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ADRENALECTOMY FOR CAH

Summary Research by

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Over 50 years ago Dr. Lawson Wilkins demonstrated that the administration of glucocorticoids (such as cortisol or cortisone) to patients with congenital adrenal hyperplasia (CAH) could suppress ACTH (the pituitary hormone that stimulates the adrenal cortex to overproduce virilizing hormones). Prior to then, most patients with the severe salt wasting form of CAH died at an early age, and females with the less severe forms of the disease became masculinized with deep voices, acne and amenorrhea. Since then, substitution therapy with adrenal steroids has saved the lives of countless patients and spared most of them from progressive virilization. Physicians with long experience in treating these patients, however, are often frustrated by difficulties in maintaining satisfactory adrenal suppression with physiologic

dosages of adrenal steroids. When these patients are then given sufficiently high dosages to halt their progressive virilization, they display the many features of hypercortisolism including progressive obesity, growth arrest, and many other features that lead to poor self-image. Affected females have pregnancy rates that are far below normal, partly because of the psychological and anatomic problems that make heterosexual attachments difficult, and partly because their elevated progesterone levels act as contraceptives. It is often not clear how many of these problems are due to poor compliance with prescribed therapy and how many are due to our limited ability to control ACTH secretion through the feedback mechanism.

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CAH Conference

October 18, 2003
Children's Hospital of Los Angeles (CHLA)

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

For conference details go to: www.caresfoundation.org
or call 1-866-227-3737

A Message from the Executive Director:



Dear Friends,

What a full summer it has been at CARES Foundation! In June, I attended the **2003 Endocrine Society's Annual Meeting** and the first meeting of the **Androgen Excess Society**. These meetings were incredible! I am so excited by the formation of the Androgen Excess Society, a professional society for physicians who have a clinical interest in androgen excess disorders. This group will focus research on these disorders and should add significantly to the body of knowledge on them and encourage further research. The Endocrine Society Meeting was amazing! I would guess that over 10,000 people attended. It took over the entire Philadelphia Convention Center for 4 days. I attended all of the lectures related to CAH and PCOS. The tapes from these meetings are available for sale (*see pg. 18*). Three of our medical advisors were key speakers—Dr. Deborah Merke, Dr. Maria New and Dr. Ricardo Azziz. Dr. New also received the prestigious Koch award from the Endocrine Society (*see pg. 14*). I met many of the physicians that care for the CAH community there. I left so excited by all of the new developments and research that will benefit those affected by CAH. We are fortunate to have such dedicated physicians studying CAH.

I also sat in on the meeting of the **Nonclassic Adrenal Hyperplasia Cooperative Group** that met in conjunction with the Endocrine Society meeting. This group cooperates on international multi-center studies on nonclassic CAH. This group has been studying the

epidemiology, physiology and genetics of NCAH for over 15 years. It was wonderful meeting this dedicated group of physicians and learning how CARES can help further the research on NCAH through this multi-center study. We look forward to cooperating with them on future studies.

In August, I attended the **Genetic Alliance Conference** in Virginia. It was fascinating to see what other disease support groups are doing for their communities. I came back with so many great ideas! It was an inspirational weekend. One thing that some groups are doing is collecting their own data from their families—blood and tissue banking and collection of epidemiological data, and then offering this data to researchers. This would allow us to help direct the research and ensure that the research is aimed towards improved health outcomes for those affected by CAH. CARES is investigating these programs, and we will keep you informed of our investigation.

CARES also nominated **Pfizer, Inc.**, the manufacturer of Cortef, Solu-Cortef and Deltasone, for a Genetic Alliance award, and Pfizer was chosen. Pfizer received the Art of Industry Partnership Award for its formation of the New Jersey Health Partnership (NJHP). The NJHP is an alliance of New Jersey voluntary health organizations, pharmaceutical industry representatives and other state and local healthcare groups. It was

formed to facilitate the exchange of information about issues of mutual concern and interest. The NJHP serves to promote strategies for ensuring accessible, quality, affordable healthcare for all New Jersey residents. I am fortunate enough to serve on its board and to see the benefit that this organization brings to organizations like CARES Foundation. We were so pleased to

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Message from the Executive Director

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help Pfizer, Inc. obtain the recognition it truly deserves for creating and supporting the NJHP.

I just returned from **Indianapolis, Indiana** where Jennifer and Mark Lynn and Penny Barrett organized the first CARES Indiana Support group meeting, with the help and support of Riley Children's Hospital. Over 50 parents attended the meeting held at the beautiful facilities at Riley Children's Hospital. Dr. Richard Rink, a Pediatric Urologist and Dr. Erica Eugster, a Pediatric Endocrinologist gave terrific presentations and answered questions. I met so many of the parents I have only known through email or over the phone. What a phenomenal day! I want to thank The Lynn's and Barrett's, Dr. Rink and his staff, Shelly King, R.N. and Trena Brim, and Dr. Erica Eugster and her staff, Nancy Bulcher, for all of their help, and the Indiana Dept. of Health, Newborn Screening Dept. for notifying the IN CAH families about the meeting. If you would like to start a support group in your state, please contact us.

I also participated in a **new clinical trial through Yale University** in New Haven, CT on carriers of CAH--all of us parents! This trial is looking at how carriers of CAH process certain steroids and the impact that this unusual processing has on our reaction to stress. It was painless--it entails two days of your time (you can schedule when you are available), and they give you a full comprehensive physical. Plus they pay you for your time. So, you benefit research for our community, get a free comprehensive

exam and get paid! If anyone is interested in this, I highly recommend it. Plus, it was fascinating learning about how carriers process these bodily chemicals differently from non-carriers. Email the researcher, Dr. Ann Rasmusson for more information at ann.rasmusson@yale.edu or call her assistant, Valinda Fox at 203-932-5711x4123.

We also welcome our new members of the **Board of Trustees**, Bonnie Stevens, Mark Engman and Dr. Diane Snyder, and the newest member of our **Scientific and Medical Advisory Board**, Dr. Scott Rivkees of Yale University! These wonderful and energetic additions to our boards will help us to move CARES Foundation forward in its goals.

Don't forget about the **October 18th Conference** in Los Angeles! I can't wait to meet our west coast members! I hope you can come!

Warmly,
Kelly



Kelly Leight with Pfizer, Inc. representatives, Joel Hastings and Kirstin Thompson at the Genetic Alliance Awards ceremony.

More on the LA Conference!!

Take a look at the topics that our panel will cover:

- ◇ New Advancements in CAH/NCCAH Treatment and Future Trends
- ◇ General Treatment of CAH/NCCAH
- ◇ Prenatal Therapy
- ◇ Psychological Aspects of CAH
- ◇ Surgical Reconstruction
- ◇ CAH/NCCAH in Women of Reproductive Age and Reproductive Issues
- ◇ Newborn Screening for CAH in California--Advocacy and Action
- ◇ Question & Answer
- ◇ Discussion (Networking)

Are you interested in attending the conference, but financial constraints are limiting you right now? Call us and let us see how we may be able to assist you. Through a generous grant from the **Alexander M. and June L. Maisin Foundation**, we have some financial aid available to individuals and families in need. Call us at 1-866-CARES37.

