

## Management of Hair Loss in Androgenic Disorders

Walter Futterweit, MD, FACP, FACE

Thinning hair due to the effects of male hormones (androgens) is called androgenic alopecia. It is a major source of psychological distress to women. This male-pattern hair loss is often seen in women with congenital adrenal hyperplasia (CAH), polycystic ovary syndrome (PCOS), other hormonal disorders (thyroid disease), those associated with androgen excess and certain drugs (e.g., propranolol, Accutane), anemias, nutritional deficiencies, and severe illnesses.

Associated with hormonal changes causing the alopecia are genetic and environmental factors which are responsible for the frequent finding of onset of hair loss at the top of the head (vertex) and the angles of the frontal hair line. In many, the pattern may start as a triangular thinning, which is called the "triangle sign." There is gradual progression of hair loss at the frontal midline which progresses towards the vertex sides of the scalp. In most women with androgenic alopecia, the frontal hair line remains intact despite diffuse hair loss.

The average number of hairs lost in a day is about 100-150. It should be noted that it may take about 20 to 25 percent

of total loss of scalp hair before it may be visibly recognized by the woman. Thus, an awareness of excessive hair loss at combing or after washing the hair, usually are the first signs of onset of alopecia. Transient hair loss (telogen effluvium) may be another cause of hair loss, for example a few months after the birth of a baby, and a return to normal hair loss may occur three to five months later.

The incidence of androgenic alopecia in CAH is not clearly defined, but in another androgenic disorders, such as PCOS, it may vary from 40 to 70 percent of these afflicted women. Experienced endocrinologists have seen similar data in women with CAH. In my experience, the severity of the alopecia appears to be somewhat more severe in CAH and appears at an earlier age than in patients with PCOS. It is important for each woman with alopecia to initially evaluate her own individual lifestyle and its relation to hair loss.

Adequate nutrition is vital for healthy hair. For example, some who are on restricted diets may require dietary readjustment with selected supplements. Another cause of hair loss may be chronic anemia and/or iron deficiency. The

*continued on page 6*

### TAKE 10 FOR CARES CARES Member Survey 2007

Please take 10 minutes to complete a survey designed to help CARES get to know our members better.

**CARES is always striving to provide better, more effective services to you and the CAH Community.**

Your input will help us achieve that important goal.

To complete the survey, please visit:

[www.caresfoundation.org](http://www.caresfoundation.org)

or call (toll free) 866-227-3737

*Ready, Set, Go* ~~Go~~  
*Relax*



**see page 13 for more information**

# Eisenbergs at Oktoberfest



**CHICAGO, September 21, 2006:** CARES members Audrey and Josh Eisenberg accept Peregrine's grant during Peregrine's second annual Oktoberfest. This award was used to fund year two of the CAH Natural History Study at the National Institutes of Health.

In October, Josh completed his second Chicago Marathon and asked sponsors to donate to CARES on his behalf. Thank you, Josh!

## IN THIS ISSUE

### CAH Articles

Managing Hair Loss	1
Parent Perspectives	4
Use of Antiemetics	22

### Debate: Carrier Testing

Ethical Issues	16
Decision Making	19

### Personal Stories

Blessings in Disguise	8
Paving the Way	9

### CAH Conference

2007 Re-cap	10
-------------	----

### Support Group News

Save the Dates	14
Worldwide Directory	15

### Fundraising

Go Buckeyes!	12
Cards for CARES	12
No Sweat Marathon	13

## Sunshine and Support in California



**September 2006:** Children from the Northern California Support Group had a great time during their picnic at Micke Grove Park.

© 2006 CARES Foundation, Inc. All rights reserved. Republication or redistribution of CARES newsletter content, including by framing or similar means, is prohibited without the prior written consent of CARES.

**Disclaimer:** Any communication from CARES Foundation, Inc. is intended for informational and educational purposes only and in no way should be taken to be the provision or practice of medical, nursing or professional healthcare advice or services. The information should not be considered complete or exhaustive and should not be used in place of the visit, call, consultation or advice of your physician or other healthcare provider. You should not use the information in this or any CARES Foundation, Inc. communication to diagnose or treat CAH or any other disorder without first consulting with your physician or healthcare provider. The articles presented in this newsletter are for informational purposes only and do not necessarily reflect the views of CARES Foundation, Inc.

a message from the

## Executive Director

Dear Friends,

### Milestones

As I reflect on this past year, I realize we have hit significant milestones. It is hard to believe a whole year has passed since the fire consumed our offices. At CARES, we tend to think in terms of “before the fire” and “after the fire.” It has become our milestone, our marker of time. With hard work, perseverance, grit and lots of support from you, we have made it to this one-year anniversary and are stronger than ever. We are so blessed.

The other milestone we have passed is our fifth anniversary! CARES was launched in 2001. How quickly we have grown and how deep the roots of our community have spread. It is due to you—our members. You have joined CARES in our effort to create the wonderful resource for those affected by CAH and helped us to expand our impact.

### Visit to NIH

In December, I had the opportunity to visit Dr. Deborah Merke and Carol Van Ryzin, CPNP at the National Institutes of Health, Institute of Child Health and Human Development. As you know, Dr. Merke is running the first and largest Natural History Study of CAH, which is also supported by a grant from CARES. To date, CARES has donated \$125,000 to this program and we will donate another \$65,000 this May to help run the study through 2008. So far, they have enrolled and studied over 150 individuals with CAH. The data and insights into CAH gained from studying such a large group of patients will be invaluable to our community and hopefully lead to better treatments or a cure. We owe special thanks to **Bill Trzos**, **Quest Diagnostics** and **Peregrine Charities** for their generous donations to support this research.

The NIH program is impressive. The Children’s Clinical Research Center is bright, clean, has beautiful facilities, along with the Children’s Inn right across the street for lodging for study participants. Dr. Merke and her staff are focused on helping the patients receive the best care possible. In addition to a complete work-up, the staff does extensive training with the



families/patients so that they understand CAH and the instructions for care. It is truly a patient-centered facility run by caring staff.

New studies will be launched in the fall, and the data collected so far is being analyzed so that preliminary results may be published in the near future.

### Conference 2006

Our conference was outstanding! Our speakers were terrific and once again graciously donated their time and expertise to help our families.

We owe our heartfelt gratitude to our generous sponsors: **Centers for Disease Control; Pfizer, Inc.; PerkinElmer; Ethicon, Inc.; Eli Lilly; Merck; March of Dimes; Genentech; Organon; The Fairhurst Family; and Patty Genovay.**

We thank Saint Barnabas Medical Center and Dr. Henry Anhalt for co-hosting this conference, for providing guidance on the program and for providing the contact hours for nurses. We also thank the Newborn Screening Programs of New Jersey, New York, Pennsylvania, Connecticut and Delaware for helping us reach new families.

I encourage you to turn to **page 10** to read more about the conference.

### Thank You, PerkinElmer !

This issue is sponsored by our friends at PerkinElmer, a dedicated supporter of CARES and the CAH community.

### New Year, New Beginnings

As we embark on 2007, I look forward to a year of calm (no more disasters, please!) while we can get back to improving and increasing our services. Thanks for bearing with us and supporting us during a tumultuous 2006.

Warmest Regards,

*Kelly*

### Founders

Kelly and Adam Leight

### Staff

Kelly R. Leight

Executive Director

Kelly@caresfoundation.org

Meryl I. Stone

Associate Director

Meryl@caresfoundation.org

Erin E. Anthony

Program Development

Erin@caresfoundation.org

Mariel Vargas

Program Development

Mariel@caresfoundation.org

Jennifer Mann Rosenblatt

Development Associate

Jennifer@caresfoundation.org

### Consultants

Stephanie Erb

Member Services Consultant

Stephanie@caresfoundation.org

Gretchen Alger Lin

Public Relations Consultant

Gretchen@caresfoundation.org

### BOARD OF TRUSTEES

Kelly Rosso Leight, President

Jami Abell-Patterson, Vice President

K.Adam Leight, Treasurer

Vivian Altman Quintanilla, Parliamentarian

Stephanie R. Fracassa, Secretary

Mark Engman

Nancy Kislin Flaum

Janet Green

Monica Heinze

Tonya Judson, R.N.

Gregory Kraff

Jayne Mackta

Alan Macy

Michelle May, M.D.

Hope Z. Raphaelian

Diane Snyder, M.D.

Bonnie Stevens

### SCIENTIFIC & MEDICAL ADVISORY BOARD

Henry Anhalt, D.O.

Ricardo Azziz, M.D.

Susan W. Baker, Ph.D.

Sheri A. Berenbaum, Ph.D.

Walter Futterweit, M.D.

Mitchell E. Geffner, M.D.

Daniel Gunther, M.D.

Claude Migeon, M.D.

Maria I. New, M.D.

Dix P. Poppas, M.D.

Richard C. Rink, M.D.

Scott A. Rivkees, M.D.

Ellen Seely, M.D.

Phyllis W. Speiser, M.D.

