



FEBRUARY 2013

BOARD BASICS UPDATE FROM OUR INTERIM PRESIDENT

I am pleased to announce that 2013 marks a major milestone in AHC history as we celebrate our 20th year as a foundation!

Throughout the year, we will be celebrating our anniversary by sharing our long history and scheduling events to acknowledge this great achievement. Our first celebratory act was to document the journey we took from the creation of the International Alternating Hemiplegia of Childhood Foundation (IFAHC) to the ultimate destination of establishing the Alternating Hemiplegia of Childhood Foundation (AHCF).

As a founding member of the foundation, I was pleased to help create a written record of our history and achievements. Since it is a long and distinguished history, please go to our website and read about how the foundation was started as an idea at a kitchen table and how we have become what we are today.

It is truly amazing that we have such dedicated and unselfish families working for the same cause - a treatment and ultimately a cure for AHC. We are truly an 'elite' group whom has bonded together for the benefit of our children.

This year, I hope you will join me and the AHCF Board of Directors in celebrating the 20th anniversary of the Alternating Hemiplegia of Childhood Foundation.

LYNN EGAN Lynn@ahckids.org
AHCF Interim President

Starting March 1st the AHCF office will be relocating to the below address. Please send all communications and donations to this address going forward. Any mail going to the Livonia address will be forwarded by USPS for 1 year.

**AHCF
2000 Town Center, Suite 1900
Southfield, MI 48075**



UPDATE FROM OUR SECRETARY

Great news. The AHCF created a new format to keep our community up-to-date on the latest information regarding AHC. Instead of doing lengthy newsletters, we will be sending out shorter updates on a more frequent basis. If you have anything you wish to share in a future update, feel free to email me your contribution.

As I prepared this month's update, the following quote resonated with me,

"There are few challenges that are solved by the efforts of one individual.

Nearly every major breakthrough in health and quality of life has been a result of teamwork - a community of caring individuals united towards a common goal."

Phil Gattone, President and CEO Epilepsy Foundation. January 2013

This quote reminds me how lucky I am to have found this AHC community and have the opportunity to volunteer on the AHCF Board of Directors. As we continue to work towards achieving all aspects of the AHCF Mission, I hope you too will find a team within the AHCF you can join. Together, we can achieve even greater breakthroughs for AHC.

VICKY PLATT Vicky@ahckids.org

AHCF Secretary

UPDATE FROM OUR MEDICAL LIAISON

- **Grant Update**

The work continues with steady progress in the research grants we approved last year for the University of Utah and Vanderbilt University. The second payment of \$47,267 to Vanderbilt will be sent March 1st to continue to develop cell lines for use in identifying drugs or drug-like compounds with potential therapeutic effects in restoring normal gene function.

The University of Utah continues to work to identify additional causative gene(s) in patients without mutations in ATP1A3.

Mollie Erpenbeck, an Executive Committee Board Member, attended a conference at Vanderbilt University in December. Dr. Kathy Swoboda, of the University of Utah, spoke about the ongoing research at Utah and the Vanderbilt team graciously gave Mollie a personal tour of the lab where their research is taking place. Mollie came away from the conference with a sense of excitement and hope for the future.



AHCF UPDATE – FEBRUARY 2013



That said, we need everyone's help to get these projects to the next level! Unfortunately, we were unable to fund an additional \$240,000 in grant requests this year. We have never had so many scientists interested in AHC and the only thing lacking is the money needed to get these projects underway.

If the cell line development is successful we would move to clinical drug trials, but that part of the research is the most expensive!! NIH has decreased the number of grants they award to 1 out of 12 and next year are going to decrease the number again to 1 out of 18, so it's crucial that we continue to raise money to keep the research going.

Your help now is more important than ever. Thank you for helping in any way you can!

- **ATP1A3 Testing**

If you would like to know if your child has the ATP1A3 gene (the first gene known to cause AHC), please contact sharon@ahckids.org and we'll arrange for your family to be tested. Tara Newcomb, who is a Licensed Certified Genetic Counselor and our Clinical Research Coordinator at the University of Utah, will let you know if there is DNA stored in the AHCF Biobank or she will send you a DNA kit and notify you of the results.

The testing and consultation are free.

SHARON CICCODICOLA Sharon@ahckids.org
AHCF Medical Liaison & AHCF Coordinator

FUNDRAISING

Update from our Fundraising Chair

Since my plea on Yahoo Groups, I've had a handful of people offer to hold a fundraiser. While I'm very thankful and excited to have new AHC fundraisers, we need more! We need much more! We have new projects that we are unable to fund due to lack of money. Please consider holding a fundraiser this year to help with research!

Our current fundraisers happening NOW:

- CharityMania NCAA Tournament Hoops tickets are available for purchase. Tickets are \$20 each. You may purchase tickets by contacting Mollie at mollie@ahckids.org. Or you may purchase E-tickets by visiting <https://www.charitymania.com/give/Y3001>.



