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Pregnancy and CAH: What every woman with Classical CAH Should Know

by Ellen Seely, M.D.
Rhonda Bentley-Lewis, M.D., MBA

Women with classical Congenital Adrenal Hyperplasia (CAH) can conceive and have healthy pregnancies and deliveries with the proper prenatal care. However, every woman with CAH should be well-informed in a few key areas prior to embarking on this journey.

Can a woman with CAH become pregnant?

Yes! However, when CAH is not well controlled with medications, overproduction of male sex hormone may interfere with ovulation and prevent pregnancy from occurring. Adjustment of the steroid dose can often lead to restoration of ovulation.

When adjustment of the steroid dose alone is not successful, clomiphene citrate, a pill that can increase the likelihood of ovulation, can be used. The use of clomiphene citrate is not specific for women with CAH and is used in many women who are not ovulating regularly.

Once ovulation is occurring, sexual intercourse with a fertile partner is usually the next step in achieving a pregnancy. However, some women with CAH have discomfort during sexual intercourse, as their vagina may be shortened due to the androgen exposure in utero. In this situation, some women may choose surgery to enlarge the vagina or to have sperm injected directly into the vagina by an obstetrician.

When should a woman with CAH seek help with family planning?

Once a woman with CAH decides she wants to conceive, she should seek out the supervision of a medical endocrinologist and an obstetrician who are experienced in the management of CAH during pregnancy. The endocrinologist will help with regularization of the steroid dose to achieve optimal ovulation and then will work with the obstetrician during pregnancy on

(continued on page 4)

You're Invited!

Saturday, October 22, 2005

5th Annual
CARES Foundation



CAH Conference

Indianapolis, Indiana

**see page 19 for details*

Calling All Federal Employees!!! Donate to CARES CFC #2937

Federal employees and military personnel can donate to CARES Foundation through the Combined Federal Campaign (CFC) operated by the US Government's Office of Personnel Management (OPM). The CARES Foundation code is #2937. This year's campaign runs from 9/28/05 to 12/15/05. 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:

Dear Friends,
I hope this letter finds you all well and enjoying the first bit of fall. The tragedy in the Southeast has had a sobering effect on all our lives, reminding us to be thankful for all we have, including each other.

Many of you have contacted us regarding Katrina and we are impressed by your willingness to help. We have been in contact with our member families in the affected area, and most of those we talked to are ok. I am particularly concerned about the families we were unable to reach and ask that you keep them in your thoughts and prayers.

To help those CAH families who are not CARES members, I have been in contact with the American Association of Clinical Endocrinologists and the Pediatric Endocrinologists from LSU. We have donated new computers and are planning specific fundraising efforts for the LSU pediatric endocrinology group, whose facilities and equipment were lost.

I have also called Pfizer and King Pharmaceuticals, who assured me they are including Cortef, Solu-Cortef and Florinef in their respective donations. In light of the disaster, I urge everyone to keep an extra month's supply of medication on hand at all times as well as extra injection kits where necessary.

Again, thank you all for your generosity and warmth. Our thoughts and prayers are with everyone in the Southeast at this time of devastation.

2005 CAH Conference

The effects of Katrina will certainly be felt throughout the country in months to come, but it is important to be optimistic about the future as well.

On that note, it's conference time! We have had an overwhelming response to our invitation and several requests for travel assistance. It is very rewarding to have so many people interested in attending and we'd like to help as many as we can. Unfortunately, the requests have surpassed our capacity. I encourage all those who can to consider sponsoring a family for the conference. For more information on sponsoring a family, please see our "Wish List" on page 12.

I am very excited to hold this year's conference at Riley Hospital for Children, a center of excellence for CAH. A great program is planned and I'm confident everyone will enjoy themselves and learn a lot. We are grateful to have Eli Lilly and Company as our Key Conference sponsor and thank them for their generosity.

Combined Federal Campaign (CFC)

As you may have seen on the front page, CARES is now a registered member of the Combined Federal Campaign (CFC). This means that all federal employees can register to donate to CARES through workplace giving. The enrollment period is approaching quickly. Please speak with your employer about designating CARES as your CFC charity of choice and encourage family and friends who are federal employees to do the same.



Warmly,
Kelly

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This newsletter is published 3 times a year.

New CARES Staff

Erin Anthony, Associate

Erin Anthony, new associate and former intern, joined the staff at the end of May after graduating with Honors from Lehigh University. A double major in Journalism/Science-Writing and Biology, she wrote and edited scientific and health related articles for the school paper, worked on a variety of community service projects, and completed a College Scholar Thesis in Biology. Erin, who is affected by CAH, brings to CARES her compassion, writing ability and eagerness to serve the community. Erin can be contacted at (toll free) 866-227-3737 or erin@caresfoundation.org.

Stephanie Erb, Member Services

Stephanie Erb serves as CARES' new Member Services Assistant. Based in California, Stephanie provides an invaluable service to CARES by responding to many of our members' e-mails and phone calls. Stephanie, an accomplished actress, has experience in theatre, television, and film. Some of her film credits include *The Ring* and *Lucky Numbers*, her television credits include *Freaks and Geeks*, *Six Feet Under*, and *Will and Grace*, and her theatre credits include work with John Houseman's Acting Company and LA Shakespeare Festival. Stephanie can be e-mailed at Stephanie@caresfoundation.org.

In Our Next Issue...

We would like to make our next newsletter the "Men's Edition," focusing our articles on the issues of men with CAH. If you have a story to share or a topic to recommend, please e-mail erin@caresfoundation.org.

Central Texas Get-together A Hit !!!



A picture from the Central Texas CARES Support Group Get-together on August 13, 2005. Families came from Austin, Houston and San Antonio.

The Central Texas CARES Support Group was a big success. The group will meet again in November and is hoping to get together every 3 months thereafter. If you live near Central Texas and would like to join the CARES Support Group, please contact Meridith Taylor at (512) 349-9719 or e-mail her at meri_taylor@yahoo.com.

Keep Your Calendar Open!

RILEY CARES TOURNAMENT FOR CHILDREN

June 1, 2006*

Heartland Crossing Golf Links
Camby, Indiana

Hosted by the Department of Pediatric Urology
Indiana University School of Medicine

*Please watch for more information in our Winter 2006 Newsletter.



Think of CARES
this Holiday Season

Do your online shopping at
www.igive.com and a donation will be
made to CARES!

Register (keyword: CAH) and enter
CARES Foundation as the cause you
want to support.

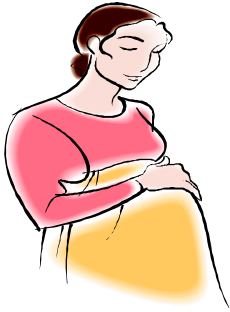
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available, \$5 each. To order, please
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*In Loving Memory
Of
Israel Chaim, SWEAK
1/03 - 8/05
son of
Yakov and Esther*

*Please keep this family in
your thoughts and prayers.*



medication adjustment. Occasionally, an Ob-Gyn trained in reproductive endocrinology and infertility may need to help if pregnancy does not occur with

regularization of menses.

Prior to pregnancy, consultation with a geneticist is also recommended to evaluate and inform the couple of the genetic risk of having a child with CAH and options for prenatal treatment as outlined in the CARES Foundation Winter 2004-05 newsletter, volume 4, issue 1.

