CARESFoundation
research, education and support for CAH

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The Potentials of Adrenal Cortical Stem Cells

by James Dunn, M.D., Ph.D.

The human body is made up of many different cell types that carry out specialized functions. Blood, for example, contains red blood cells that carry oxygen and white blood cells that fight infections. The origin of every cell within the body can be traced to the fertilized egg. Within days of fertilization, a single cell develops into a cluster of special embryonic cells called the inner cell mass. Under the right conditions, each of these cells can replicate and give rise to every cell type within the body. These powerful cells are called embryonic stem cells. Because of their origin from human embryos, embryonic stem cells have generated much controversy regarding their use for medical research.

Besides embryonic stem cells, there are also stem cells that persist in every organ of the body even after birth. For example, cells in the skin are being replaced constantly by new cells generated from skin stem cells. Such stem cells typically reside within a specific part of the organ and are present throughout life. Unlike embryonic stem cells, these organ-specific stem cells give rise to only a limited number of cell types within the body. These stem cells are usually small in numbers and are probably critical to the repair of the organ when injury occurs. There is now evidence that the adrenal cortex also contains stem cells that can give rise to the different types of specialized adrenal cortical cells.

The adrenal cortex is classically divided into three zones: the glomerulosa, the fasciculata, and the reticularis. The exact mechanism by which the three classic adrenal cortical zones arise is poorly understood. Each zone contains specialized adrenal cortical cells that possess specific enzymes that are used to produce different steroid hormones, including mineralocorticoids, glucocorticoids, and adrenal androgens, in response to physiologic demand.

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SAVE THE DATE!

Please note date change!

Saturday, October 22, 2005*

Fifth Annual

CARES Foundation

CAH Conference

Riley Children's Hospital
Indianapolis, Indiana

*Due to circumstances beyond our control, the date was changed. We apologize for the inconvenience.

Every1CARES Luncheon Raises Awareness and Funds!

(From left to right) Alyssa Ackenheil, Sarah Venit, Jami Patterson, Senator Dede Alpert, Kelly & Alyssa Leight.

More pictures and full story on pages 3 & 14

A Message from the

Executive Director:

Dear Friends,

Happy Spring!
Research, Research, Research!

Research is in the air! Our theme for this upcoming year is research and ways that we as a community can support it. We were fortunate enough to have had a very successful fundraising and awareness luncheon in March. We hope that our golf outing in Indiana, scheduled for September 22nd, will be as successful, and that many of you will hold fundraisers in your own communities. These events have and will bring in funds for CAH research. We hope that as we continue to develop financial support for CAH research, we can make a meaningful difference for ourselves, our children and those with CAH yet to be born. If we do not do this, no one else will. This is our mandate, and we need your help to make this a reality.

Every1CARES Luncheon

On March 10th, we held the very first EVERY1CARES luncheon at the Skirball Cultural Center in Los Angeles. It was extremely successful and a wonderful day. The event raised over $100,000. On behalf of CARES and its board of trustees, I want to thank the amazing Jami Abell Patterson, her terrific, supportive family, our energetic luncheon committee, the CARES Foundation staff and the fabulous event planners, The Proper Image. The event could not have happened without all of your dedication and hard work. Thank you so much! I especially want to thank Jami, who conceived of this idea and made it a reality. Jami--you are the best and we are so very grateful. I also want to thank all of those who attended and supported the event, as well as our generous corporate sponsors including Quest Diagnostics, Pfizer, Inc., PerkinElmer Life Sciences, Pediatrix, Inc., Bio-Rad Laboratories, Creative Artists Agency, Rolex, Inc., Erno Lazlo, along with the California Department of Health Services, Genetic Disease Branch, to mention just a few. We also thank all of our private and foundation sponsors. For more details on the luncheon, see the article on the following page.

Grant to the National Institutes of Health for CAH Research

The Board of Trustees of CARES Foundation has awarded a grant for $62,500 for the National Institutes of Health to support CAH clinical research. While in the past we have given smaller grants, this is the first (we hope of many) larger grants for CAH research. Now that CARES is 3-1/2 years old, through the hard work of all of the CARES members, the board and advisors, we are at a point that we can make these kinds of larger grants. It is a real turning point for us, and is a tribute to the dedication of the CAH community. This is very exciting for me and I hope for all of you as well. Working together, we are making a difference. Onward we go.....

Warmly,

Kelly

This newsletter is published 3 times a year.

Los Angeles Every1CARES Fundraiser a Great Success!

by Jami Abell Patterson, Event Chair & CARES Board Member

Our first EVERY1CARES Luncheon, held at the elegant Skirball Cultural Center in Los Angeles, CA on March 10, 2005, was an amazing success! As the Event Chair, I was overwhelmed by the outpouring of support for the CARES Foundation. With over 330 guests in attendance at the Ahmanson Hall, we honored State Senator Dede Alpert for her leadership and support of the newborn screening expansion in California, which goes into effect this August.

Our event began with a wonderful boutique which included over 20 vendors who were selling everything from fine jewelry to children's pajamas. Additionally, we received donations of over 175 items for our silent auction! We exceeded all of our greatest expectations, with such items as a trip to Hawaii with airfare, signed celebrity memorabilia, beauty and fitness packages, tickets to a taping of ABC's "According to Jim" and a private screening at a Hollywood Studio!

As lunch was served, the guests viewed the premiere screening of the CARES video. The response to the video was overwhelming, leaving many luncheon attendees in tears and with a far better understanding of why we were all gathered together for these few hours. Following the video presentation, we were honored to have Dr. Mitchell Geffner speak about the virtues of CARES. He told a heartwarming story about a newborn diagnosed with CAH, yet sent home without treatment. The parents contacted Kelly, who in turn contacted Dr. Geffner. With the proper treatment, this infant is now happy, healthy, and attended the event with her parents!

Senator Dede Alpert was presented with the CARES Foundation's first "Caring Angel" award for everything she has done in California to expand Newborn Screening. Following this presentation, Senator Alpert participated in one of the highlights of our luncheon by drawing the winning raffle ticket for the Rolex watch, kindly donated by Rolex with the assistance of one our members, Gregory Kraff. By pre-selling tickets with the invitations as well as selling raffle tickets at the event, we raised close to $8,000 from the raffle alone.

I am proud to say that over $100,000 was raised from our event, and all funds received will go towards CAH research, education and support. The success of the event is credited to so many – those that assisted in working at the event; those that attended; those that could not attend, but expressed their generosity and support of CARES by buying space in our Tribute Journal or by sending in a donation. When I first began this journey, I was hoping to raise $20,000 in profit and was praying for the minimum guarantee of 200 guests. Never in my wildest dreams did I think it would be such a success.

I am grateful to the love and support of Kelly Leight who never doubted my ability to pull this one off. I need to acknowledge and express my sincere thanks to my wonderful Co-Chairs: Jody Furie, Gloria Gurvitz, Shirley Lipstone, Marie Mandel and Jill Spiwak. None of these mothers have a child with CAH, but are merely good, loyal friends who believed that this cause is worthwhile and from the very beginning, were there when I asked for help. They, along with our hard-working committee members, made the success of this event possible. Thank you all so much!

