



CARES Foundation

research, education and support for CAH

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

Endocrinologists frequently encounter patients who present with hirsutism, but have lacked clear guidelines for diagnosing and treating this common problem. That situation changed when an expert task force published "Evaluation and Treatment of Hirsutism in Premenopausal Women: An Endocrine Society Clinical Practice Guideline" in the April issue of *The Journal of Clinical Endocrinology & Metabolism*. A pair of meta-analyses of reviews of the roles of anti-androgens and insulin sensitizers in the treatment of hirsutism accompanied the guideline to provide solid reinforcement for the recommendations.

Introduction

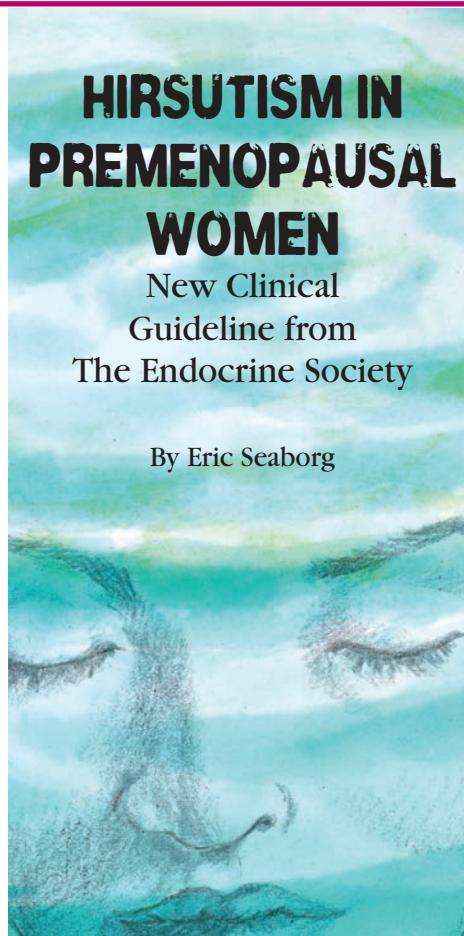
"There has never been any agreement on when to evaluate hirsutism, how to evaluate it, and the best methods for treating it," said Kathryn Martin, M.D., who chaired the task force. Dr. Martin is assistant professor at Harvard Medical School, a faculty member of the Reproductive Endocrine Unit at Massachusetts General Hospital in Boston, and senior deputy editor of *Endocrinology and Metabolism* at Up-To-Date.

The new meta-analyses and the task force's careful review of the literature were critical in developing the guideline. "The evidence is somewhat limited," Dr. Martin told *Endocrine News*. "There are clinical trials on oral contraceptives, anti-androgens, and insulin sensitizing agents, but many of them are not very high quality. This was an opportunity to review all available

HIRSUTISM IN PREMENOPAUSAL WOMEN

New Clinical
Guideline from
The Endocrine Society

By Eric Seaborg



data and, when possible, perform meta-analyses, to come up with the best possible estimate of benefits of different treatments. This had never been done before for hirsutism treatments."

Dr. Martin was selected by the Clinical Guideline Subcommittee of The Endocrine Society to lead the effort and she was joined by six additional experts, two methodologists, and a medical writer. "We first decided the important questions that we wanted to be addressed with a meta-analysis," Dr. Martin said. "The meta-analyses were essential because they gave us the evidence base to feel more confident with our recommendations and suggestions."

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Volume 8 • Fall 2008

Some of the guideline's most innovative features show the patient's perspective on her condition, the best evidence regarding when to test for high androgen levels, which pharmacological treatments are most effective, and which hair removal treatments to recommend.

Patient Importance

Hirsutism is defined medically as excessive terminal hair that appears in a male pattern, as indicated by a Ferriman-Gallwey score of 8 or more. But Dr. Martin said putting too much focus on this score has been a problem in previous recommendations for three reasons: "First of all, if you are basing your treatment approach on the severity of hirsutism, you have to be familiar with this Ferriman-Gallwey scoring, which is very subjective. But most physicians are not very familiar with it. The second problem is, by the time women come to see a physician, virtually all have already been doing some sort of cosmetic intervention on their own. They are shaving or plucking or waxing, so you can't get an accurate Ferriman-Gallwey score when you see them. The third reason is that the clinician's assessment of the severity of the hirsutism may not reflect the degree of distress it causes the patient. A score of 7 could be just as distressing as a score of 17; it just depends on the individual's perception of her problem." So the panel coined the phrase "patient-important hirsutism" as an innovative perspective for treating the condition.

Article continued on page 7

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A Message from the Acting Executive Director

Dear Friends,

There have been many changes at CARES recently. Most notably, our President and Founder, Kelly Leight, has stepped down from her senior leadership roles at CARES. Kelly will remain active with CARES as a member of our Board of Trustees, but her absence on a day to day basis is felt here in the office and she is truly missed. The staff and board of CARES Foundation thank Kelly from the bottom of our collective hearts, for her vision, commitment and countless hours she has devoted to CARES over the past seven years.

