



One Mission: End AHC!



YOUR ALTERNATING HEMIPLEGIA OF CHILDHOOD FOUNDATION NEWSLETTER

December 2016

2000 Town Center ■ Suite 1900 ■ Southfield, Michigan 48075

AHCF in Action Holiday message from Lynn Egan

The AHC Foundation has been working hard to better the lives of everyone in the AHC community for over twenty years. As this year draws to a close, we thank each of you for helping us get even closer to achieving our **One Mission: End AHC**.



While everyone is coming together for the holidays this year, we hope your celebrations will be filled with joy and good health. Thank you for continuing your support of the AHC Foundation and renewing your commitment to help End AHC in 2017.

**The Board of Directors and I wish you and your family a wonderful Holiday Season.
Lynn Egan, AHCF President and Parent.**



DOUBLE the FUNdraising **The holidays are a perfect time to help End AHC!**

List of potential projects waiting for funding

\$150,000	Dr. Kenneth Silver Univ. of Chicago Pharmaceutical Treatment
\$130,000	Dr. Alfred George Northwestern Univ. Develop Mouse Models
\$120,000	Dr. Kevin Ess Vanderbilt Univ. Accelerate Treatment Research
\$50,000	Dr. Kathryn Swoboda Harvard Univ. Database for Drug Trial Preparation
\$50,000	AHC Episode Tracker International Improve Quality of Life
\$10,000	Alice Belgrade U.S. based Behavior Study

\$510,000 Total

It is very exciting times in terms of research.

Right now we have the problem of how to raise enough money to fund it all of the potential projects that have been put in front of the foundation.

We currently have **\$510,000 in research grant requests** that we can't afford to fund.

Our end of year goal is to raise \$150K that will help us begin to fund these projects.

Doctors are excited about the discovery of the ATP1A3 gene and we want to ensure we act upon that excitement. If we don't fund the research when they have time, we will lose valuable **opportunities to End AHC!**

Our year-end campaign has kicked off and needs your help! Step Up to END AHC is a call to action for families to raise, not donate, money to fund research that will help to End AHC. We made it easy to raise as much or as little money as you can while sharing your own AHC story at the same time.

When you support the **Step Up to End AHC** campaign, you are **helping the AHCF achieve its mission** to fund research, promote year-round education, and provide families with much needed support.

www.ahckids.org

PATH to a CURE AHC campaign underway

It's time to join the momentum!

We are half way thru our Step Up to END AHC campaign! Thank you to everyone who has created a page, a team or made a donation!

As of Dec. 12, we have 67 supporters, 34 teams and 367 donors and because of you we have raised \$49,076! We are nearly 1/3 of the way to our goal of \$150,000!