Bradford L. Therrell, Ph.D.

Maria Vogiatzi, M.D.

Garry Warne, M.D.

*This newsletter is published 3 times a year.*



## Making Good Decisions about CAH: Parent's Perspectives

Laura A. Siminoff, PhD  
Mary Beth Mercer, MPH  
David Sandberg, PhD

Last summer, a group of scientists convened to discuss the issue of how families of children with CAH were making decisions about the medical treatment their children receive. We were aware of how hard it can be, in the case of daughters, to make decisions about whether or not genital surgery is needed and, if so, what type, and whether surgery should be done immediately or delayed. Moreover, issues around medical care are a continuing question for all affected children. We wanted to explore whether there were ways families could be helped to make decisions that would ultimately lead to healthier children and less stress for families. The research we conducted is a first step toward that goal. Included in the research team are Laura A. Siminoff, PhD, a public health social scientist who does research in health communications and decision

making, David E. Sandberg, PhD, a pediatric psychologist, researcher and expert in the psychological implications of CAH, Barry Kogan, M.D., a pediatric urologist, Phyllis Speiser, MD, a pediatric endocrinologist, and Miriam Schuchman, MD, a psychiatrist and bioethicist. Mary Beth Mercer, MPH later joined the team as the Research Coordinator and has been instrumental in collecting the data and analyzing it.

We decided that we first needed to understand how families are now making health care decisions for their children and to learn what the issues are from their perspectives. We interviewed with 10 family units who were recruited at the 2005 CARES Foundation Conference. The interview was designed to allow parents to tell us

about their experiences in their own words. Several issues emerged that are helping to shape the direction of future research.

First, we found that the day-to-day medical management of CAH was paramount to parents. Because CAH is a serious, potentially life-threatening condition that requires medication several times a day and constant monitoring, effective communication with healthcare providers and informed decision making about medical



management is key to helping families keep kids healthy and maintain normal activities. **Parents reported that information they received from healthcare providers was often incomplete and inadequate for their needs as parent caregivers and decision makers.** Therefore, they supplemented their knowledge with books, websites, and organizations (such as CARES) and from speaking with other parents of children with CAH. Parents also underscored the need to be informed consumers of healthcare. This was most evident in their constant quest for comprehensive up-to-date information on monitoring their child's condition and what medical and surgical treatments of CAH should be pursued. Because of the rarity of

CAH, many had difficulty finding a knowledgeable endocrinologist to provide effective on-going medical management of their child. A number of parents reported continually seeking the most qualified endocrinologists and urologists to ensure their children receive state of the art care.

In addition to daily medical management, parents of girls with Classical CAH must also navigate decisions about surgical treatment. Parents reported some problems with

healthcare providers including receiving incomplete or contradictory information on a variety of issues including the necessity of surgery on the external genitalia, optimal timing for surgery, surgical procedures and aftercare. Parents are looking for the most experienced surgeons who are also effective communicators of potential risks and benefits associated with different surgical options. This approach

allowed parents to be actively involved in decision-making. Some parents reported postponing surgery until they felt comfortable that they were fully informed and until they found the "right" surgeon. Parents' decisions were usually focused on the timing of surgery (infancy vs. pre-puberty) and which surgeon should perform the procedure. There are also questions as to how much to include older children in decision-making and whether it is better to delay surgery until the child (or teenager) can have input.

The following themes emerged as important and relevant to parents' experiences:

**Partnership:** Parents expressed a real desire to be in partnership with their children's healthcare providers in terms

*continued on next page*

*continued from page 4*

of monitoring their children's health, communications about managing symptoms and treatment, surgical interventions and overall inclusion in decision making about their children's care. Many parents expressed the need to be taken seriously by their children's healthcare providers and stated that they 'know their children best'. When this occurs, they can work quite effectively with the healthcare providers to ensure that their children are well monitored and treated.

**Advocacy:** Parents take on the role of advocate for their children in order to ensure that they receive appropriate monitoring and treatment. Parents who are well informed and assertive with their children's healthcare providers feel empowered in managing their children's CAH. **However, these parents also expressed frustration that they are often more informed than the endocrinologists, surgeons, and pediatricians.** Almost uniformly, parents mentioned that their general pediatricians were unable to provide anything more than emotional support. Parents complained that many of the endocrinologists they dealt with really did not understand the illness—that their children were either over— or undertreated. They also felt that symptoms were ignored or misinterpreted. Surgeons were often reported to be noncommunicative and failing to present the full range of options. Finally, **parents expressed concerns about their children's right to privacy and respect during physical examinations and discussions in front of or with the child.** Parents expressed ambivalence as to how to advocate for appropriate privacy without insulting healthcare providers or sounding uncooperative.

**Credibility and Trust:** Many parents reported difficulties finding physicians who were knowledgeable and experienced in effectively managing children with CAH. They expressed a reluctance to fully trust the information

they received from the healthcare providers and to fully trust their ability to provide optimal care to their child.

**Normalcy:** Many parents reported the need for their children to be 'normal kids.' Parents want their sons and daughters' CAH well managed so they can play and learn and live like healthy children and grow up to be well-adjusted healthy adults. They want their daughters to have normal looking genitals, to be able to function sexually and to be able to have children.

We asked parents what they would recommend to parents of a newly diagnosed child with CAH. The following recommendations were consistently given:

**1)** Get all of the information you can from a variety of resources including health care providers, other parents, CARES, books and websites. Parents reported that speaking with other parents of children with CAH and resources at CARES were particularly helpful to them.

**2)** Parents should trust their intuition when it comes to their child. The parent knows the child best and can sense when something isn't right. 'Follow that intuition and be assertive with healthcare providers and don't stop until you are satisfied' said many of our interviewees.


**3)** Do not blindly trust healthcare providers and assume they are doing what they are supposed to be doing. Doctors make mistakes so it's important to be informed and aware of what they are doing when providing medical care.

**4)** Find the most knowledgeable and experienced endocrinologists and urologists and make sure you feel comfortable with those healthcare providers.

### Implications of findings

CAH is a rare condition and the number of physicians and other healthcare providers experienced with caring for CAH is limited. Many children and their families will live in areas of the country where a local expert

does not reside and some will not have easy geographic or financial access to such experts. Initially, decisions are made under very emotional and stressful conditions. The need to think of decision making as an ongoing event is especially relevant to the CAH situation because children with CAH need continuous medical care. It is therefore imperative that patients (and children as they get older) become informed decision makers. One way to help is to develop educational tools and decision aids that can help parents and their healthcare providers systematically think through all the issues around treatment that are very important. These issues must be inclusive of the child and family's quality of life both immediately and in the long-term. We hope to continue our work and develop such tools.

We thank the participants who so generously shared their very personal experiences and valuable time with us and the CARES Foundation for creating this important opportunity. 

### Parent Recommendations for those with Newly Diagnosed Children

- ✓ Get all of the information you can from a variety of resources.
- ✓ Parents should trust their intuition and be assertive with healthcare providers.
- ✓ Do not blindly trust healthcare providers.
- ✓ Find the most knowledgeable and experienced physicians and make sure you are comfortable with them.

*Hair loss* continued from page 1

following are some hair care procedures and ways to improve scalp hair for everyone. Many of these listed below have been modified from the book by Philip Kingsley (*Hair*: Aurum Press, 2003).

## Hair Care 101:

*It is vital that the hair be shampooed daily and rinsed fully.*

*Conditioning the hair removes tangles, with particular attention to the ends of the hair.*

*Avoid insufficient rinsing, and minimize tangling with a wide-toothed comb.*

*Use of a brush with sharp bristles should be avoided. Smooth combs are preferable.*

*Undefined stresses, hormonal medications (androgenic-like oral contraceptives and a number of other medications) possibly may be associated with hair loss.*

*Blow drying should be done with the dryer about 6 inches away. As the hair starts to dry, reduce the heat gradually. It is necessary to avoid blow drying hair from damp to dry to avoid hair damage, brittleness and split ends.*

*Rollers have to be used carefully, and not tightly. Pins and clips are also to be used cautiously, and never on while sleeping.*

*If elastic bands and barrettes are used at all, they must not be tight. They may cause traction alopecia. Similarly, pulling the hair too tightly from the forehead may also lead to severe hair breakage.*

*A habit of compulsively touching hair and pulling it (trichotillomania) should be seriously addressed.*

## Medical Treatments of Androgenetic Alopecia

- **Oral contraceptives (OCP) in combination with spironolactone**
- **Diane-35 (containing cyproterone acetate and ethinyl estradiol)**
- **OCP in combination with a 5-alpha reductase inhibitor**
- **OCP with flutamide**
- **Multiple drug therapy**
- **Minoxidil**

The medical management of androgenetic alopecia consists of a number of options: Oral contraceptives (OCP) in combination with spironolactone; Diane-35 (containing cyproterone acetate and ethinyl estradiol); OCP in combination with a 5-alpha reductase inhibitor; OCP with flutamide; Multiple drug therapy; and Minoxidil.

