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, clomiphen citrate, a pill that can increase the likelihood of ovulation, can be used. The use of clomiphen 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

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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:

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

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!!!

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.iqive.com and a donation will be made to CARES.
Register (keyword: CAH) and enter CARES Foundation as the cause you want to support.

CARES Holiday Tribute Cards are now available, $5 each. To order, please call us at 1-866-227-3737.

Coming soon... CARES Christmas Tribute Cards!

In Loving Memory of Israel Chaim, S.W.C.A.H.
1/63 - 5/65 son of Yakov and Esther

Please keep this family in your thoughts and prayers.
CARES Foundation, Inc.

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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 testing 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, or 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. The women with who are 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 reduce 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.

Marsha Finnegan will be the first to tell you she’s “had 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 think about 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 later. Very high 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 anxiety or worries about getting pregnant or having a child with CAH?

Honesty, 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 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 are 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 treatments?

We increased dosage of medicare for my pregnancy and I did experience gestational diabetes. So, I did end up stopping 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 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 is this possible.

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

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 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, not 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 (continued on page 7)
Factors influencing parental decision making
By Elizabeth Longhin 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 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 (Longhin, E. Postel, R. Bathurst. 2004. 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 Watson, M.D. Pediatric Urologist and Professor of Pediatrics 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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News Briefs

Newborn Screening Updates

South Dakota

South Dakota has just expanded its newborn screening program from three disorders to over 30 disorders. Expanded screening began on June 1, 2005. Although parents will have to pay for the cost of the testing, it may be covered by some insurance plans.

New Hampshire

New Hampshire recently added five new disorders, including CAH, to its required screening list. New Hampshire now screens for 11 conditions, falling short of the 29 recommended.

Germany

As of April 1, 2005 the German newborn screening program was expanded to a total of 14 disorders, including CAH. German authorities have made testing for other conditions during the newborn screening process forbidden, requiring that any results acquired about other diseases be discarded immediately.

Ontario

Ontario will expand its newborn screening from two to 22 disorders. The expanded screening does not include CAH and leaves out four of the top seven disorders recommended for screening by the American College of Medical Genetics.

The new tests will be performed using three new tandem mass spectrometry machines. The tests may be covered by an Act, Bill 101, to amend the Health Insurance Act. The Bill requires supplemental newborn screening through the use of tandem mass spectrometry as an (continued on page 18)
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 (GnRHi) analogues 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 flutamide, 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 Lupon®, an FDA approved medication for the treatment of precocious puberty. In this study, Lupon was given to children with early puberty to prevent advancement of BA. A previous study in CAH looked at the effect of Lupon alone on final height and growth.

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

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

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 Lupon 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 Lupon 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. BA progression was much slower in the children who received Lupon. Predicted height improved significantly with each year of the therapy, so that at the end of the two years the gain was 11 cm (or 4.3 in.) 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.

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 recently went from 0 to 7 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 logistics of expanding Kansas’s newborn screening program. Ultimately, it will be the Kansas legislature making the final decision on the expansion.

In Ontario, it 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

Ontario
Office of the Honourable Kathleen Sebelius
Minister of Health and Long-Term Care
308 SW 10th Ave., Ste.12S
Columbia, MO 65201-1900

Ontario
Office of the Minister of Health and Long-Term Care
Fundraising 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.

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.

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 research on CAH 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!

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/caresteenchat/ and click on "Join This Group."

*the URL for this chat group has changed

CAHISTSERS2: A listserv for adult women with late-onset CAH. To learn more about the CAHISTSERS2 group, go to: http://groups.yahoo.com/group/CAHISTSERS2

CARES Spanish Group: A Yahoo Group for the Spanish-speaking CAH community. To learn more and join, go to: http://msx.groups.yahoo.com/group/hiperpilasnia

Warrants 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 splitting 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.

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 of its 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
715-341-6897
laurelmeier@ charter.net

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
Mickie Grove Park
Lodi, CA
$5 Parking Fee

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

Adria Stoner
stonerfamily@streamnet.net
(916) 434-8405

If you have a support group function you’d like advertised in our Winter 2005-2006 newsletter, please e-mail erin@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) 496-1914.

CAH Study at UNC Chapel Hill, North Carolina

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 amiodipine). Amiodipine has been shown to be safe and effective in infants treated for a variety of medical conditions.

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

For more information, please contact:
Karen Loechner, M.D., Ph.D.
(919) 216-5946 (pager)
(919) 966-4345 ext 224 (voice mail)
(919) 966-0428 (fax)

 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 non-glandular (ie adrenal cortex and extraglandular tissues) endocrine cells. This leads to an increase in adrenarche and the normalization of 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:
Kyrskie 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

Study: GH and CAH

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) of the study group in the 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 affected by the therapy.

The treated children reached a final height between the 25th and 50th percentile in contrast to 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 from 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. How this 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 thin gauge 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 more effective, it is recommended that the treatment begin prior to puberty and continue until the bone age shows fusion of growth plates. In general, the longer the therapy the higher 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 will help to determine the understanding of the disease and its impact on growth.
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 every CARES 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 Bat 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.
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Specializing in Custom Made and Engraved Medical Jewelry.
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New items added all the time... check the website!!

Don't wait... it could protect and save your child's life!
It saved Cody's life! We also carry many more items. A portion of all profits will go to CARES Foundation, Inc.

Have you recently moved, changed your phone number or e-mail? Please make sure to let us know, so we can keep our information current.

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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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