



CARES Foundation

research, education and support for CAH

Improving health...connecting people...saving lives

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Volume 10 • Spring 2009

CARES Foundation's
Continuing Medical
Education Workshop
for physicians is
scheduled
for June 9, 2009
in Washington, DC.
To register,
please visit
www.CARESfoundation.org

Attention
Non-runners!

The Annual
No-Sweat Run
for a Cure is back!



See page 7 for more
information.

Epidemiology

Nonclassic CAH (NCAH) owing to 21-hydroxylase deficiency is quite common, ranging from about 1 in 30 among Ashkenazi (Eastern European) Jews to 1 in 1,000 non-Jewish Caucasians of mixed ethnicity affected with NCAH. In comparison, the classic or severe forms of CAH occur in about 1 in 10,000 to 1 in 15,000 live births from direct newborn screening. This review will concentrate on NCAH caused by 21-hydroxylase deficiency.

Hormone profile

The gold standard for distinguishing 21-hydroxylase deficiency from other enzyme defects is the ACTH stimulation test. There are no age-related differences in the criteria for the diagnosis of 21-hydroxylase deficiency, except in low birth weight or premature infants, who tend to have higher stress-induced hormone levels, often leading to false positive newborn screening test results. The severity of hormonal abnormalities depends on the type of 21-hydroxylase deficiency. Among patients with classic salt-wasting CAH, 17-hydroxyprogesterone levels typically exceed 20,000 ng/dl (600 nmol/l), even without ACTH stimulation. In contrast, patients with NCAH have lower ACTH-stimulated levels of this hormone ranging from 1,500 to 10,000 ng/dl before glucocorticoid treatment. Non-salt-wasting, simple virilizing classic CAH may exhibit intermediate hormone levels. Your doctor will put together the hormone test results with the clinical symptoms to decide which type of CAH you/your child has. Some patients, however, cannot readily be characterized as fitting into one of these three categories.

Random blood measurements of basal serum 17-hydroxyprogesterone are often normal in patients with NCAH, unless performed in the early morning (i.e., before 8:00am).

Interestingly, infants do not

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NonClassic Adrenal Hyperplasia

Phyllis W. Speiser

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A Message from the Acting Executive Director



Dear Friends,

Spring has sprung and CARES is looking forward to all the wonderful events coming up.

The Everyone CARES Gala was a great

success that was enjoyed by all who attended and now we're moving forward to our other exciting plans. Read on to see what we've got coming up. There's something for everyone!

Everyone CARES Gala

In March, CARES Foundation honored Dr. Dix Poppas and Pfizer for their contributions to CARES and CAH. The Gala was a great success! Your contributions, ticket and silent auction purchases are helping to support all of our wonderful programs. *Visit our photo gallery in the Our Community section of our website for photos of the event.*

No-Sweat Run for a Cure

It's that time again! The Annual No-Sweat Run for a Cure is back! So, let's all NOT lace up our sneakers for another successful non-run! See page 7 for more information on No-Sweat and how to form a team.

CME Workshop

CARES Foundation's first ever continuing medical education (CME) workshop is scheduled for June 9, 2009 in Washington, DC. The workshop is for physician education and will last a full day. The main focus of the workshop is on CAH throughout the lifespan with information on other adrenal disorders as well. Our co-sponsor for the workshop is The University of Texas Southwestern Medical Center. Please don't forget to tell your doctors about this opportunity as time is running short to register. To register, please visit www.CARESfoundation.org.

Family Conference

The CARES Family Conference is scheduled for Sunday, November 1, 2009 and will take place at the College of St. Elizabeth in Madison, NJ. Planning is underway and more information will be coming soon. We look forward to seeing you there!

EMS Update

The response from the CARES Community in relation to our emergency medical response for adrenal insufficiency initiative has been absolutely overwhelming. We have received emails of interest from CARES Foundation members across the country as well as those affected with other adrenal insufficiencies such as Addison's Disease and hypopituitarism thanks to the efforts of our advocacy partners, National Adrenal Diseases Foundation (NADF) and Pituitary Network Association. Clearly this is something that is very important to the entire adrenal insufficient community and we look forward to the day we can say immediate, appropriate emergency medical treatment of adrenal insufficiency is available throughout the United States.

Personal Stories

Many of our newsletters have included inspirational personal stories about living with CAH. You may remember the one about our race car driver from the last issue. It would be really wonderful if we were able to include a personal CAH story in each and every newsletter, so I'm inviting you to tell yours. If you'd like to share your experience, please send your story, with a picture or two, to me at suzanne@caresfoundation.org. I look forward to reading your story!

