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Coping with Hirsutism

This article is a collaborative project of professionals from diverse disciplines (under the direction of Drs. Maria I. New and Heino F. L. Meyer-Bahlburg) of New York Presbyterian, the combined University Hospitals of Cornell and Columbia.

What Treatment is Available for my Excess Hair?

HAIR BASICS

Hair is really just an outgrowth of the skin layer called epidermis. In fact, hair and skin are composed of the same protein (keratin). The hair shaft is produced by the hair follicle within the skin. The hair follicle has two regions: the hair bulb and the mid-follicle region. The hair bulb contains actively growing cells and pigment (melanin) producing cells. In the mid-follicle region the actively growing cells die and harden into what we call hair.

The follicle can produce two types of hair: (1) vellus hairs, which are pale, fine, and silky; (2) terminal hairs, which are darker, coarser and larger. During its life span, hair goes through three distinct phases: anagen, catagen and telogen. In the anagen phase, protein and keratin are continuously made to promote development and active growth of the hair shaft. At any point in time, 85-90% of hair is in the anagen phase, which can last from a few months up to six years. Hair then enters a transitional, or catagen, phase, when chemical and structural changes cause the follicle to regress and stop growing. In the final part of the cycle, the telogen phase, the hair follicle shuts down and goes into resting mode. In this stage hair can shed so that new hair growth can begin. The telogen phase can last up to 100 days.

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Save the Date!!

CAH Family Workshop

October 18, 2003

Children's Hospital of Los Angeles (CHLA)

Speakers scheduled to date:

Maria New, M.D. – Pediatric Endocrinologist
Mitchell Geffner, M.D. – Pediatric Endocrinologist, CHLA
Ricardo Azziz, M.D. – Reproductive Endocrinologist
Sheri Berenbaum, Ph.D. – Psychologist
Dix Poppas, M.D. – Pediatric Urologist

Details to follow at: www.caresfoundation.org



A Message from the Executive Director:



Dear Friends,

Since our last newsletter, our world has been through some major changes. We hope for rapid peace and stability for Iraq, the Middle East, the United States and the World. However in our homes, no matter what is happening around us, our struggle to better understand and cope with the disease congenital adrenal hyperplasia goes on. At CARES Foundation, Inc., we are tirelessly working for the interests of the CAH community.

Our Conferences

On March 8th, 150 CAH affected individuals, parents, physicians, nurses and state newborn screening officials came to Johns Hopkins University Medical Center in Baltimore, M.D. to attend our recent conference. It was such a wonderful day! The speakers were so impressive and enlightening.

Dr. Claude Migeon gave a fascinating history of the treatment of CAH, including telling us about his care of the very first CAH child to live through infancy through the use of glucocorticoids. This was not that long ago—only the early 1950's. Dr. Deborah Merke's exciting presentation on the newest trends in treatment, and new and promising research on CAH gave us all hope for better clinical treatment options for us and our kids. Dr. Sheri Berenbaum spoke about her groundbreaking work to better understand the minds and behavior of those affected by CAH. She gave us all a good look into the CAH brain

and the impact of the disease on mood, behavior, intelligence and learning and sexuality.

These physicians all presented without compensation and out of their dedication to the CAH community. Dr. Migeon graciously donated the use of the facilities at Johns Hopkins, and he and his colleagues (especially Amy Wisniewski), helped us with all of the arrangements. The Inner Harbor Starbucks donated the excellent coffee and baked goodies for our refreshments. Many pediatric endocrinologists in New Jersey, Maryland, Pennsylvania, the District of Columbia, Virginia, North Carolina and Delaware notified their patients about our conference, and the State Newborn Screening programs from NJ, DE, MD, VA and PA all mailed our invitations to the positive screens in their databases. Finally, many thanks to all of the CARES families who volunteered to plan this event and helped on the day of the conference, we could not have done it without you! We thank everyone so deeply for their time and effort that made this conference such a success!

Please save the date for our *next* conference in Los Angeles on October 18th at Children's Hospital of Los Angeles. Tentatively scheduled to speak so far are Dr. Mitchell Geffner, Dr. Maria New, Dr. Ricardo Azziz, Dr. Dix Poppas and Dr. Sheri Berenbaum. We are

also planning a conference in the NJ/ NYC area for the Spring of 2004.

Solu-Cortef Shortage and Alternatives

Unfortunately, Pfizer-Pharmacia is again experiencing production and distribution problems with Solu-Cortef and the company expects the shortages to continue through the end of 2003. Many of our families have been unable to obtain this important medication. On April 16, 2003, the company announced that it would no longer provide emergency supplies of

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Executive Director's Message

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this medication for families unable to obtain it locally. I wrote to Pfizer/Pharmacia and asked them to change this policy immediately and please make arrangements to provide CAH families with emergency supplies of Solu-Cortef. I am now informed that the policy is changed and emergency supplies of Solu-Cortef will be made available to families unable to obtain it through their pharmacies. Those in need should have their pharmacists call Pfizer/Pharmacia customer service at 1-800-821-7000. They will drop ship the medications to your pharmacy. Families seeking further information about supplies should call 1-800 323-4204. The company asked, however, that families do not attempt to stockpile the medication but only ask for the medications needed on an emergency basis.

Given the seriousness of this shortage and the implications for the health of our children, one of our medical advisors, Dr. Phyllis Speiser, is now giving all of her CAH patients prescriptions for hydrocortisone suppositories. They are much simpler to use than injections, and are rapidly absorbed to therapeutic levels within an hour, longer than the IM Solu-Cortef injection, but with longer lasting effects. Parents will be on their way to ER by the time drastic measures are required, and this is better than giving oral drug which is vomited. Suppositories cannot be used when the child has diarrhea. One manufacturer is Monarch Pharmaceuticals, and the product is called Anusol-HC. It comes in packages of 12 -25 mg suppositories. Higher doses can be compounded at specialized pharmacies. Some physicians may not be that familiar with the use of HC suppositories, although they are used regularly in

lieu of IM injections in Europe. Please refer them to the medical journal article: Newrick PG, et al.; *Self-Management of Adrenal Insufficiency by Rectal Hydrocortisone*, **Lancet**, 335:212-213 (1/27/90). Please ask your doctors about this alternative/complimentary product and refer them to this article for more information. We must have the tools we need on hand to save our children's lives if necessary.

New Advisors

Dr. Deborah Merke and Dr. Claude Migeon have graciously agreed to join our Medical and Scientific Advisory Board. We are so honored to have their expertise here to better guide CARES Foundation and help the CAH community. I hope you will join me in extending a hearty welcome to them both. You can read more about them on page 15.

Please Support CARES Foundation

We have been very blessed by the financial support of our members, their families and friends. But, we are continuing to expand our services as the funds come in and depend upon your generous support to continue our work. We have provided financial assistance to those who require travel for medical purposes. We have been able to help quite a few families to obtain the quality healthcare that they deserve through these financial grants. We have also made several grants for CAH research including grants to Cornell-Weill New York Presbyterian Hospital, John Hopkins University, Penn State University, Baylor College of Medicine-Texas Children's Hospital.

Our conferences, support services and informational materials are offered without charge to all CAH affected individuals and families. Please consider making a generous tax-deductible gift to CARES Foundation, Inc. so that we can continue our important work on behalf of the CAH community.

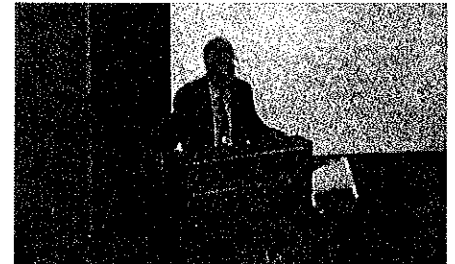
Many Blessings to All!

