

Weight Management for Children with CAH

By Michelle May, MD

Hot from the headlines: Obesity has reached epidemic proportions in our society, fast approaching smoking as the leading cause of preventable disease and death. Although this is a frightening statement, obesity can be prevented.

Prevention of obesity and the development of lifetime healthy eating habits begins in childhood. Currently, 15 percent of children and adolescents are overweight or obese, putting them at risk for high cholesterol, high blood pressure, and type 2 diabetes. They may also face social stigmatization, have low self-esteem, and face an increased chance of adult obesity.

Children with CAH are particularly at risk for weight problems due to the body's reaction to glucocorticoid therapy. Some children complain of increased appetite with medication increases, and oversuppression can cause excess weight gain. Even once the oversuppression is eliminated, excess weight may still continue to be a problem.

So what can you do? Consult your child's endocrinologist and primary care physician to discuss whether your child is significantly overweight. Then, determine if there are medical issues contributing to their weight problems (such as oversuppression), or if their weight is causing any medical problems. Then together, you can determine the best approach for helping your child reach a healthier weight.

Many overweight children do not actually need to lose weight, but instead, can maintain their weight while they "grow into it." Even for extremely overweight children, weight loss

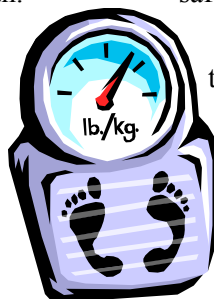
should be gradual. Since many overweight children are still growing, their diet must be nutritious and their exercise program should be safe and enjoyable.

The strategies below are important for both the prevention and treatment of childhood weight problems:

Build Healthy Attitudes

- Demonstrate your unconditional love for your child. Children—especially overweight children—need support, acceptance, and encouragement from their parents.
- Build self-esteem by focusing on all of your child's positive qualities, unique talents, and individuality. By developing interests and skills that increase their success and pleasure, they will be less likely to turn to food for fulfillment.
- Help your child develop good communication skills, encourage them to

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Save the Date!!

CARES CAH CONFERENCE

SUNDAY, OCTOBER

24, 2004

OVERLOOK HOSPITAL

SUMMIT, NEW JERSEY

Look for details in our upcoming mailings or check our website.

A Message from the Executive Director:



Dear Friends,

Best Wishes for a Happy and **HEALTHY** New Year to all! As the New Year begins, we have lots of new initiatives and many exciting changes here at CARES Foundation.

Our Conference in Los Angeles

On October 18, 2003, we held our third conference in Los Angeles at Childrens Hospital Los Angeles. It was a phenomenal day for me and hopefully for those who attended as well. We had over 165 people attend, some coming from as far away as the United Kingdom. This was the first time we offered continuing education credits for nurses, and were pleased to have a number of nurses in our audience in addition to some physicians and medical students. Our first speaker was Dr. Mitchell Geffner, our host from Childrens Hospital Los Angeles, who gave a thorough overview of CAH. Then Dr. Maria New, from Weill-Cornell New York Presbyterian Hospital in New York, spoke about her experience in treating CAH, discussing prenatal treatment, use of growth hormone and other new developments in CAH treatment. Dr. George Cunningham spoke about the newborn screening situation in California. Dr. Sheri Berenbaum, from Penn State University in University Park, PA, spoke about behavior in CAH. Dr. Ricardo Azziz from Cedars Sinai in Los Angeles explained how CAH/NCAH affects women of reproductive age. Dr. Dix Poppas from Weill-Cornell New York Presbyterian Hospital in New York gave a thoughtful explanation of surgical reconstruction for virilized girls with CAH. It was a long, but

exciting day. I enjoyed meeting so many of our members and watching the camaraderie that we all felt, being in a group who knows what we all have been through and are dealing with on a daily basis in coping with CAH.

We thank Dr. Mitchell Geffner and Childrens Hospital Los Angeles for hosting the event and supporting us all through the planning process. They were just terrific and so helpful. I also want to thank all of our wonderful speakers for donating their time to come out to California and address our group. We are incredibly grateful to them all; and they were wonderful! I want to thank the California Department of Health for their assistance in helping us arrange for continuing education credits for the nurses who attended the conference and for issuing those credits.

I also want to thank all of the volunteers who showed up to help with the set-up and clean-up, signing everyone in and greeting the attendees. I truly could not have done it without you!!! Special thanks to Starbucks for donating the delicious coffee break goodies and to Jennifer Cribbs for arranging the donations!

Our next conference is coming to Summit, New Jersey on Sunday, October 24, 2004 at Overlook Hospital.

CARES Collaborating on NIH Clinical Trial

CARES is pleased to announce

that it is collaborating with Cornell and Dr. Maria New on a clinical trial that will look at the natural history of genetic steroid disorders. We are honored to have been included in this important study and will be reaching out to the membership to help with this in the near future. You can read more about this study on Page 11.

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Executive Director's Message

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CARES Foundation Moving to New Offices Soon

We will be moving out of my basement soon! With our growing membership and the need for more and more services, we have outgrown my basement and are moving into real offices in a nearby town in the next couple of months. This is a big step for us, but will allow us to add additional staff to better serve the needs of the CAH community. Come visit us!

Fundraising Efforts and New Director of Development, Linda Mears

Many thanks to all of you who have made donations to CARES in response to our holiday letter. Our year-end campaign has raised over \$45,000 and donations are still trickling in. If you have not made a donation yet, but are able to do so, we would be most grateful. You can mail in a check or give online at our web site. Many thanks to the members who reached out to their friends and families on our behalf to contribute to CARES and to our Board Members, whose fundraising efforts have been quite successful. Special thanks to board members Adam Leight, Brad Smith and Dr. Diane Snyder, our top fundraisers this year.

Linda Mears has joined our staff as the Director of Development. Linda has 15 years experience in the field, having worked at such venerable institutions as the New York Philharmonic, Lincoln Center and the Pingry School. We are pleased and honored to have her join our staff. Linda can be reached via email at Linda@caresfoundation.org.

Our new **Fundraising Guide** is now available for those interested in helping with our fundraising efforts.

Just email/call us and we will mail one out to you. As our organization has grown, the need for our services has grown too. So, in order meet the needs of the CAH community and truly represent its interests to the rest of the world, our need for funding has increased. Also, if we want to make difference in making substantial grants for research, we must raise large sums of money to do this. We need your help. Please consider arranging a charity event for CARES. Our fundraising staff are here to help you with ideas and planning. See the wonderful story about Brad and Tina Smith on Page 16.

Our New Web Site

One of our members, Sue Bianchi, has offered to help us redesign our web site. It will have more extensive materials and will have an archive of our newsletters. Many thanks to Sue for all of her hard work and to my brother-in-law, Bruce Estes, for designing the new front page for us. We will send out an email as soon as our new website is up and running.

Future Plans

We are moving forward with our long term plans—our international drug donation program, trying to persuade the US drug companies to add CAH medications to their patient assistance programs, translating our materials and web site into Spanish, expanding newborn screening, and a Jewish awareness program. We need volunteers to help us with these efforts, so if you can help, please let us know.

