One Mission: End AHC!

YOUR ALTERNATING HEMIPLEGIA OF CHILDHOOD FOUNDATION NEWSLETTER March 2017

2000 Town Center ▪ Suite 1900 ▪ Southfield, Michigan 48075

AHCF in ACTION
Message from President Lynn Egan

Last month, we celebrated Rare Disease Day, a day to raise awareness about AHC among the general public and decision-makers. There were many of you in the AHC community who also did your part and **shared your AHC story**.

Momentum is always essential to any organization to keep raising awareness, moving research forward, and **to find answers which ultimately lead to a treatment and a cure for AHC.** Thanks to everyone, the AHCF is continuing forward with such momentum.

We have many announcements in the weeks to come and want to be sure you are up to date.
So, make sure we have your current email address,
Like us on Facebook http://www.facebook.com/AHCKids/
Join the AHC Support Group (a private chat site for parents only)
www.facebook.com/groups/1436035426663331/,
And follow us on twitter @AHC Kids.

AHC INFO EXCHANGE
Major new AHC study published with AHCF help

Neurology® An Official Journal of the American Academy of Neurology — Genetics AN OPEN ACCESS JOURNAL

On March 2, 2017, an article was published in *Neurology Genetics* titled, **"Research Conference Summary from the 2014 International Task Force on ATP1A3-Related Disorders."** Authors of the study include 9 members of the AHCF Medical Advisory Board and 5 members of the AHCF Board of Directors.

In 2014, the **AHC Foundation hosted a multidisciplinary workshop** intended to address fundamental challenges surrounding the diagnosis and management of individuals with ATP1A3-related disorders.

Workshop attendees were charged with the following: **(1)** to achieve consensus on expanded diagnostic criteria to facilitate the identification of additional patients, intended to supplement existing syndrome-specific diagnostic paradigms; **(2)** to standardize definitions for the broad range of paroxysmal manifestations associated with AHC to disseminate to families; **(3)** to create clinical recommendations for common recurrent issues facing families and medical care providers; **(4)** to review data related to the death of individuals in database to guide future efforts in identifying at-risk subjects and potential preventative measures; and **(5)** to identify critical gaps where we most need to focus national and international research efforts.

This **report summarizes recommendations of the workshop committee**, highlighting the key phenotypic features to facilitate the diagnosis of possible ATP1A3 mutations, providing recommendations for genetic testing, and outlining initial acute management for common recurrent clinical conditions, including epilepsy.

The full article can be found at: **http://ng.neurology.org/content/3/2/e139.short**

AHC ANSWERS

AHCF Medical Advisory Board answers your AHC questions

The AHC Foundation is incredibly fortunate to have **12 internationally recognized AHC experts** serving on our Medical Advisory Board (MAB) **from four different countries.** They represent leading academic facilities, clinical hospitals, and research institutions.

For years, the MAB has been **answering your questions** with the help of Sharon Ciccodicola. Under the direction of MAB Chair, Dr. Matt Sweney, and Vice Chair, Dr. Kenneth Silver, we'll be sharing answers to your AHC questions in our forthcoming newsletters.

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

AHC Double the FUNdraising

Kettle Bells for a Cure

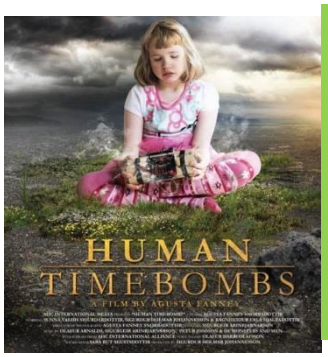
On March 11th, the *Trumbull-Monroe Daily Voice* published an article titled, "Kettle Bells for a Cure: CrossFit Gyms Raise Thousands for Trumbull Tot."

Eleven CrossFit gyms in Fairfield County went to bat and beyond to support 1-year-old **Cameron Simpson and raised more than $10,000** to help his family find a cure for AHC. The gyms competed in **"the Cam" workout** in late February to raise funds for AHC Foundation.

The Cam workout or "WOD" was designed by **Molly McMugh, Cameron's cousin**. It challenged the gym members to do 19 repetitions of a variety of exercises and to do it as many times as possible in 19 minutes. Why 19?

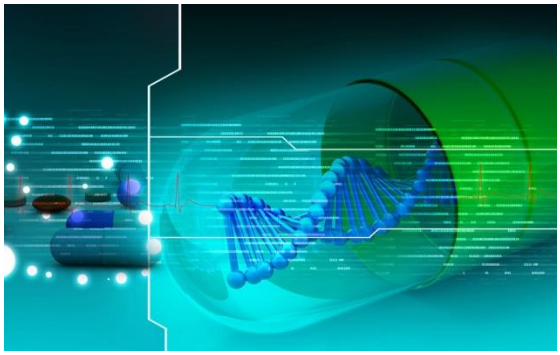
"Nineteen is the number of the chromosome that is mutated in Cam's genetic makeup and in those that suffer from AHC," said Cameron's mother, Corie.

What a great idea! Thank you to everyone who participated in the Cam workout and to Cam's family for helping to **End AHC.**



For all of you folks out east, plan on attending a local screening of the AHC documentary, "Human Timebombs." The event is hosted by Karen & Matt Shubert and will take place on June 11th.

AHC EDUCATION
Keeping AHC families informed



To help keep families informed about AHC in-between family meetings, we are now adding **a new column to the AHCF newsletter.** It will be called **"AHC Education"** and will include general information about AHC. We hope you enjoy reading it and learn something helpful at the same time.

Mutations in the ATP1A3 gene are associated with three related rare neurological disorders, rapid-onset dystonia-parkinsonism (RDP), alternating hemiplegia of childhood (AHC), and recently, cerebellar ataxia, areflexia, pes cavus, optic atrophy, and sensorineural hearing loss (CAPOS) syndrome. The ATP1A3 gene encodes the Na+/K+-ATPase α3 subunit isoform.