Top: Eric, Elizabeth & baby Maddie Hashibe with Kelly; Middle: Sarah Ackenheil speaks; Bottom: Barbara & David Abell with Jami.

Of course, I need to thank my husband Patrick, my parents Barbara and David L. Abell, who are double Caring Angels and proud sponsors of the CARES Foundation and the luncheon, and my children, Danielle, Sarah, Michael, Ariel and Shane. They are the fuel that keeps me going and the reason for everything. 💖

Medical Management of CAH: Transition from Childhood to Adulthood

by Phyllis W. Speiser, M.D.

As most readers of this newsletter know, congenital adrenal hyperplasia (CAH) is a group of inherited disorders caused by inborn errors in the natural production of essential steroid hormones by the adrenal glands. The most common form, (CAH-21), is a result of low or absent activity of the 21-hydroxylase enzyme. The classic type of CAH-21 is present in about 1 in 10,000 to 1 in 15,000 live births worldwide, and about 75% of these patients suffer from salt-wasting and potentially deadly adrenal insufficiency. The other 25% of classic CAH-21 patients, called simple virilizers, have a non-salt-wasting form recognized by genital ambiguity in affected females, and by signs of androgen (male hormone) excess in later childhood. Nonclassic CAH-21 is the mild form of the disorder, detected in up to 1-3% of certain populations. The mild form of CAH-21 may be mistaken for early adrenarche (the increase in the production of androgens by the adrenal cortex that normally occurs at ages 8-9), a generally benign form of early pubic hair development in younger children. It may also be mistaken for polycystic ovarian syndrome with unwanted facial and body hair, irregular menstrual periods, and acne in young women. Nonclassic CAH is not life-threatening, and is not usually associated with genital ambiguity. This article will address issues relating to transition of CAH care from the pediatric age group to the adult.

Issues of concern to the pediatric endocrinologist, pediatrician, and young CAH family include making sure the diagnosis is correct, getting timely medical treatment, understanding when surgical intervention may be indicated, obtaining genetic counseling, and monitoring growth. With the transition from childhood to adolescence to adult care, growth and puberty remain important, but additional new concerns develop. Among such issues are psychosexual adaptation and reproduction.

Growth: Although children with CAH grow too rapidly, they may finish growth prematurely, so that adult height is shorter than average. Balancing medical treatment to maintain appropriate blood hormone levels is often complicated in CAH. Untreated or inadequately treated children grow rapidly and may not reach their height potential, but on the other hand, those treated with excessive medication doses suffer growth retardation. Since over-zealous medical treatment is a major cause of poor growth, it is important to treat CAH children with the lowest dose effective in maintaining adrenocortical hormones in a reasonable range. Optimal levels of these hormones will change with age and sex. Although the topic of growth inhibition by excessive treatment has been studied in infants and young children, there has been no careful study of whether less stringent control at puberty is effective in promoting maximal growth. There is still only very scant information about experimental treatment regimens and how they alter adult height among CAH patients. For instance, some children have been treated with low-dose conventional therapy in addition to two other oral drugs: an aromatase inhibitor and an androgen receptor blocker.

Another experimental treatment program involves standard medication combined with daily injections of growth hormone and monthly injections of Lupron, a gonadotropin-releasing hormone (GnRH) analog that suppresses puberty. Clearly, these experimental regimens are complex and would be difficult to manage for many patients. Thus, such regimens are not the current standard of care. The heights of individuals with nonclassic CAH are not significantly different from family heights, and thus they would seldom be candidates for extreme forms of height-enhancing treatment.

Puberty: Puberty may occur early, late, or on time in CAH patients. Since pubertal hormones contribute to growth and height attainment, it is important for puberty to be properly timed. A child whose hormone levels are poorly controlled may grow rapidly, then abruptly stop growing. Girls who are inadequately treated tend to have delayed onset of their menstrual periods. In some cases, additional treatment may be necessary to induce periods. Boys should be examined for normal pubertal development, as well. One particular concern in boys is the development of testicular masses due to poor hormonal control (see below, "Reproductive function in CAH males").

Psychosexual development: Girls with classic CAH, especially salt-wasters, express more typical male play in childhood, are more physically aggressive, and show less interest in infants and maternal nurturing compared with other girls.

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CARES Foundation Awards Research Grant to NIH

CARES Foundation is excited to announce that it has awarded the NIH Clinical Center a $62,500 research grant to support ongoing research in CAH. The researcher, Dr. Deborah Merke, will be the primary beneficiary of this grant. A pediatric endocrinologist at the National Institutes of Health (NIH) Clinical Center, she is completing studies of a new treatment regimen for children with classic CAH, comparing the conventional treatment (hydrocortisone and Florinef®) with an experimental regimen of reduced hydrocortisone, Florinef, flutamide (an anti-androgen), and letrozole (an estrogen inhibitor). The preliminary results are very promising.

For the past 11 years, Dr. Merke has specialized in deepening our understanding of congenital adrenal hyperplasia and testing the effectiveness of new CAH treatments in human volunteers. Historically, CAH has been considered a disease of the adrenal cortex, in which an enzyme deficiency leads to cortisol and aldosterone deficiency and androgen excess. Over the past few years, however, her lab has defined additional endocrine problems in CAH, such as epinephrine (adrenaline) deficiency and insulin resistance.

Dr. Merke has also further explored the psychological aspects of abnormalities of adrenal function. She discovered that CAH patients have smaller amygdala volumes. The amygdala is the part of the brain that regulates emotion. Over the next few years, she plans to: 1) further explore the clinical implications of reduced amygdala size and the psychological aspects of altered adrenal function; 2) expand our understanding of the clinical implications of epinephrine deficiency; and 3) test new treatments aimed at treating the insulin resistance characteristic of many patients with CAH. She also plans to start a new protocol using metformin, a diabetes drug, to treat CAH patients.

The grant from CARES will be used to hire a Nurse Practitioner to support this research. "Additional clinical personnel will be invaluable in allowing us to see more patients," says Dr. Merke. "Thanks to the generosity of CARES and its supporters, we will now be able to begin new testing programs and add new patients to our clinical trials."

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Yet despite this early atypical female behavior, most CAH girls express female-typical gender identity (that is, they are girls in their own minds) and show heterosexual orientation. Only in very rare cases have CAH women decided to undergo sex change, negating their sex of rearing. Gender-atypical behavior has not been observed in CAH males. Psychological support from professionals experienced in treating gender-related problems should be sought by families on an as-needed basis. This is particularly important at the time of diagnosis in severely affected females, and in adolescence, especially if genital surgery is performed. It is important to know that overt psychiatric disturbances are not common among CAH patients.

Reproductive function in CAH women: Reproductive problems for women with CAH usually become apparent in adolescent women who have poor hormonal control. Some patients have a clinical picture similar to polycystic ovarian syndrome, with multiple ovarian cysts, irregular menstrual bleeding, excess facial and body hair, and acne. Adrenalectomy may be considered when medical therapies are unsuccessful in achieving adrenal suppression in severely affected CAH women. In some cases, this may allow conception and fertility. Adrenalectomized patients must still be followed with medical treatment since they are prone to adrenal crisis and death if lifelong glucocorticoid treatment is interrupted.