Comprehensive Care Center Guidelines

CARES is partnering with the Endocrine Society to develop comprehensive care guidelines for the treatment of CAH. This is exciting news as we are in the midst of planning our summit to establish comprehensive care centers for CAH. As mentioned in our last newsletter, "We believe that there should be a place for everyone with CAH to go for knowledgeable and experienced healthcare with access to sub-specialists as needed." This is yet another step.

Phoqus Pharmaceuticals

We regret to report that Phoqus Pharmaceuticals plc, a drug developer in the U.K., has put its main trading unit into administration (the British equivalent of bankruptcy) after failing to find a commercial partner for its main product, Chronocort. This is a very unfortunate turn of events as the Phase II study from Phoqus evaluating Chronocort (a delayed, sustained release hydrocortisone therapy) in patients with CAH had promising, positive results.

Support Groups

This summer marks the completion of our new Support Group Manual. Along with the manual, CARES held several webinars to introduce the manual and train our support group leaders with the goal of creating a nationwide support community. Our members will now be able to contact their local support group leader for CARES support services. We're very excited to now have the manual that our support volunteers can reference to better serve our members. For more information on how to become a support group leader in your area, please e-mail me at suzanne@caresfoundation.org

2009 CARES Family Conference

We have received many inquiries regarding our next family conference. Rest assured we will be holding our conference in the Fall of 2009! The upcoming conference will be held in New Jersey. The planning committee is in place and we are working on the details. We will be excited to share these details with you as they emerge. CARES Foundation has always underwritten the family conference which is an enormous expense. Although we have always had some event sponsors, a large portion of the cost falls solely on CARES from our annual fundraising campaigns. The current economic climate has taken a toll on our donations and funding which will have profound effects on our conference and other programs. So, we turn to you, our members, for assistance. If you believe your company might be a potential sponsor, please contact Ellie for assistance at ellie@caresfoundation.org.

ENDO 2008

For the first time, CARES had a booth at the Annual Endocrine Society Meeting. We would like to give extra special thanks to

Cedarlane Labs for sponsoring our booth and making it possible for us to be there. While there, I had the opportunity to attend several lectures. I am thrilled and excited to tell you that the lecture given by Dr. Phyllis Speiser on *CAH in the Adult* had overwhelming response. Attendance was standing room only and there were almost as many people listening out in the hall as there were in the auditorium! ENDO 2009 will be held in June in Washington, DC and I encourage all of you to try to attend as it is a wonderful conference.

CME Meeting

CARES Foundation is planning its very first meeting for physicians to earn continuing medical education credits (CME's). We expect to hold the meeting in conjunction with the 2009 Endocrine Society Meeting in Washington, DC next June. The focus of the meeting will be on the transition from pediatric to adult CAH care. The planning is underway and we are thrilled at the prospect of holding such a meeting to help educate the medical community about CAH.

Getting Ready for School/Camp Packets

CARES Foundation has created a *Getting Ready for School/Camp* packet to assist you in building a strong team of family, friends, health care providers, teachers and others to ensure the health and safety of your child with CAH while at school or camp. The packet is full of valuable information including parent tips, a sample medical information letter, medical supply checklist, a health plan worksheet and information about 504 plans. We have had great success with the packet as we have already filled close to 200 orders! We are thrilled to make this valuable tool available to our members and their families. To order the *Getting Ready for School/ Camp* packet, visit the CARES Shop at www.caresfoundation.org.

Fundraising

CARES would like to thank Rolex for their generous donation and everyone who bought raffle tickets for making this a successful fundraiser. With your help, CARES raised \$7,675 during this raffle. See our raffle winner on page 4. Our No-Sweat results can be found on page 12. Thanks to everyone who helped make this year's No-Sweat campaign a success.

It is with great enthusiasm that we move into the future. We are delighted to continue the wonderful work that Kelly began and grow in exciting new directions.

All my best,

Suzanne

Farewell to Kelly



Dearest Kelly,
From National Adrenal Diseases Foundation (NADF), we would like to extend to you our best wishes for a well-earned retirement from active service for congenital adrenal hyperplasia (CAH) patients and their families. Your patient advocacy work has been an inspiration to us all.

A brief history:

Kelly Leight joined NADF several years ago when a family member was diagnosed with non-classical CAH. After finding no organization in the United States specifically dedicated to her condition of interest, she started CARES.

That was seven years ago, and in that short length of time, she has made CARES into a comparative giant, reaching out to untold numbers of CAH patients and their loved ones. Her foundation has been involved in and the actual starting point of important CAH research. Most notably, she has saved the lives of countless newborns by tirelessly campaigning to achieve newborn testing for CAH in almost all 50 states.

Kelly, the NADF employees, board members, medical advisory board and volunteers have enjoyed working with you over the past 7 years, and will miss your help and guidance.

Thank you for all the wonderful work you have done, and know that CARES will continue to grow and save lives.