The disorders arise from autosomal dominant mutations with variable penetrance and display overlapping symptoms that **vary in severity, duration and frequency of occurrence**.

In the case of AHC, affected patients typically present in the context of an acute onset of paroxysmal, episodic neurological symptoms that include hemiplegia, dystonia, ataxia, or seizures. Some symptoms may persist after resolution, such as neurodevelopmental delays, attention deficits, trunk instability, dystonia or ataxia.

This information was quoted from:
Holm, T. H. et al. Cognitive deficits caused by a disease-mutation in the α3 Na+/K+-ATPase isoform. Sci. Rep. 6, 31972; doi: 10.1038/srep31972 (2016).

AHC INFO EXCHANGE
Amici ben fatto – Well done friends

Researchers from Italy have been very busy. In the last month, two articles were published regarding AHC. This is **an amazing accomplishment** and we are happy to let you know about this wonderful progress in AHC research.

On February 26, 2017, an article was published in **Brain Development** titled, "Alternating Hemiplegia of Childhood: Pharmacological Treatment of **30 Italian Patients**." The aim was to review the pharmacological data related to the prophylactic and acute treatment of 30 patients (16M, 14F, age range 5-42years) and to correlate them with the clinical and genetic data collected through the Italian Biobank and Clinical Registry for AHC.

A.I.S.E.A onlus
Associazione Italiana per la Sindrome di Emiplegia Alternante

The second article was published March 6, 2107 in the journal ***Seizure*** and is titled, "Alternating Hemiplegia and Epilepsia Partialis Continua: A New Phenotype for a Novel Compound TBC1D24 Mutation."

This study describes, for the first time, the **association between TBC1D24 variants and AHC,** further expanding the phenotypic spectrum of TBC1D24-related diseases and suggesting that TBC1D24 molecular analysis should be considered in the diagnostic work up of AHC patients.

Congratulations to everyone who worked on these two articles.

DOUBLE the FUNdraising
Successful event documents how fundraiser came together in community

The **Warriors for Kathryn Marszalek fundraiser**, organized by the St. Gabriel Parish on Feb. 4, **raised $17,124.43** to support the Alternating Hemiplegia of Childhood Foundation.

The **event coordinators wrote such an amazing letter** describing their success that we felt compelled to share some of it with you. Enjoy!

"We were blown away by the success of this event. Our goal was to raise $10,000. However, once our parish, St. Gabriel Catholic Church, caught wind of the event**, all prayers broke loose!"**

"We **had an army of volunteers** help with the event. Our expenses were extremely low-everyone who helped, pitched in with food, items, money, and time. Every time we turned around, **someone new was offering to help** or handing us money. This event was organized under the watch of St. Gabriel Church (all gambling and liquor licenses were held by the church). The parents, teachers, and staff of St. Gabriel School all pitched in to help!"

"One of the most amazing facts about this event is that our largest single person contribution was a check for $500. Our second largest was 2 separate donations for $250. ALL other income and donations were smaller than that.

In a county with roughly 23,000 people, and one of the 'poorest' counties in the state of Indiana, **raising over $17,000 is a huge accomplishment."**

The AHC Foundation is greatly appreciative for the achievements of this amazing community.

We hope that this success will inspire others to rally their communities to help **End AHC**. For the work of one community helps all patients with AHC.

Thank you. Thank you. Thank you!

DOUBLE the FUNdraising
Do you love food?

This fundraiser may be perfect for you if you like to eat. Or, if you know people who like to eat tasty food that is easy to prepare, you'll want to join in on this fun event.

A fundraiser to donate to the AHC Foundation and raise awareness in honor of **2.5 year old Tanner** is going on now. Check out the delicious products from Tastefully Simple and **help End AHC** at the same time.

Tanner is an amazing child who loves music and has an amazingly contagious smile. His family loves through it all with incredible fierceness. **Their hope lies in a cure**.

To buy the delicious and easy to prepare food go to: https://www.tastefullysimple.com/p/11059792

COMMUNITY CONNECTIONS

Support Epilepsy Awareness Day this year on Purple Day, March 26, 2017

The number of AHC patients with epilepsy is estimated at over 50%. It can be one of the first diagnoses AHC patients receive. It can also be one of the most live threatening and scary aspects of living with AHC.

Purple Day is an **international grassroots effort** dedicated to increasing awareness about epilepsy worldwide. On March 26th annually, people in countries around the world are **invited to wear purple** and host events in support of epilepsy awareness. Last year, people in dozens of countries on all continents including Antarctica participated in Purple Day!

If you would like more information about Purple Day, go to http://www.purpleday.org/

What are some commonly reported triggers of seizures?

Specific time of day or night
Sleep deprivation – overtired, not sleeping well, not getting enough sleep
At times of fevers or other illnesses
Flashing bright lights or patterns
Alcohol or drug use
Stress
Associated with menstrual cycle (women) or other hormonal changes
Not eating well, low blood sugar
Specific foods, excess caffeine or other products that may aggravate seizures
Use of certain medications

The Epilepsy Foundation 24/7 Helpline

Provides telephone support, answers emails and responds to posts. Some of the topics information specialists respond to most often include:

- Current treatment and alternative therapies options
- Available medications
- Support groups
- Seizure first aid and safety issues
- Employment and discrimination issues
- Emotional support
- Getting connected with local Epilepsy Foundation affiliates
- Sudden Unexpected Death in Epilepsy (SUDEP)
- Requests for print materials

Get help now by calling the 24/7 Helpline at 1-800-332-1000.
http://www.epilepsy.com/helpline