All my best,

Suzanne

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Everyone CARES Gala

More than 130 friends and supporters joined CARES Foundation members and staff at the Everyone CARES Gala in New York City on March 18th. Pfizer, Inc. and

Dr. Dix Poppas, Professor and Chief of the Institute for Pediatric Urology at the Komansky Center for Children's Health of New York Presbyterian Hospital-Weill Cornell Medical Center were honored for their invaluable contributions to the CAH community. Dr. Poppas was presented with the *Visionary Award* for his work in treating young patients affected by CAH. Dr. Michael Berelowitz received the *Corporate Partner Award* on behalf of Pfizer for its work to find new treatments to improve the lives of CAH patients. The honorees were lauded not only for their expertise, but for the compassion they have demonstrated in their work with CAH patients. The honorees were joined by family, friends and colleagues.



Left to right: Kelly Leight, Founder; Dr. Dix Poppas, Pediatric Urologist, Visionary Award recipient; Greg Kraff, CARES Board President



Left to right: Kelly Leight, Founder; Michael Berelowitz, Senior Vice President, Clinical Development and Medical Affairs, accepting Corporate Partner Award for Pfizer, Inc.; Greg Kraff, CARES Board President

Gregory Kraff, president of CARES Foundation's Board of Trustees, thanked the crowd for their support and urged them to continue to help CARES provide valuable services to the CAH community in all 50 states and 60 countries worldwide. He asked supporters to join CARES on its next crusade to ensure that EMS personnel have the proper training and medicine to provide care for patients with adrenal insufficiencies in a crisis situation.

Guests enjoyed cocktails, dinner and a video presentation on CAH and CARES, as well as a silent auction consisting of sports memorabilia, jewelry, tickets to sporting events and other goodies. The event provided an opportunity for CAH patients, affected families and professionals who are committed to helping improve the quality of life for those living with the disease to come together—many for the first time—to share stories of challenges and triumphs, as well as their hopes and dreams for a cure. The event raised more than \$50,000 for CAH education and research.

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This newsletter is published 4 times a year.

Help CARES raise funds by participating in a family fundraiser, forming a No-Sweat team, or planning your own event. Funding is critical to ensuring the future of programs and services available to CAH families.

When Ashlyn Turned Five

When Ashlyn Kinard turned 5-years-old recently, she decided, very generously, to donate all \$200 of her birthday money to CARES Foundation.

Thank you, Ashlyn, for being so selfless and thinking about others on your birthday!



POKER RUN

90 MILE
SCENIC RIDE



Join Us for the Harleyween Ride for CARES

The Harleyween Ride for CARES is a 90-mile scenic ride and Poker Run from Water Witch Fire Co. Station 7-2 Port Deposit, Maryland to Chesapeake Harley Davidson in Darlington, Maryland to raise money in support of CARES Foundation's mission. There will be music, food, 50-50, door prizes and a raffle. Registration forms must be submitted by July 31, 2009. For more information, please call Debbie Ham at 443-553-5781 or e-mail debbam57@aol.com.

Thanks to the Ward Family!

Many thanks to the Ward Family for their CARES Foundation family fundraiser! They made and sold Buckeye necklaces, bracelets and key chains during the Ohio State football season. The entire family (kids, parents and grandparents) helped out by "hunting" for Buckeyes or drilling the holes in the Buckeyes. The Buckeyes were sold in the front window of the doctor's office where Lisa Ward works and the whole staff helped out. Thanks, again, to the entire Ward family: Lisa, Ty, Olivia and Zach.



I was born with a rare disease called Congenital Adrenal Hyperplasia. At the time of my birth, it was very rare so most people had never heard of it. I was originally misdiagnosed as having severe allergies and sent on my way home. After three years of living life sick, and 15 doctors who could find nothing wrong with me, my mother finally succeeded: she found a doctor with an answer! After a series of tests, during which I suffered three adrenal crises, the doctor found



the answer. She recommended a doctor who was starting research programs at the Children's Hospital in St. Petersburg, FL. In this research program I used the hormone therapy Buserelin, which is used for people diagnosed with prostate cancer, for my CAH. This meant lengths of time in the hospital with constant blood draws and other tests. At the same time the Buserelin was started, they put me on the "drugs of life" for people who suffer what I suffer: Hydrocortisone and Fludrocortisone. These two drugs will be a part of my life until I die. Sometimes it's hard to admit to yourself that you need to depend on pills or any kind of medication to keep you alive because it's hard to think about how fragile you are. Eventually, most days on the medication become "just life" for you and you don't think twice about it, but like everything else, some days it just gets old. You wonder if you can do without them. Then, after your brief venture into nonsense, you remember, if I do not take them, it will be all over.