Warm Regards,

Kelly

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Dr. Ricardo Azziz



Dr. Sheri Berenbaum



Dr. George Cunningham



Dr. Mitchell Geffner



Dr. Maria New



Dr. Dix Poppas

Weight Management in Children with CAH

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express their feelings, and teach them effective coping skills to decrease the chance that food will serve that purpose.

- Emphasize the importance of good health, not ideal weight.
- Never tease or criticize a child or adolescent about their weight. Such comments are hurtful and can stick with a person for a lifetime.
- Be a positive role model. When your child observes you enjoying healthful foods and physical activity, they are more likely to do the same.

Develop Healthy Eating Habits

- Children have the ability to regulate their caloric intake to meet their needs. Respect these internal cues of hunger and satisfaction.
- Do not force children to clean their plates or bribe them with dessert for finishing their meal.
- Never use food as a reward. Reward desired behavior with praise, extra attention, and privileges.
- Do not comfort your child with food.
- Do not impose stringent food rules, since this may lead to rebellious eating when the child is away from parental control.
- Don't say or imply that some foods are "good" while others are "bad." Instead, teach children that some foods are healthier than others. This will help them learn to balance eating for health with eating for pleasure.
- Involve children in shopping,

meal planning, and preparation. This is a great opportunity to teach them about nutrition—and they will be more likely to try new foods if they helped make them!

- Since children (and adults!) have a natural preference for sweet and high fat foods, it's reasonable to limit the amount of sugary and fatty foods that are readily available to encourage intake of more nutrient dense foods.
- Provide a variety of delicious healthy choices for snacks and mealtimes. Suggestions include fresh or dried fruits, vegetables with tasty low fat dips, pretzels, reduced fat cheese or peanut butter and crackers, yogurt, fruit smoothies, whole fruit ice pops, granola bars, turkey roll-ups, or snack mixes made of cereal, dried fruit, and nuts.
- A healthy breakfast is a great way to start the day and is important for achieving and maintaining a healthy weight.
- Encourage children to drink water and fat free or low fat milk instead of sugary sodas, fruit drinks, and sports drinks.
- Promote a high fiber diet by giving your child whole wheat breads and pastas, brown rice, and five servings of fruits and vegetables daily. They will prefer these types of foods if that is what they are used to.
- Perhaps most importantly, sit down and eat together as a family. Mealtimes should be a pleasant time to reconnect with one another.

Enjoy an Active Lifestyle

- Help your child build a lifetime exercise habit by making

consistent physical activity a high priority in your family.

- For children that have been relatively inactive, an exercise program should be initiated very gradually to avoid injury and discouragement.
- Encourage active play like biking, swimming, and playing ball.
- Participation in individual and team sports can be a great way to build coordination, athletic skills, and self-confidence.
- Reduce the amount of time your family spends in sedentary activities like TV and video games. Instead, plan fun family activities that provide everyone with exercise and enjoyment.

While management of weight problems in childhood can be difficult, the benefits can last a lifetime!

Michelle May, M.D. is a board-certified Family Physician in Phoenix and developed the Changing Weights Weight Management Program. She speaks widely on the topic of weight management without dieting and anticipates the publication of her book "Am I Hungry?" in 2004.

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/NCAH patients. Please contact CARES Foundation for more information.

Preimplantation Genetic Diagnosis: Another Alternative

By Anonymous

I am the mother of a two-year-old boy with the Classic Salt-Wasting form of Congenital Adrenal Hyperplasia (CAH). I am also the mother-to-be of our second child. Like many CAH families, we spent a long time contemplating whether or not to have another child knowing that there was a 25-percent chance that s/he too would have CAH. I would like to share with you the story of how we came to the decision to have second child and the medical procedure – pre-implantation genetic diagnosis (PGD) – we underwent so as to be as sure as we could possibly be that our baby would not be affected by CAH.

Before I go on, I must preface everything that I am about to say with the statement that the choice we made in having this child is not necessarily the right choice for other families. It was our choice and the right one for us; however, every CAH family has to make this decision for themselves according to their cultural and religious beliefs as well as what is most comfortable for them. Therefore, I offer our story merely as another alternative for every CAH family's consideration.

It had always been my husband's and my plan to have more than one child. However, as we began to discuss the possibility of having a second baby, we both realized that given the physical, emotional and financial cost of CAH, we were not comfortable with consciously bringing another child with CAH into the world without doing everything we possibly could do to avoid this for any of our other children, especially if we were to have a girl. In discussing this with our pediatric

endocrinologist, he suggested we contact Cornell University's New York Presbyterian Hospital to learn more about genetic testing and prenatal treatment options.

From Ann Carlson at Cornell, we learned that DNA testing for CAH is available and in 95-percent of cases they are able to identify mutation(s) that result in CAH. In other words, if we sent them some of our blood, they would look at our DNA – specifically Chromosome No. 6 – and hopefully find the mutation(s) that caused our son's CAH. Moreover, if Cornell were able to identify the mutation (s) then prenatal diagnosis would be possible for us. In any case, if we were considering another pregnancy, Ann suggested, our first step should be to meet with a genetic counselor. She gave us the names of a few such counselors in our area and our journey began.

Over the course of the next two years, we engaged in extensive pre-conception counseling to learn about our options for another child. Our genetic counselor presented us with the following:

- Not having another child
- Adoption
- Gamete donation: using the egg or sperm of someone other than my husband or myself
- Spinning of sperm: separating the boys from the girls and then choosing "male" sperm
- Prenatal treatment with Dexamethasone
- Pre-implantation genetic diagnosis

After a great deal of research and lengthy consideration, we came to find that for cultural/religious reasons adoption and gamete donation were not choices with which we were comfortable. Spinning of sperm yields no better chance of having a male child than conceiving naturally.

We knew a little bit about prenatal treatment with Dexamethasone from Ann, but through our genetic counseling sessions learned more. Prenatal treatment of female fetuses is possible via maternal intake of Dexamethasone from the 7th week in pregnancy to term. This can significantly reduce the virilization of females. In other words, if I carefully monitored my cycles and started taking Dexamethasone as soon as I knew I was pregnant, it would be possible to lessen or even eliminate the "masculinization" of a CAH baby girl. There was some possibility of risk to me of side-effects such as edema (swelling), excessive weight gain, irritability, nervousness, mood swings, hypertension, glucose intolerance (diabetes), and severe striae with permanent scarring. However, research at Cornell was

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Preimplantation Genetic Diagnosis

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showing this prenatal Dexamethasone treatment to be highly effective and generally safe.

Prenatal treatment with Dexmethasone clearly was an option for us. We might still have a CAH child, but at least we could do something to minimize the in-utero effects of CAH on a baby girl. However, upon learning more about Pre-implantation Genetic Diagnosis (PGD), we knew it was the way for us to go.

Our first real conversation about PGD was a telephone conference as set up by our genetic counselor with a molecular bio-chemist at Wayne State University in Detroit, Dr. Mark Hughes. Over the course of 45-minutes or so, he explained what is PGD, how it is achieved, and how it could help us to give birth to a non-CAH child.

