



The Basics of Genetic Testing for CAH and How it May Benefit You and Your Family

By Wendy A. Conlon, M.S. and Raymond G. Fenwick, Ph.D

Although genetic testing is available for congenital adrenal hyperplasia (CAH) by having a blood test, it is not as simple or routine as it may first sound. Genetic testing for CAH is complicated by the location of the gene and by the different types of mutations or changes that can occur in the gene.

Approximately 90 to 95% of cases of CAH are due to 21-hydroxylase (21-OH) deficiency. The gene that makes 21-OH is called CYP21A2. The 21-OH deficiency form of CAH is inherited in an autosomal recessive manner. This means that parents of a child with CAH each have one working copy of the gene responsible for CAH and one non-working copy, and are referred to as carriers.

About 1 in 60 people in the general population are carriers. When two carriers achieve a pregnancy, each pregnancy has a 1 in 4, or 25% chance to be affected with CAH because the developing baby did not inherit a working copy of the gene from either parent.

The CYP21A2 gene responsible for 21-hydroxylase deficiency is located on chromosome 6. Chromosomes are the packages that contain our genes, and genes are the instructions that direct how our bodies grow, develop, and function. Each of our chromosomes come in pairs, so we each have one chromosome 6 inherited from our mother, and another chromosome 6 inherited from our father.

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On behalf of CARES Foundation, we invite your support of our first annual

EVERY1 CARES LUNCHEON

SKIRBALL CULTURAL CENTER, LOS ANGELES, MARCH 10, 2005

HONORING SENATOR DEDE ALPERT

In appreciation of her support of legislation to expand newborn screening, ensuring that all newborns in California will now be tested for CAH and many other devastating diseases.

LUNCHEON, BOUTIQUE, SILENT AUCTION, & TRIBUTE JOURNAL

All proceeds will go directly to serve the needs of those affected by CAH and their families, and to fund research for a cure and better treatments.

Please join us and bring your friends. You may also place a special ad in our CARES Tribute Journal recognizing Senator Alpert or honoring a family member or friend, whether or not they are affected by CAH. Of course, a direct donation or grant is also most welcome.

For further information, please call the CARES event office at (818) 994-4661.

CARES Foundation thanks Jami Abell Patterson and her committee members for organizing this event.

A Message from the Executive Director:



Dear Friends,
Happy 2005! I send best wishes to all of our members for a happy and healthy New Year. The new year has brought some changes to CARES, including the departure of Laurie Hitzig and the addition of Renata Blumberg and Debbie Jackman. Laurie is missed by us all, and we thank her for all of her hard work and dedication to CARES. Renata is our new administrative/programs associate, and Debbie is assisting Meryl with the bookkeeping. We are delighted to have them both working with us.

Inc., and Quest Diagnostics, Inc. for their support, along with Starbucks for once again keeping us fully caffeinated with their delicious brew. Starbucks donated the coffee and Panera Bread donated the cookies for the coffee break. We could not have had such a successful conference without you all!! Our next conference will be in Indianapolis on November 12, 2005. Details will follow.

October 2004 Conference

Our last conference was an amazing success. We had over 200 people attend from 23 states and 4 countries. We owe so much thanks to our terrific speakers, Dr. Maria New, Dr. Phyllis Speiser, Dr. Deborah Merke, Dr. Sheri Berenbaum, Dr. Susan Baker, Dr. David Sandberg, Dr. Dix Poppas, Amy Sturge, Meg Keil, Janet Green, and the fabulous panel of adults with CAH for sharing their knowledge and experience with us all. We also owe a debt of gratitude to our master of ceremonies, Dr. Luigi Garibaldi, and to Overlook Hospital, Atlantic Health Systems for hosting our conference. They were so gracious and went out of their way to help us make the conference a success. Finally, I want to thank our conference planner, Robin Levan, who once again did a magnificent job, our superb staff, Meryl Stone, Pam Knight, and Laurie Hitzig, as well as all of the volunteers who showed up to help out on the day of the conference. I also want to thank our corporate sponsors, Pfizer,

Los Angeles Every1CARES Awareness and Fundraising Luncheon

Our first Every1CARES Luncheon will take place on March 10, 2005 from 10:30am-2pm at the Skirball Cultural Center in Los Angeles. Our wonderful event committee and its energetic and passionate chair, Jami Abell Patterson, have been hard at work to ensure that this event is spectacular and raises significant sums for CAH research. We are honoring Senator Dede Alpert for her dedication to children and her sponsorship and support of legislation to expand California's newborn screening program to include CAH and many other life-threatening disorders. In August of this year, Governor Schwarzenegger signed this mandate into law and California should begin its expanded screening for CAH in July, 2005. Our luncheon will begin with a boutique and silent auction at 10:30am, followed by an elegant lunch, a few

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eye-opening and heart-tugging remarks about CAH, the premiere of our CARES video (see page 17 for details), and our special award presentation to Senator Alpert.