What is the chance of a woman with CAH having a child affected with CAH?

The genetics of CAH, including how the genetics may translate into the clinical manifestations, have been detailed in the CARES Winter 2004-05 newsletter.

Briefly, genetic evaluation will provide information regarding the risk to the fetus of having CAH. Because the carrier rate of CAH can be significant, as high as 1 in 30 in some populations, it is important to genetically evaluate (genotype) the father to determine if he has CAH, is a carrier, or is unaffected. This information will then be used in conjunction with the mother's genotype information to understand the possible outcomes for the unborn child.

For example, when the mother has CAH, if the father is a carrier of CAH, there is a 50 percent chance that the fetus will be affected and 50 percent chance that the fetus will be a carrier. If the father is unaffected,

then all children will be carriers.

What are particular medical issues that have to be managed during pregnancy in a woman with CAH?

During pregnancy in women without CAH, the steroid levels in the body normally increase. Therefore, the dose of steroid a woman with CAH takes may need to be increased during pregnancy and the dose should be monitored carefully by the woman's medical endocrinologist.

Most women with CAH are kept on their same form of steroid. Prednisone and hydrocortisone do not cross the placenta well and are not thought to affect the fetus. Some women need to increase their dose of fludrocortisone (Florinef) while others increase their salt intake alone.

Once pregnancy occurs, it is important that the mother continue to take her steroid medication regularly, not only for her well-being but also for that of the fetus. Very high levels of male sex hormone in the mother could potentially cross the placenta and make a female fetus virilized. The mother's steroid dose should be adjusted so that maternal androgens run in the upper limit of normal range for pregnancy. In addition, the pregnant woman has a "back up" system in place- the placenta has a built in enzyme called placental aromatase that metabolizes the majority of the male sex hormone produced by the mother. Therefore, with adjustment of maternal steroid doses and good placental function, a female fetus,

who is not herself affected with CAH, will not have problems with virilization.

Are there any particular concerns a woman with CAH should have about labor and delivery?

Labor is a stressful time due to exertion and, at times, discomfort. As the medical endocrinologist often advises an increase in the steroid dose at the time of stress from an illness, the doctor will increase the steroid dose during labor and delivery and then return the dose to the baseline dose in the several days after delivery.

In addition, it appears the chance of having a cesarean section may be

increased in women with CAH. This may be due to limited pelvic size and shape because of exposure to higher male sex levels during early development. In addition, some experts recommend cesarean section for women who have had prior vaginal reconstructive surgery.

Can women with CAH have healthy newborns?

Yes! Women with CAH can and do have healthy pregnancies and children. There are several reports of normal infants born to women with CAH. Many endocrinologists who care for women with CAH take care of women who have had healthy pregnancies. At the Endocrine and Hypertensive Disorders of Pregnancy Program at Brigham and Women's Hospital, of the eight pregnancies we have taken care of, all have had healthy infants. In most states, neonates are screened for 21-hydroxylase deficiency automatically as part of the neonatal

" Yes! Women with CAH can and do have healthy pregnancies and children."

screen that takes place using blood obtained by a heel stick of the neonate. Screening for 21-hydroxylase deficiency is required to take place in 42 states as of August 12, 2005 (US National Newborn Screening Report).

Summary

Women with CAH can and do

have healthy pregnancies and babies. Important to optimizing the likelihood for a healthy pregnancy is 1) family planning and counseling with endocrinology, obstetrics and genetics prior to pregnancy and 2) close follow-up with both endocrinology and obstetrics during pregnancy and delivery.

Ellen Seely, M.D. is a member of CARES' Medical Advisory Board. She is the Director of Clinical Research in the Division of Endocrinology, Diabetes and Hypertension at the Brigham and Women's Hospital in Boston and Associate Professor of Medicine at Harvard Medical School.

CAH and Pregnancy: A Short Story with a Happy Ending.

By Erin Anthony

Marsha Finnegan will be the first to tell you she "has a good life." At 51, she has a beautiful daughter, Bailey, 13, and a wonderful husband, Tim. CAH has never been something that stopped Marsha from doing anything, especially having a child, so she assures me this will be a quick conversation.

1.) *Was having a family something you thought about growing up?*

No, I actually *didn't* want a family when I was younger. I remember going to my doctor as a teenager and saying that I did not want children and wanted to have my tubes tied. My doctor, of course, wouldn't agree and told me "you'll regret it later." Fortunately, I listened to her advice.

I didn't start thinking about having children until much later. Tim and I didn't start dating until I was 28 and we dated for several years before we decided we wanted to get married and have a family. By the time we started trying I was in my late 30s and I didn't get pregnant until I was 38.

2.) *Did you have any concerns or worries about getting pregnant or having a child with CAH?*

Honestly, we didn't think much of it. We went to find out our chances of having a child with CAH. We knew we might have some problems so we did see a fertility specialist, eventually changing from

"I have a **really good** life."

one that we didn't like to another who was successful pretty quickly. We tried a few different things at first, starting with taking basal temperatures every day to see if I was ovulating. Eventually, I took pills (clomiphene citrate) every day to ovulate.

3.) *What were your concerns once you found out you were pregnant?*

Because of my age we were concerned about having a child with Down Syndrome so we did the test for that and even visited with a family who had a child with Down Syndrome. Other than that we weren't too worried. We tested for CAH, too, just because we wanted to know.

4.) *Did you consider prenatal Dexamethasone treatment? Were you concerned you might have a child with CAH?*

No, we didn't think about using prenatal dex. I have CAH and I'm ok. If my daughter did, with all that entails, that's ok too. It was something we were comfortable dealing with. I think, if people want it (prenatal therapy), and it can be done, great. But, the world shouldn't stop, life shouldn't stop.

5.) *How was your pregnancy? How did you feel? Did you undergo any special treatment?*

We increased my dosage of

medicine for my pregnancy and I did experience gestational diabetes. So, I did have to stop drinking my Pepsi, watching other stuff, and doing the blood test thing every day. Other than that I was totally fine. I didn't see my ob-gyn more than other women and was able to work up until two weeks before I delivered. I did see my endocrinologist every trimester in order to adjust my medications. All in all, it was a normal, uneventful pregnancy.

6.) *What was your delivery like?*

I knew from the beginning that I would have to have a C-section because my pelvis was small.

How did you feel about that?

I was thrilled. I really didn't have the desire to go through a "regular" labor. I healed up very nicely and it wasn't a big deal.

7.) *Did you ever consider having more children?*

We tried two more times with artificial insemination, but the clock was working against us. If I was younger we would have probably tried harder but we were more than happy having one healthy child.

8.) *Any final comments?*

I have a really good life. I have a beautiful, healthy 13-year-old daughter and a great partner. I want other women to know that this is possible.

CAH Personal Story

*Alison Winters is 23-years-old, resides in the Bay Area, and was diagnosed with CAH at birth. She agreed to share the story of her family's CAH experience with CARES Foundation members.

My older sister, Leigh-Anne, was born in 1981 and diagnosed with CAH at birth. Neither of my parents had a family history of CAH and neither were aware that they were carriers of the disease, or even that CAH existed, until Leigh received her diagnosis.