AHCF UPDATE – FEBRUARY 2013



- Auction Frog - We are in the early stages of obtaining auction items that will be up for bid on www.auctionfrog.org. Items up for bid will include vacations, gift cards, etc. If you would like to participate and put items up for bid, please contact Lynn Egan at lynn@ahckids.org, Olga Gerber at olgaagerber@cox.net, or Mollie Erpenbeck at mollie@ahckids.org.

The above fundraisers can be supported by sending the links to all family, friends, co-workers, church groups, Facebook friends, etc. PLEASE participate!

- Crazy for a Cure was held in Winchester, IL on Feb 16, 2013. The Ryan's raised \$8,000. Way to go! Thank you so much for helping the foundation this month.

In addition to fundraisers, I am in the early stages of researching various companies to obtain grant money. So, stay tuned for more exciting fundraising opportunities!

MOLLIE ERPENBECK Mollie@ahckids.org
AHCF Fundraising Chair

EASY FUNDRAISING IDEA- www.thredup.com

Turn the kids' clothes you no longer use into cash for the AHCF. If you are looking for a quick and easy way to help the AHCF and clean out the kids' closets, we have an excellent idea for you.

It is as easy as 1 - 2 - 3.

1 - Collect the children's clothing that no longer fit.

2 - To ship your clothes to ThredUp at no cost, go to the following address to request a bag: <http://www.thredup.com/groups/445-alternating-hemiplegia-of-childhood-foundation> You'll provide your shipping information and a bag will be on the way to you.

3 - When the ThredUPd bag arrives, fill it up, follow their directions, and designate the Alternating Hemiplegia of Childhood Foundation as your Cashout Group.

That's it. It's that easy to help the AHCF.





CALLING ALL AHC WARRIORS!!!

Are you looking for ways to help organize a walk or run to benefit the AHCF? Are you concerned about the time commitment for taking on such an endeavor? Are you uncertain how you would even begin?

Or, maybe you are considering hosting an event and would like to just learn more before you decide.

In either case, do we have good news for you!

The AHCF is putting together a support group called the Walk Warriors. The focus of the group will be to provide you with instruction and advice on how to host a walk, and or, run. Along the way you will receive detailed instructions, sample documents, assistance with contacting the media, direction on how to handle the finances, and much, much more.

The Walk Warriors will be led by Vicky Platt. Vicky currently serves on the AHCF Board of Directors and is the Foundation Secretary. Along with her husband Andy, and fellow Illinois AHC parents, Gene and Kelly Andrasco, they have teamed together for eight years and hosted a walk outside of Chicago. With the support of their family and friends they raise approximately \$30,000 a year. Learn from their success and have an amazing experience along the way.

To join the Walk Warriors simply send your contact information to Vicky Platt (Vicky@ahckids.org) or give her a call (847-337-9044). Upon joining the group, you'll receive a document on how to get started. The group will hold conference calls throughout the year and chat on a variety of topics via email. Walk Warriors will become a support group within the foundation where you can share concerns and exchange ideas.

Are you a Walk Warrior in Waiting?

Well, wait no more. Please contact Vicky Platt by email (Vicky@ahckids.org) or phone (847-337-9044) and we will start you on the path to hosting a hugely successful event.

The AHCF Board of Directors

Gene Andrasco, Sharon Ciccodicola, Cate Cohen, Lynn Egan, Mollie Erpenbeck, Dr. Joe George, Bill Gerber, Doug Morris, Vicky Platt, Carol Presunka, Mary Kay Riley, and Sue Roberts



AHC PHYSICIAN PROFILE

Dr. Kenneth Silver

While AHC is a rare disorder, with few professionals truly experienced in its complexities, we are fortunate to have dedicated physicians working with our foundation and AHC patients around the country. Throughout the year, our update will take a moment to profile some of these professionals and help our community become better acquainted. Enjoy!

NAME: Kenneth Silver MD, MSc, FRCPC

SPECIALTY: Child Neurology

INSTITUTION: University of Chicago, Comer Children's Hospital

HOW LONG HAVE YOU BEEN WORKING IN THE AHC COMMUNITY? 30 years

HOW WERE YOU INTRODUCED TO AHC? One weekend I was asked to see in consultation a 12 year old girl, Leslie who was previously diagnosed with hemiplegic weakness following her seizures (Todd's Paralysis). The following weekend I was asked to see her again with another episode of weakness. However this time the weakness was on the opposite side and no one actually ever saw her have a seizure. Usually seizures occur on the same side of the body so this was very unusual and I was thinking at that time more of a hemiplegic migraine disorder.