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Kelly Leight, center, with Mary Davidson, Executive Director of the Genetic Alliance and Dr. Francis Collins, Director of the National Human Genome Project at the Genetic Alliance Conference, August, 2003.

Adrenalectomy for CAH

(Continued from page 1)

In 1996 we suggested that certain patients with CAH would profit from bilateral adrenalectomy, because children with Addison's disease (a condition in which the adrenal glands are destroyed by disease) present many fewer problems in management than do many patients with CAH. Since then, we have carried out adrenalectomy in 3 young children who were proved by genetic analysis to have no possibility of making beneficial amounts of adrenal steroids. These were part of a study to compare prophylactic adrenalectomy with conventional treatment in such severely affected children. In addition, we have identified 15 other patients with CAH who, during the past 7 years, were subjected to bilateral adrenalectomy. Thirteen of these patients have been reported as case reports in the literature. The average duration of follow-up of these 18 patients was 59 months, representing an aggregate of 90 post-operative years. This, study, which was reported in the July 2003 issue of the *Journal of Clinical Endocrinology and Metabolism*, is the first long-term follow-up of patients with CAH treated by bilateral adrenalectomy.

Nine of the 18 patients were 8 years of age or younger at the time of surgery, and the others ranged from 14 yrs. to 44 yrs. Sixteen had 21-O hydroxylase deficiency, one of whom was late onset, and the other two had 11 β -O hydroxylase deficiency. Our 3 young patients were adrenalectomized prophylactically as part of an approved research protocol. The others were adrenalectomized because attempts to keep their adrenals suppressed had proven

ineffective, and they were showing signs of androgen excess as well as obesity and other signs of hypercortisolism.

Safety of the Operative Procedure:

Thirteen of the patients were adrenalectomized by laparoscopic surgery and five had traditional flank incisions. No notable operative complications were encountered. The patients operated on by laparoscopy had low post-operative morbidity and were discharged from the hospital within 1-4 days.

Responses to Adrenal Crises and Other Illnesses:

Our major emphasis in this review was to identify any immediate or long term adverse effects resulting from adrenalectomy. There were no deaths. Five patients had one or more incidents of crisis or other serious illness at some time following surgery, but all responded well to proper therapy.

One of the teenagers had an episode of "adrenal crisis" from laxness in taking medication. She responded well to regular medications.

A 30 yr old woman with Late Onset CAH (nonclassical, in the first 3 postoperative months lost 13 kgs. secondary to lowering of her cortisol dosage. She then had an adrenal crisis associated with a urinary track infection. She had no further problems after adjustment of her replacement medications.

One of the patients survived 2 crises postoperatively. She was poorly compliant and a known drug abuser who had suffered repeated crises before her adrenals were removed. Adrenalectomy had been performed because an adrenal

tumor had been suspected.

Two of our younger patients had serious illnesses postoperatively. An 8 year old girl, who had been well controlled preoperatively on a hydrocortisone dose of 20 mg/M² (20 milligrams per square meter of body surface) had been reduced to 8 mg/M² postoperatively. Three months after adrenalectomy she developed acute gastroenteritis, missed her evening meal, and probably vomited her evening dose of hydrocortisone. The following morning she was found comatose in bed with severe hypoglycemia. No further episodes of hypoglycemia have occurred after her daily dose of hydrocortisone was increased from 8 mg/M² to 12 mg/M² and the need for increased doses during stress was reemphasized. She has been left, however, with epileptic seizures that are controlled by appropriate medication.

The youngest patient of this series, who was adrenalectomized at 16 months, also had a short episode of hypoglycemia with seizures during an illness in which she had been febrile for 4 days and had fed poorly. At that time her baseline dose of hydrocortisone was 8 mg/M². While on the lower doses of HC she consistently had elevated levels of ACTH and 17OH-progesterone. Her ACTH and 17-OH-progesterone levels quickly fell when her HC dosages were increased to 11-13 mg/M². She is now a healthy child leading a normal life 6 years after adrenalectomy.

The post-operative dosage of 8 mg/M² in these patients (and in some of the others) was chosen because this has been reported to be the normal daily secretion rate of cortisol from normal adrenal glands. We have now learned that this dose is insufficient, possibly because of

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Adrenalectomy for CAH

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inherent inefficiencies in absorption from the intestine.

Activation of Ectopic Adrenal Tissue

Hyperpigmentation and elevated ACTH levels were observed in over half of the patients. This was often the result of attempts to reduce hydrocortisone (HC) dosages to 10 mg/M² or lower. When the dose of HC was increased to 11-13 mg/M² the serum ACTH levels fell to normal and pigmentation disappeared. ACTH levels proved to be the most reliable indicator of substitution therapy, since ACTH levels rose to abnormally high levels when the dosage of hydrocortisone was too low.

One might anticipate that no adrenal steroids would remain after both adrenal glands were removed. We were surprised to find that significant elevations of steroid precursors remained postoperatively in 8 of the 18 patients. This incidence is probably an underestimate because postoperative steroid levels were not routinely measured in most patients. Presumably these circulating steroids arose from abnormally located adrenal tissue. Ultrasound studies have revealed ectopic adrenal tissue in 30% of testes in boys with CAH. Previous studies have documented adrenal tissue in the broad ligament of the uterus, along the spermatic cord, and in the celiac plexus. Although such adrenal remnants theoretically have the potential of eventually nullifying the beneficial effects of adrenalectomy, re-suppression of the adrenals in this series has not proven difficult, and recurrent virilism, if present, has been far less of a problem than it had been prior to surgery.

Benefits of Adrenalectomy

These patients and their parents were nearly unanimous in their enthusiasm for adrenalectomy. In virtually all patients, signs of androgen excess have greatly lessened, and although obesity has not miraculously disappeared, it has almost uniformly become less of a problem following reduction of the glucocorticoid dosage from the very high pre-operative levels. Many of the patients and their families commented on their relief from the need for frequent monitoring.

A 28 year old infertile woman with oligomenorrhea resumed menstruation and gave birth to a normal female child. Chabre reported excellent results in a man with 11 β OHase deficiency who had developed severe hypertension. Adrenalectomy was carried out at the age of 44 because medical control of the severe hypertension had become increasingly difficult, and adrenal suppression could not be achieved without producing severe manifestations of hypercortisolism. Now, 4 years following adrenalectomy, his blood pressure is normal, and he faults his physician for not having removed his adrenals earlier.

The initial patient in this series was 3 years old at the time of her adrenalectomy. We have previously reported that prior to surgery, exogenous ACTH caused sodium loss in this patient, whereas ACTH caused sodium retention in her normal twin. This supported our thesis that in many patients with CAH the adrenals do more harm than good. Our patient has thrived in the 6 years since adrenalectomy. At the time of this report, she is a 9 year old 4th grader living a normal

life. Mother responds quickly with stress doses of hydrocortisone in response to respiratory infections. She now returns to endocrine clinic only twice a year, more out of habit than necessity.