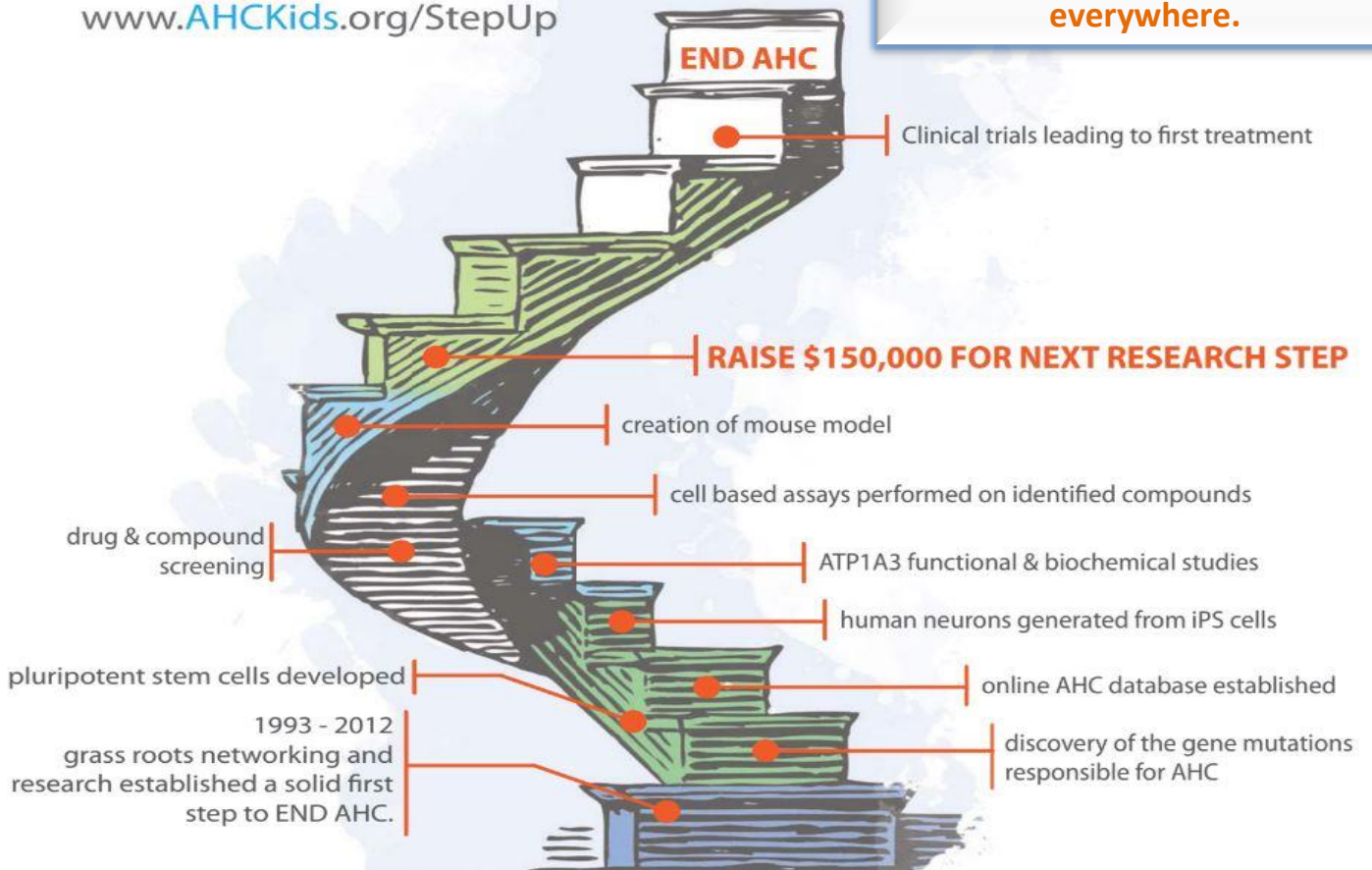
Remember to share your page with your network. With each donation you collect, you not only contribute to your child's future, but you are also raising awareness of AHC.

Step Up to END AHC...It's not too late to start your page or make a donation!



With your help, we can take advantage of these new opportunities which can bring real solutions to AHC patients everywhere.

www.AHCKids.org/StepUp



To join in the campaign, see the next pages for instructions and ideas!

PATH to a CURE

Joining the AHC campaign is simple and fast for everyone

To take the next steps to a treatment, we are asking for your help.

Go to <http://www.firstgiving.com/AHCF/step-up-to-end-ahc>

Click on **FUNDRAISE** and **create a personal page or join a team**

Create a personal fundraising page - This is your personal page where you can tell your story. You are encouraged to share the ups and downs of living with AHC. Really let people know why it is important to end AHC!

Add a photo and set a fundraising goal. **You can add a goal** for your team or leave it blank and each person who becomes part of your team set's their goal.

When people ask to join your team, remember to send them the **link to your team page**. This is where it will ask them to 'join team' and they will need to also create an account, join your team. They don't need to tell a story unless they want to, but if left blank, there is a generic one that populates.



This is an existing team page. You can customize one for your family, set your own goals and tell your own story.

Joining us is easy. Go to <http://www.firstgiving.com/AHCF/step-up-to-end-ahc>



AHC in the News

AHC family – a cut above the rest

On November 10th, **Live 5 News** in South Carolina reported on an AHC family making a huge difference for the special needs community. **The story centered on Danielle Lee**, who has a daughter with AHC, as well as a nephew with cerebral palsy.

In August, a new spa and hair salon opened in Conway, South Carolina. **Once a month, the salon focuses on haircuts for children with special needs.** "People don't realize that unless you have a special needs child, or unless you have a special needs person in your family, like how hard it is to truly get a haircut," Lee said.

The article reported that Danielle was inspired to give back to her community after finding out she was in remission from leukemia in 2010. Her initial diagnosis came in 2008. "May 22, I thought my dreams were shattered," Lee said. "I thought there would never be anything that I would ever want to accomplish."

Danielle survived and she did it by helping others. Congratulations Danielle and continued good health from all of us in the AHC Community!

