

JUNE 2013



Celebrating our 20th Year as a Foundation

UPDATE FROM OUR PRESIDENT

I hope this newsletter finds you happy and healthy as you begin summer vacation. As I reflect on our 20th year as a foundation, I am in awe of how far we have come.

In 1993, we only knew of 36 families worldwide. Today we know of over 300 patients in the US and over 800 worldwide. When our children are first diagnosed, we yearn for the knowledge of what we are dealing with and an understanding of the future. Once we have that information, we become busy with the journey to acceptance and everything else that comes with it.

Many of our families have journeyed together for some time and others have just joined us. We come from all walks of life and no matter where we are in dealing with this disorder, we

have families to raise, lives to live, and the day-to-day challenges of living with AHC.

At some point, which is different for everyone, we realize that the treatment/cure will take some time to achieve and it is the day to day, or quality of life, that takes center stage. Twenty years ago, hope of a treatment for AHC was a distant light and today we can almost touch it.

Finding the gene responsible for AHCF took over 40 years to identify. Finding a treatment won't take nearly as long, but it can't come some enough. It was once said to me, "Research is only as good as the funding behind it. The more money you have allows you to increase the speed in which you conduct the research." As we all know, funding research is the key to a treatment.

Needless to say, we also need as much help as possible raising awareness about AHC. Not only will this help families find support and information about AHC, but it also helps increase the possibility of raising funds for the foundation.

Early in the foundation's history, and before the invention of the internet, we had a much more personal connection to each other through direct phone calls. The internet is a wonderful tool that has brought us all closer together, but rather than pick up a phone we send an email or

This year, the AHCF office relocated to the below address. Please send all communications to:
AHCF 2000 Town Center Suite 1900 Southfield, MI 48075

text. I hope to talk with you this summer as we take on the project of updating our database. It will also give me the opportunity to catch up on how you, your family and your child are doing.

I will work very hard meet your expectations. I will do my best to continue the momentum that we have created. You have a wonderful board guiding the foundation, but it truly will require all of you to assist us in raising the awareness we need to find that treatment. You can also contribute to our success by fundraising, writing an article for the newsletter or joining a committee. We are here to support one another. Please feel free to contact any member of the Board, or myself, at anytime. We appreciate your ideas and are here to address your concerns.

LYNN EGAN Lynn@ahckids.org
AHCF President

REASONS TO BE THANKFUL

The Board of Directors will be holding their Annual Business Meeting shortly to plan for the foundation's next fiscal year.

While the 12 member board is busy making plans for the future, we wish to take a look back at the last six months and give thanks for the generous support we have received.

Please join us in giving thanks to some of the people who have helped move the foundation forward during the first half of 2013 through their kindness and generosity

IN LOVING MEMORY

The AHCF wishes to express our condolences to several families who lost loved ones this year. In paying tribute to these loved ones, memorial donations were received by the foundation. We wish to express our gratitude for these donations by recognizing them in our update.

Reuben Anderson
Luann Cohen
Lenora Garfinkel
Maryanne Jerabek
James Langa
Marianne Loretta
Morgan Taylor
Louis Venti
D.J. Walker, Jr.



ANONYMOUS DONATION

In May, the AHCF received an anonymous donation of \$4,000 from the Lutheran Community Foundation in Minneapolis, Minnesota. This is the second year this foundation has presented a grant to us for general purpose use. We are extremely grateful for their continued support.

MAKING A PLEDGE

The AHCF is honored to have received several recurring pledges to the foundation. This quarter, a family with a newly diagnosed child recently made a commitment to pledge 10% of all proceeds from the work of their firm to the AHCF. They took on this pledge in pursuit of a treatment and cure for AHC. The courage and generosity of all those making reoccurring donations is greatly appreciated.

REASONS TO BE THANKFUL CONTINUED...

HAPPY BIRTHDAY TO YOU

Earlier this year, an AHC family celebrated the birthday of their beautiful child. With incredible generosity, the AHCF was included in the festivities when a donation of \$15,000 was made to the foundation. Thank you for honoring us with this gift as you celebrated a happy, happy birthday.



It is because of the overwhelming support of our friends and families that we are able to begin planning for our best year yet.

Update from our Fundraising Chair

I became involved with the AHCF early upon Austin's diagnosis because I wanted to make a difference in his life; a difference that might improve his quality of living one day. I've had a gala, introduced CharityMania, Thred Up, found grants, inspired friends to fundraise for AHCF, encouraged local schools to have out-of-uniform days on Rare Disease Day in support of AHCF.