My parents thoroughly researched CAH and developed a strong relationship with pediatric endocrinologist, Dr. Debra Cohen. My mother became pregnant with me shortly after giving birth to Leigh and an amniocentesis was performed to determine if I too was affected with CAH. Test results indicated that I did not have CAH, so my mother did not pursue prenatal dexamethasone treatment. At birth it was evident that I was affected by CAH, and I began treatment.

In 1986, my mother became pregnant with Tate and decided not to have an amniocentesis. Tate was also diagnosed at birth with CAH. For Leigh, Tate and me, because our parents made treating CAH a regular part of our routine, we simply did not think about it much. As kids we never felt different from other children or had difficulties coping with our CAH.

My parents never questioned whether or not we would eventually have corrective surgery, but did question the appropriate age for our surgeries to be performed. They started researching surgeons early on, gathering information in part to be ready for a potential conflict with our

"All of us have faced **unique challenges** as a result of having CAH, but CAH has in no way dictated what **we can accomplish.**"

insurance company. By the time we were prepared to have surgery, Leigh was 8 and I was 7. My parents interviewed surgeons within the Kaiser network (all of whom would be covered easily through insurance) but could not find anyone who had performed this type of surgery. As part of this process, my parents took both Leigh and me to Stanford Medical Center to meet with Dr. Linda Shortliffe. Dr. Shortliffe was very kind to us and explained in age-appropriate terms what would happen during the surgery and what we could expect afterward. We felt scared but, with the support of Dr. Shortliffe and Dr. Cohen, also believed everything would be okay.

We stayed in the hospital for four days following surgery. Dr. Shortliffe respected our wishes in that she did not allow throngs of medical students and residents to inspect us. When it came time for her to examine us, she always asked permission if there was going to be anyone else in the room, particularly if the individual was male. She was respectful of our privacy—one reason I believe that our hospital stay was not as traumatic as it otherwise might have been.

Following our hospital stay we spent two weeks recovering at home. The recovery was difficult and painful, and there were times when we cried due to discomfort. We were both extremely frustrated at being propped up all day on an inflatable "donut". Still, we had

fun playing and having visitors.

While the entire surgery experience was difficult for my sister and me, it was even more traumatic for my parents. When we first discussed the possibility of Tate having surgery, Leigh and I expressed that it should be performed before Tate was old enough to fully remember the experience, and Tate had surgery when she was 3.

Unfortunately, CAH wasn't the only adversity our family faced. Once Leigh turned eleven, she began to experience debilitating migraine headaches. We were all frightened at the extraordinary intensity of these migraines, but assumed they were related to the onset of puberty and would likely pass. Yet the headaches did not go away: not in weeks, not in months, nor in years.

Leigh frequently missed school and had to make emergency trips to the hospital. She was checked for brain tumors and eye problems among other potential ailments. All test results were inconclusive. Her 17OHP levels were within normal range and the source of the migraines remained a mystery. Leigh was put on a variety of medications to treat the migraines, but none were wholly effective, and several caused significant side effects.

Leigh's struggles caused a great deal of fear within our family. Influenced by stress and anxiety about Leigh's condition, with perhaps some fear that our CAH might cause the

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same condition for us, Tate and I began to feel upset and to act out. I took my medication only sporadically (when I would begin to feel sick or had to take a blood test) and also began to experience difficulty in my family relationships. Tate and Leigh, were more withdrawn and became depressed and anxious. Tate also developed an anxiety disorder. At this time we started family counseling to develop strategies to repair relationships, to support Leigh, and to get through this difficult time intact. Leigh, Tate and I also had individual counseling sessions that helped us at the time.

Leigh's migraines would come and go, usually averaging two per week, but there was never a period of time when she went longer than 3 months without a migraine. This was difficult for us all. At times we simply felt helpless.

Leigh's migraines worsened in high school, but by that time we had gotten far better at dealing with them as a family. Leigh and I were involved in sports, had many friends, and were good students. Tate was also doing well.

Leigh was accepted to the University of California, San Diego and began coursework as a political science major in the fall of 1999. Even though she was in and out of the emergency room that year, she was on track to graduate from college early and with honors. Leigh was active with Intervarsity Christian Fellowship—her faith was an important influence on her life—and with Intervarsity she developed an outreach project for international students and taught a Sunday school class for inner-city youth on weekends. Leigh also completed a semester abroad at the London School of Economics and traveled throughout

London and Paris in 2001.

On February 5, 2002, Leigh was admitted to the UCSD Medical Center because she had been suffering from a migraine and vomiting for 24 hours. My father drove down to be with Leigh and stayed with her in the hospital. On several occasions during this hospital stay, my father asked the attending physician and nurse if Leigh had been given the solumedrol she needed. "Yes," they answered.

But they had not given Leigh any solumedrol. Leigh died shortly after midnight on February 6, 2002 of adrenal crisis. The hospital charts indicated that even though the physician and nurse said that Leigh had been given solumedrol, the medication was not administered. Even though Leigh had a standing order for solumedrol and was wearing a medical alert bracelet, even though the physician had emergency phone

"...we are committed to embracing future challenges with the strength and joy that Leigh so often showed..."

numbers for Leigh's endocrinologists, and even though both Leigh and my father had explained the nature of CAH to the physician, and, finally, even though our father double-checked to ensure that she was given the medication, she did not receive the dose of solumedrol which, we firmly believe, would have saved her life. My parents contacted medical experts who agreed and ultimately filed a

wrongful death lawsuit. The lawsuit was settled and the UC Regents acknowledged responsibility.

We were shocked to learn how little the hospital staff knew about treating adrenal patients. Leigh's treating physician at the hospital made a statement after Leigh's death that her adrenal condition had nothing to do with her death. He believed that the fact she had gone 56 hours in status migraine without steroids was irrelevant and she had undoubtedly gone that long without medication during previous hospitalizations. He never accepted any responsibility for his negligence. Other physicians and representatives of the hospital did admit the errors and negligence.

Leigh had drive and ambition and many great plans: plans to attend law school, get married, and someday have children. Leigh eventually wanted to be involved with an overseas ministry. It is painful to acknowledge that Leigh would have likely achieved these things had the physician and nurse that treated her taken CAH and her medical needs more seriously.

Though we are a close-knit family, being through so much together regarding CAH has been one reason for this closeness. I know that at times we all questioned whether or not we could make it through the trauma of losing Leigh. It is impossible to fully absorb and recover from the loss of so close a family member, and Leigh's absence will always be painful, but in the last three years our family, both individually and collectively, has worked to reclaim a measure of the fulfillment and joy we lost when Leigh died.

I completed my bachelor's degree a year early from UC Davis

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Letters

The Surgery Decision Melbourne, Australia

Dear CARES readers,

Australia is a long way from North America and Europe and our isolation sometimes allows us to “do our own thing” in a way that might be difficult in the larger population centers. The CAH Support Group, Australia Inc., was established at RCH, Melbourne 20 years ago and I think it was the first of its kind in the world. The warm and open relationship between CAHSG members and health professionals has been of great mutual benefit. Surgery for girls with CAH is not an issue that is much debated in Australia. Parents in the U.S would be surprised at the difference in atmosphere between our two countries. Australia is close to Asia, and we are acutely aware of the difficulties faced by children with CAH and their parents in very poor countries like Vietnam (pop. 80 million) and Indonesia (pop. 220 million). The establishment of CLAN (CAH Living as Neighbors) by dynamic Sydney-based parent, Kate Hansen, has already seen some significant advances for parents in North Vietnam. Watch this space!