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CARES Conference in New Jersey



Dr. Maria New from Mt. Sinai was a featured speaker at our New Jersey conference.



Chelsea, who turned 9 on the day of the conference, blows out the candles on her birthday cake as Josh Leight (with guitar), Adam Leight (center), and other conference-goers look on.



The main lecture hall at Overlook Hospital was filled to capacity as we hosted over 200 CARES family members and professionals.

We still need sponsors for the event and donations for our auction. Please check our web site for sponsor and donation forms. We appreciate donations of airlines tickets or miles, vacation homes/packages, dinner at restaurants, fine wines, tickets to shows and sporting events, memorabilia, priceless items like backstage tours or guest appearances, lunch with a celebrity, autographed items, golf games, professional services, beauty or spa services, decorative items for the home, fashion items, electronics (we hope someone will donate an iPod), etc.

We also would love for all of you in the Southern California area to attend if you can, and bring your friends. It will be a fun way for you to show support for CARES and help raise money for CAH research and our programs.

Newborn Screening Update

It is time for us to direct our efforts to the remaining states that do not screen for CAH and other life-threatening disorders. California and

Oklahoma, thanks to the efforts and advocacy of CARES members, have approved and funded expanded newborn screening to include CAH and expect to begin screening for these disorders soon. Nebraska is working on adding CAH to its panel and hopes to implement it in 2005. Washington, DC, New Hampshire and Utah are still working on getting expansion approved and funded. Kentucky has approved the addition of CAH, but has not appropriated the funding to implement it. West Virginia, Louisiana, Kansas, Arkansas, and South Dakota do not screen for CAH and are not actively moving towards this in the near future. Montana offers CAH screening as an option for an additional fee, but it should be mandated. Members in many of many of these states are already working on media exposure for the issue, talking to their legislators, participating on state genetics committees, etc., to move things forward. If you can help, please call the office or send email.

We have a lot to look forward to this year as a community and as an organization. Thank you all for all of your help and support during this past year. CARES continues to grow and make more of an impact through the involvement and participation of our members. You are the best!

Warmly,

Kelly

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This newsletter is published 3 times a year.

The Basics of Genetic Testing

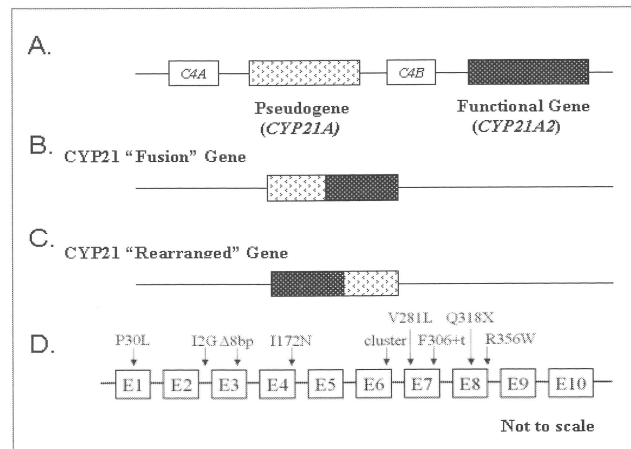
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Another gene, called CYP21A, is located very close to CYP21A2 on each chromosome 6. CYP21A is a *pseudogene*. A pseudogene is a copy of a gene that is no longer working. The most common genetic situation seen on each chromosome 6 in people who do not have CAH is one copy of CYP21A located close to CYP21A2 as shown in "A" of the diagram below. This means that the person has two pseudogenes and two functional, or working, genes in total. The pseudogene no longer works because it has acquired multiple alterations, or mutations, along its length as shown under "D" in the diagram. Although the pseudogene has many mutations that make it different from the functional gene, the two are 98% identical overall. Because both gene and pseudogene are so similar, genetic testing is more difficult and the gene is more likely to acquire changes that prevent it from working.

