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## 3 $\beta$ -Hydroxysteroid Dehydrogenase Deficient NCAH: A Myth or Reality?

Ricardo Azziz, M.D., M.P.H., M.B.A.

In contrast to classic adrenal hyperplasia (CAH), which cause symptoms that are evident either at or immediately after birth, the nonclassic adrenal hyperplasias are a series of inherited disorders that cause symptoms generally near or after puberty. Signs and symptoms associated with nonclassic adrenal hyperplasia (NCAH) include signs of premature puberty, including early growth of pubic and underarm hair and advanced bone age. More commonly, post-pubertal women develop excessive facial and body hair, irregular periods, acne, and scalp hair loss (androgenic alopecia). Some women with NCAH have difficulty conceiving due to irregular ovulation as well as the very high levels of circulating progesterones, hormones made in the adrenal glands and ovaries, which causes the lining of the uterus to be unreceptive to a pregnancy.

The most common cause of NCAH is 21-hydroxylase (21-OH) deficiency, due to a genetic

defect of the gene CYP21. In fact, well over 90 percent of all CAH and NCAH cases are due to genetic mutations of this gene. There are two other theoretical causes of NCAH including genetic defects of the genes CYP11B1 (determining 11 $\beta$ -hydroxylase [11-OH] activity) and HSD3B2, (determining 3 $\beta$ -hydroxysteroid dehydrogenase [3 $\beta$ -HSD] activity). To date, few subjects have been found to be affected with 11-OH deficient NCAH, the vast majority of them are children, and no patient with genetically confirmed 11-OH deficient NCAH has been diagnosed in adulthood.

Previous reports have suggested that 3 $\beta$ -HSD deficient NCAH is one of the most common causes of androgen (male hormone) excess in women. This assumption was based on the finding that many hyperandrogenic patients had elevated levels of 17-hydroxypregnenolone and DHEA; both of these steroids are immediate precursors to the 3 $\beta$ -HSD enzyme. However, others and we

*continued, see **Overdiagnosis** on page 16*

## You're Invited!



*"bringing the CAH community together"*

Seton Hall University  
South Orange, NJ

**\*see page 23 for details**

### Attention Federal Employees: Donate to CARES CFC #2937

CARES has been approved for a second year as a member of the Combined Federal Campaign (CFC) operated by the Office of Personnel Management (OPM). The CARES Foundation code is #2937. **The 2006 campaign will run from Sept. 1st to Dec. 15th.** Please speak to your employer about enrolling and encourage family and friends to enroll as well. For more information, visit the OPM website:

<http://www.opm.gov/cfc/index.htm>

## A Message from the Executive Director

*“When written in Chinese, the word ‘crisis’ is composed of 2 characters. One represents danger, and the other represents opportunity.”*

*(John Fitzgerald Kennedy, 35th US president, 1917-63)*

Dear Friends,

### The Crisis

The quotation above epitomizes the experience of CARES Foundation over the last few months. The morning of February 17, 2006 changed so much for us. Watching our office building burn down, and with it all of our possessions, five years of work product, our server and our precious database, was devastating. Our on-site server back-up was destroyed as well, so we irretrievably lost all data. After calling our insurance agent, we realized we were dreadfully under-insured. An overwhelming anxiety and fear gripped the staff. We were heart-broken and thought perhaps this was the end of CARES.

Within hours of the fire, the phone calls from our friends began, and you opened up your checkbooks, gave of your time and rescued CARES. Our community—and indeed we are a community in the truest sense—came to our rescue and provided the moral, volunteer and financial support we so needed. You cannot imagine how much your calls, emails and letters meant to us all. Many made us cry—tears of joy to know that you cared about us and the work we did for the CAH community. You kept us going during a time when it was often tempting to give up. The financial support was and is essential, as we could not have made it without the extra help. I, we, cannot thank you enough for keeping us going during that difficult time. Please know how much you all mean to us and how much we appreciate your support. This is the meaning of community—being there for each other. You were there for us, so we can be there for you. May God Bless you all.

### New Opportunities

Thanks to your generosity and the



generosity of the local community, our foundation and corporate donors, we have been able to move into new offices in Union, New Jersey and buy new (used) furniture, computers and office equipment. The local community came out in force. Rabbi Daniel Cohen from Temple Sharey Tefilo-Israel dropped off two new computers and two new printers just days after the fire, paid out of his own pocket. The youth group from the synagogue also held a fundraiser for us. The local Rotary Club, Junior League, local school groups and Whole Foods Market all raised funds for CARES. The Healthcare Foundation of New Jersey, the Karma Foundation, RolexUSA, Inc. and PerkinElmer, Inc. all made generous donations to help get CARES back on its feet. With these generous gifts, we have been able to re-establish our operations and are stronger than ever!

We also thank the volunteers who helped with countless hours of data entry and, most of all, our dedicated, hardworking and tireless staff for never giving up and working so hard to rebuild CARES under these extremely difficult circumstances.

We are settled into our new offices, just a couple of miles from our old office and are so happy to be in our new home! Please come for a visit anytime!

### Celebrating Five Years of Saving Our Babies

On June 17, 2006, we held our first fundraiser in New Jersey. It was a wonderful evening. About 200 people turned out to honor former Governor Donald DiFrancesco for signing an

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*This newsletter is published 3 times a year.*

executive order five years ago that has led to saving the lives of almost 300 New Jersey babies, including many with CAH. Special thanks are due to our chairs, Adam Leight and James Raphalian and to our hard working committee members, as well as our generous corporate sponsors, including: **Pfizer, Inc., Keyspan Energy, and Commerce Bank.** We also thank all of those who gave generously to the event, attended and to those who donated to the auction. We raised over \$117,000 for CARES and for CAH research!

#### Casino For CARES

Many thanks to our chair of this lovely event, Monica Heinze, who almost single-handedly organized this event. About 80 people joined her on July 29th in Denver, Colorado for a fun-filled evening of casino games to raise over \$14,000 for CARES! Many thanks to Monica and to all who attended and supported the event!

#### New Members of the Board of Trustees, Medical and Scientific Advisory Board, and New Staff

We are pleased to welcome Gregory

Kraff and Monica Heinze to our Board of Trustees. Both Monica and Greg have children affected by CAH and are long-time, active members of CARES. We are delighted to have them join our board. We are also honored to have two new members of our Scientific and Medical Advisory Board, Dr. Henry Anhalt of St. Barnabas Medical Center and Dr. Maria Vogiatzi of Weill-Cornell New York Presbyterian Hospital. You can read more about them on page 13. We also welcome new staff members, Jennifer Mann Rosenblatt, our Director of Development and Mazal Wolfskehl our bookkeeper. They make wonderful additions to our CARES team!

#### In Other News

We mourn the loss of our dear friend and wonderful supporter David Abell. Our hearts and minds are with his family and friends. Please see page 13 to learn more about David and his wonderful contributions to humanity.

We thank Peregrine Charities for their generous grant that will allow us to continue funding a natural history protocol at NIH

(see below).

We thank Dan Hackett and his father Steve Hackett for approaching Bibbero Systems on our behalf. Bibbero selected CARES for its employee giving/ matching gift program. To learn more, please see page 14.

Finally, we thank the Cornell Institute for Pediatric Urology for generously sponsoring this edition of the newsletter. Our newsletter costs several thousand dollars to print and distribute and remains completely free of charge for all CARES members. Thank You!

#### Gratitude

*"At times our own light goes out and is rekindled by a spark from another person. Each of us has cause to think with deep gratitude of those who have lighted the flame within us."* (Albert Schweitzer, M.D., German theologian, philosopher, and physician, 1875-1965)

With Warmest Regards,

*Kelly*

## CARES Awarded \$50,000 from Peregrine Charities

CARES Foundation is excited to announce its receipt of a \$50,000 grant from Peregrine Charities to aid in funding clinical research for Congenital Adrenal Hyperplasia (CAH).

This grant will be used to continue the funding of seminal new research, a Natural History Protocol of CAH, as being conducted by Deborah Merke, MD, MS, Chief of Pediatric Services for the National Institutes of Health Clinical Center.

The outcome of this research protocol will be a foundation for the study, treatment, and hopefully, a cure of individuals with CAH.