Kelly asked for Australian perspectives on how parents make decisions on behalf of their children, about legal attitudes to surgery, and how our surgeons approach the subject. My colleagues and I hope that you will find the responses interesting and helpful.

*-Garry Warne, M.D.
Senior Endocrinologist
Royal Children's Hospital
Melbourne*

Factors influencing parental decision making

*By Elizabeth Loughlin
Endocrine Social Worker*

Their prior social and emotional life influences how all parents respond to the birth of their new baby. When the sex of the newborn baby is unclear, some things in particular seem to color the parents' response to the birth. These include whether the parents' family of origin is understanding and the maternal grandmother is physically available, whether the parents can talk of their worry for their daughter's future to each other, and whether there is particular cultural concern with privacy or secrecy. Hospital factors

“We have talked to both sides, and I am not completely certain what is best to do, but we can tell our daughter later on, we really thought over the decision.”

beyond the control of the parents include: Was the medical information clear or messy in the first few days after the birth? Did it take several days before the parents could tell or retell others the sex of their infant? Were other unexpected birth events perceived as traumatic by the mother?

Clinical experience indicates that when parents can talk at length with the medical team and the social worker in the first few days about their shock and confusion, they are more able to move on to other uncertainties about the place and timing of genital surgery. When parents do not have this opportunity

they may close off further thinking about surgery and want to “go home” with a definite certain plan mapped out for future treatment.

A social work assessment and interview program (Loughlin, E. Poster, *Who is my baby? A social work intervention program for infants of indeterminate sex and their families*. WAIM Congress, Melbourne, January 2004) offers early counseling for the individual family situation. At the same time it indicates whether a family is vulnerable or able to cope with future uncertainties, and secondly, whether the family has or will find information from a wide range of sources and voices about the pros and cons of surgery.

One mother said about her daughter's impending surgery, “We have talked to both sides and I am not completely certain what is best to do, but we can tell our daughter later on, we really thought over the decision.”

The Surgical Plan

*By John Hutson, M.D.
Pediatric Urologist and Professor
of Pediatric Surgery*

The surgical plan for CAH patients is to resolve as quickly as possible the underlying diagnosis shortly after birth and obviously establish them on medical treatment. My plan thereafter is to see the families for discussion in the clinic about the various surgical options and the surgery required with regard to the amount of surgery and timing of surgery, etc. I put to the parents the alternative views of having surgery in early infancy or delaying this until later. I also ensure that they understand my own personal bias which is that early surgery has many significant advantages and very few disadvantages if done well.

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Following lengthy discussion, if the family is happy to proceed with surgery in infancy, I would normally arrange the surgery sometime between 6 and 10 weeks after birth. This is a good time for rapid healing and the only concern is ensuring a low risk from infection by aggressive antibiotic management and perineal hygiene and antiseptic management. With these simple steps we have achieved a high rate of anatomical success which I think would be hard to beat if we had delayed the surgery to older age groups. In addition, having gotten the surgery over with in early infancy, the children are able to grow up without being frightened of the hospital system, because they do not remember any of the unpleasantness of their management.

Legal Aspects of Informed Consent for Genital Surgery

*By Elizabeth Kennedy
Corporate Counsel
Royal Children's Hospital
Melbourne*

Informed consent is a basic requirement when any surgical or invasive procedure is to be performed on the patient. A health professional who performs an operation without the patient's express or implied consent is liable under Australian Law in trespass.

A health professional may also be liable in damages if he is negligent in failing to inform the patient of the material risks involved in the treatment and if the patient, having been so informed, would not have consented.

In the case of children, it is the parents who give consent to surgery. Note that under Australian Law special medical procedures would require authorization from the Family Court of Australia. For

example, where a procedure is considered sensitive or ethically contentious, a Family Court Order would be required. For example, when a young baby has been born with both male and female characteristics and the parents wish to determine the child's gender by surgery, a Court Order protects the surgeon from the legal liability outlined above when a health professional acts without informing the patient of all material risks.

Note that in *re Alex (2004)* the Family Court of Australia authorized hormone treatment of a 13 year old girl wishing to suppress menstruation and undergo gender reassignment surgery at a later stage.

Recently the County Court of Victoria Australia considered the case of a 37 year old man who at age 21 underwent a medical operation whereby his penis and testes were removed and he was fitted with an artificial vagina as part of a gender reassignment process. He was granted permission to bring an action in negligence out of time against the hospital and the treating medical practitioners, including the surgeon and the psychiatrist involved, in assessing his suitability for the surgery, his attitudes and behaviors.

Parents of girls with ambiguity due to Congenital Adrenal Hyperplasia need therefore to have a proper understanding of the condition, the treatment options and proposals offered by the health care professionals, so as to act in the best interests of the child and be able to give proper consent. It may be that the taking of legal as well as medical advice is appropriate.

*If you would like more information on the work being done in Australia, or have questions about genital surgery, please e-mail
info@caresfoundation.org .*

Controversy

by Erin Anthony

In recent months there has been much debate about the appropriateness of early genital surgery for genital anomalies as well as controversy over the term "intersex." Those of you who read and participate on the CAH message board are no strangers to these topics, and many of you have contacted CARES about both.

As a support organization, it is CARES mission to support its members in any way possible. It is not our role to say whether early surgery for girls born with genital anomalies is right or wrong, only to provide the best, most comprehensive information available. We rely on our medical advisors and the most respected literature, such as the LWPES Consensus Statement on 21-OH Deficiency, for the information we distribute. As it is, both of these sources currently recommend surgery between 2-6 months of age because it is technically easier, cautioning that extreme conservatism be exercised when considering clitoral reduction because of the potential loss of sexual sensation.

While following these guidelines, we also recommended that no surgery be done until: 1) the child is medically stable; 2) the parents are fully informed of the risks and benefits; and 3) an expert surgeon is found. Often, this takes longer than the recommended time frame.

Ultimately, the decision for or against early surgery is intensely personal, only to be made by the parents in close conjunction with the physician, and not to be rushed. Whatever they choose, parents must

(continued on page 18)

Growth Hormone and Height in CAH: A study of the effect of GH and GnRH on final adult height.

by Maria Vogiatzi, M.D.

Children with CAH are frequently tall before puberty, but complete their growth prematurely and are ultimately short as adults. This growth pattern can be caused by elevated adrenal androgens that result in bone age (BA) advancement and fusion of the epiphysis (growth plates in the bones). Despite treatment with steroids, adrenal suppression may not be perfect, allowing androgen levels to rise, leading to bone age advancement. When the bone plates are completely fused, growth is finished in the child. Furthermore, over-treatment with steroids can result in growth deceleration (slowing of the rate of growth). An additional problem can be early puberty, which frequently occurs in CAH and may contribute to advanced bone age (BA) and eventually to short adult height. The combination of these factors (advanced bone age, early puberty and slow growth rate) lead to a short adult height prediction in many children with CAH.