Discussion

Objections to adrenalectomy in CAH have been based primarily on surgical risk and deprivation of the protective actions of the adrenal. Although adrenalectomy by any procedure should not be taken lightly, the operative procedure carries no unacceptable risk if adequate provision has been made for exogenous steroids. With modern laparoscopic procedures the postoperative morbidity is very low, and in the hands of skilled laparoscopic surgeons, the laparoscopic approach is clearly preferable to the open flank approach.

Following adrenalectomy we have found that glucocorticoid therapy can generally be maintained at replacement dosages that are lower than the suppressive dosages that were often required before adrenalectomy. Mineralocorticoid supplementation with mineralocorticoids such as fludrocortisone is mandatory, but dosages rarely need adjustment, and can be easily monitored by infrequent renin measurements.

Bilateral adrenalectomy removes the adrenal medulla as well as the cortex. This deprives the patient of

(Continued on page 8)

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This newsletter is published 3 times a year.

The Other Side of the Stethoscope: Communicating with Your Child's Doctor

by Michelle May, M.D.

Nine years ago, I found myself in the unexpected position of being on the “other side of the stethoscope.” As a Family Physician, I had helped many patients and families through difficult diagnoses and had celebrated many recoveries. With one healthy son and an uneventful pregnancy, we were stunned when we learned that our daughter Elyse had Congenital Adrenal Hyperplasia. Since then, we have had to navigate the medical system like many other families affected by chronic disease. However, knowing how things work from the inside has made it a little easier. I would like to share a few things with you as a CAH parent who happens to be a doctor too.

First and foremost, remember that you and your child's doctors are all on the same team, with the same objective – achieving the optimal health of your child. Good communication among team members is critical to ensuring that this very important goal is accomplished.

An important member of the team is your child's primary care physician, either a family physician or a pediatrician. Your child's primary care physician will do routine check-ups and immunizations, and provide care for acute illnesses and injuries. However, when your child has a relatively rare condition like Congenital Adrenal Hyperplasia, which affects only one in 15,000 children, it is possible that your primary care physician may have only read about this disease. This

does not imply that he or she is not competent to care for your child. It simply means that one of your roles on the team is to help educate them about the details of CAH. In this computer age where medical information is so readily available, I have found that my patients with unusual medical problems often know as much, if not more, than their doctors about their particular condition. On the first visit, respectfully offer an article about



CAH and a copy of the “illness guidelines” your endocrinologist has given you so that everyone on the team knows exactly what is to be done in the event of fever, serious illness, or injury.

Due to the complexity of Congenital Adrenal Hyperplasia, most children have a pediatric endocrinologist who will primarily deal with CAH related matters such as laboratory monitoring and medication dosages. They should communicate regularly with your primary care physician about their findings and recommendations. Girls with CAH will also likely have a pediatric urologist if there are genitourinary issues.

You and your child should feel comfortable and confident with the physicians you have chosen. Clinical expertise, effective communication skills, and a warm bedside manner are essential. Likewise, since you may interact frequently with each physician's staff, you should feel that they are competent and efficient. If you

have any concerns in this area, address them promptly with the doctor or office manager; physicians depend on their staff but may be unaware of problems.

It helps to clarify your understanding of the physician's office policies to avoid unnecessary breakdowns in communication. There are a few things you may wish to ask the staff and/or physician: How far ahead should routine appointments be scheduled? Are same day appointments available for urgent problems? What about after hours? How are phone calls handled? Will my child usually see the same physician or will they see other physicians, physician assistants, or nurse practitioners? Even procedures like insurance billing and referrals should be clear to prevent inconvenience and disruptions in the best possible care of your child.

Prepare for the first visit by gathering your child's immunization record, a list of their medications, and details of their medical and family history. If necessary, arrange to have previous medical records transferred to the new office. You will also need to bring your insurance information to each visit.

To help make sure adequate time is available to address your concerns, be specific when scheduling appointments for your child. Most offices book the length of the appointment based on the type and number of problems. For example, if you schedule your daughter for an ear infection, but decide to ask about a study you read on monitoring bone age in CAH patients while you're

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Communicating with Your Child's Doctor

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there, don't be surprised if the doctor seems rushed or asks you to schedule another appointment. (You wouldn't schedule an appointment with your hairdresser for a haircut and expect to get a perm too, would you?)

Some people find it helpful to make a list of concerns and questions before each office visit. Be sure to bring your list but address the most important ones first. This will ensure that those are taken care of, while the less urgent ones can be reserved for another visit if necessary due to time constraints.

Family physicians and pediatricians are used to having more than one child in the exam room, but if you feel distracted by having your other children present, it may be difficult for you to pay full attention during the visit. Likewise, asking questions about another child in the family or asking the doctor to "take a quick look" at another child's ear or rash, not only detracts from the appointment time set aside for the first child, but without the second child's chart, review of their medical history and good documentation cannot occur.

Understand that while most physicians strive to be on time, unexpected situations do occur. Remember that at some point in the future, your child may require extra time while others have to wait. However, if you feel that your wait times are always excessive, discuss the problem openly with the physician or office manager. If the problem cannot be corrected, or if your schedule does not allow that flexibility, it is best to find a new office, rather than always feeling frustrated by the time you see the doctor.

Schedule regular preventive check-ups for your child. These are particularly important for children with chronic medical conditions like CAH. Preventive visits allow your family and your child's doctor to develop a good working relationship and rapport under less stressful conditions than may occur during visits for acute illnesses. Even more importantly, these "wellness" visits ensure that your child is healthy, developing normally, and that future problems are prevented since normal growth and development are key markers for how well the cortisol levels are being maintained. Furthermore, other important health markers can be addressed during preventive visits, for example, checking immunizations and talking to adolescents about smoking.

Repeat instructions back to your doctor to be sure that you understand what you are supposed to do and make notes or request handouts that you can refer to later. If anything is not clear, do not hesitate to ask questions. This prevents confusion and can help avert serious errors.

Be sure to ask what to expect after you leave the office. Will you be notified of all test results? How do you obtain a copy of your child's test results for your personal records? Will the report be sent to your child's primary care provider? If your child was ill, how soon should they improve? What symptoms should you watch out for? What should you do if he or she gets worse? Is a follow-up appointment necessary to ensure that the treatment is working or the problem has resolved?

If you wish to discuss information that you have obtained from other sources such as the

Internet, bring the article and/or source with you to help your child's doctor address your questions objectively. Be aware that while the Internet has been wonderful for helping patients and families become more educated and involved in their health care, many websites offer inaccurate or incomplete information. Always discuss any concerns with your child's doctor, especially if you are getting conflicting information. A few reputable sources on the web are American Academy of Family Physicians at <http://www.familydoctor.org>, American Academy of Pediatrics at <http://www.aap.org>, and <http://www.hopkinsmedicine.org/pediatricendocrinology/cah/index.html> and <http://www.emedicine.com> and of course, <http://www.caresfoundation.org>.

Sometimes, you and your child's doctor will disagree about a treatment plan or a parenting issue. You should feel comfortable expressing your concerns and know that you are being heard. Likewise, your child's doctor should take the time to explain why they are making a particular recommendation and whether other options may be equally acceptable. Frequent or irresolvable conflicts may signal a need to find a physician with whom you see "eye to eye."