As with any disease, research is imperative to understanding the basic

cause. Through a complete and thorough examination of a disorder, effective and groundbreaking treatments can be established.

CARES Foundation is extremely grateful to Peregrine Charities for its commitment to the continuation of such research.

Peregrine Charities has been formed as a grant-making family foundation to help under-funded and underserved populations—families afflicted by rare or "orphan" diseases. The Wasendorf family chose to start this philanthropy to assist needy organizations.

Peregrine Charities was established as a 501(c)3 in May of 2004 and hopes

to give worthy organizations over one million dollars in funding from this private Wasendorf family foundation on an annual basis.

For more information on Peregrine Charities please visit [www.peregrinecharities.org](http://www.peregrinecharities.org).

To learn more about the CAH Natural History Study, please see **page**

**22.** 



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# We Get By With A Little Help from Our Friends

It's during tough times when you find out who your true friends are, and CARES has definitely seen some tough times. Although we faced a devastating fire in February that destroyed our offices, we never closed our doors—**not even for one day**. The only way we were able to do this was through the generous support of our friends, who rallied on our behalf with emergency funding, donations, and help with fundraisers. We cannot thank you all enough!



**Rotary Club to the Resuce!** The Rotary Club of Chatham, Millburn & Short Hills provided CARES with emergency funding in the wake of the fire that destroyed CARES offices.



**Junior League Saves the Day!** The Junior League of The Oranges and Short Hills came to CARES' aid in the weeks following the fire with the donation of supplies and a generous grant for rebuilding.

Special thanks to our corporate and foundation donors who helped us through this most difficult time.

*Healthcare Foundation of NJ*

*Karma Foundation*

*Rolex USA, Inc.*

*Pfizer, Inc.*

*PerkinElmer*

We would also like to thank our numerous members who so generously gave and allowed us to continue our work as the only US organization devoted entirely to the CAH community.



**Grilling for a Good Cause:** Kelly, Josh and Alyssa Leight help Whole Foods employees sell snacks. Whole Foods of Millburn, NJ chose CARES for its "Good Will From the Grill" community fundraiser. Whole Foods employees volunteered their time to grill burgers and sell healthy snack foods, raising almost \$500 for CARES Foundation.

*\*\*\*Fundraising Updates continued on pages 14-15*

# Celebrating Five Years of Saving Our Babies

On June 17, 2006 CARES Foundation celebrated five years of expanded newborn screening in New Jersey with “Celebrating Five Years of Saving Our Babies,” which honored former Governor Donald T. DiFrancesco.

The event, held at the Maplewood Country Club, Maplewood, NJ, was fantastically successful and featured a casino and raffle as well as silent and live auctions.

In 2001, Governor DiFrancesco signed an Executive Order that dramatically expanded newborn screening in New Jersey and set up a structure for future expansion. Because of this action, hundreds of New Jersey babies were saved.

In addition to the awareness the event brought to New Jersey’s newborn screening program, attention was also brought to CARES and its mission of Research, Education and Support. Through the hard work of the Event Committee and our dedicated staff, CARES raised over \$117,000 for its programs. This is due, in large part, to our wonderful corporate and private sponsors which include:

**Pfizer, Inc.**

**KeySpan Home Energy Services**

**Adam & Kelly Leight**

**Commerce Bank**

**James & Hope Raphalian**

**Rolex USA, Inc.**

**John & Carolyn Ferolito**

**Joel & Ethel Sharenow**

Our sincerest gratitude goes to everyone who helped make our second major fundraiser such a tremendous success. We couldn’t have done it without you! 💙



**From Left: Erin Anthony, Meryl Stone, Kelly Leight and Mariel Vargas pose with Governor Don DiFrancesco at the Maplewood Country Club, Maplewood, NJ. A 2001 Executive Order signed by Gov. DiFrancesco dramatically expanded New Jersey’s Newborn Screening Program, saving hundreds of babies.**



**Alyssa Leight pulls the winning raffle ticket. Jami Abell-Patterson (CA) was the lucky winner of the Women’s Rolex Watch generously donated by Rolex USA, Inc.**

## Personal Story

## Shelby's Trips

*Julie Boutilier is a mother from Indianapolis, IN. The following was adapted from an email she shared with us while traveling with her daughter to North Carolina. We are very grateful she offered to share her family's story with us.*

By Julie Boutilier

To those of you out there that know Shelby and know about our trips to Chapel Hill, I thought you should hear about our most recent trip. To give a little background, Shelby is participating in a clinical trial study for a new drug that could someday be very beneficial to those with CAH. It could lower doses of steroid, while promoting increase in growth and lowering weight gain—a very worth while effort by the doctors of UNC at Chapel Hill.

In order to participate, Shelby takes this drug daily and we make a visit to UNC once a month. The tests and the drugs cost nothing, but the trips can be quite costly. There are people that pour out their hearts and pocket books and skills to people they do not know. I want to tell you about these people, although I can not tell you their names—they wouldn't want me to.

#### From the Beginning

It all started with Shelby and me needing a flight. My dad knew a pilot who was part of an organization called Angel Flight. These people ARE angels. They fly kids in need to different hospitals around the country, free of charge. This pilot, that my dad knows, set up one such flight for us. However, due to bad weather, the flight was cancelled. Even though we couldn't use Angel Flight for this trip, this gentleman, along with some other pilots at the Greenwood Airport, took up a collection for Shel. It was not a small amount of money and it is what we use to pay for food and expenses when we make the trip.

We were finally able to take one of the Angel Flights. It is two legs of

flights, one from Greenwood, IN to Beckley, WV and one from Beckley to Raleigh, NC. Our trip home was the same. I was humbled by the kindness those men showed Shelby and me. They fly their own planes, with their own gas and time for total strangers.

#### About Our Pilots

Our first pilot was in his 60's with grown children and a beautiful plane. He was active in volunteering for Ducks Unlimited and other organization and flew for us. Why? I don't know.

The second man's aircraft was much older and a little slower, but the man had the same background. He, too, was in his 60's,

volunteered for a soup kitchen and was on it's board, had a wife of 40 years, grown kids and had cancer. Yeah, he had his own problems, but he wanted us to be his 45th Angel Flight.

#### Our Journey

This journey keeps getting more and more intense for us. We stayed at the Ronald McDonald house. In our room was a journal of those who have been here before us. They speak of little lives that are in one place—in God's hands. No one else knows what will happen. Every one of the letters acknowledges a total lack of control.

So, that's it. That is what I'm here to tell. This woman, who a year ago was in tears and panic stricken at the thought of flying, has now been on over 20 flights in 5 months. I'm not over it yet, but through the knocking knees and bloody noses I'm learning. Like each parent in the journal knows, we just aren't in control.

#### Good News

Shelby has lost a pound and gained an inch as of this appointment. Hopefully, this is a sign of good things to come. Our flight going home on

Angel Flight was cancelled due to

ice in the clouds (SCARY), so we were left in


Chapel Hill a day longer than expected. (It was

70 degrees there, and the cherry

blossoms were in bloom, so this

was not a hardship). We took a commercial flight

home the next day. The people at Enterprise Rental

knew we were with Angel Flight and waved any extra fees for our already reduced car rental, and the Ronald McDonald house is let us stay an extra night. 

*"There are people that pour out their hearts and pocket books and skills to people they do not know. I want to tell you about these people. . ."*

If you have a story you think would be beneficial to our audience and would like to share it, please email [info@caresfoundation.org](mailto:info@caresfoundation.org) or call (toll free) 866-227-3737.



## Quest Indigent Patient Program Helps with Lab Costs

As many families know, managing the medical costs of CAH can be difficult—especially when without health insurance. Quest Diagnostics' Indigent Patient Program is designed to help uninsured patients who cannot afford to pay for their clinical laboratory testing manage their laboratory costs.

### Eligibility

- *The program is available to uninsured patients who cannot afford to pay their bill.*
- *Certain information, such as your W-2, is required to verify your income.*
- *Those eligible for federal food stamps, Medical Assistance, or the Hill-Burton Program will qualify.*
- *You may be required to submit proof of your eligibility for one or more of these programs.*

If you do not currently qualify for these programs, Quest will take into account your income and family unit size based on guidelines provided by the US Department of Health and Human Services. These guidelines are updated on an annual basis and are available at the HHS website.