Reproductive function in nonclassic CAH is more consistently normal. In fact, many cases of mild 21-hydroxylase deficiency, both male and female, go undiagnosed for lack of clinically important symptoms. At present, there is no test to predict which individuals affected with nonclassic CAH will progress and suffer adverse consequences of their hormonal imbalance.

Pregnancy outcome in women with CAH: A recent review found that up to about 80% of simple virilizers and 60% of salt-wasters can bear children. French investigators found that about 50% of women affected with nonclassic CAH became pregnant before the diagnosis of mild 21-hydroxylase deficiency was made, without receiving any specific treatment. Among the other 50%, those who desired pregnancy conceived during hydrocortisone treatment; and only one in twenty women required

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Treating Hirsutism Permanently With Electrolysis

by Stephanie Fracassa, C.P.E.

Hirsutism is often an early symptom of non-classical CAH, and may appear after puberty in classical CAH. Excess facial and body hair is not just a "cosmetic" issue. Hirsutism has profound psychological and sociological side effects for the afflicted patient.

In treating CAH, medication is used to lower the overall level of androgens in the blood. This prevents the hirsutism from becoming worse. However, *it will not reverse the excess hair growth.* Once a hair follicle has been stimulated to grow a pigmented hair, it will continue to do so unless the hair follicle is permanently destroyed. To date, there is only one proven method of hair removal that can achieve this, and that is ELECTROLYSIS.

Electrolysis is **permanent** hair removal achieved by the insertion of a fine, sterile probe into a hair follicle and the application of electrical current, which destroys the follicle through a *chemical reaction*, *heat*, or *combination* of both. Electrolysis, also known as Electro-epilation, is gentle to the skin, comfortable for the patient, and yields beautiful results.

To appreciate how Electrolysis works, it is important to understand that hair grows in cycles. At any given time, a hair will be in one of the following three stages of growth: *anagen* (active), *catagen* (transitional), or *telogen* (dormant). In any given area, not all hairs are in the same stage of growth at the same time.

To prevent a hair from growing, the technician must disengage 2/3 of the hair follicle where hair germ cells exist. To achieve this, a hair is best treated during the anagen phase. Because not all hairs are in this active phase at the same time, a hair may need to be treated several times over a period of months before all hair germ cells are effectively destroyed. For this reason, it could take 12- 18 months to complete a large project such as the bikini and inner thighs. At the beginning of treatment, the patient will come in for sessions that are longer in duration and more frequent. Eventually, she will come in for 'clean-up' sessions perhaps once every 6- 8 weeks. To complete Electrolysis treatments in the shortest amount of time, it is important to commit to a regular treatment schedule. In addition, the CAH patient must be in good hormonal control throughout the treatment period and beyond. This is to make sure androgen levels do not rise too high and stimulate new hair to grow in untreated follicles.

Electro-epilation encompasses three methods or *modalities*: Electrolysis, Thermolysis, and Blend.

Electrolysis was the first modality applied to permanent hair removal in 1875. Dr. Charles Michel, an ophthalmologist of St. Louis, MO, used it to treat ingrown eyelashes. Electrolysis is the application of direct current (DC) to a solution of salt water, which produces the byproduct sodium hydroxide (lye). In this method, a sterile probe is inserted into the follicle and DC flows through the probe. When DC meets the natural salt and moisture in the skin, a tiny amount of lye is produced. Since lye is a caustic solution, it causes damage to the hair germ cells. Electrolysis utilizes a *chemical reaction* to destroy the hair. It is gentle and comfortable because the chemical reaction takes place slowly. The probe remains in the follicle for several minutes while the lye develops. Today's epilators use multiple probes simultaneously to speed up the process.

Thermolysis uses oscillating current or high frequency (HF) to produce *heat*, which coagulates the hair germ cells in the hair follicle. This is the most common modality of permanent hair removal practiced today because it is fast. A single, pre-sterilized, disposable probe is inserted into the follicle. HF flows through the probe and a small amount of heat is produced for fractions of a second. The heat disengages the hair germ cells and cauterizes the follicle's blood supply. This method is also known as *shortwave* or *flash.*

Blend is a *combination* of direct current and high frequency. In it, DC and HF are sent through the sterile probe simultaneously. This method takes advantage of the strengths of both modalities. All three methods of Electro-epilation can achieve permanent hair removal. Which modality a patient is treated with depends on her hair type, her tolerance, and her Electrologist.

One of the biggest misconceptions about Electrolysis is that it is very painful. The hair follicle is surrounded by nerve endings, so the patient will feel something. Most patients, however, find the treatment quite comfortable. If a patient is very sensitive, her

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electrologist can offer sugestions to make treatment easier, for example, the use of a topical anesthetic such as LMX Cream. Without fail, new clients are always surprised at how comfortable Electro-epilation really is.

In addition to being comfortable, Electrolysis is also cost-effective. Remember, it is a *permanent* solution to unwanted hair. While temporary solutions like shaving and waxing are less expensive up-front, over a lifetime a patient will spend thousands of dollars more on these methods than on Electrolysis treatments. Electrologists charge by the amount of time a patient spends in a treatment session, not by the area being treated. Hourly rates will vary according to geographic location. While HMOs generally do not reimburse for Electrolysis, some might allow a patient to seek reimbursement if it is deemed medically necessary by a physician as part of the overall treatment of a disorder such as CAH. A precedent has already been set to this effect (Abernathy vs. The Prudential Insurance Company of America, No. 21178, Supreme Court of South Carolina, March 31, 1980).

Immediately following an Electrolysis treatment the skin might be slightly red and/or swollen. This usually disappears within 1- 2 hours. In the days following, the treated area may develop tiny scabs. These scabs are part of the normal healing process and should not be touched. Your Electrologist will go over specific post-treatment instructions with you to ensure proper healing.

Electrolysis is safe for all skin types and effective on all types of hair. It has been studied extensively and found to have no long-term negative side effects. It can be done safely on all parts of the body except the inside of the ears and nose because of the risk of possible infection. You should not have Electrolysis if you are on Accutane because of the skin's compromised ability to heal on this medication. Diabetics should get their doctor's written permission before beginning Electrolysis treatments for the same reason.

In the United States, regulation of Electrologists varies from state to state. Most states require licensure for Electrologists, but some do not, i.e. New York.

If you are seeking treatment in an unregulated state, it is especially important to look for a practitioner who is a Certified Professional Electrologist (CPE). This is a national board certification credential established by the American Electrology Association (www.electrology.com). It is a voluntary credential and requires ongoing continuing education to be maintained. It shows that the practitioner adheres to specific standards of practice established by the AEA such as those for Infection Control. These standards include the sterilization of all Electrology instruments which may have parenteral contact, the use of pre-sterilized, single-use disposable probes, hand washing, and the use of disposable gloves for each treatment.

