July 2006
Newsletter

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Issue 1

THE ALTERNATING HEMIPLEGIA OF CHILDHOOD FOUNDATION

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www.ahckids.org

"Our vision is to find the cause(s) and a cure for AHC while providing support to the children with AHC and the parents who care for them."

INSIDE THIS ISSUE:

AHCF
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Raising Awareness....Raising Funds

By Lynn Egan

How much money has been given to AHC research over the past 3 years? $203,870!!! To move ahead with this critical research, we desperatly need help from AHC families because research costs more than we are raising.

Have you ever considered having a fundraiser? Writing a solicitation letter? Having a car wash? A bake sale? A garage sale? Texas Hold-em Poker party? You would be surprised how easy and fun it is.

Carol Presunka and I hosted a fundraiser a few years ago and raised about $3000. Carol found a venue, Bay Meadows Race Track, which allows organizations to sponsor fundraisers. So we had "A Day at the Races" This was probably one of the simplest fundraisers as the racetrack did most of the work. All we had to do was make flyers and invitations and sell tickets! To make it easier on ourselves due to time constraints, we provided the items for the raffles ourselves. We found friends and family happy to attend and others who were disappointed not to be included in the invitation. The first time, we sold about 60 tickets and had a few raffle items. We plan to do it again October 29. Our goal is to double the amount of tickets and increase our raffle items by soliciting our local businesses. Yes, it did and will take a lot of work and time but it is for a worthy cause...our children!

"If every family raised $1000 today in the United States, we would have $165,000!!!"

How about a solicitation letter? You could write your own but you can also use the fundraising letter example in this newsletter or see the other examples that are up on our website http://ahckids.org/AHCFfundraising.htm. All you need to do is sign your name and send it to family, friends, neighbors and acquaintances. We all receive solicitation/donation letters, postcards in the mail or phone calls from organizations. Encourage those to whom you send the letter to, to make AHC their charity of choice this year and make sure you tell them it is tax deductible. You could even take this same letter, changing the salutation to 'Dear Local Business Owner'. It is an amazing how stores that you visit every week are willing to support and possibly donate to your cause.

(Continued on page 4)

Family meeting in Great Britian Saturday, July 29, 2006

Contact:
rjoanexx@aol.com
Or
Mrs. Gillian Bailey, 80 London Road
Datchet, SLOUGH, SL3 9LQ
Tel: 01753 546268

Save this Date!!

Next Family meeting
July 28-29, 2007
San Francisco, California U.S.A.

Information about hotel, exact location and agenda will be in the October newsletter.

Sharon Ciccodicola RN, Medical Liaison

I'm very outspoken when it comes to the belief that each individual family member MUST have a knowledgeable, strong, supportive patient advocate. It is a necessity in medicine these days and from the calls I've received, the people I've met and observing the discussion group online I feel many of you are at the forefront of this task and doing it very well. I have the utmost admiration for all of the AHCF families and the struggles you face dealing with this cruel disease in addition to the stresses of everyday life. The level of information sharing and support you have for each other is something I have rarely experienced before in any other group.

I'm going to help Lynn update the foundation family list this year and develop an e-mail notification list for those families interested in receiving e-mails regarding news and updates about the foundation between newsletters. If you are interested in this please e-mail me with your current e-mail address and I will put you on the list.

If you are willing to be a family support or reference person for your state or region please give me this information also. No e-mail addresses will be distributed without your permission.

I would also like to extend this invitation to e-mail me with questions, comments and suggestions @ any time. My e-mail is cicco@rualfree.net

Bulletin Board

The AFHA hosts a bulletin board. Everyone can subscribe. Families post their questions, concerns and ideas regarding AHC and their children. You must have a yahoo account to sign up. Go to www.yahoo.com and sign up and get an account. Next, click on 'Groups' on the left hand side under Connect. Type 'AFHA' in the box under join a group. Follow the directions and subscribe.

Weighted Blankets are available from "Salt of the Earth Weighted Gear". For a discount call Annie at 402-723-5229 and mention Richard George, President, AHCF

AHCF FUNDS TWO GRANTS

By Sharion Ciccodicola, RN

In December of 2005 Dr. Kathryn Swoboda was sent a check for $66,890.00 to continue research into Alternating Hemiplegia for 2006, the last year under her current grant proposal. In reviewing her current update you can feel the excitement about the research project. Significant progress has been made.