Kelly

CARES Conference Photos

John's Hopkins University

March 8, 2003



**Dr. Claude Migeon at the lecturn
(above); at break (below)**



Dr. Deborah Merke

Adrenal Crisis

*Phyllis W. Speiser, MD Director, Pediatric Endocrinology, Schneider Children's Hospital,
NY Professor of Pediatrics New York University School of Medicine, New York City, NY*

One of two adrenal glands sits on top of each kidney with a combined weight of only about 1/3 ounce in healthy adults. There are two main parts of the gland, the cortex (outer portion) and medulla (inner portion). The adrenal cortex is where the vital hormones, cortisol and aldosterone, are produced. These hormones are contribute to maintenance of blood pressure and heart muscle tone, and to sugar and salt balance. The adrenal cortex is also a secondary site for sex hormone synthesis. The adrenal medulla is responsible for production of the stress hormones, epinephrine and norepinephrine, also important in cardiovascular and nervous system regulation.

Adrenal insufficiency or "crisis" is important to recognize because of its potentially life-threatening implications. Crisis occurs when the adrenal is prevented from producing normal amounts of its vital hormones. Symptoms and signs of adrenal crisis are varied and nonspecific. In infancy these include lethargy, vomiting, poor appetite and failure to thrive. Doctors may mistake these problems for formula intolerance or inadequate lactation, or alternatively, primary infectious or gastrointestinal disorders. In older children chronic fatigue, headache,

gastrointestinal symptoms, salt-craving and excess skin pigmentation may be noted. Congenital adrenal hyperplasia is only one of many potential causes of adrenal crisis. Patients may undergo extensive evaluation before a diagnosis is made.

Often adrenal insufficiency has a slow, insidious course, but in some cases develops rather suddenly. The underlying problems include low blood sugar, low blood sodium, dehydration, low blood pressure, all predisposing the individual to heart failure and shock (collapse).

The classic form of congenital adrenal hyperplasia (CAH) should be considered in the differential diagnosis of adrenal insufficiency or crisis, especially in the infant. These days, with many states performing newborn screening for CAH, it is much less likely for CAH patients to go into shock. With newborn screening, most cases of severe or classic "salt-wasting" CAH are detected before crisis occurs. These individuals lack an enzyme, steroid 21-hydroxylase, and cannot adequately produce either cortisol or aldosterone, and must take hormone replacement therapy. Failure to begin treatment

in a timely manner, medical non-compliance, inadequate dosing, or failure to adequately absorb oral medications may all contribute to adrenal crisis. Thus, it is essential for the CAH patient to receive regular check-ups with measurement of appropriate blood hormone levels to assess whether he/she is receiving the right medication dose. Recent studies also have shown that CAH patients are also somewhat deficient in epinephrine and norepinephrine, although doctors do not replace these hormones.

Among all patients with classic CAH (21-hydroxylase deficiency form), about 25% actually have sufficient aldosterone to maintain salt balance. These people termed "simple virilizers." Because testing to distinguish salt-wasters from simple virilizers is somewhat complicated, and because physicians often do not wish to risk an adrenal crisis when they are uncertain of the child's aldosterone status, many "simple virilizers" will be treated with cortisol replacement and added oral Florinef (an aldosterone substitute known generically as fludrocortisone) to enhance salt balance. Over time, as the child grows accustomed to eating foods with higher salt content, the Florinef dose is tapered, and

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Adrenal Crisis

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eventually discontinued, although cortisol replacement is continued lifelong. Simple virilizers rarely experience adrenal crises. There are many known cases of such individuals having grown to adulthood and old age without ever having had the benefit of diagnosis or treatment, and without ever having a crisis, even during or after major surgery. Among the problems untreated classic CAH patients have are adrenal, testicular or ovarian tumors; untreated women with CAH have excess body hair and fertility difficulties. Thus, it is not advisable for individuals diagnosed with classic CAH to ever abandon cortisol replacement treatment. Salt-wasters most often also require additional Florinef and/or salt tablets indefinitely.

With increasing awareness among physicians, and with more sensitive newborn screening methods, there are now many individuals diagnosed at an early age with mild, or nonclassic, CAH. In contrast to classic CAH, this is not a life-threatening disease. Many individuals have no obvious symptoms of the mild 21-hydroxylase enzyme deficiency, and can live quite normally without any medical treatment. There is no danger of adrenal insufficiency or crisis. Symptomatic individuals may come to medical attention in childhood with early onset of puberty, in the teen years with acne, young women with excess facial and/or body hair, irregular menstrual periods, or in young adult life with fertility problems. These complaints can be remedied with hormone therapy, usually a low dose of cortisol replacement medication. Such

medications include hydrocortisone (favored in treatment of children for the low incidence of side effects), prednisone or dexamethasone. Once a person has been taking cortisol replacement drugs for an extended length of time, the body becomes somewhat dependent on the medications. This means that one's own adrenal gland is "lazy" and cannot respond to stress as well as it might have done before taking the medications. If a patient were to abruptly stop taking cortisol replacement medications after many months or years, adrenal crisis could ensue. Nonclassic CAH patients, because they have only a mild impediment to producing cortisol, can often discontinue their medications, but this should be done gradually under medical supervision, so as not to precipitate adrenal crisis.

Parents and physicians should be aware of clinical clues to the diagnosis of adrenal insufficiency. **Infants may be jittery, listless, or have convulsions due to low blood sugar or blood sodium. These chemical changes can be detected by blood testing. Chronic adrenal insufficiency will result in failure to gain weight. Older children may exhibit fatigue, malaise, muscle aches, headache, abdominal pain, nausea, vomiting, salt-craving, and weight loss with cortisol and/or aldosterone deficiencies.**

In suspected chronic adrenal insufficiency, careful review of growth and weight gain is critical to understanding the nature of the problem. **A severe drop in blood pressure after the person rises from lying down is a sensitive indication of dehydration and potential signal of adrenal crisis. In untreated primary adrenal insufficiency, physical examination**

often shows dark skin

pigmentation, especially of non-sun-exposed areas such as palms, soles, and gums.

Prompt recognition of adrenal insufficiency is the key to a good outcome. People with CAH or any other condition associated with adrenal insufficiency (such as Addison's disease) should always carry medical identification cards or wear medical identification tags to alert emergency medical personnel to their needs. Timely initiation of stress doses of cortisol replacement and salt and sugar intravenous fluid therapy are the most important factors, and are life-saving. At home emergency therapy can be administered, if there will be a delay in reaching the hospital. This can be accomplished with an intramuscular injection of hydrocortisone (25 mg for infants, 50 mg for children, 100 mg for adults), or more readily with hydrocortisone-containing rectal suppositories. The latter are simple to administer and rapidly absorbed, and do not require special equipment or training to give. If the individual is conscious, clear electrolyte fluids should also be provided (sports drinks such as Gatorade or Powerade are ideal). The patient should then be seen by his/her physician, or in an emergency treatment facility. CAH patients and other with adrenal insufficiency should consider carrying a letter from their physician describing their condition and treatment needs whenever travelling.

Primary care practitioners and school health personnel should be informed of necessary measures in case of shock. Classic CAH and Addison's disease are unremitting conditions, requiring lifelong treatment. Nevertheless, most

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Multi-Center Study on Non-Classical CAH in Women of Reproductive Age

Ricardo Azziz, M.D. Chair OB/BYN, Cedars Sinai Medical Center, Los Angeles, CA

Non-classic (a.k.a. delayed or late onset) adrenal hyperplasia (NCAH) due to 21-hydroxylase deficiency affects between 2% and 10% of hyperandrogenic women, depending on ethnicity. Dr. Ricardo Azziz, currently Chair of the Department of Obstetrics & Gynecology at Cedars-Sinai Medical Center, has been studying the epidemiology, physiology, and genetics of the disorder for the past 15 years. Some of the highlights of Dr. Azziz's research include:

1) Determining the prevalence of NCAH in unselected hyperandrogenic women in the Northeast (at the Johns Hopkins Hospital) and Southeast (at the University of Alabama at Birmingham) United States, and Puerto Rico (at the University of Puerto Rico). These studies indicated that the prevalence of NCAH in hyperandrogenic women in the United States is approximately 2%, although it may be lower in the African-American population.