Unlike acne and hirsutism, medically controlling hair loss is much more difficult to treat. The listed drugs and options are more successful in slowing the progression of androgenic alopecia than actually reversing it. In CAH, controlling the overproduction of male hormones and stabilizing the disease is an essential first step prior to the use of these drugs.

### Oral contraceptives (OCP) in combination with spironolactone

The most commonly used treatment is spironolactone in combination with OCP. Only those OCP with low androgenic potential should be used. Monotherapy with spironolactone alone, or OCP alone, is of little value in arresting alopecia and the use of spironolactone may be associated with fetal abnormalities in the genital development of a male fetus. **It is important to realize that antiandrogens should be stopped at least 4-6 months prior to attempting to become pregnant.**

Spironolactone is a diuretic that has been in use for a long time and found to have anti-androgenic effects. It works by blocking entry of the active metabolite of testosterone, namely

dihydrotestosterone (DHT), into the hair follicle. It has only a minimal effect on the hormone production of androgens and therefore the use of spironolactone with an OCP is indicated. The latter suppresses ovarian stimulation by pituitary hormones which stimulate ovarian androgen production and also have a direct effect on androgen synthesis in the ovaries and to some extent in the adrenal glands.

Some investigators also claim that spironolactone may increase insulin resistance, which is often seen in CAH. For maximal effects on alopecia the dosage should be 150-200 mg daily, in divided doses. A gradual dosage incremental program should be instituted.

The most commonly encountered side effect of spironolactone is dizziness on getting up quickly or suddenly bending over. Its diuretic effect also usually makes one urinate frequently, and in hot weather increased water consumption with an increased salt intake is indicated. A rare side effect is a possible increase in serum potassium which should be monitored at three to four month intervals. **Women with salt-wasting CAH should be advised to have their physician check electrolytes regularly and watch closely for symptoms of dizziness and low blood pressure.**

An effect on slowing the progression of alopecia may be seen in five to seven months. This treatment program is frequently helpful and the most used by endocrinologists in the treatment of alopecia, as well as



continued from page 6

hirsutism and stubbornly resistant cystic acne.

The combined use of any antiandrogen with OCP has the advantage of reducing the effect of hair shedding by a number of actions of the OCP: 1) they suppress the pituitary hormones, namely luteinizing hormone (LH) which stimulates the ovary to produce androgens; 2) they increase a substance called sex hormone binding globulin which allows more binding of testosterone to this protein, and 3) they further prevent biochemical effects of conversion of testosterone to DHT. The use of OCP alone has only a minimal effect in reducing alopecia. Other benefits of OCP, however, are the reduction of the incidence of uterine and ovarian cancer.

### Diane-35 (containing cyproterone acetate and ethinyl estradiol)

Although it is **not approved by the U.S. Food and Drug Administration (FDA) and not available within the US**, cyproterone acetate (CPA) is a potent progestin and antiandrogen which is effective when combined with an estrogen such as ethinyl estradiol in the form of Diane-35. It may be obtained in Canada and many other countries including those of Europe. There are conflicting and no conclusive data as yet indicating Diane-35 as a more effective antiandrogen treatment when compared to the combined use of OCP and spironolactone.

CPA blocks the binding of the active androgen DHT at the receptor site of the hair follicle and has other hormonal effects in the synthesis of androgens in the ovary and some effect on the release of LH by the pituitary gland. Some common side effects of Diane-35 include light-headedness, fluid retention, weight gain and rare reports of adrenal insufficiency.

### OCP in combination with a 5-alpha reductase inhibitor

The effect of the non-hormonal “5-

alpha reductase inhibitors” is the reduction of the formation of DHT from testosterone, and this inhibits the interaction of DHT and the receptors of hair follicles, which in the scalp may reduce the intensity of shedding hair. Many clinicians have expressed the view that there are no major differences of the clinical effects of reducing excessive hair growth in women (hirsutism) or reducing alopecia in women with this therapy when compared to spironolactone. The earliest agent used in this category for alopecia as well as hirsutism has been finasteride (Proscar), a commonly used drug in men with prostate enlargement. There are no drug company statements suggesting its use in women, but neither is there for spironolactone.

The earliest effects of this combination may be noted in six months, and the side effects usually are minimal with no change in the menstrual cycles. It is also essential for 5-alpha reductase inhibitors this drug to be combined with OCP to prevent conception, because the effect on fetal genital development may be significant. **In fact, it should again be stressed that any woman considering fertility should stop the drug for at least four to six months prior to trying to have a family.** Monotherapy with finasteride alone may be an option for some postmenopausal women with alopecia.

Finasteride is available in a 1.0 mg dosage form in men with rapid hair loss (Propecia), while 5mg daily is recommended for women with androgenic alopecia. Several early studies suggest that another 5-alpha reductase inhibitor, dutasteride (Avodart), may be tried in women whose hair loss is not controlled with finasteride. The dosage is 1 capsule of 0.5 mg daily. Definitive studies of the effect of the drug as an antiandrogen for androgenic alopecia should be forthcoming.

### OCP with flutamide

Flutamide (Eulexin) is a nonsteroidal antiandrogen that is a pure antiandrogen in that it inhibits male hormonal effects in all tissues responsive to testosterone by inhibiting the effect of binding to the nucleus of these tissues. It has an earlier onset of action than all other antiandrogens, usually within three months after start of treatment. The dosage may be effective in most patients taking a 125 mg capsule twice a day. Its side effects include abdominal distress, diarrhea, and rarely fatal liver toxicity. Liver profiles in the blood must be tested frequently in those receiving the drug. My personal view is to use it only in the most severe presentations of alopecia and those with such great emotional stress that it interferes with their lifestyle.

### Multiple drug therapy

The use of several antiandrogens in combination with an OCP may be tried by experienced endocrinologists familiar with these drugs.

The regimen I consider the best is a combination of spironolactone and a 5-alpha reductase inhibitor together with OCP. There are few adequate reports regarding this form of treatment for androgenic alopecia.

### Minoxidil

The topical use of minoxidil (Rogaine), an over-the-counter preparation, may be considered in early forms of alopecia either as a solo treatment or in combination with some of the above treatment choices. It is also used frequently in women with various

continued on page 22

*Dr. Walter Futterweit is a Medical Advisor to CARES Foundation. He is also Clinical Professor of Medicine in the Division of Endocrinology at Mount Sinai School of Medicine, NY.*

# Blessings In Disguise

Tonia Drake

Two weeks after our oldest daughter's first birthday, the eight EPTs I took and the doctor's blood test said I was pregnant. I called the doctor right away. We worked out a plan, and my husband and I saw a genetic counselor and got the Amnio as soon as possible.

I think it was late summer or early fall of 1997 when we got the phone call from our genetic counselor. She happily announced "You're having a girl!" I smiled and wondered what was next because her voice didn't sound like she was done. "Tonia, I need to let you know the new baby does have salt-wasting CAH, just like Abby." The rest of the conversation was a blur. I left my office, promptly cried and asked my boss if I could go home.

When my husband got home from work with our one-year-old, he found a blubbering mess. I told him the news. We were both overcome with emotion. Not because we thought there was anything wrong with Abigail ("father's joy") but because we had been so overwhelmed with the care she took—the worrying, the doctor and ER visits, and the financial strain. How would we do it with two! After we put Abby to bed, we prayed and prayed—hard—together.

The months went by and finally Gabriella ("gift from God") was born. She was beautiful—a shining light! Things were tough at first. Many times we would be at the hospital with two children. But we noticed something as the girls got older: their bond with each other. They seemed at an early age and even at eight and ten to develop their own secret way of communicating and supporting each other.

In July of 1999, Ellie woke us at 4 am with a gurgling cry. Her temperature was 106.7, she was convulsing and threw up on herself.

Brett called our (volunteer) fire department and they told us it would be 10 to 15 minutes before they could get to us and could not administer any medications or IVs. I injected Ellie with Solu-Cortef and drove her to the hospital myself, knowing I could get there sooner.

Ellie was admitted immediately into the PICU and it was determined she had an ear infection that had spread to her blood stream (septic) and was resistant to everything but the strongest antibiotic available at that time—vancomycin. On her second day in the hospital, I was awakened by the nurse very early. She said, "Your husband is in the ER..." I was trying to wake and

figure out why he would be there! Then she finished her sentence, "Abby has a very high fever and is vomiting profusely. I can stay here with Ellie if you want to go down." I went down and there was my Abby, looking just like Ellie did two days before. They made an exception in the PICU and put the two together in the room.

It turned out, after testing everything under the sun, that there was nothing diagnostically wrong with Abby! The endocrinologist had a theory that the stress of her sister being in the hospital and their connection with each other, sent Abby into a full blown crisis. I was glad to hear she was okay.

*continued on page 22*

## Letters:

### NEW Medical Alert Card Really Works

I wanted to thank you so much for the Medical Alert card for adrenal insufficiency and steroid dependence. I went to the hospital yesterday due to projectile vomiting and high fever. I was able to advocate for myself with the card. Without the card I would have waited for 3 hours to get into the ER. I got in right away. Thank you so very much again. The organization can truly save lives.

