

Prenatal Treatment of Congenital Adrenal Hyperplasia

Svetlana Lajic, M.D., Ph.D.

Ever since the first case was described, more than 20 years ago ⁽¹⁾, where prenatal virilization was prevented with dexamethasone (DEX) in a CAH affected female fetus, the treatment has been intensely debated. On the one hand, the treatment offers a means to prevent genital surgery in affected female infants, but in order to be fully effective it has to be initiated in the early first trimester before prenatal diagnosis with chorionic villous sampling (CVS) is possible. Thus, a majority of treated fetuses (7 out of 8) will not benefit at all from the treatment, since CAH due to 21-hydroxylase deficiency is an autosomal recessive disorder. The question is whether early prenatal exposure to glucocorticoids such as DEX may harm the developing fetus and lead to an increased risk of somatic (physical) and neuropsychological disease later in adult life.

The rationale for prenatal treatment is to treat the fetus with a glucocorticoid via the mother, in order to suppress the fetal adrenal androgen production that is increased in fetuses with severe forms of CAH (the salt-wasting and simple virilizing variants). Fetuses with mild non-classical CAH do



not benefit from the treatment and it is therefore important that each family affected by CAH is genotyped prior to planning a pregnancy, so that a correct genetic counseling can be offered. There are clear genotype-phenotype relationships in 21-hydroxylase deficiency and the degree of virilization can be predicted based on experience from previous cases. A

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2007
No-Sweat
Run for a Cure
a virtual event for a real cause

Form a team, shape the future!

Your participation is vitally important.
Every team raising \$500 would insure
\$1 million for CAH research,
education and support.
We can't do it without you.

On Your Mark, Get Set, Go!

See page 23 to make a difference.

2007 CAH Conference

Cedars-Sinai Medical Center
Los Angeles, CA



CAH Through the Life Cycle

November 10, 2007

See page 11 for more information.

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New Addition to Medical Advisory Board

CARES Foundation is pleased to welcome **Karen J. Loechner, M.D., Ph.D.** to its medical advisory board. Dr. Loechner is the Director of the University of North Carolina at Chapel Hill Pediatric Osteoporosis Clinic and is also an Assistant Professor in the Department of Pediatrics. Dr. Loechner received her medical degree from Yale University School of Medicine as well as a Ph.D. in Neuroscience from Yale University. Upon receiving her degrees, she was trained in pediatric endocrinology at Massachusetts General Hospital in Boston. Dr. Loechner is Board Certified in Pediatrics and Pediatric Endocrinology as well as a member of The Endocrine Society and The Lawson Wilkins

Pediatric Endocrinology Society.

Dr. Loechner has a longstanding interest in CAH and relationship with CARES. She is the principal investigator for an ongoing clinical trial using Calcium Channel Blockers as therapeutic targets to reduce steroid requirements in kids with CAH and is examining the mechanism of calcium channel regulation of ACTH release and pituitary function (supported by CARES). Additionally, Dr. Loechner is preparing a clinical trial to examine the relationship between CAH and osteoporosis.

CARES Foundation is excited about the addition of Dr. Loechner to its medical advisory board and looks forward to working with her.

In Memory of Shelton Uptmore

Pam Kowalski- *Shelton's Aunt*

Shelton was a special child who almost did not survive when he was born because his disease was not diagnosed at first. We prayed and hoped that he would live, and live he did! He was compassionate and fun-loving. He did not like to see anyone sad or suffering and would try to make people smile. Shelton's love for life was infectious. He was very giving, raising money for the less fortunate and working as a counselor at a muscular dystrophy camp, where he personally attended to the every need of his campers. Many people were unaware Shelton suffered from CAH, as he did not let his disease hinder his ability to live a full life. **We thank God for the gift of Shelton.**



CARES Foundation member Shelton Uptmore (SW-CAH) died in a car accident earlier this year. His family expresses their gratitude for all your thoughts of sympathy and donations in his memory.

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a message from the
President & Founder

Dear Friends,

The most savage controversies are those about matters as to which there is no good evidence either way.

- Bertrand Russell

We at CARES Foundation continually strive to bring you the most up-to-date information in CAH care and research. Often, this information is controversial—mainly because there is so much we don't know. Articles like the one on this issue's cover serve to remind us of the need for continued research and education. Just as we work to educate you, our members, we also need to continue the education of the healthcare community.

Endo 2007

I was overwhelmed by the Endocrine community's will to learn when I attended Endo 2007 (the annual meeting of the Endocrine Society) and the Androgen Excess Society meeting (both in Toronto) during the first week of June. The "Meet the Professor" sessions on CAH and NCAH were some of the most popular. Both sessions were run twice, with audience members lining the aisles and spilling into the hall for all sessions. It was wonderful to see so many healthcare practitioners receiving expert information on everything from the diagnosis and treatment of CAH and NCAH, to the appropriate use of prenatal dexamethasone therapy.

CAH Conference 2007

Continuing with the theme of education, you all should be making your plans to attend our 2007 CAH Conference on Nov. 10th at Cedars-Sinai Medical Center. Hosted by Dr. Ricardo Azziz of Cedars-Sinai and Dr. Mitch Geffner of the Children's Hospital of Los Angeles, we have a terrific program for you focusing on "CAH Through the Life Cycle."

Membership Survey 2007

The response to the Membership Survey was terrific. If you (or your spouse) haven't yet filled out the



survey, please do so ASAP. We are analyzing the data now and will have an update in the Fall 2007 newsletter. Remember, CARES Foundation is working for you. We need your input to make sure we are directing our programs and services where they are needed most.

Survey of Adult Women

In partnership with Penn State University, CARES Foundation received internal review board (IRB) approval to begin its first original research by distributing a questionnaire, entitled "*Experiences of Women with Congenital Adrenal Hyperplasia.*" We will be calling our adult members with Classic CAH to inquire about participation. This study is vitally important in making our voices heard within the healthcare community. If you'd like more information, please call our office (toll free) 866-227-3737.

No-Sweat Run for a Cure

Everything we do at CARES, including our annual conference, is completely free-of-charge. We rely on the generosity of our donors and membership to ensure that **cost is never a barrier to information**. With that in mind, I encourage your participation in our *No-Sweat* benefit. Ask friends and family to join your team and make an impact. If you'd like, you can share your own CAH story, like young Jason Lin (page 13), to motivate team members. Imagine, if every team raised \$500 we'd have \$1million for Research, Education and Support. Prizes will be awarded to team captains raising the most money as well as to those with the most participation.

Warmly,
Kelly

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Kelly and Adam Leight

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Speak Up

Become a Partner in Your Health Care

Many people go to the doctor ready to just listen and let the doctor take the lead. But the best patient-doctor relationships are partnerships. You and your doctor can work together as a team that includes nurses, physician assistants, pharmacists and other health care providers to address your medical problems and keep you healthy.

Your first step is to find a main doctor (your primary doctor or primary care doctor) that you feel comfortable talking to. Your doctor needs to understand your health concerns and problems. He or she will help you make medical decisions that suit your values and daily habits, and will keep in touch with any other specialists you may need. So spend some time finding a doctor you can trust and with whom you can talk openly.

Try drawing up a basic plan to help you make the most of your appointments, whether you're starting with a new doctor or continuing with the one you've seen for years. Make a

list in advance of the things you want to discuss. Do you have a new symptom? Are you concerned about how a treatment is affecting your daily life? If you have more than a few items to discuss, put them in order with the most important ones first.

Good communication is key to good health care. Tell your doctor if you have vision or hearing problems so he or she can accommodate you. Ask for an interpreter if the doctor doesn't speak your language.

Some doctors suggest you put all your prescription drugs, over-the-counter medicines, vitamins, and herbal remedies or supplements in a bag and bring them with you. You should at least bring a complete list of everything you take. A recent survey found that nearly two-



thirds of Americans use some form of complementary and alternative medicine—health practices outside the realm of conventional medicine, such as herbal supplements, meditation, homeopathy and acupuncture. Less than one-third of them, however, discuss these practices with their doctors. This news is a cause for concern because your doctor needs to have a full picture of everything you're doing to manage your health.

