



In Search of a Cure for CAH: Cell-Based Therapy and the Role of Stem Cells

David T. Breault, M.D., Ph.D.

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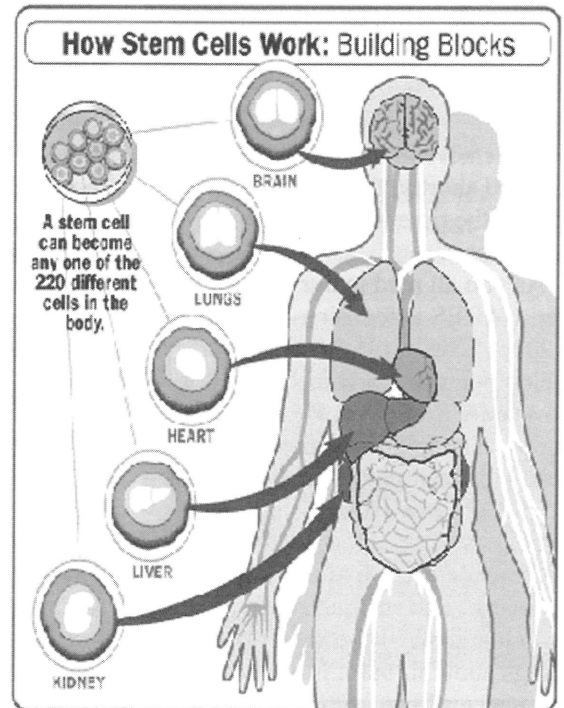
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Stem cells are unique given their capacity (1) to give rise to other cells in the body and also (2) to replicate (self-renew) themselves, which provides for a continuous supply of new cells over time. Stem cells play important roles in the normal development of an individual and help maintain the essential functions of tissues and organs throughout one's life.

Two major types of stem cells exist: Adult Stem Cells and Embryonic Stem Cells.

Adult Stem Cells are likely located within many tissues of the body serving as self-renewing cells over time. These cells have been definitely found in bone marrow and skin, although many more tissues are expected to contain adult stem cells. The clinical use of adult stem cells from bone marrow is well established to treat a variety of diseases.

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Is Congenital Adrenal Hyperplasia Screening Worth It?

Katherine Kirk, RN, MSN

Screening for Congenital Adrenal Hyperplasia (CAH) due to 21-hydroxylase deficiency, the most common form of CAH, started in Oklahoma on Valentine's Day 2005. I was the state's Coordinator for CAH, a new position, and I was trying desperately to ensure everything was prepared for the first newborns in Oklahoma to be identified at risk for this life-threatening disorder through screening.

The page came at 8 a.m. as I was making my way to the office. Only eleven days after launching the Oklahoma newborn screening program for CAH, the first preliminary abnormal screen had been identified. The result was pre-released as a panic value warranting immediate follow-up. Further testing was underway on the newborn's specimen, but the result was too critical to wait for the results of the second screening test. I wrote down the name Aidan, and hurried to

contact the reported primary care doctor about the abnormal screen result.

The Oklahoma State Department of Health (OSDH) achieved this milestone for Aidan and other Oklahoma newborns after six years of planning and promotion. The Board of Health (BOH) approved adding CAH to the newborn screening panel after an adequate funding stream was achieved, a requirement of the Oklahoma law that governs newborn screening. In addition to funding obstacles, screening for CAH stirred controversy in the state's pediatric endocrinology community. The medical community anticipated countless hours ruling out large numbers of false positives, particularly in premature infants. Some also questioned whether newborn screening for CAH was really necessary. For example, females

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A Message from the

Executive Director:



Dear Friends,

Happy New Year! I hope this newsletter finds you all well and excited to welcome 2006. A lot has happened since our last newsletter and there is so much more in store for this year.

As we **grow as an organization**, it becomes increasingly important to keep our finger on the pulse of all of our families. To help us **better serve you**, we have developed a comprehensive membership survey, to be mailed shortly, that we hope you all will take the time to complete and return to us.

As you all read in our year-end letter, CARES is focusing on research for 2006. One of the most promising developments is a new technique for **Embryonic Stem Cell Research** (see page 1). The implications for this research are astounding and likely hold the **key to a cure** for future generations.

Many of you are familiar with the controversy that surrounds this type of research and may have very strong feelings about it, but it is our hope that these new **embryo-sparing** methods for obtaining stem cells will help to alleviate ethical concerns. I hope you will read Dr. Breault's article with an open mind and come to your own conclusion.

We all agree that **research** is a priority for our community and I'd like to extend a big "Thank You" to everyone who has given to this campaign to date. We so appreciate your enthusiasm to keep us thriving as the only US organization devoted entirely to this disorder. **Your contribution is so important** as we continue to support the most promising research while adding new programs and staff to fit **your support and education needs**.

While I am sad to say goodbye to Renata Blumberg who was such an integral part to our success in 2005 and the Indiana Family Conference, I am excited to welcome three new members to our team—Mariel Vargas,

Cheryl Karch and Gretchen Alger Lin.

A graduate of Arizona State University, Mariel is leading our **Hispanic outreach** efforts—working on our Spanish website as well as printed materials for new families.

In September, we welcomed Cheryl Karch to assist with our development efforts. Cheryl has a comprehensive background in planning and development and we look forward to a long and fruitful relationship with her.

I'm also pleased to welcome Gretchen Alger Lin, who has a SWCAH child, to our staff. Gretchen brings 15 years of marketing, advertising and public relations experience to CARES. Expanding our staff is very important as we work to **increase our outreach** and provide important programs, such as our regional conferences, for all of you.

Our conferences are always the highlight of my year. I get to catch up with old friends and experience the energy of being with our members who all want to make the world a better place for those with CAH. Our 2005 CAH Family Conference was a huge success, and we are so thankful to **Riley Hospital for Children and Clarian Health Partners** for donating their beautiful conference space. We extend special thanks to our Riley hosts: Dr. Rick Rink and Dr. Erica Eugster. Your support in the months and weeks leading up to the conference, your graciousness and patience helped to make it such a tremendous success, and we are so grateful. In fact, we thank all of our presenters and panelists for all of your hard work, time and travel to educate and support our membership. Every year, it seems like our conferences get better. Can't wait 'til next year!!!

Warm Regards,

Kelly

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This newsletter is published 3 times a year.

2005 CAH Family Conference Riley Hospital for Children, Indianapolis, IN

The 2005 CARES Conference was held on October 22, 2005. Sponsored by The Riley Hospital for Children and Clarian Health Partners, the day featured several of the country's leading CAH researchers.

We thank all of our distinguished presenters and panelists for their hard work and dedication to educating families and healthcare providers about CAH.

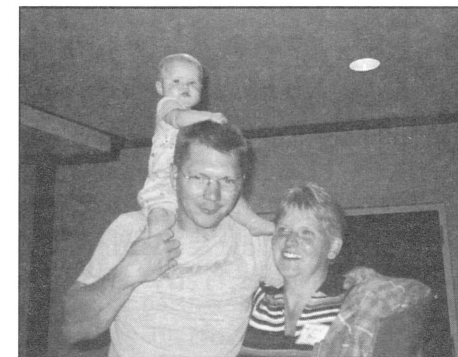
Integral to the success of the conference were our generous corporate sponsors **Eli Lilly and Company, Serono, Genentech, and Perkin Elmer**, who allowed us to keep the conference free for attending families. Their dedication to the CAH community is truly appreciated.