— Robyn  
(Maryland)



# Paving the Way...One Step at a Time

Constantino Tejeda

“You will know them by their fruit,” is a biblical phrase that throughout time gains more significance; and I am certain that it forms part of the philosophy and mission of the CARES Foundation. It is worthy to admire and recognize the role that this foundation has played for children affected by CAH. Though CARES Foundation hasn’t been established for very long, without a doubt, they have earned an honorable reputation within the community. Their immeasurable impact on affected children and their parents has exceeded the boundaries of their mission and their helping hand has extended throughout the globe.

Now is a good time to make mention of the magnificent conference that CARES hosted on November 12, 2006 in New Jersey. This event surpassed my expectations of CARES and their ability to organize an international conference of this level. In attendance, were the medical experts within the fields of endocrinology, gynecology, and psychology. Each speaker displayed skillfulness in their specialty, and it was amazing to see the enthusiasm of each parent to learn more about CAH and give their children better care. We were fortunate to instantly reap the benefits of this conference; within three days after the conference our little son, Alan, had an adrenal crisis and was on the verge of death. Fortunately, my wife who also attended the conference learned the steps to take in case of emergency. Our

eternal gratitude goes to the CARES Foundation – through this conference, their constant guidance, research, and services, we today have our beloved Alan Scott Tejeda back home safe and sound.

## **Haciendo Camino Al Andar**

Constantino Tejeda

“Por sus frutos los conoceréis,” es una frase bíblica que con el paso de los



**“...through this conference, their constant guidance, research, and services, we today have our beloved Alan Scott Tejeda back home safe and sound.”**

años adquiere más notoriedad. Y tengo la certeza que forma parte de la filosofía y misión de CARES Foundation. Es digno de admirar y reconocer el papel que esta jugando esta fundación dentro de la comunidad de los niños afectados de CAH. Sin lugar a discusión CARES Foundation se ha ganado un sitio preponderante en la sociedad porque a pesar de tener poco tiempo a llevado a cabo una labor

*amplia y grande a favor de los niños afectado con CAH y sus padres; y porque su misión a tras pasado fronteras y su brazo humanitaria se ha extendido a una gran parte del planeta.*

*Oportuno es el momento para hacer mención de la majestuosa actividad que llevo a cabo la fundación el día 12 de Nov. 2006, en el estado de NJ. Donde se dieron citas las grandes autoridades medicas dentro del campo*

*de la medicina endocrinológica, ginecología, y psicológica.*

*En este evento se puso de manifiesto la capacidad de la fundación y de si material humano para organizar un certamen internacional de esta magnitud; también la pericia exhibida en el manejo de los temas de cada conferencista; y el entusiasmo de cada padre de saber mas sobre CAH para darle un mejor cuidado a sus hijos. Para fortuna nuestra los resultados de la actividad surtieron sus efectos al instante; pues a los 3 días después de la actividad mi pequeño hijo, Alan, presento una crisis adrenal y se vio al borde de la muerte. Gracias a la destreza de mi esposa que estuvo en la conferencia y*

*aprendió como proceder en caso de emergencia. Nuestra eterna gratitud a CARES Foundation; pues por medio de esta actividad y el trabajo constate de orientación e investigación que están propiciando por diferente medios; hoy tenemos a nuestros querido Alan Scott Tejeda de regreso a casa sano y salvo.*

*\*\*Originally in Spanish. Translated to English by Mariel Vargas.*

# 2006 CARES CAH Conference, November 12, 2006

CARES 6th CAH Conference, “Bringing the CAH Community Together,” was held on November 12, 2006. The largest and most comprehensive gathering of its kind, the 2006 conference featured several new programs and was met with an overwhelmingly positive response by participants and speakers.

“When I walked into the cafeteria and saw close to 300 people affected by CAH sitting together, laughing, sharing, and making friends, my heart was filled with pride and happiness,” said Kelly Leight, Executive Director.

Our Spanish-speaking families were especially grateful for the translation services. Many understood CAH for the first time because it was presented in their native language. In addition to translators for each plenary session, Dr. Alejandro Diaz spent over two hours with these families, answering all of their questions.

Other new programs included in-depth sessions on growth, weight management for CAH and emergency care. Our one-on-one Solu-Cortef injection and sick-day trainings made a significant impact on our families, as you read about on page 9. Families were thrilled to be the first to receive our new Medical Alert cards. We were very fortunate to have “dummy” Act-O-Vials (filled only with saline), donated by our friends at Pfizer, to make the trainings as real as possible. The nurses from NIH, Saint Barnabas Medical Center and Children’s Hospital Los Angeles were fantastic teachers.

Of course, all of this would not have been possible without the beautiful, very large space donated by Seton Hall University. ♥





# Seton Hall University, South Orange, New Jersey



**Standing Room Only:** Most break-out sessions, like the "Adult Care" session pictured above, were completely filled as participants feverishly took notes from some of the world's leading CAH experts.



**Playtime:** Alyssa Leight and Catherine Peterson help out in the Children's Recreation Room. CARES was incredibly lucky to have help from numerous volunteers. Many thanks to everyone who pitched in!

## 2006 CAH Conference Sponsors

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



**ETHICON, INC.**  
a Johnson & Johnson company

*Lilly*



*The Fairhurst Family*



**Genentech**  
IN BUSINESS FOR LIFE

*Patty Genovay*





## GO Buckeyes! Necklaces Score

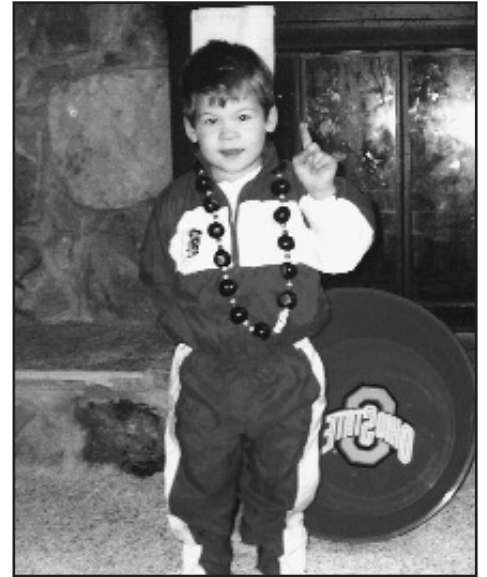
Lisa Ward, *parent*

My son Zack is a 3-year-old with Salt-Wasting CAH. We live in Columbus, Ohio—home of the Ohio State Buckeyes, and we are huge OSU fans, as is the entire state. I have been told that Ohio State fans are some of the most devoted in the country. Because of this, I decided to incorporate this enthusiasm into a way to raise money for CARES.

We have a few buckeye trees in our backyard and every fall the kids and I collect the buckeyes to make “Buckeye Necklaces.”

Every Friday during football season, most people dress in OSU colors to show their support. One of the most unique ways to show your pride is

by wearing Buckeye necklaces adorned with Scarlet and Gray beads. I decided to capitalize on this phenomenon by selling necklaces for \$10 and key chains for \$5 at the doctor’s office where I work. By doing something as simple as hanging them in the check-in window with a sign telling about CARES, I was able to raise \$1,000! I was amazed to see such an outpouring of support. Not only was I able to raise the money, but I also educated people about CAH and CARES. I really believe that I could have raised even more money, but I ran out of buckeyes! I already have plans to repeat this effort next year, scouting out more buckeye trees in neighbors’ yards. Hopefully we will surpass our contribution next year! ❤️



Zach Ward, 3, roots for his favorite team, the Ohio State Buckeyes, while wearing his “Buckeye Necklace.”

## Artistic Endeavor: Cards for CARES

Lorraine Penoro, *parent*

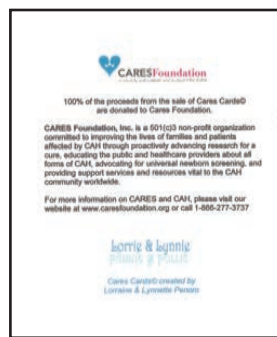
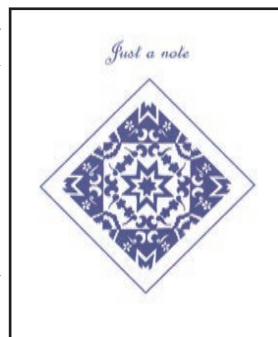
When we heard about the tragic fire that struck CARES headquarters, we knew that we had to do something to help. My daughter, Lynnette, and I tapped into our artistic talents and came up with the idea to create note cards with our original artwork, print out the cards with a notation about CARES Foundation on the back, package the cards in sets of 4, sell the packages for \$5.00 each, and donate 100% of the proceeds to CARES. When we contacted Kelly about the idea, she was very excited and we went ahead with our fundraising project.

