

CARES Foundation



Congenital Adrenal hyperplasia Research, Education and Support Foundation, Inc.

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Our First Newsletter is Here!
December 2001
Happy Holidays to All!



The Lecture/ Support Group Meeting Was a Huge Success!

On November 14, 2001, CARES Foundation, Inc., in conjunction with Dr. Maria New and New York Presbyterian Hospital-Weill Cornell Medical Center, hosted its first lecture/support group meeting for CAH families. Dr. New presented the findings of her newly published study on prenatal therapy for mothers at risk of giving birth to a CAH virilized females. For discussion of that study, please see article below. Dr. Susan Baker, the psychologist at Cornell who works with Dr. New and counsels many CAH patients and their families, also presented her findings on a study of frequently asked questions by adults who suffer from CAH and non-classical CAH. Most affected individuals were concerned about treatment options and side effects of medications and the disease. Dr. Baker is preparing a booklet that will address the most frequently asked questions. Dr New also discussed her gene therapy research that holds the promise of a potential "cure" for CAH. CARES Foundation, Inc. presented Dr. New with a check representing its first research grant for the Gene Therapy program at Cornell-Weill Medical Center to support Dr. New's research.

The lecture was videotaped. Copies will be available for a small fee from CARES Foundation in mid-December.

The best part of the evening for me (Kelly), however, was meeting the many families who attended. There were several whom I had spoken to over the phone or corresponded with, but I was finally able to put a face to the name. People brought photos of their beautiful children and we shared our experiences. One couple attended who had just given birth to a CAH daughter 13 days prior. Their daughter was still in the hospital. Parents who had been through the same experience were able to reassure them that their daughter would be fine. It was a wonderful, heartwarming, supportive and educational experience for me and I hope for the others who attended. I hope many more will be able to attend our next lecture/support group meeting on February 27th.

Our Next Lecture/ Support Group Meeting with Dr. New is Scheduled for February 27th! Save the Date!

On February 27th, 2002, CARES Foundation, Inc. will be sponsoring another lecture and support group meeting with Dr. New at NY Presbyterian Hospital -Weill Cornell Medical Center. The lecture will address issues of concern for non-classical CAH individuals and families, but most topics will be of interest to classical CAH patients as well. Since Dr. New ran out of time at the last lecture and was unable to discuss Growth Hormone therapy, we hope she will address this subject at this lecture. Please mark your calendars and plan to come! An Email alert with details will be sent out closer to the event.

New Study By Dr. Maria New on the Benefits and Safety of Prenatal Therapy

In the December edition of the Journal Of Clinical Endocrinology and Metabolism, Dr. New will be publishing her study *Prenatal Diagnosis for Congenital Adrenal Hyperplasia in 532 Pregnancies*. Her study confirms the safety and efficacy of prenatal treatment for expectant mothers and their babies at risk for virilization from CAH. It shows no apparent risk to child (CAH-affected or not) or mother.

The treatment involves the pregnant mother taking dexamethasone starting at or before the 9th week of pregnancy to suppress the excess adrenal androgen secretion and prevent virilization should the fetus be a CAH-affected female. Once diagnosis is made via chorionic villus sampling or amniocentesis and the fetus is an unaffected female or a male, the treatment is discontinued. If the fetus is diagnosed as an affected female, the treatment is continued through the remainder of the pregnancy. The mother must be carefully followed by during the pregnancy. The study shows that there were no real differences in the symptoms during pregnancy between those treated with the dexamethasone and those untreated, except that additional weight gain (which was lost after pregnancy), edema, and striae (stretch marks) were somewhat greater in the treated group. There were no lasting side effects in the fetuses proving that the treatment is safe. Most importantly, of those mothers who began the treatment at or before the 9th week of pregnancy, the amount of virilization was substantially reduced or in many cases completely eliminated. The vast majority of female CAH-affected babies whose mothers were treated with Dex beginning by the 9th week of the pregnancy and completed the course of treatment did not require any surgery for virilization. The

prenatal treatment spares the female the consequences of genital ambiguity, potential surgery and possible misassignment.

We at CARES Foundation, Inc. thank Dr. Maria New for her groundbreaking research on prenatal therapy that will improve the quality of life for so many CAH affected females.