An article on Electrolysis would not be complete without mentioning laser hair removal. The most important fact that must be clarified about laser hair removal is the following: It is not proven permanent. The FDA has ruled that laser manufacturers are not allowed to claim that laser hair removal is permanent (http://www.fda.gov/cdrh/consumer/laserfacts.html). No long-term studies have been conducted on laser hair removal's efficacy or safety.

Since its creation in 1875, the benefits of permanent hair removal with Electrolysis have been proven again and again. The CAH patient who suffers from hirsutism can rest assured that Electrolysis will provide a solution that is safe, comfortable, cost-effective and above all, **permanent.** ❤

Kelly Wins Pfizer's Unsung Hero Award!

On April 15, Pfizer Inc. honored Kelly Leight by awarding her their first Unsung Hero Award, which was presented by soccer star, Mia Hamm. Pictured above are Pfizer's Greg Duncan, Kelly Leight, and Mia Hamm. This program recognizes one individual each year who provides outstanding support and resources to those affected by illness or disability. The award comes with a grant of $10,000 to CARES.

Introducing CLAN (CAH: Living As Neighbours)

by Kate & David Hansen
CARES Members from Australia

What If…?

Have you ever reached for that bottle of hydrocortisone or fludrocortisone and felt that involuntary shiver down your spine as your mind played those irrational "what if?" games? What if you couldn't get any more medicine? What if you knew this was your last bottle and it had to somehow last you the next few months? What if…?

Thanks to two articles in the CARES Foundation Fall Newsletter of 2004 (see the newsletter archives at www.caresfoundation.org) we, the international CAH community, were made aware that this scenario is in fact a horrible reality for hundreds of families living in Vietnam with CAH.

The Situation in Vietnam

In Vietnam CAH can be an enormous burden on patients and families who are unable to access affordable medication. Fludrocortisone and hydrocortisone are not routinely imported into the country, and so families are forced to purchase their drugs on the black market at huge expense. Just one bottle can cost a family over one third of their already modest monthly income. Unable to access hydrortisone, parents are left with no choice but to use inferior substitutes for their children – when it's all you can get you take it. Problems with drug availability and affordability also mean parents are rationing doses, and chronic under-suppression comes at a great cost. Short stature is common amongst older patients. Several children with CAH die each year – rates unheard of in wealthier countries of the world. It was the enormity of this inequality that sparked CLAN.

CAH: Living As Neighbours

CLAN is an acronym for **C**AH: **L**iving **A**s **N**eighbours. It is a charitable organization based in Australia, that is dedicated to the dream that all persons living with CAH in the developing countries of the world have affordable access to essential medications. CLAN is based on the concept that families affected by CAH belong to a worldwide community – a human family scattered around the earth. But more than this, we can choose to *live* as a community, and care about each other as neighbours.

Sometimes it is easy to forget things that are painful and hidden by distance. But the international CAH community has the opportunity – and responsibility – to speak out and take action to support our members in developing countries.

We have seen examples already of the power of the CAH community when it unites. Consider organizations such as CARES; the various National CAH Support Groups that meet around the world; the numerous newborn screening lobby groups; many web-sites sharing information and support, and the various CAH Conferences that present the latest treatments and advances in technology for all to share.

What is CLAN doing to help?

There is a big movement worldwide currently looking at improving affordable access to essential medication for people living in developing countries. Most notably these discussions relate to patients affected by HIV/AIDS, malaria and TB.

In all these international policies and good works, CAH rarely rates a mention. Most babies die before accurate diagnoses are made, and the numbers that survive are so small as to make them virtually powerless. Cultural taboos related to ambiguous genitalia also mean that parents in some countries are reluctant to speak out. Even in the formulation of the World Health Organisation's List of Essential Medicines, fludrocortisone was recently removed, apparently because it was thought only patients with Addison's Disease needed it, and their numbers were so small as to not warrant the drug's inclusion.

As the international CAH community – and members of civil society - we have a responsibility to understand the situations facing our members in developing countries, and speak out when they need our support to right wrongs. Long-term sustainable solutions will require a dedicated, collaborative approach involving: developing country governments; international organisations (such as the WHO); health professionals; pharmaceutical and biotechnology companies; non-government and philanthropic organisations, as well as the broader private sector. As caring neighbours, we need to monitor the activities of the various stakeholders mentioned above, lobby when they fail in their duties, and do what we can to effect change.

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To this end, CLAN has worked first to put a halt to the suffering of children with CAH in Vietnam by securing access to free medication for all 220 CAH families in Hanoi for the next 2 years. This has been enabled by the generous support of two large drug companies in Australia, as well as some basic fundraising. Through our association with the Royal Children's Hospital International (RCHI) (the philanthropic arm of the Royal Children's Hospital in Melbourne, Australia) we are able to offer receipts for tax deductions on all donations over $2. We are also able to access the enormous experience and energy of the staff of RCHI, and tap into their strong relationships with the National Hospital of Paediatrics (NHP) in Hanoi.

However this has just been the first step, and unfortunately could prove nothing more than a temporary band-aid measure unless long-term solutions are found. Consequently we are now communicating widely to understand the specific barriers to affordable access to medication in Vietnam, and lobby for change within the capacities of existing health systems and budgets. Physicians in Vietnam are also working hard for change, and CLAN is keen to support their efforts. We are also sponsoring the next annual CAH Support Group meeting in Hanoi – an active and vocal local group will be essential for the future.

CLAN's Vision for the Future

We hope that CLAN is really only just beginning. Already we are liaising with doctors in the Philippines who want to help their patients access hydrocortisone and fludrocortisone – these drugs are not routinely imported into the Philippines either. Presently the doctors are purchasing the drugs themselves when traveling abroad, or through friends overseas – that's commitment! - and one Florinef tablet can cost a family $2. Because donations of all drugs to treat all CAH patients in the developing world is not a viable answer, CLAN is communicating with drug companies and other responsible stake-holders with the hope of finding sustainable solutions. We hope to support the establishment of a CAH support group in the Philippines so the people can harness their own power to bring about change.

Where to next? No doubt we will learn of other CAH families in other developing countries who need support, and we hope in the future to work with them also.

What can you do to help?

Please consider what YOUR role could be in this united international CAH community. Do you have special skills or contacts that could help? Do you feel especially called to be involved? Are you good with fundraising? Could you afford to make a regular donation? Please visit CLAN's website (www.cahclan.org) and contact us if you have any queries, suggestions or offers.

Kate, David and their 3 children (eldest, age 6, has CAH) live in Sydney, Australia. David works in food manufacturing as a food technologist like his hero Clark W. Griswold, and Kate is a practicing doctor.

Issues in Drug Access

For many CAH patients throughout the world, obtaining access to medications is a continuous problem due to expense and availability. Although drug donations can be effective, they cannot supply all the long-term pharmaceutical needs of developing countries. Many governments and advocacy organizations are developing and initiating alternative strategies to increase worldwide access to essential medicines. In order to address barriers to drug access, the British government has issued a new report outlining several possible alternatives to improve access (*Increasing people's access to essential medicines in developing countries: a framework for good practice in the pharmaceutical industry*, UK Government policy paper, March 2005). The report urges pharmaceutical companies, as well as international organizations, to promote access by considering all of the options available, such as: differential pricing, increasing research and development for diseases affecting developing countries, considering voluntary licenses, and increasing access to essential medicines.