PGD is a medical technique whereby embryos can be screened for specific genetic defects prior to transfer to the womb. It has been being done for over 10 years and has proven to be a most effective method of diagnosing embryos for known genetic mutations. To-date there have been over 2500 PGDs performed around the world resulting in over 1600 children born without the disease for which they were screened. The error rate for PGD is less than two-percent; therefore, PGD would reduce our chance of having a CAH child from 25-percent to less than two-percent.

A PGD cycle begins with in-vitro fertilization (IVF). This involves inducing ovulation through maternal intake of hormones, the harvesting of eggs, and the fertilization of those

eggs in a culture dish. On Day 3 after retrieval, when the embryo is eight-cells or so in size, a single cell is biopsied from each embryo. These cells then are sent to Detroit where Dr. Hughes tests the single cell from each embryo for the genetic defect in question. From biopsy to diagnosis would take approximately 48 hours. In the meantime, our embryos would be sitting in their petri-dishes at the fertility center growing.

PGD can be done for any number of diseases including CAH. The only pre-condition to undergoing PGD is that the specific DNA mutation(s) that cause the disease must be known. As Dr. Hughes explained it to us, the genetic code of each of our DNA is made up of 3.3 billion "genetic letters." Sometimes, these letters or even entire paragraphs or volumes of these letters get moved around, duplicated or deleted. When this happens a genetic mutation occurs. If the mutation carried by prospective parents is known, then Dr. Hughes could test the DNA of an embryo for the same mutation simply by comparing sequences of genetic letters.

The goal? By the end of Day 5 to have at least one and hopefully two or three high-quality embryos that were either unaffected by CAH or carriers of CAH for transfer. These then would be moved from the lab to my uterus. From there, based on the fact that we have no known fertility problems and I would be 35 years of age at the time of delivery, we would have a 45-55-percent chance of having one or more embryos implant themselves in my uterine lining thereby achieving pregnancy.

"Would biopsying a cell from a Day 3 embryo cause damage?" we asked. After all, this meant removing one of only eight cells from the embryo. Dr. Hughes asked us to think about how identical twins are formed. At some point in the very early stages of development, an embryo splits in half, each half losing far more than a single cell, yet two fully formed babies result. Similarly, when a single cell is taken in the PGD process, the remaining cells merely "pick up the slack." He added, however, that it was very important that we be working with an IVF center with laboratory personnel practiced and skilled in this biopsy procedure. As we soon learned, no such center exists in our state; however, Dr. Hughes was able to provide us with the names of several IVF centers in our part of the United States that did have the proper staff and experience to take us through a PGD cycle.

Our next question of course was, "How much does all this cost and will our insurance company cover it?" The cost of the pre-implantation genetic diagnosis as done in Dr. Hughes' lab is incredibly inexpensive given the level of technology employed and their 48-hour turn-around time. It is ~\$2800; the cost of their materials. (Note: This cost is subject to change and must be confirmed with the lab.) However, the cost of IVF itself is incredibly high and for many prohibitively so. Upon further research, we calculated that by the time one paid for pre-cycle testing, the various fertility drugs (hormones) required, egg retrieval, biopsy, embryo transfer, travel/lodging, and other miscellaneous expenses, a single IVF cycle would cost approximately \$20,000. PGD is

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not a procedure covered by most insurance companies; however, some will pay for it and others can be incited to do so. We were advised that if we were going to undergo a PGD cycle the insurance coverage approval process might be lengthy. If we could not wait for a final approval or denial to start our cycle, then we should plan on having to cover the full \$22,800 ourselves; the majority of which is due in full in advance.

In other words, if Cornell could identify our CAH-causing genetic mutation(s), then not only was prenatal diagnosis possible but also pre-pregnancy diagnosis. PGD could reduce our risk of having a CAH child from 25-percent to less than two-percent. For us, this meant that PGD afforded us the best chance possible of giving birth to a healthy child.

We immediately had our blood drawn and sent it to Cornell. They performed their DNA test and found me to be the carrier of a heterozygous Exon 3 (8 basic pair deletion) mutation and my husband to carry a heterozygous Intron 2 (A or C to G) mutation. In short, we are carriers of CAH mutations and they are clearly identifiable; therefore, the “biomedical” path to PGD was clear.

As everyone we came in contact with while undergoing this process told us repeatedly, every PGD cycle is different. In all cases, however, it is a complicated, physically and emotionally intense, and relatively expensive procedure.

First and foremost, we had to face the fact that IVF is a prerequisite to PGD as a single cell cannot be

biopsied from an embryo in-utero. Having no known fertility problems, undergoing the IVF portion of this process was a strange and often disquieting experience. It began with more genetic counseling. I explained that we had already undergone extensive genetic counseling, but the IVF center was insistent that we go through a session with their counselors. They also insisted that we do a comprehensive pre-cycle assessment including testing for sexually transmitted diseases, several ultrasounds and blood tests to determine my fertility status as well as complete analysis of my husband’s semen. Moreover, we had to send blood off to Dr. Hughes’ lab for him to create the probe that he would use to test our embryo samples. Finally, there were all sorts of forms to be read, filled out, signed and notarized.

At every step of the way, I worried that something else “wrong” with us might be found. I was now 35 as opposed to 31 like I was when my first son was conceived. In the interim had my eggs diminished in number significantly or become too old for conception or had my husband’s sperm been compromised in some way? Should we do the Tay-Sachs, Cystic Fibrosis, and other disorder testing just to be sure there is nothing else lurking in our genes? For some reason would Dr. Hughes be unable to create an effective probe? What do we want the IVF center to do with any embryos we do not transfer? If we freeze embryos – assuming we even have any to freeze – what do we want the IVF center to do with those embryos if we decide we do not want to keep paying for their

cryopreservation? Would my irregular periods somehow keep us from starting a cycle?

The moment of greatest apprehension came, however, when we were told we would have to undergo not only IVF but also Intracytoplasmic Sperm Injection (ICSI). The form we were asked to sign indicated, “ICSI is a micromanipulation technique developed to help couples with severe male factor infertility...[It] involves precise maneuvers under the microscope to inject a living sperm directly into an egg to effect fertilization.” In other words, my eggs would not simply be put in petri dishes with some of my husband’s sperm. Rather, a lab technician would examine my husband’s sperm and select individual ones, break the tails off them, and then inject one sperm into each egg.

“But we have no fertility problems, why would we need to do ICSI?” we asked. The purpose of this, we were told, was to avoid having more than one egg and one sperm’s DNA present in any sample sent to Dr. Hughes. In “normal” IVF conditions, it is possible for one sperm to penetrate and thereby fertilize the egg while others get stuck in the outer layers of the egg or onto the outside of a growing embryo. The presence of these “extra” sperm could interfere with Dr. Hughes’ analysis and therefore diagnosis.