As a team, you and your child's doctors will deal with minor, as well as potentially serious illnesses, and together you will guide your child through their growth and development. Developing a warm and effective professional relationship requires a team effort, but providing the best possible care for your child is well worth it!

Adrenalectomy for CAH*(Continued from page 5)*

epinephrine and probably results in reduced ability to respond to stress. It has recently been shown by Merke et al, however, that patients with CAH on glucocorticoid substitution already have greatly reduced secretion of epinephrine. Thus they are no worse off in this respect than they were prior to surgery.

The present studies reinforce the necessity for preventing sustained high levels of ACTH. This is particularly important because many patients have adrenal remnants that are capable of responding to ACTH. In our patients ACTH suppression could usually be achieved with hydrocortisone dosages of 11-13 mg/M². Most of the patients in this series were on much higher doses of glucocorticoid prior to surgery. It is difficult to overstate the importance of promptly instituting stress dosages of glucocorticoids at the onset of illnesses.

Summary

We believe that these long term studies document that bilateral adrenalectomy is a safe and efficacious method of managing patients with severe forms of congenital adrenal hyperplasia. It should be considered in patients who have repeatedly escaped from adrenal suppression and who are now suffering from progressive signs of both androgen and glucocorticoid excess. Adrenalectomized patients will require close medical supervision for life since they will remain at risk for serious consequences or death if not given adequate substitution therapy. Prophylactic adrenalectomy of young patients should be limited to academic centers with established research protocols. Most of the

Mississippi Support Group**Family Picnic****Sat. Oct. 18****Liberty Park Madison****11 AM- 2 PM***Lunch will be provided**RSVP to:*

Gina Murray: (601) 829-9850 e-mail: cgmurray@bellsouth.net
or

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

CF

patients in this series report a better quality of life without adrenals than had been their experience prior to adrenalectomy.

Acknowledgements

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The Surgery Controversy Continues...

In the July 2003 edition of the *Journal of Clinical Endocrinology and Metabolism*, two Letters to the Editor debated the recommendations on early reconstructive surgery for virilized CAH girls contained in the 2002 CAH Consensus Statement. We obtained permission from the *Journal* to reprint these letters for you to read in their entirety. Please note that the CAH Consensus Statement is now available online for free at <http://jcem.endojournals.org/cgi/content/full/87/9/4048>. We thank the *Journal of Clinical Endocrinology and Metabolism* for the opportunity to reprint these letters in our newsletter.

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Regarding the Consensus Statement on 21-Hydroxylase Deficiency from the Lawson Wilkins Pediatric Endocrine Society and The European Society for Paediatric Endocrinology

Sarah Creighton, Philip Ransley, Patrick Duffy, Duncan Wilcox, Imran Mushtaq, Peter Cuckow, Christopher Woodhouse, Catherine Minto, Naomi Crouch, Richard Stanhope, Ieuan Hughes, Mehul Dattani, Peter Hindmarsh, Caroline Brain, John Achermann, Gerard Conway, Lih Mei Liao, Angela Barnicoat and Les Perry

Clinicians from the Multidisciplinary Intersex Clinic at Great Ormond Street Hospital and University College London Hospitals, London, United Kingdom

To  the

Editor:

This consensus statement is a comprehensive review of a range of issues involving the management of 21-hydroxylase deficiency from before birth until adulthood. Many sections of this article are constructive and helpful, but the section on "surgical management and psychology" may be misleading and potentially detrimental to patient care.

The surgical management of ambiguous genitalia is controversial because few long-term follow-up data are available on the effects of surgery on sexual function and psychological outcome. There is increasing concern from intersex consumer groups about possible detrimental effects of genital surgery. Adult patients and parents of affected children should have a central role in this debate.

The authors list three goals of surgery (page 4050, first paragraph): 1) genital appearance compatible with gender; 2) unobstructed urinary emptying without incontinence or infections; and 3) good adult sexual and reproductive function.

1) The authors use the word gender but presumably mean sex of rearing as decided by the clinicians

and parents. There is, to date, no evidence that surgery to render the genital appearance compatible with sex of rearing improves psychological or psychosexual outcome or promotes a stable gender identity 2) Unobstructed urinary emptying without incontinence or infection is an important quality of life issue for the child. Until now clinical instinct that surgery is beneficial has led to early surgery, although there are no data to support this. Surgery itself can result in urinary infections and fistulae (1).

3) The final goal is good adult sexual and reproductive function. There is no evidence that reconstructive surgery gives a better outcome if performed in an infant rather than an adolescent. Clitoral surgery may not promote good sexual and reproductive function, and some studies suggest there may be damage to sexual function (2). Surgery performed in an infant may require revision in adolescence in a significant number of patients (3). An additional major advantage of surgery in an adolescent or adult is that informed consent can be obtained.

Early surgery may be the appropriate course of action and may or may not be supported by long-term outcome data in due course. For the moment, the apparently rigid guidelines in the consensus statement remove flexibility and potentially prejudice the possibility of constructive debate between specialties and with patient groups.

The only consensus attainable at the present time is that of a dedicated

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multidisciplinary team addressing an individual case including full participation of the affected family who will be responsible for the nurture of the child in the modern world.

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References

1. Passerini Glazel G 1989 A new 1-stage procedure for clitorovaginoplasty in severely masculinized female pseudohermaphrodites. *J Urol* 142:565-568
2. May B, Boyle M, Grant D 1996 A comparative study of sexual experiences. *J Health Psychol* 1:479-492
3. Creighton SM, Minto CL, Steele SJ 2001 Objective cosmetic and anatomical outcomes at adolescence of feminising surgery for ambiguous genitalia done in childhood. *Lancet* 358:124-125
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LETTER to the EDITOR

Authors' Response: Regarding the Consensus Statement on 21-Hydroxylase Deficiency from the Lawson Wilkins Pediatric Endocrine Society and The European Society for Paediatric Endocrinology

Walter L. Miller, Sharon E. Oberfield, Phyllis W. Speiser, Laurence S. Baskin, Patricia K. Donahoe, Claire N-Fekete, John M. Hutson and Dix Phillip Poppas

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To the Editor:

We thank Dr. Sarah Creighton and her colleagues for their views concerning the consensus statement on 21-hydroxylase deficiency from the Lawson Wilkins Pediatric Endocrine Society and the European Society for Paediatric Endocrinology, which was published in the September 2002 issue of the *JCEM* (1). The consensus statement was prepared by a group of 40 endocrinologists, psychologists, and surgeons concerned with the management of congenital adrenal hyperplasia (CAH), representing 35 institutions in 12 countries on four continents. This highly qualified and diverse group, which included one of the signatories to Dr. Creighton's letter, represented a broad range of views and experience.

We disagree with the assertion that the guidelines concerning surgical management and psychology are misleading, and we strongly disagree that they might be detrimental to patient care. Dr. Creighton's letter refers to "increasing concern from Intersex consumer groups." These groups primarily represent the experience of women with disorders other than CAH, primarily androgen insensitivity; many of these women were subjected to ill-advised mutilating surgery by inexperienced surgeons. By contrast, the consensus statement deals only with 21-hydroxylase deficiency and emphasizes focusing

surgical care in the hands of a small number of highly experienced surgeons. Whereas the care of patients with ambiguous genitalia who do not have CAH may be controversial, there should be little controversy regarding CAH patients who are 46,XX with normal female internal structures and variable masculinization of the external genitalia. Only a small number of these patients are extremely masculinized.