AHC in the News

AHC family makes local news

On November 23, the *Cherry Hill Sun*'s headline read, "Cherry Hill mother determined to help daughter afflicted with rare disorder."

The article shared Meredith and Emilia Schalick's story about living with AHC. For Emilia, her biggest trigger is a change in the weather. Meredith was happy to share that Emilia receives great support and encouragement from her classmates at Barclay Early Childhood Center.



Meredith has recently spent a lot of time and energy fundraising. In November Meredith has raised more than \$2,600 for the **Step Up to End AHC campaign**. To raise more funds, the Schalick family is partnering with Painting with a Twist in Haddonfield to hold a fundraiser on Dec. 1st.

Through fundraising with the Alternating Hemiplegia of Childhood Foundation, Meredith is determined to help support research efforts so better treatment and a cure may be found for her daughter and others afflicted with the disorder. **Our thanks go to the Schalicks for sharing your story with the community and helping to find solutions for all AHC patients.**



AHC in the News

AHC international research consortium at work in Europe

On October 26, 2016, **Rosaria Vavassori and Tsveta Schyns** gave a presentation at the **EuropaBio's Patients Bio-Forum**. The forum was a closed-door event which gathered representatives from European patients groups and industry as well as other stakeholders (e.g. regulators, policymakers, academics, etc.) to discuss patient-centered issues in healthcare biotechnology. This recurring European event allows stakeholders to present their perspectives on a given topic, while engaging in constructive and inquisitive round-table discussion.

Rosaria and Tsveta **gave a thorough presentation regarding the IAHCRC Cloud Registry Platform**. This specific platform is a network of interconnected registries serving the international studies of the IAHCRC Consortium.

Patient registries have long been used in academic settings to generate observational data for research purposes. However, there has been **a recent shift** towards utilizing patient registries for more than just traditional recruitment or sample collection, and **towards opportunities to explore population behavior, product usage as well as monitoring quality of healthcare.**

With the multitude of different types of registries that exist today, it is important that we optimize the use of these patient registries and understand the link between the importance of setting up high quality registries with different steps of the drug development cycle. **Great job Rosaria & Tsveta!**

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

INFO EXCHANGE

Important research just published

Below you will find a number of recent articles relevant to the AHC Community. The text is copied directly from the article's abstract and the full citation is provided for your reference. Be sure to discuss any specific questions you have with your physicians.



“Ketogenic diet and childhood neurological disorders other than epilepsy: an overview.” *Expert Review of Neurotherapeutics*, 2016 Nov 21, pp. 1-13.

In the last years, ketogenic diet (KD) has been experimentally utilized in various childhood neurologic disorders such as mitochondriopathies, alternating hemiplegia of childhood (AHC), brain tumors, migraine, and autism spectrum disorder (ASD). The aim of this review was to analyze how KD can target these different medical conditions, highlighting possible mechanisms involved. Recently, few cases suggest the potentiality of KD in decreasing paroxysmal activity in children affected by AHC. A few data support its potential use as co-adjuvant and alternative therapeutic option for brain cancer, while any beneficial effect of KD on migraine remains unclear. KD could improve cognitive and social skills in a subset of children with ASD.

“Recognizable Facial Features in Patients with Alternating Hemiplegia of Childhood.” Gurrieri F, Tiziano FD, Zampino G, Neri G. *American Journal of Medical Genetics, Part A*. 2016 Oct, Vol. 170 No. 10, pp. 2698-705.


While the neurological profile of this condition is well defined, the question whether a recognizable pattern of physical anomalies does exist in this condition is still open. We performed a morphological evaluation of 30 patients at different ages. All patients were evaluated independently by each author and evaluation sheets were compared, discussed, and agreed afterwards. On physical ground, almost all patients shared a similar physical phenotype consisting of hypotonia, long face, thin eyebrows, strabismus, hypertelorism, long palpebral fissures, downturned mouth, and slender habitus.



“Mosaicism in ATP1A3-Related Disorders: Not Just a Theoretical Risk.” Hully M, Ropars J, Hubert L, Boddaert N, Rio M, et al. *Neurogenetics*, 2016 Oct 10.

Mutations in ATP1A3 are involved in a large spectrum of neurological disorders, including RDP, AHC, and CAPOS. In AHC, a few familial cases of autosomal dominant inheritance have been reported, along with cases of de novo sporadic mutations. In contrast, autosomal dominant inheritance has frequently been

associated with RDP and CAPOS. To our knowledge, mosaicism has not previously been reported in ATP1A3-related disorders. This report, therefore, provides evidence that germline mosaicism for ATP1A3 mutations is a likely explanation for familial recurrence and should be considered during recurrence risk counseling for families of children with ATP1A3-related disorders.