Update on Newborn Screening

Beginning on January 1, 2002, a number of states will add CAH to their newborn screening programs including Oregon, Virginia, Arizona and Missouri. Arizona and Oregon will be doing a 2-part screen that detects a greater number of simple virilizers and non-classicals than the standard screen. In April 2002, New York will finally begin screening for CAH, and the state will be doing genotyping as an adjunct to regular testing. This will help to determine the type of CAH affecting the child. Vermont just approved adding CAH screening, but has no set start date finalized. We also applaud the Health Departments of Maryland, Mississippi, New Jersey, Delaware and Maine that began CAH newborn screening in the last 6 months. With the addition of Vermont, the number of states that screen for CAH is 34!

Dr. Piero Rinaldo of the Mayo Clinic is completing research on the usefulness of Tandem Mass Spectrometry technology in CAH screening and its ability to eliminate up to 95% of all false positive results, as well as its ability to detect a greater number of simple virilizers and non-classicals. We are looking forward to seeing the results of his research.

In addition, NeoGen Screening, Inc., a private laboratory in Pennsylvania, is doing research on genotyping in CAH newborn screening. This would provide better accuracy in newborn screening for CAH.

CARES Foundation's Advocacy Efforts for Expanded Newborn Screening

CARES Foundation is stepping up its efforts to advocate for CAH newborn screening in all states. Unfortunately, many states still do not screen for CAH at birth despite its life-saving benefits. That means that all classical CAH male babies and those females who are so virilized they are sent home as males born in those states are at risk of dying or suffering traumatic and life-threatening adrenal crisis.

For more information about CAH newborn screening, please visit the CARES Foundation website at <http://www.caresfoundation.org/nbs.html>.

In California, we are working with the March of Dimes, which is making CAH newborn screening one of its top legislative priorities. I need California residents to help CARES Foundation advocate for the addition of CAH screening to the California newborn screening program. It involves letter writing, making telephone calls to legislators, reaching out to and speaking to the media, speaking at hearings if necessary, and taking advantage of any personal contact we might have. It would be especially helpful if someone who has a connection to the Governor of California, as he is the person holding things up right now.

We should also get our California pediatric endocrinologists, OB/GYNs and pediatricians to get involved. Ask them to write letters and make phone calls.

I need one person to act as the State coordinator, who will work closely with the March of Dimes and coordinate the efforts of the other parents. Beyond that I just need lots of "noise makers" It may take a while, but if we can make enough noise, they can't ignore us.

Also, any parents in any state who have, unfortunately, lost their child because they were not identified as having CAH in time, please contact me. If you would be willing to speak out about your tragedy, you might be able to help save the life of another family's child.

We have parents in Ohio and Utah coordinating the efforts there as well, but they need more help. If you live in a state that does not screen for CAH at birth and you are willing to work on advocacy, your efforts would be truly helpful. CARES Foundation will instruct you on advocacy if needed.

Please email or call Kelly Leight if you can help out at Kelly@caresfoundation.org or call 1-866-227-3737 (outside NJ) or in NJ - 973-912-3895.

Kelly Leight To Speak as A Panel Member at Robert Wood Johnson Medical School on January 8, 2002.

Kelly Leight has been invited to participate in panel discussion of the issues and controversy surrounding surgical options for children born with ambiguous genitalia before the medical students and faculty at Robert Wood Johnson Medical School in New Brunswick, New Jersey. The other panelists include Dr. William Reiner from Johns Hopkins, Dr. Nina Williams, a therapist who counsels women with CAH, and several CAH adult women. Kelly Leight's presentation will focus on the necessity for support for parents and the importance of sensitivity to the concerns of parents. The panel discussion is from 9-11 am at the medical school. The lecture is not open to the public, but if you wish to attend, contact Kelly Leight, and she will make entry arrangements for you.

Volunteers Needed!

Please Contact Kelly Leight if you can help with any of the following:

We need someone to work on getting the charity registrations in key states. This involves phone calls (out of state) and sending the needed documents to the states.

We need someone to work with another NY state mom to organize a family day event in the spring. Soup to nuts--do everything. Call Susan Donohoe at 845-265-3306 if you can help with Family Day. Susan is also coordinating the creation of a video about living with CAH. If you have special skills related to video and film, please call her as well.

We need someone to put together our newsletter (semi or bi annual) with updates via email. I also need written contributions from parents on topics of interest.

We need someone to coordinate fundraising activities and someone to write some grants. Contacts with drug companies and medical insurance companies help.