Although there are few studies of long-term outcome showing that improvement of genital appearance improves psychological or psychosexual outcome or promotes a stable gender identity, competent surgery can provide an excellent outcome in the most severely affected children (2) and is compatible with normal reproductive function (3). Therefore, we believe that it is cruel to leave children in a state of gender uncertainty until "they can participate in an informed consent," particularly in this disorder. While a third sex may be acceptable in some cultures, we believe that it is not so in either North America or Western Europe. Body image, while growing up, particularly through stormy adolescence, is very important to confidence and identity of self.

It is also true that surgery can result in urinary infections and fistula and that clitoral surgery may damage sexual function. The decision to reduce clitoral size is not taken lightly, and every precaution is made to preserve the nerve supply and, hence, sexual function (4). As emphasized in the consensus statement, if highly experienced surgeons do these procedures, the incidence of such complications is low.

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The decision for surgery is made jointly among parents, surgeons, and endocrinologists. Most surgeons inform the parent that simple revision or introitoplasty will probably be needed at adolescence. However, revision surgery is far simpler than having to do the entire pull through at adolescence, because the distance from the urogenital sinus to the perineum has elongated with growth. We agree that a dedicated multidisciplinary team is needed to address each individual case and that the affected families should participate fully. This indeed was recommended in the consensus statement.

We agree that outcome data are exceedingly important, but they must be based on present, improved techniques, by experienced pediatric surgeons or urologists, not on those used three decades ago. We suggest careful follow-up of all surgically reconstructed patients and urge a National Institutes of Health-funded long-term outcomes study of CAH patients.

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References

1. **Joint LWPE/ESPE CAH Working Group (Writing Committee: Clayton PE, Miller WL, Oberfield SE, Ritzin EM, Sippell WG, Speiser PW)** 2002 Consensus statement on 21-hydroxylase deficiency from The Lawson Wilkins Pediatric Endocrine Society and The European Society for Paediatric Endocrinology. *J Clin Endocrinol Metab* 87:4048-4053
2. **Woelfele J, Hoepffener W, Sippell WG, Brämwig JH, Heidemann P, Deiss D, Bökenkamp A, Roth C, Irle U, Wollmann HA, Zachmann M, Kubini K., Albers N** 2002 Complete virilization in congenital adrenal hyperplasia: clinical course, medical management and disease-related complications. *Clin Endocrinol* 56:231-238
3. **Lo JC, Grumbach MM** 2001 Pregnancy outcomes in women with congenital virilizing adrenal hyperplasia. *Endocrinol Metab Clin North Am* 30:207-229
4. **Baskin LS, Erol A, Li YW, Liu WH, Kurzrok E, Cunha G R** 1999 Anatomical studies of the human clitoris. *J Urol* 162:1015-1020

CAH Study at UNC

Chapel Hill, North Carolina

Karen Jane Loechner, M.D., Ph.D.

We are recruiting children with CAH who are 6-12 years old (bone age <14 years), are still growing, and have not yet started puberty. Children will be enrolled in the study at the General Clinical Research Center at the University of North Carolina, Chapel Hill.



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

Although cortisol replacement (hydrocortisone, prednisone, or dexamethasone, for example) and Florinef have virtually eliminated

Based on studies of the regulation of release of ACTH (that, in turn, drives the production of hormones from the adrenal cortex), we have found that we can decrease the amount of ACTH using calcium channel blockers (medications typically used to treat high blood pressure, such as amlodipine). Amlodipine has been shown to be safe even in infants treated for a variety of medical reasons.

Lifetime Magazine Feature on the Colello Family's Journey with CAH

The September/October edition of **Lifetime Magazine** has published a sensitive, heartwarming story of one of our own—the Colello family-- and how they coped with their daughter's diagnosis of classical, salt-wasting CAH. This is a must-read for all CAH families. It really hits home. The magazine is on the newsstands now. The magazine web site is www.lifetimemag.com.

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

For more information, please contact *Dr. Karen Loechner* at (919) 216-5946 (pager) or (919) 966-4435 ext. 224 (voice mail); fax (919) 966-2423. All visits, including laboratory testing, research medication, and parking will be paid for by this protocol.

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Adrenalectomy: My Personal Story

by Amy Aivale

I was diagnosed with non-classical congenital adrenal hyperplasia when I was 14 years old. I was so relieved to actually have everything I was feeling rolled into a little package and given a label. I don't think at that point I would have cared what diagnosis I got, just as long as they told me something was actually "wrong" with me and it wasn't all in my head. I have a younger brother with non-classic CAH and 2 maternal cousins with classic CAH. This is *my* story.

The thing I remember most about growing up is being sick all the time. It was usually the topic of conversation with my parents, teachers, grandparents, doctors, etc. No one ever knew what to do. I never made it to a full week of school throughout my entire school years. Usually by Thursday I would be so exhausted I just couldn't make myself get out of bed. I would spend from Thursday to Sunday recuperating and then be back to school on Monday to start the cycle again. Being sick became my lifestyle and I got used to it. I began to mature at a very early age. I started my period in the 2nd grade, which was accompanied by pneumonia. My menstruation cycle was irregular at first and then became regular in the 3rd grade when I was 9 years old. I developed severe acne about this time, which continued, throughout my teenage years. I also had horrible hirsutism that started appearing the same time. Needless to say, I felt like a walking disaster. By the time I was in 5th grade I reached my final height of 5'9". I felt like an 11-year-old trapped in a 25-year-old woman's body. I was often treated much older than I was because I

looked so much older than my classmates. This added a lot of responsibility and stress to an already stressed out 11-year-old girl. I was never really able to act like a "kid".

Junior High years are awkward for almost all kids, but my feelings of not fitting in became almost overwhelming. I missed more and more school because of illnesses and probably because of depression. My self-image and self-esteem were very low. I started to feel like maybe it was all in my head, maybe nothing was really wrong with me. It was about this time my mom watched a report they did on a news show about non-classical CAH. My brother and I fit every sign. We went into see Dr. Rawlison at Primary Children's at the University of Utah. As I said earlier, I have two maternal cousins with classic CAH, and he was their endocrinologist. Sure enough, we were both tested and we both were diagnosed with non-classical CAH. I was put on .5mg dexamethasone and 15mg hydrocortisone. I didn't feel much of a change, except that I had started to gain a lot of weight. I had battled with weight before, but when I was put on the medications it became much more of a significant weight gain. I gained a lot of weight from the ages of 15-18. At the age of 19 I started only eating once a day and took my medication about once a week. I did this for about a year and a half. I lost about 60 lbs. and as soon as I began eating normally again I gained it back plus more. I also was very sick and very weak most of the time.

I was still dealing with many

illnesses. When I was 18 I came down with viral meningitis and ended up in the hospital for about a week. If I got a cold, it would take me about 2 weeks to get over it. Nothing seemed like it had changed. I was often taking high stress dosages of my medications (50-100mg hydrocortisone) to battle illnesses. I saw an endocrinologist about 3-4 times a year. My levels were consistently high. Nothing was helping. I was beginning to think it was basically hopeless.