Special recognition is also due to our hosts **Drs. Rick Rink and Erica Eugster**, whose hard work and enthusiasm are truly inspiring. ♥

Next: November 2006, NJ



Top Left: Mayté Villaseñor with baby Marianna **Top Right:** (from left) Scott Rivkees, MD; Richard Rink, MD; Kelly Leight; Sheri Berenbaum, Ph.D. **Bottom Left:** Tod Hogan, Kristie Day and baby Lucy **Bottom Right:** (from left) Omar Campos, Erin Anthony, Mayté Villaseñor, Kelly Leight, Meryl Stone and Renata Blumberg.



Katrina Update...Your Generosity At Work

Dear Kelly, I certainly want to extend our thanks to you and the other kind and thoughtful members of CARES who have sent assistance to help get the pediatric endocrine services in New Orleans back in order and tide us over in the coming years of rebuilding. We foresee a multiyear rebuilding program at the hospital. We have made an effort to find our former patients scattered around the area and extend services to them. We have new clinical outreach to Baton Rouge and Lafayette, Louisiana. In addition, we had the only fellowship training program for pediatric endocrinology in the area. Two of our fellows have returned and are busily engaged in their training. The contribution of the CARES program will be most helpful to us in maintaining the quality of the fellowship program. The monies will help us bridge the gap in local resources in the aftermath of the hurricane.

We were the recipients of a number of computers from members of CARES and we will use them to facilitate the transition to electronic medical records. When we were displaced from the main hospital we lost all access to records and charts of our patients. We hope that in the new era we will have access to records electronically wherever we may be. In addition accessibility of the staff to extra computer work stations should enhance our research efforts as well. The donation of those computers has obviously freed up resources to be used in other aspects of the recovery.

Once again, thanks to all the membership from the faculty and staff of Pediatric Endocrinology at the Children's Hospital of New Orleans and best wishes to all for the upcoming holiday season.

-Stuart Chalew, M.D., Director of Endocrinology, Children's Hospital New Orleans

From the Office

¡Se Habla Español!

Tenemos una empleada que habla Español. Para hablar con Mariel, llámela 1-866- 227-3737 o escribala Mariel@caresfoundation.org.

New Phones and Copier!

In the Fall of 2005, CARES received grants from **E.J. Grassman Trust** and the **Smart Family Foundation**. Thanks to these generous gifts, we have purchased a professional phone system and are in the selection process for a commercial printer. These tools will enable us to better serve our membership as we grow as an organization. We are truly grateful for these generous gifts!

Welcome!

CARES is pleased to welcome three more employees to its family, Mariel Vargas in our main office and consultants Cheryl Karch and Gretchen Alger Lin.

Mariel, a graduate of Arizona State, brings to CARES her excellent bilingual skills and tremendous enthusiasm for non-profit work. To contact Mariel, call her at 1-866-227-3737 ext. 104 or email Mariel@caresfoundation.org

Cheryl Karch has extensive experience in the non-profit sector with planning and development and we look forward to a long and beneficial relationship. To contact Cheryl call her at 1-866-227-3737 ext. 105 or email Cheryl@caresfoundation.org

Gretchen Alger Lin not only brings her 15 years of advertising, marketing and public relations experience, but a personal touch as a mother of a child with CAH. To contact Gretchen, email her at Gretchen@caresfoundation.org .

CARES Family News

♥ **Thank You, Sue!**

Sue Bianchi, our Webmaster, Designer, Master Technician and wonderful friend, is stepping down from her position at CARES. Sue has volunteered her time tirelessly for 3 years, putting in countless hours (often times tearing her hair out) to make sure our website is up-to-date, beautiful and easily accessible.

We are working feverishly to hire a replacement for Sue, but these are hard footsteps to follow! Sue, we will miss chatting with you on a regular basis and we thank you from the bottom of our hearts for all your hard work and dedication to the CARES community.

We Love You!

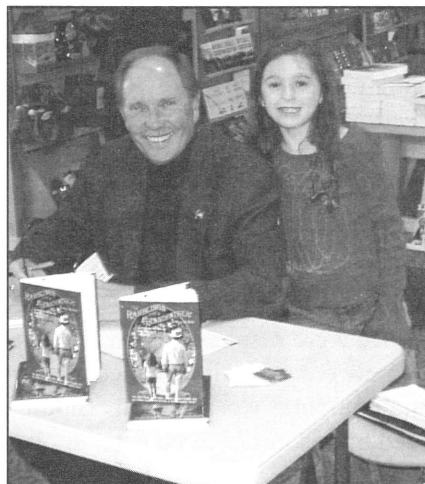
2005 CAH Conference Indianapolis, IN

Our 2005 conference was a huge success and we couldn't have done it without the hard work of all of our wonderful volunteers. Thank you to everyone who helped out in the weeks before and the day of the conference.

(In alphabetical order): Penny Barrett, Laurie Ember, Louise Fleming, Stephanie Fracassa, Stephanie Grubler, Monica Heinze, Jenny Hendricks, Cindy Klem, Jami Abell Patterson, Alesia Pinson and Shalimar Scott.

(Babysitters): Cody & Destiny, Lindsay Beemer, BethAnn Bolton, Nicole Deel, Gina Dreger, Terry Krist, Chelsea Naley, Samantha Naley and Leah Samples.

We were very fortunate to have several impromptu volunteers throughout the day and may not have gotten everyone's name. If you helped out on the day of the event, please let us know. We truly appreciate all of your hard work and would like to recognize all those who helped.



Lauren Doss and her grandfather Carson Doss pose at the signing of "The Ramblings of a Grandfather."

"The Ramblings of a Grandfather"
by Carson Doss

CARES Foundation thanks Carson Doss for donating several copies of his book "The Ramblings of a Grandfather" for sale at the Indiana Conference.

Sales of the book were a huge success and several conference attendees took order forms once we sold out.

We are so grateful to Mr. Doss and his family for their continued generosity and support of CARES Foundation.

Thank You!

If you would like to purchase a copy of "Ramblings," please email Lin@thedosses.com.

Legislative Alert

Support The Reconstructive Surgery Act of 2005 (HR 4022)

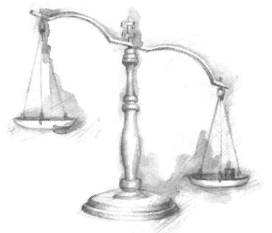
CARES Foundation encourages all members to contact their US Representatives today in support of The Reconstructive Surgery Act of 2005 (HR 4022).

The legislation has been reintroduced by Congressman Mike Ross (D-AR) to require insurance companies to cover medically necessary surgery for congenital defects, developmental abnormalities, trauma, infection, tumors, or disease. This legislation is most important for females affected by CAH who are born with

genital differences as a result of androgen excess.

Unfortunately, many health insurance companies define some surgeries as "cosmetic," making it necessary to define these reconstructive surgeries as "medically necessary." This Act defines medically necessary reconstructive surgery as surgery "performed to correct or repair abnormal structures of the body caused by congenital defects, developmental abnormalities, trauma, infections, tumors, or

disease, to (1) improve functions; or (2) give the patient a normal appearance, to the extent possible, in the judgment of the physician performing the surgery."



To take action, please write a letter to your U.S. Representative by visiting <http://www.house.gov/writerep> and indicate your support of HR 4022.