During your visit, make sure to ask questions if anything is unclear to you. Bring up any problems or concerns you might have, whether or not the doctor asks about them. Ask about different treatment options. And don't hesitate to tell the doctor if you have concerns about a particular treatment or change in your daily life.

You might also consider bringing a


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Tips for Getting the Most out of your Appointment

- ✓ Make a list in advance of the things you want to discuss at your appointment.
- ✓ If you don't understand something your doctor is saying, ask questions until you do understand.
- ✓ Don't forget that other members of your health care team, such as nurses and pharmacists, can be good sources of information. Talk to them, too.
- ✓ Take notes, or get a friend or family member to take notes for you.
- ✓ Be First! Picking the first appointment of the morning or afternoon means that the doctor is less likely to be running late—a good thing if your child gets restless with long waits.

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family member or close friend to your appointment with you. Let him or her know in advance what you want from your visit. Your companion can remind you what you planned to discuss with the doctor if you forget, or take notes for you and help you remember what the doctor said.

Take an active role in your own health care. Do everything you can to get the best care possible. 

This article first appeared in *NIH News in Health*, May 2007, and was adapted and reprinted with permission from the National Institutes of Health.

Parent Tip Giving Medicine

submitted by:
Carrie Koenig, CA

Our Daughter is 20-months-old and has Salt-wasting CAH. We found a creative way to give her Cortef and Florinef tablets—sticking them inside gummie vitamins.

Our kids love gummie vitamins. We have been giving them a brand of whole food nutrition gummies that contain 17 raw fruits and vegetables and 2 grains for a little while now.

In addition to Reese and her brother Cole being cold and flu free, Reese loves eating the gummies. It was a struggle to get her to take her medicine, and now, with this trick, it's so easy. She would eat the whole jar if she could!

Do you have a tip that could be helpful to other families affected by CAH? Submit it to Daina Bunges, daina@caresfoundation.org.

Parent Tips for a Painless trip to the Doctor

- ✓ For long appointments or to discuss behavior or discipline problems, AVOID the late afternoon. You, your child and the doctor are likely to be too drained at the end of the day.
- ✓ Prepare your child ahead of time. Discussing that “Dr. Joe will look in your ears, pat your tummy, and listen to your heart...” or reading a book like “Blue’s Checkup” by Sarah Albee and Ian Chernichaw, makes the child feel better by letting her know what’s going to happen at the appointment.
- ✓ One word: **VELCRO**. Dress your child in clothes that are easy to get on and off. Avoid laces and too many buttons.
- ✓ **Eat**. Hungry kids are cranky kids. Make sure your child has eaten before the office visit or, if bringing a snack, choose the mess-free kind so you don’t have to think about crumbs. The staff will appreciate it.
- ✓ Let Barbie go first. Doing a checkup on a child’s stuffed animal or doll shows just how easy an exam can be. Ask your doctor if she wouldn’t mind giving Buddy Bear his checkup first.
- ✓ **Visual aids**. For issues that come and go, like skin problems or acting up only while not in the doctor’s office, bring along a photograph or home video. (Remember to bring the camcorder to show it on.)

*Tips adapted from *Parenting.com* .

Drug Update

Cortef® Generic Now Available

Generic Cortef® (hydrocortisone tablets, USP) is now available. The tablets, manufactured by Steifel Labs, come in 5mg and 10mg amounts. Reports from our members indicate the tablets are smaller than the brand name and may be harder to split. If you prefer the brand name Cortef® please make sure your prescriber indicates Cortef® with no substitutions. We have no information on the efficacy of the generic so patients should be careful when switching from the name brand.

FLORINEF® Discontinued

King Pharmaceuticals® has decided to discontinue the manufacture of Florinef® Acetate (Fludrocortisone Acetate, USP) **in the United States**. Florinef® will remain available through pharmacies until current supplies run out. Florinef® is distributed through Monarch Pharmaceuticals, Inc. a subsidiary of King.

Generic Fludrocortisone is still available, manufactured by both Barr Laboratories, Inc. and Global Pharmaceuticals.



DEX continued from cover

mother that has previously given birth to a child with severe CAH can thus be offered treatment with DEX at 6-7th week of pregnancy in the next coming pregnancy. The dose given is 20 µg/kg body weight/day, based on pre-pregnancy weight and maximum 1.5 mg/day, in three divided doses. A few weeks later, around week 12, prenatal diagnosis is performed on fetal DNA obtained from a chorionic villous biopsy (CVS). In healthy fetuses and in CAH affected boys, treatment will be stopped while affected girls will be treated until term.

The positive effects of the treatment in reducing or ameliorating genital virilization in affected girls are nowadays undisputable. However, should we be concerned whether DEX may have negative side-effects in the fetus, the growing child or later in adult life? The fact that only 1 out of 8 treated children will have a direct benefit of the treatment per se has added to these concerns. Treatment safety has been reported to be acceptable, at least in the short term perspective, based on findings of normal pre- and postnatal growth (2-5) and the reported side

effects that have been observed do not appear to follow a particular pattern (2, 4, 6, 7). The treated children are not born premature, and they do not present with an increased incidence of anomalies. In approximately 25 percent of treated mothers, side effects can be observed, such as excessive weight gain during the first trimester of pregnancy, striae (stretch marks), mood swings, irritability, insomnia, edema (swelling) and general discomfort (4). We have not seen an increased incidence of gestational diabetes mellitus or hypertension in treated mothers. Thus, the current follow-up studies on humans suggest that DEX treatment does not have major teratogenic effects (does not cause birth defects). Nevertheless, long-term effects are just starting to be addressed due to the fact that the oldest children treated are now reaching early adulthood. Results from experimental animal models have also raised concerns regarding the impact of the treatment on fetal programming and the possible effects on future metabolic and

“ An ethical dilemma regarding the future use of this treatment is opened-up . . . ”

cognitive (brain) functions in adult life (8-10). Rats and monkeys exposed to prenatal corticosteroids have shown adverse effects both on somatic (physical) development, cognitive functions and behavior. However, animal experiments aimed at identifying possible risks with prenatal glucocorticoid treatment have primarily been designed to mimic the treatment used for reducing the risk of respiratory distress syndrome in preterm infants and the glucocorticoid has thus been


given late in the pregnancy. There is scarce clinical evidence concerning the effects of low-dose prenatal DEX treatment as used in CAH.

Few follow-up studies on developmental outcome of the DEX-exposed children have been performed and only one has been based on direct assessment of the treated children (11). Direct assessment of treated children, that have reached school-age, using a wide range of tests in addition to child and parent completed questionnaires will give valid data regarding cognitive functions, behaviour and prevalence of psychopathology (psychological differences from the norm). In our Swedish group of prenatally treated children (40 cases) and healthy non-treated controls (35 cases) we found that prenatal DEX treatment did not affect psychometric intelligence (IQ), short-term memory and long-term memory in treated cases. **However, we could assess a negative effect on verbal working memory (sentence comprehension) and on children's perceptions of their scholastic competence in addition to increased social anxiety. These effects were observed in short-term treated, CAH unaffected children, when compared to healthy controls.** Parents did not observe any negative effects on school performance. When long-term emotional and behavioural development of these children was assessed we could not observe any increase in psychopathology, but the parents of treated children reported that the children were more social compared to controls. We could not make any final conclusion regarding plausible negative effects in the CAH affected children, most probably due to a small study group.