The form continued, “[B]abies conceived through ICSI have a 6/1000 chance of having either an extra or missing sex (X or Y) chromosome. This is three-fold higher chance than that seen in the general population...Babies with sex

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chromosome abnormalities can have a variety of symptoms, including but not limited to cardiac and other problems which may require surgery, behavioral and learning difficulties or infertility.”

“Whoa!” I said. “No way. We have already done one in 15,000 [the statistical probability of having a CAH affected child]. The whole purpose of doing PGD is to reduce risk not increase it!” The genetic counselor stepped in. Explaining that the form was standard for all ICSI patients, she noted that recent data is revealing that the chromosomal problems generally show up in couples with “male factor” (low to zero sperm count/motility) infertility not couples like us with no known fertility problems. Bringing it all into perspective, she concluded, “By undergoing PGD and thereby reducing your risk of having a CAH affected child from 25 percent to less than two percent, your greatest risk of birth defects is due to your [my] age.” We signed all the documents and moved on to reviewing IVF drug dosages, cycle schedules, and injection instructions.

Which IVF drugs one takes and the timing of their administration depends purely on each individual couple’s situation; therefore, I cannot say what your experience might be should you do IVF. In our case it was daily prenatal vitamins and baby aspirin and nearly three weeks of follicle stimulating hormones overlapped one week with an ovulation suppression drug for me. Then I took a single egg release trigger dose the night before our harvesting appointment. Throughout this hormonal therapy I was closely

monitored as to my follicular development and reaction to the drugs via ultrasounds and blood tests. Once the embryos were transferred I also would take a uterine lining growth hormone. As for my husband, he took a single antibiotic dose the night before his sperm was needed and otherwise spent the cycle dealing with a semi-crazed woman on way too many hormones. While I did not experience sudden weight gain, I was exceedingly hungry while on these drugs, often felt heavy in the lower abdomen, and ran to the bathroom to empty my bladder constantly. Moreover, I was often queasy and not particularly pleasant to be around. It was all worth it though when 16 days after starting our cycle 15 eggs were retrieved.

The time period from retrieval to transfer was nerve-wracking. We had been forewarned by others who had undergone IVF that it was a roller coaster ride, and we had prepared ourselves for this experience both physically and emotionally well in advance. I had quit my job a month prior to the start of our cycle to remove all work related stress from my system. We had chosen an IVF center with extensive IVF and PGD experience in a location where we had a numerous family members to help us so as to reduce emotional and physical strain to a minimum. Moreover, we had spent hundreds of hours discussing our thoughts, feelings and beliefs about conception, life, birth; so as to be as clear and unified as possible when making decisions in the midst of the cycle. However, we still were not prepared for the “count down” as I called it – the race against nature and time to have unaffected

viable embryos to transfer – and the inexplicable complexity of PGD.

We started with 15 eggs of which 12 achieved fertilization. All twelve were biopsied on Day 3 but really only ten of them were looking viable by Day 5 when we went in to the IVF center to hear the results of Dr. Hughes’ analysis and undergo embryonic transfer. We had spent the entire night before steeling ourselves for the momentous decisions we would have to make the next day; decisions that would have to be made within a matter of an hour or so. Would any of the “good looking” embryos be unaffected? If so, how many? How many would we ask to have transferred? Would we have any left over to freeze? Or would it turn out – as statistics predicted – half the fertilized eggs would be unviable...of those six 25 percent would be affected by CAH and another 18 percent would have ambiguous PGD results eliminating another three...and we would lose another few by the time we got to transfer day leaving us with nothing?

We were completely unprepared, however, for what the doctors said when we walked in the door on the morning of Day 5, “We have not heard anything from Dr. Hughes’ lab yet.” We waited several hours and still no results. Meanwhile our embryos were dividing and growing and moving from the embryonic to blastocyst stage. Dr. Hughes called. He was having trouble with his probes. He did not want to give us a false analysis; so, he asked us to allow him to rerun the tests from scratch over night. We were perfectly clear that to transfer embryos without an analysis was the same as throwing the genetic dice and conceiving naturally; so, we

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would not transfer without a diagnosis. But we had come this far and had viable embryos and now we might have to throw the entire cycle away because the PGD was not successful? We had no choice but to wait.

At 8:00 the next morning, our doctor at the IVF center called. She had PGD results for us: five affected, five unaffected and two carriers. Of the five unaffected, three looked good; the other two they considered poor choices for transfer. The two carriers were also candidates for transfer. In all cases, however, overnight the blastocysts had hatched – they had broken out of their protective zona pellucida shell - and rapidly were becoming too advanced in their development for transfer. As we had already agreed that “selective reduction,” the removal of one or more fetuses from the womb to reduce the number of babies being carried, was out of the question for us and that we were perfectly happy with having twins, we decided to transfer the two good unaffected embryos and freeze the other unaffected and two carriers. From here on it was all up to Mother Nature and given our past history of becoming pregnant within a month of trying to conceive, we both were sure we were well on our way to a second and third child despite the statistical fact that we had only a 45 to 55-percent chance of achieving pregnancy.

Two weeks later, I had my blood draw and the next day we received a telephone call from the IVF center. My HCG levels were rising. My body was showing signs of pregnancy! I had my blood drawn again and continuing rising HCG

levels confirmed what the IVF center called a “chemical pregnancy.” We visited our obstetrician to get “visual” verification of pregnancy two weeks later. On the ultrasound we saw a heartbeat – “Yes!” – but only one. We asked the obstetrician to look again. No there’s only one. “It could not be hiding behind the one we see,” I asked. No. As I had experienced no bleeding or cramping or any other signs of having lost one of the two, we were both surprisingly dismayed at this news. We were definitely pregnant but one of the two embryos had dissolved sometime between transfer and this first ultrasound. The uncertainty of it all suddenly hit home.

We abruptly became adverse to the idea of doing chorionic villi sampling (CVS) or amniocentesis to reconfirm Dr. Hughes’ diagnosis as well as anxiously awaited each and every ultrasound appointment so as to be sure the baby was indeed still there. In the end we did do CVS at 12 weeks – CVS can be done between 10 and 12 weeks allowing earlier detection of any problems with a fetus than amniocentesis which cannot be done before 15 weeks – as we had signed paperwork indicating that we would, but it was with great trepidation that we might be one of the one in 200 who suffer a spontaneous miscarriage therefrom.

The CVS results for Down’s Syndrome and other chromosomal abnormalities was presented to us shortly after the procedure. The baby’s chromosomes looked completely normal. It would take several weeks though for Cornell to complete their CAH analysis; so, we asked not to be told the sex of

the baby until after Cornell’s report was in. In fact it was over a month before we received a call from the genetic counselor, “Great news! The baby is an unaffected carrier of the maternal mutation.”

My heart stopped. We had transferred two unaffected embryos and froze the two that were carriers. Was Dr. Hughes or Cornell right? Or worse, were they both wrong? I had not taken dexamethasone as there was such a low probability of a misdiagnosis. What if the baby actually is a CAH-affected girl? We are too late to minimize or prevent virilization. I quickly asked to have the results along with the CVS analysis faxed to our home. Gender: Male. Even if they were both wrong and the baby was CAH-affected, we were definitely going through with the pregnancy. A pregnancy that was to end much sooner than we expected.