The World Health Organization maintains a list of essential medicines, which could provide treatment for the majority of diseases. This bi-annually updated list serves as a recommendation for governments and healthcare organizations. Although hydrocortisone is on the list, fludrocortisone was taken off during the last review. CARES Foundation will work with other organizations, such as CLAN, to promote access to medications used for CAH by applying to get fludrocortisone back on the list of essential medicines, and by developing strategies to serve the CAH community worldwide.

The Potentials of Adrenal Cortical Stem Cells

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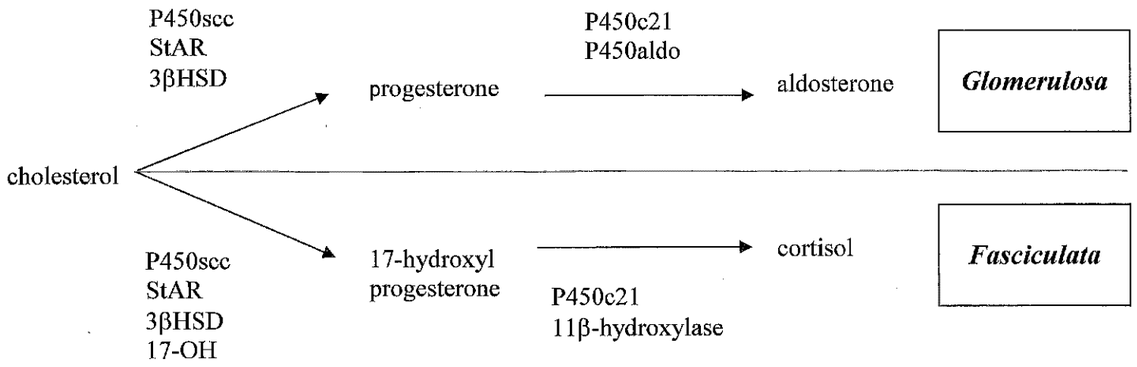

Figure 1. Diagram of the biochemical pathways of steroid production in the adrenal cortex. Only the glomerulosa and the fasciculata are illustrated here.

All adrenal steroid hormones are synthesized from cholesterol (Figure 1).

Cholesterol is first converted to pregnenolone by cytochrome P450scc, which is present in every adrenal cortical zone. Pregnenolone is converted to progesterone by 3b-hydroxysteroid dehydrogenase, which is also present in all adrenal cortical zones. Progesterone is converted to deoxycorticosterone by cytochrome P450c21, which is present in both the glomerulosa and the fasciculata. Aldosterone, a mineralocortiocoid released by angiotensin II stimulation, is synthesized from deoxycorticosterone by cytochrome P450aldo, which is present only in the glomerulosa. On the other hand, cortisol, a glucocorticoid released by the adrenocortictropic hormone (ACTH) stimulation, is synthesized from deoxycortisol by the enzyme 11b-hydroxylase, which is found only in the fasciculata.

When these biochemical pathways are disturbed, the production of the steroid hormones is altered, often leading to states of adrenal insufficiency. In congenital adrenal hyperplasia, most patients have mutations of the 21-hydroxylase gene that encodes P450c21, a key enzyme in the production of both aldosterone and cortisol. Current medical therapy for adrenal insufficiency employs the administration of steroid hormones to replace the missing hormones. Such treatment is life-saving but is not ideal because the body's need for steroid hormones varies with the physiologic states. During periods of stress, the need for steroid is increased considerably, and the normal feedback regulation in the body would increase the production of steroids automatically. The current medical therapy for adrenal insufficiency does not have such automatic adjustment. Instead, extra steroids are empirically given during stress states such as a significant illness.

Traditionally the adrenal cortex is viewed as a static organ where the cells do not grow in response to physiological changes. More recently, it has been shown that this view is incorrect. The functional zones within the adrenal cortex are dynamic layers of cells that do respond to changes in physiologic need by cell growth. For example, when excess ACTH is present, the fasciculata increases in size to produce additional glucocorticoids. Alternatively, when a salt-deficient diet is given, the glomerulosa increases in size to produce extra mineralocorticoids to retain salt within the body. It is speculated that the additional cells that expand the functional zones are generated from stem cells that reside in an area between the glomerulosa and fasciculata. Adrenal cortical cells in this area are actively growing on a continuous basis. This can be demonstrated by using a special tracer, bromodeoxyuridine, to label the adrenal cortical cells that are making new DNA. With special staining, such labeled cells appear as dark brown spots between the glomerulosa and the fasciculata soon after the label is given, and the labeled cells migrate toward the

(Continued on page 11)

(Continued from page 10)

Figure 2. A cross section through the adrenal cortex. Newly produced cells in the adrenal cortex are labeled as dark brown spots. Most of these cells are located between the glomerulosa (G) and the fasciculata (F). These cells also spread out towards the outer edge of the adrenal cortex and the reticularis (R).

adrenal capsule and medulla over a period of several days (Figure 2).

There are numerous other studies in the literature that strongly suggest the existence of stem cells within the adrenal cortex. These stem cells are capable of making new specialized adrenal cortical cells that produce different steroid hormones. If such adrenal cortical stem cells could be used to regenerate functional cortical tissues in patients with adrenal insufficiency, then the physiological secretion of steroid hormones could be restored.

In our laboratory, we have developed methods to isolate and to grow adrenal cortical cells from mice. Furthermore, we have transplanted these cells into other normal mice. We found that these adrenal cortical cells continued to express many characteristics of the normal adrenal cortex after transplantation. These observations suggest that it would be feasible to develop similar techniques for humans whereby adrenal cortical stem cells can be transplanted into patients with adrenal insufficiency. As a first step toward that goal, we are working to identify the adrenal cortical stem cells that can regenerate the adrenal cortex. If successful, the next phase of the experiments will utilize such stem cells for gene transfer so that a normal copy of the gene such as 21-hydroxylase can be inserted into these stem cells. Ultimately, the objective is to procure adrenal cortical stem cells from patients with congenital adrenal hyperplasia so that these cells may be genetically modified in the laboratory. The corrected adrenal cortical stem cells that carry a normal copy of the gene will be transplanted back into patients with congenital adrenal hyperplasia. For those patients who do not have adrenal glands, it may still be possible to transplant adrenal cortical stem cells from another donor, but this will likely require immunosuppressive drugs to prevent rejection. With the proper technique, the transplanted adrenal cortical stem cells will able to regenerate functional adrenal cortical tissues as a potential cure for adrenal insufficiency.

New Website for KIDS Explains CAH!

The Hospital for Sick Children in Toronto, Canada, has launched an interactive website (http://www.sickkids.ca/childphysiology/) to educate children about their own physiology, and about certain medical conditions. In addition to a an extensive and informative section on genetics, the website also includes information about CAH, with animated and colorful adrenal glands, enzymes, as well as an interactive description of genital development.