This article summarizes the results of a study that used growth hormone (GH) in combination with gonadotropin-releasing hormone (GnRH) analogue to improve the final height in children with CAH. In addition to GH and GnRH, all children continued their standard therapy with steroid replacement and flornidone, as needed, with the goal of maintaining satisfactory suppression of adrenal steroids. GH was used to improve growth rate that could be decreased because of steroid therapy. GnRH analogues are medications that suppress puberty as long as they are being administered. The most

common medication in this group is Lupron®, an FDA approved medication for the treatment of precocious puberty. In this study, Lupron was given to children with early puberty to prevent advancement of BA. A previous study in CAH looked at the effect of Lupron alone on final height and

Despite treatment with steroids, children with CAH have significant difficulties reaching their genetic potential in terms of height.

failed to show a positive outcome. This result was contributed to the growth deceleration that is usually seen with Lupron therapy. Therefore, this study examined the combination of GH and Lupron therapy.

Approximately 30 children with CAH, both with the classical and nonclassical form of the disease, enrolled in the study that was conducted at the New York Presbyterian Hospital/ Weill Medical College. The results of the first two years of therapy in the first 20 enrolled children (13 boys and 7 girls) were reported in 2001. Changes in growth rate, BA advancement and height prediction in the group of children who were treated with GH and Lupron (treatment group) were compared to those of children who were treated only with steroids in the past by the

same investigator (control group). For statistical analysis, each child of the treatment group was matched at the start of the study with a child in the control group for type of CAH (classical vs. nonclassical), age, BA, sex and stage of puberty.

The average age of the children at the beginning of the study was 8.6 years in both groups. The average BA was 11.8 years for both groups. The predicted adult height was just below the 3rd percentile in the treatment group and a little above the 3rd percentile in the control group. GH therapy was started first and Lupron was added with the onset of puberty. Twelve children of the 20 enrolled were not in puberty at the start of the study and remained prepubertal during the study. Therefore, they were treated with GH alone. The rest of the children received combined GH and Lupron treatment.

Growth rate increased by the end of the first year of the study. At the end of two years, growth rate was higher in the treatment group compared to the control group, and BA progression was much slower in the children who received Lupron. Predicted height improved significantly with each year of the therapy, so that at the end of the two years the gain was 11cm (or 4.3in.) in average over the initial height prediction. This corresponds to an improvement of predicted final height from just below the 3rd percentile to close to the 25th percentile. On the contrary, predicted height remained the same in the control group, close to the 5th percentile.

(continued on page 17)

Newborn Screening

by Connie Souder, CARES Intern

There are 29 genetic disorders, including CAH, for which the American College of Medical Genetics (ACMG) recommends nationwide newborn screening. All 29 disorders are treatable, but if not detected early, can result in death or lifelong disabilities.

Over the years many people have advocated for a uniform screening standard so that no baby suffers for having been born in the “wrong” state. However, problems such as delayed results, false positives, human error, and financial, legal, and ethical issues have delayed adoption of a uniform standard. Mississippi is currently the only state that requires screening for all 29 conditions.

Since its beginning in 2001, CARES has advocated for newborn screening for CAH in all 50 states and internationally. Twenty states have expanded their newborn screening programs to include CAH since CARES began advocating for expansion, and there are only four states (Arkansas, Kansas, Louisiana and West Virginia) plus the District of Columbia that do not currently screen for CAH. New Hampshire, Nebraska and Utah have approved screening for CAH but have not yet implemented it.

CARES is focusing its attention on Kansas and Ontario. Kansas requires screening for only 7 of the recommended 29 disorders and has not yet taken any steps towards expanding its requirements. In August, a team from the National Newborn Screening and Genetics Resource Center of Texas met with the Kansas Department of Health and Environment to study the

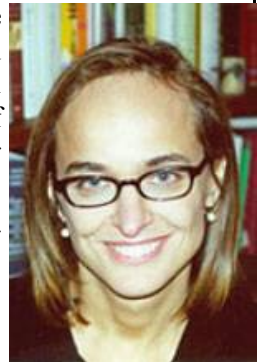
Meet Our Newest Medical Advisor!

**Kyriakie Sarafoglou, M.D. Pediatric Endocrinologist, Geneticist
and Inborn Errors of Metabolism Specialist**

CARES is pleased to welcome Dr. Kyriakie Sarafoglou to its medical advisory board. Dr. Sarafoglou is the director of the Center for Congenital Adrenal Hyperplasia and Intersexuality at the University of Minnesota Medical School, a multi-disciplinary center that specializes in the care of CAH patients.

In addition to her role as director at the Center for Congenital Adrenal Hyperplasia and Intersexuality, Dr. Sarafoglou is an active researcher. Her current research, which is both NIH and privately funded, examines growth and fertility. She is also collaborating with scientists at the Mayo Clinic on a study that is working to improve the monitoring of CAH and is leading a workgroup of Minnesota Pediatric Endocrinologists to standardize the care of CAH patients.

Dr. Sarafoglou serves as the Advisor to MDH Newborn Screening Program and Advisor to Upper Midwest States on 2nd tier CAH Newborn Screening. She is also the author of “Congenital Adrenal Hyperplasia: A Handbook for Patients and Their Family,” which will be available in the near future through CARES Foundation, Inc., and is editing/writing a textbook for McGraw Hill (“Essential Pediatric Endocrinology and Inborn Errors of Metabolism”). CARES is very pleased to have Dr. Sarafoglou on its Medical Advisory Board and looks forward to working with her.



logistics of expanding Kansas’s newborn screening program. Ultimately, it will be the Kansas legislature making the final decision on the expansion.

NBS in Canada leaves a lot to be desired as well. Under provincial jurisdiction, the number of disorders screened for ranges from 3 to 28, and only 5 provinces have formal NBS advisory committees and state mandates.

Ontario is set to expand its newborn screening program from 2 to 21 tests but has not included CAH. In fact, Ontario’s expansion does not include 4 of the top 7 disorders recommended for screening by the ACMG.

We encourage you to write letters of support to Kansas and Ontario. Please contact your legislators and

encourage them to expand newborn screening. The contact information for Kansas and Ontario can be found below.

Contact Information:

Kansas

Secretary of Kansas Department of Health and Environment

Roderick L. Bremby
Curtis State Office Building
1000 SW Jackson
Topeka, KS 66612
Phone (785) 296-1500
Fax: (785) 368-6368

Governor

The Honorable Kathleen Sebelius
Office of the Governor
Capitol, 300 SW 10th Ave., Ste.12S
Topeka, KS 66612-1590

Ontario

Minster of Health and Long-Term Care

(continued on page 16)

Office NEWS

Feedback Needed!

CARES Foundation needs your feedback! What would you like to know more about? What topics are most interesting or important to you? Contact us! We would appreciate it!

Newsletter Correction

In the CARES Foundation Newsletter V4:2, Spring 2005, page 3, Alyssa Ackenheil was mistakenly called Sarah Ackenheil. We apologize for the mix-up and thank ALYSSA for speaking at the EVERY1CARES luncheon. Great job, Alyssa!

Fundraisers Falling

Family Fundraisers have declined in recent months. We encourage anyone considering a fundraiser to contact our office for a fundraising guide. We are very fortunate to have generous corporate sponsors but still rely on the hard work of our member families.

Support Group Manual

CARES is currently developing a manual to be used by all support group leaders. We are including a section of "Frequently Asked Questions" and invite your input. Please contact Erin Anthony at (toll free) 866-227-3737 or erin@caresfoundation.org for more information.