 <p>I just supported Alternating Hemiplegia of Childhood Foundation Inc at AmazonSmile. I shop. Amazon gives.</p> <p>When you shop at AmazonSmile, Amazon donates to... SMILE.AMAZON.COM</p>	<p>When you shop online, consider using AmazonSmile (smile.amazon.com). The AmazonSmile Foundation will donate 0.5% of the price of eligible purchases to the AHC Foundation. It is that easy.</p> <p>Simply go to smile.amazon.com, select AHC Foundation, and make your purchases. Amazon does the rest.</p>
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INFO EXCHANGE

Research summaries continued

“De Novo p.Arg756Cys Mutation of ATP1A3 Causes an Atypical Form of Alternating Hemiplegia of Childhood with Prolonged Paralysis and Choreoathetosis.” Kanemasa H, Fukai R, Sakai Y, Torio M, Miyake N, Lee S, Ono H, Akamine S, Nishiyama K, Sanefuji M, Ishizaki, Y Torisu H, Saitsu H, Matsumoto N, Hara T. *BMC Neurology*, 2016 Sep 15, Vol. 16, pp.174.



A 7-year-old boy presented with recurrent symptoms of generalized paralysis since 1 year and 5 months of age. Hypotonia, dystonia, and choreoathetosis persisted with exacerbation under febrile conditions, but no cerebellar ataxia had ever evolved in 6 years. Whole-exome sequencing (WES) was performed to determine his genetic background. WES identified a de novo pathogenic mutation in ATP1A3 (c.2266C > T:p.R756C) for this patient. A literature overview of two reported cases with p.R756C and p.R756H mutations showed both overlapping and distinct phenotypes when compared with those of the present case. Conclusions: This study confirmed that p.R756C mutation of ATP1A3 cause atypical forms of AHC-associated disorders. The wide spectra of neurological phenotypes in AHC are linked to as-yet-unknown deficits in the functions of mutant ATP1A3.

“A Novel de Novo Mutation in ATP1A3 and Childhood-onset Schizophrenia.” Smedemark-Margulies N, Brownstein CA, Vargas S, Tembulkar SK, Towne, MC, Shi J, et. al. *Cold Spring Harb Mol Case Stud*, 2016 Sept, Vol. 2, No.5, pp.1008.

We describe a child with onset of command auditory hallucinations and behavioral regression at 6 yr of age in the context of longer standing selective mutism, aggression, and mild motor delays. We discuss the literature on phenotypes associated with known variants in ATP1A3, examine past functional studies of the role of ATP1A3 in neuronal function, and describe a novel clinical presentation associated with mutation of this gene.



“Flunarizine Versus Topiramate for Chronic Migraine Prophylaxis: A Randomized Trial.” Lai KL, Niddam DM, Fuh JL, Chen SP, Wang YF, Chen WT, Wu JC, Wang SJ. *Acta Neurologica Scandinavica*, 2016, Jun 16.

Chronic migraine (CM) is a prevalent and devastating disorder with limited therapeutic options. This study explored the efficacy of 10 mg/d flunarizine for CM prophylaxis as compared with 50 mg/d topiramate.

Results: Sixty-two subjects were randomized (n=31/group). Patients treated with flunarizine showed significant reductions in the numbers of total headache days (-4.9 vs -2.3, P=.012) and migraine days (-4.3 vs -1.4, P=.001) compared with those treated with topiramate. Patients treated with flunarizine also showed significant reductions in the numbers of days of acute abortive medication intake (-2.3 vs -0.2, P=.005) and acute abortive medication tablets taken (-4.6 vs -0.5, P=.005) and had a higher 50% responder rate in terms of total headache days (58.6% vs 25.9%, P=.013) and migraine days (75.9% vs 29.6%, P=.001), compared with topiramate-treated patients. Flunarizine was generally well tolerated and had a safety profile comparable to that of topiramate.

Conclusions: Our results suggest that, in an 8-week study, 10 mg/d flunarizine is more effective than 50 mg/d topiramate for CM prophylaxis.

Your Dollars Matter Most. Our foundation is 100% donor-supported. Help improve the quality of life of children with AHC today... with your donation.