This educational resource is also suited well for adults who seek a more visual understanding of CAH. In order for the website to function interactively, Flash player needs to be installed on your computer. The section on CAH can be accessed at: http://www.sickkids.ca/childphysiology/cpwp/Genital/GenitalCAH.htm .

In loving memory of
Katelyn Ireland
Age 14 mos., SWCAH

With deepest sympathy, CARES Foundation sends condolences to family and friends.

Medical Management of CAH: Transition from Childhood to Adulthood

(Continued from page 5)

additional treatment with Clomid, a standard fertility drug, to conceive. Despite high levels of maternal testosterone (male hormone) during pregnancy, unaffected female offspring, even of classic CAH mothers, show no genital ambiguity. This is because the placental aromatase enzyme prevents maternal testosterone from reaching the fetus.

Reproductive function in CAH males: Impaired reproductive function is less frequent among men with classic CAH compared with affected women. Most CAH males are able to father children, or at least have adequate sperm counts. Low sperm counts, observed in both classic and nonclassic CAH, do not preclude fertility. Testicular adrenal rests (also referred to as "testicular tumors of adrenogenital syndrome"), may occur in CAH males, especially if they are inadequately treated salt-wasters. For this reason, it is recommended that by adolescence all CAH boys have careful testicular examinations and a baseline testicular sonogram. The preferred treatment consists of effective adrenal hormone suppression. Male infertility and testicular adrenal rest tumors are uncommon in nonclassic CAH.

Other problems associated with CAH: The incidence of adrenal masses increases with age, and is higher in CAH than in the general population. Most adrenal masses associated with CAH are benign.

Medical treatment: With the completion of growth, CAH patients may be managed with more potent glucocorticoids (for instance, prednisone or dexamethasone, rather than hydrocortisone). These drugs are not often used routinely in children because of their greater tendency to cause growth suppression. Treatment during pregnancy requires special consideration, depending on whether one is treating the mother with classic CAH (where hydrocortisone or prednisone are used), or a potentially affected fetus (where dexamethasone is used because it crosses the placenta intact). Patients treated for nonclassic CAH before or during the child-bearing years may not require treatment throughout adult life, if symptoms abate. Patients on long-term prednisone and dexamethasone should be carefully monitored for potential development of excess weight gain, hypertension, and high blood glucose. Some patients may be able to reduce or discontinue fludrocortisone (Florinef®) in later childhood or adolescence. The reason for this is not entirely understood, but may relate to increasing salt consumption with age, and to better conservation of salt by the mature body. There are other treatments for CAH women that supplement glucocorticoid therapy. These include cosmetic hair removal, topical and oral anti-acne medications, and drugs that either reduce production (e.g., oral contraceptives) or block action (e.g., flutamide or cyproterone, not approved for this use in the U.S.) of male hormones.

Surgical treatment: In the past, surgical correction of genital ambiguity was considered an emergency to be repaired, at least in part, before the infant was discharged from the hospital, or in the first few months of life. Current thinking and practice are evolving to allow the families to play a greater decision-making role in situations that do not require immediate attention. In some centers, a concerted surgical repair may be performed in a single stage in early life, while others will delay vaginal reconstruction until nearer the time a girl is mature enough to become sexually active. The decision of whether, when, and what type of genital surgery is desirable needs to be decided by the family and/or patient, experienced surgeons, and endocrinologist; surgery is not necessary in many cases.

Genetic counseling: The diagnosis of CAH is most often made on clinical grounds and on the basis of hormone measurements. Genetic counseling is useful in helping the family understand the implications of the diagnosis. There are several circumstances when genetic testing is important, especially, prenatal diagnosis and newborn screening with questionable results (see "The Basics of Genetic Testing for CAH," Winter 2004-05 newsletter). Another common situation arises when a woman has been told she has "CAH" and is treated with glucocorticoids, but the hormonal measurements upon which the diagnosis was made are either unavailable or in doubt. Since hormonal diagnosis is unreliable during or immediately following glucocorticoid treatment or during pregnancy, genetic testing is a viable alternative. Genetic testing may also be done for those who have been diagnosed with nonclassic CAH who wish to know whether they carry a classic trait or allele. The estimated risk of a nonclassic CAH parent and a partner of unknown genetic status of having a child affected with classic CAH is about 1 in 1,000. Since this overall risk is relatively low compared with the 1 in 8 risk to a couple who have already produced a child with classic CAH, prenatal

(Continued on page 13)

(Continued from page 12)

treatment and invasive prenatal diagnosis seem unwarranted, especially now that newborn screening is performed in most states.

Carriers or Heterozygotes: Family members often wish to know their risk of transmitting CAH. Parents of a child with CAH have a 50% (1 in 2) risk of transmitting the CAH trait to offspring, and this risk is the same in each successive pregnancy. Thus, there is a 25% (1 in 4) disease risk for each sibling of a CAH patient born to the same parents. Carriers of a single CAH trait do not usually suffer from any significant symptoms or hormone imbalance, and are consequently not candidates for hormonal replacement therapy.

Classic CAH is a congenital disease with far-reaching ramifications in childhood, adolescence and adult life. Much progress has been made in understanding various aspects of CAH, but several areas remain to be studied. Nonclassic CAH is of variable severity, and should only be treated in cases with overt clinical evidence of androgen excess. Whereas treatment duration is lifelong in classic disease, this is not necessarily so for the nonclassic disorder. Patients, parents and family members should be aware of the changing needs of CAH patients throughout life, and seek appropriate medical care.

Phyllis W. Speiser, M.D., a member of CARES Foundation's Scientific and Medical Advisory Board, is Chief of the Division of Pediatric Endocrinology at Schneider Children's Hospital in New Hyde Park, NY.

CARES Foundation Continues Campaign to Expand Newborn Screening

Ever since CARES began its work in 2001, one of its prime goals has been to ensure that all newborn babies are tested for CAH. The importance of newborn screening for CAH, as well as 28 other heritable diseases, is emphasized in a recent report by the U.S. Department of Health and Human Services ("*Newborn Screening: Toward a Uniform Screening Panel System,* Federal Register: 3/8/05). Our efforts have been an uphill battle, but we are doing extremely well: In the past few months, Kentucky, Utah, and Nebraska have enacted measures to expand newborn screening. CARES Foundation will continue to offer its services to support these newborns and their families by working with the states' Departments of Health.

Between July 1, 2005 and January 1, 2006, Kentucky will begin to expand its newborn screening panel to incorporate 29 additional disorders, including CAH. In March, Governor Ernie Fletcher signed the newborn screening bill, and also set aside $1 million in tobacco settlement funds to help pay for the initial costs of the program. In Utah, an expanded newborn screening panel which includes CAH will be fully implemented by January 1, 2006. Lawmakers in Utah mandated this expansion as a result of the advocacy efforts of CAH-affected families. In Nebraska, the Health and Human Services System Policy Cabinet approved adding CAH to its newborn screening panel and expects to begin screening in January of 2006. In a letter from Nebraska Governor Dave Heineman to CARES Foundation, the Governor states that the "Newborn Screening Program will be working over the next several months to revise regulations and ensure quality education, testing, follow-up, diagnosis and treatment are in place to effectively meet the needs of newborns and their families."