Dr.Swoboda will present the Board of Directors with a new grant proposal by June to take us through the next three years. However there are several factors that I would like to discuss that may have an impact on this.

First, her team is changing. Justine Johnson, laboratory specialist for five years has left due to her husbands change in jobs. She will be missed. We are however very fortunate to have as her replacement Minh Hang Ph.D who has worked in Dr. Swoboda's lab for two years completing her postdoctoral fellowship training in genetics. By excellent timing, her training grant ended in March so with unanimous board approval she has begun work on the Alternating Hemiplegia project April 1st. Dr. Swoboda feels that she shares in the commitment to this project and brings to it a high level of knowledge and enthusiasm.

Second, as you can tell by reading the update, we are advancing into the "meat" of the research, providing new directions for identifying abnormalities in the genetic make-up of Alternating Hemiplegia. Most of the data collection is in place allowing more time for the actual scientific genetics and biochemistry work.

Third, Dr. Swoboda has indicated that in the next grant proposal she will put together an even more aggressive strategy for the next three year period.

These issues coupled with the fact that we are certain Dr. Swoboda herself has had to contribute to making this grant go forward leads us to expect a grant proposal that is substantially more than what has been spent in each of the previous three years.

REMEMBER: The single most important purpose of The Alternating Hemiplegia Foundation is to generate research funds to find a cure and/or treatment, therefore if we are to move forward into the next phase of this research we need everyone's help! We have come so far and are getting closer to an answer but may not have the resources to go the distance unless everyone contributes in someway!

The second grant that was approved by the Board in March was submitted by Josh Magleby MS. It is in the amount of $3,100.00. Many of you are familiar with Josh' work in the area of cognitive, adaptive, and behavioral characteristics of children diagnosed with AHC. His research began in 2002. The proposed study hypothesizes that children and adolescents diagnosed with AHC display unique cognitive and behavioral sequelae that separate AHC from other similar disorders and may aid medical and mental health professionals in the diagnosis of Alternating Hemiplegia."

Though the grant request technically falls outside of "medical research" most of the board members felt that it was closely related to the medical research and may one day provide a common thread or clue to enhance or confirm a direction the research has taken. Also as AHC has become more well known, (There are over 1600 related responses now when using internet search engines) Doctors, educators and parents have a need for understanding the characteristics of this disease and providing relevant interventions. Josh' research is the first and only one of its' kind specifically related to Alternating Hemiplegia.

This approval however was not without dissension because although we unanimously agreed that the research was very important there was a voice that reminded us to be careful how we spend the money available in the foundation.

This again brings me to a plea for your help. We need to build up the bank! Everyone needs to contribute, we have no corporate or celebrity sponsor. We are a grass roots, parent driven organization. There are a few people who do outstanding fund raising and seem to have endless creative ideas for making money. This foundation performs many different functions from education to family support but clearly our overwhelming objective is to make the research possible. Dr. Swoboda has developed a great research team. Every person is dedicated and selfless when it comes to this project. We are very, very lucky, let's make sure it does not slip away!

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Medical Liaison
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Raising Awareness...Raising Funds (continued)

(Continued from page 1)

Another letter that is being used by a family is the Birthday Letter. Each year, the family mails a child's yearly update on their birthday and asks for a donation to the foundation, instead of gifts.

Here are some other ideas? How about a car wash? bake sale? garage sale? Ok, for the car wash and bake sale, you need a venue. Check with the school your child attends or with your church. Many functions are held on these grounds and you won't know until you ask. As for a garage sale…we all have lots of 'stuff' hanging around. Ask family members to donate their unwanted items too! One last garage sale tip, sell donuts and coffee manned by your child/children! Who could refuse!! With any of these three, place an ad in your local "free or throw away" newspaper and advertise the sale is for AHC.

You can order these donation cans and walk in to your local businesses and ask if you can leave one. I'm sure we have all put extra coins in these types of dispensers to support one organization or another. Every coin helps!!!!

Order donations cans from Richard George

You might be asking yourself, what is my point of all these suggestions and ideas? The research grant for Dr. Swoboda was for three years and will be expiring December 2006. The grant was for $66,890 each year or a total of $200,770 for the three years. With the genetic research ready to go to the next level and with the hiring of Dr. Minh Hang (see Grant Funding Update), the probability of the grant increasing two-fold or more is inevitable. In 2005, revenues were $76,747; expenses (including funding Dr. Swoboda) were $74,297; leaving $2,450 towards the next years funding. In 2004, revenues were $41,253; expenses (including funding Dr. Swoboda) were $73,255 – we spent $32,002 more than we brought in for the year. If we had several 2004 years, we would no longer be able to afford Dr. Swoboda. I don't need to spell out the consequences. Research would be slowed down tremendously or be stopped all together. We would be back to square one….we can't let our children down.