Newborn Screening: 4 States Fail for CAH

Gretchen Alger Lin

CARES continued its campaign for universal expanded newborn screening throughout the final quarter of 2005. Expanded testing for 29 disorders including CAH began on December 5, 2005 in **Kentucky**. This was a full month before testing was scheduled to begin. Initially positive results will be sent to the Mayo Clinic in Rochester, MN for confirmation; however, by the end of the year Kentucky intends to move the entire program in-state. With the start of the new year, both **Utah** and **Nebraska** will begin screening for CAH and **Washington D.C.** is in the process of implementing an expanded newborn screening program that includes testing for CAH.

Newborn screening in **Canada** continues to lag far behind the times. Of the 13 provinces in the country only two and a half – Manitoba, North West Territories and the

western part of Nunavut – currently are screening for CAH. Thanks to the efforts of our newborn screening advocate partners and CARES members, on November 2, 2005, Ontario approved expanded newborn screening. By the end of 2006, Ontario will test babies for 27 disorders including CAH.

While the majority of our efforts are in the form of encouraging governments to expand their screening programs to include CAH, we also monitor existing newborn CAH screening programs, scientific publications and legislative action to ensure the best possible screening programs for all children.

In December, CARES responded to a paper entitled "Newborn Screening for Congenital Adrenal Hyperplasia Has Reduced Sensitivity in Girls" that argued against lowering thresholds and requiring a second screen in the state of **Wisconsin**. CARES sent letters to

the secretary of the Department of Health & Family Services and the supervisor of Wisconsin's newborn screening laboratory encouraging them to aggressively pursue expanding screening for CAH to include lower 17-OHP threshold levels and a required second screen.

National Newborn Screening and Genetics Resource Center records as of November 2005 include 17-OHP threshold tiers for 27 states. For normal-birth-weight babies 21 of 27 states have set their cut-off at 50ng/mL or less. The average threshold for low-birth-weight babies in these states is 129ng/mL. Wisconsin is at 55ng/mL and 135ng/mL respectively.

Moreover, CARES made it clear that we believe the goal of any newborn screening program should be to detect as many Classical CAH-affected infants as possible: both salt-wasting – the focus of

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Profiles

"Having CAH, for me, was another part of my life, like being near-sighted."

Calm And Healthy: Living with CAH

Josh Eisenberg

My wife, Audrey, gave birth to our first child, Andrew Gehrig Eisenberg, in February 2005. At 7.5 pounds and 20 inches, he was a healthy, bouncing baby boy. Our only worry was put to rest when his newborn screening test showed negative for CAH. He would be a carrier but would not face a lifetime of medicine and salt-cravings.

Andrew's birth prompted me to consider my own life with CAH – what did it mean? How had my life been different because of it? Looking back, I can say, with few exceptions, that I have lived a normal life. Sure there were the incidents of severe acne, the shorter stature (5' 7 1/2" thank you very much), and my strange food cravings (you haven't lived until you've tried a bowl of croutons, pickles and French dressing), but I have generally lived my life without much CAH-related trauma.

While awaiting the birth of our son, I spoke to my own parents about my early childhood experiences with CAH. I remember a blur of episodes of sickness, urine samples, and bone density tests. Fortunately, my

parents never made it scary. Even now, when they tell me how sick I was as a newborn and young child, they gloss over the trauma and calmly recount some of my worst episodes concluding, "but it all turned out OK..." Having CAH, for me, was another part of my life, like being near-sighted.

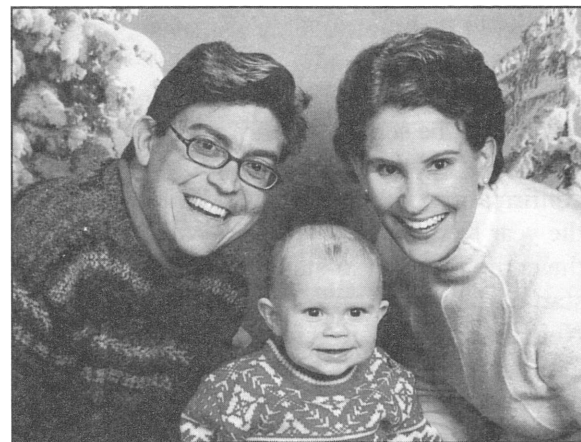
My parents loving, "we'll just deal with it," approach set the tone for my life experience with CAH. Because I had such a casual relationship with the disease, I was never the best patient. During college and graduate school, I would go days without taking my medicine. If I felt really weak I'd load up on Prednisone or throw some salt into my Coke. I knew why I was feeling poorly, but I also knew that I would be fine.

The worst scare I have had with my CAH came in the summer of 2002, when my endocrinologist, Dr. Jeng Su, found testicular growths. Like my parents, Dr. Su did not overreact, but presented me with several options for their cause, the most likely of which was my medication. A few worst case scenario options included the possibility of cancer, or that the growths would render me

sterile. We were relieved when tests verified Dr. Su's theory related to improper dosages of my medicine.

So here we are, a few days before Christmas and my wife and I are ready to celebrate Andrew's first holiday. I continue to live my life as I always have, taking my medicine, (extra when I feel ill), eating salty foods, and keeping active—running marathons, climbing the Hancock Tower stairs, and chasing after a 10-month-old. I know I will always live with CAH, but it is manageable. Life with CAH may have its rough spots and even some scares, but it is manageable with good patient-doctor communication...and the occasional salted Coke. ♥

The author, a CARES member, is a Stay-At-Home Dad and tour guide and resides with his family in Chicago.



From left: Joshua, Andrew and Audrey Eisenberg

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My Experience

Bobby Covitz

When asked to write this article, the question posed to me was: how has CAH impacted my life? Despite maybe not being as tall as I would like to an assortment of daily pills, I consider myself a healthy, normal 20-year-old college student for someone born with a congenital condition. Over the years, my CAH has caused my family and I a few scares; however, the hardest part has been the relative obscurity of the condition.

I was born on December 23, 1984 at Brigham and Women's in Boston and was back in the hospital a month later. My aunt said I resembled a "flick chicken." Regardless, it was clear to my parents and my pediatrician that something was wrong. Without much help from the doctors (an unfortunate trend during my adolescent treatment), my mom began to give me Pedialyte. My mom is still unsure as to what propelled her to give me the electrolyte solution, but it was obvious that my classical salt wasting CAH wasted no time manifesting itself.

Now, Bostonians are fortunate enough to have access to some of the best teaching hospitals in the country, if not the world. My first few years of treatment were at Children's Hospital. My memories of my appointments include round-table discussions with teams of doctors. At one point, a doctor even went so far as to accuse my parents of not giving me my medications because my bone age was running out of control. With not much idea of where to turn to next, we visited the late, great Dr. John Crawford at

Mass General. Aside from being a great doctor, Dr. Crawford was a great human being. It is because of him that I stand 5'8 1/2" instead of 5'3". And most importantly, it is because of Dr. Crawford that I was physically able to play football and

"For me, CAH is a part of my very nature and character."

baseball, to have the same opportunities as every other teenage boy.