DVDs Available

Fundraising and other informational DVDs are available. The DVDs feature some of our Medical Advisors speaking about various aspects of CAH. For a complete list, or to request copies, please contact us (toll free) 866-227-3737 or e-mail info@caresfoundation.org.

Fundrai\$ing Corner

Help us make our wishes come true!

CARES Foundation

Fall 2005 Wish List

Education & Programming

CARES Newsletter—\$5,000 per issue

Underwrite the cost of printing and mailing our comprehensive, 20 page newsletter to families affected by CAH.

Family Financial Assistance—\$500 per family

Help a family affected by CAH attend the CARES conference by sponsoring their travel and accommodation costs.

Conference Lunch—\$1000

Help by sponsoring a healthy lunch for our families who attend the CARES conference.

Research

Research Endowment—\$50,000

Make a substantial contribution towards CAH research, helping us get another step closer to a cure.

Office Needs

Computers—\$1,500

Help replace obsolete computers and printers for program planning and support.

All wish list supporters will be prominently featured in our next newsletter.

We'll Miss You, Connie!

Many thanks to our Summer Intern, Connie Souder. A Junior at Middlebury College, Connie worked very hard on various projects for CARES, such as researching NBS in Kansas, New Hampshire and Ontario (see her article on page 11). Connie also got us involved with Recycle For Free, a program in which families can recycle their inkjet cartridges to raise money for CARES. It was a pleasure having Connie in our office, and we wish her the best of luck in everything. Thank you, Connie. We hope you come back!

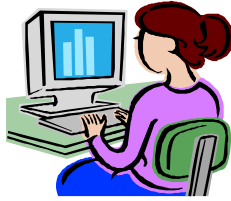
Thank You!

CARES Foundation thanks Sonya and Mickey Matson for their generous fundraising efforts. The Matson's garage sale was a huge success and proof that every effort helps!

We'd also like to thank Mickey's employer, Degussa, for generously matching the Matson's gift twofold.

CAH CHAT GROUPS

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."



**the URL for this chat group has changed*

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

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/> .

Warners speak at NBS Press Conference

by Susan Warner

Newborn screening saved my son's life. Bradley, now a healthy, happy 6-month-old, was the only child in California to have been diagnosed with CAH through a private screening test.

It's a miracle we caught it in time.

Bradley, my husband Matt and I have just returned from an exciting trip to the State Capitol in Sacramento to tell our story.

At the press conference we attended, state officials announced that California will now be testing all children for 75 metabolic and genetic disorders, including CAH.

Bradley was one of the "stars" of the show along with three other families with other disorders. While waiting for the cameras to roll, we met another child with CAH, several people from the March of Dimes, members of the press, and representatives from the Department of Health. Bradley gave his best full-body smile to all the new people he met, before spitting up all over my new pink shirt. Luckily, I told our story with only one hiccup: trying to say Congenital Adrenal Hyperplasia with my heart beating fast and Bradley pulling my hair. Since then, our story has been in several news papers and on the evening news! We even saw the "Governator" walk through the hall of the Capitol! (He's not as tall as I thought he'd be.)

We feel lucky to have the CARES Foundation, which has created a community for those of us who have children with CAH and helped mandate newborn screening throughout the country. It's wonderful that now every child in California will be screened like Bradley. Hopefully, the recent publicity regarding the new law will help alert and educate parents about early screening for genetic and metabolic disorders.



Susan, Matt and baby Bradley Warner spoke at the press conference kicking-off California's expanded NBS.

Support Group NEWS

South Carolina

If you live in SC and would like to get to know other families affected by CAH, join the SC Support Group for it's Fall Bash. Support Group leaders, Kevin and Johnette Kinard, will host this event at their home. Please contact them if you would like to attend an afternoon of fun, food and fellowship.

Hope to see you and your family there!!

Saturday, October 15th

1pm.

1988 Mt. Pilgrim Church Road

Prosperity, SC

803-364-9945

kevin1@backroads.net

Wisconsin

Support group leaders Lisa Jaskie and Laurel Meier are planning their 5th CAH Family get together. It will be held at:

Treasure Island Waterpark Resort

Wisconsin Dells, WI

Saturday, Nov. 5th

2pm.

For more information please contact:

Laurel

715-341-9697

laurelmeier@charter.net

or

Lisa

414-645-0782

Lisa1273@msn.com

<http://groups.msn.com/CAHFamiliesfromWisconsin>

Northern California

Families in the Northern California area are invited to attend a CARES CAH Picnic.

Saturday, Oct. 1, 2005

10am to 5pm

Micke Grove Park

Lodi, CA

\$5 Parking Fee

Activities include: Horseshoes, Playground, Japanese Garden, Zoo and Museum. For more information and directions visit www.mgzoo.com. Please bring a picnic lunch, drinks and chairs/blankets for your family. Dessert will be provided. Please RSVP to:

Adria Stoner

stonerfamily@starstream.net

(916) 434-8405

If you have a support group function you'd like advertised in our Winter2005-2006 newsletter, please e-mail erin@caresfoundation.org .



CARES US Family Support Groups

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¡Atención Personas que Hablan Español!

La Fundación CARES quiere ayudarlo a que aprenda acerca de HAC. Si usted tiene alguna pregunta por favor llámenos (866) 227-3737, o escribanos (renata@caresfoundation.org) para organizar una conferencia telefónica con un traductor. También existe un grupo de Yahoo en idioma español que recibe familias e individuos afectados por la Hiperplasia Adrenal Congénita. Para suscribirse vaya a <http://mx.groups.yahoo.com/group/hiperplasia/>.

*New groups are added all the time. For the most up-to-date list, visit www.caresfoundation.com/support.html.

For information on starting a support group in your area, please call us (toll free) 866-227-3737 or e-mail info@caresfoundation.org.

NIH Study of Hormones and Emotion Regulation

Investigators: Dr. Deborah Merke, M.D., M.S.
Dr. Monique Ernst, M.D., Ph.D.
Dr. Daniel Pine, M.D.

We are currently evaluating the effects of hormones on emotions, memory, and attention in patients with CAH. This study can be completed in either one or two visits to the NIH Clinical Center in Bethesda, Maryland and includes:

- **Updated endocrine evaluation by Dr. Deborah Merke**
- **Structured psychiatric interview**
- **Questionnaires**
- **MRI (does not involve radiation)**

In order to participate in this study, volunteers must:

- **Have the classic form of CAH (21-hydroxylase deficiency)**
- **Be between the ages of 9 to 25 years old**
- **Not be on other medications besides CAH-related medications**

Participants will be paid \$350 for completion of the study and partial payment is available if only part of the study is completed.

For more information or to volunteer, please contact Julie Hardin or Liza Golan-Green at the NIH at (301) 451-9194.

NCAH study at Children's Hospital of Los Angeles

The Division of Endocrinology at Children's Hospital Los Angeles is currently recruiting subjects for a research study aimed at determining the stress-fighting ability in subjects with Non-classical Congenital adrenal hyperplasia (NCAH) and comparing these responses to those in subjects with Classical Congenital Adrenal Hyperplasia (CAH) and those in carriers of either disorder. If you have NCAH, CAH or are a family member (parent or sibling) of someone with either disease, and are interested in participating in this study, please contact: Dr. Maria Karantzis (323) 644-8705 or Dr. Mitchell Geffner (323) 669-7032.