Eventually, we moved and my mother swore she wouldn't put me on another research grant, if she could find me a new treatment. She had seen enough of me being treated like a lab rat. Looking back, I was treated like a circus freak most of the time. The research I was part of was published in

on the daily pills that have been, and will be, a permanent part of my life.

Now, with all these therapies and treatments, including the terminal use of Corticosteroids, I began to gain weight. If anyone reading this has ever been treated for asthma or used an inhaler and noticed a sudden increase in appetite, it is from the corticosteroids they contain, so you know what I mean. I was starving, every day, all day. I could never eat enough to feel satisfied. Now, I do not blame all the weight I gained on my medicine. I could have shown more self-control, but I have to tell you, taking

medicines that mess with your brain and your stomach is not such an easy thing to deal with or to control. All of my life, every doctor has always said "we need to get that weight off of you" and my mother has always sort of said "yeah, right, you try that." Did I mention I was a grumpy little jerk most of the time? I slowly ate my way up to 295 pounds. That's right, five foot six inches and two hundred ninety-five pounds. I couldn't take it anymore. I didn't like what I saw when I looked in a mirror or how I felt. I decided to drop some pounds.

I joined a gym attempting to train, but in my ignorance, I more or less starved myself. I wasn't eating much and I definitely wasn't going about it the right way, but in the end I lost 100 pounds. I got comments and compliments about how I looked and was even asked what I was doing. Though they all saw a great change

Life Building... Surviving and Thriving with CAH

By Ian Kauffman

continued on page 6

in me, I still wasn't happy. I was weaker than ever and so tired all the time. In the end, I gave in and ate. I didn't eat right since I was still lacking knowledge about nutrition. I gained back about 50lbs, mostly in fat. Eventually, I got my first trainer.

My trainer taught me a lot about lifting and form. She pushed me really hard with the programs she set up, and I learned a lot from her. Just when I started to get going on the road to a new and improved way of life, she left the gym where I was training. I kept at it. I started training with two people that eventually became my friends and for a full year we pushed each other. Finally, even though my diet wasn't great, things were happening. I was dropping fat, super slowly, but it was happening at least. During this time, I went to see my current endocrinologist. I was curious about the skin and fat that seemed a permanent hurdle I could never overcome. I asked if it would ever go away. I was told then that the only way to get rid of some of the weight was to have plastic surgery. I was shocked, and upset. It was just one more thing to add to my worries. I went and got a consultation done by a recommended surgeon, and he too told me the same thing. I didn't want to believe them, so I put it on the backburner for over a year. Eventually, work and other commitments got in our way, and we had a hard time getting together to lift. And to make things worse, the gym we were all at was starting to lose its shine in our eyes. They left first, and tried other gyms. Shortly after, I left too.

I started a new way at a new gym. It was there my life would really take a major turn. After months and months of fighting to improve my health and well being, a road block would present itself. It was a Friday night in the winter of 2006. I was coming home from work and my stomach started to get really upset. I tried to ignore it, but it wouldn't stop. I was at home with my parents, having just suffered a break-up and a herniated disc. Anyway, stress is deadly to me because my body can't handle it. Suddenly, I was throwing up uncontrollably. My mother ran and got me some water and some cranberry juice. I was starting to feel weak and tired. It had been years since I had my last adrenal crisis, and I had just started to educate myself some more on my condition. I had no idea what was going on, or how close I would come to losing it all. Things started to get hazy, and I had been basically vomiting for hours. I finally asked my mother to read up on the details of an adrenal crisis. A few minutes later, she came running upstairs, shook me