A very high point in my life was when I got married in the summer of 2001. However, shortly after my wedding, I stopped having my periods. This was very devastating to my husband and me as it seemed there would be no chance of pregnancy. After not having a period for about 4 months, I went to see a fertility specialist. Provera was prescribed to me and this forced me to menstruate. My periods were very light and only lasted about 2 days. I could tell that it was just not working. I had been seeing many different endocrinologists who just didn't know what to do with me. Finally, I was able to change insurance companies and see Dr. James Grua again. I had seen Dr. Grua when I was younger during the time I was seeing Dr. Rawlison at the University of Utah. Dr. Grua was also treating my 2 cousins with CAH and one of them had just had a bilateral adrenalectomy and was doing extremely well. I wondered if this could be a possibility for me. We went over my lab work and blood tests for the past couple years. He understood my frustration and he felt

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Adrenalectomy: My Personal Story*(Continued from page 12)*

that a bilateral adrenalectomy would be a good option for me. I was ready to do anything at this point. I felt like maybe this would be the answer I had been looking for. I made an appointment to consult with the surgeon and we set the date for surgery on September 6, 2002. I had a CAT scan done a couple weeks before the surgery to make sure my adrenal glands were accessible to the surgeon. Everything was going well and I felt very confident about the choice I had made along with my doctor.

The surgery went well without any complications. It took about 6 hours for both adrenal glands to be removed. I remember waking up feeling as if I had been beat in the stomach with a baseball bat. I felt very sore and miserable for about a month. I couldn't sleep, I had no appetite, and it hurt to even move. I took stress dosages of hydrocortisone for about 3 weeks and then went down to a maintenance dosage of 20mg per day.

Since the surgery I have noticed many changes. The most promising thing for me was that I started my period one month after surgery! I have been having them regularly since. The hirsutism has become less of a problem and I just feel more relaxed. Before the adrenalectomy, I was not a salt waster. I have been getting use to being a salt waster now and dealing with being able to stay hydrated in the hot summers of Utah. I do feel tired a lot more than I used to, but I am hopeful that this will become less of a problem as time goes on. I am still getting used to all the changes my body has made, especially hormonal changes. I feel a lot more emotional that I used to, and cry more at sad movies. I suppose

you could say I feel more like a "girl".

I am happy with my decision to have an adrenalectomy. I am not saying that everyone should rush out to their endocrinologist and schedule for surgery. It was an individual decision that I put a lot of thought and prayer into. I also know that nothing will ever "cure" my condition. I basically traded one condition for another, but I was willing to do that. I have learned that doctors will not always have every answer for my condition, so it is up to me to become educated and knowledgeable about my own body and what it needs. I have also learned that for me to be able to overcome my weaknesses I must be in the best spiritual, mental, and physical health I can be in. Without my faith and trust in God, I don't think I would have been able to make it this far. I am hopeful for a long and happy life. I look forward to someday having children and teaching them the patience and perseverance I have learned from my own experiences. Most of all I have learned that having this condition is a part of my life. It doesn't have to *be* my life. I do have a life beyond being sick and I am going to be out there living it!

Editor's note: *While Ms. Aivale has nonclassical CAH and has chosen adrenalectomy, this choice is not often recommended for those with nonclassical CAH. Unlike classical CAH, NCCAH is not life-threatening, but removing the adrenal glands gives the person a life-threatening condition.*

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Growth Hormones Approved by FDA for Use in Healthy, but Short Kids

Numerous CAH kids with advanced bone ages or very poor growth are treated with growth hormone to improve their height predictions. However, this use of growth hormone has been considered "off label" since these children are not growth hormone deficient. The FDA just approved the use of growth hormone in healthy, but short kids.



Insurance companies have been reluctant to cover the costs of growth hormone for CAH kids, which can run from \$10,000-\$20,000 per year. Now that such use is no longer, "off label", we can hope that this will encourage more coverage for these costs by the insurers.

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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.

New CARES Medical and Scientific Board Advisor!

SCOTT A. RIVKEES, M.D.

Pediatric Endocrinologist, Yale University School of Medicine

Dr. Scott A. Rivkees, is a board-certified specialist in pediatric endocrinology, is an Associate Professor of Pediatrics, in the Section of Pediatric Endocrinology at Yale University School of Medicine. Dr. Rivkees received his M.D. from the University of Medicine and Dentistry of New Jersey, and completed residency in Pediatrics at Massachusetts General Hospital and Harvard Medical School in Boston. His postgraduate training includes a fellowship in Pediatric Endocrinology at Massachusetts General Hospital and Harvard Medical School in Boston. He also received postdoctoral training in neuroscience at Massachusetts General Hospital.

Dr. Rivkees is the recipient of multiple National Institutes of Health and foundation-sponsored research grants and has trained more than 10 postdoctoral fellows. He has an active clinical and basic science research program. He has published more than 100 original and review articles in scientific journals and serves on multiple editorial boards and National Institutes of Health review panels. Dr. Rivkees is a fellow of the American Academy of Pediatrics and the American Society for Clinical Investigation. He currently is a member of the State of Connecticut's General Advisory Committee and is active in the Lawson Wilkins Pediatric Endocrine Society. Dr. Rivkees also has been an expert witness for the United States Senate Children and Family Subcommittee.

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Maria I. New, M.D., Receives The Endocrine Society's Highest Honor—The Fred Conrad Koch Award

Maria I. New, M.D., is the 2003 recipient of the Endocrine Society's highest award—the Fred Conrad Koch Award. This award is presented annually to recognize exceptional contributions to endocrinology and includes a \$25,000 honorarium. The award was presented to Dr. New at ENDO 2003, the 85th Annual Meeting of the Endocrine Society, which took place from June 19-22, 2003 in Philadelphia, Pennsylvania.



Dr. New is currently the Harold and Percy Uris Professor of Pediatric Endocrinology and Metabolism at the New York Presbyterian Hospital: New York Weill Cornell Center. Her work has made important contributions to endocrinology. She discovered apparent mineralocorticoid excess, a disease with severe low-renin hypertension, hypoaldosteronism and low secretion of all adrenal steroids, resulting from a pathogenetic deficiency in the 11 β hydroxysteroid dehydrogenase enzyme. She also discovered a second form of low-renin hypertension known as dexamethasone-suppressible hyperaldosteronism.

Dr. New is also a leading authority on congenital adrenal hyperplasia (CAH). Her achievements include establishing aldosterone deficiency as a cause of salt wasting; delineating hormonal norms for classical and nonclassical CAH; demonstrating mutations of *CYP21* causing 21-hydroxylase deficiency; and pioneering prenatal diagnosis and treatment for CAH based on molecular genetic analysis. Additionally, she recently reported two sisters with partial resistance to all steroid hormones, but not to thyroid hormones or to vitamin D. This important finding suggests the first global transcription factor defect in humans.

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CARES Foundation is so very proud to have Dr. Maria I. New on our Scientific and Medical Advisory Board.