As of this writing, there are four fundraisers happening – Olga & Bill Gerber in Connecticut just hosted a dinner/dance May 5th—they raised $25,000!!!!;

- Dave and Cindy Ryan are hosting their annual Golf Tournament June 10th (see page 8);
- Todd & Sara Wagner of Minnesota are hosting the first annual "DockWorks Charity Classic" invitational golf tournament on July 17th;
- and Brenda Vaughn, aunt to Spencer of Virginia, is having Tupperware parties and the profits are going to AHC—she has raised $1100 to date.

In a phone call today, Brenda asked me how many families were in the U.S. At this time we have 165 knowenfamilies. Brenda then said, "If every family in the US were to raise $1000 this year, we would have $165,000.

We are half way through 2006. Our fundraising goal for this year is $200,000. In order to continue to find a cause/cure to help our children and those in the future we MUST find ways to raise more money. If we don't do it, no one else will. We are one big family, connected by our children. Please take part and help us achieve this goal!!!

Copies of fundraising letters are on the fundraising page on the website. If you do not have a computer and would like copies, please contact Richard George.

Grant Writing

Does anyone have any experience in grant writing? Do you know anyone who might - a relative, friend, co-worker or neighbor? Besides the fundraising, grant writing is a fantastic way to get needed funds for our foundation.

RESEARCH UPDATE: MARCH 2006

Kathryn J. Swoboda, M.D.

Over the past few months we have made significant new progress in evaluating "candidate genes" which may play a role in causing AHC. Candidate genes are those which are considered more likely to cause the disease for a variety of reasons including 1) location of the gene near a known place where a chromosome problem has occurred in a patient with AHC, or 2) the gene of interest has been found to cause similar symptoms, such as those seen in hemiplegic migraine, closely related to AHC, or 3) the gene seems to perform a function in the brain that, when disrupted, seems likely to cause such symptoms, or finally, 4) the gene is associated with similar symptoms in an animal model in which that particular gene has been disrupted in some way. We have continued to use this strategy as we move forward in evaluating promising gene candidates. I will highlight our progress in order to better help the committee assess our progress toward the outlined goals below. We are pleased with our progress, and I think that the speed and efficiency of evaluating new gene candidates in our laboratory and in collaboration with others has greatly increased over the past few months. Increasing interest in AHC on the scientific front is also a major goal, which will encourage additional investigators to become involved in research. Thus, presenting our ongoing work at meetings, such as the International Child Neurology Meeting to be held in June of this year, is another way in which we can help to keep this disease at the forefront and encourage others to participate in such research.

As mentioned in the previous report, we have established two important collaborations with other research laboraties, one at Harvard in Boston with Dr. James Gusella, and the other with Drs. Jen and Baloh at the University of California Los Angeles. We continue to work together with them, and taking advantage of resources at both of those institutions as well as at the University of Utah, to work together to better increase our chances of identifying a genetic cause of AHC as soon as possible.

Our work with the Gusella laboratory and Dr. Kim Hyung-Goo in Boston has focused on two major areas: 1) we have some indications that there may be a "position effect" of a specific chromosome abnormality rather than a direct disruption of a single gene. What this means is that instead of a simple error in the genetic message, the chromosome breakage may actually be interfering in a different way with the normal function of genes in the general area. 2) we continue to work to check every single gene in the region by more standard techniques to decode the message to look for any small errors that could be interfering with the function of one or more genes in the region. Thus, we have applied different approaches to move forward using the strengths of this particular laboratory.

"We propose to dedicate 60% of her time to the AHC project".

As mentioned also in our last report, Drs. Jen and Baloh from UCLA have recently identified a new gene candidate, a glutamate transporter, which they determined to be causing the disorder in a single child in California. The symptoms in this child are very similar to that seen in AHC, and include episodes of alternating hemiplegia. This has given us an entirely new direction in evaluating a whole new group of gene candidates which could play a role in AHC. We continue to work together with our colleagues at UCLA to sequence gene candidates (decode the genetic map dictating how these genes are made into proteins) and also to use newer techniques to identify small and difficult to find changes in these gene candidates which could prove critically important.