Adrenal Crisis

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patients lead unrestricted and productive lives. Patients receiving low dose maintenance cortisol and aldosterone replacement therapy are not at risk for serious side effects from these medications. There are no significant interactions of these medications with other commonly used drugs.

CF

2) Using prospective studies, the technique of screening for the disorder among hyperandrogenic women with the use of a basal 17-hydroxyprogesterone measurement, was studied. Essentially, a cut-off value of approximately 2 ng/ml for a basal 17-HP level obtained in the follicular phase of the menstrual cycle has a positive predictive value for NCAH of 16%; hormone levels below this cut-off value essentially exclude affected patients.

3) The early genetics of the disorder of NCAH were studied, and a unique mutation (PRO-453 to Ser) was first reported.

4) The risk of clinically-evident hyperandrogenism in carriers (heterozygotes) for congenital 21-hydroxylase deficiency was also assessed and was found to be relatively minimal.

5) The mechanisms underlying the excess androgen production in NCAH, in spite of relatively normal circulating ACTH levels, were studied and indicated that the androgen excess was primarily due to intrinsic defects in enzyme kinetics and to associated ovarian hyperandrogenism.

6) The clinical presentation of NCAH was studied in a large multi-center study and it was found that, contrary to earlier reports, the hyperandrogenic symptoms of the disorder appear to be progressive over time.

7) Other studies also indicted that there appears to be a loose association between the severity of the phenotype (appearance of the disorder) and the severity of the

genotype (type of genetic abnormality).

Dr. Azziz has also coordinated a large multi-center collaborative group for the study of NCAH. The NCAH multi-center international study group includes investigators from the following sites:

- The University of Alabama at Birmingham, Birmingham, AL, USA.
- Instituto Mexicano del Seguro Social, Mexico City, Mexico.
- The University of Palermo, Palermo, Italy.
- Centre Hospitalier et Universitaire de Lille, Lille, France.
- University of Pisa, Pisa, Italy.
- Hospital Sant Joan de Deu, Barcelona, Spain.
- Hospital das Clinicas, Sao Paulo, Brazil.
- Hospital de l'Antiquaille, Lyon, France.
- Centre Hospitalier Universitaire D'Angers, Angers, France.
- North Shore University Hospital, Manhasset, NY, USA.
- Faculty of Medicine of Porto, Porto, Portugal.
- Hospital Ramon y Cajal, Madrid, Spain.
- University of Pittsburgh, Pittsburgh, PA, USA.

For further information about these studies or to participate, please call 310-423-9964.

CF

Evaluation of Clitoral Sensitivity and Viability Following a Modified Reduction Clitoroplasty in 21 Patients

Dix P. Poppas, New York, NY; Michael Chaar, New York, NY; Cathy Kelly, New York, NY; Maria I. New, New York, NY

Enlargement of the clitoris is a prominent manifestation of virilizing congenital adrenal hyperplasia (CAH) and other disorders resulting in ambiguous genitalia. Controversy persists as to the viability and sensitivity of the clitoris following reduction clitoroplasty. To date, no single large group study using a single technique has been evaluated for post-surgical clitoral viability and sensitivity. We report on a modified technique for reduction clitoroplasty based on the understanding of the female clitoral anatomy that results in exceptional cosmetic appearance while providing reliable viability and sensation. Our group evaluated 22 patients who underwent reduction clitoroplasty from 1996 to 2002 using the same technique by a single surgeon. The youngest patient was 5 months and the oldest 24 years with a mean age of 6 years. Eighteen patients had CAH. One patient was a 46 XX true hermaphrodite and two patients were 46 XY and underwent sex

reassignment surgery. Simple modifications to the conventional technique included: total mobilization of the neurovascular bundles (NVB) through parallel ventral midline incisions of Buck's fascia, no reduction or excision of the glans clitoris, irrigation of the NVB with papaverine and subtotal excision of the corpus cavernosal tissue. All patients were evaluated post-operatively for clitoral viability by gross examination and capillary perfusion testing. Of the 22 patients, 8 patients were above 5 years of age and considered candidates for clitoral sensory testing (CST). CST was performed using a cotton-tip stimulator. On a scale of 0 to 5, the patient was asked to report the degree of sensation at various points of the genitalia and inner thigh. Mean follow-up time of these patients was 2 years. Our results in the 22 patients that underwent reduction clitoroplasty have been tremendous. No postoperative complications

have been reported. All patients were confirmed to have a viable clitoris with normal capillary perfusion. Of the 8 patients that were evaluated with CST, all reported a degree of sensation of 3/5 at the inner thighs, 4/5 at the labia minora and 5/5 at the clitoris. These results suggest that the modified reduction clitoroplasty provides a safe, reliable approach to managing the enlarged clitoris. The positive viability and sensation outcomes in the older patients suggest this technique will be successful in the younger children undergoing the procedure. This data supports the belief that clitoral reconstructive surgery using this technique does not result in loss of sensation or viability. Continued long-term evaluation of our patients remains an important focus for our group. We are grateful to our patients and their parents for the continued enthusiasm and shared interest in this work.

CF

Will Restricted Distribution of EMLA® Cream Affect You?

Stephanie Fracassa, New York, NY

Regular blood work is a necessary part of managing CAH. The pain and discomfort associated with venipuncture can be reduced with the use of a topical anesthetic cream. EMLA, manufactured by Astra Zeneca, is one such cream.

On November 15, 2002 Astra Zeneca ceased distribution of EMLA Cream to all Drug Wholesalers and direct buying Retail Pharmacies because it had failed to comply with

regulations regarding Child-Resistant Closure (CRC) packaging. After January 31, 2003 EMLA could no longer be dispensed for use outside of the hospital, in-patient setting.

As an alternative, **ELA-Max (Lidocaine 4%) Topical Anesthetic Cream** is available from Ferndale Laboratories, Inc.

ELA-Max is Prilocaine-free for safe use in children and adults. It is non-toxic with minimal systemic

absorption and less vasoconstrictive than EMLA. ELA-Max delivers safe, effective topical anesthesia in 30 minutes or less **without** the need for occlusive dressings.

ELA-Max is available in a variety of sizes:

- *15-gram tube
- *30-gram tube
- *box of five 5-gram tubes
- *box of five 5-gram tubes with 10

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Why Not Knowing the Implications of my Adrenal Hyperplasia Almost Caused Me to Lose My Testicles

Reprinted with permission from the Melbourne CAH Support Group

The Underlying Condition

I am a 34 yr old male, and I was born with Congenital Adrenal Hyperplasia (CAH). I have been treated for it my whole life.

The Diagnosis

I found a lump on my right testicle. After getting a referral from my Endocrinologist, I went to see a Urologist to have the lump checked. The Urologist felt the lump and decided to order an ultrasound. I had the ultrasound performed and was concerned to see lumps in both testicles.

The Phone Call

I had a very stressful week waiting for a return call from the Urologist. I was finally told to call him Friday morning at 9am. I called my Urologist and he told me I had lumps in both testicles. The lump had spread from the right testicle to the left testicle. He told me I would have to have both testicles removed as soon as possible. Paraphrasing the doctor he told me "This cancer moves fast, we have to get them both off right away". He told me to schedule the surgery and head to the sperm bank if I wanted any chances of kids. I don't currently have any children. I remember sitting at the phone suddenly covered in sweat.