NADF looks forward to continuing in our relationship as "Sister Organization" to CARES for many years to come.

With deep admiration and affection,

The Employees, Board Members, Medical Advisory Board Members and Volunteers of National Adrenal Diseases Foundation (NADF)

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Family Fundraisers

CARES Foundation is able to offer support to families affected by CAH, fund medical research for new treatments and a cure for CAH, and educate healthcare professionals because of family fundraisers like these. We want to thank all of the individuals and donors who participated in these events. If you'd like to raise funds and awareness in your community, please contact Ellie Avitan, Development Director, at ellie@caresfoundation.org for assistance.



Alexandra's 1st birthday!

Alexandra Thiel's first birthday was made even more special by many of her party guests: to advance research, education and support of CAH, they made gifts in Alex's honor to CARES Foundation totaling \$290. Though she has salt-wasting CAH, Alex is doing great. She is growing and developing well, and couldn't be prettier in pink!

Mrs. Suhovsky's

5th Grade Class

Though the money came in nickels, and though they're only 10 years old, Mrs. Suhovsky's fifth grade class managed to raise nearly \$200 for CARES Foundation. The students unanimously decided to collect coins for CARES Foundation in honor of a classmate's sibling, who is affected by CAH. Every little bit counts!

Pampered Chef

Sonya Matson of Florida, whose son Erik has salt-wasting CAH, opened her home to family and friends for a Pampered Chef party that raised over \$260 for CARES Foundation. Who knew so many people needed fancy kitchen gadgets and cookware? Great job Sonya!

Mathies Airman Leadership School

The students of Mathies Airman Leadership School class 08E raised close to \$2900 for CARES Foundation through various lunch fundraisers. There were many hot dog, hamburger, and Frito pie days at the flight school.



Cleveland Marathon

As Beth Silverman trained for the Cleveland marathon this past spring, crossing the finish line wasn't the only goal in mind. Beth was running to make a difference in her boyfriend Bobby's life by asking friends and family to sponsor her efforts with a donation to CARES Foundation. "I've seen what Bobby goes through each day" said Beth, and CARES Foundation "is the organization that means the most to me".

Beth earned \$670 for CARES.

And the winner is... Eden Santa Cruz!

CARES would like to congratulate Eden on winning the beautiful Rolex watch.



CAH and OSTEOPOROSIS SCREENING STUDY

UNC Chapel Hill, North Carolina

WHO: Children with CAH who are 8-12 years old (bone age 14 years) and are still growing. Siblings (6-14 years old, bone age 14 years old) of those children with CAH who otherwise meet the same eligibility criteria except that they do not have CAH and are not on glucocorticoids.

WHY: Although cortisol replacement is essential to treat children with CAH, there is the potential risk of over-treatment with glucocorticoids that can result in abnormal weight gain, decreased linear growth and, more recently reported in adults, the risk of osteoporosis. We are now testing if there exists a risk for osteoporosis in children with CAH and if this risk is related to the dosing of glucocorticoid used, as would be expected with any medical condition in which steroids are required for long-term treatment. We are also examining if the subtype of CAH contributes to the risk for osteoporosis.

WHERE: Children will be enrolled in the study at the General Clinical Research Center at the University of North Carolina, Chapel Hill.

WHAT: Your child would have:

1. Bone Age X-ray
2. DXA scans (to screen for osteoporosis and for subtle spine fractures).
3. Special X-ray of his/her arm to look at the effects of glucocorticoid dosing (Cortef, for example) on bone structure itself

4. Blood and urine tests to determine the degree of his/her "control" of CAH
5. Blood test for genotyping for all children in the study. In this way, "control" siblings can find out if they are "unaffected" or "carriers".

WHEN: This would all occur in a one-time visit (3 hours) for your child with CAH and/or sibling.

HOW MUCH: The clinical visit, including laboratory testing, radiologic evaluation and physical exam will be paid for by this protocol. Overnight accommodations can be arranged, a rental car to/from the airport and parking at UNC will be covered. Travel assistance is possible (please inquire for details). There is a \$50 compensation provided for incidental costs for each child enrolled.

For more information, contact:
 Karen J. Loehner, M.D./Ph.D.
 Director, UNC Pediatric
 Osteoporosis Clinic
 Assistant Professor, Pediatric
 Endocrine Unit
 (919) 216-5946 (*pager)
 (919) 966-4435 x 224 (voice mail)
 (919) 966-2423 (fax);
 or
 Roxanne Schock, CDE/RN
 Study Coordinator
 (919) 966-0428 (voice mail)
 (919) 966-0971 (fax)

Classical Adult Women's Quality of Life Study

CARES Foundation and Dr. Sheri Berenbaum from Pennsylvania State University have launched a quality of life study of women with classical CAH. It is open to women with classical CAH (Salt wasting and simple virilizing forms) over the age of 18, and entails answering a written survey. If you have questions about the study or want more information, please contact Suzanne Levy at 1-866-227-3737 or email suzanne@caresfoundation.org