**Because of certain federal and state regulations, Quest cannot waive co-payments or deductibles from insurance companies. Therefore, Quest's Indigent Patient Program is only available to patients who do not have health insurance coverage.**

### How to Apply

If you cannot afford to pay your bill and you are uninsured, call the telephone number listed on your invoice. Ask the Billing Representative for a Quest Diagnostics Indigent Patient Program application. Return the application to Quest within 30 days of receipt, so that they can place your bill on hold while they make their determination.

Based on your request and financial income, Quest will work with you to determine whether or not you qualify for their program. They will notify you whether or not you qualify for financial assistance from Quest Diagnostics and indicate what portion of the invoice has been credited. If you do not qualify, they will develop a payment schedule for you. For more information on this program, please visit [http://www.questdiagnostics.com/patient/indigent\\_program.html](http://www.questdiagnostics.com/patient/indigent_program.html).



*Angel Flight® America (AFA) provides access for people in need seeking free air transportation to specialized health care facilities or distant destinations due to family, community or national crisis. For more information on the organization, please call 877-858-7788 or visit*

*[www.angelflightmaerica.org](http://www.angelflightmaerica.org).*

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A portion of all proceeds will go to CARES Foundation, Inc.

# Interpreting research results: *The dangers of scientific jargon*

Melissa Colman, M.A. and David E. Sandberg, Ph.D.  
Psychiatry & Pediatrics  
School of Medicine and Biomedical Science  
University at Buffalo, The State University of New York

Recently, Dr. Knickmeyer and colleagues conducted a research study involving people with congenital adrenal hyperplasia (CAH); the study was titled “Androgen and autistic traits: A study of individuals with congenital adrenal hyperplasia.”<sup>1</sup> For parents of affected children, titles such as these can sound quite alarming – leaving parents to wonder if, just because their child has CAH, he or she might also have or develop autism. Autism is a developmental disorder characterized by severe impairment in communication, social interaction, and creative or imaginative play. **In short, the answer to that question is “no.”**

Prior to conducting this study, other research showed that autism is more prevalent in males than females. It was thought that increased testosterone during prenatal development might be a factor in why more males have autism than females. The authors attempted to test the hypothesis that autistic traits (eg, poor social skills, communication, imagination, and attention switching) are associated with high prenatal testosterone levels by measuring those traits in people whose prenatal testosterone levels were higher than average due to CAH.

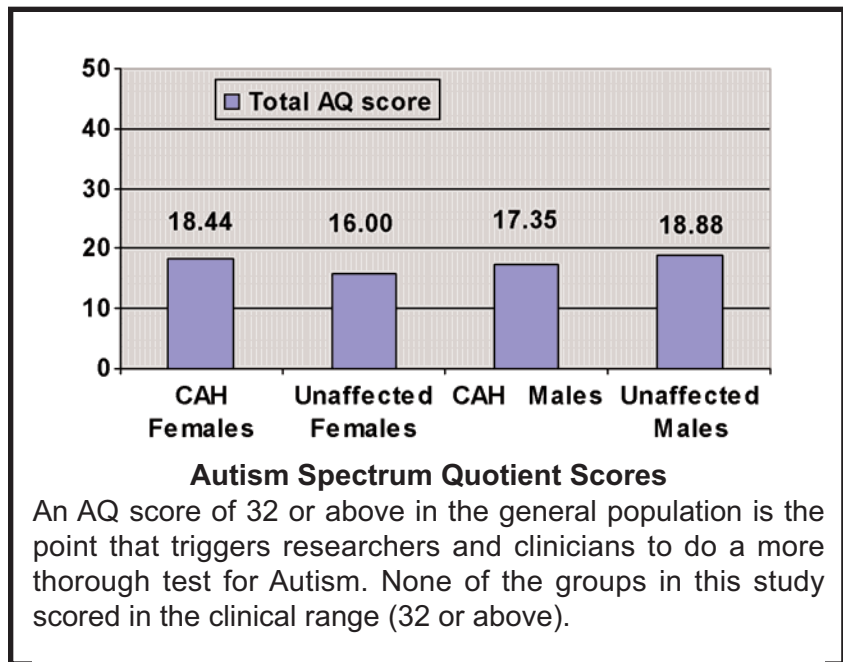
Sixty research participants with classical CAH (34 females, 26 males; 53 with 21-OH deficiency salt-wasting CAH), and 49 unaffected relatives (24 females, 25 males), ages 12 to 45 years, were recruited through pediatric endocrinology clinics and a CAH support group in the UK. Information

regarding participation rates was not recorded. Each participant filled out a questionnaire called the Autism Spectrum Quotient (AQ) that assessed five areas: social skills, communication, imagination, attention to detail, and attention switching. On the AQ, scores can range from 0 to 50 with higher scores indicating more characteristics labeled as “autistic.” A score of 32 or above in the general population is the point that triggers researchers and clinicians to do a more thorough test for autism.

After participants completed the questionnaire, researchers compared the

unaffected males in this study had significantly higher total AQ scores than unaffected females. In addition, females with CAH scored higher (that is, in the male direction) on total AQ than unaffected females. In contrast, total AQ scores of males with and without CAH did not differ statistically. Another way to look at it is shown in the figure below.

As you can see, on average, while affected females scored higher than the unaffected females, none of the groups scored in the “clinical range” (the clinical range is the range at which you might suspect participants were



scores of four groups of participants: CAH-affected females, unaffected females, CAH-affected males, and unaffected males. They found that

autistic). In other words, this study shows a subtle shift in some behaviors for girls and women who have CAH. Females with CAH showed AQ scores

*continued on next page*



continued from page 8

similar to that of males (affected and unaffected by CAH); **it did NOT show that they behaved like individuals with autism or associated disorders in the autistic spectrum.**

Studies like this one help illustrate an important lesson to keep in mind while reading research: **the goals of science and the goals of parents are often very different.** Much of the scientific research on CAH was initiated

so that we could learn about how biological factors (for example, androgens) affect our behavior and psychological development. Scientists who study and conduct research on CAH are most interested in general principles that influence human development. In contrast, your goal as parents is quite different: you are interested in how your own child will grow and develop. There is a lot of

research and information out there that speaks to the general principles of development for people with CAH and some of this will apply to your son or daughter; other parts won't. Your child is special. He or she, with your help, will grow and learn. And you'll learn more about your son or daughter as he or she grows. ❤️

<sup>1</sup>Knickmeyer R, Baron-Cohen S, Fane BA, Wheelwright S, Mathews GA, Conway GS et al. Androgens and autistic traits: A study of individuals with congenital adrenal hyperplasia. *Hormones and Behavior* 2006; 50:148-153.

## Insulin Pump for Hydrocortisone Delivery?

Peter Hindmarsh, M.D.

CAH is a challenge to manage, as the current medications available (hydrocortisone, prednisone and dexamethasone) cannot mimic easily the normal circadian rhythm of cortisol in the circulation. This problem arises because of the way in which these medications are administered, so that inevitably there are periods of over, normal and under exposure to the glucocorticoid during the 24-hour period.

The critical time period for glucocorticoid exposure is from midnight through to the late afternoon, when the ACTH drive from the pituitary is at its maximum. With the exception, perhaps, of dexamethasone, it is not easy to deliver glucocorticoid in a way in which ACTH suppression can easily be achieved, as this would, in many instances, require dosing after midnight in order for suppression to last through to the next morning. The situation can worsen during puberty when the clearance of hydrocortisone from the circulation is increased, necessitating more frequent hydrocortisone dosing.

As the basic principle of endocrine

therapy is to deliver replacement therapies in as physiological a manner as is possible, consideration needs to be given as to how this might be achieved. Two options are available. One is to modify the properties of hydrocortisone tablets into a slow release preparation. The other is to use other technologies to deliver hydrocortisone.

The latter is what we have done recently in someone who cleared hydrocortisone very quickly. We used an insulin pump system to deliver small amounts of hydrocortisone, with variable amounts programmed in on an hour-by-hour basis, so that we could mimic the normal circadian rhythm and get good control of the CAH in terms of 17-OHP and androgen concentrations.