awake and told me to get up or they were going to call an ambulance. I wanted to go to the hospital where my endocrinologist was, but the ambulance would only take me to the nearest hospital, without a choice. So the fight against my own body began. I asked for my shoes and help to sit up. I slowly got dressed and slid my shoes on. I had gotten so weak and dehydrated that I could barely walk. I started my slow and painful trip down three flights of steps. I stopped on the first landing to sit and rest after only five steps. I sat at my father's desk on the second floor. I went to the bathroom to throw some cold water on my face, as I was so hot by this point. As I stood in front of the mirror, my eyes barely open, I noticed I had turned a gray color. I almost looked like a corpse. I got to the first floor and I sat on the last step to rest. Meanwhile, there was a snowstorm going on outside. It was like mother nature was out to get me that night. My father tried to pull his van around, but the snow was too deep, so he had to switch to my truck. I took my last breath, stood up, and did my best to charge my way out to the car. In all, it took me about 30 minutes to get down all those steps, all the while one phrase echoed in my head "get up and move or you are going to die." I basically got shoved into the back of my truck. My parents had brought a bucket and blanket for me. I was falling in and out of sleep while trying my best to fight it all off. At this point, all I could think about was how thirsty I felt and how weak I was. We finally pull up to my hospital's ER, my mother runs in and grabs a wheelchair for me, I crawl out of the back and basically fall into the chair. We rush inside, I get pushed up to the counter. My stupid pride back then kept me from having a life alert necklace of any kind, so when they tried to talk to me, I could only say "thirsty, tired." My mother sat down and started answering every question they threw at us. All I could do was beg for water. Eventually I got a cup of ice to chew on, but it was never enough. They put me in a room after only a few minutes. I was shivering uncontrollably so they had to put seven heated blankets on me to get that to stop. The nurses told my mother my temperature was 103.9, my heart rate was through the ceiling, and my blood pressure was through the floor. My parents took turns yelling at the doctor, explaining that I needed a hydrocortisone shot, but she swore it was a bad virus, nothing more. After hours of this arguing, they finally gave me my shot, and changed my saline over to sugared saline. I was so dehydrated that it took hours of IV liquids to get me back to somewhat normal. All night I would fall asleep for five to ten minutes, but it would seem like days. I swore I was done, but I survived. I eventually got home, but it took me 5 days to get back to my normal self. This is just one of my survival stories, but you know all I could think about was getting healthy and getting back into the gym to continue

my journey. Eventually, I was back on my feet and training harder than ever.

After months of doing my own research and asking advice from people who knew better than I did, I met someone. She is just as driven as I am. She knew more about nutrition, and I knew more about the weights which made us a great team. We started training together and from there we developed a relationship outside the gym. We provided support and motivation to one another, but when she noticed that my body was not shaping up the way she expected I had to explain my situation to her. She supported me in going through the plastic surgery, and even stayed at my side while I recovered. Now, understand, I did this surgery as a last resort. It took me over a year to decide to do this. Besides money, I was concerned that with my condition, I could honestly die. Luckily, when I decided to do it, the surgeon my endocrinologist had recommended knew about my condition. Even better, the day of the surgery, the anesthesiology nurse said he knew about it as well, having just been to a seminar about CAH, so I felt a lot safer. It cost me out of my own pocket, even though what I had done was clearly a direct effect of both my CAH and my medicine's side effects. If you ask me, personally, having experienced the surgery myself, it is not a shortcut. Overall, I only lost 10lbs from the surgery. It hopefully added some years to my life, and it has taken some strain off of my body. While I was healing I focused on writing my program for when I could return to the gym. I continued to heal, and slowly worked my way back into training, starting with muscle groups that could be worked without fear of tearing or hurting my abs as they finished healing. Eventually, I was going full bore again, and loving it. Working out is my lifestyle, I love it and live for it. I would go on to lose another 15lbs after the surgery. I had finally gotten to a point where I could try to bulk for the first time. So, I turned to *bodybuilding.com* to look around some more, as I usually did. This time I saw something, an advertisement for free training. So, I decided to try it. I signed up with Team Scivation, and started my first bulk.

Eventually, I became tired of your basic fitness center. I wanted more. I wanted to be surrounded by like minded people. So, I went searching for a new gym. I found a trainer there, a natural world champion: Luis Santiago, and under his guidance and with the support of my friends over at Team Scivation, I have been improving more and more with every week. I have been training there for about two months now, and show no signs of slowing or stopping.

Contact Ian—Staticsbifter@hotmail.com

www.bodybuilding.com/fun/male_transformation_ian_kauffman.htm

Free Diet and Nutritional Help—www.TeamScivation.com

The **2009 No Sweat Run for a Cure** launched on May 8th and will run through September 14th. This virtual run provides an opportunity for CARES members and supporters to raise both awareness of CAH and funds to advance CARES' mission. And you don't even have to break a sweat!

To compete in our annual *No Sweat Run for a Cure*, form a team and ask your friends, colleagues and neighbors to join it. The goal is to raise as much money as you possibly can. Prizes will be awarded to the teams raising the most money and those with the largest number of participants.