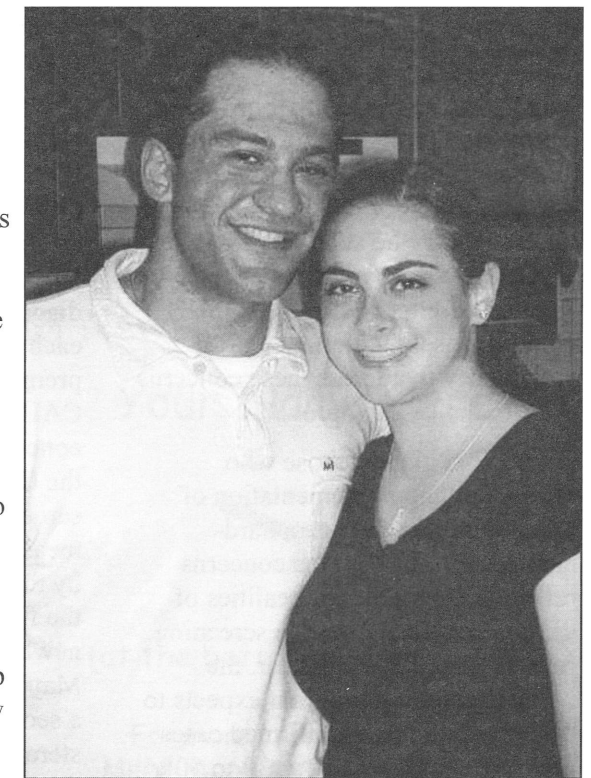
My first real experience with the potential deadliness of CAH was in the summer of 2002. While working in a camp, I contracted a bug bite that eventually brought upon adrenal crisis. I entered the hospital with a blood pressure of 70/30, and for the first 24 hours I was under nonstop care. About two weeks later, I was discharged with a month's worth of IV antibiotics. Combined with the accompanying fear of an experience such as this, it was an eye-opening experience to say the least.

This new experience led to some investigating into this new side of CAH my family and I had never seen before. Eventually, this investigating led us to CARES. Our first trip to a CARES event for the New England region left my family in shock of how much we had been missing out on. From

certain precautionary measures I can take to just meeting people face to face with CAH, it was a great learning experience. It lifted this shroud of obscurity I had concerning CAH and made me much more aware of how my condition can affect me.

Thus, how has CAH impacted me? Aside from the occasional annoyance of having to change doses, CAH has shaped who I am as a person more than any book, teacher, or movie. My day begins and ends with CAH; as I take my pills twice a day, I am constantly reminded of what I had to go through just to get to this day. For me, CAH is a part of my very nature and character. ♥

The author, a CARES member, is a Junior at Ohio State University majoring in Jewish Studies.



Left: Bobby Covitz, 20, salt-wasting CAH.

Newborn Screening Update

(Continued from page 5)

Wisconsin's newborn screening program – and simple virilizing – largely ignored by this program.

Of the 50 states, only four - **Arkansas, Kansas, Louisiana and West Virginia** - remain that do not screen for CAH. We encourage you to write letters of support to these states and contact your legislators (see box to right).

If you live in a state that does not currently screen for CAH and would like to contact your legislator, you can find the contact information here: <http://www5.lsb.state.ok.us/legislators/lbaddress.asp>. You can also visit <http://www.newborn-screening.org/letter.html> to obtain a sample letter to send to the governor or other government official of any state. ♥

Please contact the officials in these states and voice your support for universal newborn screening including congenital adrenal hyperplasia (CAH). For help, please email the CARES office at info@caresfoundation.org.

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Fax (304) 558-1130

CAH Screening

(Continued from page 1)

with CAH are frequently born with ambiguous genitalia, and are diagnosed promptly with this clinical presentation. For males with CAH, a diagnosis may be achieved by alert clinicians who recognize the early signs of adrenal crisis. The OSDH worked closely with the medical community to address these concerns prior to the BOH approval.

At first glance, those who questioned the implementation of this program might seem hard-hearted. But, in fact, the concerns reflect some pragmatic realities of the challenges inherent in screening for this disorder. Each year, the OSDH screening program expects to detect two to three confirmed cases of CAH, with as many as 30 to 50 additional newborns with abnormal

screen results that do not have CAH (i.e., false positives).

These false positives are most often associated with prematurity. It is known that premature infants have elevated levels of 17-OHP (CAH screening involves detecting elevated levels of 17-OHP). However, a diagnostic workup must be done on each abnormal screen result because premature infants might also have CAH. To address this and the concerns of the medical community, the OSDH implemented a two-tier screening test. The first test, measuring the 17-OHP level, is done by the Public Health Laboratory. If the first test is abnormal, then the newborn's specimen is sent to the Mayo Laboratories in Minnesota for a second test. Mayo performs a steroid profile to assist in determining those infants at risk for CAH versus those with elevated 17-

OHP values related to other conditions, such as prematurity. This second-tier test reportedly will dramatically reduce the number of newborns that must undergo further testing for CAH. This is done by utilizing a steroid profile ratio to determine those newborns at risk for CAH. It is thought that false positives are decreased by 70% to 80%. Oklahoma is the second state to use this new technology for CAH screening.

Now the time had arrived to act and as the OSDH Coordinator for CAH, I had the responsibility to ensure Aidan received the needed health care services to ensure a timely diagnosis and treatment if needed. When the pediatrician was contacted, Aidan and his mother happened to be in the office waiting to see the doctor.

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Despite some medical complications in the first few days of life, he appeared to be a resilient newborn with few outward signs of the potentially fatal path for which his genes were coded. Aidan's doctor understood the gravity of the screening results and acted promptly. Within two hours the necessary diagnostic tests to rule-out CAH were completed and mom and baby were on their way to see a specialist. Dr. Kenneth Copeland, M.D., Chief of OU Children's Pediatric Endocrine Program, was waiting for Aidan's arrival to ensure all health needs were met to avert a potential life-threatening crisis. Dr. Copeland has years of experience treating children with CAH and was actively involved in the implementation of CAH screening in Oklahoma.

Only five hours had elapsed from that 8:00 a.m. page until Aidan's arrival at the OU Children's Physicians Diabetes Center. His electrolyte lab results were available and after a physical exam, Dr. Copeland confirmed that Aidan had the beginning clinical signs and symptoms of the most severe form of

CAH, classical salt-wasting. Dr. Copeland immediately began steroid treatment and hospitalized Aidan briefly as a cautionary measure. Within 48 hours of diagnosis, Aidan was on his way home, ready to resume his exploration of the new world into which he had arrived just 10 days before.

"I have only to look at the face of this bright-eyed little baby and his devoted mother and the answer is clear."

My next responsibility to Aidan is to provide care coordination services to ensure his health care needs are met. This long-term follow-up program is unique when compared to other state screening programs.

Over the next few years, assessment of the CAH screening program will require the painstaking gathering of data and the meticulous analysis of cost/benefit ratios and

outcome measures assessing health benefits of screening and long term follow-up care coordination services. From a funding standpoint, the scientific data will be critical in determining the program's success. From the human standpoint, Aidan benefited immediately from the expanded newborn screening program. He will also benefit from the long term follow-up services that will facilitate access to needed services to achieve optimal health throughout childhood.

For me, there's no question of the program's success. I am privileged to work with Oklahoma's newborns identified with an abnormal screen for CAH, whether the condition is confirmed or a false positive. After all, what is the value of a human life? This first and unforgettable experience remains foremost in my mind as I consider the question. I have only to look at the face of this bright-eyed little baby and his devoted mother and the answer is clear. ♥

Katherine Kirk is a CARES support group leader and the Endocrine Long Term Follow-up Nurse for CAH in the Oklahoma State Department of Health.