As Dr. Reyna has indicated in a recent conversations with both the AHC Foundation President and Medical Liason, approximately two weeks ago the lab specialist who has been working closely with me for the past five years on this project

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Pictures Needed

Dr. Swoboda needs pictures of your children. She needs frontal head (from shoulders up) and a profile shot. She has begun to build a data base of this information to compare features of AHC children.

Also needed are any videos you have of your children during an episode.

Please send them to:
Mark Wright/Kathryn Swoboda, M.D.
University of Utah
Department of Human Genetics
15 N. 2030 E. Rm. 7160
Salt Lake City, UT 84112
801-585-9717 phone
801-581-7404 fax

swoboda@genetics.utah.ed

RESEARCH UPDATE: MARCH 2006 (continued)

(Continued from page 5)
unexpectedly moved. This provides us the opportunity to hire someone with experience in new areas we would like to pursue for the AHC project. We have been actively interviewing new candidates, and have tentatively identified a young scientist with excellent credentials to replace her. She has a PhD, and would bring a number of new skills to the project. We propose to dedicate 60% of her time to the AHC project. Our former laboratory specialist, Justine Johnson, was extremely organized, and we foresee no difficulty in continuing work uninterrupted.

Thus, we report the following progress, and continue to pursue the following goals.

Specific Aim 1. Genome wide linkage analysis and further characterization of resulting gene regions of interest in selected families with more than one affected child:

Simply put, genome wide linkage analysis is a method in which we utilize families with more than one affected individual to help us zero in on a particular area. Newer and better techniques are continually being identified to achieve this task, even using small families, because the tools to perform such work has become better and better in the last year or two. We are actively using a newer strategy, known as SNP analysis (or single nucleotide polymorphism analysis) that takes advantage of unique changes between individuals that occur throughout our genetic map to help identify subtle clues as to how those with AHC may be different than those without within families. Using this technique we are narrowing on focus on at least 3 different areas that could have critical genetic changes causative in AHC.

Specific Aim 2. Analysis of "gene candidates" which cause similar symptoms to those seen in AHC patients, or have an important function that has been implicated as possibly affected in these patients.

All three laboratories have been working on this effort. As a result, we have identified a calcium channel mutation in one of our families with two affected children with AHC. Neither parent seems to carry the mutation, leading us to assume the mutation could have newly occurred in either the eggs or sperm of one of the parents without he or she actually being affected by the disease. We are currently working on additional studies to better understand what this mutation is doing. In addition, we have redoubled our efforst in looking at this and other calcium channels as potential cause of AHC in other children within the database. If the PhD we mentioned above agrees to take the position, then we will have her focus her efforts on this front over the next few months, as it seems particularly promising.

In addition, we have been pursuing new directions with regard to regions within the genetic map holding genes in the following categories: the ATPases and glutamate transporters, using the families with more than one affected child to help us determine whether these additional candidate genes may be important to examine.

3) Cytogenetic analysis of AHC patients without a family history of the disease.

Cytogenetic analysis involves using techniques in which we isolate cells from the blood (we can use cells we've previously banked from our database) and then use a variety of chemicals to fix the chromosomes in place that hold all the genetic information in the cell together. We have used older techniques to screen patients in the past, but newer tools now provide improved power to identify very small missing pieces of chromosome. These small missing pieces are known as "microdeletions". Disorders like Angelman syndrome, which has many symptoms in common with AHC, have been found to be due to such microdeletions, and we continue to try to use better and better techniques to help us find answers. To use an analogy: suppose you were to fly a plane over New York City, and instead of identifying entire missing buildings from the air, you could identify the equivalent of a broken window in one of the buildings....such techniques that dramatically improve our ability to detect changes among the 30,000 candidate genes are coming on line. So far, we have used high resolution karyotyping and fluorescent in-situ cytogenetic techniques to investigate a subgroup of AHC children. Tests performed in this screen include high resolution karyotype (to the 650 band level), and telomere FISH studies. We hope to use even higher resolution techniques in the months ahead to further explore this possibility.