As it turns out, my placenta was low lying and anterior (in the front of the womb) and started to come apart in the fourth month of pregnancy; causing pre-term labor and the delivery of our baby boy at 31 weeks of gestation. These conflicting diagnoses became the center of attention and anxiety immediately thereafter as 17-hydroxyprogesterone levels are elevated in premature infants. Blood drawn at 60-hours after birth showed his 17OH-P to be 1841. Maybe too high; maybe not. We reran the test at 10 days of age: 382. It was going down and his electrolytes were looking really good, but we would have to do the 17OH-P again at one month of age.

We still have not seen a 17OH-P level that is within the “normal

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Newborn Screening Update

Our grassroots advocacy in California is moving forward quickly! We printed postcards on Hot Pink paper addressed to Governor Schwarzenegger asking him to expand newborn screening to include CAH and "SAVE OUR BABIES". These were then signed by California residents and mailed to the Governor. Our members have distributed over 8,000 postcards so far! We are working in coalition with other groups, the Urea Cycle Disorders Foundation, PKU Southern California and the PKU Network. These groups liked our postcard idea so much that they have printed their own postcards asking the Governor to expand newborn screening! So, Governor Schwarzenegger is being inundated with neon colored postcards asking him to Save Our Babies! We still have a lot of postcards left, so if you can help

Preimplantation Genetic Diagnosis

(Continued from page 9)

range" for an infant, but now at two months of age, our little boy is nearly double his birth weight, his 17OH-P level has not gone up, his electrolytes are perfect, and he is a happy, healthy child. As for his carrier status? Dr. Hughes might have been mistaken in his diagnosis due to a phenomenon I still do not understand called allele drop-out. Cornell also could be wrong. Either way, we have decided not to ask until he reaches reproductive age. Until then, we plan on enjoying our miraculous bundle of joy.

Anonymous

distribute them, please call or email us and we will mail them out to you.



While on vacation in California last month, I met with an aide to Governor Schwarzenegger about newborn screening. We met for over 1-1/2 hours and the aide told me that expanding screening was a "no brainer" and in line with the Governor's current priorities. I felt quite confident that the Governor will address the issue, but funding will still be a problem. We need to keep up our advocacy efforts! Keep the cards and letters to the Governor coming! If you want to write personalized letters about the benefits newborn screening and your family's experience without the screening, please write to:

The Honorable Arnold Schwarzenegger
Governor, State of California
State Capitol Building
Sacramento, CA 95814

Vermont added CAH to its screening panel along with several other disorders in November of 2003! Way to go Vermont! The Newborn Screening Program of the Oklahoma State Department of Health is moving forward with an expanded screening panel. Funds have been secured for the lab and follow-up program to hire staff and buy equipment. They plan to

implement CAH newborn screening by July 1, 2004. Many thanks to all of the Vermont and Oklahoma families who called and wrote in support of expanded NBS.

As you can see, our efforts are making a difference! We are saving babies, one state at a time!

Legislation/Appropriations Update: Newborn Screening Saves Lives Act

Senator Chris Dodds (CT) introduced the Newborn Screening Saves Lives Act on May 15, 2003, bill number S. 1068. This bill amends the Public Health Service Act to establish grant programs to provide for education and outreach on newborn screening and coordinated follow-up care once newborn screening has been conducted, and for other purposes. The co-sponsor is Senator Michael DeWine (OH). It was referred to the Senate Committee on Health, Education, Labor and Pensions. It has not moved. Please call your local US Senators and ask them to co-sponsor this important legislation. If you do not know who your US Senator is, you can look them up at www.senate.gov.

Reconstructive Surgery Act

The Reconstructive Surgery Act, sponsored by Congressman Mike Ross (AR), bill number HR. 1499 is pending in the Committee on Energy and Commerce, Subcommittee on Health, and in the Committee on Education and the Workforce Subcommittee on Employer-Employee Relations. It has 18 co-sponsors. This bill would require insurance companies to cover reconstructive surgeries performed to correct or repair abnormal structures

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Newborn Screening Update

(Continued from page 10)

of the body caused by congenital defects, or to improve functions or give patients a normal appearance to the extent possible in the judgment of the physician performing the surgery. Please call your Congressional Representative and urge him/her to sign on a co-sponsor of this legislation. You can find your Congressional representative at www.house.gov.

Genetic Information Nondiscrimination Act

Genetic Information

Nondiscrimination Act of 2003, Senate bill number S. 1058 passed in the Senate on October 14, 2003 and was sent to the House of Representatives. This bill would prohibit discrimination on the basis of genetic information with respect to health insurance and employment. A comparable bill, sponsored by Congresswoman Louise Slaughter (NY) was introduced in the House in May 2003 and has 236 co-sponsors. The sponsors of the two pieces of legislation are trying to make changes to help this legislation move through the House quickly. Unfortunately, the Chambers of Commerce came out against this bill because they feel it would hurt employers. Please call your Congressional representatives, (look up at www.house.gov) and urge them to support this important legislation!

Appropriations

The FY 2004 Omnibus Appropriations bill passed in the Senate on January 22, 2004. In this bill, the NIH budget was given a 2.6% increase over last year for a total appropriation of \$27.8 billion. The President has proposed a 2.7% increase for FY 2005. This poor appropriation for NIH means, essentially, that few new grants for research will be approved this year or next. We need to let President Bush know that this is

CARES Foundation to Collaborate with Cornell on Clinical Research

On February 27, 2003, Office of Rare Diseases in response to the Rare Diseases Act of 2002, P.L. 107-280, released a Request for Applications (RFA) for a Rare Diseases Clinical Research Network together with the National Center for Research Resources (NCRR)/General Clinical Research Centers (GCRC) Program and in collaboration with other NIH Institutes. On September 28, 2003, ORD, NCRR, and the National Institute of Child Health and Human Development (NICHD), National Institute of Neurological Disorders and Stroke (NINDS), National Institute of Arthritis and Musculoskeletal and Skin Diseases (NIAMS), and National Institute of Diabetes and Digestive and Kidney Diseases (NIDDK), all components of the NIH, funded seven Rare Diseases Clinical Research Centers and one Data and Technology Coordinating Center at a cost of \$51 million over a five-year period. Cornell was chosen as one of the clinical research centers to study genetic steroid disorders, including CAH.

The purpose of the network is to facilitate clinical research in rare diseases through:

unacceptable. In order to gain a better understanding of CAH, we need more research. This is devastating to our community. This bill also had a \$2 million appropriation for grants to states for newborn screening improvement and expansion.