While this approach of wearing a pump and delivering the hydrocortisone subcutaneously may

not appeal to everyone, it is certainly worth thinking about in situations where hydrocortisone drug delivery is difficult. ❤️

*Peter Hindmarsh BSc, MBBS, MD, FRCP, FRCPC is a Paediatric Endocrinologist specializing in Adrenal Diseases and Diabetes at the Great Ormond Street Hospital in London, UK.*

*He has notified CARES that he will advise any physician wishing to try the pump for his or her patient.*



The pump shown here is the MiniMed Paradigm® made by Medtronic MiniMed, Inc. It is about the size of a cell phone.

*Personal Story*

# Chris' Story

Chris is a teenager from the UK whose CAH is now managed using a pump normally reserved for diabetes. The following is his mother Kathy's account of his treatment. We are very grateful that Chris and his family offered to share their story with us.

I think one of the hardest things in life is to see someone you love ill and not be able to help them. So, as I relate the following, know that it does not touch the emotions and anxieties our family has felt over the past few years. I feel that CAH is such an individual disorder, in that every person has it to varying degrees and reacts to the medication differently, which makes it such a difficult condition to treat. Some children seem to have no problems and manage very well, whereas others do battle as my son Chris has.

However, I would like to share with you all what I consider an exciting new treatment for CAH! It is a pump, which is used mainly for diabetics but delivers hydrocortisone subcutaneously at a continuous rate of infusion.

My 18-year old-son Chris has SWCAH and has been using this pump since October 2004. For us, it has been nothing short of miraculous! In Chris's words, the pump has given him back his life!!

Almost from the time Chris could first talk, he complained of very bad headaches. He would also always tire very quickly and continually battled with his weight. When he started puberty, things got even worse. Chris's levels became almost impossible to control and his general health deteriorated significantly. He started to lose weight rapidly; had no energy and no muscle tone; often suffered from severe nausea and lived with a constant headache. At times, he would faint for no reason. During this period, I often had to stress dose him with

hydrocortisone.

As you can imagine, I became extremely desperate. We then changed endocrinologists to Professor Peter Hindmarsh, who took the time not only to listen but hear all our problems and luckily took a special interest in Chris. After a lot of testing, he discovered that Chris was not absorbing the cortisol properly and was metabolising his medication extremely quickly. At one point, Chris was on a total dose of over

**“In Chris' words, the pump has given him back his life!!”**

100 mgs of hydrocortisone, given over 6 doses a day, yet still his CAH was completely out of control.

Prof Hindmarsh then spent months developing a formula for Continuous Subcutaneous Hydrocortisone Infusion via a pump. He worked out the doses so that the pump could deliver hydrocortisone at rates which mimic natural circadian rhythm.

In October of 2004, Chris was started on the pump. Miraculously, his 17-OHP level came down to normal ranges within hours! His androstenedione and other adrenal hormone levels all also came down to within normal range. Even more amazing: after several days, Chris was headache free for the first

time in years and his nausea disappeared. After missing 2 years of school because of illness and requiring a home tutor, he is now back at school and has just written his AS levels. As I said, he has been literally given his life back!!

The pump is so efficient, that we were able to reduce Chris's total hydrocortisone dose after two weeks. Over time, we have been able to continue to reduce the dose and his CAH now is very well controlled!

The other remarkable thing is that Chris has also started to grow again, after starting on the pump. He has even gone up a shoe size and after years of battling weight, either being too heavy or too thin, his weight is now stable.

Everyone is amazed at the dramatic improvement in Chris's health and his appearance! The terrible dizziness and nausea he used to suffer is a thing of the past! Chris had sinus surgery earlier this year which he sailed through, whereas previously he was always unwell after an anaesthetic. Chris's blood pressure is now stable, his heart rate has improved dramatically, and he no longer suffers the terrible headache that even painkillers would not touch. It is fair to say that he has never felt as well in his life as he does now he is using the pump!

Chris was at first apprehensive when changing the infusion site, (where the fine cannula enters his skin), however it is now part of his normal

*continued on page 20*

## Kid's Corner

Hello, my name is Lillian Nicole Kelly. I am 13-years-old and I have salt-wasting CAH.

I think sometimes it's not fun to have CAH, because I have to go to the doctor all the time and always have to have blood work done. Some of the kids at school don't understand why I miss school so much.

In the summer, I have to be careful not to get too hot. One time when I was playing softball, I got too hot and passed out. When I play basketball, I get too hot from running.

I wish that all of us with

CAH would have a place to go in the summer so we could get together and have time to know more kids our own ages with CAH. Maybe some day, I got to meet Kelly and her daughter once, it was great. They are so nice.

If you have a story, picture or poem to share in our next newsletter, please send it to [erin@caresfoundation.org](mailto:erin@caresfoundation.org).



CARES thanks Ohio members **Laura Williams & Alicia Meddles** for organizing

### BRYAN'S BENEFIT COUNTRY CONCERT IN THE PARK

an extremely successful fundraiser that featured Blue Grass and Country Music by

**Huckleberry Blue  
Pickin' N' Grinnin'  
and  
Up "4" Grabs**

The event raised \$3,000 for CARES and CAH research.

**Way to go, Laura and Alicia!**

## Run, Josh, Run!!!

CARES congratulates Josh Eisenberg on running his second Chicago Marathon, with over 35,000 runners and 1.2 million spectators.

Josh has generously asked his sponsors to donate to CARES. If you would like to sponsor Josh, please let us know. We would also like to encourage all members living in the Chicago area to come out and support Josh on race day!

**LaSalle Bank  
Chicago Marathon**

**Sunday, Oct. 22, 2006  
Millenium Park**

For more info:  
[www.chicagomarathon.com](http://www.chicagomarathon.com)



On March 4, 2006, the **Florida Family Support Group** got together at Lake Seminole Park to hear guest speaker Dorothy Shulman, M.D. and enjoy each other's company.

Thanks to Sonya Matson and Pat Tovar, the Florida Family Support Group continues to be one of CARES most active groups. (Way to go, FL!!!) If your support group is getting together, please let us know!

If you're interested in starting a support group or advertising an event, please give us a call so we can get you started.

**(toll free) 866-227-3737**



## News From the CARES Office

### New Offices!

We are thrilled to report that we have moved out of Kelly's home and into new office space. Our office is big and bright and we look forward to filling up the space as CARES expands. Our new address is:

**CARES Foundation, Inc.  
2414 Morris Ave., Suite 110  
Union, NJ 07083**

Please note that our phone numbers remain the same (In NJ) 973-912-3895 and (toll free) 866-227-3737.

### New Staff!

We are excited to welcome two new members to our staff, Mazal Wolfskehl and Jennifer Mann Rosenblatt.

Mazal brings extensive accounting experience to our organization as our new bookkeeper and Jennifer adds her experience from The Christopher Reeves Foundation and The March of Dimes as our new Director of Development.

### New Website

We are in the process of upgrading our website and developing a mirror site in Spanish. The new site will be easier to navigate and have a more updated look. This project should be completed shortly, so please let us know what you think!

### Remember to Re-register

We are making progress rebuilding our database but still need your help! If you haven't re-registered (so we have your current address, phone number, email and info about your family), please do so ASAP. Visit [www.caresfoundation.org](http://www.caresfoundation.org) and click "Join" or give us a call 866-227-3737.

### 2007 Conference

Believe it or not, we've already begun planning our 2007 CAH Conference. Mark your calendar for November 3, 2007 in Los Angeles, CA.



**From left: Erin Anthony, Meryl Stone, Kelly Leight, Mariel Vargas and Mazal Wolfskehl meet in CARES' large conference room. Our larger and brighter space inspires our already motivated staff and finally allows us to host our own board meetings!**



**Hot off the presses: Kelly Leight poses with our new Ricoh copier, provided by a generous grant from the Smart Family Foundation. The copier has been instrumental in getting us back on our feet, and we are forever grateful to the foundation for allowing us the ability to purchase this essential tool.**

**Thank You!!!**

## Remembering a Dear Friend and Remarkable Man

We are sad to report that David L. Abell, a very dear friend to CARES, passed on February 11, 2006.