Summary

I hope this above report outlines our progress for the committee in sufficient detail to assure them of our continued dedication and progress in trying to identify the genetic cause or causes or AHC. It increasingly appears from our data that more than one gene may be involved. So far, we and colleagues have identified mutations in 3 separate genes in association with AHC symptoms: ATP1A2, CACNA1A, and now the glutamate transporter SLC1A3. This represents considerable progress over the past three years as we approach the end of our current funding cycle, and provides new directions for identifying abnormalities in all AHC patients. Mutations in the SLC1A3 gene are directly responsible for our pursuit of a new therapeutic direction, since we can more specifically expand our focus to medications known to impact on glutamate related processes. We continue to be optimistic that with time, we will make progress in treating this disease by better understanding the genetics and biochemistry underlying symptoms in these children. I apologize for the previously highly technical report, and will make every effort to make our work more understandable in future reports.

OUR FUTURE DEPENDS ON YOU!! By Richard George

I hope this Newsletter finds everyone happy and healthy.

So, what's new at the AHCF Headquarters?

Our 2006 Board Meetings was held on March 11. Many things were discussed, including funding of a new Grant to Josh Magleby to study the behavior of our children. As far as Dr. Swoboda's Grant, we are in the 3rd and final year of funding. She will be asking for an additional 3-year grant with substantially more money because she is in the lab now. We are most likely looking at more than 100% increase to continue funding her research. Research is the life-line for our children and our future now depends on your support.

We also reviewed our financial position which revealed, unfortunately, depleted funds. We are making every effort to make our money work for us (as we always have), but now we desperately need help from you. At the rate we're going, our funds will run out after we fund Dr. Swoboda's next research extension. We have come so far..... and for what?? To let our children down?? I can't even bear the thought of this and since the day my son was born, I have used every waking second to find money to help our children. I've run out of steam. I'm at a dead end. Please help me find a way.....I'm lost and I need you. I know that God will lead the way, but I also know that it will become easier if you help.

Another key is grant writing. This is one critical form of raising dollars, and we need someone who has some experience in grant writing (or is willing to be trained) to step up and accept this position or to help find someone to accept this task to help take us into the future. Grants = dollars and there are millions of dollars out there just waiting for someone to ask for them. That someone needs to be us!!

We also need volunteers for our Newsletter and for our Website. We are so behind and a small few are doing everything. We all know people we can ask in time of need, be if a family member, a friend, a co-worker or a neighbor. The time is now – WE NEED YOU – OUR CHILDREN'S FUTURE DEPENDS ON YOU!!

So few have done so much. THANK YOU to all of my friends who have helped carry me to this point. I look forward to your continued help and support. May God bless you and keep you always.

10th International Child Neurology Congress

We will be hosting a pre-congress workshop June 11, 2006 at the 10th International Child Neurology Congress in Montreal, Canada. The title of this workshop is 'Alternating Hemiplegia of Childhood: Where do we go From Here?' and is for attendees of the conference only.

Presentations will be made by Dr. Swoboda, Dr. Silver, Drs. Fredrick and Eva Andermann plus 22 others from around the world and will be followed at the end of the session by an hour long 'Think Tank' and summary of the days events.

Our goal is to perhaps find some new direction for research. Secondly, to educate the medical community about AHC, the foundation, and all of you.

As of this printing, we have 120 delegates signed up to attend. We anticipate an additional 10-20% more to sign up the day of the event. We will report to you the success of this meeting in the October newsletter.

Dear friends,

The alternating hemiplegia of children foundation
And
Jacksonville area insurance financial advisors

Cordially invite you to attend and support

Golf 2006

At the Plum Creek Golf course on Saturday, June 10, 2006

This year's event will once again support research for Alternating Hemiplegia of Childhood, which is a neurological disease that has afflicted Michael Ryan, 10, from Alsey, IL. Michael continues to have some paralysis every couple of weeks no matter what and still if he gets over tired, too excited or from an illness. He have had a lot of firsts in his life this year! He continues to attend his multiple health impaired class in Murrayville with Speech, Occupational and Physical Therapies, however this year he has become potty-trained, which means a raise for mom and dad…good thing with the current gas prices!!! This easter was the first time he was able to join the family easter egg money hunt at mom's, usually it is too much for him. Also, Michael just turned 10 on April 23rd and was able to have a weiner roast at the river with the family and friends with no spells! We thank God and the support of good friends like you every day for these milestones!

To date over $47,000 has been used for research just from our golf tournaments. We cannot begin to tell you what that means to Michael, his family and all the other families around the world! We continue research with Dr. swoboda at the university of Utah, where she continues to gather data! (see page 6). For more information about AHC, we can be found at www.ahckids.org!