Donating is easy. We provide you with many ways to reach out to your friends. We will send you *No Sweat* invitations to mail out or you can set up your own *No Sweat* team page with Kintera, an online fundraising site. You set up your team page, encourage others to donate to it and then you can track your team's progress.

This is a wonderful opportunity to engage your children in raising funds for CAH research. Encourage them to form teams with friends and classmates. They can organize an event, such as a car wash, basketball game, lemonade stand, etc. to raise money for their team.

Your family and friends can celebrate a special occasion such as a birthday, bar or bat mitzvah, anniversary, etc. in the life of a loved one by contributing to *No Sweat Run for a Cure*.



**Ready,
Set, Go....**

**No-Sweat
Run for a Cure**

To learn more or to sign up, please contact: dina@caresfoundation.org.

**Help us meet our goal of \$85,000.
Every "step" counts!**

exhibit day-time variations in adrenal cortical hormone levels at birth, but develop this pattern within the first year of life. Although newborn screening programs have detected very few NCAH infants, test sensitivity and specificity may improve as more screening programs shift to newer diagnostic methods. Other hormones that are usually elevated in 21-hydroxylase deficiency include progesterone, androstenedione and, to a lesser extent, testosterone.

Clinical Manifestations

Mild or nonclassic CAH may be asymptomatic or associated with signs indicative of postnatal androgen (male hormone) excess. Typically, female infants with mild forms of CAH do not have genital ambiguity.

As a consequence of the variable adrenal androgen excess, some affected children grow rapidly or have advanced bone age, and some prematurely develop pubic or axillary hair (pubarche). It is important to rule out NCAH in any child who has unusually early and rapidly progressive signs of puberty, e.g., a growth spurt, acne, or excess facial or body hair. Despite advanced bone age in some children with NCAH, the average adult height attained is well within the normal range for most children with this mild condition.

At adolescence, females are more obviously affected than males, and may develop distressing features of androgen excess, such as progression of facial and body hair growth (hirsutism) and acne. Infrequent menstrual periods, common in many adolescent girls, may be more severe in NCAH. Hirsutism is most common (found in approximately 60% of cases at diagnosis), followed by infrequent menstrual periods (54%) and acne (33%).

It should be noted that these non-specific signs of androgen excess are also associated with the even more prevalent polycystic ovarian syndrome. Moreover, women from certain ethnic groups are more prone to excess body and facial hair. Thus, as with the early appearance of pubic hair in children, not all women with androgenic symptoms have NCAH as the cause.

Despite advanced bone age in some children with NCAH, the average adult height attained is well within the normal range for most children with this mild condition.

Reproductive problems are more common in NCAH women than in the general population. Women desiring fertility should seek treatment from a reproductive endocrinologist; conception is possible without hormone therapy, but if this does not occur, simple treatment is available. Although there is no sex partiality for CAH, males are not readily detected with androgen excess after childhood. During adolescence,

young men are most often diagnosed because a family member is known to be affected. Low sperm count and infertility have been described, although less often than in classic CAH. Testicular adrenal rests, common among classic CAH boys and men, have not been reported in NCAH men, perhaps because of low rates of screening.

Natural History

The natural history of this common, mild form of CAH is not precisely known. A few NCAH cases are detected by newborn screening programs, but most are missed because of the relatively low baseline levels of 17-hydroxyprogesterone. It is not known what proportion of cases ascertained in this manner eventually become symptomatic. At present, the consensus among pediatric endocrinologists is that healthy asymptomatic newborns with minimally elevated 17OHP levels do not require hydrocortisone treatment, but should be closely followed.

Based on retrospective family studies, many subjects with genetic and hormonal traits diagnostic of NCAH never seek medical attention for androgen excess. Indeed, a substantial proportion of affected people with NCAH have experienced normal growth, puberty and reproduction without glucocorticoid treatment. In one survey, 68% of 203 pregnancies occurred before the mother's diagnosis of NCAH. Also of interest is the variable severity of androgen excess and the age at presentation. There are no published reports of death, or even acute adrenal insufficiency, attributed to NCAH. In light of these observations, clinicians should not equate NCAH with more severe forms of the disease when counseling families. It is unclear why

some affected individuals show signs of androgen excess and others do not.

The risk of a parent with NCAH producing offspring affected with classic forms of CAH is estimated at about 2-3%. About 15% of children born to mothers with NCAH are similarly affected.