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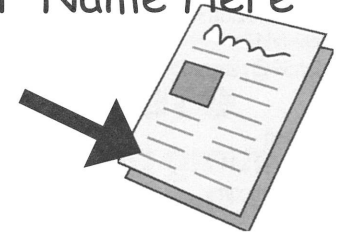
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Support Group News and Events

Florida Family Support Group
guest speaker

Dr. Dorothy Shulman, M.D.
All Children's Hospital
University of South Florida College of Medicine

Saturday, March 4, 2006
1-4 pm*

Lake Seminole Park
10015 Park Blvd.
Seminole, FL 33777

RSVP:
Patricia Tovar (727) 541-1683
Sonya Matson (941) 722-7560

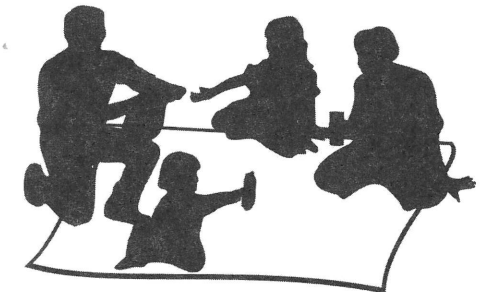
We look forward to hearing from you. If you can't make it for this date, please let us know if you would be interested in hearing about future meetings.

**A snack and refreshments will be provided.*

Oklahoma Support Group

What: Family Picnic
When: June 3, 2006 11:30am—1:30pm
Where: South Lakes Park, Event Center
4210 SW 119th Street
Oklahoma City, OK

RSVP: Kathy Kirk, RN, MSN
(405) 271-8001 ext43051
Katherine-kirk@ouhsc.edu



Event Idea . . .
Breakfast, Lunch or Dinner Fundraiser!

Food is always a good fundraiser, so try a fundraising breakfast, lunch or dinner event.
Remember: KEEP IT SIMPLE!

Example:
What: A Hotdog event fundraiser is a great way to start.

How: Have your local grocer donate the hotdogs, buns, condiments, chips and pop. You can use a propane BBQ or boiler for the cooking. Offer up a package deal like "1 Hotdog, bag of chips and a can of pop for only \$3!" **This way, you can raise more, faster.**

Where: Set up in a busy area, even the parking lot of your grocer.

IMPORTANT: Be sure to thank your grocer by giving them recognition for their food donation.



Thank You, Florida

We thank Sonya Matson and the Florida Family Support Group for all their hard work and dedication.

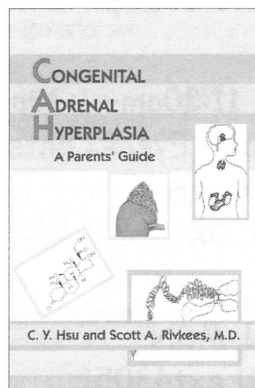
The group's BBQ fundraiser in November was a finger-lickin' success, raising almost \$700. Great Job!

Your Ad Here

Want to start a support group or advertise an event? Send us an email or give us a call and we will get you started!

Email: info@caresfoundation.org
Toll free: (866) 227-3737

Book Review

**CAH: A Parents' Guide**

Written by one of our medical advisors, **Scott Rivkees, MD**, and **Carol Hsu**, a parent of a child with CAH, this book is an excellent resource for both parents of newly diagnosed children as well as the more seasoned.

Written in language that is easy to understand, the book explains in detail all aspects of CAH care from genetics to prenatal diagnosis and therapy.

We use the book in our office almost daily and recommend it to all new members who join our organization. Comprehensive and well written, *CAH: A Parents' Guide* is a must have for all families and individuals affected by this disorder.

This book is available online through www.AuthorHouse.com, www.BarnesandNoble.com and www.Amazon.com.

New Product Update

E-HealthKEY®

MediAlert has developed a new comprehensive medical tool to track and manage your health. The E-HealthKey uses a USB device attached to your key chain to organize and manage your medical information including medications, insurance, vaccinations and family history.

While the E-HealthKEY is an excellent tool, it will take some time before all medical offices and hospitals will use it. It is highly recommended that all people with CAH wear a medical ID bracelet to alert medical personnel in an emergency.

The Emergency Screen, which contains your basic life-saving medical information (ie Adrenal Insufficiency) and the MediAlert emergency telephone number, appears each time the E-HealthKEY program is run. In an emergency situation, medical personnel can call MediAlert for other pertinent information, but the remaining information contained on the E-HealthKEY will remain secure and confidential.

The E-HealthKEY also allows for easy update of medical files and synchronization with MediAlert. You can update your records, attach and email documents, print customized emergency cards and even develop graphs using medical tests.

To order the E-HealthKEY or MediAlert bracelets call 1-888-633-4298 or go online to www.MediAlert.org.

Parent Tips

Emergency Kits

Keep your Solu-Cortef vial and syringe in a travel toothbrush holder. It will help to protect the needle and vial in your purse.

Create business card sized information cards in bright colors to staple to permission slips and give to caregivers. Include your emergency numbers, your physician's name and a brief explanation of adrenal crisis and physical stress.

- Natalie Rechner, WI

Breastfeeding

The Medela Starter Supplement Nursing System™ is a great tool for mothers who want to breastfeed but also need to supplement their baby's salt intake. The system, which is attached to you while breastfeeding, allows you to deliver a salt solution while breastfeeding (but you will need to ask your doctor how to mix the correct concentration). The Medela system is available online or through your lactation consultant.

-Robin Hendricks, AZ

**Emergency Instructions**

Those of you who received a Year End fundraising letter or recent new member packet also received our new Emergency Instructions.

The response we received from distributing this information has been overwhelming and we thank you all for your enthusiasm and kind words. Several of you have requested additional copies for caregivers and school officials and we are happy to provide them free of charge (but donations are always welcome).

If you would like additional copies or have not received this pamphlet, please contact our office and we will be happy to send them to you.

Email: Erin@caresfoundation.org
Toll Free: (866) 227-3737

Clinical Trials

**CAH Steroid Study
Riley Hospital for Children
Indianapolis, IN**

Everyone who has CAH is treated with a type of medication known as a "glucocorticoid." There are several different glucocorticoids available, including hydrocortisone (Cortef), prednisone and dexamethasone. Traditionally, many physicians treat children with CAH with hydrocortisone, which is a short-acting glucocorticoid that is usually given three times a day. Longer-acting glucocorticoids, such as prednisone or dexamethasone, are not as commonly used because of potential concerns related to growth. Little is known about the effects on growth, the pituitary gland, or CAH control of these different types of glucocorticoids. Improved understanding of the effects of hydrocortisone, prednisone, and dexamethasone in children with CAH would lead to better treatment options, including the potential of more simplified therapy with only once or twice daily dosing.

Our goal is to compare different glucocorticoids in children with CAH in order to optimize growth and control of CAH so as to avoid the consequences of over-treatment and under-treatment.

We are recruiting children who have classic CAH, are not yet in puberty, and are between the ages of 5 and 12 years old. Children will be enrolled at the General Clinical Research Center (GCRC) at Indiana University School of Medicine affiliated with Riley Hospital for Children in Indianapolis, IN. The study lasts for 18 weeks, but requires only 4 visits to Indianapolis. Three of these visits will consist of an overnight stay. Each participant will randomly be on a 6-week course of each glucocorticoid (hydrocortisone, prednisone, or dexamethasone).

All overnight stays, including labs, medications, and participation compensation, will be paid for by the study. If one is traveling a long distance, arrangements can be made to spend the night at the GCRC the night prior to an overnight stay at no cost.