I have many more ideas, and would be willing to share them with anyone that wants to get involved! I'm sure we all share the same dream of finding a treatment.

We CAN make this a reality if we all do some part, whether small or large. I was very hesitant at first to ask people to donate, but after sharing Austin's story, I have seen and experienced nothing but love and the willingness to help. It's very humbling. I encourage you all to do the same!

Here are some ways families are participating in foundation fundraising activities.

Thred Up- Thred Up is like a consignment store but all online. It's very simple, Order a free bag by following the link provided below. When the bag arrives in your mailbox, fill the bag with gently used clothing, and set the bag out for the mailman.



Thred Up, will then access a value to clothing you donated and then send it along to the AHCF. It's that easy to help the foundation.

To learn more about Thred Up, go their website or shoot me an email at mollie@ahckids.org . To date, we have collected \$166.98. Thred Up matches up to \$1000, so let's all participate!

<http://www.thredup.com/groups/445-alternating-hemiplegia-of-childhood-foundation>

CharityMania- We collected over \$2500 for the March Madness Basketball Tournament through CharityMania! Way to go! I would like to thank all of those that either sold or bought tickets.



Auction Frog- This spring, the AHCF raised \$4300 on Auction Frog, an online auction website! Thank you Lynn, Olga, and Sharon for all your hard work and dedication in putting this new event together.



Upcoming Events:

Renegades Baseball- Paul Hodes, NY
Comedy Fundraiser- Shannon Leigh, NY
Golf Scramble- Rik, Greenwood, MI
Golf Tournament- Terry Sharo/Gene & Kelly Andrasco, IL
San Francisco Walk- Lynn Egan and family
Chicago Walk- Andrasco/Platt Families
Austin's Angels Gala- Erpenbeck Family

MOLLIE ERPENBECK Mollie@ahckids.org
AHCF Fundraising Chair

ONE REASON WHY RAISING MONEY FOR THE FOUNDATION IS SO IMPORTANT

The Global Genes Project reports that it is estimated that 95% of all rare diseases do not have a single FDA approved drug treatment, and there are currently less than 400 treatments approved by the FDA for the nearly 7000 rare diseases which have been identified.

According to estimates from the NIH, it will take 10,000 years at the current rate of FDA drug approvals to find therapies for all people suffering from rare and genetic diseases.

ARE YOU WILLING TO WAIT THAT LONG?

MAKING FUNDRAISING EASY

Everyone has to pitch in at small foundations like the AHCF to garner the funds needed to quickly find a treatment for AHC. In order to help you feel more comfortable participating in foundation fundraising activities, I am sharing some tips on how to be successful at fundraising.

These tips were adapted from a recent posting on *The NonProfitTimes*' website. Enjoy.

- **People give to people.** A donor needs to know the donation is in good hands.
- **Know your story,** then articulate it. It's important to translate the foundation's impact on AHC into relatable, digestible bits of information.
- **Have a plan.**
- **Connect** with donors face-to-face.
- **Identify, cultivate, ask** and then do it again. Your work doesn't stop with the first gift; turn your donors into advocates for the foundation and they'll be more valuable, both monetarily and otherwise.
- Remember, you are brilliant. Recognize how important your work is, and **have confidence** that you can get the job done.
- Recordkeeping is of vital importance. Make sure you **thank those that help.**

To see the entire article, go to:

<http://www.thenonproffitimes.com/management-tips/10-rules-to-fundraise-by/>

UPDATE FROM OUR SECRETARY

Welcome to the AHCF June Update. I am pleased to share with you the latest information about AHC events, conferences, and issues important to our community.

As secretary, one of my responsibilities is to prepare the Family Newsletter. To make it truly awesome, I would love to share your stories and pictures with our community. If you have any favorite vacation pictures or great stories of achievement, please be sure to send those along to me for inclusion in our next newsletter.

The foundation can also use your help in locating our next Vice President and member of the Board of Directors. The search is on for finding an AHC enthusiast to fill this exciting position vacated by Lynn Egan.

The board is comprised of parents, professionals, and friends of AHC patients. If you are reading this newsletter, you probably fit into one of those categories and could be our next Vice President.

If you're not our next Vice President, maybe you know a person that would be perfect for the job. Do you know anyone who is eager to help promote the AHC cause? Do you know of someone who is a great volunteer and ready for a challenge?

If so, we would love to hear from you. Please contact Lynn or myself for additional information.