In the beginning, we made about \$50.00- \$75.00 between sales and some cash donations, but then we decided to expand our fundraiser and present the idea to the Art Honor Society of Lynnette’s high school, H. Frank Carey H.S. of Franklin Square, NY. We thought that if we could have art students create new designs, we could sell the note cards in school to teachers,

and to family and friends at home. The Principal of H. Frank Carey High School, Mr. Douglas Monaghan, was immediately in favor of this project and offered his full support. He told Lynnette, “Remember, the first four

Kaufman, the club advisor, had scanned to a CD ready for me to take home for printing. The next week, we had a packaging party complete with home-baked cookies, (I had to feed them!) and each student took home packages of cards to sell. We were able to sell some at the senior high winter concert, and at Matthew’s Cub Scout pack night.

So far, we have collected over \$500.00 in sales and cash contributions, and the students have told me that they want to do new designs for an in-school sale. They are a very talented and genuinely giving group of kids who are very enthusiastic about making this project a success, which just might become an ongoing fundraiser. I am so proud of Lynnette, and all of these fine young people for all of their efforts and I know that CARES Foundation will benefit greatly from this fundraiser. ❤️



letters in Carey are C-A-R-E.” The Art department, PTSA, Dad’s Club and teacher advisors were all on board and helped out in countless ways.

The students were excited to take on this project and decided they wanted to do some holiday designs before the winter holiday vacation. Within a week I had 8 original designs, which Mr. John

**DO NOT**

**START TRAINING**



**10K**

**NO WAY !!!**

# **No Sweat Run for a Cure**

## ***Spring 2007***

This is CARES' first "non-event" fundraiser. Rather than asking you to attend another event, we are providing a fun way to participate from the comfort of your home.

**Event committees forming now!**

We need help securing corporate sponsors, prize donations, feedback on the concept and people to encourage participation.

**Email [mariel@caresfoundation.org](mailto:mariel@caresfoundation.org) for more information.**

# We couldn't do it without YOU.

Jennifer Mann Rosenblatt

As the previous articles illustrate, fundraising is certainly a grassroots endeavor (or in the case of the Ward Family it included a new drill, help from the entire family and buckeyes). CARES is humbled by the support that we continue to receive from our members and families. These contributions are the backbone of our organization and truly come from the heart. CARES is thrilled to share with you the following snapshots of some fundraising efforts that are in the process of being planned or have

already yielded impressive results! We are grateful to have the opportunity to recognize these generous donors, and it is our hope that their dedication and creativity will serve as an inspiration.

### Buy a Chance for CARES

CARES member, Louise Fleming, along with husband, Joe are collaborating with the Raleigh, N.C. office of KPMG to conduct a raffle that they hope will raise significant funds for CARES. This raffle will feature 3 weekend get-away packages (which include fine dining, spa and golf) at

premier hotels in the Raleigh area. Tickets are \$10 each. Since you do not need to be present to win, CARES families and members are encouraged to contact Louise directly at [nutmeg0822@bellsouth.net](mailto:nutmeg0822@bellsouth.net) to purchase your raffle tickets before Labor Day 2007!

### Coins for CARES!

Thanks to John Krise for hosting a coin drive held at his Seattle, WA office.

### Happy 1st Birthday, Wyatt!

The Harper Family of Snoqualmie, WA celebrated their son, Wyatt's 1st

*continued on page 18*

## MICHIGAN

### CAH Family Education Day

Saturday, May 19, 2007  
2:30-5pm

Palmer Commons  
University of Michigan – Ann Arbor

Please join us for a fun and educational afternoon and an opportunity to meet other children and families who live with congenital adrenal hyperplasia.

There is no cost for this meeting; we do ask you to let us know if you are coming.

### RSVP

Rose Harlow  
(Michigan Newborn Screening Program)  
734-647-8938.

*More details will follow.*

Directions to Palmer Commons:  
<http://www.umich.edu/~info/maps.html>

*This program is supported by the  
Michigan Department of Public Health*

## SOUTH CAROLINA

### SC Support Group Family Fun Day

Saturday, May 19, 2007  
11am

The home of Kevin and Johnette Kinard  
1988 Mt. Pilgrim Church Road  
Prosperity, SC 29127

### Games, Food and Fun for All

RSVP 803-364-9945

**We hope to see you there!**

## COLORADO

### Supper Club

*Very informal. Very informational. Very Helpful.*

Denver/Boulder has a new Sunday Supper Club for parents of kids dealing with CAH. The group will meet the first Sunday of each month for dinner at restaurant in Denver at 6:00pm.

For more information, or to get on the contact list, please contact Anna Payne at [paynedomain@mac.com](mailto:paynedomain@mac.com).