If you are interested in learning more about our study, please contact either Dr. Erica Eugster at (317) 274-3889, or Dr. Todd Nebesio at (317) 274-3889 or by e-mail (tdnebesi@iupui.edu).

**CAH Volunteers Needed
for Natural History Study**

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

We are currently recruiting patients of all ages with CAH (both classic and nonclassic) for a natural history study at the National Institutes of Health (NIH) in Bethesda, Maryland. By studying the natural history of CAH in a large population of patients, we hope to define new aspects of the disease. This will allow us to develop new management and treatment approaches. Participants will be seen as outpatients at the NIH Clinical Center in Bethesda, Maryland and may be seen once, twice, or for ongoing care. Patients seen for a limited number of visits will have a full report sent to their private physician. All testing is free of charge and will include:

- Genotyping (patients with 21-Hydroxylase deficiency only)
- Hormonal evaluation including evaluation for insulin resistance
- Psychological and cognitive testing
- Bone age (growing children) and ultrasound

For more information or to volunteer, please contact Carol VanRyzin at the NIH at (301) 451-0399

The studies advertised on this page are new, as provided to us by the researchers. Other studies (previously advertised) are also available and can be viewed on the CARES Foundation website.

The studies listed on the website include topics such as (but not limited to): Women's Hormones, Prenatal Diagnosis, Children with CAH, Reproduction in Males with CAH, and NCAH.

For more information, please visit http://caresfoundation.org/clinical_trials.html.

Controversy Update

CARES Letter to Consensus Meeting Receives International Support

The Lawson Wilkins Pediatric Endocrine Society and the European Society for Pediatric Endocrinology held an "Intersex Consensus" meeting in October, the results of which will be published in 2006. Although congenital adrenal hyperplasia is the most common "intersex" disorder, no representative from the CAH community was allowed to attend. Instead, Cheryl Chase, a consumer advocate from the Intersex Society of North America (ISNA), who does not represent the CAH community, was invited on behalf of all people under the intersex umbrella. CARES Foundation, which objects to use of the term "intersex" and inclusion of CAH as an intersex disorder, protested the meeting with a letter to the organizers and participants. The letter objected to the exclusion of a CAH-specific advocate and made recommendations about surgery, terminology and development of centers of excellence. A copy of the letter is featured below.

October 2005

CARES Foundation, Inc. is the only US organization devoted entirely to congenital adrenal hyperplasia and is in constant contact with affected individuals and families. CAH is the most frequent disorder causing genital ambiguity and is substantially more common than other disorders of sexual differentiation.

As you are aware, the CAH community—those affected, caregivers and professionals—relies tremendously on the recommendations of LWPES [Lawson Wilkins Pediatric Endocrine Society] and ESPE [European Society for Pediatric Endocrinology]. The CAH Consensus Statement published in 2002 has become a vital tool in the care of CAH patients. Today, I am writing to express concern about certain critical aspects of the upcoming LWPES/ESPE Intersex Consensus Meeting. I am concerned that these issues I raise have the real potential of damaging the integrity of the consensus process and the good names of LWPES and ESPE. In the interest of time, the focus of this letter will be limited to: unbalanced representation by

consumer advocates; the need to base recommendations on scientific data; the terminology used to identify those with disorders of sexual differentiation as "intersex"; and creation of guidelines for designating centers of excellence for the care of those with these disorders.

In January 2005 I contacted Dr. Peter Lee to ask that CARES Foundation be included as a consumer advocate at the upcoming Intersex Consensus Meeting. I was very surprised to hear in September that no representative from any of the major worldwide CAH organizations (CARES, CLIMB, CAHSGA, or CAHNZ) was included in the conference. I have since learned that other special interest groups, namely ISNA, will participate in the meeting as the consumer representative. It is my opinion that the unbalanced representation of activists with their own socio-political agendas, poses a threat to the integrity of this meeting and the Consensus Statement that will result.

As we all know, medicine is far from an exact science, especially when it concerns disorders of sexual differentiation.

Recommendations about such complicated issues as genital surgery are far too serious to be left to lay opinion. Instead, physicians must adhere to credible, scientific data. To my knowledge, the consumer advocates included in this meeting have no such data to present but will offer only opinion and anecdotal evidence.

The topics to be discussed at the Intersex Consensus Meeting are surrounded in controversy and emotion. Because of this, I cannot stress enough how imperative it is the leaders of the conference focus on research-based evidence and not the personal agendas of those present. That said, its leaders should also recognize the lack of data and follow-up studies pertaining to outcome and to the cultural, familial and community differences of those born with these disorders. In addition to encouraging the study of these issues, leadership of LWPES/ESPE must encourage sensitivity and mental health training in the medical community, recognition of the evolution of treatment protocols and ethical issues, and increasing education of affected

(Continued on page 15)

(Continued from page 14)
individuals, families and the community at large.

The inability to make specific recommendations for disorders included under the umbrella "intersex" indicates a problem with generalization. Disorders of sexual differentiation vary widely, each having its own physical, emotional and social ramifications. What is true for one affected community may not be true for another. In that regard, we cannot allow representatives from other consumer groups to speak for the CAH community. Because a CAH advocate has not been granted participation in this meeting, I must respectfully ask that the issues discussed by consumer representatives exclude congenital adrenal hyperplasia. As a consensus statement has already been published on CAH, I expect it will

not be the focus of this conference and such exclusion will not be a problem.

In addition to rejecting representation by intersex advocates, the memberships of CARES Foundation, CAH New Zealand, CAH Support Group Australia and CLIMB reject the term "intersex" as their classification. Affected individuals find the characterization offensive and misleading, implying gender confusion—not just genital ambiguity. Clearly, the term has social connotations that can adversely affect females with CAH. As it has before, it is time for the nomenclature to evolve.


We urge those attending the meeting to recommend the designation of Centers of Excellence for the care of those with disorders of sexual differentiation that will have the appropriate tools, training and

experience to care for those with these disorders and support them and their families. This esteemed group of experts should be able to determine what guidelines are necessary to deem a medical center as a Center of Excellence. These centers may be disorder-specific as few medical centers have vast experience in all of the disorders of sexual differentiation. This should vastly improve the care of these individuals by consolidating their care in the hands of those with substantial experience, allow for the development of further understanding, improve support of families, and enhance opportunities for research.

Thank you for taking the time to review my comments, and I hope you will find them helpful. Please feel free to make the comments available to those at the meeting along with my contact information. ♥

In Support:

 Sue Elford, Chairwoman
CAH UK Support Group

 Helen Mann, Executive Director
CAH New Zealand Support Group

 Linda Powell, President
CAH Support Group Australia

Sincerely,



Kelly R. Leight, Executive Director
CARES Foundation, Inc.

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CAH CHAT GROUPS



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

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

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

New CAH Greek Group: Listservs for Greek speaking families and individuals affected by CAH. To learn more and join, go to <http://groups.yahoo.com/group/cahgreece> or <http://groups.msn.com/cahgreece> .

Continuous Treatment for Men with CAH

Erin Anthony

Historically, the focus of CAH research and treatment advancement has generally centered on women, as symptoms such as genital virilization and infertility were thought to be more problematic. While continuous treatment for women with classical CAH has never been questioned, there has been some debate as to whether men who are not salt wasting require lifelong treatment. However, researchers are now saying there is no question men with classical CAH should receive continuous treatment and are indicating that what we once thought

were "women's issues" are troublesome for men as well.