NCAH study at Children's Hospital of Los Angeles

The Division of Endocrinology at Children's Hospital Los Angeles is currently recruiting subjects for a research study aimed at determining the stress-fighting ability in subjects with Non-classical Congenital Adrenal Hyperplasia (NCAH) and comparing these responses to those in subjects with Classical Congenital Adrenal Hyperplasia (CAH) and those in carriers of either disorder. If you have NCAH, CAH or are a family member (parent or sibling) of someone with either disease, and are interested in participating in this study, please contact:

Mitchell Geffner, M.D.
 at 323.361.7032 or
mgeffner@chla.usc.edu

New additions to Board of Trustees

CARES Foundation is delighted to welcome Michael Wajnrajch, Dan Taylor, Victoria Shenderovich, Deborah Brown and Karthik Radhakrishnan to our Board of Trustees!

Michael Wajnrajch, M.D.

Mike is a pediatric endocrinologist with extensive clinical experience in CAH. He is currently a medical director for endocrinology at Pfizer Inc, with responsibility for the United States, where he oversees clinical and preclinical activities relating to endocrinology. Mike is also a Clinical Associate Professor of Pediatrics at NYU.

Mike has lived in Argentina, the US and Israel. He enjoys spending time with his wife and three children.

Dan Taylor

Dan brings a wide range of experience in planning/ strategy, general management, and finance from his 14+ years in information technology and financial services industries. Dan currently oversees capacity planning and operations for all of Dell's consumer tech support centers.

Dan is a Michigan native who resides now in Austin, Texas with his wife, Meridith, and two sons, Luke and Colt. His oldest son is a happy, healthy boy that was fortunately diagnosed and treated for SWCAH as a result of newborn screening and follow up care in Texas.

Deborah Brown, R.N.

Deborah is a registered nurse and a mother to two wonderful children. As both a medical professional and mother of a daughter with CAH, she is looking forward to the opportunity to help advance the CARES mission.



Victoria (Vika) Shenderovich



Vika works as a financial analyst in Financial Guarantors group at Moody's Investors Service. She's been with the company for 11 years. Prior to that, Vika worked briefly at Lehman Brothers where she was a coordinating liaison between trading floors and technology groups.

Vika holds a BA degree in Economics from Columbia University. Born in St. Petersburg, Russia, she moved to Israel in 1990, and came to the US in 1993.

Karthik Radhakrishnan



Karthik is a Senior Research Analyst at ING Investment Management in New York. Karthik's research focus is now primarily centered on healthcare investment opportunities. On the academic front, Karthik received a BS in chemical engineering from the Indian Institute of Technology, a MS in civil and environmental engineering from the State University of New York at Buffalo and a MBA from the University of Michigan Business School. Karthik holds the Chartered Financial Analyst designation.

Advocacy

World Health Organization Considers Adding Hydrocortisone and Fludro-cortisone to Essential Drug List

CARES Foundation advocacy partner, Dr. Kate Armstrong, President & Founder of Caring & Living As Neighbours (CLAN) in Australia, has submitted an application to the World Health Organization for the inclusion of hydrocortisone and fludro cortisone in WHO's Essential Drug List. Over the past several months, CARES member families and medical professionals have been adding their voices to others around the world to make these medications available to children in resource-poor countries. Without these essential drugs, children affected by a wide variety of conditions face serious morbidity and often death.

The Second Meeting of the Subcommittee of the Expert Committee on the Selection and Use of Essential Medicines ended 3 October 2008. To follow this process and the outcome of the subcommittee's meeting, please visit: www.who.int/selection_medicines/committees/

Androgen Testing

A patient's first focus may be on hair growth, but the endocrinologist is likely to think first of any underlying conditions that might be behind it. The male-patterned hair growth of hirsutism depends on the presence of androgen, but the guideline does not suggest testing for androgen in every case: "We suggest against testing for elevated androgen levels in women with isolated mild hirsutism because the likelihood of identifying a medical disorder that would change management or outcome is low." Testing for elevated androgen levels should be reserved for moderate-to-severe cases, or for cases of any severity that are sudden in onset, rapidly progressive, or associated with menstrual irregularity, infertility, obesity, acanthosis nigricans, or clitoromegaly.

"Excess androgen production is most often caused by polycystic ovary syndrome (PCOS)," the guideline notes. Dr. Martin said that PCOS is frequently discovered because a patient presents with hirsutism and that physicians' approach to PCOS has changed: "We used to worry only about their cosmetic problems (hirsutism and acne), their irregular periods, and anovulatory infertility," Dr. Martin said. Now physicians worry about the potential metabolic complications associated with PCOS. These women tend to be obese, most are insulin resistant, they are at high risk for diabetes, and many have abnormal lipid profiles. "So we are also worried about long-term risk of coronary heart disease in these women," she noted.