CAH Study at UNC Chapel Hill, North Carolina

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 the 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-

**Now Open to
Younger Children!**

blind/placebo-controlled crossover study.

For more information, please contact

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

(919) 216-5946 (pager)
(919) 966-4435 ext 224 (voice mail)
(919) 966-0428 (fax)

or

**Roxanne Schock, CDE, Clinical
Coordinator**

(919) 966-0428 (voice mail)

All visits, including laboratory testing, research medications, 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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Reproductive Function Study

Clinical researchers at the Center for CAH and Intersexuality are recruiting males with Congenital Adrenal Hyperplasia (CAH) and decreased spermatogenesis (process of forming sperm cells in the testes) for a NIH-sponsored research study performed at the University of Minnesota Medical School. In CAH due to 21-hydroxylase deficiency (21-OHD), the body produces excess androgens (male hormones) because of the defect in the 21-OH enzyme. In all humans, androgens are converted to estrogens through a process called aromatization. In 21-OHD, the increased levels of androgens are aromatized/converted in glandular (ie testes) and extraglandular tissues and result in elevated estrogen levels. Ideally the production of adrenal androgens is normalized in CAH patients by glucocorticoid replacement therapy. However, even well controlled CAH patients still manifest the adverse effects (compromised final height, polycystic ovarian disease, male infertility, etc) of elevated androgens/estrogens. We hypothesize that these chronically elevated estrogen levels negatively affect spermatogenesis in males with CAH. The research study examines whether reducing the body's estrogen level with an estrogen-reducing medication will improve overall fertility by reducing the negative effects of elevated estrogen levels.

Eligible Participants:

- Males 16-50 years of age
- Diagnosis of CAH

Principal Investigator: Kyriakie Sarafoglou, M.D.
 Director, Center for CAH and Intersexuality
 Division of Pediatric Endocrinology
 Division of Genetics and Metabolism
 University of Minnesota Medical School
 General Clinical Research Center

For further information, please call (612) 626-6522

Update on Newborn Screening

(continued from page 11)

The Honorable George Smitherman
 Phone: (416) 327-4300
 Fax: (416) 212-4255 (Senior Policy)

Chair, Newborn Screening Committee

Joe Clarke, M.D.
 joe.clarke@sickkids.ca
 Sick Kids Main: (416) 813-1500

If you live in a state that does not currently screen for CAH and would like to contact your legislator, you can find the contact information here:

<http://www5.lsb.state.ok.us/legislators/lbaddress.asp> .

You can also visit <http://www.newborn-screening.org/letter.html> to write to the governor or other government official of any state.

Attention! Keep Extra Meds, Injection Kits

In light of the recent events in the Southeast, we would like to remind all CAH families to keep an extra month's supply of medication on hand at all times. In addition to this, families affected by Classical CAH should also keep extra injection kits in the event of an emergency.

We have been in contact with almost all of our member families in the Katrina affected area, and most are okay. We will do everything in our power to help those member families affected by Katrina and invite your help as well. If you'd like to help please e-mail info@caresfoundation.org or call (toll free) 866-227-3737.

Parent Tip:

Sports and Medical "Jewelry"

Our 11-year-old twin daughters, Lisa and Annie, have played on a boys baseball team for the past three years. Lisa, who has SWCAH, has worn a medical alert bracelet since she was born. When she was little she wore it around her ankle, and for about the past 7 years she has worn it on her wrist. Last year, an umpire told her she'd have to take off her "jewelry" before she could play. We explained to him that it was a medical alert bracelet and it had to remain on her. We put a sweat band over it, and the problem was solved. Now along with having to have hair ties that match her uniform, we also have to have sweat bands that match. Just another good reason for a girl to go shopping! And, of course, Annie has to have some too.

-Susan Zaremski, North Carolina

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Study: GH and CAH

(continued from page 10)

The study was continued to determine if the beneficial effects of GH and Lupron therapy were temporary or could be sustained until final height, and a report on the final height of the first 14 children (8 boys and 6 girls) was published this summer. Their final height was compared to that of children who were treated only with steroids (control group). As in the first report, each child of the treatment group was matched at the start of the study with a child in the control group for type of CAH, age, BA, sex and stage of puberty.

This paragraph summarizes the growth characteristics and outcome of these 14 treated children and their controls. For simplicity, the results, which are reported here, refer to the average values for each group. The age of the treated children at the beginning of therapy was 9.7 years, their bone age was 12.5 years and their predicted final height was close to the 5th percentile. The children in the control group had the same growth parameters at baseline. In the first year of the therapy, the growth rate of the experimental group was a little more than one inch faster than the control group. In the following years, growth rate slightly decreased for the experimental group but remained higher than that of the untreated children. The change in BA did not differ between groups, or otherwise, was not affected by the therapy.

The treated children reached a final height between the 25th and 50th percentile in contrast to close to the 5th percentile for the control group. Final height in treated boys averaged 67.5 inches and treated girls averaged 64.5 inches. In contrast, the final height of untreated boys and girls was 64 inches and 62 inches respectively.

The treated group, as a whole, improved their height by 2.9 inches compared to their initial height prediction. However, the final height in the control group was practically the same with their initial height prediction. Length of therapy was approximately 4.5 years.

Comparisons were also made between the final height of each group and their corresponding target height. Target height is an adjusted average of the height of both mother and father and reflects the genetic potential of the child. The treated group reached a final height that was one inch shorter than its target height. Contrary, the control group's final height was about 3.6 inches shorter than its target. Both groups had similar target heights.

These studies are the first to show that GH therapy, combined with Lupron in case of early puberty, improves the height potential and final height of children with CAH. The treatment had excellent short-term results that could be sustained until the end of growth. There were not reported adverse effects except pain at the injection site with Lupron. As the rest of the children of this study complete their growth, we will gain additional information about the long term efficacy and safety profile of this therapy.

Despite treatment with steroids, children with CAH have significant difficulties reaching their genetic potential in terms of height. GH therapy, combined with Lupron in case of early puberty, can be an attractive alternative option for those children with CAH, who despite treatment with steroids, suffer from an advanced BA, and therefore, are projected to have a poor final height. Otherwise, not all children with CAH are candidates for this therapy. It can be considered, however, for those who

continue to have low predicted final height despite steroid therapy. It is of interest that the studies included a number of children with non-classical CAH: 12 out the 20 children in the first report and 5 out of the 14 in the second report. The children with NCAH have a milder disease, but also tend to present late in childhood, and may experience significant BA advancement before the diagnosis is established. However, the form of the disease did not appear to affect the response to the GH therapy

Beyond the medical outcome, problems with the GH and Lupron therapy include its high cost, difficulties obtaining approval by the insurance and the intensity of the treatment. GH therapy requires a daily injection, once at bedtime, that is very similar to insulin injection in terms of administration. It is typically a lengthy therapy (i.e. until the child reaches near adult height). Lupron is given monthly as an intramuscular injection that is frequently painful. In order to be most effective, it is recommended that GH treatment begin before puberty and continue until the bone age shows fusion of growth plates. In general, the longer the GH therapy is, the more significant is the effect on final height.