The Concern

Due to the fact I have CAH, about two years ago a CAT scan was taken of my adrenal glands. The CAT scan showed that I had multiple nodules on my adrenals. Not a concern for a CAH patient. Remembering this, I thought why couldn't my disease cause lumps in my testicles too? For some reason I remember being told in

Biology that the testicles and adrenals are made up of very similar material.

The Research

I did some quick searches on the internet. It did not take long to find quite a few articles on CAH males having possibilities of lumps in their testicles. The problem was that the studies did not show it was a common occurrence with CAH males. The best supporting information I could find was that up to 50% of CAH males had been shown to have these lumps. In addition, I don't usually listen to everything I read on the internet. I had to know more before I could dismiss what the doctor was telling me.

The Goal

I scheduled the surgery for my testicles to be removed. The surgery would be performed in two weeks. My goal was to get my Urologist or Endocrinologist to listen to me and understand these may not be malignant tumours. If I did not get the doctors to listen to me, I would go through with the surgery. I did not want to be in denial and regret putting off the surgery.

The Calls

Getting a hold of either my Endocrinologist or Urologist turned out to be a nightmare. It took forever to get either one of the doctors to call me back. Time to the surgery was ticking away and the stress began to build. Finally my Endocrinologist called me. I explained to him about the reports that I had found online. He told

me he found it could be possible, but never actually came across such a thing. He told me that he would talk to my Urologist. He instructed me to ask my Urologist to call him. However, if after the discussion the Urologist wanted to proceed, then it would be between me and my Urologist. My Endocrinologist did give me two second opinion Urologists that I could go see. I scheduled a second and third opinion with the two doctors. Time was running out and I did not want to take any chances.

The Frustration

I was still waiting to hear back from my Urologist. I had left several messages for him to call me and/or to call my Endocrinologist. (By the way, their offices are 3 feet apart.) Another week had gone by and I was getting closer to the surgery. One more week of desperation until the surgery. Finally I got a call through to my Urologist. He admitted the lumps could be caused by my CAH, however, he quoted statistics that 95% of the time lumps in testicles are malignant. He told me he never did get the messages I left him about calling my Endocrinologist. After some desperate pleading with my Urologist, he told me he would only remove the right testicle, do a biopsy during the surgery, and then decide whether to keep the left testicle. This was some progress, however, I still felt uneasy about being asleep during the decision process. My biggest fear was waking up and finding both of my testicles removed.

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The Second Opinion

Luckily I was able to schedule the second and third opinions very quickly. The referral from my Endocrinologist helped speed the process. While waiting for the second opinion Urologist to come into the exam room, I felt the stress taking its toll. Feeling like I was going to pass out was combined with a burning sensation in my stomach. Was this stress or the cancer spreading to other parts of my body? The doctor came in and examined my testicles. He also felt the lump the first Urologist found. I gave him a copy of the study I found on the internet. The best evidence I could find at that point. The study showing up to 50% of CAH males could have benign lumps in their testicles. He quickly read the study and admitted of not knowing about such a thing. A ray of hope shined through. The ray of hope was quickly squelched. The Urologist told me that the functionality of the right testicle was so minimal from the lumps that it should be removed anyway. He basically repeated what the first Urologist told me. The right testicle could be removed and biopsied and then the left testicle could be cut open and examined. His concern was that cancer could be spread during the removal of the right testicle if the supporting plumbing was not clamped off properly. He praised my first Urologist as being a very good Urologist just as his father had been before him. He told me that whatever my first Urologist wanted to do was probably best.

The Last Hope

I finally contacted John Hopkins University in Baltimore. I found a contact on the internet that worked in the Children's Hospital there, more

specifically with children that have CAH. She was a psychologist that had written a book on the subject of dealing with CAH. She co-wrote the booklet with a specialist that worked primarily in CAH research and treatment. Her phone number was the only one published on the website, so I decided to give her a call. I was surprised, shocked that she called back within the hour. Having talked with her associate, she recommended a doctor in the Minneapolis area. Her associate had trained this doctor in CAH and would be the best person to see.

I contacted the doctor that was referred. I mentioned to his receptionist that I was an adult patient with CAH and needed any help they could provide. After she talked with the doctor, I was scheduled to come in the very next day. The next day I went to see Dr. Brown. It did not take long for Dr. Brown to verify the study I had been reading and to schedule me to see another urologist. A paediatric urologist that was familiar with testicular tumours associated with CAH. I was scheduled to see the Urologist the next day.

A Happy Conclusion

I was still a bit nervous, but I was feeling much better that Dr. Brown listened to me and understood that the tumours could be caused by the CAH. The urologist examined my testicles and to my delight announced "It doesn't even feel malignant". I was thrilled and amazed that such a simple exam resulted in such a finding. To be sure he scheduled a testicular biopsy to be performed. He assured me not to worry, he was 90% sure it was adrenal rest material being aggravated by excess androgens.

I called my first urologist and cancelled the original surgery. I went through with the testicular biopsy and was very happy to be told 20 minutes after the surgery that preliminary examination found benign tumours. Further pathological examination verified that the tumours were benign adrenal rest material.

A Road to Recovery

Since the benign tumours were removed it took about a week to recover, however recovery with both my testicles intact. Currently I am seeing Dr. Brown on a regular basis in which he is adjusting my daily Hydrocortisone and Florinef replacement therapy. The hope is to reduce my androgen production as much as possible and restore my fertility.

Name withheld

EMLA Cream

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Tegaderm dressings

*5-gram tube with 2 Tegaderm dressings

ELA-Max is available **WITHOUT PRESCRIPTION** in child-resistant packaging. Look for it at your local pharmacy or purchase online at:
www.dermadoctor.com
www.dermstore.com

Ferndale Laboratories Inc. manufactures ELA-Max Cream. For more information, call Ferndale Laboratories toll-free 1-877-ELA-MAX4 (352-6294) or (888) 548-0900 or go to their website: www.ferndalelabs.com. On their website, there is a press release dated Oct. 11, 2002 that explains why EMLA is no longer available.

CF

Newborn Screening and Legislative Update

Newborn Screening. Once again, CARES Foundation members have been active in advocating for expanded newborn screening including CAH. In a mere 2 months, our members in **Nevada**, lead by dynamo Gretchen Alger Lin, influenced the NV Department of Health to make CAH newborn screening a priority in its expansion efforts. In January, the Department of Health had issued an RFP (request for proposals) for their newborn screening laboratory contracts including an expansion to include tandem mass screening for inherited metabolic diseases, but not including CAH. The Department of Health was so swayed by the efforts of our members that they recalled the initial RFP and issued an amended one that included CAH. Now all of the labs interested in the NV newborn screening contract must include CAH screening in their pricing. What a fabulous victory! But this does not insure that CAH will be screened for at birth in NV. The expansion must be approved by the Governor and receive the funding needed in the budget. So, please write to the Governor and urge him to approve the newborn screening expansion and to fund it generously in the budget. This is time sensitive, so please try to write soon.

Governor Kenny Guinn
State Of Nevada
Capitol Building
Carson City, NV. 89701
(775) 684-5670
(775) 684-5683 FAX
Or to email him: <http://gov.state.nv.us/mailgov.htm>

Another fantastic volunteer, Sue Bianchi from California, has created a web site to make advocacy for expanded newborn screening easy for

the average person. Please visit this web site <http://www.newborn-screening.org>. You can click on the icon for "letter writing campaign" and can email a letter to the Governor of your choice.

Advocacy must be continued in Nebraska, (thanks to our NE families and pediatric endocrinologists for all of their efforts so far in NE); Vermont; New Hampshire; Utah; Oklahoma and the District of Columbia. So, check out this web site and let your state leaders know that newborn screening saves babies' lives.