- Collaborative clinical research in rare diseases, including longitudinal studies of individuals with rare diseases, clinical studies, phase one and two research studies, and/or pilot and demonstration projects to respond to the very differing level of research and to facilitate research that is sorely needed;
 - Training of clinical investigators in rare diseases research;
 - Clinical data management that incorporates novel approaches and technologies for data management, data mining, and data sharing across rare diseases, data types, and platforms; and
 - Access to information for basic and clinical researchers, academic and practicing physicians, patients, and the lay public across the U.S. and abroad. This cooperative program should facilitate many advances including the identification of biomarkers for disease risk, disease severity/activity, and clinical outcome and encourage development of new approaches to prevention, diagnosis, and treatment of many rare diseases beyond those being studied. The easy and free availability of data from the center should also spawn many new research ideas and subsequent applications to NIH Institutes and Centers.
- Project Title:** The Natural History of Rare Genetic Steroid Disorders
Principal Investigator/Project Director & Contact Information:
 New, Maria I, M.D.

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CARES to Collaborate with Cornell on Clinical Research

(Continued from page 11)

Weill Medical College of Cornell University
1300 York Avenue, Box 103
New York, NY 10021
212-746-3450

Description from the Grant Application:

A consortium of investigators, institutions, and patient support groups will constitute a Rare Disease Clinical Research Network focused on a diverse group of disorders characterized by defects in steroidogenesis. We will study the longitudinal history of these rare disorders and determine the outcome of treatment on height, fertility and gender. Long-standing informal collaboration between investigators at Weill Medical College, Rockefeller University, Columbia University, the University of Texas Southwestern Medical Center, the University of Quebec, Hospital Debrosses (Lyons), and the Hospital das Clinicas da FMUSP (Sao Paulo) will facilitate the creation of a productive cooperative research network that draws on the extensive experience of each investigator. Clinical Research Centers at Weill, Rockefeller, and the University of Texas Southwestern Medical Center will participate. Each investigator in the consortium has followed a large group of patients with a specific genetic defect affecting steroid synthesis over many years, encompassing the natural history of these diseases from prenatal life to death. Creation of a storage and management database will constitute a scaffold for ongoing research, enabling the preservation and use of this large body of clinical data assembled by experts in each disorder. Moreover, design of

templates for a standardized clinical description of these disorders will permit prospective studies which can offer open enrollment to affected individuals or individuals at risk. Our research group includes the investigators who have identified the molecular genetic defect for each disorder, where known, and who maintain laboratories dedicated to the identification of new mutations. The combination of clinical and molecular genetic information will raise the standard of medical care and may permit development of novel treatments based on detailed knowledge of the natural history and molecular genetic basis of these disorders. Important elements of our plan are to

1. Establish the clinical research network which pools data from our sites in cooperation with the DTCC and analyzes this data,
2. Educate young investigators in the management and clinical research of steroid disorders, and
3. Strengthen our connections with patient support groups to enable individuals affected or at risk to have new kinds of input and access to optimal medical care.

Performance Sites:

Weill Medical College of Cornell University at the New York Presbyterian Hospital, New York, NY
Rockefeller University, New York, NY
Laval University, Québec, Canada
University of Texas Southwestern Medical Center at Dallas, TX
Hospital Debrosses Lyon, France
Hospital das Clinicas da FMUSP, São Paulo, Brazil
Columbia University College of Physicians & Surgeons, New York

State Psychiatric Institute, New York, NY

Patient Support Organization: Congenital Adrenal hyperplasia Research, Education, & Support, CARES Foundation, Inc.

The Rare Diseases Clinical Research Network is funded by the Office of Rare Diseases, National Center for Research Resources, National Institute of Child Health and Human Development, National Institute of Neurological Disorders and Stroke, National Institute of Arthritis and Musculoskeletal and Skin Diseases, and National Institute of Diabetes and Digestive and Kidney Diseases, all components of NIH, an agency of the Department of Health and Human Services.

The creation of the network responds to the Rare Disease Act of 2002, which directed NIH to support "regional centers of excellence for clinical research into, training in, and demonstration of diagnostic, prevention, control, and treatment methods for rare diseases." The term "rare (or orphan) disease," as defined in the Orphan Drug Act, is a condition affecting fewer than 200,000 in the United States or a disease with a greater prevalence but for which no reasonable expectation exists that the costs of developing or distributing a drug can be recovered from the sale of the drug in the United States.

Congratulations!!

Dr. New was inducted into the Hall of Honor of the NICHD (National Institute of Child Health and Human Development) on Sept. 22, 2003 on the occasion of their 40th Anniversary.

Laura's Story

I was born with Congenital Adrenal Hyperplasia (CAH) in 1953, as the second child of my parents. The first child, a boy, died after living only a few days, from undiagnosed causes (probably also with CAH). When my mother became pregnant with me, she hoped strongly to have another boy, rejecting the possibility of having a girl. When I was born she had a terrible shock. Being educated within a religion with a God as a chastiser she looked at me as a severe punishment for committing the sin of not accepting His will. At that time my parents lived in a foreign country and the only member of family they had here was my father's aunt. When she learned about me, she accused my mother of being responsible because my mother had been a tomboy when she was an adolescent. The culpability of my mother was aggravated when my father told his aunt, that when making love, my mother liked to stay above him, which was - they said - a male behaviour. Crushed by her "faults", with her sexual life destroyed, and completely alone, my mother spent her days crying and asking God to forgive her and to take me away. Very early I realized that my mother wanted me to be dead, and I began wanting to die.

We lived on a farm. Whenever I felt that my mother was feeling hopeless I went to the forest since I heard that snakes killed people. I thought how nice it would be for my mother if one killed me. In the forest I cried for hours, ultimately dropping to the ground from sheer exhaustion. Then I began sleep. Usually I woke up when it became dark and then, frightened, I returned home, trying to reach my bed without being seen. People used

to play with me asking me where my mother was, as I was young, I replied, "She has gone". No one could imagine how I was suffering!

At two years old I became very sick, and my mother took me to a doctor. He told her that I had a liver crisis - it was my first bad adrenal crisis - and a new phase began in my torment. Each time I had a new crisis - I usually had two per year - my mother took me to this doctor, and the routine was always the same. He would say to my mother "What do you want? I already told you to put her on a diet", and after "pull down her panties so that I could examine her". When all I wanted was to ease my crisis, he was only interested in the examination of my genitals. At that time I understood that something was wrong with me and I tried to ask my mother about it. However she always showed a very severe face and replied "Know that you are not like the other children". Soon I learned not to ask anything about me, but I began thinking about my differences. As I thought more and more about them, they grew, like a big monster which consumed me completely.

Between my four and fifth years of age the same doctor told my mother to take me to one of the most important hospitals in the country to be examined. I stayed there alone (at that time parents were not allowed to stay with their children at the hospitals) terribly frightened thinking that my mother abandoned me here to be killed (many years later I had still had nightmares that my mother wanted me to be killed). I don't know how many times I stayed at that hospi-

tal - maybe several weeks or some months - or what was done to me. Some images are present in my mind but, in general, in an indistinct way, except for those who caused me more suffering. I remember that some nurses were nurturing to me, however the doctors who usually arrived in groups to examine me, frightened me a lot. I heard about being subjected to a laparotomy. The doctors told my mother to take me away and to wait to see what will happen to me when I grew up.