## CARES US Support Groups

**ALABAMA**

Tonya Judson  
(205)991-8674  
tjudson@charter.net

**ALASKA**

Sarah Brown  
(205) 452-7772  
lrichardson@gci.net

**ARIZONA**

Caren West  
(602) 332-8615  
cjsilentheart@cox.net

**ARKANSAS**

Gail Blucker  
(501) 835-6679  
gailtcb@sbcglobal.net

**NORTHERN CALIFORNIA**

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

Tammy Hupp  
(916) 784-1813  
tammypoo@mac.com

**SOUTHERN CALIFORNIA**

Jami Abell-Patterson  
(818)906-8668  
jami@caresfoundation.org

Jennifer Cribbs  
(714)968-6794  
jcribbs@starbucks.com

Jennifer Cribbs  
(714)968-6794  
jcribbs@starbucks.com

**COLORADO**

Jennifer Butler  
(303) 690-7829  
jbutler70@hotmail.com

Julie Adams  
(303) 439-9816  
jcc4adams@aol.com

**CONNECTICUT/NEW ENGLAND**

Lynn Torony  
(203) 264-6898  
Ltorony@charter.net

**FLORIDA**

Patricia Tovar  
(727) 541-1683  
rubentovar.1@netzero.net

Sonya Matson  
(941) 722-7560  
sonya\_mickey@yahoo.com

**GEORGIA**

Heather Carpentieri-Sorrows  
(770) 655-3672  
hjacarpentieri1@msn.com

Michelle Corbitt  
(912) 473-2495  
ivy31543@yahoo.com

**IDAHO**

Amanda & Thomas Hopper  
(208) 882-6617  
Amandah59@yahoo.com

**ILLINOIS**

Wendy Herst  
(847) 945-2221  
mwherst@aol.com

**INDIANA**

Penny Barrett  
(317) 865-9320  
onecent721@yahoo.com

**IOWA**

Sandy Mostaert  
(319) 265-7768  
mozzdesigns@yahoo.com

**KENTUCKY**

Dana Rush  
(502) 499-0313  
prush04@peoplepc.com

Donna Burkhead  
(502) 338-1500  
donna Burkhead@charter.net

**KANSAS**

Tonia Kroll  
(913) 240-9318  
mommykroll@aol.com

**LOUISIANA**

Bonnie Blanke  
(985) 845-0667  
barn415@bellsouth.net

**MAINE**

Jeff & Jody Spear  
(207) 832-2006  
jrspear@adelphia.net

**MARYLAND (DC METRO AREA)**

Eden Santa Cruz  
(301) 801-0741  
Edeneig@aol.com

**MINNESOTA**

Karen Wilson  
(612) 671-3602  
K.E.Wilson@aexp.com

**MISSISSIPPI**

Gina Murray  
(601) 829-2409  
cgmurray@bellsouth.net

Susan Aycock  
(601) 833-8373  
shaycock822@aol.com

**MISSOURI**

Cassy Wanstreet  
(660) 339-7449  
wanstreet@cebridge.net

**NEBRASKA**

Paige Young  
(402) 813-2902  
pandkids@cox.net

**NEVADA**

Alesia & Jeremy Pinson  
(775) 813-5031  
Pinsona@juno.com

Julie Tacker  
(702) 399-8553  
julie110@cox.net

April Perrucci  
(702) 658-3997  
aprilandnickp@aol.com

**NEW JERSEY**

Michelle Cascarelli  
(732) 516-0916  
mcascarelli@yahoo.com

Vanessa Perez (Se Habla Español)  
(973) 485-1651  
vanesa.perez@att.net

**NEW MEXICO**

Betty Herrera  
(505) 647-8314  
bettyc54@msn.com

**NEW YORK (UPSTATE)**

Dona Miller-Cameron  
(716) 773-7372  
dm0070@yahoo.com

**NORTH CAROLINA**

Louise Fleming, R.N.  
(919) 363-6447  
nutmeg0822@bellsouth.net

Amanda Russo  
(910) 262-3757  
amandawilmington@aol.com

**OHIO**

Tonia Drake  
(614) 920-1584  
TDrake4475@aol.com

Lisa Phillips  
(440) 871-3659  
phillipsmandl@wowway.com

**OKLAHOMA**

Katherine Kirk, R.N.  
(405) 271-8001  
Katherine-kirk@ouhsc.edu

Margaret Williams  
(580) 965-5360  
maggieann02@hotmail.com

**OREGON**

Rossana Wong  
(503) 239-3707  
wongfam4@msn.com

**PENNSYLVANIA**

Lisa Stipetich (Pittsburgh Area)  
(412) 630-8287  
lisas@americanfastener.com

Joseph Thibodeaux (Pittsburgh Area)  
(412) 488-1694  
joetdeaux52@comcast.net

Jennifer McCleod (Allentown area)  
(610)530-1833  
sideshowjenn@gmail.com

**SOUTH CAROLINA**

Johnette & Kevin Kinard  
(803) 364-9945  
kevin1@backroads.net

**TENNESSEE**

Jane Ann Luttrell  
(865) 689-9493  
luttreltj@bellsouth.net

**TEXAS**

Lesly Stevens (Dallas)  
(817) 472-0253  
mwlastevens@prodigy.net

Meridith Taylor (Austin)  
(512) 349-9719  
Meri\_taylor@yahoo.com

Sandra Billings (Houston)  
(281) 861-6043  
billings6@sbcglobal.net

**UTAH**

Trisha Irving  
(801) 915-8950  
utahcah@yahoo.com

**VERMONT**

Terri Chaney  
(802) 440-8057  
terrichaney@adelphia.net

**VIRGINIA**

Alesha Pierson  
(757) 439-7977  
apierson@chubb.com

Cherry Lane  
(757) 416-9630  
CLane10407@aol.com

**WEST VIRGINIA**

Karen Bozarth  
(304) 252-5922  
3beez@charter.net

**WISCONSIN**

Lisa Jaskie  
(414) 645-0782  
lisa1273@msn.com

Laurel Meier  
(715) 341-9697  
Laurelmeier@charter.net

## International

**BRAZIL**

Isabel Wagner  
(552) 126-19-3423  
isabelsw@provide.com.br

**CANADA**

Alison Weatherall  
(905) 713-2304  
kiddo71@sympatico.ca

Tina Haslip  
(905) 465-0927  
tinahaslip@cogeco.ca

**CHILE**

Valeska Rojas  
(56) 998263754  
valerj33@hotmail.com

**COLOMBIA**

Diana Oesch  
(574) 334-7294  
dianaoesch@une.net.co

**ECUADOR**

Sandra Merizalde Paredes  
5939 982 8886  
smerizalde@andinanet.net

**FRANCE**

Nina McPherson-Quanton  
331 40 27 08 75  
nina@easynet.fr

**GREECE**

Siomos Kostas  
00306396739581  
kostasdis63@hotmail.com

**INDIA**

Matthew Jacob  
009 938781838  
00 91 484 23433  
jacobbhai@gmail.com

Sanjeev Sahni  
091-11-98-11774  
Sks\_ps@hotmail.com

**MEXICO**

Mayté Villaseñor  
5281-8367 5790  
maytevillaesear@yahoo.com

Sergio Varela  
52 01 662 260782  
varel@correobancomer.com

**URUGUAY**

Mariela Sessa  
598-2-6134176  
mariesessa@montevideo.com

Be a resource for families in  
your area.

If you would like more information on  
starting a support group,  
call 866- 227- 3737  
or email [erin@caresfoundation.org](mailto:erin@caresfoundation.org).

# Carrier Testing in Minors: *Ethical Issues*

Pascal Borry, Ph.D

Kris Dierickx, Ph.D

In a previous article in this newsletter (“The Basics of Genetic Testing for CAH and How it May Benefit You and Your Family” Vol 4 Issue 1 Winter 2004-2005) we could read that 90 to 95 percent of the cases of congenital adrenal hyperplasia (CAH) are due to 21-hydroxylase deficiency (made by the gene called CYP21A2). As a consequence, genetic testing for CAH can be used to confirm the diagnosis of CAH or to identify the mutations in a person who is suspected of having CAH. Detection of an affected person allows testing of other family members for the specific mutation(s) identified. This might allow relatives to know if they are carriers of the disease.

Carrier tests are intended for healthy people who have no symptoms of a disease, but who are known to be at high risk because of a family history of a genetic disorder. To be a carrier mostly doesn't affect the health of the carrier but could have consequences for the carrier's offspring. In the case of autosomal recessive disorders (such as CAH) each offspring of two carrier parents has a 50 percent risk of being a carrier, a 25 percent risk of developing the disorder and a 25 percent risk of being neither carrier nor affected by the disease.

Due to the familial character of genetic diseases, when a genetic disorder is diagnosed in a family, an immediate question usually asked is whether other family members are at risk. When the relatives in question are adults, they can decide for themselves whether or not to undergo a genetic test. Children at risk, however, are dependent on others, usually the parents, for decisions about (genetic

testing for) their health.

## Concerns

Because of concerns that carrier knowledge at young age may be harmful, carrier testing in minors remains controversial. It has been argued that it might harm a child's self-esteem or affect his/her self-concept, for example by reinforcing latent feelings of unworthiness. Knowing carrier status may also cause “survivor guilt” or heightened anxiety of a child. In addition, it has been argued that it might distort the family perception of a child, might lead to psychological maladjustment or stigmatization, and might lead to significant misunderstanding and consequently erroneous interpretation of the carrier status.

Concerns have also been raised about the potential for unfair genetic discrimination in the sphere of employment or when applying for life insurance or related products. There is however a considerable lack of empirical evidence about these potential harms. The Genetic Interest Group (GIG), a U.K.-based alliance of patient organisations which supports families affected by genetic disorders, for example considers that there are no reasons to be “...overly preoccupied with psychological considerations and the harm that knowledge of genetic disorders can cause within families. With little evidence, this seems to reflect more the fears of doctors that they will be held responsible for negative reactions, rather than the needs

of families.” For the GIG, the potential psychological impact for of carrier testing is then different than for predictive or presymptomatic genetic testing (in which persons at risk are tested if they will develop the disease). Although the GIG also affirms that “children should only be tested when of an age to be involved in the decision,” they consider that, “Parents have the right to make an informed choice about whether or not to have their children tested for carrier status.”

The position of the GIG contrasts with existing clinical guidelines (*See P. Borry et al. Carrier testing in minors: a systematic review of guidelines and position papers. Eur J Hum Gen 2006; 14 (2): 133-138*). The guidelines are in agreement that children preferably should not undergo carrier testing. The child's future autonomy tended to be the main ethical argument at stake. As carrier testing has the potential of affecting the future reproductive prospects of a child, the guidelines emphasised that the decision to test should be made by the child when he/she reaches reproductive age. This view is based on the basic ethical principle of informed consent, by which an individual can freely and voluntarily give, without external pressure, their consent to be tested after being informed of the benefits, risks, procedures, and other pertinent information relating to the carrier test. As carrier testing performed during childhood only affects the (reproductive) future of that child, the

*continued on next page*

**“Letting children decide about their reproductive health means respecting them as humans with their own individuality and responsibility.”**

continued from page 16

guidelines state that it is wiser to defer testing until the child is able to give proper informed consent than to acquiesce to the wishes of his parents or guardians to go forward with testing. The child's personal consent takes precedence over the wishes of third parties, including parents, either to carry out or to refuse genetic testing.

Knowledge of carrier status critically impacts future decisions concerning reproduction (e.g., carrier testing of partner, prenatal diagnosis, artificial insemination, pre-implantation genetic diagnosis, adoption, not to have children). Some guidelines also suggest that carrier testing performed during childhood also denies the child of confidentiality, a right he would receive if tested as an adult.

### Parental responsibility

Many parents feel responsible for the health of their children. (see P. Borry et al. *Attitudes towards carrier testing in minors: a systematic review. Gen Couns* 2005; 16, 341-352). It might therefore be difficult for many parents to accept delaying the carrier test. In a study by McConkie-Rosell (*Parental Attitudes Regarding Carrier Testing. Am J Med Gen* 1999; 82, 206-211) most parents of individuals with Fragile X syndrome were very concerned that their child(ren) knew the genetic risk before becoming sexually active and/or should be able to marry informed of their carrier status. They appear to want this information in order to help their children adjust to the knowledge of their carrier status. The majority of the parents indicated to be very or somewhat concerned about informing and raising the children with knowledge of their carrier status. A strong belief in a parental right to make the decision regarding carrier testing in their children was found. About half of the

parents felt that parents should have the right to decide when their child should be tested and informed of the result. Only 15 percent of the respondents were very concerned that making the decision to test their child would violate their child(ren)'s autonomy by depriving the child(ren) of the right not to know.

### Telling and testing

Parents have argued that carrier testing in childhood might help the child



to adapt to the knowledge of being a carrier slowly, while receiving the information later could be more shocking. However, the option to perform a carrier test in childhood does not take into account the alternate option to inform children about their genetic risk, without performing the carrier test in childhood.

On the one hand, this option takes into account the parental responsibility of communicating the genetic risk to their children and to support them in dealing with that risk. This underlines the familial character of genetic information and emphasizes that genetic risk information should be discussed openly in a family context. On the other hand, this also emphasizes the individual character of this information, the individual decision to undergo a genetic test, and the individual choice to share the result of the genetic test with others.