Mr. Abell, whose daughter Jami is a member of CARES' Board of Trustees, was described in the LA Times as "the music vendor to the stars" and "an angel to the community."

Founder of David L. Abell Fine Pianos, Mr. Abell showed his tremendous commitment to the jazz community through the establishment of several educational programs and scholarships in the Los Angeles area. He was honored in 2005 by the Friends of Jazz at UCLA with their Humanitarian Award as well as by the NAACP with its Community Service Award.

Since the beginning of his

involvement with CARES, Mr. Abell was a very generous supporter of the organization and instrumental in the success of CARES' first major fundraiser, the EVERY1CARES luncheon. At the request of his family, those paying tribute to David were asked to continue his legacy with donations to CARES, and the David L. Abell Memorial Fund for Research was established. We continue to be touched by the amazing generosity of David's friends and family.

Our thoughts continue to be with the Abell family as they look forward and celebrate the life of a truly remarkable man. 💙



David was a great humanitarian and friend to CARES. He will be sorely missed by all who knew him.

## CARES Welcomes New Medical Advisors

Henry Anhalt, D.O. & Maria Vogiatzi, M.D.

CARES Foundation is pleased to welcome Drs. Henry Anhalt and Maria Vogiatzi to its medical advisory board.

Dr. Henry Anhalt is the Director of the division of pediatric endocrinology and diabetes at the Saint Barnabas Medical Center. He is Associate Professor of Clinical Pediatrics at the State University of New York at Downstate Medical School, and Co-Director of the Healthy L.I.F.E. program at Saint Barnabas Medical Center, a new multi-disciplinary obesity treatment program.

Dr. Anhalt completed his post-doctoral fellowship in pediatric endocrinology at the Lucille Salter Packard Children's Hospital at Stanford University and was a recipient of the

prestigious Dean's fellowship award. Prior to his appointment at Saint Barnabas Medical Center, Dr. Anhalt was the Director of the division of pediatric endocrinology at the Infants and Children's Hospital of Brooklyn at Maimonides Medical Center.

Dr. Anhalt's expertise in the treatment of childhood obesity will add depth to the advisory board.

Dr. Maria Vogiatzi is Chief of the Komansky Center for Child Health at NewYork-Presbyterian Hospital Weill Cornell Medical Center. She conducts clinical research in the areas of growth, congenital adrenal hyperplasia (CAH) and osteoporosis.

Dr. Vogiatzi received her medical degree from Aristotelian University Medical School in Greece. She was

trained in Pediatric Endocrinology at NYPH/Cornell University and at Baylor College of Medicine in Houston. She is Board Certified in both Pediatrics and Pediatric Endocrinology.

Dr. Vogiatzi joined the department of Pediatrics at Cornell University in 1997. She has great experience in the treatment of children with short stature who may require therapy with growth hormone, in the management of patients with disorders of puberty, and in congenital adrenal hyperplasia. She has published on numerous topics in Pediatric Endocrinology.

CARES Foundation is excited about the addition of Dr. Anhalt and Dr. Vogiatzi to its medical advisory board and looks forward to working with them. 💙

# Blue Jeans & Sequins a Success!!!

On July 29, 2006 the "Blue Jeans and Sequins" Fundraiser was held at the JW Marriott Cherry Creek in Denver, Colorado.

The night included a silent auction with a casino as well. We raised over 14,000.00 after all the expenses were paid.

I'd like to extend a special "Thank You" to our sponsors: Quest

Diagnostics, Belles and Beaus Academy, Tom and Kim Konkell, Tom and Tricia McCann, Chip and Melinda Hunnings, and Dean and Colleen Pisciotta.

We had close to 80 friends and family join us and we hope to double the numbers with our next event.

Most of all, we, the Heinze Family, would like to thank CARES for all they

do and have done for us and all the other CARES members.

Please remember if you are in the Denver area to join us for our next event and bring your friends! Everyone that attended the event had a great time and we're sure to have an even better time next year.

*~Monica*

Monica Heinze, Event Chair

## Employee Giving

We thank members

**Stephanie Grubler**

**Dan Taylor**

**Mary Lynn Holly**

**Karen Wilson**

**Laura Shine**

for designating CARES in each of their companies' employee giving programs. We also thank all members, their friends and families who donate through the United Way and CFC (#2937).

**We truly appreciate your support.**



**Gabi Flaum raised money for CARES with a Snack Sale at Temple Sharey Tefilo-Israel, South Orange, New Jersey. Thank You, Gabi!!!**

## Bibbero is the Best!

We are tremendously grateful to the wonderful people at Bibbero Systems, Inc. for choosing CARES in their employee giving campaign. Donations from Bibbero employees are matched by the company and making a huge difference in our ability to provide free services to the CAH community and fund research for a cure.

We are particularly grateful to Steve Hackett for approaching Bibbero on our behalf and Dan Hackett, Steve's son, for educating Bibbero employees about CAH and CARES.

Of course, none of this would be possible without help of Bibbero President Mike Buckley for choosing to match employee donations and the Bibbero employees for choosing CARES.

**Thank You!!!**



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NEW

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### Overdiagnosis of 3 $\beta$ -HSD

have demonstrated that the elevations in 17-hydroxypregnenolone and DHEA present in hyperandrogenic women are relatively mild, generally not exceeding two-fold the upper normal limit found in healthy women. In contrast, the level of 17-hydroxypregnenolone (17-OHP), the immediate precursor to the 21-OH enzyme, is 3-fold to 10-fold greater than the upper normal limit in NCAH patients. In addition, a number of investigators, including ourselves, have noted that between 25 percent and 40 percent of women with the polycystic ovary syndrome (PCOS), another fairly common cause of androgen excess in women, have elevated levels of DHEA and DHEA sulfate (DHEAS), suggesting that the adrenal may be overactive in patients with PCOS, not related to the presence of a genetic mutation.

This data indicates that we were over-diagnosing patients with 3 $\beta$ -HSD deficient NCAH, and that many of these women simply have PCOS with a hyper-reactive adrenal, similar to the hyper-reactive ovary also observed in PCOS. More recently, Dr. Songya Pang and colleagues have done very careful genetic studies of patients with supposedly 3 $\beta$ -HSD deficient NCAH and observed that the patients who had genetic defects of the 3 $\beta$ -HSD gene had 17-hydroxypregnenolone values that were at least 20-50 standard deviations above age-matched controls (at least 10-fold the upper normal limit), and all of these presented in childhood. **In fact, to date, there has been no patient with 3 $\beta$ -HSD deficient NCAH who presented as a hyperandrogenic adult woman.** These and other investigators have also observed that the vast majority of individuals who


**"This data indicates that we were over-diagnosing patients with 3 $\beta$ -HSD deficient NCAH..."**

have elevated or exaggerated 17-hydroxypregnenolone or DHEA values following ACTH stimulation **actually have a form of PCOS and do not have NCAH.**

What is the difference? Patients with PCOS generally have insulin resistance and, while they have a genetic basis that underlies the disorder, the genetic cause is a complex mixture of various genes and the environment. Furthermore, the use of glucocorticoids (dexamethasone, prednisone and hydrocortisone) in patients with PCOS is generally not recommended since it can worsen the insulin resistance. Alternatively, patients with true NCAH may often (but not always) benefit from glucocorticoids and should also be counseled carefully regarding their chance of having a child who is also affected with NCAH or CAH. Consequently, making and treating the true diagnosis that underlies a patient's

hyperandrogenic symptoms is very important.

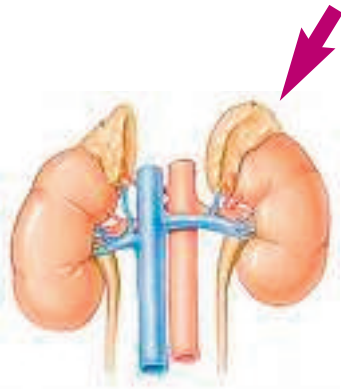
In summary, the 3 $\beta$ -HSD deficient NCAH has been woefully over-diagnosed, particularly considering modern studies which are able to determine whether a patient actually has a genetic defect of the 3 $\beta$ -HSD gene. In fact, to date, no woman presenting with hyperandrogenism in her adulthood (or even adolescence) has been confirmed to have 3 $\beta$ -HSD NCAH. **The vast majority (if not all) of women with an exaggerated (above normal) 17-hydroxypregnenolone or DHEA levels have PCOS.** This should be a relief to many patients who are being inappropriately treated with glucocorticoids based on older criteria.