VICKY PLATT Vicky@ahckids.org
AHCF Secretary

UPDATE FROM OUR MEDICAL LIAISON

Grants Awarded for International Symposium!

The AHCF Board of Directors is pleased to announce the funding of three travel grants to the Second Symposium on ATP1A3 in Disease: Genotype/Phenotype Correlations, Modeling, and Identification of Potential Targets for Treatment. In addition to be a proud sponsor of the symposium, the AHCF is honored to participate on the organizing committee for this meeting.



In 2012, the AHCF was pleased to fund and sponsor one researcher's attendance at the first symposium. After witnessing the success of the first event, the AHCF decided to increase our participation by funding three wonderful representatives travel to Catholic University's School of Medicine in Rome, Italy.

This September, AHCF President Lynn Egan will represent the foundation at the symposium. In addition, the AHCF is sponsoring Dr. Swoboda from the University of Utah and Dr. Al George from Vanderbilt University. Dr. Swoboda will be among the speakers at the symposium and Dr. George will present a poster.

Grants Awarded for International Symposium! Continued...

The objective of this meeting is to discuss the future direction of AHC research, foster collaboration between research groups, and to accelerate the research process to benefit all AHC patients. This is a very exciting time for AHC research as the number of worldwide projects continues to increase.



Grant Update

The two currently funded grants to Vanderbilt University and the University of Utah are winding down for this fiscal year. We anticipate comprehensive reports from each by the middle of July.



I can share that the cell line development was successful and that we are anticipating a new grant proposal from Vanderbilt University. It would include the next steps in this research, which will target drug screening as the first step toward a treatment for AHC.

Dr. Swoboda's team is continuing to look for the remaining genes that cause AHC for the 25% of patients without the ATP1A3 mutation. In addition, they are conducting functional studies that define the characteristics of AHC based on the location of the gene mutation.



We greatly appreciate the hard work of these research teams and that of all researchers working on the behalf of AHC patients everywhere.

What is ATP1A3 and How Can Your Child Be Tested?

ATP1A3 is the genetic mutation recently identified to cause AHC in approximately 75% of AHC patients. In order to provide our families with as much information as possible about the mutation, a presentation was created for use by our families. To find the presentation and learn more about ATP1A3, please visit <http://ahckids.org/medical-research/ahc-in-charts/>

If you would like to know if your child has the ATP1A3 gene, please contact Sharon Ciccodicola at Sharon@ahckids.org and we can assist you in beginning the testing process.

The first step is to work with Tara Newcomb, a Licensed Certified Genetic Counselor, who is our Clinical Research Coordinator at the University of Utah. Tara will introduce you to the AHCF Biobank and can send a DNA kit to your home. Upon completion of the screening, Tara will notify you of the confidential results.

The testing and consultation appointments are free to our families and are funded by the AHCF's continued support of the work completed at the University of Utah.

Additional information is available at <http://ahckids.org/atp1a3/>

MEDICAL LIAISON UPDATE CONT

AHCF Assists in Creating the AHCIA

AHC family foundations and associations from around the world recently formed an organization called the Alternating Hemiplegia of Childhood International Alliance, or AHCIA. The goal of the alliance is to promote collaboration in research and exchange ideas for improving AHC patient's quality of life. China is the latest country to join the alliance and brings the total number of countries represented to twenty seven.

The best way to find information on the alliance is on the AHCIA's website, <http://ahcia.org>. The site is a portal to each countries foundation or association with links to the individual websites.. The website was created and funded through the courtesy of AHCF.



For those countries without a formal association or foundation, individual volunteers agreed to be the contact person with the hope that they will connect with others in their country and grow their contact list.

International meetings are held via teleconference to discuss common issues and develop a strategy in promoting our role as the impetus behind the researchers and clinicians, making sure the parent's voices represented by the associations are heard.

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

AHC PHYSICIAN PROFILE

Dr. Mary Zupanc



The AHCF Board of Directors is pleased to announce the addition of Dr. Mary Zupanc to our Medical Advisory Board. We hope you will enjoy reading this profile on Dr. Zupanc and join us in welcoming her back to our AHC community.

Dr. Mary Zupanc joined the PSF-CHOC-UCI faculty on February 1, 2011 as Director of the Comprehensive Epilepsy Program and Chief of the Division of Child Neurology. Dr. Zupanc is one of the country's most highly regarded pediatric epileptologists. She received her MD degree at UCLA, her pediatric training at the University of Washington and her pediatric neurology training at the University of Wisconsin.