It is important that minors should

be able to participate in health decisions that affect their offspring. When an adolescent understands the information given to him/her and the implications of a positive and negative carrier test for him/her, his/her partner and eventual offspring, he/she can make a decision about performing a carrier test. It is also possible that adolescents prefer to postpone the carrier test, but it is important that minors are informed about their genetic risk.

Choices and actions around sex, marriage and reproduction belong to the most personal and intimate aspects of life. Because the goal of a carrier test is situated in reproductive decisions, it is important that children, when growing up to be adults, are able to request a test personally and keep full control over that information and share the information with who they want. This should, however,

not restrain parents to discuss the opportunity of carrier testing openly with their children later in life. The children should be aware of their genetic risk from a young age even though the actual testing is being postponed.

The fact that parents should delay carrier testing in order to reserve the opportunity for the child to choose among as many options as possible once he or she reaches adulthood, can be based on the fact that in caring about their children, these should be fully respected as care-receivers from their parents, but also as care-givers who are caring about their own offspring. Parents are expected take the future attentiveness, responsibility and competence of their children in caring about, taking care of and giving care to their own offspring serious. Letting children decide about their reproductive health means respecting them as humans with their own individuality

continued on page 18




Debate continued from page 17

and responsibility. Parents and genetic services are invited to be supportive so that individuals at risk are informed about their genetic risk and are able to make use of the opportunities of genetic testing if necessary. The presence of severe anxieties and uncertainties in parents about the possible carrier status might be an indication for further psychological support in order to address these emotions rather than a clear indication for testing.

**Carrier testing for CAH**

As carrier testing only affects the individual’s reproductive health, carrier testing of incompetent children for CAH should preferably not be performed. However, carrier testing in adolescents and young adults who have undergone adequate genetic counseling, who understand the information given to them and the implications of a positive and negative carrier test, and who can participate in a carrier test without external pressure can perform a carrier test. This gives the parents the

role to inform and support their children concerning their genetic risk, but lets minors participate in this decision.

The research for this contribution was supported by the Eurogentest Network of Excellence of the EU, FP6-512148. 

**Corresponding author:**

Pascal Borry  
K.U. Leuven Center for  
Biomedical Ethics and Law  
Kapucijnenvoer 35/3  
3000 Leuven  
Belgium  
[Pascal.Borry@med.kuleuven.be](mailto:Pascal.Borry@med.kuleuven.be)  
Tel. 00 32 16 33 69 51  
Fax 00 32 16 33 69 52

*Pascal Borry Ph.D., works in Ethical and Legal Unit of the research project Eurogentest ([www.eurogentest.org](http://www.eurogentest.org)) funded by the European Commission and is involved in the Erasmus Mundus Master of Bioethics ([www.masterbioethics.org](http://www.masterbioethics.org)).*

*Kris Dierickx, Ph.D. is associate professor and a postdoctoral fellow of the Belgian National Fund for Scientific Research (NFWO). His teaching and research focuses on the ethics of genetics and reproductive medicine.*

**Keep Reading** →  
Read the other side of this debate on page 19.  
For questions or comments please email [erin@caresfoundation.org](mailto:erin@caresfoundation.org) .

continued from page 14

Birthday by requesting in that, lieu of birthday gifts, friends and family make donations to CARES in honor of Wyatt’s milestone. Thank you to the Harpers for including CARES in your family’s celebration!

**Tickle the Ivories for CARES!**

John Rollo of Glen Ridge, NJ has graciously offered to sell a used, 5 ft. Cable & Sons Baby Grand piano. All proceeds from this sale will be donated to CARES. Please email Jennifer, [jennifer@caresfoundation.org](mailto:jennifer@caresfoundation.org), for more information and details.

**Shop for a Cause!**

Dan Hackett of Santa Rosa, CA sponsored CARES as a participating charity in the “Evening of Giving” at the Santa Rosa Plaza (a Simon Mall). Dan and his family sold tickets to a

special evening at the mall that featured exclusive discounts, food tastings, entertainment and door prizes. Thank you for including CARES in this fun event!

**Cook for CARES!**

Pampered Chef consultant, Bernice Sosa-Izquierdo is using her home-based business to fundraise for CARES. The fundraiser will be held from now until March 23, 2007. Please contact Bernice directly at: 646-483-8074 or [bsimom@gmail.com](mailto:bsimom@gmail.com) to learn how your Pampered Chef purchases can benefit CARES. You can visit [www.pamperedchef.biz/bsikitchen](http://www.pamperedchef.biz/bsikitchen) to view all products.


**Reduce...Reuse...Recycle...**

This is the motto of resale shop, The Red Geranium in Cleveland, Ohio.

Led by CARES grandmother, Virginia Phillips, The Red Geranium donated a nickel to CARES (and 2 other charities) whenever a customer declined to take a bag for their purchase.

**You Can Make a Difference**

Our members continue to amaze us with their creativity and passion. No matter the size of the endeavor, your hard work and dedication are helping us to make a larger impact on behalf of the CAH community.

If you are interested in conducting a fundraiser for CARES, please feel free to ask us for help. For more information, please email Jennifer [jennifer@caresfoundation.org](mailto:jennifer@caresfoundation.org). 

# Carrier Testing in Minors: *Decision-making within a Familial Context*

Sharon F. Terry, M.A.

Issues in genetic testing are all about ‘family’, since genetics reveals information about the whole family. As genetic information is integrated into family’s

lives, there are many decision-making points. Understanding carrier status, particularly for children, must include psychosocial, familial and community benefits. Decisions must reflect a broad understanding of health and should take place in a family-centered, community-based, discernment process. This article addresses carrier testing of children from the family’s point of view.

Many decisions affecting health are made using something that is sometimes called the “medical model.” In the medical model, healthcare providers make definitive judgments about diagnosis and treatment. It is expected that families will make decisions based largely on the medical community’s assessment. Families who spend a great deal of time seeing specialists, making many decisions about health, tend to move away from a

**“Decisions about carrier testing in children need to be made by evaluating not only the benefit to the child being tested, but to the family as a whole.”**

narrowly defined “medical model.” For these families, health is more than the presence or absence of a disease or diagnosis. In fact, families who have experienced disease have a very different sense of disease, sometimes embracing it as a ‘normal’ part of life .

When making decisions, families do not shy away from considering all aspects of a child’s life in the present and in the future. Families do not make decisions based solely on whether or not medical treatment is available or necessary. There are other benefits they consider, and since there is no “medical treatment” or even perhaps a medical reason to know, carrier testing stretches the medical paradigm. Moreover, there seems to be a double standard: individuals and families may decline carrier and other types of genetic testing primarily because of psychosocial concerns such as fear of discrimination

or stigmatization rather than lack of medical benefit, but the decision to choose carrier testing for children based on psychosocial benefits is not equally regarded.

While medical benefits can be easily identified by health professionals, psychosocial benefits may be more obvious and recognizable by the family. Therefore, if psychosocial benefits are given the same weight as medical benefits, then the opinion of family members should be equally considered during the decision-making process. The unique position of parents to make these decisions should not be usurped or disregarded by healthcare providers. Decisions about carrier testing in children need to be made by evaluating not only the benefit to the child being tested, but to the family as a whole. There are differences between the medical community’s value system and the family’s, and this can result in a dilemma.

Guidelines for testing in general consider the bioethical principles of autonomy, beneficence, and non-maleficence. Children do not have complete autonomy and their parents often ultimately decide whether the benefits and risks are acceptable on their behalf. Families generally do not use these principles in their decision-making process, nor are they measuring risks and benefits by psychological metrics. Instead, families think strengthening relationships is more important than following rules or



Debate continued from page 19

principles. In addition, autonomy is a western value, and individuals who share genetic mutations experience a “kinship of affliction” and tend to value family and even the ‘disease’ community above the individual’s right to make decisions about testing.

When the medical community makes a decision about an issue such as carrier testing in children, they do so after considering ‘harm.’ It is probable that families, particularly those who have experienced genetic diseases, define harm differently after their experience with disease. Furthermore, the assumption that a positive carrier status will lead to devastating psychosocial consequences for the child are not supported by testimony from families.

It is sometimes said that parents may confuse the best interests of the child with their own interests. But when parents make decisions within the family framework, the parents are placing a higher value on the child, nurturing him or her in the present. If the benefits of carrier testing will not be substantially decreased by postponing testing until the child can comprehend some of the benefits or risks of testing, delay of carrier testing should be strongly considered in order to include the child in the decision-making process. Sharing of information

“There are differences between the medical community’s value system and the family’s, and this can result in a dilemma.”

between families and health professionals is important as families make decisions that affect the future of their children.

It is thought that one of the major benefits of genetics is the ability to someday prevent disease. Medicine is expected to become tailored to the individual and allow early prevention and intervention. It may also be true at some point that being a carrier of a disease might have medical or other implications. Understanding carrier status can give individuals information needed to make reproductive decisions, and although children do not need reproductive information, there are

some families who believe that a child should grow up knowing their carrier status. Certainly, families who have children with genetic diseases have seen those diseases integrated into their lives with perhaps less trauma than those who learn about it more suddenly later in life.