Patients should not be surprised at the general change in diagnostic criteria for 3 $\beta$ -HSD deficient NCAH, since it is difficult to establish the presence of adrenal hyperplasia without proper genetic tools, only made available to us in the past decade and often only in research laboratories. As these genetic tools become more widely available for clinical use, it is highly likely that the reported incidence of "3 $\beta$ -HSD deficient NCAH" (and "11-OH deficient NCAH") will drop dramatically. 

**"This should be a relief to many patients who are being inappropriately treated with glucocorticoids based on older criteria."**

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The adrenal glands are small, walnut-sized organs that sit on top of each kidney. They may be hyper-reactive in some patients with PCOS.

### Points to Remember:

- The level of 17-OHP in patients with confirmed NCAH is 3-fold to 10-fold greater than the upper normal limit.
- Patients with confirmed genetic defects of  $3\beta$ -HSD have been shown to have 17-hydroxypregnenolone values of at least 10-fold the upper normal limit.
- To date, there has been no patient with  $3\beta$ -HSD NCAH who was first diagnosed as an adult woman with androgen excess.

*Dr. Azziz is the Helping Hand of Los Angeles Chair in Obstetrics & Gynecology, Director of the Center for Androgen Related Disorders, Chair of the Department of Ob/Gyn at Cedars-Sinai Medical Center, and Professor and Vice-Chair in the Department of Ob/Gyn and Professor in the Department of Medicine at The David Geffen School of Medicine at UCLA.*

## Controversy Update:

# Say Goodbye to the “I” word

In October 2005, the Lawson Wilkins Pediatric Endocrine Society (LWPES) and the European Society for Paediatric Endocrinology (ESPE) met to review the management of intersex disorders. The meeting was composed of several different working groups made up of 50 international experts in the field. These groups prepared prior written responses to a defined set of questions from an evidence based review of published reports. The result of this meeting is the “Consensus Statement on Management of Intersex Disorders.”

As CAH is the most common cause of genital ambiguity, CARES protested the exclusion of a CAH-specific advocate at this meeting (“CARES Letter to Consensus Meeting Receives International Support” Volume 5, Issue 1 Winter 2006). Within the protest letter, CARES also made recommendations about addressing issues related to: terminology, development of centers of excellence, and surgical management.

The “Consensus statement on management of intersex disorders” was published in *Archives of Disease in Childhood* in April 2006. Of particular interest to the CAH community are the recommendations made about nomenclature, multidisciplinary care and surgical management.

### Nomenclature

The working group agreed with CARES and other patient advocacy groups that the word “intersex” and other arcane classifications are “...perceived as potentially pejorative by patients, and can be confusing to practitioners and parents alike.” Instead, the LWPES/ESPE group proposes the term “disorders of sex development” (DSD), defined by congenital conditions in which development of chromosomal, gonadal, or anatomical sex is atypical.

### Multidisciplinary Care

The consensus statement also stresses the need for a multidisciplinary approach when caring for a child with a

DSD. According to the consensus statement, the team should include “paediatric subspecialists in endocrinology, surgery or urology or both, psychology/psychiatry, gynaecology, genetics, neonatology, and, if available, social work, nursing, and medical ethics.” In addition to recognizing that this team has a responsibility to educate other health care staff in the appropriate initial management of affected newborn infants and their families, the consensus statement also recognizes the important role support groups have in the delivery of care to DSD patients and their families.

### Surgical Management

In alignment with the consensus statement on Congenital adrenal hyperplasia, this consensus statement asserts that only surgeons with expertise in the care of children and specific training in the surgery of DSD should undertake these surgeries. According to members of LWPES/ESPE, parents

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# Update on Newborn Screening

*Louisiana Gets On Board (Listen Up Arkansas, Kansas and West Virginia!)*

Gretchen Alger Lin

Over the past several months, huge strides forward have been made in the area of newborn screening; though, of course, there still is a great deal of work to be done.

## Victories

In the United States, only three states now remain that do not test for CAH. As announced in May 30, 2006, letter from the **Louisiana** department of health

(<http://www.dhh.louisiana.gov/offices/miscdocs/docs-263/EXPANDED.SCR.LETTER.5.30.06.pdf>), the state expanded testing to 27 conditions, including CAH, effective August 1, 2006. Special thanks go out to members across the state that assisted CARES in both our advocacy and media-based efforts there.

## Advocacy Continues

Advocacy work continues in **Arkansas, Kansas and West Virginia**.

Thanks to the efforts of CARES member Gail Blucker in Arkansas, CARES is actively involved in efforts to expand newborn screening. All branches of government have agreed on the need to expand and to include CAH

in all of those plans. CARES has placed the leaders of this effort in contact with several experts in the field of CAH newborn testing to make sure Arkansas creates an efficient and effective testing program.

## Stumbling Block

The stumbling block, it appears, is funding for follow-up. When an infant comes back with a positive result, the state needs to have people and systems in place to inform the physician of the positive test result, conduct confirmatory testing and make sure the family gets the support and services they need. CARES is collaborating with the Arkansas Department of Health, March of Dimes, Easter Seals and other stakeholders in the state to encourage the crafting of legislation to mandate universal, comprehensive screening that is properly funded. Please contact legislators across the state [www.arkleg.state.ar.us](http://www.arkleg.state.ar.us) to ensure their support of expanded newborn screening.

## Outside the US

Outside of the United States, momentum is building in the area of

newborn screening. In early 2006, expanded newborn screening, including testing for CAH, began in Russia. In May, Canada celebrated the opening of an expanded newborn screening center at Children's Hospital of Eastern Ontario (CHEO). With a commitment of \$7 million per year, the government will implement comprehensive testing for 27 disorders, including CAH, by the end of this year. As of July 1st, financial coverage of expanded infant testing including screening for CAH began in Taiwan. At the Sheba Medical Center in Israel, universal, comprehensive testing will soon begin.



*If you live in one of the 3 states that does not test for CAH and would like to help advocate for comprehensive Newborn Screening, please feel free to email Gretchen, [gretchen@caresfoundation.org](mailto:gretchen@caresfoundation.org).*

## Fund Research While You Shop!!!

Shop Online at [iGive.com](http://iGive.com) and Help Us Find a Cure

CARES Foundation, Inc. is a registered member of iGive. At the mall at [www.igive.com](http://www.igive.com) you can shop over 600 stores and help CARES at the same time. There is no cost or obligation to join, and a percentage of your purchase is donated to CARES. To Designate CARES as your charity of choice, search the keywords: CARES, CAH or congenital adrenal hyperplasia. Since 2005, CARES has awarded over \$150,000 for research. Please help us keep it up!

**Shop, Shop, Shop!**

## Use GoodSearch and Raise Money for CARES

***Did you know that you can raise money for CARES every time you search the Internet?***

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**Thank you!**

## In the News. . .

### Genetic Nondiscrimination Campaign Gains Momentum

The campaign for protection from genetic discrimination continues. Please add your voice in support of the Genetic Information Nondiscrimination Act of 2005 (H.R. 1227)! To learn more about this legislation and how you can help build support for it, visit:

<http://www.geneticfairness.org/act.html>.

### Stem Cell Bills Blocked

#### *HR 810: The Stem Cell Research Enhancement Act of 2005*

July 18, 2006 marked passage by the Senate of The Stem Cell Research Enhancement Act of 2005 and also spurred the President's first veto.

HR 810 would have eliminated the restrictions the President put into effect five-years-ago, which limited the use of federal money to embryonic stem cell lines created before August 9, 2001. Supporters say the current bill would have increased the number of stem cell lines available for scientific embryonic stem cell research.

However, scientists have said the bill would have continued to prevent the use of federal money to create new cell lines if it involved the destruction of embryos as well as preventing the use

of federal money to create stem cells for therapeutic cloning.