While Newborn Screening Programs throughout the United States expand their programs, two states – Minnesota and California – face significant worries. The state of **Minnesota** expanded its NBS program to more than 30 disorders last year. Unfortunately, they've met some serious opposition from a consumer advocacy organization that now threatens to jeopardize the entire program.

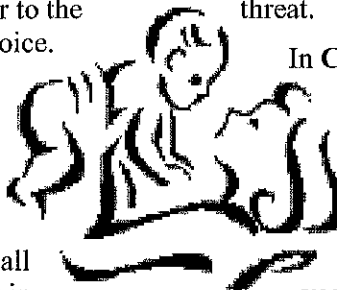
Twila Brase, president of the Citizens' Council on Health Care (CCHC) in St. Paul, MN, has raised loud concerns about patient's rights and the risks of personal genetic information stored in state NBS databases. CCHC opposes newborn screening because they believe that the new HIPAA guidelines do not offer adequate privacy protections. She called newborn screening "defect testing of children". Our MN members jumped right in and wrote to their legislators. They were able to thwart the efforts of this group to undermine Minnesota's newborn screening program.

Thanks to all who wrote letters and made phone calls. Again, your advocacy made a difference and protected MN's program from this threat.

In **California**, budget cuts are also endangering expanded newborn screening. California has the largest newborn screening program in the nation and accounts for over 500,000 births each year. Through a pilot program, this state has been providing expanded newborn screening for those diseases screenable via tandem mass spectrometry (though it does not screen for CAH) to its infants since last July. If California does not receive continued and permanent funding, they will not be able to provide this screening to its infants and will most certainly prevent the state from adding CAH. This would mean that once again, efforts in newborn screening would take a giant step backwards.

Our members from **Michigan** are working to improve CAH newborn screening in their state. Michigan has screened for CAH for many years. However, it has the most restrictive laboratory cut-off levels for CAH in the country. Currently, the goal of its newborn screening program with respect to CAH is to detect only salt-wasting male babies. Every other state that screens for CAH sets its cut-offs broadly to detect all cases of classical CAH—males, females and simple-virilizers. The program director has asked for our help to improve the follow-up of CAH babies, and we are more than happy to work with the state to make improvements in follow-up procedures and resources. But, we

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Newborn Screening Update

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also want to make sure that the state broadens its lab cut-offs so no classical CAH goes undiagnosed in the newborn period. Member Lynn Schroeder is spearheading the effort there.

Other Legislative Issues (from the Genetic Alliance Newsletter, NORD's *Up and Running in the 108th Congress...* and the March of Dimes' *Advocacy Update*):

NIH Funding. Many thanks to our members who wrote letters to their Congressional Representatives to urge them to appropriate generous funding to the National Institutes of Health. For the fiscal year 2003 that began last October 1, the National Institutes of Health will receive \$27.16 billion — an increase of \$3.79 billion or 16.2% over last year's funding, and the final installment in the agency's five-year budget-doubling campaign. This was a confusing budget year as the President's proposed budget for fiscal year 2004 was released on February 3rd, despite the fact that Congress had not completed work on the current's years' budget (FY 03). Fiscal Year 2003 funding was finally approved by Congress on February 14 and signed by the President on February 20, 2003. Now we need to begin our work on Congressional support for FY 2004 funding for NIH so that they can continue their exciting research into new clinical treatments and gene therapy for CAH.

HIPAA Privacy Regulations Went into Effect on April 14, 2003!

The Privacy Rules portion of the Health Information Portability Assurance Act (HIPAA) is the first major federal law to protect the privacy of peoples' medical records in this age of information technology.

The HIPAA Privacy Rules grant consumers a number of significant new rights, although in less sweeping form than hoped for by most patient advocates. It is critical that health care consumers understand their new health privacy rights and that they know what to do if their rights have been violated.

You can learn more about HIPAA and its importance to your medical care by reading a new brochure, *Know Your Rights*, that was created by The Health Privacy Project (HPP) at Georgetown University to make HIPAA more useful to the lay public. To access this easy-to-understand outline of our new privacy rights under HIPAA go to http://www.healthprivacy.org/usr_doc/KnowYourRights.pdf. For a more comprehensive posting that covers about 120 pages, including Q&As, and provides a sense of the overall impact of the regulations, go to <http://www.hhs.gov/oct/hipaa/privacy.html>.

Consumers Call for Bipartisan Genetic Non-Discrimination Legislation – Mark-up by Senate HELP Committee Delayed. On March 31st, the mark-up on legislation to outlaw discrimination in health insurance and employment on the basis of genetic information was postponed for the fourth time. A new mark-up date has not yet been announced. We have been assured, however, that senior staffers as well as Republican and Democratic members of the Health, Education, Labor and Pensions (HELP) Committee are continuing their attempts to bridge the difficult issues that divide them with respect to privacy and enforcement. Consumer groups – the Genetic Alliance, the Genome Action Coalition, the Coalition for Genetic

Fairness and the National Breast Cancer Coalition – have each called for bipartisan negotiations to create a bipartisan bill that has the potential for successful passage on the Senate floor. Keep up to date on genetic non-discrimination legislation by visiting http://www.geneticalliance.org/GND_Respage.html.

Reconstructive Surgery Act of 2003. Inspired by a constituent with a rare congenital disease, Representative Mike Ross (D-AR) reintroduced this legislation. It would require insurance companies that cover surgery to cover medically necessary reconstructive surgeries for congenital defects, developmental abnormalities, trauma, infection, tumors, or disease. The provisions of this bill would cover reconstructive surgery for virilized females with CAH. The bill number is H.R. 1499.

Volunteer Pilot Organization Protection Act. This bill provides liability protection to nonprofit volunteer pilot organizations flying for public benefit and to the pilots and staff of such organizations in order to promote the activities of nonprofit volunteer pilot organizations flying for public benefit and to sustain the availability of these services, including transportation at no cost to financially needy medical patients for medical treatment, evaluation, and diagnosis, as well as other flights of compassion and flights for humanitarian and charitable purposes. The Bill number in the House is H.R. 1084 and in the Senate, S. 955. Volunteer pilots offered their services to transport some of our families to our Baltimore Conference.



Coping with Hirsutism

(Continued from page 1)

Hair growth in humans is asynchronous, meaning that growth and shedding of each follicle is independent of surrounding follicles. The number of hair follicles throughout the body is genetically determined. Men and women typically have the same number of hairs. However, in men, high androgen levels cause hair follicles in the androgen-sensitive areas of the body to produce terminal, coarser hair. In women, in whom androgen levels are typically low, those same hair follicles produce less visible, vellus hair.

EXCESS HAIR

When the delicate hormonal balance of the body is disturbed, hair production, among other things, can be affected. In the case of CAH, excess androgen (e.g., testosterone) production causes excess "terminal" hair to grow in the androgen-sensitive areas of the body. Excess hair can be categorized as either "hirsutism" or "hypertrichosis". *Hirsutism* indicates the presence of excess terminal hair growth in areas of the body that are androgen-sensitive. Androgen-sensitive areas of the body are the face, chest, areola, lower back, buttock, inner thigh and external genitalia. *Hypertrichosis* indicates excess terminal hair growth in areas of the body that are not androgen sensitive. Androgen-insensitive areas of the body include forehead, forearms, tops of the hands.

In women with hirsutism, the hair follicles that would normally produce pale, fine vellus hairs have switched to producing the darker, coarser terminal hairs. On the scalp the effect is the opposite, with hair follicles switching to production of vellus hairs in the presence of high

androgen levels. This is why temporal balding is seen in untreated women with CAH. In general, hypertrichosis is not a problem in CAH since it does not depend on high androgen levels.