In other states, the successes of past newborn screening campaigns are now being realized. In California, where CARES members and other newborn screening advocates waged a successful campaign in 2004, the expanded newborn screening panel will be fully implemented this August. In Oklahoma, the Newborn Screening Program of the Oklahoma State Department of Health, in collaboration with the Oklahoma University Department of Pediatrics, Section of Endocrinology, has not only expanded newborn screening to include CAH, but has committed to providing long-term follow-up services. Effective February 14, 2005, the program offers a new resource of information, support, and advocacy, and plans to educate healthcare professionals as well. CARES members in Oklahoma should contact Kathy Kirk, Endocrinology Long-Term Follow-up Coordinator for CAH, at (405) 271-8001 ext. 43051, to learn more about the available services.

These lifesaving advances in CAH detection would never have come about were it not for CARES members. By giving their time and energy to provide testimony, write letters, and personally meet with state officials to make them aware of the need for screening newborns for CAH, these volunteers have been largely responsible for pushing newborn screening measures through the various state legislatures.

Volunteers for Research Studies Needed!

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

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

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

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

To find out more information about participating in the study, please contact Barbara O'Brien, M.D. (**bobrien@tufts-nemc.org**) or Diana Bianchi, M.D. (**dbianchi@tufts-nemc.org**) at Tufts-New England Medical Center (**phone number: (617) 636-1468**).

Jenny Lynn thanks everyone who offered her support and well wishes. She is now living at home and continues to recover. Soon her family will be moving to Texas. For updates, visit her website at www.jennylynn.org .

More Photos from the Every1CARES Luncheon

From Top to Bottom: Board member Alan Macy makes an auction bid; Kelly Leight and Dede Alpert pose together; Luncheon attendees evaluate auction items; Guests at the boutique admire purses.

CARES Family Support Groups in the US

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

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

Are you interested in starting a support group in your state or country?
Contact the CARES office and we will get you started.

International CARES Family Support Groups

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

CANADA
Alison Weatherall
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Aliweatherall@rogers.com

Tina Haslip
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¡Atención Personas que Hablan Español!

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

Update on Genetic Discrimination Legislation

by Erin Anthony

The Genetic Information Non-Discrimination Act of 2005 (GINA), was introduced in the House of Representatives in March. Despite winning unanimously in the Senate (98-0), the legislation is said to face heavy opposition in the House, where a similar effort died last year.

The legislation bars health insurers and employers from discriminating against people with a genetic predisposition to disease. It prevents health insurers from excluding people from coverage or charging higher rates because of genetic risk. Under the legislation, insurers cannot require customers to take genetic tests prior to enrollment, and employers are barred from basing hiring and firing decisions on genetic information. The current measure covers public and private health plans, employers, employment agencies, labor organizations and training programs. It also tightens protection of medical information privacy.

GINA is the work of Sen. Olympia J. Snowe (R-Maine) who has introduced similar legislation in each of the last four congresses. The first version of this type of bill was introduced in April 1996.

According to Snowe, the legislation is necessary to insure that advances in treatment and prevention of disease do not constitute a new basis for discrimination. Medical experts say that many people who could benefit from certain genetic screening tests do not take them for fear of losing health coverage for themselves or their families. For example, The National Institutes of Health report that nearly 32 percent of women offered genetic testing for breast cancer risk decline to take it citing concerns about health insurance discrimination.

Since its introduction into the House, GINA has gained 24 new cosponsors bringing the total to 76. Although the legislation is supported by President Bush, Michael Maves the chief executive of the American Medical Association, The National Society of Genetic Counselors, The Coalition for Genetic Fairness, and many others, it faces heavy opposition from business lobbyists and some Republican leaders.

Business groups argue that the legislation will add regulatory burdens without improving consumer protection. But, supporters think national rules, rather than state regulations are in everyone's interest. Currently, about 15 states have laws specifically protecting against genetic discrimination. To find out if your state has laws against genetic discrimination, log on to the homepage of the National Conference of State Legislatures at www.ncsl.org .

How does genetic discrimination affect people with CAH?

Genetic testing is particularly important in families with a history of CAH. Testing in the parents allows for early detection and treatment in the child—especially critical in states without newborn screening. However, some people may fear discrimination by health insurers and avoid testing for CAH. The Genetic Non-Discrimination Act of 2005 would prevent insurers from dropping coverage or charging higher rates for families known to carry mutations for CAH.

What can you do?

Right now, GINA is in the House of Representatives and faces heavy opposition from business lobbyists and some Republican leaders. It is important that state representatives know how important it is that GINA passes. Legislation like GINA has been introduced in each of the last four Congresses and never passed. It also won unanimously in the Senate last year but then died in the House. To find your Congressperson, log on to www.house.gov/writerep .

Medical Bracelets: Underused Lifesavers

Accidents and medical emergencies can happen anywhere, anytime. Given the potential seriousness of such emergencies, all people who have an allergy or medical condition should wear some sort of medical ID. For a minimal amount of money, medical bracelets, necklaces, or tags, can save your life. However, those wearing medical bracelets today do not nearly represent the tens of millions of people who should be wearing these lifesaving bracelets.

In a recent *Wall Street Journal*, article, Kevin Helliker details how use of the Medic Alert bracelets is lower today than it was ten years ago (4/5/05). Helliker states that physicians prefer medical warning tags over electronic files. Even though paramedics are taught to look for the tags, often doctors fail to recommend their use to patients. For CAH patients, CARES Foundation strongly recommends wearing a medical ID bracelet, such as those made by CodyCares (see p. 19).

Highlights of 2004

January 1 - December 31, 2004

Programs

- *Conference*: Hosted all day (free) conference for individuals and families affected by CAH at Overlook Hospital in Summit, NJ. Over 200 people attended from 23 states and 4 countries.
- *CARES Support Groups*: Added 12 new support groups, totaling 21 CARES support groups across the United States and Canada.
- *Newborn Screening*: CARES advocacy efforts resulted in 4 additional states adding newborn screening for CAH.
- *Research*: Provided grants to 6 medical institutions for CAH related research.
- *Newsletter*: Developed and distributed 3 comprehensive newsletters to members and physicians across the United States and 40 countries abroad.
- *CAH Awareness:* Initiated bracelet and holiday card programs in October, 2004 to raise awareness about CAH. Four thousand bracelets were sold in 2004.

General

- Moved from basement to "real" office space able to accommodate additional staff and volunteers.
- Redesigned and expanded website including archived newsletters and improved navigation.