In conclusion, this study indicates that prenatal DEX treatment has long-term effects on verbal working memory and on aspects of self-perception, both of which were previously not described, but no major behavioural problems or

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effects on long term memory or IQ have been found. An ethical dilemma regarding the future use of this treatment is opened-up because the ultimate clinical significance of our findings with increased sociability, social anxiety and affected verbal working memory among the healthy, CAH unaffected, short-term treated children, cannot yet be determined. It is therefore important that additional studies of larger groups are designed to make more decisive conclusions on the safety of this controversial treatment, and preferably the treatment should only be given within the frames of a clinical study in order to ascertain accurate follow-up of the mother, the fetus and the growing child. A prospective, European, multi-centre study, PREDEX, is currently on-going (12). Until then, it is important that the parents are thoroughly informed about the potential risks and uncertainties as well as the benefits of the treatment. 

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“ It is therefore important that additional studies of larger cohorts are designed to make more decisive conclusions on the safety of this controversial treatment . . . ”

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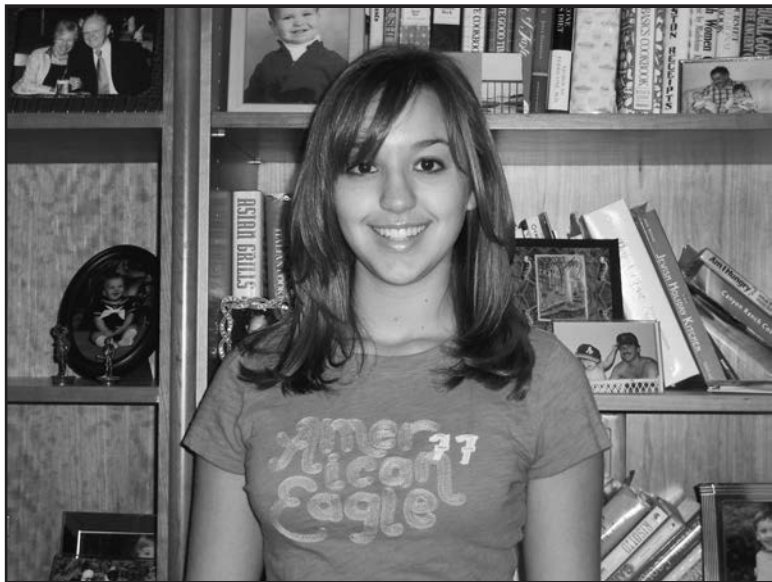
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Teen Perspective: Growing up with NCAH

Alyssa Leight

I am a girl. I am an American. I have congenital adrenal hyperplasia. For most people the words “congenital adrenal hyperplasia” are complete gibberish, but to me they’re who I am. I was five-years-old when I began showing symptoms. My mother was scared. I was confused. At six, I began going to the hospital regularly. The nurses knew my name, my school, and my friends. By the time I was seven I had seen more of my own blood leave my body and more needles than any person, no matter their age, ever should. Every night I cried, especially before I would have to go to another visit to the hospital. I prayed to G_d to make me different, to make me normal. All I wanted was to be like everyone else. At 7-and-a-half the men and women in the long white lab coats, who had become far too familiar, had an answer. I had congenital adrenal hyperplasia. At the time, those words were gibberish to my parents, but to me they were much more.



Alyssa is 14-years-old. She is entering her Sophomore year of high school.

I often think how can three simple letters change ones life forever? The letters C-A-H most definitely changed mine. The moment I was diagnosed I felt the cold hand of shame. I was told I had to wear a MedicAlert bracelet, and to me nothing screamed “LOOK AT ME. I AM DIFFERENT!” more than that.

By the time I was nine, I was in the height of the terrible pre-teen period. The two years from ages 9 to 11 were the years when my self-esteem and self-confidence evaporated as water in the sun. When people would ask me about my MedicAlert, I would lie and say it was just a bracelet. My lowest point was when my best friend in the world asked

me about my bracelet and I lied and told her it was nothing. My biggest fear was someone figuring out that I had CAH. I didn’t want to be different. I wanted to be like the rest of the girls in the world. I wanted to be normal.

Now that I am older, I am no longer afraid of having congenital adrenal hyperplasia. I no longer cry, and I no longer pray to be different. I still pray to

G_d though, but now my prayers are in thanks to G_d for blessing me with the gift of CAH. I often look back over the years of my life and wish I hadn’t been ashamed. My father told me that everyone has “something.” I didn’t understand this then, but now I realize that I was lucky that my something was mild in the scheme of things.

Now that I think back upon the time when I was a lost boat in the sea of pre-teen and teen propaganda, I wonder why I let the shame get to me. I realize now what an impact our society makes on us. They tell us we must be a certain way. They tell us that it is wrong to be different, that who we are isn’t good

enough. All I ever wanted when I was young was to be “normal.” But what is normal? Movies, television, books, and magazines teach that all women and men must fulfill this inconceivable idea of normal. Is it the movie stars with eating disorders, or those with silicone for breasts? Is it the sports stars on steroids, or the 14-year-old girls who dance with no clothes on behind Ludacris? I was a young girl and I hid who I was to be “normal.”

I am a girl. I am an American. I have congenital adrenal hyperplasia. I was once ashamed and now I am not. I let society’s view of what and who I should be cover up who I was. Now I am thankful to be different. I don’t want to be normal. Media gets the best of most of us, at one time or another, but I know now that I will never let that happen to me again. Because of the three simple letters that changed my life, and made me who I am: C-A-H. ❤️

“All I ever wanted when I was young was to be “normal.” But what is normal?”

Parent Perspective: Tough Decisions

Lisa Smith

Samantha Lynn is our daughter's name, but that wasn't always so. When she was first born her name was Ryan, because we thought we had a little boy. (That name is still so hard to say or even write.) We thought this because Samantha was born with salt-wasting congenital adrenal hyperplasia. She was very sick and we didn't even know it. We went for all kinds of tests for what we thought at the time was our son Ryan, just to find out Ryan, now Samantha, was very sick, needed medication and had this birth defect called ambiguous genitalia.

I still don't understand how I even could breathe after hearing that. We were in a room full of doctors, who announced, "You don't have a son you, have a daughter." The doctors all just looked at me while I told them they were crazy. How could this be? I left the room thinking what do I do from here? How do I feel?

Her father John took the news better than I did, saying to me, "At least she's ok." I was so upset that he said that. Why was it so easy for him? I was such a mess.

We went back into what we thought to be Ryan's hospital room. My grandmother was sitting there. I remember thinking how was I ever going to tell my family what the doctors had just told us. Then my grandmother looked up at me with this peaceful look and said, "Why do the doctors keep referring to Ryan as a girl?" So, I just went on to explain the best way I could. She made it very easy for us, but not all of our family was like that.

Right at that moment we decided that we needed to change her name, find the medication that she needed every

six hours, and have surgery as soon as we could. But then her doctor said, "What is your hurry?" She really didn't want Samantha to have surgery, but I did. The more the doctor would try to convince me not to, the more I wanted to go through with surgery.

So, that was the plan. We changed

“The doctors all just looked at me while I told them they were crazy.

How could this be?

I left the room thinking what do I do from here?

How do I feel?”

Ryan's name to Samantha. Well, her father picked out her name because I couldn't bear to even think about that at that point.

Later, at John's mother's house, we watched a show about adults whose parents chose surgery for them when they were very little. These people hated their parents for making that choice without their input, and talked

about how their parents did such a horrible thing to them. Watching this, my heart broke. All I could think was that I couldn't do this to my baby who has been through so much already. I always felt like this had to somehow be all my fault. There was no one else to blame. The thought that she would grow up to hate me stopped me right in my tracks.

A short time after the television program and fearing my daughter would hate me, I found a web site called Bodies Like Ours.com, and they reinforced everything that I had seen. I went to New York to hear people speak about their stories. With all of this, I decided to let my daughter make her own choice. I was even interviewed by the New York Times, where I explained proudly that we made our choice to let Samantha choose.

Well, that was then and this is now. Things are much different now. Sam was only a baby then. She had me there with her all the time, keeping her safe from the mean words and glares. But, no matter how much you try to protect your child, you can't be there every second. Others may say you teach your kids not to be swayed by other's words or thoughts, but, remember, we're not talking about everyday kid's stuff here.