The tournament is scheduled for June 10, 2006 at the Plum Creek Golf Course in Winchester, IL. 217-742-9018 to sign up your team. Teams consist of 4 members at $50 a person. You may also sponsor a hole for $50.00 or more. This is good advertisement for you while showing your support for our cause. All golfers receive a gift bag and any donations for advertisement would be welcomed. 100% of the donations will go to research, our foundation is ran by families, all donations are tax deductible and receipts will be given upon request. For more information contact us, Dave and Cindy Ryan at 217-742-3253 or email at dcryan@wincoinet.com

1st Place $300, 2nd Place $150, Coed Team 1st Place $100, Oldest team 1st Place $100

Prizes based upon full capacity flights; subject to reduction due to the number of golfers
Coed Teams must include 1 female, oldest team is combined ages must equal at least 160, then best score

Tee times will be 7:30 AM and 1:00 PM

Hole in one for $20,000

All you can eat cook out with mike lonergan
Auction starting at 6:oo PM

Our auction items are donated by local businesses, family and friends. WE appreciate any Items that are donated, ranging from Tucker's famous wine to fishing trips!

The Jacksonville Area Insurance Financial Advisors continue to show their support and we are very grateful. We look forward to another great day!

Respectfully,
Dave, Cindy, Rachel and Michael Ryan And
Jacksonville Area Insurance Financial Advisors

A VARIENT OF AHC????

BY KELLY COSTEDIO

My name is Kelly Costedio. Over the past five years myself along with my three children diagnosed with alternating hemiplegia of childhood.
This is a very hard syndrome for others to understand. It has effected each person in my family in different ways. Although the episodes are so similar, the toll it takes on our body and minds is so completely different. Alec,unlike other AHC children, was not diagnosed until he was three years of age. It was a long process just like everyone else has gone through. First, they thought seizure but finally the diagnosis of AHC. Alec is now eight years old and participates in a substantially separate classroom in the second grade at the Ditson school in Billerica. He is know by all of his friends to have a funny sense of humor and his favorite hobby is collecting jewelry. He is the most effected psychically and cognitively.
Camryn, my youngest, was diagnosed at 8mo. Old. She started having episodes that were quite similar to Alec but just to be sure she went through the same test as Alec because we did not want to jump to any conclusions. Sure enough after a long process she was diagnosed as well. She attends a preschool in Billerica where she can receive all of her services. She is a real spit fire and keeps us all on our toes.
Then, Maxwell, at three, started having episodes that were identical to the other children. We were shocked but knew the drill. We, however, did not put him through all of the test. We decided, along with his doctors that in our hearts knew what we were dealing with. He also attends the Ditson school in an integrated classroom. He loves to take care of his cat jack.
This past year I started having AHC symptoms. These symptoms vary from day to day but are very similar to those I read in your post every day online. My first episode was in October and lasted 6 days. It was a full body. I was unable to use any part of my body. Unresponsive, although I could hear what was going on round me. I have had left side facial droop. I have had significant short term memory loss issues. I have had pain in my limbs. I have paralysis in my left leg when walking long distances. I do have residual weakness in my left side. I have not had a normal day since October. I have episodes that lasts hours-days. They are generally one sided but I have had about 7 full body. My local neuroligist that treats my children has taken me on as a patient as well as an adult neuroligist and they have run all of the test that all of our children have been through and more. They believe that it is some form of AHC. I have started on Flunarizine and Topamax. This has made my days a lot easier and there truly is a difference with these drugs for me. (15mg am 15 mg pm Flunarizine) (100mg am 100mgpm topamax) The only good that comes from my diagnosis is finally knowing what our kids are going through . It helps me to be far more patient with them, and them with me.

Getting Flunarizine from Canada

by Lynn Egan

It has become increasingly difficult to get flunarizine into the United States from Canada.. During the months of February and March, three families had their shipment of flunarizine stopped by the United States customs.

After some phone calls and some intervention by Mark's Pharmacy, we **must** add to our yearly prescription, a letter of necessity. Most physicians should know what that is and what to include however just in case....it needs to include: that the drug is for a child (they are more apt to send it across for children); that flunarizine is not available in the U.S., that it is for Alternating Hemiplegia of Childhood and there is nothing else available in the U.S. that works to control episodes the way flunarizine does.