Supporters of the bill have said it is essential for the productive continuation of embryonic stem cell research that new cell lines be open for use. Because these older lines were grown using animal cells or serum, they hold the possibility of being unsuitable for use as medical therapy. In addition, some of these cells accumulate genetic abnormalities over time, making them hard to use even for simple research.

#### *Santorum bill (S. 2754):*

#### *A bill to derive human pluripotent stem cell lines using techniques that do not knowingly harm embryos*

Overlooked in the hype about the President's veto of HR 810 was S.2754 (the Santorum Bill), intended to direct NIH funding to research studying how to derive human pluripotent stem cell lines using techniques that do not knowingly harm embryos.


On July 18, the Senate passed this bill unanimously and turned the bill over to the House for its first consideration, where it failed to pass. There is currently a motion to suspend the rules and pass the bill failed by Yeas and Nays (2/3 required). The bill is cosponsored by Senators Richard

Burr (NC), James M. Inhofe (OK), Arlen Specter (PA), Susan M. Collins (ME) and Jeff Sessions (AL).

### Chem Div, Yale Receive Phase II NIH Grant

#### *Research to Focus on Development of Novel Drugs for the Treatment Of Adrenal Gland Disorders*

SAN DIEGO and NEW HAVEN, Conn., Aug. 30 /PRNewswire/ —

ChemDiv, Inc. and Yale University have announced that they have been awarded a joint Phase II NIH grant to further develop small molecule inhibitors of the G-protein coupled receptor, melanocortin-2 receptor (MC2R). ChemDiv will be responsible for medicinal chemistry optimization of the lead molecules using a panel of in vitro biochemical and cell-based assays, while the Yale scientists will perform the in depth follow-up studies on the mode of action of the discovered hits, with the ultimate goal being the development of effective drugs to treat congenital virilizing adrenal hyperplasia (CAH) and Cushing's diseases in children. 

routine. It does take effort and discipline, which would make it unsuitable for people who are noncompliant with taking medication. Chris prefers wearing the pump to taking medication several times a day. (I suspect that it is also because he now feels so much better!) He also develops lumps under the sites, which I have been assured by a dermatologist will cause no problems; I believe this happens with some diabetics too.

As his mum, it is also reassuring to know that he is getting a programmed amount of hydrocortisone every day, as we don't have to worry about missed doses! The pump also has a bolus button so that Chris can self-administer extra hydrocortisone if he starts to feel unwell. The pump allows you to switch to double dose or triple dose so that, even in sickness, you know that the medication is going into the body. Before the pump, Chris would have to

be admitted to hospital if he was vomiting as the tablets would not have been absorbed.

When we need to fine tune the doses it can be done precisely, with no worries about trying to cut quarter tablets. Another big plus factor with the using the pump, is that Chris has been able to program it with different rates for different situations. For example, during his exam period, with Prof Hindmarsh's help, we worked out a special rate to enable Chris to have more hydrocortisone during the most stressful periods of the day.

The pump may not be suitable for all children with CAH but I am so pleased and grateful for how it has helped Chris. I am sure it will have a secure place as a new treatment for CAH for those who need it! I am also indebted to Professor Hindmarsh for his time interest and hard work he has put into developing this method and for

helping Chris and extend a special "Thank you" to Sinead Bryan, Chris's endocrine nurse, for her unfaltering support, encouragement and help. ♥



Chris celebrates his 18th birthday. He has been using a pump to deliver a continuous infusion of hydrocortisone for management of his CAH.

## CAH Chat Groups

**Classical Women's Group:** A place for women with classical CAH to talk about the issues that affect them. To join, send an email to [http://health.groups.yahoo.com/group/classicalwomen/](mailto:health.groups.yahoo.com/group/classicalwomen/).

**CAHSisters2:** A place for adult women with late-onset CAH. To learn more about this group, go to <http://groups.yahoo.com/group/CAHSISTERS2>.

**CARES Teen Chat Group:** A place for teens with CAH to talk about feelings, questions, and life experiences with CAH. To join, go to: <http://health.groups.yahoo.com/group/caresteenchat1> and click on "Join this Group."

**CARES Spanish Group:** A Yahoo Group for the Spanish-speaking CAH community. To learn more and join, go to <http://mx.groups.yahoo.com/group/hiperplasia/>

**Greek CAH Groups:** Places for Greek speaking families and individuals affected by CAH. To learn more and join, visit <http://groups.yahoo.com/group/cahgreece> and <http://groups.msn.com/cahgreece>.





# Clinical Trials



The studies advertised on these pages have appeared in previous editions of the CARES newsletter. We update the clinical trials section of the newsletter and website as studies become available. For an up-to-date list, visit [http://www.caresfoundation.org/clinical\\_trials.html](http://www.caresfoundation.org/clinical_trials.html).

## CAH Study at the University of North Carolina Chapel Hill

**Karen Jane Loechner, M.D./Ph.D.**

We are recruiting children with CAH who are 4-12 years old (bone age <14 years), are still growing, and have not yet started puberty. Children will be enrolled in the study at the General Clinical Research Center at the University of North Carolina, Chapel Hill.

Although cortisol replacement (hydrocortisone, prednisone, or dexamethasone, for example) and Florinef have virtually eliminated mortality, there are at least two reasons for continuing to try to perfect our treatment regimens: (1) the linear growth of children is suboptimal and the end result is an adult who is too short, and (2) over-treatment with glucocorticoids in children may increase risk of osteoporosis in later years.

Based on studies of the regulation of release of ACTH (that, in turn, drives the production of hormones from the adrenal cortex), we have found that we can decrease the amount of ACTH using calcium channel blockers

(medications typically used to treat high blood pressure, such as amlodipine). Amlodipine has been shown to be safe even in infants treated for a variety of medical reasons.

Our hypothesis is that addition of amlodipine will allow us to decrease the amount of glucocorticoid medication that your child is currently taking to control his/her CAH. Such a decrease should translate into better growth and bone strength. This new medication would be added to your child's current CAH treatment program and evaluated in a double-blind/placebo-controlled crossover study.

For more information, please contact *Dr.* Karen Loechner at (919) 216-5946 (pager) or (919) 966-4435 ext. 224 (voice mail); fax (919) 966-2423; Roxanne Schock, CDE, Clinical Coordinator at (919) 966-0428 (voice mail). All visits, including laboratory testing, research medication, and parking will be paid for by this protocol. Limited "off-site" testing at your local care provider may be available subject to IRB approval. Travel funds available (please inquire for details).

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## CAH Steroid Study at Riley Hospital for Children Indianapolis, IN

Everyone who has CAH is treated with a type of medication known as a "glucocorticoid". There are several different glucocorticoids available, including hydrocortisone (Cortef), prednisone, and dexamethasone. Traditionally, many physicians treat children with CAH with hydrocortisone, which is a short-acting glucocorticoid that is usually given three times a day. Longer-acting glucocorticoids, such as prednisone or dexamethasone, are not as commonly used because of potential concerns related to growth. Little is known about the effects on growth, the pituitary gland, or CAH control of these different types of glucocorticoids. Improved understanding of the effects of hydrocortisone, prednisone, and dexamethasone in children with CAH would lead to better treatment options, including the potential of more simplified therapy with only once or twice daily dosing.

Our goal is to compare different glucocorticoids in children with CAH in order to optimize growth and control of CAH so as to avoid the consequences of over-treatment and under-treatment.

We are recruiting children who have classic CAH, are not yet in puberty, and are between the ages of 5 and 12 years old. Children will be enrolled at the General Clinical Research Center (GCRC) at Indiana University School of Medicine affiliated with Riley Hospital for Children in Indianapolis, IN. The study lasts for 18 weeks, but requires only 4 visits to Indianapolis. Three of these visits will consist of an overnight stay. Each participant will randomly be on a 6-week course of each glucocorticoid (hydrocortisone, prednisone, or dexamethasone).

All overnight stays, including labs, medications, and participation compensation, will be paid for by the study. If one is traveling a long distance, arrangements can be made to spend the night at the GCRC the night prior to an overnight stay at no cost.

If you are interested in learning more about our study, please contact either Dr. Erica Eugster at (317) 274-3889, or Dr. Todd Nebesio at (317) 274-3889 or by e-mail ([tdnebesio@iupui.edu](mailto:tdnebesio@iupui.edu)).