About the time she started school, Sam became afraid that someone would see her when she changed for swimming or for other activities. And someone did.

Since then, the same person has taunted my child, along with telling all the other kids in the class, who also joined in doing the same thing. This girl

Personal Story continued on page 22

CARES Foundation, Inc. Year In Review: Key Accomplishments of 2006

Thanks to our dedicated staff, board, and donors across the country, CARES Foundation recovered from a devastating February fire to finish the year with an impressive number of accomplishments. We were up and running at full speed in a new office a mere three months after losing everything. Here are some 2006 highlights by program area:

Education & Support CAH Conference

On November 12, 2006, CARES Foundation held its largest free, full-day educational conference for affected families, individuals and healthcare professionals in South Orange, New Jersey. With over three hundred people in attendance, the agenda included presentations relating to daily living issues, treatments, and research. Simultaneous Spanish translation was offered for the first time.

Newborn Screening

Effective, August 1, 2006, Louisiana was added to the list of states that screen for CAH due, in large part, to CARES Foundation's advocacy efforts and its members across the state.

Support Groups

Twenty-three new support groups were formed across the US and in four countries abroad.

Newsletter

Two new issues of the CARES Foundation newsletter (20+ pages) were distributed to members and healthcare providers across the U.S. and 38 countries.

Emergency Care Cards

CARES Foundation developed

Medical Alert wallet cards that outline emergency treatment for electrolyte disturbances or adrenal insufficiency. The development of this product, available free from CARES, was prompted by stories from members about frustrating and dangerous delays of life-saving care. Over two thousand cards have been distributed already.

Research Landmark Study moves forward

The research design for the Experience of Women with CAH study, a partnership between CARES Foundation and Penn State University, passed the university's Internal Review Board, an important prerequisite for moving ahead. This study will increase understanding of quality of life for adult women with classical CAH by means of detailed questionnaires, to be sent during 2007.

CAH Natural History Study

Conducted by the National Institutes of Health, this study chronicles aspects of development in children and adults with CAH over several years. CARES Foundation has provided funding for a Nurse Practitioner research assistant since 2005. To the end of 2006, CARES Foundation donated \$125,000 to this important research, which enrolled 150 affected children/adults in the first full year of the protocol. Five percent of enrolled patients were found to be misdiagnosed, an interesting unintended outcome of this project.

University of North Carolina Study

CARES Foundation research

dollars supported a double-blind study of affected children to test a modified treatment protocol that could decrease glucocorticoid medication levels. Benefits of such a decrease include improved growth and bone strength.

Advocacy Pediatric Awareness

Kelly Leight's editorial on the benefits of CAH newborn screening was published in *Pediatrics*, the official journal of the American Academy of Pediatrics with a U.S. circulation of 63,000.

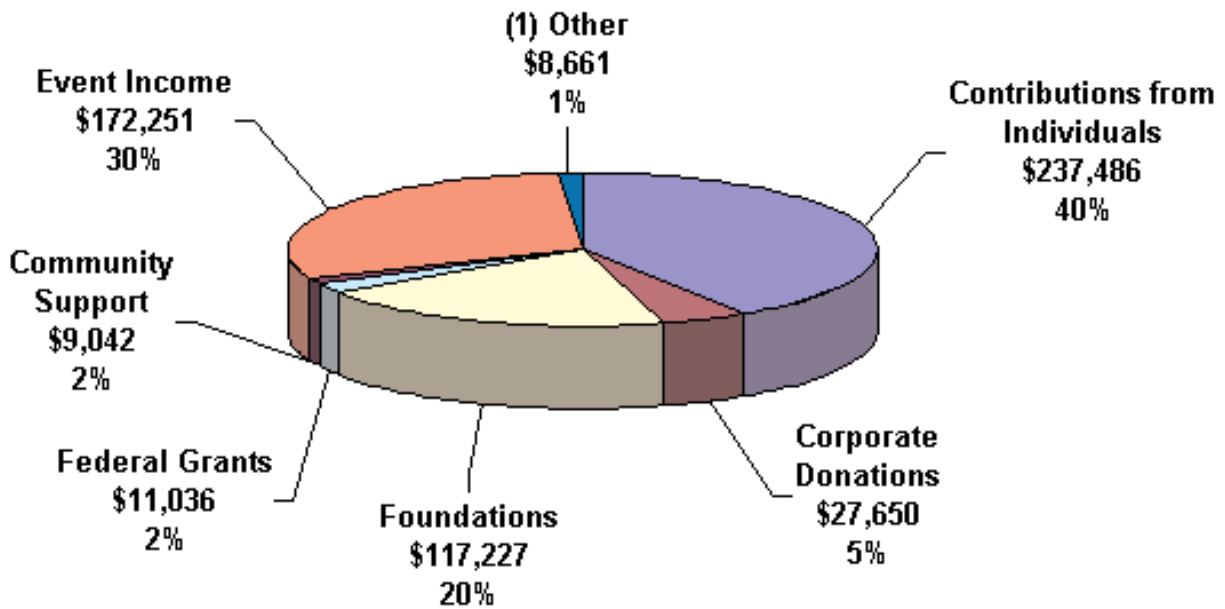
Legislation

In October 2006, landmark legislation, co-written by Kelly Leight, was introduced in the New Jersey State Assembly. The 2006 New Jersey Newborn Screening Act is one of the most important pieces of NBS legislation ever introduced as not only does it expand newborn screening by statute, but also mandates appropriate follow-up, treatment, education, and an annual review of all aspects of the screening program by an advisory committee.

Nomenclature

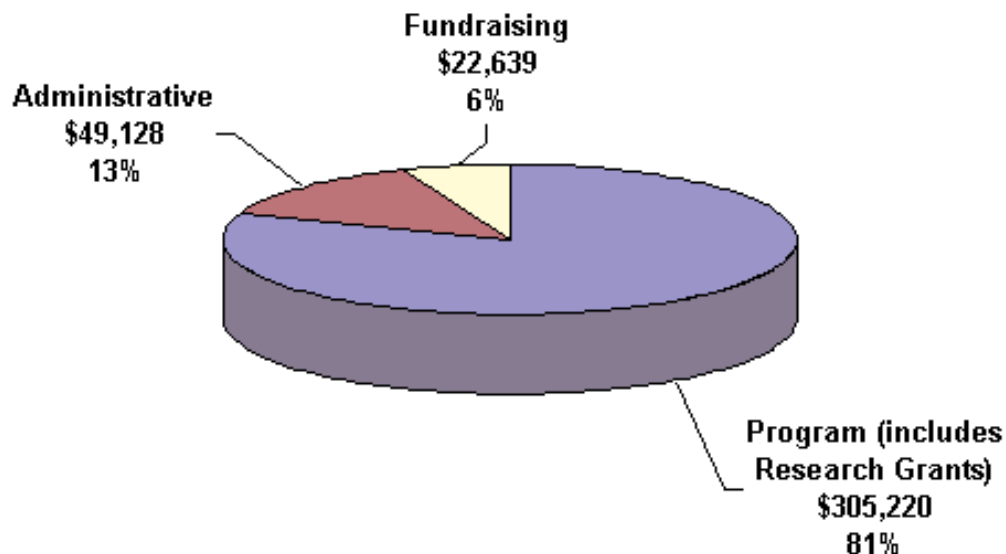
CARES Foundation's letter to the Lawson Wilkins Pediatric Endocrine Society received international support and was instrumental in forging consensus on the use of the term 'disorders of sex development' in place of the misleading classification 'intersex'. The letter also highlighted the need for more research and support services for those affected by CAH.

**CARES Foundation, Inc.
2006 Total Income: \$583,353**



(1) Includes: In-kind donations, Interest, Product Sales

**CARES Foundation, Inc.
2006 Expenses: \$376,987**



Fundraising: CARES Foundation members get creative!