Mutations in the functional gene occur because one or more mutations have moved from the pseudogene to the functional gene. Sometimes this involves little bits and pieces of the pseudogene and other times it involves major rearrangements of the gene and pseudogene. The major rearrangements can delete or add an entire copy of the gene or pseudogene, or can leave behind changed structures that are part gene and part pseudogene as illustrated in "C" and "D" of the diagram.

Labs that do testing for CAH use some or all of the following genetic techniques in order to provide results. Genes are made of DNA, so the first step of the process is isolating DNA

(deoxyribonucleic acid). Usually a blood sample is used as the DNA source, but other bodily tissues and fluids can be used as well. Then, different methods are used to detect small genetic changes, or large alterations such as whole gene deletions. Labs that offer testing for CAH should be able to detect 90% to 95% of the mutations causing the classic form of the disease.



Glossary of Genetic Testing

The **polymerase chain reaction (PCR)** is a common method used for genetic testing that can tell one gene apart from another. PCR can be thought of as a "genetic Xerox machine." By using PCR, the laboratory can make up to a million copies of a specific gene or piece of a gene from a DNA sample for studies.

Some laboratories use a technique called a **Southern blot**. In this method, extracted DNA is cut at specific sites near or within the gene and pseudogene. A specially made piece of DNA, referred to as a probe, is used to detect the specific DNA pattern of the gene and pseudogene. A Southern blot will usually find large gene deletions and rearrangements. After a lab performs a Southern

blot, it will also need to perform PCR and some other method, like sequencing, in order to find those smaller changes.

PCR can also be used to distinguish the pseudogene and functional gene, and in most cases can determine the number of functional genes and pseudogenes a person has in their DNA without using the Southern blot technique. Unique landmarks outside the gene and pseudogene are used to separately copy and identify the gene, the pseudogene, as well as any rearranged copies of gene and pseudogene that might be present.

The copies of the gene and pseudogene that have been made by PCR can be closely looked at using a technique called **sequencing**. Through sequencing, labs look for the presence or absence of the most common "small" genetic alterations seen in individuals affected with CAH. These are known as P30L, Intron 2 "G", G110del8, I172N, Exon 6 cluster mutations, V281L, F306+1, Q318X, R356W, and P453S. Different labs may choose to look for some or all of these small mutations. This technique can also be used to carefully review the entire gene for less common mutations; this is generally referred to as "full gene sequencing" or "sequencing the entire coding region." This approach would be valuable for individuals with a confirmed diagnosis of CAH due to 21-OH deficiency who do not have one of the common mutations. Full gene sequencing is labor intensive, generally takes longer to obtain results, and might cost significantly more than the tests for

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the common mutations.

In families where the diagnosis of CAH due to 21-hydroxylase deficiency has been proven but a mutation cannot be identified, another test called **linkage** can be used to determine if a relative has the mutation or if a pregnancy is affected. Linkage cannot be performed without testing multiple family members. At least both parents must be tested, as well as a previously born affected child. Unique genetic landmarks either within the gene or pseudogene, or close to, or "linked" to the CYP21A2 gene are used to identify and follow copies of the mutated gene causing CAH from generation to generation without actually knowing the exact mutation involved. Sequencing small areas of the pseudogene and functional gene can provide good genetic information for linkage analysis. Linkage can be used to predict whether or not a subsequent child will be affected with a high degree of accuracy. Because linkage does not test for the actual mutation, however, there is a small risk that the linked genetic landmark will become unlinked and thus an incorrect result will be obtained.

Who Might Consider Genetic Testing?

Genetic testing can be used to confirm the diagnosis of CAH and identify the mutations present in a person who is suspected of having classic or non-classic CAH. For example, genetic testing can help confirm the diagnosis in infants that have a positive newborn screen for CAH. Adults with suspected CAH due to infertility problems or women who have symptoms of androgen excess might also have their

diagnosis confirmed through genetic testing.