# CAH Volunteers Needed for Natural History Study

Principal Investigator: Dr. Deborah Merke, M.D  
 Pediatric Endocrinologist  
 Chief of Pediatric Services  
 National Institutes of Health Clinical Center  
 Bethesda, Maryland

We are currently recruiting patients of all ages with CAH (both classic and nonclassic) for a natural history study at the National Institutes of Health (NIH) in Bethesda, Maryland. By studying the natural history of CAH in a large population of patients, we hope to define new aspects of the disease. This will allow us to develop new management and treatment approaches. Participants will be seen as outpatients at the NIH Clinical Center in Bethesda, Maryland and may be seen once, twice, or for ongoing care. Patients seen for a limited number of visits will have a full report sent to their private physician.

All testing is free of charge and will include:

- **Genotyping (patients with 21-Hydroxylase deficiency only)**
- **Hormonal evaluation including evaluation for insulin resistance**
- **Psychological and cognitive testing**
- **Bone age (growing children) and ultrasound**

For more information or to volunteer, please contact Carol VanRyzin at the NIH at (301) 451-0399.

*This research is funded in part by grants from CARES Foundation, Inc. If you would like more information, or to speak with individuals who have participated in the Natural History Study, please call (toll free) 866-227-3737.*

*continued from page 17*

## **Controversy Update**

seem to be less inclined to choose surgery for less severe clitoromegaly. The group is firm that clitoral surgery should only be considered in cases of severe virilization (Prader III and up) and should be carried out in conjunction with repair of the common urogenital sinus when appropriate. According to the consensus, "Emphasis is on functional outcome rather than a strictly cosmetic appearance." Additionally, the group recognizes that surgical reconstruction in infancy may need to be refined at the time of puberty and that vaginal dilatation should not be undertaken before puberty.

## **The Need for Better Data**

While the Consensus statement on management of intersex disorders is fairly comprehensive, the authors continually highlight the need for better

long-term outcome data than is currently available. Within this data is a tremendous need for information on quality of life which, according to the authors, encompasses falling in love, dating, attraction, ability to develop intimate relationships, sexual functioning, and the opportunity to marry and raise children, regardless of biological indicators of sex. To that end, the report urges health care staff to offer adolescent patients the opportunity to talk confidentially without their parents as well as to encourage participation in support groups so that they may discuss their concerns comfortably.

If you would like more information on any of the issues discussed here, please call (toll free) 866-227-3737.

## **Flu Season Tips**

CARES reminds all its members to get their Flu vaccinations early (either by injection or nasal mist). Children and adults with CAH require special care during illness and the vaccination is a good first line of defense.

Please remember to **check the expiration date on all Solu-Cortef Act-O-Vials**. All households should check their Solu-Cortef prescriptions and ask for new prescriptions if necessary. Back to school means time for cold and flu season Don't be caught without your Solu-Cortef!

If you would like a copy of CARES' Emergency Instructions, please call (toll free) 866-227-3737 or email [info@caresfoundation.org](mailto:info@caresfoundation.org).

# 2006 CARES CAH Conference

*"bringing the CAH community together"*

Jubilee Hall, Seton Hall University

South Orange, NJ

Sunday, November 12, 2006

8:30am-5pm

## Topics Include:

- New Advancements in CAH Treatment & Future Trends in Research
- Research Roundtable
- Behavioral & Psychological Aspects of CAH
- Monitoring & Treatment of CAH/NCAH
- Weight Management Issues with CAH
- Pregnancy & Reproduction in CAH/NCAH
- Surgical Reconstruction in CAH
- Transition to Adult Care & Adult Healthcare Issues
- Panel Discussion: Adults with CAH/NCAH...and much more!

<b>Henry Anhalt, D.O.</b> (host)	Division Director, Pediatric Endocrinology St. Barnabas Medical Center, Livingston, NJ
<b>Susan Baker, Ph.D.</b>	Psychoendocrinologist Mount Sinai Medical Center, New York, NY
<b>Linda Burkett, R.N.</b>	Pediatric Endocrine Nurse Children's Hospital of Los Angeles, CA
<b>Patti Criswell, A.C.S.W.</b>	Certified Clinical Social Worker, Author Kalamazoo, MI
<b>Meg Keil, CPNP</b>	Pediatric Endocrine Nurse Practitioner National Institutes of Health, Bethesda, MD
<b>Sheri Berenbaum, Ph.D</b>	Professor of Psychology and Pediatrics The Pennsylvania State University, PA
<b>Deborah Merke, M.D.</b>	Chief of Pediatric Services National Institutes of Health, Bethesda, MD
<b>Dix P. Poppas, M.D.</b>	Chief, Institute for Pediatric Urology Children's Hospital of New York-Presbyterian/Weill Cornell, NY, NY
<b>Scott Rivkees, M.D.</b>	Chief, Section of Developmental Endocrinology & Biology Yale University School of Medicine, CT
<b>Richard Ross, M.D.</b>	Professor of Endocrinology University of Sheffield, United Kingdom
<b>Phyllis Speiser, M.D.</b>	Director of Pediatric Endocrinology North Shore Long Island Jewish Health System, NY
<b>Carol VanRyzin, CPNP</b>	Endocrine Nurse Practitioner National Institutes of Health, Bethesda, MD
<b>Maria Vogiatzi, M.D.</b>	Chief, Pediatric Endocrinologist Children's Hospital of New York-Presbyterian/ Weill Cornell, NY, NY

## Registration Form

**RSVP by October 29th**

**Mail:** CARES Foundation, Inc., 2414 Morris Ave, Suite 110, Union, NJ 07083

**E-mail:** mariel@caresfoundation.org or erin@caresfoundation.org

**Fax:** 973-912-8990

Name: \_\_\_\_\_

Address: \_\_\_\_\_

City: \_\_\_\_\_ State: \_\_\_\_\_ Zipcode: \_\_\_\_\_

E-mail: \_\_\_\_\_ HomePhone: \_\_\_\_\_

Work Phone: \_\_\_\_\_ Cell phone: \_\_\_\_\_

How many adults and older children will be attending the conference? \_\_\_\_\_

How many young children will need childcare?(please indicate age) \_\_\_\_\_



**Cost:** There is no charge to attend this event, but your tax-deductible donation is greatly appreciated.

**Accommodations:** Marriott Courtyard West Orange \$89/night, 1-800-321-2211 to request group code CFGCFGA. This discounted rate is available on a first-come, first-serve basis. Please reserve your room by Oct. 11th to receive this rate.

**Travel:** Newark International Airport is approximately 8 miles from the Marriott Courtyard West Orange (<http://www.panynj.gov/CommutingTravel/airports/html/newarkliberty.html>). Those traveling by Amtrak should use Newark-Penn Station. The closest train station to Seton Hall is South Orange (Morristown Line/GladstoneBranch) ([http://www.njtransit.com/rg/rg\\_servlet.srv](http://www.njtransit.com/rg/rg_servlet.srv)).

**\*\*\*Financial assistance for travel to the conference is available on a case-by-case basis. Please call or email for details.**

**Directions:** Seton Hall University is located in South Orange, NJ at 400 South Orange Ave and is convenient to Rt. 78, the New Jersey Turnpike and the Garden State Parkway. For directions to the university, please visit [www.shu.edu/visiting](http://www.shu.edu/visiting). For directions to the Marriott Courtyard West Orange, please visit <http://marriott.com/property/mapandnearbyairports/default.mi?marshaCode=ewrwo>.

*This program is supported in part through grants from:*

**Centers for Disease Control and Prevention (CDC)**





## Thank You

This newsletter has been generously sponsored by  
**The Institute for Pediatric Urology**  
**Cornell University**

We are truly grateful for their long standing and continued  
commitment to the CAH community.

For information on sponsoring an upcoming issue of the CARES  
newsletter, please call (toll free) 866-227-3737.



**EVERY1CARES** bracelet sales continue to rise thanks to the dedication of our members. Bracelets are available in Adult and Youth Sizes for \$3 each (minimum order of 5, plus shipping & handling).

To order:  
call (toll free) 866-227-3737  
or order online  
[www.caresfoundation.org](http://www.caresfoundation.org)

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