Happy Birthday Caleb and Brenden! Brothers Caleb (3) and Brenden (7) Adkins (both front center, facing forward) held a combined birthday party in March and asked their friends to donate to CARES Foundation. Thank You, Caleb and Brenden!



Flying Pig Marathon: (from left to right) Adrienne Gates, Lisa Ward (mom), Lynn Gates, Jane Ward, Ty Ward (dad), Zach Ward (in dad's arms), Gigi Cali (Grandma), Ross Cali (Grandpa) and sister Olivia (front).

Lin is In It to Win It! Six-year-old Jason Lin of Las Vegas, Nevada (see next page) has taken the lead in the *No-Sweat Run for a Cure*. Jason has sent sponsorship requests and emails with his own note to friends and family across the country and as far away as Japan. Great Job, Jason!

Raffle tickets still available Louise and Joe Fleming of North Carolina are selling raffle tickets for three weekend get-away packages (which include fine dining, spa and golf) at premier hotels in the Raleigh, N.C. area. Tickets are just \$10 each, you do not need to be present to win. Please contact the CARES Foundation office at (toll free) 866 – 227 – 3737 to purchase your tickets. The drawing will be held September 1, 2007 .

Wearing Blue to raise some Green. CARES Foundation member Bernie Berrigan of Michigan encouraged his company to sponsor CARES Foundation for its “Jeans Day” fundraiser on February 9, 2007. Each month the Human Resources Department at Tenneco sponsors a “Wear Your Jeans to Work Friday” and the proceeds from the \$5 donation needed to participate go to a pre-determined charity. Bernie used the opportunity to raise awareness of CAH among his co-workers and support CARES Foundation. Thank You, Bernie!

“When Pigs Fly” On May 5, 2007, Lisa Ward and her family ran in “The Flying Pig Marathon” in Cincinnati as a way to raise support for CARES Foundation. The Flying Pig Marathon is the country’s largest first-time marathon and welcomes runners of all skill levels and abilities. Way to go Ward Family!



Congratulations Paisley and Douglas!

Paisley Dillon and Douglas Blank were married on June 3, 2007 in Belize. In lieu of gifts, they asked guests to make donations to CARES Foundation. We wish Doug and Paisley all the best as they begin their new life together.

Cook for CARES! Pampered Chef consultant Bernice Sosa-Izquierdo is using her home-based business to fundraise for CARES. The fundraiser will be held from June through August. Please contact Bernice at: 646-483-8074 or bsimom@gmail.com to learn how your Pampered Chef purchases can benefit CARES Foundation. You can visit www.pamperedchef.biz/bsikitchen to view all products.

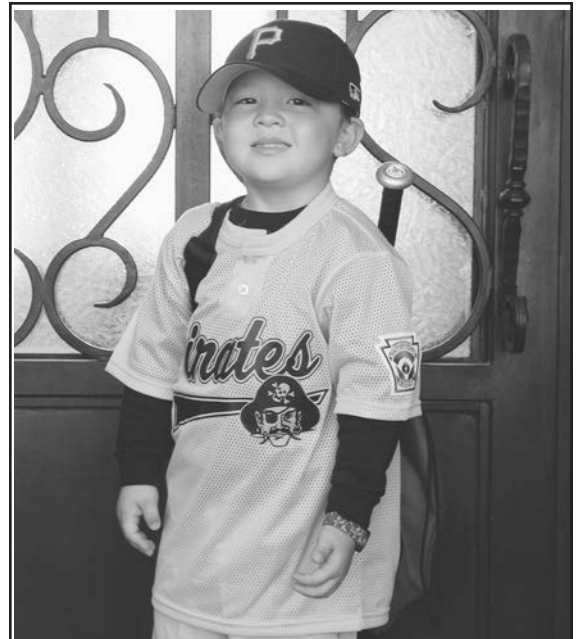
If you have pictures or events you'd like to share in the CARES Foundation newsletter, please contact Daina Bungs at (toll free) 866 - 227 - 3737 or email Daina@caresfoundation.org .

Lin Takes Lead: 6-year-old raises \$2,000 in No-Sweat Run

My name is Jason Lin. I am (almost) 6 years old. I have Congenital Adrenal Hyperplasia (CAH). I need medical care to stay alive.

CARES saves the lives of people like me. Please help me raise money for CARES by sponsoring me for a 5k walk I am doing as part of CARES' "No-Sweat Run for a Cure."

Thank you!
Jason



Happy Birthday Jason! Jason Lin turned 6-years-old on June 8th. Jason's team "Team Racin' Jason" is in the lead for the **"No-Sweat Run for a Cure."** In addition to collecting over \$2,000 in sponsorship (so far), Jason is holding his own 5K walk on July 8th. **Left:** Jason solicited donors by including his personal note with the sponsorship form.

Save the Date!

2007 CARES CAH Conference

Saturday, Nov. 10, 2007

8am - 5pm

Cedars-Sinai Medical Center, Los Angeles, CA



CAH Through the Life Cycle

California Department of Health



ChildrensHospitalLosAngeles
International Leader in Pediatrics



Life is our life's work.

- The Basics
 - Treatment & Monitoring: Child
 - Treatment & Monitoring: Adult
 - Transition to Adult Care
 - Growth
 - Just the Girls Session
 - Prenatal Diagnosis & Treatment
 - Fertility & Reproduction
 - Menopause
 - Men's Session
 - Research Roundtable
 - Solu-Cortef Injection Training
- ...and much more!

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MORE SUPPORT GROUPS NEEDED!

We need volunteers to help run support groups in the following states:

- Delaware**
- Hawaii**
- Massachusetts**
- Montana**
- New Hampshire**
- North Dakota**
- Rhode Island**
- South Dakota**
- Washington**
- Wyoming**

Please contact Daina at
daina@caresfoundation.org or (toll free)
866 - 227 - 3737 for more information.

WOMEN HELPING WOMEN

Would you be willing to help women with CAH by coordinating a support group?

Share information and empower adult women as they conquer CAH.

Please contact Daina for more information

at

866-227-3737 or

daina@caresfoundation.org.

Attention Support Group Leaders!

Send us your photos, updates and event announcements. We will publish them in the next newsletter as well as on the CARES Foundation website.

Please contact Daina at 866- 227- 3737 or
daina@caresfoundation.org.

Coming Soon: Support Group Manual

Nerve Sparing Reduction Clitoroplasty: Analysis of Clitoral Sensitivity and Viability

Jennifer Yang, M.D. and Dix P. Poppas, M.D.

Institute for Pediatric Urology
New York Presbyterian Hospital

Introduction

Enlargement of the clitoris is often a prominent manifestation of virilizing congenital adrenal hyperplasia (CAH) and other disorders of sex development (DSD).

Management of the physical and potential psychological affects faced by these children and their families requires a compassionate and multidisciplinary approach. The surgical management of patients with ambiguous genitalia has been the focus of debate for many years and remains in large part unresolved.

Surgical correction of the enlarged clitoris has undergone many changes in the past three decades, from clitorectomy that was typical until the 1960s, to recession clitoroplasty, to today's most widely accepted and used reduction clitoroplasty that preserves the glans, nerves and vascular bundle (blood vein) while excising part of the erectile tissue. Despite these advances, however, the published literature describing surgical outcomes and functional results reflects data collected on a wide variety of outdated, currently unused surgical techniques by various surgeons at numerous institutions. Currently, there is no published long-term follow-up data on modern techniques used at a single center.

In this study, we report our results using a nerve-sparing technique for reduction clitoroplasty based on the current understanding of female clitoral anatomy. Unlike other studies in the literature, all of the patients in this study underwent a nerve-sparing reduction clitoroplasty using **modern techniques performed by a single surgeon**. While

this is the largest group studied with follow-up clitoral viability and sensitivity ever reported, one limitation of this study was the lack of a normal population for comparison. Although the results presented here suggest relative normal clitoral sensitivity in these patients, continued follow up, as well as use of validated instruments to determine sexual function once the patient becomes sexually active, are indicated. At that time, it may be feasible to obtain similar test results from an age-matched normal control group for comparison.