Treatment

The development of hirsutism depends not only on androgen concentrations, but also on the way an individual's hair

follicles respond to the androgen, so the guideline says: "There are two main approaches to the management of hirsutism, which may be used either individually or in combination: (a) pharmacologic therapies that target androgen production and action, and (b) direct methods to reduce and remove hair including cosmetic approaches, electrolysis, and photepilation (laser and intense pulsed light)."

"There are two main approaches to the management of hirsutism, which may be used either individually or in combination..."

The meta-analyses' findings on drug treatments are of particular interest. Examining the value of anti-androgens revealed "weak evidence (that) suggests anti-androgens are mildly effective agents for the treatment of hirsutism." Because of the association of hirsutism and PCOS's diabetes risk, insulin sensitizers have often been used in treatment. But the other meta-analysis concluded, "Imprecise and inconsistent evidence of low to very low quality suggests that insulin sensitizers provide limited to no important benefit for women with hirsutism."

Hair Reduction Therapies

One area of the guideline that endocrinologists may find particularly helpful is the description of cosmetic therapies for hair reduction. Most patients will try their own methods

such as plucking, waxing, and shaving, but if these are not satisfactory they might want guidance on choosing longer-lasting hair removal therapy. The guideline notes that laser removal "appears to be superior to conventional treatments such as shaving, waxing, and electrolysis." The evidence that laser removal is better than electrolysis is fairly thin, however. "Our suggestion to use laser over electrolysis places a relatively higher value on efficiency, convenience, and minimizing pain, and a relatively lower value on cost," the guideline says, noting that patients who place a higher value on cost might well choose electrolysis over laser.

Patient Guide

In addition to the guideline aimed at physicians, the task force created a consumer-friendly patient guide. This guide concludes with a cautionary note useful to patient and physician alike that "hirsutism is usually an ongoing problem. It can be managed, but may not be permanently resolved."

To view or order copies of this and other Endocrine Society clinical guidelines, visit www.endo-society.org/publications/guidelines/index.cfm. The Hormone Foundation's companion patient guides, including one on hirsutism are available at www.hormone.org/Resources/patientguides.cfm.

Eric Seaborg is an award-winning writer living in Charlottesville, Va.

Members of the expert panel were: Kathryn A. Martin, M.D., R. Jeffrey Chang, M.D., David A. Ehrmann, M.D., Lourdes Ibanez, M.D., Ph.D., Rogerio A. Lobo, M.D., Robert L. Rosenfield, M.D., Jerry Shapiro, M.D., Victor M. Montori, M.D., and Brian A. Swiglo, M.D.

Reprinted with permission from *Endocrine News*—complete article can be found at http://www.endosociety.org/endo_news/endo_news_past.cfm (May 2008)

DuoCort Gets Orphan Drug Designation in the US for Treatment of Adrenal Insufficiency

Helsingborg, Sweden, June 27, 2008—DuoCort Pharma AB, a privately held Swedish drug development company, today announced that the US Food and Drug Administration (FDA) has granted an Orphan Drug Designation for the company's DuoCort™ hydrocortisone dual-release oral tablet in development for the treatment of adrenal insufficiency—the inability of the body to produce sufficient amounts of the essential hormone cortisol. Orphan drug designation will give DuoCort™ market exclusivity in the US for seven years after the product attains marketing authorization.

"Current research confirms that conventional therapy is suboptimal and is not serving patients as well as it should. There is scope for improvement in the delivery of cortisol replacement and we are convinced DuoCort™ will offer a better treatment option for patients with adrenal insufficiency and deliver better long-term outcomes," said Dr. Gudmundur Johannsson, Chief Medical Officer of DuoCort Pharma.

"Orphan designation in the US will facilitate this development aimed at demonstrating the significant benefit of cortisol replacement which effectively mimics the physiological diurnal release profile of cortisol in a convenient once-daily tablet. The new DuoCort™ therapy will be the first real innovation for adrenal insufficiency patients in over 30 years," continued Dr. Johannsson.

The orphan-designated product is a novel, dual-release, oral hydrocortisone tablet in 5 mg and 20 mg dosage strengths currently in late stage clinical trials in Europe. DuoCort™ will improve today's cortisol replacement therapy with a once-daily tablet that delivers a more physiological dose of the active substance, better mimicking the body's own cortisol serum profile.

DuoCort™ 5 mg and 20 mg dual-release hydrocortisone tablets are currently undergoing a pivotal clinical Phase II/III trial in Europe assessing the pharmacokinetics, safety and tolerability of once-daily DuoCort™ in comparison to conventional thrice-daily oral hydrocortisone therapy in patients with adrenal insufficiency. The trial commenced in August 2007 and is being conducted at five University endocrinology clinics. Data from the study is expected to be available in the second half of 2008. With normal regulatory review and approval times, DuoCort™ could be on the market by 2010.

Orphan Drug legislation in the US provides incentives to encourage the development and marketing of medicines for rare diseases. Designation as an orphan medicinal product does not indicate that the product has yet fulfilled the efficacy, safety and quality criteria required for drug marketing. These remain to be fulfilled in the pharmaceutical and clinical development of the drug and assessed by the FDA at the marketing authorization stage.