Once all of our materials are together, we will need someone to mail them out whenever we get a request. We also need people to bring brochures to their endocrinologists' offices.

We need a national newborn screening coordinator--someone to keep track of CAH screening in the states.

Kindly specify exactly which of the above projects you are willing to help with.

About CARES Foundation, Inc.

Two years ago, our daughter, Alyssa, was diagnosed with a genetic disorder called non-classical Congenital Adrenal Hyperplasia (CAH). As we attempted to research the disease, we found that there were no active support organizations for families and affected individuals and few resources for learning about the disease despite the fact that the non-classical form of the CAH is the most common genetic disease identified to date. My husband Adam and I felt that we could make a difference in the lives of those affected by all forms of CAH and their families and could help to publicize the non-classical form of the disease in the most affected ethnic groups. So, this past year, we formed CARES Foundation, Inc. (Congenital Adrenal hyperplasia Research Education and Support), a non-profit 501c(3) organization. Its purpose is to educate the public and physicians about all forms of Congenital Adrenal Hyperplasia, its symptoms, diagnostic protocols, treatment, genetic frequency, the necessity for early intervention and benefits of newborn screening. It is also dedicated to providing support and information to affected individuals and their families. We also will provide medical scholarships to those in need and donate funds to CAH-related research. CARES Foundation, Inc. can only continue its work through your tax-exempt donations and support.

Thank You,

Kelly R. Leight, Founder and Executive Director

Please Consider a Donation to CARES Foundation or Help with Fundraising

This holiday season, please consider making a tax deductible contribution to CARES Foundation, Inc. So far, all expenses of CARES Foundation have been subsidized by the Executive Director, Kelly Leight and her husband, Adam, with the exception of a small grant from the PerkinElmer-Wallac company, a company that produces newborn screening equipment. A donation form is included in this newsletter. We have many plans in the works for the future, including a video on the subject of Living With CAH aimed at families with newly diagnosed children; printing of brochures and purchasing of educational materials for families; outreach and educational efforts; a family day event for the spring to be held in the NYC vicinity; the NYC lecture/ support group meeting set for February and similar events to be held in Maryland, California and Texas; preparation of a nationwide list of pediatric endocrinologists with experience in treating CAH; continued advocacy for CAH newborn screening; grants for CAH related research; and medical scholarships for families in need of specialist care. CARES Foundation, Inc. has been chosen to be the featured support organization for the March 2002 issue of Exceptional Parent Magazine.

CARES Foundation Accomplishments for 2001:

Organization Established 3/01;

Tax Exempt status --501(c)3 received from IRS 8/01;

Grant received from PerkinElmer-Wallac 8/01;
Played a major role in the expansion of newborn screening in New Jersey including CAH;
Contributed to two New York Times articles on Early Puberty written by Gina Kolata;
Featured in CBS Evening News Segment on Early Puberty;
Featured in New Jersey Jewish News article on Non-classical CAH and its prevalence in the Jewish population;
Establishment of CARES Foundation web site, www.caresfoundation.org 4/01;
Organizational Board Member of Tyler For Life Foundation (advocacy group for newborn screening);
Pending memberships in NORD and Genetic Alliance;
Lecture and Support Group meeting 11/14/01;
First CARES Foundation Research Grant given to CAH Gene Therapy Program at NY Presbyterian Hospital-Weill Cornell Medical Center 11/01;
First Newsletter 12/01.

CARES Foundation, Inc cannot continue these efforts without financial support from you. Please consider giving generously.

You might also wish to assist in fundraising. Adam and Kelly Leight will be putting a fundraising letter in all of their Holidays cards this year and we hope you will consider doing the same. Here is a form of fundraising letter:

Dear Family and Friends,

In this holiday season, our family has chosen to support an organization dear to our hearts. It is a relatively new organization, so any gift you can make would be particularly meaningful. CARES Foundation, Inc. (Congenital Adrenal hyperplasia Research Education and Support) is a non-profit 501c(3) organization dedicated to educating the public and physicians about all forms of Congenital Adrenal Hyperplasia, its symptoms, diagnostic protocols, treatment, genetic frequency, the necessity for early intervention and benefits of newborn screening. It is also dedicated to providing support and information to affected individuals and their families. CARES Foundation, Inc. can only continue its work through your tax-exempt donations and support.