Results

A total of 51 patients, ages 4 months to 24 years, with clitoromegaly associated with ambiguous genitalia underwent a modified reduction clitoroplasty. Thirty-two of these patients were between 0 to 2 years of age, five patients were 2 to 5 years of age and 11 patients were greater than 5 years of age. Of these patients, 46 (90%) were genetic females with congenital adrenal hyperplasia. On initial examination, four (8%), 22 (43%), 11 (21%) and six patients (12%) were rated Prader Score of II, III, IV and V respectively. The remaining patients (16%) were undocumented.

Our evaluation of post-operative results included assessment of the overall cosmetic appearance, capillary perfusion (blood flow) testing and, in selected patients, sensory testing of the clitoris. Follow up of the patients ranged from 1 week to 8 years with a mean follow up time of 24.4 months (two patients were lost to follow up). No infections, tissue death from lack of blood flow or other postoperative complications were identified. None of

the patients in the study group were identified as requiring further revision of clitoris.

Both clitoral sensitivity testing and vibratory testing results revealed heightened clitoral sensitivity relative to surrounding genitalia.

Of the group we reviewed, a total of 49 patients (96%) had blood flow testing of the clitoris. All patients had a viable clitoris with normal blood flow when compared to the blood flow received by the nail bed.

Ten of the 49 patients (20%) were older and considered candidates for clitoral sensory testing (CST) using a cotton-tip stimulator. Using a scale of 0 (no sensation) to 5 (maximum sensation), the patient was asked to report the degree of sensation at various points of the inner thigh and genitalia (labia majora, labia minora, vaginal introitus and clitoris). Inner thigh stimulation was set at level 3 for each patient and used as a baseline to compare other areas tested. Patients reported increased sensitivity at the labia minora and clitoris (degree of sensation of 3.6 +/- 0.9 on average at the labia minora and 4.8 +/- 0.4 at the clitoris). The mean time after surgery for the patients who underwent CST was 2.0 ± 0.8 years. No variations in the sensibility results were reported on follow-up visits. Two patients reported no change in sensation when tested before and after clitoroplasty.

In addition, 9 of these 10 patients also had vibratory sensory testing performed using a Bio-thesiometer designed to quantify the ability of patients to detect vibratory stimuli. A

Surgery continued next page

Surgery continued from page 16

quantitative measure was established by recording the amplitude of vibration (on a scale from 1 to 10) which correlated to the threshold. For this variable, greater amplitude of vibration (a higher number) meant a lesser degree of patient ability to sense vibration. The results indicated increased vibratory sensitivity at the clitoris and introitus compared to the labia and thigh (the average values for the introitus, clitoris, labia and thigh were 3.56, 1.61, 5.08, and 5.83 respectively.)

Conclusion:

To our knowledge this is the first report of a follow-up testing of clitoral viability and sensation after a reduction clitoroplasty. The nerve sparing reduction clitoroplasty described in this

report leaves the dorsal neurovascular bundles of the corporal bodies and the glans clitoris intact. This is a safe and reliable approach to correct the enlarged clitoris. Sexual and social function of our patient cohort is difficult to assess until all patients reach sexual maturity and adolescence. Continued, long term follow-up is on going to document long-term sexual function using this nerve sparing approach for clitoroplasty. 💙

New Staff:**Welcome Daina!**

CARES Foundation is pleased to welcome Daina Bung to the Program Development staff.

Daina joined the CARES Foundation staff in May and will be providing support to families and individuals affected with CAH, as she is also affected by the condition. She comes to CARES from the Hunterdon County YMCA, where she worked as part of the Financial Development team which raised over \$200,000 in 2006 for the financial assistance program. Daina is a graduate of Rutgers University, having received a Bachelors Degree in Economics with a minor in Political Science. In her free time, she works with a local Boy Scout Troop with the Venturing Crew, a co-ed program for youth from 14 to 20 years of age.

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/>.

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>.

Why we do what we do...

Jason: Mommy, I want to wear this bracelet to school tomorrow.

Mommy: You know you are not allowed to wear jewelry to school, but maybe you can wear it on the next free dress day. You will have to be able to explain what it is though.

Jason: It says "Every One Cares." It is from CARES.

Mommy: What is CARES?

Jason: A place.

Mommy: And what do they do at CARES?

Jason: Save lives.

Mommy: Whose lives?

Jason: People like me, Mommy. People with CAH.

NIH Update: Natural History Study of CAH

Since July 2005, CARES Foundation has generously provided an annual gift to the National Institutes of Health Clinical Center in support of clinical research in congenital adrenal hyperplasia. This annual gift was used to hire a Nurse Practitioner, Ms. Carol Van Ryzin, and allowed Dr. Deborah Merke to start a new clinical research protocol to study the natural history of CAH. Prior to July 2005, 20 percent of one pediatric nurse practitioner's time was dedicated to supporting one CAH clinical research study at the NIH Clinical Center. New clinical research protocols were not in the realm of possibility because of the lack of clinical personnel and budget cuts. In October 2005, Dr. Merke received final approval of this Natural History Study. As of June 2007, Dr. Merke and Carol have seen 174 patients with CAH (150 classic CAH and 24 nonclassic CAH) on the Natural History Protocol. Many patients have also returned for follow-up visits.

Although Dr. Merke has only accepted patients with an established diagnosis of CAH, 8 patients have been seen who were misdiagnosed (about 5 percent of patients evaluated to date). These patients include: an 8 year old male with a testicular tumor, a 10 year old female with precocious puberty, 3 young adult females with polycystic ovary syndrome, 2 unaffected carriers (1 year old male and 7 year old female) and a 27 year old female with 21-hydroxylase deficiency who was misdiagnosed with 11-hydroxylase deficiency. Seven of these 8 patients were receiving glucocorticoid medication unnecessarily. These patients represent the need for a greater understanding of CAH in our medical community. The Natural History Study is helping to educate physicians in numerous ways including: physicians-in-training in endocrinology (fellows)



As of June 2007, Dr. Merke and Nurse Practitioner Carol Van Ryzin have seen 174 patients with CAH.

are seeing the patients with CAH who come to the NIH; letters are being sent to all patients' local physicians; and findings will lead to publications and expand our general knowledge of CAH.

All patients enrolled in the Natural History Study have a comprehensive medical evaluation including physical examination, a review of prior medical records, confirmation of the diagnosis, genotyping (21-hydroxylase deficiency only), endocrine evaluation including adrenal hormones and an evaluation for insulin resistance, bone age for children, ultrasound of the ovaries and uterus in females and testes in males, neurocognitive testing and extensive one-on-one teaching regarding sick day rules and management of illnesses. Full reports are provided to the patients and their local physicians.

NIH Accomplishments to Date as a Result of the CARES Donation

Because of the large number of CAH patients being seen and also because of the contributions of Carol VanRyzin and the gift by the CARES Foundation, 2 studies were completed this year that further evaluate the adrenal medulla in patients with CAH. One manuscript, *Patients with Classic Congenital Adrenal Hyperplasia Have*

Decreased Epinephrine Reserve and Defective Glycemic Control During 90-Minute Moderate-Intensity Exercise, was recently accepted by the *Journal of Clinical Endocrinology and Metabolism*. This manuscript describes abnormal glucose regulation in patients with classic CAH during 90-minutes of exercise equivalent to brisk walking. This study shows that epinephrine (adrenaline) secretion is impaired with mild to moderate stressors, or in situations other than high-intensity physical stress. This study also reports further evidence of insulin resistance in patients with classic CAH. A second manuscript, *The Degree of Epinephrine Deficiency Observed in Patients with Classic and Nonclassic CAH in Response to High-Intensity Exercise is Associated with the Severity of Disease*, is in preparation. The results of the latter study was presented at the Endocrine Society's 89th Annual Meeting in June 2007. It is the first study ever evaluating the function of the adrenal medulla in patients with nonclassic CAH and is the first description of abnormal epinephrine (adrenaline) secretion in patients with nonclassic CAH.

