC Family Meeting in New Jersey. If interested please reach out to Shannon@ahckids.org for an application. Applications must be received by May 1<sup>st</sup> to be considered for the Family Meeting this June. **Grants will be reviewed by a committee and awarded as available.**

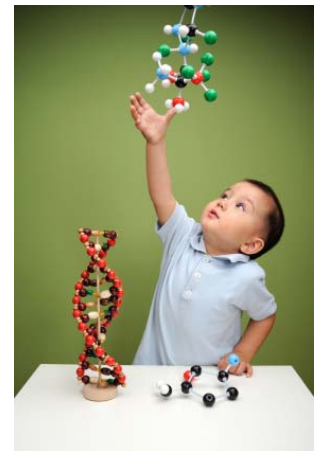
## **AHC INFO EXCHANGE**

### **ATP1A2 Research Published this Month**

In the April 2018 edition of *Pediatrics*, Ueda, Serajee, and Hug published an article titled, “Clinical Benefit of NMDA Receptor Antagonists in a Patient With ATP1A2 Gene Mutation.” **They hypothesized that glutamatergic excitotoxicity due to the ATP1A2 mutation contributed to the pathogenesis of their patient's condition.**

Mutations in the ATP1A2 gene cause familial hemiplegic migraine type 2, alternating hemiplegia of childhood, and cerebellar function deficits, epilepsy, and mental retardation.

The patient was started on N-methyl-D-aspartate receptor antagonists (memantine and dextromethorphan), as well as coenzyme Q10. One year later, he showed significant improvement in sustained attention, learning efficiency, general cognitive efficiency, and fine motor dexterity. They postulated that N-methyl-D-aspartate receptor antagonists were effective for behavioral, cognitive, and cerebellar symptoms in our patient with ATP1A2 gene mutation.



**The AHCF is grateful to all of the researchers working towards our One Mission: End AHC!**

We are looking for suggestions on which topics to present during future webinars.

If you have ideas about what would be most helpful to you and other AHC families please let us know.