Parents of a child with CAH may want to know the genetic alterations present in their affected child. This information can be used, early in pregnancy, to determine whether a subsequent child has CAH by testing the baby through amniocentesis or chorionic villus sampling. The information could help direct prenatal treatment with dexamethasone or help families and doctors anticipate and prepare for the birth of an affected child. Genetic information can also be used for preimplantation genetic diagnosis in future pregnancies in order to significantly reduce the chance of a having another child affected with CAH (see "Preimplantation Genetic Diagnosis: Another Alternative" Winter 2003-04 newsletter).

Finding the mutations present in an affected child allows testing of other family members, such as the aunts and uncles of an affected child, for the specific mutation(s) identified. This allows other relatives to know if they are carriers so that they can use the information in the way that is best for themselves and their own family planning. If an affected child has not been tested and a relative decides to pursue their own testing, problems with interpretation of the results can occur. It is always best to test the affected individual. For example, if the relative is negative, it is still possible that the family carries a mutation that cannot be detected with current methods, and that would not be known unless the affected individual was also tested and was negative.

Genetic testing can also be used to screen an unaffected person who

has no family history of CAH to determine if they carry CAH. This can be especially useful for the partners of individuals who are either affected by CAH, or are known carriers of CAH, for the purpose of family planning and pregnancy management. If both members of a couple are known carriers, they can consider the option of starting dexamethasone treatment early in the pregnancy which would reduce the degree of masculinization of the female genitalia in an affected female infant.

For couples that have no known personal or family history of CAH, but are currently pregnant with a female fetus that has ambiguous genitalia detected by prenatal ultrasound, genetic testing for CAH may be appropriate after other causes have been ruled out. The advance knowledge can help the family and physicians prepare for the medical, social and emotional issues related to the diagnosis and birth of an affected child.

Prenatal Testing for CAH

Prenatal testing can be performed by two methods. **Chorionic villus sampling (CVS)** is a procedure that obtains fetal cells by sampling cells from the developing placenta. The procedure is usually done with ultrasound guidance to see the physical structures of the patient and fetus. CVS is typically offered at 10-12 weeks from the last known menstrual period. As with any prenatal procedure, CVS carries with it a small risk of miscarriage. The advantage of CVS is that the procedure takes place in the first trimester and genetic test results can be obtained early in the pregnancy.

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How CAH Genes Translate Into Symptoms: The Genotype/Phenotype Connection

By Erin Anthony

Congenital adrenal hyperplasia is a fairly well characterized disorder, but its effects are far from simple. CAH is caused by mutated recessive genes and the physical effects can range from severe cases evident at birth to those that are never diagnosed. Even within the same family, the characteristics of CAH can be markedly different.

Understanding the different ways in which CAH can occur requires understanding the difference between phenotype and genotype—between what you see and what you get. In other words, phenotype refers to observable physical characteristics, while genotype refers to the genes a person carries within their body's cells.

The Basics

Humans are “diploid,” having two copies of every chromosome—one from their mother and one from their father—46 in all. Within each chromosome are genes, the body's directions for operation. Because every person has two copies of each chromosome, they also have two copies of each gene.

A gene can be dominant or recessive. As you might expect, a dominant gene takes precedence over a recessive gene. In fact, recessive genes only get the opportunity to show themselves when there are two copies of them (double recessive genes) for a given trait.

For each gene there are variations, or alleles. Some genes may have only one or two variations,

while others, like those responsible for CAH, may have several variations.

A person's genetic combination is called their “genotype.” Sometimes genes acquire glitches or mutations. And, while the body is usually pretty good about fixing these problems, a few persist and are passed down from generation to generation—such as with CAH.

Which alleles a person receives depends on the combination of parental gametes (eggs or sperm). Depending on which alleles a person receives, their genotype (and as a result, their phenotype) will be different.

The phenotype is the physical expression of the genotype. Blue eyes, a birthmark and CAH are all examples of phenotypes. Depending on the combination of alleles, whether they are dominant or recessive, as well as other complicating factors, phenotypes will vary.

Genotypes and Phenotypes of CAH

CAH is a recessive disorder that results in the deficiency of one of five enzymes required for the production of cortisol. Because it's recessive, a person with CAH (having the phenotype) must have two mutated copies of the gene, one from each parent.

The most common cause of CAH is mutation of the gene CYP21A2, which causes deficiency of the enzyme 21-hydroxylase.

According to researchers, CYP21A2 is one of the most polymorphic (“many-formed”) human genes studied to date. This means it has many different variations, or alleles.