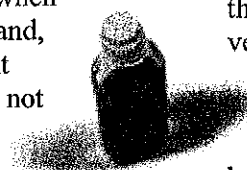
Endocrinologists use the Ferriman-Gallwey (FG) scale to assess the degree of hirsutism in CAH patients. Measuring the degree of hirsutism in 11 areas of the body, the FG scale can range from 0 – 44 (the higher the score, the more severe the hirsutism). A typical score for someone with hirsutism is between 8 and 29. While this scale is somewhat subjective, it does allow the physician to monitor the improvement in hair growth.

TREATMENT FOR HIRSUTISM

In CAH patients, excess androgen levels can cause an increase in terminal hair production. Therefore, the first step doctors take to treat hirsutism is to ensure that the patient is adequately controlled with replacement cortisol (cortef, prednisone or dexamethasone).

MEDICATIONS

Even with optimal replacement of cortisol, however, there are certain times during the day when androgen levels rise, and, therefore, replacement cortisol alone is often not sufficient to combat hirsutism once it has begun. Elevated androgen levels can trigger a condition called "Polycystic ovarian syndrome" or PCO. In PCO, the ovaries become enlarged with multiple cysts and begin to produce androgens. For this reason, if replacement therapy alone is not sufficient to combat hirsutism, you may be tested for PCO and, if found, oral contraceptives would be prescribed.



If hirsutism persists even with adequate replacement therapy and oral contraceptives, drugs which block androgen action may be tried. By blocking androgen action, terminal hair growth decreases with each passing cycle. However significant improvement can take several months to two years because of the cyclical nature of hair growth. If the androgen blocker is discontinued, hair growth will recur.

Spironolactone is an androgen blocker which is also a weak diuretic (a drug that causes excess urination). While taking Spironolactone, electrolytes should be monitored periodically. Cyproterone acetate is another androgen blocker which is used in Europe and Australia but not approved in the United States. Cyproterone acetate also counteracts androgen action and is often combined with ethinyl estradiol (a type of estrogen) which counteracts the androgens produced by polycystic ovaries. The combination of cyproterone acetate and ethinylestradiol is found in Diane and Dianette. These drugs therefore operate as both an oral contraceptive and an androgen blocker at the same time. Contraindications for cyproterone acetate (mainly due to the estrogen action) are varicose veins, uterine fibroids, smoking and cardiovascular disease. Side effects for both spironolactone and cyproterone acetate can include breast tenderness, decreased libido (sex drive), fatigue, headaches, depression, weight gain and irregular periods. In high doses, anti-androgens have been linked to liver toxicity. However, this is usually not a problem at the doses used to treat hirsutism. It is also extremely important that a woman **does not** become pregnant while on these

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Coping with Hirsutism

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medications as they will interfere with the normal development of a male fetus.

Flutamide, another anti-androgen medication, is no longer used by our center as other centers have reported two deaths tentatively associated with its use.

HAIR REMOVAL METHODS

Bleaching

Many women use hydrogen peroxide to bleach their facial hair so that it is less noticeable. The only downside to this method is that sometimes the hair develops a yellow hue.

Shaving

This method offers temporary benefits and can sometimes cause skin irritation and infections. Many women have the mistaken belief that repeated shaving will cause the hair to grow in darker and coarser. After shaving, the only hairs that immediately regrow are the ones in the anagen phase.

Plucking

Plucking stimulates hairs that were in the telogen phase to begin their anagen phase. This means that plucking a hair that happens to be in the telogen phase actually speeds up the re-growth of that hair. Facial hair has a long telogen phase, so it is best to shave before plucking so that the only hairs plucked are already in the anagen phase.

Depilatory creams

The most often used depilatory preparations contain thioglycolates, which target the keratin in hair. Since skin also contains keratin, depilatory creams often irritate the skin,

sometimes causing dermatitis. A new depilatory cream called Vaniqua has been getting good reviews. Vaniqua must be prescribed by a physician.

Waxing

Waxing involves the application of warmed wax to hair-bearing skin. Upon cooling of the wax, hairs are imbedded within the wax and when the wax is pulled away in a quick motion, the trapped hairs are pulled away with it. Waxing can be painful and may cause hyperpigmentation, folliculitis, scarring and, if performed improperly, thermal burns.

Electrolysis

Electrolysis has been an option for over a century and is considered the only permanent form of hair removal. It uses an electrical current to disrupt individual hair follicles in the anagen (actively growing) phase. Since only a percentage of hair follicles are in the anagen phase at any given time, electrolysis must be done over several visits to steadily destroy all follicles in a given area. For instance, removing excess hair from the upper lip and chin could take approximately 18 monthly treatments, with the initial visits lasting longer than subsequent visits. Most patients find it mildly uncomfortable, and some take anti-pain medication (e.g., Tylenol) before their appointment. For the minority of patients who experience electrolysis as painful there is the option of EMLA cream (for which you need a prescription from an MD) or ELA-Max cream (over the counter but more expensive).

Some states require that electrologists be licensed. In these states we advise that you only use those who have a license. However, many states, including New York State, do not require electrologists to obtain a license. In these states it is

recommended that that an electrologist be accredited by the American Electrology Association (AEA) and be a Certified Professional Electrologist (CPE). The AEA hosts a website at www.electrology.com with information about electrolysis and with a search engine to find AEA members in a given area. Questions to ask of an electrologist when choosing one are: Are you a CPE? Do you use disposable probes and do you sterilize your forceps? Is your equipment new (at least within the last 10 years)?

In unskilled hands, electrolysis can cause folliculitis (a painful, red swelling of the hair follicle) and scarring. Typical complications include a mild redness that lasts for about 1 hour afterward, occasional breakouts, and minor temporary scabbing. Many electrologists will require a doctor's note if you have diabetes, are a pregnant woman, are on blood thinners, or have mitral valve prolapse (a common heart condition which usually causes no symptoms and doesn't need to be treated). Electrologists should offer a free consultation (where you can ask all these questions). They have variable fees, and, while electrolysis is not usually reimbursed by insurance, some people have successfully lobbied their insurance companies for reimbursement.

Laser Treatment

Laser treatment is the newest method of hair removal. There is less data on safety. Laser uses light waves to target the melanin (pigment) in the hair follicle and disrupt the follicle bulb. Because skin also contains melanin, light-skinned dark-haired individuals usually have the best outcomes with laser therapy. Like electrolysis, laser only destroys

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Coping with Hirsutism

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hairs in the anagen phase so multiple treatments are required to achieve hair removal in a given area. Approximately 50 – 70% of the excess hair can be removed. With this method follicles do not need to be treated one at a time, and therefore each treatment is relatively quick and usually only mildly uncomfortable.

There are several kinds of lasers used in treating hirsutism. The type of laser is chosen based on skin type. Laser treatment does not cause permanent hair loss since with time, hair tends to re-grow to a variable extent. This is why touch-ups are required on a yearly basis. Complications could include permanent scarring and hypopigmentation (loss of normal skin color); however, based on very short term data, these risks appear to be low if the procedure is done by a trained dermatological surgeon.

At the moment there are no regulations governing the use of laser technology, and in fact it is offered in any number of settings from local spas to beauty parlors. We strongly recommend that laser treatment be done only by a dermatological surgeon. Surgeons should be a member of the American Society of Dermatological Surgeons (ASDS) and have had specific medical training in the use of lasers. The ASDS hosts a website (www.asds-net.org) which contains information about laser hair removal and it provides a search engine which enables the user to find local ASDS approved dermatological surgeons.

Hair Loss

Women with untreated CAH often experience temporal balding due to the action of androgens on the

hair follicles of the scalp. Unlike the other androgen sensitive areas of the body, the hair follicles of the scalp respond to high androgen levels by making vellus (soft, pale and fine) hair instead of the usual terminal hair found on the scalp. Usually, when the underlying hormonal imbalance is treated with replacement cortisol (i.e., Cortef, prednisone or dexamethasone), women have a fairly rapid improvement in scalp hair re-growth.