For more information, please contact:

Greg Batcheller, CEO, DuoCort Pharma AB

Telephone: +46 46 288 5008

E-mail: ggreg.batcheller@duocort.com

Fall Michigan Families CAH Gathering

An Afternoon of Bowling and Casual Conversations

Date: Sunday, November 9th

Time: 2:00 pm to 4:00 pm

Location: An Ann Arbor Bowling Alley (exact location TBD)

Contact:

Cathy Berrigan (734) 477-9992

bcberrigan@msn.com or

cjberriigan@umich.edu

Thank You Stephanie! Some kind words...

The following is a portion of an e-mail we received at CARES thanking us (and one of our support people in particular) for what we do. Helping people through difficult situations makes what we do so gratifying.

I have a 5 1/2 year old son with SWCAH. About a month ago, we discovered that our son had been very badly under-dosed and had 5 1/2 year advanced bone maturity. In desperate need of help, I turned to the internet for help and found CARES. This has been a Godsend. I wanted to let you know how much help we have received from Stephanie Erb. She has been absolutely awesome! She has helped us over and over again. From getting us an appointment with a specialist to letting me call and e-mail anytime, and answering any questions I have had. I just thought I'd let you know what a help she has been to us. I know in our busy lives, we don't pause long enough to thank those that help us. I have told her many, many times how much she has meant to our family. But I thought I'd pass this along and also thank everyone at CARES for all that you do!!

From the Emergency Room to the Race Track

By Robbi Converse

When Keith was born, there was no screening at birth for congenital adrenal hyperplasia (CAH). The first sign we had that something was wrong was when he had his first adrenal crisis at about two and a half weeks old. That was followed by weeks in the hospital to diagnose and stabilize his condition. That was followed by months of middle-of-the-night trips to the emergency room and years of wondering what the prognosis really was for our little boy with salt-wasting congenital adrenal hyperplasia.

Were the doctors being completely honest with us? This was years before the internet was available for research, so we were left to learn what we could understand from medical books and trust the doctors for the rest. This is nothing unusual to parents of children with CAH. Wondering. Always wondering, what can my child's life be? What will his limitations be? I write this to encourage you that your child's life, with managed CAH, can be whatever he or she desires it to be!

Keith received his pediatric endocrinology care at Emory University Medical Center and what was then Egleston Children's Hospital in Atlanta, GA. People travel from all over the world to see doctors at Emory and we were blessed enough to have it as our local hospital. However, even with their expertise in so many areas, their familiarity with CAH was limited. As years passed, I never stopped seeking more information on this disorder. When the internet became available, I would search and search for information, but with very little success.

Then came Google! Someone had told me about this search engine that had reference to untold numbers of websites, so I typed in that long string of letters and waited (things were slow back in dial-up days)...five or so hits. I read them all and was disappointed that most of the sites were just parents like me wanting more information. I would regularly visit various search engines and type those same three words. One day when I tried it out, I thought surely I had typed something incorrectly...more than a thousand hits! I double checked my typing and it was correct—every hit was a site pertaining to congenital adrenal hyperplasia. Somewhere in that list of sites, I came across information on a CAH Natural History Study at the National Institutes of Health (NIH) that receives funding support from CARES Foundation. The day of Keith's first visit to NIH for



evaluation was truly a turning point in his life. At NIH he has received top notch treatment from professionals who care about Keith as a person as much as they care about their pursuit of more information on his disorder.

Keith is 20 years old now...a grown man...and he is a race car driver. You see, Keith was born with a passion for cars; far exceeding the normal little boy fascination with the machines. His

career path was sealed when he was in fifth grade and he attended his first Petit Le Mans at Road Atlanta in Braselton, Georgia. He told us then that he knew he wanted to be a race car driver.

Just after his junior year in high school, he raised the money to travel to Spain where he attended Formula BMW's licensing course (for open-wheel cars) and, later that summer, worked to pay for the Panoz Racing School (for sports cars) in Georgia. I'm not sure what most graduates spend their monetary graduation gifts on, but Keith spent his on racing gear. Along the way, the whole family caught the fever and we all work hard to assist Keith (and other young drivers) to acquire sponsorship to help them pursue their goal of a career as a professional race car driver through our family business, American Patriot Motorsports.

Of course we have concerns regarding his medical care should he be in an accident, but we always communicate his medical requirements to the racing and medical personnel at each race. Sure, we worry about him. Racing can be a dangerous sport, but I don't think we worry any more than the parents of any other racing driver. We are confident that if he were to have an accident, he would receive the medical care that he requires. Other than that, we marvel that this young man who was so weak and helpless when he was first diagnosed has achieved so much more in his twenty years than most of us will achieve in a lifetime.

So, what can your child's life be with managed CAH? Whatever he or she is willing to work hard to make it!