CYP21A2 is found on the short arm of chromosome 6, in a region that also carries many other important genes whose products control immune function. Mutations of CYP21A2 affect males and females equally and has been found at a frequency of 1 in 60 in the general population—meaning in a group of 60 people, one will carry a mutation of CYP21A2 on one of their chromosomes. A few different terms are used to describe different genotypes. For CAH, the terms refer to the presence of mutations. People with two copies of the same mutation are called “homozygous” while people with two different kinds of mutations are “compound heterozygous.” A person with only one mutant copy of a gene is “heterozygous,” or a “carrier,” and has no symptoms of the disorder.

Knowing a person's genotype allows physicians and genetic counselors to calculate the probability of a child inheriting a particular trait. In this case, knowing the genotype and predicting the phenotype may influence prenatal care.

For two carriers of a CYP21A2 mutation, there is a 25 percent chance they will have a child with CAH. A child born to a carrier and non-carrier would have a 50 percent chance of

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also being a carrier. Two affected individuals have a 100 percent chance of having a child with CAH.

Although genotypes are easily predicted, the resulting phenotypes are not as simple. Because CYP21A2 has so many different variations, there are several different mutations that can occur. Here, only the most common mutations—associated with 21-hydroxylase deficiency—will be discussed.

CAH phenotypes (classic salt-wasting, simple virilizing, and non-classical) depend on the severity of the mutations. The type of mutation determines the level of enzymatic activity (or inactivity). Enzymes are produced according to what the gene “says”—so if a gene is defective the resulting enzyme will also be defective.

For CAH, the phenotype is usually the result of the less severely affected allele. CYP21A2 mutations can be grouped into three categories according to how enzymatic activity is affected.

The first group is most often associated with salt-wasting CAH. It consists of severe mutations such as deletions (loss of all or parts of the gene's information) or nonsense mutations (changes that “don't make sense” to the molecules that “read” the gene) that completely stop enzyme activity needed to make cortisol and aldosterone.

The second group is usually associated with the simple virilizing form of CAH. It consists mostly of “mis-sense” mutations, where the enzyme-making instructions are changed but the enzyme still has some function. Here, the enzymes usually have 1 to 2 percent of their

normal activity and so permit the body to make aldosterone, the main sodium-retaining hormone made by the adrenal glands.

The third group of mutations is usually associated with nonclassical CAH (NCAH). It consists of mutations that produce enzymes retaining 20 to 60 percent of their normal activity—so the resulting phenotype is usually less severe and usually detected later in life.

It is important to note that dividing phenotypes into distinct groups is a simplification. In reality, there may be a continuum of phenotypes even within the same genotype. Humans are highly dynamic and unique in the expressions of their genes. Thus, the rest of a patient's genetic background will influence their CAH phenotype. Modifying genes, such as those that regulate the production of androgens and estrogens, as well as individual sensitivity to these compounds, will influence a patient's phenotype.

A French study of women with NCAH emphasizes the complicated genotype-phenotype relationship. Studying the genotype-phenotype correlation in women with NCAH is important because those carrying a severe mutation of CYP21A2 risk giving birth to children with the classical form of the disease. Here, screening for carrier status in the partner is crucial.

Deneux, et al., reported in *The Journal of Endocrinology and Metabolism* in 2001 that phenotype cannot always be predicted by genotype and in some cases may be milder than expected.

According to Deneux, 2 subjects in the study were found to carry 2 potentially severe mutations

often associated with the simple virilizing form of the disorder, but showed no virilization at birth. The clinical and biological phenotypes of the patients were particularly marked and may indicate a phenotypic continuum among the three forms of 21-hydroxylase deficiency, the researchers said.

According to Deneux et al., “This failure of strict correspondence of phenotype and genotype must be borne in mind when performing a prenatal fetal DNA analysis. Predicting phenotype must remain cautious.”

The complicated relationship between genotype and phenotype in patients is highlighted in families with both symptomatic and asymptomatic forms of NCAH. According to Deneux, et al., this confirms that variability in phenotypic expression is influenced by factors other than heterogeneity of CYP21A2, such as modifying genes, individual differences in adrenal and extra-adrenal chemical pathways, cortisol requirements, and individual sensitivity to androgen oversecretion.