If you are ordering from Mark's Pharmacy, they are even willing to take your prescription to customs to insure that it goes through with no problem.

Regardless of whether you are ordering from Mark's or elsewhere, I would suggest that you get a letter of necessity from your doctor now and send it so that when you need a refill, they will have the letter on file. Please let them know that a copy of the prescription and letter of necessity must be sent with your order to insure that it crosses the border with no problem.

If you have further questions or need more information, please do not hesitate to call me. 888-225-3353

Letter of Necessity

To Whom It May Concern:

(child's name) has been a neurology patient followed by (Name of physician and Hospital) since (year). She/He is now (age) years old. (child's name) has a diagnosis of Alternating Hemiplegia of Childhood from which she/he takes Sibelium (flunarizine). Sibelium is the only medication that has proven effective in treating this disorder. As you know, flunarizine is not FDA approved and thus, must be purchased from sources outside the United states and then imported into this country.

We request that you please allow the import of Sibelium (flunarizine) for (child's name) personal use only.

If I can answer additional questions, please feel free to contact me at (neurologist's phone number).

Sincerely,

Overview and Impressions of the meeting in Boston

By Sharon Ciccodicola, RN

I was very grateful for the opportunity to attend the Family meeting in Boston last July. It really helped define the Alternating Hemiplegia of Childhood Foundation for me. Meeting the research team led by Dr. Swoboda, the families and children themselves was invaluable.

Lynn Egan, Vice President, welcomed everyone. All of the participants and families then introduced themselves and said a little bit about their family, their affected child or children and where they were from. There were 22 new families in attendance and almost every region of the globe was represented.

Dr. Kenneth Silver was the first speaker. He gave an overview of the disorder. He has been working with children with this disease for 23 years. Interesting points from his presentation:

- There was no defining trouble @ birth or trauma during delivery There was a family history of migraines in 90% of children with AHC
- This disorder affects more females than males
- Half of the patients develop true epilepsy and have severe seizures of 30 minutes or more in duration as they age
- After a prolonged period of seizures usually 6 weeks or more there is noticeable increase in frequency and duration of paralysis, usually occurring during the teen years
- There is an increase being seen in families that have multiple children affected with AHC though each child may differ as to symptoms and severity
- There is no one etiology of an underlying disease that causes AHC

Dr. Silver was also the final speaker. He summarized the current treatments used and reviewed the history of Flunarizine, it's use and effects on the disease, and it's availability, including the current non-interest by the US pharmaceutical companies to produce the drug because of the limited use and low profitability. This was a major concern of the parents attending.

The next speaker was Stephanie Porter RN who addressed the health care issues in schools. She is the "Mass Start" director of Children's hospital in Boston. Stephanie and her team pioneered the use of child specific care plans to be utilized in schools for children of special needs, and showed how to adapt this to AHC children. I have since referred several families to her for help with their own school issues, mostly stemming from their fear of liability and uncertain handling of AHC as a disease since:
1) It has limited common symptoms and the disease itself is still relatively unknown
2) Each child has unique triggers, symptoms, and responses
3) There is a knee-jerk reaction by the schools to have an imprimatur from a physician when in reality few are knowledgeable or comfortable approaching this arena outside of obvious safety issues.

The parents were left with an affirmation to trust their own instincts and not be afraid to voice their concerns, to actively participate and oversee the care their child's school provides.

This led right into the next presentation on Neuropsychology by Josh Magleby MS. He was actively testing affected children and their siblings throughout the conference for many long hours. His presentation included an overview of the tests used, the results of these tests in previous meetings and the importance of increasing the family database to compile accurate findings. I noted that the parents were very interested in his presentation, even asking him questions that sometimes put him on the spot, looking for any positive outcomes to offer hope for their children having some normalcy and especially parents of newly diagnosed children were outwardly anxious to know what lies ahead. He also reminded the families that it is important to help siblings deal with their brother or sister that has AH and to provide an avenue for them to express their feelings.

Josh is to be commended as he is the only speaker to post a report on our website. We eagerly await the results from his testing in Boston. He has currently applied for a grant to further his work in this field. (See page 3.)

Dr. Kim from Harvard was introduced and reviewed the criteria for the molecular studies he is conducting. He is one of the collaborators sharing research with Dr. Swoboda. He defined atypical cases found in families and reviewed exclusions to the study. He is currently working on identify-

OVERVIEW AND IMPRESSIONS (continued)

(Continued from page 10)

ing four additional gene candidates and further narrowed his research to critical regions found on two chromosomes, especially chromosome #3, looking for translocations and deletions. He also compared odds ratios and linkage studies with markers found in families.