For more information about the NIH Natural History Study:
Carol VanRyzin • CVanRyzin@cc.nih.gov • 301-451-0399

For more information about Keith and American Patriot Motorsports, write to: American Patriot Motorsports, LLC
P.O. Box 1096, Grayson, GA 30017
Or e-mail: robbi@americanpatriotmotorsports.com

NEWBORN SCREENING UPDATE IN BRAZIL

In Brazil, neonatal screening started in the 70's, and in the year 2001 the Public Neonatal Screen Program was created. This program found that the number of screened newborns presented a wide variation depending on the state, as well as, the date of the blood sample collections. At the present time, most Brazilian states are performing screening for hypothyroidism and phenylketonuria (phase I of the national screening program), some performing tests for hypothyroidism, phenylketonuria and hemoglobinopathies (phase II), and a few are in phase III, which includes tests for the abovementioned diseases and for cystic fibrosis. However, screening for CAH is not mandatory in Brazil, and generally is done in private laboratories.

Analysis of CAH patient cohort from the Hospital das Clínicas in São Paulo state (the largest referential medical center in Brazil), evidenced that the salt wasting form comprises only 50% of cases with the severe form; and surprisingly, no more than 20% of patients are males. These data indicate that due to the absence of neonatal screening in São Paulo, patients with salt wasting form are probably dying in the neonatal period, mainly of the male sex. Despite the abnormalities of external genitalia at birth in females, which could be easily identified by clinical examination, we identified 13 girls presenting sex assignment errors at birth, and in half of them, the CAH diagnosis was performed too late to permit the correction of social sex.

To my knowledge, there are only two states in Brazil performing CAH screening with their own resources, namely Goias and Santa Catarina.

Tânia Sanchez Bachega, MD, PhD

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Nowadays, approximately 90% of the live-births are screened for CAH in Goias. Recently published data (Silveira et al, JPEM 2007) revealed the CAH incidence in 1:10,300 live-births in Goias and demonstrated the neonatal screening benefits, in increasing the frequency of the salt wasting form diagnosis and also of the number of males diagnosed. However, it was observed that the samples were collected mainly at 3 to 7 days of life, but some of them were collected after 15 days of life, which could be late for the salt wasting form diagnosis.

These data demonstrated that the CAH screening should be considered to be included in the public neonatal screening program in Brazil. Moreover, it is also important to create timely educational programs for pregnant women and pediatricians, explaining the importance of neonatal screening and the need for precocious sample collection. When there is a public neonatal screening program, the cost of CAH test inclusion represents additional US \$9.00/test in Brazil, which is not expensive. The costs should not be evaluated considering only the filter-paper measurements, but should be considered in evaluating the number of deaths avoided, reducing hospitalization

time and improving life quality, which is priceless.

If you are interested in learning more about newborn screening in Brazil and how you can help, please contact Brazil CARES Support Group Leader Isabel Wagner at (552) 126-19-3423 or isabelsw@provide.com.br.

?Se você está interessado em obter mais informações sobre triagem neonatal no Brasil e como poderia ajudar, por favor, entre em contato com Isabel Wagner, responsável pelo Grupo de Apoio e Pesquisa de Pacientes com Hiperplasia Adrenal Congênita—CARES, (tel: (21) 2619-3423; mobile: (21) 9138-6798; email: isabelsw@oi.com.br)?

Acknowledgments:

Professor Berenice B. Mendonca M.D. (Full Professor, Division of Endocrinology, School of Medicine, University of São Paulo), Eliane Santos (APAE-Goias Screening Neonatal Program), Elizabeth Silveira M.D. (Rio Grande do Sul University), Giselle Y Hayashi (APAE-São Paulo Screening Neonatal Program).

Presidential Candidates' Health Care Proposals and Positions on Science

As we try to decide how to vote in the upcoming presidential election, the following side-by-side comparison may help you to better understand the candidates' health care proposals:

www.health08.org/sidebyside.cfm
For information on their positions related to science, genetic testing, and stem cell research, please visit:

www.sciencedebate2008.com/
www/index.php?id=42.

Newborn Screening Expansion Initiatives Around the World

GO CANADA! On July 3, 2008, British Columbia Premier Gordon Campbell officially announced the expansion of newborn screening in his province from 6 to 19 conditions including testing for CAH. Thank you to all CARES' members, especially Diana Aspen, and our advocacy partners across Canada who worked so hard on this initiative. It is through your voices that this has been made possible.

While we are celebrating the inclusion of CAH in the newborn screening programs of Alberta, British Columbia, Manitoba, Northwest Territories, Nunavut (West), Ontario and Yukon Territory, the campaign for newborn screening expansion goes on in other Canadian provinces.