Treatment

There is currently no strong evidence to suggest that preventive early glucocorticoid treatment of asymptomatic or mildly symptomatic NCAH children is superior to watchful waiting in terms of long-term outcomes with respect to growth, fertility, or related metabolic conditions. Low dose glucocorticoid treatment should be reserved for children with early onset and rapid progression of pubic and body hair, growth, and/or skeletal age. Treatment should not be instituted merely for elevated hormone levels. Similarly, in adults, hormone treatment should be reserved for women who suffer from symptoms of androgen excess not readily remedied by other methods. Infrequent menstrual periods are readily responsive to glucocorticoid treatment. In contrast, patients with extensive hirsutism should be aware that glucocorticoid therapy alone will not instantly improve their condition; additional treatment with lasers, electrolysis, depilatories or other therapies may also be needed.

In children, the preferred drug is hydrocortisone. The dose must be individualized and carefully monitored. Children should also have an annual bone age x-ray. Statural growth and growth rates should be graphed on age and gender appropriate charts to make sure that growth is neither too slow nor

too fast. The therapeutic goal is to use the lowest glucocorticoid dose that adequately suppresses adrenal androgens and maintains normal growth and weight gain.

Once growth is complete or nearly complete, adolescents and adults may be treated with prednisone or

*Nonclassic
CAH is a mild
condition that
should not be
regarded as a
life-threatening
medical
condition...*

dexamethasone (dex). Patients on long-acting glucocorticoids like prednisone and dex should be monitored carefully for signs of medically-induced Cushing Syndrome, such as rapid weight gain, high blood pressure, pigmented skin stretch marks, and fragile bones, although these problems are typically quite rare among appropriately monitored patients with CAH.

Individuals who are affected with NCAH, but have never been symptomatic or treated with gluco-

corticoids, have no requirement for stress dosing.

Stress doses of hydrocortisone are recommended for glucocorticoid-treated NCAH patients who undergo surgery requiring general anesthesia or critical illness. It is assumed that such patients have medical partial adrenal cortical suppression owing to long-term glucocorticoid therapy. Patients on treatment should carry a prescription from their endocrinologist for emergencies, in case they need to be treated away from their home hospital. The patient's family members should be given a supply of injectable hydrocortisone and instructions for appropriate emergency use at the start of oral glucocorticoid maintenance therapy. Conventional wisdom is that for a child in adrenal crisis, infants and toddlers receive 25 mg by intramuscular injection, school-age children 50 mg, and adolescents and adults up to 100 mg. The patient should be taken for emergency medical evaluation and further treatment, including continued intravenous hydrocortisone every 6 hours during critical illness. When the acute illness resolves, usually within 48 hours, the dose can be rapidly tapered to maintenance doses.

Conclusions

Nonclassic CAH is a mild condition that should not be regarded as a life-threatening medical condition, nor is universal glucocorticoid treatment required. Rather, each case must be carefully evaluated for the relative risk versus benefit to aggressive treatment or observation.

EMS Campaign Update

We have made great strides in our EMS for Adrenal Insufficiency Campaign over the past several months, including having had the opportunity to present our appeal directly to the NYS EMS medical advisory board (SEMAC) in early March. Subsequent to this presentation we were invited to appear before the Emergency Medical Services for Children (EMSC) Advisory Committee. We believe they will recommend to SEMAC training in adrenal crisis response for all EMTs in NY as well as expansion of existing protocols for treatment of shock to include glucocorticoid treatment for adrenal insufficiency. SEMAC will be voting on our appeal in June. Special thanks to members Debbie Brown, Nancy Ginsberg and Dr. Susan Stred who have been working tirelessly on this campaign in New York.

Even more exciting, in Nevada our appeal has made it all the way to protocol development. Special thanks goes out to Drs. Alan Rice, Rola Saad and Lewis Morrow, Gretchen Alger Lin, and Julie Tacker and family who have provided vital testimony as well as letters of support from CARES Foundation and National Adrenal Diseases Foundation members, the Pituitary Network Association and American Association of Clinical Endocrinologists (AACE)—Nevada Chapter.

Our efforts are being noticed at the national level as well. CARES Foundation and this campaign were featured in the Winter issue of the Emergency Medical Services for Children (EMSC) Family Advocacy Network (FAN) newsletter (http://www.childrensnational.org/files/PDF/EMSC/ForFamilies/FAN_Mail_Winter_2009.pdf) distributed nationwide.

Finally, two CARES Foundation family members have been confirmed as presenters before an audience of EMS providers, EMSC Coordinators and Program Managers, family representatives, physicians, nurses, grant managers, and many connected with the EMSC program at the national EMSC conference to be held in Washington, D.C. this coming June. From each state, three representatives are required to attend this meeting; EMSC program manager, State family representatives, and the principal investigators of the grant (usually the State EMS Director).