By the time I was five years old, I had the body of a ten year old. Being bigger than my peers I was severely reprimanded when I played like them. "What will people think about you, with such a big body?" said my mother. Then I become more quiet than ever, living much like an autistic. Most of the day I played alone in the field with wild flowers which I pretended to be people. Sometimes I took refuge in the forest to cry.

At that time my mother had to work hard. My father lost his job and she finally got the son she had wished for - one who she wanted to give a nice future. My frequent "liver crisis" exasperated her. As I was on a diet, she accused me of eating unripe fruits at the farm. "You are sick, because you want to be" she said "and I have no time to take care of you, as I need to work". She began mistreating me severely and she told my father that "his daughter" was an idler as "she has a body big enough to help me in domestic tasks, but she is never at home". When more exasperated she beat me violently with a switch until her revolt had passed. I remember crying for hours without understanding why I was so severely

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Laura's Story*(Continued from page 13)*

punished.

At puberty I began to seriously masculinize: facial and body hair growing, voice cracking, no breasts and no periods. Then, my years in school were a minefield of secrecy and emotions. I felt immense shame and the lessons of gymnastic and singing were tremendously hard, in a such a way that, when recently a colleague told me to go to a gymnasium, I began crying in an uncontrolled way. I remember vividly the day a colleague, who had a sister older than us, approached me and dying with laughter said "my sister told me that there are persons who are partially males and partially females; how funny is it: you with male legs, and female arms". My wish was to disappear into the soil.

By the time I was 17 years old I begin to "play" flirt with a neighbour. It was really to play, as at that time I felt that I was so strange a creature that I couldn't think of having a boyfriend. However it was so nice to have an illusion! When my mother discovered this she became furious with me. Her words still hurt me. "What were you thinking? You know that you are different from the other persons and that you can never get married. Well, I will take you to the hospital where you stayed as a child, to see what they can do for you".

At that time it was not uncommon that people made a game of me, commenting "Look, is this a male or a female?" and than dying with laughter. At the buses, trains and elsewhere, they avoided sitting near me and I begin avoiding staying near those that I liked, so I would not cause shame to them.

At the hospital I was sent to a young doctor who, without having a word to me – he was talking with his colleagues – told a nurse to give me a room. I stayed there for several months running test after test. No one ever explained what was being done to me, nor why. However I must also confess that I never asked about it, because I was so fearful about what was wrong with me.

At that time I had entered the University. When classes were beginning I told the doctor that I had to go home. He told me that what I needed was "to be treated". This surprised me because I had never realized that I had a disease, except for the "liver crisis". I had always been told that there was nothing to do, except diet. However I thought, why should I leave the hospital when life outside was so difficult and if I was living in one of the best periods of my life? Several of my fellow patients were very sick, and I could actively give them comfort. So, the first time in my life, I felt needed and useful.

However difficulties followed me. Before I left the hospital, I had a laparotomy. The day after it was done, when I was on bed in pain, a nurse took my bed into the "treatment room". After removing the sheet which covered me, she opened my legs saying that doctors wanted to see me. After this, several of her colleagues arrived and looked into my genitals, where hairs were removed, and had a good laugh, while I felt crushed by the humiliation.

Some days later, another nurse looked into my room and dying with laughter said "I heard about you. You will not be allowed to live here. If I was in your place I would

go to the outskirts of London where there are lots of strange people and you will escape one's notice". After that, I became more isolated than ever.

At the University I became a good student who studied hard. It was the only means to alleviate my permanent anguish. It was also the same time that I found a job. However, at times this anguish became so strong that I slipped into a depression. As I was put on estrogen and corticosteroids my look became less "strange" and in general I escaped people's notice. But even so, sometimes I perceived that some colleagues were commenting about me, and one day when I was recovering from a strong depression, one of my colleagues said to me "people are commenting that you are changing your sex; let me see your karyotype". Again humiliated, the only thing that I wanted at that time was to be left in peace.

Several years have passed. About two years ago I began having serious health problems and I was told that they were due to the dexamethasone I was taking. I decided to ask why I had to take it. I asked the doctor who had prescribed it to me, who replied "you have Congenital Adrenal Hyperplasia". The meaning of these words was completely unknown to me and I decided to type them in the internet. The results obtained caused to me an enormous astonishment. What I realized for the first time was that there were other people like me! For a long time I couldn't do anything as tears didn't stop. After, I begin thinking that it couldn't be true... probably those were only stories created by some writer, and those people didn't exist. However, after the warm messages that I've received from them, I realized that they are real, and how grateful I felt to them. I have also concluded that I'm simply a person equal to everyone else living in this world, only with a genetic muta-

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Employers can help Parents of Special Kids

Up to 20% of workers care for children with physical or emotional problems

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Each of Bart Reid's three children had unlikely childhood emergencies, ranging from a kidney defect to the need for openheart surgery. And three times, his employer, Deloitte, gave him all the time off he needed.

"They said, 'Family first. Come back when you're ready,'" recalls Reid, now the human resources director for the firm's three south Florida offices.

He did go back to work after each crisis passed, and he has stayed—so far—for 20 years. "It's given me such a sense of loyalty to the firm," he says. "I don't think for my situation there could be a better place to be all these years."

Now Reid bends over backward to accommodate other Deloitte parents whose children have special healthcare needs by extending their family leave or fashioning part-time jobs. Reid's experience helped him recognize something most employers do not: Up to 20 percent of working parents care for a child with a physical, mental or emotional problem that can wreak havoc on an employee's routine.

"One of the things we discovered is that many companies did not even know about employees who might have a child with special needs," says Christina Fluet, director of a federally funded study sponsored by MassGeneral Hospital for Children. Many people consider only children who use wheelchairs to be disabled, she says. But parents of kids with asthma, congenital heart disease,

obesity or attention-deficit disorder routinely face extra doctor visits, meetings with teachers, physical therapy assessments, high medical bills and problems at day-care. Yet they're reluctant to ask their employers for help, says Marianne Stook, vice president of marketing services for resource and referral service LifeCare. "Employees are somewhat embarrassed about admitting it," she says. Those parents depend on their employer-sponsored healthcare plans, says Linda Roundtree, president of Roundtree Consulting in Renton, Wash., whose son has Down's syndrome, and might fear they will lose their jobs if they ask for help.

Flexibility tops list of needs

Yet employers can help, she says, by granting flexible schedules and allowing parents to work from home—or, in Roundtree's case—from her son's hospital room, where she spent many nights. "That was an amazing benefit to me," she says. Fluet says she expected parents of special-needs kids to tell her about problems with healthcare coverage during interviews. Instead, they talked about their work/life problems.

The good news, she says, is that many companies already allow the flexibility these parents need. "Their needs are really not different from employees who are caring for an elder parent or other relatives with a chronic disease or

disability."

Still, they need help with the paperwork and assessments that are necessary before their children can enroll in community and school programs that cater to their situations.

A company's resource and referral program, notes Stook, can educate parents about what they need to do and find caregivers who are qualified to look after a child with a disability. "We save them all the legwork," says Stook.