Unique Center Targets Both Excessive and Insufficient Levels of Male Hormones in Women

Reprinted with permission from Cedars Sinai NEWS

LOS ANGELES (July 7, 2003) – An occasional facial wax or bikini wax is the only treatment most women need to control unwanted hair. But those who find themselves in a constant battle with coarse, dark hairs that grow in patterns typical of men may be dealing with a common symptom of an often-overlooked but usually treatable hormone imbalance.

Androgens are known as male hormones but they also circulate in lesser levels in women's bloodstreams. In fact, several necessary male hormones are produced by the ovaries and other organs and glands. If they exist in lower-than-normal amounts, a woman may experience decreased sex drive and fatigue and be vulnerable to osteoporosis. In excess, androgens often lead to acne, balding or thinning hair on the scalp, irregular or absent menstrual periods, fatigue, decreased sex drive, and the abnormal hair growth called hirsutism.

The intricate mechanisms producing androgens and other hormones that impact androgen availability and strength are the first links in an entwined chain of events that ultimately affect individual sites such as hair follicles, skin pores and eggs in the ovaries. The effectiveness of treatment, therefore, depends on finding and addressing the defective point or points in the sequence while providing therapy for bothersome symptoms.

To offer in-depth testing, comprehensive treatments and support, and research into molecular mechanisms and future therapies,

Cedars-Sinai Medical Center has launched the Center for Androgen-Related Disorders. It is believed to be the only program of its kind in the country to specialize in both up- and down-regulation of androgen levels in women.

Ricardo Azziz, M.D., who holds the Helping Hand of Los Angeles Chair in Obstetrics and Gynecology, directs the new center and its programs in androgen-excess disorders. Director of Cedars-Sinai's Department of Obstetrics and Gynecology, Dr. Azziz joined the medical center in late 2002 after serving for 15 years at the University of Alabama at Birmingham. In addition to his role at Cedars-Sinai, Dr. Azziz also serves as Executive Director of The Androgen Excess Society, an organization dedicated to offering recommendations for therapy, identifying research priorities, and helping to provide education on this often-misunderstood and misdiagnosed disorder.

Glenn Braunstein, M.D., holder of the James R. Klinenberg, M.D., Chair in Medicine at Cedars-Sinai, offers expertise in causes and complications of androgen deficiencies. Board-certified in internal medicine with a subspecialty in endocrinology, diabetes and metabolism, Dr. Braunstein has conducted extensive research in reproductive endocrinology. Currently, three Phase III clinical trials are underway to assess the effectiveness of testosterone replacement therapy in women.

Although androgen excess –

which typically is seen in the years from adolescence to menopause – causes many symptoms that are readily visible, underlying causes are more challenging to detect. The inability to become pregnant, for example, may stem from a variety of causes, and many women who suffer from hirsutism believe they are destined for a life of plucking, tweezing or paying for expensive treatments, never knowing that a serious medical condition may be responsible.

“The overwhelming majority of women with hirsutism have an underlying androgen disorder,” says Dr. Azziz. Non-classical congenital adrenal hyperplasia and polycystic ovarian syndrome are causes of androgen excess.

Cedars-Sinai is one of the largest nonprofit academic medical centers in the Western United States. For the fifth straight two-year period, it has been named Southern California's gold standard in health care in an independent survey. Cedars-Sinai is internationally renowned for its diagnostic and treatment capabilities and its broad spectrum of programs and services, as well as breakthroughs in biomedical research and superlative medical education. Named one of the 100 "Most Wired" hospitals in health care in 2001, the Medical Center ranks among the top 10 non-university hospitals in the nation for its research activities.

Newborn Screening Update

Ohio and Oregon:

Began Screening for CAH
July 1, 2003.

Nevada:

Nevada is now screening for CAH!! In January of this year, the Nevada Department of Health put out a Request for Proposals (RFP) for the newborn screening contract for the state. At that time, the RFP included a request for bids on an expanded panel including screens for metabolic disorders and hemoglobinopathies (Sickle Cell Disease and Thalassemia) but not CAH. In reaction to a call for the inclusion of CAH from families affected by this disorder and Nevada medical professionals, the state recalled the RFP in April and issued a new one that included CAH. The state accepted a bid from the Oregon State Laboratory and began the expanded screening on July 1.

Deeming an expanded panel "a sensible addition to our screening program," the Health Division implemented testing for over 25 new disorders including CAH (see <http://www.savebabies.org/states/nevada.htm> for a complete list). However, the state decided to stop testing for CAH after a month due to budgetary restraints.

At the public hearing on Sept. 12, 2003, the Nevada Department of Health asked the State Board of Health to consider and approve an amendment contained in Nevada Administrative Code to increase the fee charged by the state for the registration of births from \$28 to \$60. This fee increase would cover the cost of the expanded newborn screening panel as well as the

establishment of a state birth defect registry. However, due to a negative fiscal situation, the Department of Health indicated that testing for CAH would not begin until July 1, 2004.

In an emotional session, a number of CARES families affected by CAH then presented testimony. While each of their stories differed slightly, all told of holding a dying baby in their arms with no explanation as to what was wrong with their newborn child and implored the board to prevent this from happening to another Nevada family by approving the fee increase inclusive of the immediate resumption of testing for CAH. Dr. Alan Rice, a pediatric endocrinologist from the University of Nevada, Las Vegas and Dr. Michael Skeels, director of the Oregon State Lab that does the screening for Nevada, also testified in support of reinstating CAH newborn screening.

Showing broad-based, unqualified support for the entire expanded newborn screening panel, the board immediately approved an amendment calling for an initial fee of \$64 (to be reduced to \$60 July 1, 2004) to enable the continuation of testing for metabolic disorders and hemoglobinopathies as well as the immediate reinstatement of screening for CAH.

This is an incredible victory for the babies of Nevada. The amazing Gretchen Lin, who so artfully organized the advocacy efforts in Nevada, the families who testified and wrote letters, the doctors who supported CAH screening and the members of the Nevada Dept. of Health and Board of Public Health, who supported the CAH screening

and came up with the funding necessary to screen for CAH all deserve our deepest gratitude. Thank you all so much!

California:

California has taken a huge step backwards in its newborn screening program. Governor Gray Davis pulled the plug on funding for the state's successful pilot expanded screening program. Although CA did not screen for CAH in its pilot, the fact that he took away what little funding the program did have does not bode well for an expansion to include CAH anytime in the near future under the current administration. Perhaps the recall election in CA will give the state a more child friendly Governor. Make sure you vote!

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To all our Members:

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>.



Put the "Fun" in Fundraising!!

As a non-profit, we are always in need of financial support. One of our members, Brad Smith came up with a wonderful fundraising idea to raise funds for CARES Foundation. Brad, along with his wife, Tina, have arranged a pork burger and bake sale in his home town – Albion, Indiana. Way to go Brad and Tina!!

We would love for more of our members to help with fundraising. Please consider coordinating or hosting a fundraising event in your area. We will do whatever we can to assist you in this effort. We are starting to compile event sheets from CARES members and will produce a Fundraising Guide for others interested in running a fundraiser so that no one has to re-invent the wheel.