Transition to Adult Care

Perhaps the most fundamental reason for poor treatment for men and women with CAH is a lack of adult CAH specialists. In the US, patients found to have CAH are usually followed by a pediatric endocrinologist until the time of transition, around 18 years of age. Unfortunately, after 18, patients are often left to find their own doctor. Many end up seeing an internist unfamiliar with the condition or forego treatment altogether.

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In a recent article in the journal *Hormone Research*¹, German researchers Mönig and Sippell propose special considerations for the time of transition. The researchers recommend the patient be transferred during the course of an extensive consultation with the new physician, with special attention paid to a few major problems: adrenal crisis, impaired gonadal function, and adrenal tumors.

Adrenal Crisis

Adrenal insufficiency and adrenal crisis are obvious problems

(Continued on page 17)

(Continued from page 16)

for all CAH patients, but, according to Mönig and Sippell, there are no reports in the literature on the frequency of adrenal crises in adult CAH patients.

That said, the basic biochemical defect of CAH, insufficient cortisol production by the adrenal glands, is a lifelong condition. According to the authors, there is no reason to believe that the adrenals will begin to function in adulthood even though steroid replacement requirements may change.

A 2004 article in *Experimental and Clinical Endocrinology & Diabetes*², to which Mönig and Sippell contributed, indicates a possible decrease in dosage for adults. The reasoning for the change is that, with adulthood, the goal of treatment shifts from optimizing growth and preventing virilization to preservation of fertility and good general well being and physical performance. However, the authors are also careful to highlight the need to repeatedly inform adult patients about the symptoms of adrenal insufficiency and the need to increase steroid doses in cases of physical stress.

Impaired Gonadal Function

Although it has long been expected that persistent increase of adrenal androgens results in small testes and impaired spermatogenesis, normal testicular function has been described in untreated CAH patients, Mönig and Sippell said. However, this should not be the only reason for infertility concerns in men with CAH. Fertility may be further compromised by testicular adrenal rest tumors, caused by undersuppression of ACTH, the authors said. Continuous treatment, according to the authors, is necessary

to prevent both occurrences and preserve normal fertility.

Another complication the authors cite is development of testicular hypertrophy, frequently encountered in classical CAH. This condition, which may at first be visible only on ultrasound but can grow into palpable masses more than 10cm in diameter, is usually due to insufficient steroid treatment. It is important to note that while these lesions are not harmful in themselves and do not necessarily preclude fertility, they may, as the authors note, impair spermatogenesis and Leydig cell function (testosterone production).

Another problem arises when a CAH patient sees a urologist unfamiliar with the condition, the authors said. While the tumors are of a hard consistency and resemble malignancy, they are usually benign and reversible if steroid therapy is optimized. It can also be very difficult to distinguish between Leydig cell tumors and adrenocortical rest hyperplasia—putting patients at a high risk for unnecessary removal of the testicles.

Adrenal Masses

CAH may also be associated with increased incidence of adrenal tumors, Mönig and Sippell said. While most adrenal masses in patients with CAH are benign, a mass over 5cm in diameter requires surgical removal. This makes the prevention of such tumors another argument for continuous treatment of male patients with classical CAH.

Osteoporosis

Osteoporosis is typically thought of as another "women's issue," but men with CAH should also take note.

According to the 2003 article referenced before, bone loss is one of the major complications of steroid treatment in pharmacological doses. At risk are patients who were obviously over-treated for years.

Summary

CAH is a disorder affecting both sexes equally. However, because the symptoms of androgen excess appear to be more of a problem for women, men have not received the attention they deserve. The article by Mönig and Sippell emphasizes the fact that men with classical CAH need to receive continuous treatment and should be aware of problems surrounding adrenal crisis, impaired gonadal function and adrenal tumors. It also highlights the need for better education of internists, adult endocrinologist and urologists concerning treatment of adults with CAH. Integral for good healthcare is smooth and conscientious transition from pediatric to adult care. ♥

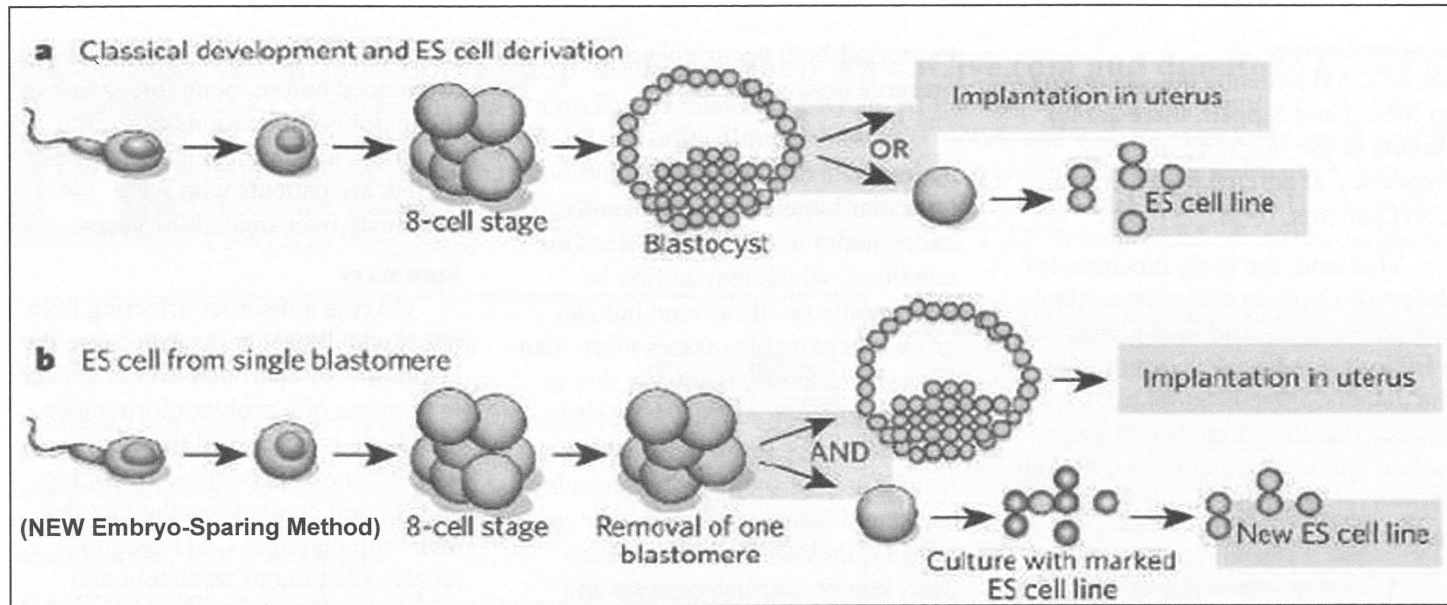
Issues for Men with Classical CAH

- Transition to Adult Care
- Adrenal Crisis
- Adjustment of Dosage
- Impaired Gonadal Function
- Adrenal Tumors
- Osteoporosis

References:

¹Mönig, Heiner and Wolfgang Sippell, "Congenital Adrenal Hyperplasia in Adulthood: Do Men Need to Continue Treatment?" *Hormone Research* 2005; 64 (suppl 2):71-73.

²Kruse, B. et al. "Congenital Adrenal Hyperplasia—How to Improve the Transition for Adolescence to Adult Life," *Experimental and Clinical Endocrinology & Diabetes* 2004; 112: 343-355.



a) Embryonic Stem Cell derivation using previous techniques without the possibility for embryo viability. b) NEW embryo-sparing method allows cells to be obtained at an earlier stage for viability of the embryo, as used in pre-implantation genetic testing.