Fundraisers

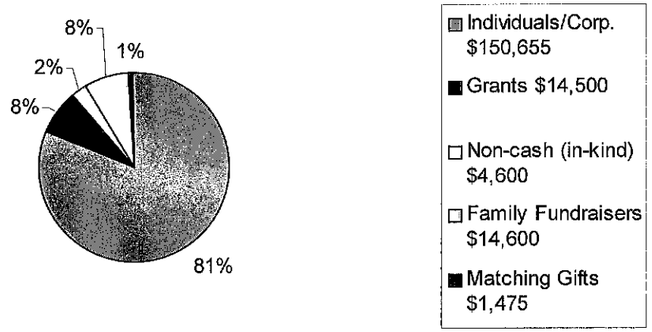
- Year-End Appeal
- Instituted new mid-year appeal
- CARES members initiated 11 local family fundraisers around the country.

If you would like a copy of our audited financial statement, please contact the CARES Foundation office.

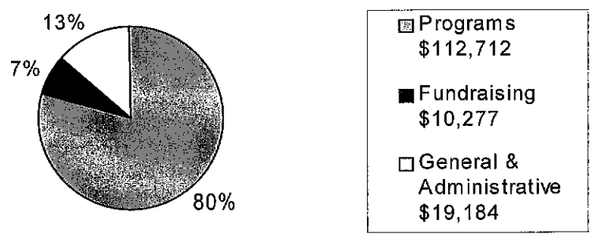
CARES Foundation 2004 Annual Report

CARES Foundation, Inc. is committed to education and research for Congenital Adrenal Hyperplasia while providing the resources and the latest information available for managing life with CAH.

2004 Sources of Income



2004 Expenses



News from the CARES Foundation Office

Feedback Needed!

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

Newsletter Correction

In the CARES Foundation Newletter V4:1, Winter 2004-05, page 13, there is an error in the listing for Esoterix, on the chart entitled "U.S. Laboratories Offering DNA Testing for CAH." Esoterix does not perform Southern Blotting, nor do they perform complete sequencing of the 21-hydroxylase gene. The mutation detection rate should be 90-95%.

CARES Foundation apologizes for this error and any inconveniences it has caused. Updated copies of the chart can be printed from the website (http://www.caresfoundation.org/news_letter/winter04-05_page_8.htm) or requested from our office.

Updated Physician Listings Available

CARES Foundation has compiled a large list of pediatric endocrinologists, some adult endocrinologists, urologists and psychologists with experience in treating CAH/NCAH patients.

Recently, we updated the listing to ensure that we distribute the most accurate and detailed contact information available. We sincerely thank all the healthcare professionals who responded to our letter, and promptly returned our questionnaire.

Fundrai$ing Corner

Attention Golfers: Save the Date!
Golf for CARES on September 22nd!

Dr. Richard Rink, Event Chair
Indianapolis Area, Indiana

For more information contact Committee Chairs:
Trena Brim, (317) 278-8554 or tbrim@iupui.edu
Jenny Hendricks, (317) 783-7702 or jennylhendricks@sbcglobal.net

Want to run a CARES Fundraiser?

Call us for a copy of our NEW Promotional Video!

The CARES Foundation video premiered to great acclaim at the Every1CARES Luncheon. This short video is a great introduction to CARES and CAH. Many thanks to Dr. Phyllis Speiser, Paul Duffy at Altered Image, Crazy Duck Productions, and to all our CARES members who participated in the filming.

For great fundraiser ideas, you can order the CARES Foundation Fundraising Guide, or look at http://www.fundraising-ideas.org/DIY/.

Happy Birthday Brenda Siegal!

Thank you for asking your friends and family to make a donation to CARES in honor of the occasion! Brenda is the mother of Bonnie Stevens, an active member of the CARES Foundation Board of Directors. Now we know where Bonnie's generosity and enthusiasm come from!

Thank you Daniela!

Daniela Quintanilla, daughter of Board member Vivian Altman, made CARES the subject of her Bat Mitzvah project. She spent several weekends slapping labels on our Every1CARES bracelets, selling CARES bracelets and jewelry that she had made. She gave a portion of her Bat Mitzvah gifts to CARES. She collected over $1,000 for CARES while raising awareness of CAH.

RUNNERS WANTED!

Need a little extra motivation to train for the New York Marathon? Let's start a team of runners to come together and run the Marathon to raise money and awareness for CARES Foundation, Inc. I look on this as a long-term idea, which will gain more and more interest over the next few years. Any ideas or interest please contact John Rollo, at jrollo@comus-intl.com

CAH CHAT GROUPS

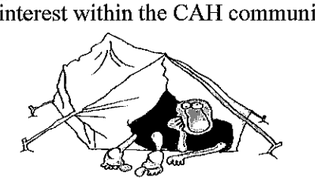
CARES Teen Chat Group: A place for teens with CAH to talk about feelings, questions, and life experiences with CAH. To join, go to: http://health.groups.yahoo.com/group/caresteenchat/ and click on "Join This Group."

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

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

Help Start a Camp for Kids with CAH!

CARES Foundation is starting to plan an exciting long-term project: to establish a summer camp for kids with CAH. Interested members, healthcare professionals, social workers, psychologists, camp counselors are urgently needed to help make this dream a reality. If you are interested in becoming a part of this project, or if you have expertise or access to resources that you can contribute, please contact Renata ((866) 227-3737, renata@caresfoundation.org). We need to assess interest within the CAH community!



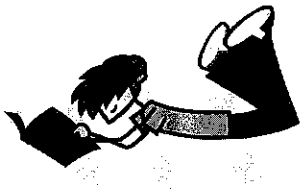
CARES Thanks Sue Bianchi, Our Volunteer Webmaster Extraordinaire!

Many families affected by CAH first learn about CARES by finding our site on the internet. Our website hosts numerous useful articles, archived newsletters, parent tips and resources. Sue Bianchi, our volunteer webmaster enthusiastically strives to update the site, while maintaining its artistic design. The CARES Foundation is extremely grateful for all of her work, her willingness to respond to our requests, and her patience.

Sue lives in Northern California. She has three children and two grandchildren. Ashlee, her oldest grandchild, has salt-wasting CAH. "Being a small part of an organization that is involved in saving babies lives through newborn screening, supporting appropriate research projects for CAH, providing CAH education, and supporting families is very fulfilling," says Sue. "I really enjoy working with the people at CARES Foundation and believe they are making a difference in the lives of our families."

Extra Paper Newsletter Copies Are Now Available

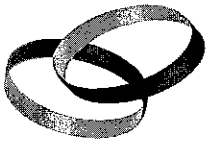
A limited number of our archived newsletters are available for our members. If you would like to request extra copies for distribution to your doctors, nurses, family members, teachers, etc., please let us know by contacting Renata (renata@caresfoundation.org or (866) 227-3737). Remember to include the exact number of newsletters, the edition you would like, and where the newsletter should be sent.



The following issues are available: Winter 2004, Spring/Summer 2004, Fall 2004, Winter 2004-05. As always, you can find the contents of these newsletters online: www.caresfoundation.org/news_letter/ or click on **Newsletter… Archives** on the home page.

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

BRACELET SALE RAISES MONEY, AWARENESS



EVERY1CARES bracelets are still selling thanks to the energy and dedication of CARES Foundation members!

So far, we have distributed a total of 5,604 EVERY1CARES bracelets, resulting in $17,106 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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