One Mission: End AHC!

# INSIDER'S EDGE

YOUR ALTERNATING HEMIPLEGIA OF CHILDHOOD FOUNDATION NEWSLETTER www.ahckids.org FEBRUARY 2015

2000 Town Center ■ Suite 1900 ■ Southfield, Michigan 48075

*Working Together as a Larger Community*

## Join AHCF on February 28th as we celebrate Rare Disease Day!

As we all know, AHC is a very rare disease. With less than 1,000 known cases, it can be frustrating and isolating. However, we are not alone.

Groups of patient organizations facing similar challenges have come together for one day to celebrate our perseverance and fortitude. It is known as Rare Disease Day.

The main objective of Rare Disease Day is to raise awareness amongst the general public and decision-makers about rare diseases and their impact on patients' lives. The campaign targets primarily the general public and also seeks to raise awareness amongst policy makers, public authorities, industry representatives, researchers, health professionals and anyone who has a genuine interest in rare diseases

Rare Disease Day was started by patient organizations and is patient-led. Therefore, the primary drivers and beneficiaries of the international campaign are people living with a rare disease.


2015 marks eight consecutive, successful years of Rare Disease Day. Rare Disease Day 2015 puts the focus on the daily lives of patients, families and caregivers who are living with a rare disease.

Over 6000 different rare diseases have been identified to date. The complex nature of rare diseases, coupled with limited access to treatment and services, means that family members are often the primary source of solidarity, support and care for their loved ones.

The Rare Disease Day 2015 theme **Living with a Rare Disease** pays tribute to the millions and millions of parents, siblings, grandparents, spouses, aunts, uncles, cousins, and friends whose daily lives are impacted and who are living ***day-by-day, hand-in-hand*** with rare disease patients.

Join us in recognizing the amazing achievements of this rare disease community as we promote the importance of finding a cure for AHC and all of those who suffer with a rare disease.

*A Great Start for AHC Community as Clinicians and Researchers Continue to Have Success*

# AHC research making headlines in 2015

## GRAND ROUNDS

Dr. Kathy Swoboda

On January 23, 2015, **Dr. Allison Brashear**, the Neurology Chair at Wake Forest Baptist Health, was joined by **Dr. Kathryn Swoboda**, Director of Pediatric Motor Disorders at University of Utah, in a grand rounds presentation on ATP1A3. Dr. Swoboda focused her lecture on AHC and generated a lot of interest on the topic from those in the audience.

We thank the doctors for spreading the word about AHC and other ATP1A3 related diseases.

Dr. Allison Brashear

## MOUSE MODEL

Director of the AHC Clinic at Duke Children's Hospital, **Dr. Mohamad Mikati,** recently published an article in the journal *Epilepsia*.

The article is titled, "Knock-in Mouse Model of Alternating Hemiplegia of Childhood: Behavioral and Electrophysiologic Characterization."

The article discusses how his mouse model reproduces the major characteristics of human AHC, indicates ATP1α3 dysfunction, and a predisposition to more severe spreading depression responses (potential mechanism for hemiplegias).

Thank you for spreading the word about AHC.

## RESEARCH PAPER

Dr. Matt Sweney

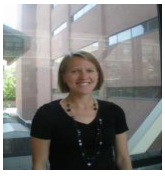
In January, doctors **Matt Sweney, Tara Newcomb and Kathryn Swoboda** wrote an article in *Pediatric Neurology* titled, "The Expanding Spectrum of Neurological Phenotypes in Children with ATP1A3 Mutations, Alternating Hemiplegia of Childhood, Rapid-onset Dystonia-Parkinsonism, CAPOS and Beyond."

The article discusses while classically defined phenotypes associated with AHC, RDP, and CAPOS syndromes are distinct, common elements among ATP1A3-related neurological disorders include characteristic episodic neurological symptoms and signs that vary in severity, duration, and frequency of occurrence.

Dr. Kathy Swoboda

The paper concludes stating that additional work is needed to better identify and classify affected patients and develop targeted treatment approaches.

Thanks to the team at the University of Utah for your fine work on this paper.



Tara Newcomb

*Amazing Event to Benefit AHCF Taking Shape*

# Don't miss out on a new event taking place outside of San Francisco, California on June 6, 2015

**AHCF is sponsoring a major event that has the potential to raise $50,000 - $100,000 for research!**

Lynn Egan is spearheading an event called **Wine, Women & Shoes** to be held June 6, 2015 in Atherton CA at the stunning Menlo Circus Club.

Wine Women and Shoes (WWS) is a non-profit national organization that has raised more than $25 million for women and children's causes. The event has been hugely successful in raising more money for non-profits than traditional fundraising events. Our partnership with WWS will not only help raise awareness but also much needed funds for research for AHC!

The event features a sophisticated evening of wine tastings, a live and silent auction, and a fashion market place for your shopping pleasure and a fashion show presented by Macy's. Wine, Women and Shoes is for those that enjoy fine wine, great style, female camaraderie all the while supporting a noble cause.

All proceeds will go directly toward research.

Why am I telling you all this? Because in order to make this a super success and secure research monies for many of your projects we need you to help us by tapping into your friends, relatives and well to do colleagues in the San Francisco Bay Area.


We expect 400 guests to enjoy strolling wine & food, boutique shopping, Wall of Wine, silent & live auctions, fashion show, swag bags, VIP tables, "Shoe guys" and more.....

If you would like to help make this amazing event even more successful, there are numerous ways for you to participate.

We're looking for people who are fun-loving and charity minded *fashionistas* who'd love a few fabulous hours of shopping, networking and fundraising for a great cause – AHC!

Please contact Lynn Egan at lynn@ahckids.org on how to participate.

*Five Part Series on Getting Diagnosed with AHC by Doug Morris*

# Part two: What is wrong with my child?

*This is the story of how one family went through various phases of learning about their children's AHC diagnosis to thriving in life.*

*Many of you may relate to their experience while others of you may see hope for all of our AHC community.*

*By the end of the series we hope you all will see that our kids can lead a fulfilling life as we all work towards ultimately finding a cure.*

**PHASE 2 ANGER**

Our first child, Haley, was diagnosed with AHC in January 1994. Six weeks later, our second child, Caroline was born. Within 18 months, she too was diagnosed with AHC.

In last month's article, I discussed how initially we went through denial. As parents we searched for every piece of information we could find on AHC and talked to various doctors to understand the symptoms better. We wanted to be able to predict and provide a better future for Haley and Caroline.

After denial came anger. Why me? What did I do in the past to bring this on to our first child? Why did Haley come genetically into this world with AHC? Why did Caroline come with AHC too? It's not fair to our kids, their mother, or to me.

We didn't understand the certainty of the diagnosis. I was angry at the world and stored it in a silent but definitive spot in my mind. I felt the need to blame someone, something, but found that need to be unfulfilling.

To this day, our kids never once expressed anger at me or Chris on their condition. I really could have used their maturity of acceptance when I first learned about their conditions.

Anger subsided and I started bargaining. Next month, I'll share more about how I bargained my way with AHC.


**Be sure to join us next month as Doug talks about bargaining and how it played a role in dealing with the diagnosis of AHC for his family.**

**Donors:** Your gifts fund 100% of our effort to help children with AHC. It is our privilege to coordinate the support and services you find critical to improving their quality of life. Thank you for your steady support. Everything we achieve depends on you.