If you are interested in adding your voice to that of CARES Foundation and our family and healthcare professional members in either New York or Nevada, or learning more about initiative planning in your own state, please contact Gretchen Alger Lin at gretchen@caresfoundation.org.

Newborn Screening Blood Spot Debate Heats Up



Over the past decade, CARES Foundation has fought tirelessly for expanded newborn screening including testing for CAH across America and around the world. As of 2008, we were able to celebrate the fact that every child born in the United States is tested for CAH at birth. This does not mean, however, that our work in this arena is done. With every state facing budget shortfalls, newborn screening programs throughout the country are being scrutinized. In Georgia, lab hours were cut back to five days-a-week from six. In Utah, the government discussed increasing newborn screening fees, but vowed the economic crisis would not affect its newborn screening program otherwise. CARES Foundation continues to monitor programs across the country and looks forward to your continued support of universal, mandatory screening.

Simultaneously, the debate over newborn screening program blood spot storage has moved from Minnesota to Texas and beyond. As noted in a recent article entitled, *Science Gold Mine, Ethical Minefield*, by Jennifer Couzin-Frankel and published in the April 10, 2009, issue of *Science*, "In Minnesota, a group promoting confidentiality in health care has been engaged in a 6-year battle with the state legislature and the courts over whether its newborn screening program violates privacy by storing and disseminating samples. Last month, a civil rights group sued the state of Texas, charging that its screening program is unconstitutional because it stores samples long-term without obtaining informed consent." Minnesota legislators are now considering a bill (HF1341/SF1478)

that will maintain a strong newborn screening program, while also stepping up parent and provider education regarding the storage of blood spots. The Texas lawsuit has resulted in legislation (HB 1672/ SB 2421) requiring full disclosure to parents regarding the storage and potential use of these blood samples as well as providing parents with the opportunity

to limit the use of this genetic material or request the complete destruction thereof.

What is done with the drops of blood obtained for newborn screening varies widely from state to state and country to country. In some, filter papers are destroyed within a year of screening and others, such as California, samples

are being stored for more than two decades. Couzin-Frankel writes, “This growing treasure trove of samples is catching the attention of researchers, who are turning to them to study everything from the origins of childhood leukemia to toxin exposures in utero and the development of a better, more accurate CAH newborn screen.”

Mabuhay CAHSAPI!

CARES Foundation is pleased and honored to announce CAH Support and Advocacy in the Philippine Islands (CAHSAPI) has joined our list of international support groups. Established through the joint effort of the Philippine Society of Pediatric Metabolism and Endocrinology (PSPME), Section of Pediatric Endocrinology, University of the Philippines-Philippine General Hospital (UP-PGH), CARES sister organization CLAN of Australia, and devoted families in the Philippines, CAHSAPI has been providing invaluable support to individuals affected by CAH and their families across the Philippines since 2005. We look forward to a mutually rewarding relationship dedicated to improving health, connecting people and saving lives in the Philippines and providing much needed support to Filipino families affected by CAH around the globe.

A Message from CAHSAPI President Alain Benedict Yap

CAHSAPI. The term is a wordplay on the Filipino word, *kasapi* which means 'being a member or part of' and an acronym for CAH Support and Advocacy in the Philippine Islands.

I don't remember the exact circumstance when I chanced upon that word but I definitely believed that Filipino parents dealing with CAH needed to group ourselves together. Out of work to commit ourselves to caring directly for our daughter, my wife and I were blessed for the support from family and friends that came our way. Many times, we have relied on online information to help calm our fears and make the right decisions. We have seen and felt true dedication from our doctors and even from persons we've just met. Because of this, we also felt that we need to do our share to help other families, however small our parts may be. We've come to accept that dealing with CAH isn't just a family matter but one that required us to live as true caring Filipinos.

CAHSAPI not only supports around 80 CAH patients already—most of them kids of Filipino families who barely have enough resources to meet the day's basic needs—but in a greater sense, a lifeline. Through the group, members are able to receive the medicines at a subsidized cost through the regular donations by CLAN represented by Kate Hansen. Other than the CAH information from booklets made available to members, the group derives a lot from the experiences shared

among them from concerns about surgery, crises interventions, and parenting. Currently, we hold pocket meetings every quarter and conduct a general assembly once a year.