Another accomplishment is the development of an extensive database for the Natural History Study.

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One advantage of having a center of excellence for a rare disease is that clinical observations across a large number of patients can lead to the discovery of new aspects of the disease expression. In the Natural History Study, Dr. Merke and her colleagues at the NIH are exploring new clinical aspects of CAH in relation to specific genetic mutations and possible risk for cardiovascular disease. Preliminary results are promising and they hope to be able to report new findings in the coming year.

Goals of the NIH Natural History Study

The ultimate goal of the NIH Natural History Study is to achieve a comprehensive description of CAH in the largest ever group of patients leading to novel findings and a greater understanding of CAH, and to the development of new treatment approaches.

In the next year, Dr. Merke will be continuing to recruit patients for the Natural History Study and will also be seeing those enrolled for follow-up

visits; her group will be analyzing data and writing a manuscript to report the Natural History Study experience to date; and they will be completing a novel phenotype/genotype study. ❤️

Drug Update

Phoqus announces positive results of Chronocort™ pharmacokinetic study

Chronocort™ pivotal pre-registration studies on track to start in H2 2007.

West Malling, UK, 31 May 2007: Phoqus Group plc, the oral drug delivery and development company, announces that the clinical development programme for Chronocort™, its lead in-house development project for certain cortisol deficient endocrine disorders, continues to progress to plan and that the Company remains on track to commence pivotal pre-registration studies later this year.

Phoqus recently completed successfully a detailed pharmacokinetic (“PK”) study (#002) in healthy volunteers that has further validated the delayed-and-sustained release profile of Chronocort™ for all the dose strengths that are in development. Study #002 demonstrated that Chronocort™ is safe and well tolerated in healthy volunteers and produced the necessary profile of cortisol levels in the blood required for Chronocort™ to be capable of replicating the normal daily cycle of cortisol secretion including the important sustained rise seen over-night and in the early morning. Study #003 has now started and will assess PK dose

proportionality and food effect in healthy volunteers. Planning is underway for the full clinical development programme which includes pivotal pre-registration studies in both congenital adrenal hyperplasia (“CAH”) patients and patients suffering from adrenal insufficiency (“AI”) that are scheduled to commence in the latter half of 2007 in both the EU and the US, subject to regulatory approval.

Phoqus appointed a medical advisory board earlier this year comprising leading endocrinologists from Europe and the USA to assist the Company in the design of the clinical development programmes able to demonstrate patient benefits and clinical superiority of Chronocort™ over existing therapies for both CAH and AI. This medical advisory board has reached consensus on the required clinical programmes and Phoqus is now liaising with the EMEA and FDA to ensure that the clinical protocols for the subsequent full clinical development programme including the pivotal pre-registration studies for both CAH and

AI will satisfy regulatory requirements.

CAH is a serious genetic disorder leading to the deficiency of an enzyme responsible for cortisol production. AI refers to the failure of the adrenal glands to produce sufficient steroid hormones. Both conditions require patients to take life-long corticosteroid hormone replacement therapy. However, current therapy does not provide steroid in a natural physiological manner and as a result often only poorly controls disease symptoms and can lead to the unwanted side effects of steroid therapy. Chronocort™ is designed to provide a new form of corticosteroid hormone replacement therapy by releasing

CHRONOCORT continued on page 21



Newborn Screening

Way to Go West Virginia!

Gretchen Alger Lin

On April 2, 2007, West Virginia Governor Joe Manchin III placed his signature on HB2583 completing the legislative process necessary to make screening of infants for 29 life-threatening, treatable diseases (including Congenital Adrenal Hyperplasia) mandatory in West Virginia. This expansion of the state's newborn screening program from testing for only six diseases will save the lives of about 25 babies every year in West Virginia.

CARES Foundation would like to express sincere gratitude to those who made passage of this most important legislation possible: CARES members as well as the leadership and members of other national rare disease organizations that joined in this effort.

CARES member Gretchen Murphy took the lead on this and followed HB2583 as it made its way through both houses of the legislature lobbying hard for expanded newborn screening at every turn. CARES members across the state helped with calling Senators when the bill was pulled off the legislative calendar in the days before the close of the legislative session. Classic CAH-affected members Timothy Wyatt Nichols and Max Murphy were vital to this effort, giving real faces to the cause.

The Organic Acidemia Association also brought the voices of numerous affected individuals and families out in support of the bill. Leslie Pierce and her daughter Cadence who is affected by Propionic Acidemia, as well as Josh Mayfield, who has Glutaric Acidemia, Type 1, shared their personal stories with legislators encouraging their

Every baby deserves a healthy start. Way to go West Virginia!

Kansas

Less than a month after West Virginia's victory, Kansas' Governor Kathleen Sebelius signed SB11 – 2007 Health Care Reform Act – into law expanding her state's newborn screening program too. The effort there was led by CARES Foundation member Tonia Kroll who, along with her husband Michael, traveled to Topeka to testify in favor of expanded screening. Their moving story of the first weeks of their daughter's life along with that of other parents who had suffered due to the state's failure to screen weighed heavily in the deliberations that followed. THANK YOU Tonia & Family!

And that brings us to **Arkansas: the only state left** in United States that does not screen for

CAH. CARES Foundation member Gail Blucker along with the March of Dimes, Easter Seals, Save Babies and other national organizations are working hard to make expansion a reality, but we need your help. The time for action is now! If you would like to join the fight for Arkansas, please contact Gretchen Alger Lin at gretchen@caresfoundation.org.

Also, **congratulations to Ontario, Canada**, where they officially began



April 2, 2007: West Virginia, CARES Foundation members Gretchen (3rd from right) and Glen Murphy led the fight for expanded NBS in West Virginia. **Center:** Governor Manchin **First row, left:** Max Murphy holding the signed bill.

support of the bill, and they were present at the governor's signing.

Joined by FOD Family Support Group, March of Dimes, National Urea Cycle Disorders Foundation and Save Babies it was through a united effort that newborn babies across the state will be saved from death, mental retardation and severe disability by early intervention and timely treatment of devastating genetic diseases.

Newborn Screening continued on next page

CHRONOCORT *continued from page 19*

hydrocortisone in a manner that will enable doctors to achieve a daily cycle (circadian rhythm) of cortisol levels in patients that closely matches that of the normal population. This in turn should improve disease symptom control and may also increase the accuracy of the disease treatment and monitoring regimen, potentially reducing the incidence of over or under exposure to steroids.

Chronocort™ is the first circadian endocrine treatment for CAH and AI. Chronocort™ uses Phoqus' proprietary Qtrol™ modified release technology to provide a delayed-and-sustained release profile of hydrocortisone to mimic the natural over-night and early morning hormone levels found in healthy individuals that are considered important in controlling both actual disease symptoms and also reducing unwanted side effects resulting from excess steroid treatment.

Phoqus' CEO, Dr. Richard Mason, commented:

“The results of the #002


pharmacokinetic study give us confidence that Chronocort will achieve the desired product profile when given to patients in the pivotal pre-registration trials expected to commence later this year. Chronocort's product profile is unique and represents an entirely new steroid hormone replacement therapy, designed to achieve physiological hormone levels in patients with absent or impaired endogenous cortisol production. As a result patients should benefit from both improved disease symptom control and also a reduction in side effects compared with current steroid therapy.”