In Summary

In summary, there are many ways to battle hirsutism or hair loss. The first and most important step is to consult your endocrinologist and make sure that the replacement medication you are taking is adequate. Then, if hirsutism is the problem, discuss the various options presented here with your endocrinologist and/or dermatologist and find a treatment plan that you are comfortable with. If you chose electrolysis or laser therapy, your doctor may be able to recommend a well trained local professional. If you have found an intriguing suggestion on one of the message boards (see newsletter number 7), discuss it with your doctor before jumping right in. What works for one woman, may not be the treatment of choice for someone else.

GLOSSARY

Androgens: Male sex hormones, made in the testes in men, ovaries in women, and the adrenals in both men and women.

Anagen phase: the period when hair is actively growing. Protein and Keratin are continuously made in this phase.

Catagen phase: a transitional period where the hair undergoes chemical and structural changes and ultimately stops actively growing.

Cyproterone acetate: a medication that blocks the action of androgens such as testosterone; found, in combination with ethinylestradiol in the drugs Diane and Dianette. (Not approved for use alone in United States.)

Diuretic: a medication which usually causes excess urination

ELA-Max cream: a topical analgesic (painkiller) which can be purchased over the counter.

EMLA cream: a topical analgesic (painkiller) which can be purchased with a prescription. While not currently available, EMLA should be put back on the market in late '03.

Ethinylestradiol: a form of estrogen found in some oral contraceptives.

Ferriman-Gallway scale: a method of rating the severity of hirsutism on a scale of 0-44 with a score of 44 indicating the most severe hirsutism.

Folliculitis: Inflammation of the hair follicle. Inflammation involves redness, swelling and pain.

Hirsutism: excess terminal hair growth in response to high androgen levels.

Hypertrichosis: excess terminal hair growth in parts of the body not androgen sensitive.

Hypopigmentation: loss of normal skin color. Hypopigmentation is often permanent.

Keratin: a protein found in both hair and skin.

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Coping with Hirsutism

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Melanin: dark brown to black pigment (color) found in both hair and skin.

Mitral Valve Prolapse: a common condition involving the mitral valves of the heart. Most people with mitral valve prolapse have no symptoms and never need treatment but, if the condition is diagnosed, antibiotics are usually prescribed before surgery or dental work to prevent the possibility of infection of the heart.

Polycystic Ovarian Syndrome (PCO): a condition, often caused by excess circulating androgens, involving enlarged ovaries with multiple cysts. Polycystic ovaries usually produce excess androgens too.

Spirolactone: a medication which blocks the action of androgens such as testosterone

Telogen phase: the final phase of the hair growth cycle where the hair has completely stopped growing and enters a resting phase prior to falling out.

Terminal hair: dark, coarse and thick hair

Vellus hair: fine, pale and silky hair

If you have any questions or issues with any of the information presented in this newsletter, please call Ann Carlson, genetic counselor, at (212) 746-3495, or Dr. Susan Baker, psychoendocrinologist, at (212) 746-3481.

CF

New CARES Medical and Scientific Board Advisors!

DEBORAH P. MERKE, M.D.

Pediatric Endocrinologist, National Institutes of Health

Dr. Deborah Merke is Chief of Pediatric Services for the Clinical Center of the National Institute of Child Health and Human Development (NICHD). Her primary research interests have focused on studies of the pathophysiology and treatment of congenital adrenal hyperplasia (CAH), an autosomal recessive disorder of the adrenal gland.

Dr. Merke also maintains a joint appointment as a clinical investigator in the Pediatric and Reproductive Endocrinology Branch of NICHD. Central to her work is the study of the pathophysiology and treatment of congenital adrenal hyperplasia. The goal of a group led by Dr. Merke is to understand the mechanisms involved in diseases of the adrenal gland. Insights into the mechanism are used to generate hypotheses for new approaches to treatment.

CLAUDE MIGEON, M.D.

Pediatric Endocrinologist, John's Hopkins Hospital

Dr. Migeon received his M.D. from the University of Paris (France) and received Pediatric training at the Hopital des Enfants Malades/Necker. He came to Johns Hopkins Hospital in 1950 as a Fulbright Fellow and support of the American Field Service.

From 1950-52, he was Fellow-in-Residence in the group of Lawson Wilkins. This was the time when CAH patients were treated with cortisone and salt-retaining hormones for the first time in the world. From 1952-55, he learned steroid chemistry and biology at the University of Utah in Salt Lake City under the direction of Dr. Leo T. Samuels. In 1955, he returned to Hopkins to direct the laboratories of Dr. Wilkins.

For 20 years (1974 – 1994), he was Director of the Pediatric Endocrine Clinic at Hopkins. During the past 50 years, he has been involved in many aspects of Endocrinology, particularly gonadal and adrenal function. With more than 100 Fellows trained in the clinic, he published about 400 scientific papers.

At this time, he continues to treat children with endocrine problems and he is involved in the study of follow-up to patients with intersex and with CAH.

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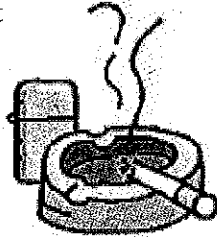
Physician Listings Available from CARES

CARES Foundation has compiled a large list of pediatric endocrinologists, some adult endocrinologists, urologists and psychologists with experience in treating CAH/NCCAH patients. Please contact CARES Foundation for more information.

Two Adrenal Studies by Yale University

Effects of Gender and Post Traumatic Stress Disorder (PTSD) on Executive Function During Smoking Withdrawal

Research has shown that new or infrequent smoking activates the adrenal gland, while heavy regular smoking tends to suppress reactivity of the adrenal gland. When the adrenal gland is activated, persons with cortisol synthesizing enzyme defects may shunt chemicals normally used to make cortisol into other pathways. Some of these pathways produce steroids that increase arousal and anxiety, while other pathways produce steroids that decrease arousal and anxiety. Therefore, smoking patterns may vary, depending on where an adrenal gland enzyme block is located. Some people may smoke heavily to suppress the adrenal gland and decrease the production of certain steroids that induce arousal and anxiety. Others may smoke less often, but regularly, to increase the production of steroids that increase arousal or reduce depression. In either case, smoking is used to improve mood and cognition and therefore may be difficult to stop. Research studies are therefore being conducted to see how common alterations in adrenal steroid production influence mood and cognition during smoking and smoking withdrawal. It is hoped that this research will lead to development of new techniques to improve smoking cessation success as well as treat mood and anxiety disorders in persons with alterations in adrenal gland function.



HPA Axis Function in Men and Women with Chronic Post Traumatic Stress Disorder (PTSD)

Women and men between 18 and 55 are invited to participate in a Yale/VA research study about the way that traumatic events, such as rape or incest, domestic violence, criminal assault, natural disasters, motor vehicle accidents, etc. change the way the stress hormone system functions. Subjects with post-traumatic stress disorder and healthy control subjects with no medical or psychiatric problems will be needed. You will be paid up to \$485 for participating. If you have a trauma history, a comprehensive evaluation for post-traumatic stress disorder is included. If you are interested, please call and leave a message at the VA National Center for PTSD Research Office at 932-5711, ext. 4447.

VA Connecticut Healthcare System
Protocol AR#0004 and Yale School of
Medicine HIC#12819.

This study, and other related studies conducted through Yale University and the West Haven Veteran's Hospital are examining the impact of adrenal functioning on stress and reaction to major stress experiences. Specifically, they are looking for carriers of CAH gene mutations to participate. They are seeking healthy subjects and subjects who have suffered major life stresses (whether you have had resulting emotional distress or not). This is a good chance for carriers of CAH (for example, parents of an affected child) to help research that is looking at the adrenal glands' role in dealing with stress, anxiety disorders and depression.