My colleague, Dr. Fred Andermann, is an international expert on the relationship between epilepsy and migraine who had actually seen Leslie previously. After we discussed the case it was clear that what was responsible for Leslie's weakness was the newly described syndrome of AHC. This stimulated us to see many more patients, find better treatments, define different subtypes of AHC and research some of the pathophysiology of AHC and teach other neurologists what we had learned about AHC.

When I left Montreal to come to Chicago I expected my interests to change, but it quickly became apparent that there was a continued great demand for expertise in treating patients with AHC. Additionally, this developed into a close collaboration for patient care and research with my colleagues, Dr. Swoboda and her group at the University of Utah.

WHAT INTERESTS YOU MOST ABOUT AHC? The extreme diversity and paroxysmal nature of the symptoms of AHC.

WHAT COMMONALITIES DO YOU SEE AMONG AHC PATIENTS? The extreme similarities of the paroxysmal nature of the symptoms of AHC.

HOW CAN THE AHCF HELP YOU ADVANCE A BETTER UNDERSTANDING OF AHC?

Collaboration in patient care, research, support community and governmental action, publicity, and fundraising.

To learn more about Dr. Silver, go to <http://ahckids.org/medical-research/medical-advisory-board/> or view his presentation from the San Francisco family meeting at: <http://ahckids.org/events/>

Thank you Dr. Silver for your time and your service!



INFO FROM AROUND TOWN

- **AHC SYMPOSIUM UPDATE** Last December, there was a medical symposium held on AHC in Brussels, Belgium. The following summary about the symposium was shared with us by Dominique Poncelin, president of the AHC family foundation in France.

Following the first genetic discovery of a gene responsible for AHC (ATP1A3), this symposium was organized by Dr. David Goldstein (Duke University) and Tsveta Schyns (ENRAH). The symposium was sponsored by ENRAH and various AHC patient organizations from Europe and the United States (including the AHCF). Approximately 50 researchers/doctors from the United States, Canada, Europe, and Japan attended the symposium along with some AHC patient representatives.

While it was not possible to appreciate some of the presentations (too much technical), I found those interesting points:

1. German and Japanese groups (25 cases each) found mutations on 100% of screened cases while Utah and Europe groups on about 75% only.
2. Italian group (Rome) found that there should be a relationship between type of mutation and severity of the disease; however this point is not confirmed up to now by others.
3. Japanese doctors made contacts with Europeans and might be interested in joining the Genotype/Phenotype study.
4. German group did not answer clearly if they are interested or not to collaborate.
5. Geneticists from the Netherlands, France, Italy, and Duke did some improvements in their collaboration for further research to find new genes responsible for AHC.
6. The Genotype/Phenotype current project will involve several geneticists and clinicians from France, USA (Duke), England, Italy, Spain. It will concern at least 100 AHC cases.
7. One aim of the meeting was to point out common points between RODP and AHC diseases as they are concerned by the same gene ATP1A3. However, it was rather disappointing that clinical manifestations and progression of the disease seemed to me rather different.
8. I concluded the symposium with my presentation. On behalf of all AHC patients' representatives, I so dedicated the first gene discovery to passed away children (31 during those past 15 years) and giving their first name and country of origin. Not easy to do, but I'm glad it has received a very positive echo from the researchers (some of them told directly to me that involvement of families is the most important help for their research.)

Best wishes,
Dominique Poncelin

INFO FROM AROUND TOWN CONTINUED

- **February 28, 2013 marks the sixth international Rare Disease Day** coordinated by EURORDIS and organized with rare disease national alliances in 24 European countries. On and around this day hundreds of patient organizations from more than 60 countries and regions worldwide are planning awareness-raising activities converging around the slogan "Rare Disorders without Borders". Activities will take place across Europe, all the way to Russia, continuing to China and Japan, in the US and Canada, and as far as Australia and New Zealand! To find out how to get involved, go to www.rare diseaseday.org



- **Approximately half of all patients with AHC also have epilepsy. Here is some info from our friends at the Epilepsy Foundation (EF).**

Washington, DC, December 26, 2012 - The Epilepsy Foundation (EF) and the Epilepsy Therapy Project (ETP) announced today that their Boards of Directors have unanimously approved the merger of the two organizations.

The new organization, known as the Epilepsy Foundation, will fight for more than two million Americans and their families living with epilepsy today and the one in 26 Americans who will develop epilepsy at some point in their lives.



The new mission of the Foundation is to stop seizures and SUDEP, find a cure and overcome the challenges created by epilepsy through efforts including education, advocacy and research to accelerate ideas into therapies. This strategic merger of the EF and ETP has garnered tremendous support from leaders in the healthcare advocacy and research communities.

DONATE TODAY

Help find the remaining causes of AHC and viable treatment options.

PRIVACY

Your privacy is important to us. We will not provide your e-mail address to anyone else.

SHARE

Share this e-mail with friends and family.

CONTACT US

Send an e-mail to:
contactus@ahckids.org