PROGRESS IN THE UK This June, principals involved with the United Kingdom's Newborn Blood Spot Screening Programme met to discuss adding testing for CAH to the country's screening panel. In anticipation thereof, CARES submitted a letter strongly encouraging the implementation of universal, comprehensive newborn screening including testing for CAH in the UK, but also expressing some concerns related to the inclusion of screening for CAH under current testing protocols. Current UK Newborn Screening Programme Centre guidelines call for blood spot samples to be "taken on day 5 and certainly between day 5 and day 8 for all babies." While some specialists may feel newborn screen results by day 16 of life may be good enough—at least receipt of results would coincide with crisis—CARES has made it clear that this will be too late. We expect the full panel

(including CAH) will go to the National Screening Committee in October.

IN AUSTRALIA, the fate of newborn screening appears to be in the hands of the masses. While we thought expanded screening was going to be implemented by the end of this year, the word we are receiving now is that only a pilot program is going to be run and even its start date is unclear. We are in desperate need of your stories and voices to bring attention to the plight of hundreds of babies born in Australia each year who are dying unnecessarily due to the country's failure to screen for CAH.

If you are interested in joining the campaign for expanded screening in Canada (especially our next target: Quebec), the UK, Australia or Brazil, please contact Gretchen Alger Lin via gretchen@caresfoundation.org.

Online GINA Resource

Earlier this year we celebrated the passage of the Genetic Information Non-discrimination Act (GINA) which prohibits discrimination on the basis of genetic information by health insurers and employers. Genetic testing is particularly important in families with a history of CAH as testing in the parents allows for early detection and treatment in the child. GINA prevents insurers from dropping coverage or charging higher rates for families known to carry mutations for CAH. It also prohibits employers from making decisions related to hiring, firing, promotions or training based on your CAH status. Thanks to the Coalition for Genetic Fairness, there is now an online guide to GINA, "What Does GINA Mean? A Guide to the Genetic Information Nondiscrimination Act." This tool

provides a functional outline of GINA's protections and how the legislation impacts every individual in the nation as well as hypothetical situations of genetic discrimination and key examples. To view the resource, please visit:

www.geneticfairness.org/ginaresource.html

Newborn Screening Programs Under Fire

This past July, CARES Foundation celebrated the fact that as of 2008 every baby in the United States is being tested for CAH at birth. A monumental achievement, especially considering that a mere seven years ago barely half of all states were screening for CAH! With the economic turmoil facing our country and state budgets in dire straits, however, there has been no chance to rest on our laurels. In September we received word that both the Georgia and Virginia newborn screening programs were facing possible budget cuts that would affect the timeliness of diagnosis and follow-up once a child was identified as having a condition tested for by those state's programs. CARES members and our advocacy partners across the nation have fought so hard to ensure no family has to face the physical, emotional or financial trauma of a CAH-affected child going undiagnosed at birth. The fight, however, goes on! CARES Foundation will continue, as always, to monitor the programs of every state in our nation. If we send you an email asking for your support in the face of changes to your state's newborn screening program, we hope you will add your voice to ours to make sure every baby in our nation receives a healthy state at life.

No-Sweat 2008

Thanks to all of the (over 60!) families who took part in this year's Run and made it a wonderful success!

If you didn't have the opportunity to take part but would like to do your part, there are many, many ways you can make a difference. Consider hosting a bake sale, an election party with a \$20 cover donation to CARES Foundation, a Halloween celebration where your guests treat CARES Foundation to a monetary goodie, or a fundraiser of your own creativity.

Though CARES Foundation runs fundraising campaigns at certain points of the year, programs and services to the CAH community are provided 365 days a year, and only with the support of our member families.

No-Sweat Run for a Cure

We're counting on YOU to care about our community.

If you need any help getting started, we are here to help! Please contact Ellie Avitan, Development Director, at ellie@caresfoundation.org.

Thanks to the wonderful work of our community, over \$83,000 has been raised to date through the 2008 No-Sweat Run for a Cure! Donations continue to come in for CAH research, education and support, but the team competition for prizes ended on Friday, October 3rd. Congratulations to all of our teams—you who have made great strides in advancing our mission. Thank you for taking time out of your busy lives to make a difference!

Join us in congratulating the 2008 No-Sweat Run for a Cure prize winners:

HIGHEST FUNDRAISERS:

Kimura/Ohana Team—raised \$8,616 *David Yurman earrings*

Paulson/Schauer Team—raised \$3,104 *Apple iPod Nano*

GREATEST NUMBER OF DONORS:

Team Racin' Jason—39 donors (\$2,922) *Designer Luggage*

Kenny's Krusaders—29 donors (\$2,564) *\$100 Bed Bath & Beyond gift card*

BIGGEST IMPROVEMENT OVER LAST YEAR:

Team Elise raised \$1,535 during No-Sweat 2008

versus \$1,050 in 2007—a 32% increase! *Sterling Picture Frame*

And though they didn't qualify for the team competition prizes, several CARES Foundation board members deserve special recognition for leading outstanding teams and making tremendous contributions to our total:

Jessica Upchurch (\$11,762)

Kelly Leight (\$8,122)

Diane Snyder (\$4,261)

Greg Kraff (\$3,097)

**2008 NO SWEAT
RUN FOR A CURE**

Results!



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