As many of you know, [I am] our child[ren], _____, is [are] affected by a form of Congenital Adrenal Hyperplasia. Congenital Adrenal Hyperplasia is a family of inherited disorders affecting the adrenal gland. The most common form is 21-hydroxylase deficiency, which is inherited in severe or mild forms. The severe form, called Classical CAH (CAH), is usually detected in the newborn period or in early childhood. The genetic frequency of Classical CAH is approximately 1 in 15,000 births. The milder form, called Non-Classical CAH (NCCAH), may cause symptoms at anytime from infancy through adulthood. Non-Classical CAH is a much more common disorder than Classical CAH. It affects 1 in 100 in the general population of New York City and is most common in certain ethnic groups. It affects 1 in 27 Ashkenazi Jews, 1 in 40 Hispanics, 1 in 53 Croatians, and 1 in 300 Italians. This frequency makes NCCAH the most frequent genetic disorder identified to date.

Congenital Adrenal Hyperplasia (CAH), in its severest form, is a life-threatening disorder. It is caused by a defect in the gene coding for an enzyme, steroid 21-hydroxylase, important to cortisol production and without which adrenal insufficiency results. Its severest form, called Classical CAH, carries with it a salt-wasting factor-- an inability to produce aldosterone, which, if undetected at birth, can lead to adrenal crisis and death. Frequently, these babies present for urgent medical attention at a time when they are beyond resuscitation. Classical CAH children with careful medical care can live a long and normal life. There are many health issues that families and individuals are presented with throughout their lives, but the disease can be managed with medication.

Non-Classical CAH (NCCAH), is much more common than Classical CAH. But-perhaps because it is less severe and harder to recognize than the classical form of the disease, NCCAH has not received the attention it deserves as a widespread condition that affects health and quality of life. Symptoms of NCCAH include: premature puberty, premature adrenarche (manifest as body odor or pubic hair in young children), abnormal bone aging leading to rapid growth in early childhood but ultimate short stature, severe acne, oily hair and skin, and later in life, infertility in men and women. Anxiety and depression are also common symptoms. These symptoms are treatable with glucocorticoids.

CARES Foundation, Inc. provides many services to the CAH community including a physician referral service for parents and affected adults, educational lectures, support groups, maintenance of a web site with many resources, and providing of free informational materials. It also provides medical scholarships to needy families for specialist care and grants to medical institutions for CAH-related research. We hope to expand the services CARES Foundation, Inc. offers to the CAH community and its efforts to educate the public, the medical community and the most affected ethnic groups about NCCAH. It also supports parent efforts to expand newborn screening programs to include screening for CAH at birth as it did with great success in several states this past year.

We ask you to consider supporting CARES Foundation, Inc. and its efforts to provide these services and support to those affected by this disease. Your tax deductible contribution to CARES Foundation, Inc. will help the organization to continue its work.

Happy Holidays to all!

[Your Names]

To our CAH adults and families:

We are trying to create a workable database with the full names and addresses of the CAH community. Please help us to help you. For many of you, I only have a first name and email address. You can cut and paste this form and put it in an email to Kelly Leight at Kelly@caresfoundation.org or call and leave us a message with the information at **1-866-227-3737**. Thank you for your help! All information will be kept confidential.

Please tell us the following:

Name _____
Address _____
Telephone number _____
email address _____
Affected adult _____
Child[ren] with CAH and how many _____
Type of CAH:
Non-classical/late-onset CAH _____
Salt-Wasting _____
Simple Virilizing _____

Please put us on your email alert list _____
Snail mail only _____

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NAME: _____
ADDRESS: _____
TELEPHONE: _____
EMAIL: _____

I/we would like to learn more about CARES Foundation, Inc and CAH. _____
Please put me on the CARES Foundation, Inc. email list. _____

Your Tax Deductible Contribution:
\$1000 _____ \$100 _____
\$500 _____ \$50 _____
250 _____ \$OTHER _____

Matching Gift Program Available? Name of Company: _____

Check enclosed \$ _____

Please charge to my credit card. MC _____ VISA _____ AMEX _____
Number _____ Exp. date _____

Signature (for credit card authorization) _____

**From Our Family To Yours--Our Best Wishes of the Season
And a Happy, HEALTHY New Year!
Kelly, Adam, Josh and Alyssa Leight**