Healthy smokers, 18-55 years, needed for research on effects of 24-hour smoking withdrawal on brain function. Pays up to \$300. Call Marty: 203-932-5711x4447. YaleHIC#19974, VAHSS#AR0011. Carriers of CAH who smoke especially needed.

Doctors and Nurses Needed!!

We are looking for volunteers to help us respond to the growing number of emails and phone calls we are getting from new families with CAH. Specifically, we are looking for parents or relatives of CAH individuals who are medically trained or are otherwise well versed in the medical aspects of CAH. If you can spare just a couple of hours a week, please consider volunteering your services by making a direct impact on other peoples lives.



CF



Parent Tips!!

(solutions for common problems)

ADMINISTERING MEDICINE TO AN INFANT

I have the solution for easy medication administration! It is called "The Medicator" by *Munchkin*. I found it at Wal-Mart, but I've seen them at other places too. My daughter is nearly 3 yrs. old and I still use it, just because it is so easy. (Unless, a child will chew or swallow the pills, this is the next best thing.) It is a small 10 ml. container connected to a screw-on nipple (like the nipple on a bottle, not a pacifier). Since the Cortef and Florinef dissolve so easily, I just put the pills in the little container and put a little clear juice in it. (I use clear liquid so I can see if all of the particles have dissolved.) If you leave the pills in the liquid long enough, they will dissolve completely, but if you don't wait, then I have a solution for that too! I took the plunger out of a syringe (like an extra syringe you may have for the solu-cortef injectable) and the flat part of it fits perfectly down into that little medicator for crushing those pills! No medication is lost from crushing the pills in a crusher, then trying to put that powder into another container to give it to the child. And this can be used for very small infants, so you don't have to worry about the medicine dribbling out of a baby's mouth, like you do if you are squirting it in their mouth with a syringe. Here's a tip though-Don't put any other kind of medication in this

dispenser!! You don't want your child to associate something that tastes bad with this particular dispenser. So, find a different kind of dispenser for other meds such as Tylenol, Benadryl, etc. Hope this helps!

Gina Murray, Mississippi

BREASTFEEDING AN INFANT WITH SALTWASTING CAH

My 7 week old son was diagnosed with SWCAH at 13 days of life. My biggest hurdle since his diagnosis was getting salt into him. I had made the choice to breastfeed before I ever even conceived my son and wasn't willing to give it up without a fight. I tried everything I could think of short of pumping all his feedings to get the salt down. Nothing seemed to work. Without fail he would get horribly congested and end up with salty milk coming out his nose. In the end I decided to post a message on the CAH main message board.

I got tons of responses, but very few from breastfeeding moms. Most the responses suggested I break it up and mix it in with all of his bottles. Not much help for a breastfeeding mom. I did have one response from another mother of a breastfed baby who suggested I try wetting my finger, dipping it in the salt, and wiping it in the baby's mouth. My son didn't like it, but the

salt went down and the whole ordeal was over in 5 minutes. This worked for about a week at which time he started spitting the salt out as soon as it was wiped in his mouth. So, it was back to the drawing board (and message board) again. Among the responses to my post was a message from a mom who now pumps all her bottles and mixes a little salt in each. She mentioned a method that hadn't worked for her in the post as well. She said she tried dissolving the salt in water and using a dropper to sneak it in her son's mouth while he was on the breast. I wasn't ready to pump all my son's feedings yet, so I thought I'd give the dropper a try. Once again it wasn't my son's favorite thing, but it did go down and has continued to since. One thing this ordeal has taught me is to be open to suggestions. If it weren't for people sharing their experiences, I might still be struggling with worry and frustration. For now I can relax a little and enjoy my son instead of constantly looking at him for signs of crisis. That said, I hope my experience can help one of you.

Elise Jaudon, Florida

If anyone is interested in sharing their ideas/solutions with other families, please send us an email or a letter and we will print in our next newsletter. The CARES newsletter is published three times a year.



Financial Assistance Available

Often, the most experienced physicians/surgeons are at a great distance from the homes of CAH patients and seeing them requires travel and lodging expenses. CARES Foundation offers small grants to families who have legitimate financial need to help cover the costs of travel for this purpose. CARES has also negotiated reduced rate rooms at the Helmsley Hotel in New York for families needing to travel to Manhattan for specialist care. Visit our website for more information about travel assistance for medical care.



Genetics Home Reference Web Site

Making Genes, Chromosomes & DNA Easily Understood (from NIH)

(Bethesda, Md.)--When you hear "gene map," do you think it's a guide to finding the nearest Gap store? Are you the kind of person who thinks that "genetic markers" are sold at office supply stores?

Now, thanks to the National Library of Medicine (NLM) you can find answers to your genetic questions. With the click of a mouse you can go the NLM's newest consumer web site, "Genetics Home Reference," at <http://ghr.nlm.nih.gov>. Genetics

Home Reference joins Medlineplus.gov (the consumer site for general medical information) and Clinicaltrials.gov (the site that lists clinical research trials) in the lexicon of NLM's consumer medical web sites.

To read the entire article visit: http://www.nlm.nih.gov/news/press_releases/GenhomeRefPR03.html.



Pharmaceutical Patient Assistance Programs

The organization called PhRMA has a list of patient assistance programs offered by each drug company for prescription drugs at <http://www.phrma.org/pap/>. If you cannot pay for medication, these programs can help.

On that same internet page in the right hand column, it has links for other types of government supported and private assistance programs.



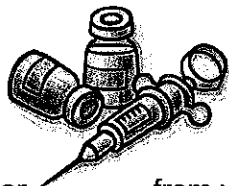
More Volunteers Needed!!

We have invitations to host Family Workshops in Northern New Jersey, Los Angeles, Dallas and Indianapolis. We are looking for volunteers in these areas to help coordinate these events. As a volunteer-run organization, we can only provide these conferences with support from you. If you would be willing to help arrange a Workshop in one of these locations or elsewhere, please email Kelly at: kelly@caresfoundation.org or call on the toll-free line. Please consider volunteering.



When your child is facing adrenal crisis, there's *no time* to waste... **You NEED to be ready....**

Keep **SoluCortef** nearby
AND LEARN HOW TO USE IT!
(also ask your doctor about **Anusol-HC** suppositories)



As a back-up, keep a letter from your doctor outlining emergency procedures.

Visit the Adrenal Crisis page on our website for a sample emergency letter:

<http://www.caresfoundation.org/adrenalcrisis.html>

To all our CAH Adults and Families:

We are trying to create a workable database with the full names and addresses of the CAH community. *Please help us to help you.* For many of you we only have a first name and email address. If you haven't already done so, *please register on our database at:* <http://www.caresfoundation.org/form.html>.



CARES Contributors October 1, 2002 – April 30, 2003

Thank you to all of our wonderful contributors – your financial encouragement means so much as we work together to make a brighter tomorrow for the greatest gift of all — our families!

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We rely on your commitment and generosity. If you would like to make a tax-deductible contribution, please make your check out to: CARES Foundation, Inc. and mail to: PO Box 264, Short Hills, NJ 07078

CAH Family Support Groups Around the Country

ALABAMA

Contact Susan Davenport
205-665-1934
susand@sepcousa.com

ARIZONA

Contact Michelle May
480-759-0870
michlmay@aol.com

INDIANA

Jennifer Lynn
317-823-1317
jenannlynn@aol.com

Tentative plans for *CAH Family
Support Group Event: June
28th, Indianapolis, IN*
Call Jennifer for more
information.

MISSISSIPPI

Susan Aycock
601-833-
8373
5HAycock822@aol.com

New

TEXAS

Contact Sandra Billings
281-861-6043
billprop1@msn.com

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(414) 645-0782
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CARES Foundation, Inc.

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