Stem Cell Research
(Continued from page 1)

Embryonic Stem (ES) Cells are derived from the early embryo, prior to the time of implantation into the uterus, and are able to become any cell within the body. Given this enormous potential, scientists hope to one day be able to coax ES cells to develop into a wide range of tissues, which will serve as the basis for treating many diseases.

The Ethical Debate Surrounding the use of ES Cells

Until recently, the generation of ES cells required the destruction of the early embryo, which raises significant ethical and moral issues for many individuals. **Important advances have recently demonstrated that embryonic stem cells can be obtained from an 8 cell stage embryo without destroying it in the process.**

The procedure to generate ES cells without destroying the embryo uses well-established methods to isolate a single cell from the embryo

before it is implanted into the uterus. This technique is already widely employed by doctors to identify genetic mutations in embryos without their destruction. In addition, other promising approaches, which are designed to avoid the use or destruction of embryos, are currently being developed.

Cell-Based Therapy for Congenital Adrenal Hyperplasia (CAH)

The potential role of stem cells in the treatment of a wide variety of diseases, such as diabetes and Parkinson's disease, has recently gained considerable attention. Cell-based therapy involves the use of highly specialized cells to replace a tissue's missing function, such as hormone replacement.

CAH is an ideal condition for cell-based therapy given (1) the need for precise hormonal replacement, (2) the need to increase steroid levels during times of stress and (3) the highly complex daily variation in normal cortisol production rates. Each of these functions could ideally

be provided by cells, which are responsive to normal hormonal signals.

Because individuals with CAH may have the ability to regulate cortisol in a normal manner, it is hoped that replacement with either (1) adult stem cells from the adrenal gland or (2) ES cell-derived steroid producing cells might restore normal adrenal hormone production. Ideally, cell-based therapy would also result in partial-to-complete adrenal androgen suppression from the patient's own adrenal glands, which may be possible if sufficient cortisol levels are produced by these cells, though this remains to be proven.

Adult Stem Cells from the Adrenal Gland

The adrenal gland has long been assumed by researchers to contain a population of adult stem cells, which are thought to maintain this essential tissue for the lifespan of the individual. Proof of their existence, however, has been lacking. Recent studies from our laboratory have confirmed the presence of adult stem

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cells in the adrenal glands of rodents, raising the possibility that they also exist in humans.

While there is little doubt the tools required to study adult stem cells will emerge during the next decade, it is quite clear the field of adult stem cell biology, for tissues other than the bone marrow, is still in its infancy. In the meantime, ES-cell based strategies may offer the best potential for cell-based therapy for individuals with CAH and other conditions.

ES Cell-derived Steroid Producing Cells

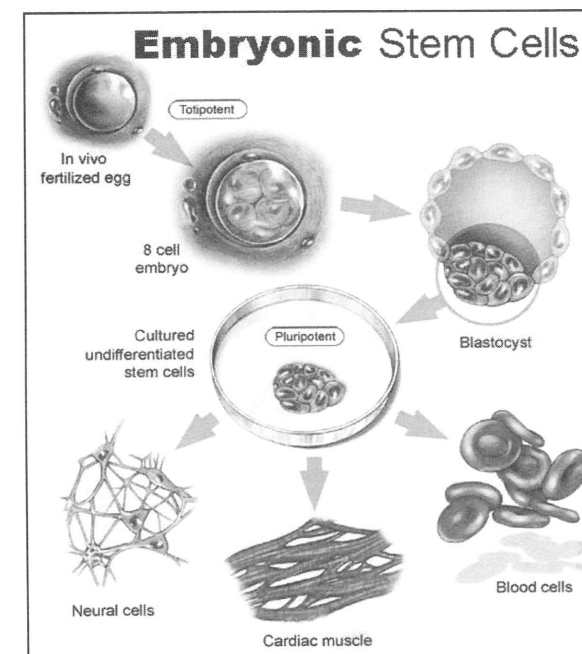
The ability to coax ES cells into becoming cortisol-producing cells would represent a major step towards cell-based therapy for individuals with CAH.

The methods required to turn ES cells into mature cells of any particular tissue will ultimately require determining how a tissue normally develops. For most tissues, including the adrenal gland, these methods are currently being established.

In our laboratory, we have developed a highly specialized screening system to help identify the specific developmental steps (factors) required for adrenal development. After identifying these factors, we hope to establish the methods required to coax ES cells into cortisol-producing cells and ultimately test their ability to restore normal adrenal function in model systems of CAH.

Advantages of Cell-Based Therapy for CAH

Unlike current cell-based therapies (like bone marrow transplantation), which require individuals to share common immune markers to avoid rejection, cell-based therapies offer the theoretical advantage of being tailored to the individual, thus potentially avoiding immune rejection and the need for immune suppressing drugs. For example, someday it may be possible to correct the genetic mutations causing CAH in individuals by applying gene therapy to an individual's own stem cells (adult stem cells or ES cells), and therefore fully cure CAH.



The therapeutic potential of ES cells, whose unique power to self-renew and to develop into any human tissue or organ, is expected to transform the way that we treat, prevent and cure disease. Instead of drugs, cell-based therapy hopes to provide the means of restoring normal function to tissues and organs – including the adrenal gland. To help ensure that patients with CAH are not denied the potential future option of ES-derived cell based therapy, we have initiated fundamental research in this emerging area. ♥

Stem Cell Questions and Answers

What are Stem Cells?

Stem cells, the body's master cells, have the potential to form one, many or all cell types of an organism and have the ability to replicate (self-renew).

What are Adult Stem Cells?

Cells derived from multiple tissues with the capacity to self-renew and to give rise to specific cell types within each tissue.

What are Embryonic Stem Cells?

Cells derived from the early embryo with the potential to give rise to all cells in the body.

What is Cell-Based Therapy for CAH?

The ability to replace adrenal gland function with highly specialized steroid producing cells.

Can Embryonic Stem cells be created without destroying the embryo?

Yes! Using well-established methods scientists have recently been able to generate embryonic stem cells without destroying the embryo.
(See diagram B on page 18.)

Dr. Breault is an Assistant in Medicine in the Division of Endocrinology at Children's Hospital of Boston and an Instructor in Pediatrics at Harvard Medical School. In addition, he is an Affiliate of the Children's Hospital Boston Stem Cell Program, and a member of the Harvard Stem Cell Institute and the International Society for Stem Cell Research.

“What’s that bracelet on your arm?” . . .





...“It’s an Every1CARES bracelet that raises awareness about CAH.”

This is a dialogue that goes on every day, when people wear their CARES bracelets in public. In addition to accessorizing your family’s attire, Every1CARES bracelets are a great way to make people aware of CARES’ mission. CARES’ blue silicone bracelets are available for sale in both adult and youth sizes (\$3 each). They make great party favors and even napkin holders.

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To order, call toll-free 1-866-227-3737 or order online at www.caresfoundation.org.

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



Thank you, Thank you!

To those of you who have already sent in your donation to our Year End campaign.

It’s never too late.

For those of you who have not yet done so, please send in your donation. We need your help!! We still have so much work to do supporting families like yours who are affected, raising awareness about CAH and funding research for a cure.

Please send your donations to:
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189 Main Street, Millburn, NJ 07041
or logon to our secure website:
www.caresfoundation.org.

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