Finally, the response to the GH/ GnRH analogue therapy was quite variable. Factors that may influence the outcome, such as adrenal control or type of CAH, could not be identified. This speaks to the fact that there are multiple, still poorly defined factors, that may affect growth overall and response to GH therapy, and represents a challenge for all those who care for these children. The answer to this challenge lies in continuous commitment to research that can lead to a better understanding of the disease and its impact on growth.

Personal Story (continued from page 7) in 2003 with Dean's List Honors and got married in 2004. I am employed as a researcher through the State of California and plan to attend graduate school to earn a master's in Public Health after my husband completes law school.

Tate just graduated from high school with a strong scholastic record and will be attending University of Nebraska-Lincoln in the Fall. She

plans to pledge a sorority and to pursue a degree in education.

My parents are nearing retirement and have plans to travel. They also plan to achieve Leigh's goal of working in short-term missions overseas.

All of us have faced unique challenges as a result of having CAH, but CAH has in no way dictated what we can accomplish.

Through accomplishing my own individual goals, and with the support of my family and friends, I feel that CAH is not a barrier to any possible attainment. Having CAH has presented obstacles for our family, by far the greatest and most painful one has been the loss of my sister, but we are committed to embracing future challenges with the strength and joy that Leigh so often showed while reaching her goals.

Controversy (continued from page 9) be comfortable and confident in their decision. It is important also to recognize that social and cultural conditions vary widely and each family is different. What may be acceptable for one family is often unheard of by another.

Much of the conversation about surgery has been conducted by intersex groups, such as ISNA, who choose to include CAH as an "intersex" condition.

Use of the word intersex is itself being debated, and most people with CAH have rejected it entirely. An umbrella term, the word carries with it a certain amount of stigma and ambiguity and many want it phased-out—a point not lost on intersex advocacy groups. In fact, in two handbooks due out shortly from ISNA, intersex has been avoided entirely, using instead the terminology Disorders of Sexual Differentiation (DSD).

CAH is a disease with several forms, only one of which, classical CAH, could even be loosely considered an "intersex" condition. Even then, only females are affected by genital anomaly and to widely varying extents.

Again, it is not our role to decide who is right or wrong, only to support the CAH community in the best way that we can.

A Very Special CARES Friend

by Jami Abell Patterson

CARES would like to thank Hannah Mandel for all her work on behalf of CAH. Hannah learned that her friend and soccer teammate Sarah Venit was affected by NCAH after Sarah sold Every1CARES bracelets to her team. Taken by Sarah's courage, Hannah decided early on that it was a cause she felt she needed and wanted to undertake.

Hannah was studying for her Bat Mitzvah and had already identified a project she wanted to do for her Mitzvah project. She decided midstream to go to her Rabbi and explain that she wanted to change her project to raise awareness and funds for the CARES Foundation.

Hannah wrote a speech, after doing research, that she presented to the entire Religious School community at Leo Baeck Temple in Los Angeles. After the speech, she sold whatever bracelets she had on hand and then took orders for more. Hannah also read her speech in each of her classes at school. She sold bracelets to our clergy, temple members, family and friends and still continues today.

Most important to Hannah was the need to educate everyone about the disease and how important early screening and research is. At the end

of the religious school year, the 7th grade class decided to put on a Mitzvah Fair. Hannah decided that she wanted to come up with something that would draw a lot of attention (and money). She put together a pie throwing booth, which she decorated with pamphlets and bracelets, and asked our clergy and our director of education to allow the kids to throw pies at them. They all agreed and, needless to say, Hannah's booth was the most popular. At the end of the day, all the tokens were tallied, and she raised the most money, \$100 for CARES Foundation. On her own, Hannah has sold over 200 bracelets, using another 100 as napkin rings at her Bat Mitzvah.

After speaking with Kelly, it was decided that this is the best way to honor and thank Hannah for her dedication to the CARES Foundation. For a child to become so dedicated to a cause without being affected is rare, a true testament to Hannah's character. Perhaps after reading this article, others will be inspired by her enthusiasm and dedication to raising awareness for CAH and CARES.

Hannah, on behalf of everyone at CARES Foundation, thank you.



CARESFoundation

2005 CAH Conference

Riley Outpatient Center ♥ Ruth Lilly Learning Center ♥ Indianapolis, Indiana

Saturday, October 22nd, 9AM to 5PM

Join Us For This Event, With Speakers:

Susan Baker, Ph.D., Psychoendocrinologist, Mount Sinai Medical Center, New York

Sheri Berenbaum, Ph.D., Professor of Psychology, Pennsylvania State University, Pennsylvania

Erica Eugster, M.D., Section Director, Pediatric Endocrinology/Diabetology, Associate Professor of Pediatrics, Riley Hospital for Children, Indiana

Deborah Merke, M.D., Chief of Pediatric Services for the Clinical Center of the National Institute of Child Health and Human Development (NICHD); National Institutes of Health, Maryland

Maria New, M.D., Pediatric & Adult Endocrinologist, Mount Sinai Medical Center, New York

Richard Rink, M.D., Robert A. Garrett Professor of Pediatric Urologic Research, Chief, Pediatric Urology, Indiana University School of Medicine & Riley Hospital for Children, Indiana

Presentation Topics Include:

New Advancements in CAH Treatment & Future Trends in Research

Research Roundtable

Behavioral and Psychological Aspects of CAH

Monitoring & Treatment of CAH/NCAH

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...

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

Accommodations: *University Place Conference Center & Hotel, 1-800-627-2700 (select Option #1) for \$99.00 per night plus tax. This discounted rate is available on a first come, first serve basis. Please reserve your room for October 21st and/or October 22nd and indicate that you are with the CARES Foundation group.*

Travel: *Angel Flight America has generously offered its resources to help CARES members attend the conference. Interested members should be within 1,000 miles of Indianapolis and have financial need. For more information, call Angel Flight America at 1-877-621-7177 or visit the website at www.angelflightamerica.org.*

Childcare: *This event is intended for adults and older children. Snacks/ lunch will be provided. A children's recreation room will be staffed by volunteers, but spaces are limited. Please RSVP as soon as possible to guarantee a space for your children.*

Directions: *Riley Outpatient Center is located next to Riley Children's Hospital, at 575 West Drive, Indianapolis, IN. For detailed directions, please go to: http://rileychildrenshospital.com/guide_outp.jsp?locid=590 or call 1-800-248-1199.*

Special thanks to Riley Hospital for Children. This event is sponsored by an educational grant from Eli Lilly and Company.



Registration Form

RSVP by October 8th, by: Mail: CARES Foundation, Inc. 189 Main St., Millburn, NJ 07041; E-mail: renata@caresfoundation.org; Fax: 973-912-8990.

Any questions related to the conference can be directed to Renata Blumberg.

Name: _____

Address: _____

City: _____ State: _____ Postal Code: _____

E-mail: _____ Home Phone: _____

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)? _____

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Child Sizes Now Available!



Child-sized EVERY1CARES bracelets are now available. Our staff has found the bracelets are also nice for women with smaller wrists.

Bracelet sales continue to rise thanks to the energy and dedication of CARES Foundation members! So far, we have distributed a total of 7,063 EVERY1CARES bracelets, resulting in \$21,624 in sales. Bracelets are available at a cost of \$3 each (minimum order of 5, plus shipping and handling.)

**To order, call
(866) 227-3737 or order online at
www.caresfoundation.org .**

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