About Phoqus

Phoqus is an oral drug delivery and development company. It creates new therapeutic products for both in-house and collaborative development programmes using its proprietary electrostatic powder coating technology, LeQtracoat®. This technology provides Phoqus with an extensive range of innovative drug delivery systems that are applied to pharmaceuticals to provide benefits

such as controlling the release of a drug into the body, enhancing patient compliance and improving the performance and efficacy of an existing drug. In turn, these benefits enable the development of new products that better meet the unmet medical needs of patients. They can also allow pharmaceutical companies to extend the life cycles of their products, strengthen their patent protection and thereby enhance the value of their marketed products and development pipelines. Phoqus is the only company using electrostatic powder coating technology for pharmaceutical applications and has over 120 granted patents.


Based in Kent, Phoqus was established in 1998 as a spin-out from Colorcon, a division of Berwind Pharmaceutical Services Inc. The Company was admitted to trading on AIM in November 2005 and is listed under the symbol “PQS”.

Further background on the Company can be found at www.phoqus.com. 

Newborn Screening *continued from previous page*

screening for CAH on May 14, 2007!

Newborn Screening Packets for Parents

"Packets for Parents" is a program that provides free newborn screening packets to expectant parents in the United States who are unable to pay for them. Certain restrictions apply, and quantities are limited to a specific number of packets each month. For information please visit <http://www.savebabies.org/NBS/packetsforparents.php> or contact Save Babies at 1-888-454-3383. 


Other News: NIH, Personalized Medicine & Stem Cell Research**Legislative Action**

Over the past several months, CARES Foundation has been continually involved in the fight to ensure funding of the National Institutes of Health. This is of particular importance to orphan disease-affected communities like ours as most research would never even get started without appropriate funding of NIH.

We also have been keeping an eye on Senators Obama and Burr's Genomics and Personalized Medicine Act of 2007 (S 976) which calls for improving the safety and effectiveness

of genetic tests; something of great import to all CAH Community members who undergo such testing.

Finally, if anyone is interested in letting their voice be heard by President Bush in favor of or against the Stem Cell Research Enhancement Act (S. 5), please contact CARES Foundation for some options on how to influence him.

CARES Foundation has not taken an official position on this legislation. We are awaiting the results of our membership survey. 

CAH Studies



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

Prenatal Diagnosis Research Project to Target which Women Should Take Dexamethasone During Pregnancy

We are seeking the help of couples at risk for having an affected child with congenital adrenal hyperplasia to participate in a research project in Boston. As you know, some pregnant women are offered the opportunity to take dexamethasone early in pregnancy to reduce the risk of masculinization of a female fetus affected with congenital adrenal hyperplasia (CAH). The problem is that only 1 in 8 fetuses will be female AND affected, so 7 out of 8 possible fetuses will receive unnecessary treatment. Steroids, while effective, do cause side effects in pregnant women and some children. Our research aims to target dexamethasone treatment to only female fetuses at high risk of having CAH.

The purpose of this research study is to develop a simple prenatal test that will use blood samples from a pregnant woman and her partner. The pregnant woman's blood will be used to determine if the fetus is male or female using cell-free fetal DNA testing. This can be done as early as 7 weeks following the first day of the last menstrual period. If both

parents have different mutations, the partner's blood will be used to test for the presence of his mutation in the pregnant woman's blood, which was inherited by the fetus.

Please note that this is a research study. In the first phase of the study, we cannot release results of fetal gender testing to you. Thus, the results will not affect your clinical care. However, if we get enough patients enrolled, and our study is accurate, our hope is that in the near future this will transition to early fetal gender diagnosis that can be used clinically.

The study involves 1-2 blood samples from the pregnant woman and 1 sample from her partner. There is no travel required. We will arrange to have the blood drawn in your hometown.

To find out more information about participating in the study, please contact Helene Stroh (Hstroh@tufts-nemc.org) or Diana Bianchi, M.D. (dbianchi@tufts-nemc.org) at Tufts-New England Medical Center.

Personal Story *continued from page 9*

and the others would tell Samantha that she "wasn't a girl, she was a boy." We had to go as far as to bring Samantha to her doctor to explain to her she was a girl. These kids were so cruel as to ask Samantha to go into the bathroom with them and show them her "penis."

When I look back, I should have done what I first wanted to do. There are still statements that will always stay with me, like when she was going to have her vaginoplasty done and her doctor said, "Why hurry? What does she need a vagina for?" My response was, "Because she does. You have one, don't you?"

People who are not going through this will never know what it feels like to not be what other people call "normal." As a mother, even I don't know. But

what I do know is she is my little girl and I'm going to keep her safe. Yes, safe from doctors making comments, from people like Bodies Like Ours, from those who lump CAH with "intersex" people who are much different. This isn't a case of I don't know my gender. These are baby girls who are born with a birth defect. A birth defect, just like any other birth defect, that can and should be fixed.

Please don't misunderstand me—my heart goes out to the intersex community, but my daughter, or any other girl with CAH, isn't intersex. That word means in-between, and there's nothing in-between about CAH girls. These terms are making parents more confused than they already are. Recently, Samantha has chosen to have

surgery. Since she made her decision, she asks everyday, "When is surgery? Are we going tomorrow?" She begs to get it done as fast as we can, and that is what I will do.

I also want to say thank you to everyone who never pushed me and just listened, even if they may have thought differently. CARES Foundation has been great. Kelly, what would the CAH community do without you? Also, my thanks go to www.CongenitalAdrenalHyperplasia.org. They have also been a great help. Families who are affected by CAH need to stay together and never judge each other. We need each other. Thanks to everyone who has ever given me kind word. God bless. ❤️

2007
No-Sweat
Run for a Cure
a **virtual** event for a real cause

a fundraising campaign that allows everyone to participate

Help CARES further its mission by asking friends and family to “join your team.”

To “compete” in the **2007 No-Sweat Run for a Cure**, CARES members will first receive one invitation per household (team) to fill out as *if* they were running a real race. To form larger “teams”, members are encouraged to request additional invitations to distribute to their friends, family and co-workers. Motivated by the time and money they will save by *not* training and *not* purchasing new clothing and sneakers, supporters will be asked to donate per kilometer they *don't* run. Prizes will be awarded to the members whose teams raise the most money and for the teams with the most participation.

We hope that you will participate in this *non*-event and help us move our mission forward.

Form a Team! 3 Easy Steps:

1. Request more invitations to give to family, friends and co-workers (see below).
2. Distribute the invitations with a personal note and team number.
3. Team members return the sponsorship cards & donations to CARES in the envelope provided.

Finish Line: Call CARES or check our website to see your team’s status.



We appreciate your participation.

Yes! Please send me ___# of invitations to send to my **TEAM** of family, friends, and co-workers.

Email: Marisel@caresfoundation.org

Fax: 973 - 912 - 8990 **Phone:** 866 - 227 - 3737

Mail: 2414 Morris Ave, Suite 110, Union, NJ 07083

Make sure to include YOUR:

Full Name _____

Home Address _____

City, State, Zip _____

Phone Number _____

Email Address _____



Use Your IRA Distribution to Support CARES Foundation

Are you planning to take your 2007 required IRA distribution?

- ✓ You can make gifts up to \$100,000 per taxpayer and the gift will count toward the required minimum distribution

Would you like to support CARES Foundation's programs?

If you answered "yes" to both these questions, you may wish to consider special opportunities created by recent pension legislation using your IRA funds to make gifts. For many of our friends, gifts from IRAs provide extra benefits.

To qualify:

- ✓ You must be 70 ½ or older at the time of the gift.
- ✓ Transfers must go directly from the IRA to CARES Foundation

- ✓ Gifts must be outright

Would you like to know more?

Please contact us today toll-free at 866-227-3737 or Meryl@caresfoundation.org to find out how you can benefit from this special opportunity, which may be available only in 2007. CARES Foundation, Inc. is a not-for profit, 501(c)(3) organization.



If you (or your spouse) haven't done so already, please complete the **2007 CARES Foundation Member Survey** and help us serve YOU better.

Visit www.caresfoundation.org for more information.

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Have you recently moved, changed your phone number or email? Please make sure to let us know!

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