The Personal Toll

The employer also can refer an overstressed parent to the employee assistance program for help in finding a support group, counselor or even respite care. "The first feeling these parents express is extreme frustration and confusion," says Stook. "They just get so much relief from talking to an expert who can calm their fears." At Blue Cross Blue Shield of Massachusetts, Employee Relations Director Terri Ireton doles out comfort. She works out flexible schedules with parents of special-needs children on a case-by-case basis, and says she's sure most of them would have to quit their jobs if she didn't. And her firm, she says, gets "loyal, dedicated employees" in return. "We have not gotten a tremendous number of requests, but the good will that those people feel and express goes miles for us."

Fluet isn't surprised. "The discretionary effort that they exert far

(Continued on page 16)



New CAH Study from Marc Breedlove, Ph.D., Michigan State University

We are looking for families that have one daughter who has CAH and one daughter who does not have CAH. The survey would be completely anonymous and no travel would be necessary, so any such families could participate. Women with CAH, or the parents of girls with CAH, can learn more about how to participate in the study by visiting : <http://www.marcbreedlove.com>, and clicking on links to "CAH Study".

Employers Can Help Parents of Special Needs Kids

(Continued from page 15)

exceeds employees in a different situation because these employees are so thankful, and they're willing to put out the extra effort for their employer," she says.

Contact: Christina Fluet at cfluet@partners.org

Laura's Story

(Continued from page 14)

tion! This crushed me. So many years of shame, humiliation and a lot of other strong emotions, just for a mutation!

Presently what makes me happy is to see that people with Congenital Adrenal Hyperplasia and their parents are receiving more and more support, thanks to the efforts being developed in this field in some regions; which I am confident will spread to other ones. My hope is that my story would become more and more a story from the past.

Laura T.

Fundrai\$ing Corner

The News-Sun

Friday, September 19, 2003

KENDALLVILLE - In gratitude to an organization helping them learn how to live with their baby son's unusual illness, Brad and Tina Smith of Albion will hold a bake sale this weekend to raise funds for the Congenital Adrenal Hyperplasia Research, Education & Support Foundation, also known as the CARES Foundation.

The bake sale will be at the Kendallville Wal-Mart, located on Fairview Boulevard, from 10 a.m. to 4 p.m. Saturday and Sunday.

This appeared in Indiana's *The News-Sun* newspaper when Brad and Tina Smith held their bake sale this past September. The Smiths and their neighbors must be excellent bakers, because they raised \$2,000 for CARES Foundation and brought a heightened awareness about CAH to their community. In fact a few new people learned about CARES and signed up as members as a result of this event.

As if that weren't enough, the Smith's went on to raise another \$166 through selling candles at their local Wal-Mart. Our gratitude and deepest appreciation go out to them on their great success, support and enthusiasm.

New Fundraising Guide

Interested in having fun and helping CARES? Here is your chance. CARES has a **NEW Fundraising "How to" Guide** that can help you, help us. The Guide has over 20 ideas to entice your imagination, and "how to" explanations to guide you through the process. Not only does it explain how to run an event but it also has resources to assist you, template letters for the media and so much more! It is simple, comprehensive and has a convenient check list to help you organize and plan your event. If you are interested in holding a fundraising event and raising awareness in your community about CAH, please contact CARES to receive a copy of the Fundraising Guide. You can email: info@caresfoundation.org or call toll

Attention New Jersey CARES Members

We are looking for a volunteer to help us in the office part-time. Hours/days are flexible. If you or perhaps a retired family member/friend is looking for a way to make a difference, please consider helping us out. Our office is now located in Short Hills, but will be moving shortly to a nearby town.



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 name and email address. All information is strictly confidential. If you haven't already done so, **please register on our database at:** <http://www.caresfoundation.org/form.html>.

Parent Tip!!

(Solutions for Common Problems)

Please remember to keep the Solu-Cortef with you and or your child at ALL times. My son broke his arm, I was in such a panic to get him to the emergency room that I brought the Solu-Cortef with me to the ER, instead of giving the stress dose at the time of the injury. Within 20-30 minutes, the signs of crisis were appearing and the emergency room physicians thought he had diabetes and needed insulin. Once the injection was administered, he was feeling better within 30 minutes. We now keep the Solu-Cortef in the car, backpack, athletic bags, and my purse.



Monica Heinze
Parker, Colorado

A NEW CAH BOOKLET FOR FAMILIES

A new booklet on CAH has been published by British Columbia's Children's Hospital, in Canada. The 32 page booklet includes medical details such as the Adrenal Hormone Pathway, parental management tips such as how to give salt to a baby, and a detailed emphasis on illness management. There is also a sensitive section on special issues for girls that are very difficult for families.

The booklet has been written by Sheila Kelton, RN BScN, Nurse Clinician in Pediatric Endocrinology. She has also written a chapter on CAH for the Pediatric Endocrinology Nursing Society Manual.

For pricing and ordering information contact: Family Resource Library ■ BC's Children's Hospital ■ 604 875-2345 Local 7644 ■ email: famreslib@cw.bc.ca

New Family History Tool

Those of us affected by genetic conditions are often interested in family history. The Genetic Alliance, in collaboration with the National Society of Genetic Counselors and the American Society of Human Genetics, has developed a new family history tool. We believe that a tool like this one can help all of us teach our members (and families and friends) that knowing one's family history, one can learn what health problems one may be at increased risk for in the future and how to reduce one's risks.

Mississippi CAH Support Group News

The first Mississippi CAH picnic was held on Saturday, October 18th, in Madison, MS, at Liberty Park. We had a beautiful fall day for our first (hopefully annual) picnic. Dr. George Moll, pediatric endocrinologist at the University Medical Center, sent our invitations out to all his CAH families. Seven families out of thirty attended. Dr. Moll joined us for a brief update on CAH. Krogers graciously donated all of our food. We hope to meet other MS families next fall when we meet as a group again!

Congratulations!!!

Members, Dr. Diane Snyder and her husband, Dr. Al Steren were included in **The Washingtonian** listing of the D.C. area's best physicians. Congratulations to both of them on being chosen. Diane is also a member of the CARES Foundation Board of Trustees.

Disclaimer:

Any communication from CARES Foundation, Inc. is intended for informational and educational purposes only and in no way should be taken to be the provision or practice of medical, nursing or professional health-care advice or services. The information should not be considered complete or exhaustive and should not be used in place of the visit, call, consultation or advice of your physician or other health-care provider. You should not use the information in this or any CARES Foundation, Inc. communication to diagnose or treat CAH or any other disorder without first consulting with your physician or healthcare provider. The articles presented in this newsletter are for informational purposes only and do not necessarily reflect the views of CARES Foundation, Inc.

CARES Foundation Contributors – 2003

Thank you to all of our wonderful contributors – your financial support means so much as we work together to make a brighter tomorrow for the greatest gift of all – our loved ones.

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Contact Michelle May
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michlmay@aol.com

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Contact Lynn Torony
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Ltorony@earthlink.com

**NE/CT Family Gathering
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317-823-1317
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Susan Aycok
601-833-8373
SAAycok822@aol.com

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mwlastevens@prodigy.net

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