Dominique Poncelin the President of AFHA in France reviewed the current number of families in Europe by country, their fundraising efforts and frequency of meetings. He reviewed the workshops initiated and scientific projects in play currently. Mr. Poncelin addressed the difficulty of too many countries, languages and cultures and stressed the need for collaboration on clinical information on a large number of cases. He proposed that already existing groups reach out and provide help in the point collection of blood specimens, diagnosis and treatment especially in remote countries of Asia, South America and Africa.

Scientist Tsveta Schyns of Austria reviewed European research and problems i.e. delayed diagnosis, sporadic population, no clear pathophysiology, proper treatment or cure. Management of the disease is all trial and error and the research is not collaborative. There is low interest by pharmaceutical companies to develop new drugs to treat symptoms and genetics is hampered by the lack of studies.

In 2003 a virtual organization was formed called ENRAH with contacts in each country. There are 14 participants working toward funding to support research in other countries. So far they have the involvement of 9 Euro countries, 2 patient organizations and 9 clinical centers. In 2 years with the $358,000.00 raised so far they would like to establish a European patient registry with clinical data and videos and put this online for use by the scientific community. It will be coded, secured and stored, accessible only for genuine research but available worldwide. She advocated a homogenous data sheet that would be language translated and also would like to merge with the American database to eventually create universal treatment standards. Her group is currently looking for an industry partner willing to fund research.

Dr. Brian Katchan an Intensivist from Toronto had a newly diagnosed 10 mo. old with AH. He gave an interesting and exuberant talk throwing out for discussion possible treatments for the disorder, that in some cases have worked on other diseases. These included the GHB drug, creatinine and succinate. He was open to any suggestion or path no matter how wild it seemed if there was a chance it would help. He would like to look into listing meds on the AHCF website and have parents comment for other parents.

Dr. Kathy Swoboda reviewed in lay terms, the research progress her team has made since the database was formed in 1999. There are over 200 families enrolled now. She described Dr. Kim and Dr. Jen's involvement. Dr. Swoboda also would like an open database to encourage foreign collaboration but cautioned to proceed very guardedly with the sharing of information. She freely answered questions and addressed the concerns of parents even discussing candidly the seemingly new increased risk of sudden death and the need for autopsies and tissue recovery to help other families. Ironically we have lost two children to this disease since then.

I had the opportunity to talk at length with Dr. Joanna Jen PhD, a genetics Neurologist at UCLA. Her specialty was random eye movements associated with autosomal dominant migraines. She is a bundle of energy who sought out Kathy Swoboda after seeing a single 10-year-old patient. This patient's history stated that he had exhibited at age 5 and was hospitalized for 1 month. She noticed an overlap of symptoms between migraine episodes, epilepsy and AHC. She will collaborate with Dr. Swoboda with genome sequencing using a current NIH grant.

I also had the opportunity to participate in a group discussion with Dr. Jen and Dr. Reyna. No question was ridiculous or silly to them. They made the parents feel at ease and created an atmosphere of open exchange. Their answers were knowledgeable and intuitive.

At the end of the meeting we reassembled, shared our discussions and asked any last questions of the speakers. Everyone was given a questionnaire to evaluate the family meeting. It should be useful for planning future meetings as well as how the family's view the foundation in general.

All of the Team assembled for the family meeting worked extremely long hours, never turned away a family member or refused to answer that one last question. They are truly a dedicated group with a genuine interest in finding a cure for this cruel disease. The foundation is very lucky to have them as researchers and advocates to advance the voice of this disease so it doesn't become one of the 6000 rare diseases that are forgotten.

My overall impression was one of awe; the level of family involvement and knowledge continues to impress me. This is truly a parent driven organization. I'm sure that all who attended this family meeting overwhelmingly felt it worthwhile. I believe there will be a DVD available soon to purchase for those who could not attend.

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www.afha.org

Italy
A.I. S.EA— Associazione Italiana per la Sindrome di Emiplegia Alternante
www.aisea.com

Denmark
www.ahckids.dk

Netherlands
www.ahckids.nl

Spain
Associacion Espanola del Sindrome de la Hemiplejia Alternante
www.aesha.orgfree.com

ENRAH
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