There are many different ways to raise funds and your contribution makes a difference. Not only will it raise funds to expand our services, but it will also raise awareness about CAH throughout the community.

Here are some ideas that you might consider: Bake Sale, Garage Sale, Car Wash, Walkathon, Bike Ride, Progressive Dinner Party, Casino Night, Auction, Treasure Hunt, Karaoke, Variety Show, Dance Marathon, Sport Tournament, Raffle, just to name a few. A fundraiser can be great fun and so rewarding. It's a wonderful way to involve the whole family in such a worthwhile activity. Call us to let us know what you are planning. YOU can do it!

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Attention Arizona Families!!

We need your help! **Basha's Supermarket** is starting up its annual fundraising program called: **"Thanks A Million For Friends and Neighbors"**. As a non-profit organization, CARES Foundation will benefit when members, such as yourself, link their Basha's Thank You Cards to our organization. Every time you make a purchase at Basha's, the supermarket will donate 1% of your total to CARES Foundation, up to \$2,500 per organization. It costs you absolutely nothing – Basha's makes the donation on your behalf. It's a win-win situation for everyone.



Here is how the program works. Basha's assigns us a 5-digit group identification number (**the CARES Group Id Number is 29261**). You need to link our group id number to your Basha's Thank You Card. Each time you make a purchase, Basha's will

credit our account with the donation. *Make sure to check your receipt after your purchase to see CARES*

Foundation listed-- this will

ensure that the donation has been properly credited.

This program will run from **September 1, 2003 – April 30, 2004**. Please consider shopping at Basha's during this time period and encourage your family and friends to do the same. Remember to link your card to the **CARES Foundation Group Id Number: 29261**.

This is a wonderful way to participate in fundraising for the organization while raising awareness about CAH. If you would like some brochures from us to distribute to your friends and family, call or email us and we would be happy to send you some in the mail.

Your participation and support means so much for our organization and it will allow us to continue to expand our services to individuals and families with CAH.

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Every Dollar Counts – and we Count on You

If you know of a supermarket or store chain that has a fundraising program that will contribute to non-profit organizations such as Basha's in Arizona (see article above), please contact us to let us know. Or if you have an idea for a local fundraising event, contact us to see how we can support you. As a non-profit organization we are always looking for ways to raise money so that we can continue to provide you with services and meaningful programs. Every dollar counts. Help us help you and your family.

CF

Audio Tapes from the 2003 Endocrine Society Annual Meeting Available for Purchase

You may wish to order the audiocassettes from the 2003 Endocrine Society meetings. The tapes of the keynote speeches are available through www.CMEunlimited.org/es. The tapes that pertain to CAH and androgen excess disorders are:

#25--Diagnosis of Adrenal Disorders—first lecture is by Dr. Ricardo Azziz on diagnosing nonclassical CAH;

#43--Diagnosis and Management of Sexual Ambiguity, Dr. Melvin Grumbach, Dr. Peter Lee and Dr. Gary Berkovitz;

#57-- Behavioral and Reproductive Consequences of Prenatal Androgen Exposure, David Abbott, Kim Wallen and Lourdes Ibanez;

#76-- Treatment of Congenital Adrenal Hyperplasia, Dr. Deborah Merke, Dr. Gerard Conway, and Dr. Stefan Bornstein;

#120 --Meet the Professor: Congenital Adrenal Hyperplasia, Dr. Patricia Donohoue; and

#142-- Polycystic Ovarian Syndrome, Dr. Ricardo Azziz.

The tapes can be ordered for \$12 each plus shipping online at www.CMEunlimited.org/es or by calling 1-800-776-5454 or by FAX at 818-504-2771 or by mail at 7764 San Fernando Road, Suite 13, Sun Valley, CA 91352.

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Parent Tips!!

(Solutions for Common Problems)

ARE YOU HAVING TROUBLE GETTING SOLU-CORTEF FROM YOUR PHARMACY? WE WERE!!

For months our local pharmacy would call us and say, "Sorry, still not in, we'll call you as soon as it gets here." The pharmacy called around to other pharmacies and still no luck. Finally a bright idea came to mind - our hospital has had it when we needed it, wonder if the hospital pharmacy could sell a vial to me? So I made a call and sure enough the hospital was stocked with it, but they could not sell it out right to me. BUT, they could transfer it to our local pharmacy and they could sell it to us. So if you are having trouble getting Solu-Cortef give you hospital pharmacy a call.



Note: We were also told my another CAH family, if you call the product maker of solu-cortef (Pfizer), they will drop ship you an order. We have not had to do that, but that is another option.

*Rob Simmons, Father of a 2 year old daughter with CAH
Elburn, IL*

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.

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New Genetics Museum

The Stetten Museum of the Office of NIH History is pleased to announce a new internet exhibit based on existing physical exhibits in the NIH Clinical Center: **A Revolution in Progress: Human Genetics and Medical Research** <http://history.nih.gov/exhibits/genetics/>.

Genetics research is important in the prevention and treatment of disease. This exhibit provides information on what DNA, genes, and chromosomes do in our bodies, explains how basic research has led to a better understanding of genetic research, and discusses the scope, purpose, and techniques of the Human Genome Project to map and decode our genes. The exhibit poses such questions as: How do genes cause disease? Can gene therapy work? How do we manipulate genes and should we? The exhibit has a "Just for Kids" section that helps to teach kids about the history and role of genetics in our world. This would be a good tool to teach our children about the genetic aspects of CAH.

This exhibit was produced by the Stetten Museum in collaboration with the National Institute of Allergy and Infectious Diseases, the National Cancer Institute, the National Human Genome Research Institute, the National Institute of General Medical Sciences, and the National Heart, Lung, and Blood Institute.

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Volunteers 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.




We need members who have experience dealing with international programs in developing nations or who have international contacts. We are looking into providing drug donations to CAH patients in developing countries where these drugs are not available or exorbitantly expensive. We need people to help us set up this program. If you can help, please let us know! Email: Kelly@caresfoundation.org or call: 1-866-227-3737.

CF

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>

Cody Cares Medical ID Jewelry

4155 Carson Avenue
 Indianapolis, IN 46227
 317-783-7702 • codycaresid@aol.com

Specializing in **Custom Made and Engraved Medical Jewelry.** *Choose from:*

- ✓ ID Bracelets
- ✓ Shoetags
- ✓ ID Wristbands (*perfect for infants & sports*)

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

Financial & Pharmaceutical 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.



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.

CF

CAH Family Support Groups Around the Country

ALABAMA

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

511Aycoc822@aol.com

TEXAS

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

ARIZONA

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

WISCONSIN

Contact Lisa Jaskie
(414) 645-0782

INDIANA

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

MISSISSIPPI

Susan Aycock
601-833-8373

Family Picnic - Oct 18
see pg. 8 for details

PROFESSIONAL TOUCH ELECTROLYSIS, INC.

STEPHANIE R. FRACASSA, C.P.E.

30 East 40th Street, Suite 801
(bet. Park & Madison Aves.)
New York, NY 10016
Tel. 917-821-2409

By Appointment Only

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**DO NOT DELAY
MEETING NOTICE**

Address Service Requested