Her training in advanced epilepsy has been broad, with additional training at the University of Wisconsin, UCLA, and Stanford University. She has been instrumental in the development of comprehensive pediatric epilepsy programs at Mayo Clinic, Columbia University, and NYU School of Medicine.

Dr. Zupanc is highly esteemed for her scholarship and contributions to the advancement of Pediatric Neurology. She has presented at numerous nationally and internationally recognized lecture series and is a much sought after speaker.

AHC PHYSICIAN PROFILE cont.

Among Dr. Zupanc's many awards are those for teaching recognition and clinical care at each of the major institutions she has served. She has published many articles in peer reviewed journals, most notably the Journal of Pediatric Neurology regarding pediatric epilepsy surgery and outcomes, the efficacy of Felbamate, and infantile spasms. Dr. Zupanc is the author of 15 textbook chapters including the chapter on epilepsy surgery in the primary textbook in the field: Pediatric Neurology.

Links to Dr. Zupanc's videos and articles can be found

here: <http://www.choc.org/findadoc/index.cfm?id=P00348&pid=1643>

Dr. Zupanc in Her Own Words

I first became involved with the AHC Foundation in the mid-1980s when I identified a patient with Alternating Hemiplegia of Childhood (AHC). The patient was a young baby with episodes of alternating hemiplegia that were initially mis-identified as the post-ictal phase of seizures. We admitted the child to the hospital for long term video EEG monitoring and discovered that the episodes were not associated with any epileptiform discharges on EEG. In fact, the brain waves demonstrated slowing in the contralateral side of the hemiplegia. This suggested a temporary disruption of brain activity.

We actively searched the literature and discovered some articles written about Alternating Hemiplegia of Childhood. This diagnosis fit our baby perfectly. Subsequently,

we relooked at our long term video EEG monitoring. We wanted to find out more about the pathophysiology of AHC, so little was known about it at that time.

The finding of transient/temporary brain wave slowing at the time of the hemiplegic episode



prompted us to perform an ictal SPECT scan (single proton emission computerized tomography), at

the onset of a hemiplegic episode. We used a radio-isotope that crossed the blood-brain barrier in direct proportion to blood flow. To our surprise, the ictal SPECT scan showed a significant decrease in isotope uptake at the time of the hemiplegic attack.

This finding strongly indicated that there was decreased cerebral perfusion in the contralateral hemisphere (the hemisphere that controls movement on the opposite side of the body) at the time of the hemiplegia. This had never been reported before and resulted in several publications—one of which drew the attention of Jean Aicardi, MD, the French physician, who was among the first to describe Alternating Hemiplegia of Childhood. He invited me to an international symposium on AHC in Rome, Italy in the late 1980s. It was an experience that I will never forget. It piqued my interest in Alternating Hemiplegia of Childhood. Subsequently, I saw multiple patients from all over the country and Canada who had AHC. Ultimately, Dr. Uri Kramer, Dr. Mohammed Mikati, and I published our collective findings on a cohort of patients with AHC.

Dr. Zupanc in Her Own Words cont.

I have kept in touch with several of my patients with AHC. Some of them have fared much better than others. Many of my patients with AHC have developed epilepsy, often difficult to control. In addition, some of my patients have had stroke-like episodes. AHC can be a cruel disease. It is with renewed interest and a belief in a hope for a cure in this "decade of the brain" that I have re-pledged my commitment to the AHC Foundation.

**Thank you Dr. Zupanc for
your time and your service to
the AHC community!**

PRIVACY

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

CONTACT US

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

INFO FROM AROUND TOWN

AHC BIBLIOGRAPHY UPDATED

On June 4th, in honor of the twentieth anniversary of the Alternating Hemiplegia of Childhood Foundation, the AHCF was happy to present a bibliography of articles on Alternating Hemiplegia of Childhood. The articles included in this bibliography were selected to help parents locate information on a variety of topics related to this disorder.

The AHCF would also like to congratulate several authors well-known to the foundation for having published articles in the last year. Congratulations!

Dr. David Goldstein, July 2012

Dr. Erin Heinzen, July 2012

Dr. Mohamad Mikati, July 2012 and May 2013

Tara Newcomb, July 2012

Dominique Poncelin, May 2013

Dr. Sandy Reyna, July 2012

Dr. Kenneth Silver, July 2012

Dr. Matthew Sweeney, July 2012

Dr. Kathy Swoboda, July 2012

The 2013 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

DONATE TODAY

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

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