Carrier testing of children is not a decision that is lightly made. To help limit controversial or unethical uses of carrier testing of children, many things need to be considered including maturity level of the child, presence of a strong family support system, and access to other support services. ❤️

*Sharon Terry is President and CEO of the Genetic Alliance, a coalition of over 600 disease specific advocacy organizations working to increase capacity in advocacy organizations and to leverage the voices of the millions of individuals and families affected by genetic conditions. She is also the founding Executive Director of PXE International, a research advocacy organization for the genetic condition pseudoxanthoma elasticum (PXE).*

## Use GoodSearch and Support CARES

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

GoodSearch.com is a search engine that donates half its revenue, about a penny per search, to the charities its users designate. GoodSearch is powered by Yahoo! and can be used like any search engine.

**Visit [GoodSearch.com](http://www.goodsearch.com) and designate CARES as your charity of choice.**

Whenever you search the Internet, use GoodSearch.com or, even better, make GoodSearch.com your homepage and **download the GoodSearch toolbar at <http://www.goodsearch.com/toolbar/>.**

**Thank you!**



# Update on Newborn Screening

Gretchen Alger Lin

## All Eyes on Arkansas, West Virginia and Kansas

Arkansas is on the path to expanded newborn screening! Thanks to the continued efforts of CARES member Gail Blucker in Arkansas, and through collaboration with the Arkansas Department of Health, March of Dimes, Easter Seals and other stakeholders, the state is moving slowly toward the passage of mandated universal, comprehensive screening. It is expected that Arkansas will join the list of states screening for CAH in 2007.

On January 26, 2007, legislation was introduced in both West Virginia and Kansas to expand newborn screening. West Virginia CARES member Gretchen Murphy - in conjunction with the West Virginia Chapter of the March of Dimes and PerkinElmer - is working hard to make newborn screening a priority in her state. Also, Kansas CARES member Tonia Kroll is lending her voice to the cause in Kansas.

Now, more than ever, we need your support in these states! Please add your voice to ours by contacting legislators to ensure their support of expanded newborn screening by visiting the websites below:

Arkansas - [www.arkleg.state.ar.us](http://www.arkleg.state.ar.us)

West Virginia - [www.legis.state.wv.us](http://www.legis.state.wv.us)

Kansas - [www.kslegislature.org/legsrv-legisportal/redistricting.do](http://www.kslegislature.org/legsrv-legisportal/redistricting.do)

## New Jersey Leading the Way

On October 23, 2006, landmark legislation - co-written by Executive Director Kelly Leight - was introduced in New Jersey to further expand newborn screening and codify the process by which the lives of hundreds of children are saved each year. The 2006 New Jersey Newborn Screening Act is one of the most important pieces of legislation ever to be introduced as not only does it expand newborn screening by statute but also mandates appropriate follow-up, treatment, education, and an annual review of all aspects of the screening program by an advisory committee.

## Canada!

Ontario, Canada, is set to go live with CAH newborn screening in January 2007!

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

## In the News . . .

### Stem Cell Research

On January 11, 2007, the House of Representatives passed H.R. 3, the Stem Cell Research Enhancement Act, by a vote of 253-174. The Senate is expected to take up its version of the Stem Cell Research Enhancement Act (S. 5) within the next few weeks.

### Show your support of GINA!

The Genetic Information Nondiscrimination Act (GINA) will benefit every American and directly improve the lives of those concerned with the misuse of their genetic information in health insurance and employment decisions. As an autosomal recessive, inherited disease the most effective route to diagnosis and successful management of CAH is through genetic testing. Moreover, diagnosis implies the existence of genetic information. The current lack of federal legal protections restricts individuals' access to health information, clinical trials, targeted drugs and advanced treatments. In order to fully promote personalized medicine and the use of genetic information in healthcare, we must pass GINA legislation. Ask your representative and senators to cosponsor GINA!

Learn how to contact your representative:

[www.senate.gov/senators/senator\\_by\\_state.cfm](http://www.senate.gov/senators/senator_by_state.cfm)

Find Senator information at

<http://www.house.gov/writerep/>

## Fund Research While You Shop

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

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

**Shop, Shop, Shop!**

*Hair loss continued from page 7*

degrees of alopecia. In some women there may be a mild degree of hair regrowth. The patient using Minoxidil should be careful to apply it carefully so as not to allow any drops to drip to the face which may lead to hirsutism of the affected areas.

### Conclusions

Although a number of treatments are effective in arresting androgenic alopecia, they are not FDA approved for use in women with alopecia, nor in hirsutism or acne. It requires careful study to exclude causes that may worsen its presence and a thorough evaluation by your physician followed by an endocrinologist is an important first step. Future studies will hopefully advance the introduction of new formulations which will benefit the woman with alopecia. 💙

*Blessings continued from page 8*

They eventually downgraded the girls and kept them together in a room (even though Abby was fine) and they were discharged together. It did wonders for Ellie's well being.

This is one of many similar stories, but the one that stands out in my mind the most. When the girls get blood draws, they hold hands and do rock, scissors, paper to see who will go first. When one is sick, the other takes a quiet place to pray. When one needs an injection, the other is getting a popsicle and holding her hand with encouraging words.

I look back to when Brett and I thought we couldn't do it, and realize God gave us the gift of two because He knew not only could we but they could. My friends ask me all the time if they fight and drive me nuts. For the most part, they don't. They continue to have a bond that is unexplainable to most.

And I realize that when I was questioning, I was given a gift and a blessing in disguise! 💙

## CAH CHAT GROUPS

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

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

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

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

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

## The Use of Antiemetics to Prevent Dehydration in CAH

*contributors*

Henry Anhalt, D.O.  
Mitchell Geffner, M.D.  
Phyllis Speiser, M.D.  
Maria I. New, M.D.

Recently, CARES has received several inquiries from families about the use of antiemetics (drugs effective against vomiting and nausea) by patients with CAH. With no published reports either recommending or discouraging their use in this population, we posed the question to our Medical Advisory Board.

There are many different types of antiemetics. Some you might be familiar with are: Zofran (Ondansetron), Tigan (Trimethobenzamide), and Compazine (Prochlorperazine).

According to our advisors, while these medications are often effective in treating nausea and vomiting, physicians should be very hesitant to prescribe them in cases of CAH. Using

these medications may mask the need for stress dosing or Solu-Cortef injection and delay a visit to the emergency room for intravenous rehydration, which is usually required in a patient with salt-wasting CAH. Additionally, nausea and vomiting can be symptoms of an adrenal crisis which, if covered-up, could lead to a false sense of security and the overlooking of dehydration and shock.

Acutely ill patients with CAH must always be given a stress dose of steroids (or Solu-Cortef injection when necessary) and seek medical attention immediately. 💙

# CAH Studies



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

## CAH Volunteers Needed for Exercise Study

### *Catecholamine Reserve and Exercise Tolerance in Subjects with Congenital Adrenal Hyperplasia and in Healthy Controls*

Principal Investigator: Dr. Deborah Merke M.D. Pediatric Endocrinologist and Director of Pediatric Services at the National Institutes of Health, Bethesda, Maryland

At the National Institutes of Health in Bethesda, Maryland, we are currently investigating the effects of high-intensity short term exercise over 20 minutes in patients with CAH. Since participation in the study does not confer immediate benefit to the volunteer, CAH participants will be paid \$200 upon completion of the study.

In order to participate in this study volunteers must:

- have the **non-classic** form of CAH (21-hydroxylase deficiency)
- be between 9-40 years old
- demonstrate good clinical control defined by: 17-OH-progesterone level between 100 and 1500 ng/dl, plasma renin activity within the normal reference range, growth rate within 2 standard deviations for age, and no new signs or symptoms of virilization in females.
- not be on other medications besides their CAH-related medications.

Travel is paid by the NIH.

**For more information or to volunteer, please contact Liza Green-Golan at the NIH at (301) 496-0610.**

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

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

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

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

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

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

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

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



## Thank You

This newsletter is generously sponsored by  
**PerkinElmer Life and Analytical Sciences**

PerkinElmer is a long-time supporter of CARES  
and the CAH community.

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

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



© 2007 CARES Foundation, Inc.

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



**Medical Alert**  
**Adrenal Insufficient**  
**&**  
**Steroid Dependent**

patient name / nombre

physician name / nombre del médico      phone number/número de teléfono

TIME IN A WAITING AREA NOT APPROPRIATE - MUST BE SEEN AND TREATED IMMEDIATELY

### NEW Medical Alert Cards

CARES has developed wallet-sized Medical Alert cards for CAH patients. The cards feature ER instructions, as well as areas for personal information and emergency contacts.

If you have not already received a card, or would like additional copies, please email Erin Anthony, [erin@caresfoundation.org](mailto:erin@caresfoundation.org) or call (toll free) 866-227-3737.

Remember, everyone with CAH should wear a medical ID and carry a medical alert card.