CAHSAPI, along with the doctors at PGH (Phil Gen Hospital), is working for drug registration for hydrocortisone and fludrocortisone, both of which are not available nor commercially-approved for sale in the Philippines.

CAHSAPI: Ang pangalang ito ay hango sa salitang '*kasapi*' na nag kahulugan ay 'pagiging miyembro o kasama' at bukod dito ang mga binuong mga letra ay maari din mangahulugan ng 'CAH Support and Advocacy in the Philippine Islands'.

Limot ko na kung paano sumagi sa isip ko ang salitang iyon ngunit lubos ang aking paniniwala na kailangan magsama-sama kaming mga magulang na Pilipino upang matugunan ang aming mga problemang dulot ng CAH. Kaming mag-asawa ay sabay na iniwan ang aming mga trabaho para tumutok maalagaan mabuti ang aming anak na sanggol nuon at malaki ang utang namin sa mga tulong na mula sa aming mga kapamilya at kaibigan. Maraming beses rin na ang mga impormasyon na mula sa 'internet' ay sya naming gabay sa pagde-desisyon at upang mapanatag ang aming mga loob. Amin ring nadama ang pagmamalasakit ng aming mga duktor at maging mga taong panandalian pa lang

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naming nakilala at nabuo sa aming pag-iisip na ang CAH ay hindi lamang isang suliranin pang-pamilya ngunit isa na nagangailangan na kami ay magbahagi ng aming buhay bilang mga nagmamalasakit na mga Pilipino.

Ang CAHSAPI ay nagbibigay tulong sa may humigit kumulang na (80) walumpung miyembro na, karamihan dito ay mga batang galing sa mga mahirap na pamilya na hirap na matugunan ang kanilang pang-araw-araw ng pangangailangan. Dahil sa samahan, ang mga miyembro ay nakakabili ng murang gamot sa tulong ng donasyon na mula sa CLAN sa pamumuno ni Kate Hansen. Bukod sa mga pampletang ipinamamahagi tungkol sa CAH, importante din ang dulot sa mga kasapi ng mga pagbabahagi ng karansan tungkol sa siruhiya, pag-tugon sa krisis at maging sa mga payong pang-magulang. Sa ngayon, maroon kaming maliit na pagpupulong tuwing ika-tatlong buwan at nagdaraos ng isang malaking pagtitipon kada taon.

Ang CAHSAPI, gayun man ang mga duktor na aming gabay mula sa PGH ay nag-tutulong lakarin ang rehistro para sa mga gamot na *hydrocortisone* at *fluodrocortisone*, na parehong hindi matatagpuan o ipinagbibili sa mga lokal na botika.

What Does the Economic Stimulus Package Mean to You?

As many of our families tell us, the challenge of getting medical coverage for a chronic disease such as CAH is often frustrating and extremely stressful. With recession in the United States, rising unemployment and increasing healthcare costs, what is already a tough situation has become even worse. Among the provisions of “The American Recovery and Reinvestment Act of 2009” is premium subsidies for COBRA continuation coverage for unemployed workers. Through this act, workers who have been involuntarily terminated can receive assistance to cover up to 65% of the cost of COBRA continuation premiums for up to 9 months. To qualify for this premium subsidy, you must be involuntarily terminated between September 1, 2008 and December 31, 2009. To learn more about this and other provisions designed to assist families and unemployed workers, see page 15 of the act’s “Full Summary” at http://finance.senate.gov/press/Bpress/2009press/prb_021209.pdf.

Additionally, the act provides \$10 billion to the National Institutes of Health for medical research, \$19 billion for health information technology including the adoption of electronic medical records, and \$1.1 billion for comparative effectiveness research.

Oklahoma Support Group

An Oklahoma support group meeting took place last month. Fifteen patients and family members attended as well as a local endocrinologist, Dr. Chalmers, and a CAH nurse, Traci Schaeffer. Their discussions included 504 plans and working with the school, different ways to ensure kids get enough salt, and some confusing parts of stress dosing. The CARES newsletter and camp packets were handed out. Children were kept busy in another room with games and arts & crafts.

The next meeting will be in Tulsa, OK on Saturday, August 8, 2009. Please contact Traci at 405-271-8001 x43043 or e-mail traci-schaeffer@ouhsc.edu if you’re interested in attending.

Please remember that CARES Foundation has “Gone Green” and that our newsletters are now only available electronically. Please make sure we have your most current e-mail address and contact information to ensure that you receive newsletters and other important information from CARES. Send your updated information to Odaly Roche at Odaly@caresfoundation.com.

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