Understanding the relationship between genotypes and phenotypes for CAH is important for better management of the disorder. When physicians and genetic counselors know more about the relationship between genotypes and phenotypes, quality of life for patients improves.

CF

The author, a CARES member, is a senior at Lehigh University, majoring in biology and science / environmental writing.

CAH and Me

What It's Like Being A Teen With CAH

By Jennifer Kilmartin



Jennifer Kilmartin is 17 years old, a resident of Calgary, Alberta, Canada, and was diagnosed with CAH at birth. She agreed to answer a few questions and give us her take on the issues people with CAH face as they enter adolescence and look forward to adulthood. Of course, everyone's experience will be different, but her views may get you thinking about what to expect as your child with CAH grows up and begins to take responsibility for his or her own health. The proper perspective is key: As Jennifer says, "Live by the rule that CAH doesn't define who you are or what you will become in the future; CAH is only a small fraction of what you have to deal with."

"How does CAH affect your life right now?"

Being a teenager with CAH doesn't affect every aspect of my life, for CAH does not define who I am. I am still like any other teenager you come across and can do pretty much anything that any other teenager can do.

Still, injuries are still out there for me, meaning that when I play sports I need to be more careful than someone else, since I, as well as any other CAH person, react differently to any kind of injury.

Through childhood to adolescence having CAH can be very hard emotionally and mentally – especially for a teenager. With any disease/condition anyone will feel different or perhaps out of place.

That is normal – at least it was for me. As your child gets older they will gain more responsibility for taking medication, watching out for signs of an adrenal crisis, and many other responsibilities that will come into place as they get older. As a child, my mum would administer my medication – but now that I am 17 years old I have to remember to take my medication three times a day. Some parents may ask when they should start to let their children be responsible for their medication. Good question: for me, I was in about grade two or three when I started to do things on my own. It will vary among children – as some develop quicker than others in maturity.

Sometimes I would forget to take a dose in one day, but that only happened every so often and didn't become a threat to my well-being. If I missed a dose, I would usually just take it at the most convenient time. As I stated before, I take medication three times a day, which means that, yes, I do take my medication while in school. Many

people say that I am being really brave to show that I have to take medication – but really, I wasn't always that brave. I used to skip out on my medication at school because I didn't want anyone to know.

It can sometimes be hard for a parent to let their children take responsibility because they never know if they are taking the medication and they don't have as much control. But all people must learn to be responsible at some point in their life. Parents, start training your children to be responsible while they are young!

"In what ways might having CAH enhance your life?"

Having CAH might enhance my life in my not complaining about it because I could have been born with something worse. Being a CAH teenager has gotten me curious about other diseases/conditions and has got me into researching and trying to understand what other people go through. In my spare time I sit down and study different diseases/conditions in the world because I want more people to know and understand what CAH is – so in return I want to know and understand what other people go through.

Being affected by CAH has also given me the opportunity to share my knowledge with others who show interest in what I have and how it affects me. Although not everyone understands, they can continue to learn and share with others.

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"What are your worries and fears for the future?"

My worries and fears for the future are mainly just the curiosity about whether or not my children will be affected by this same disease that I have and have to go through the same thing I go through now, with CAH. I also worry that that having CAH will interfere with my profession of choice, which is being part of the Calgary EMS. I start my training summer of 2005 for the EMR/EMT testing – will CAH affect my results in the tests that I have to take? I worry about what my future husband will say when he finds out I have CAH, will he stay with me or will he leave me. How will my children react? Those are all my fears, they may change as the years go by – and not everyone's fears are the same. Talk with your children about their worries and fears for the future!



"Is body image a concern for you?"

Being a female, body image is always a concern. Being a female with CAH is a different story and sometimes brings out the worst. I was on prednisone for about two years and I gained a lot of weight and

became very chubby. I wasn't too happy about that, to say the least. Just a few months ago, I was switched over to hydrocortisone (Cortef®) and it has brought me a long way. I have lost some weight, I look a lot healthier, and I also feel a lot better and am happy with myself. The Cortef has helped me emotionally and mentally in life.

All in all, body image is just a small portion of life – it doesn't at all define who you are or who you will become. If your daughter is concerned that she will never get a boyfriend because of the way she looks – well, to say the least, she will find a guy (or a guy will find her) who likes her for who she is and not what she looks like. Live by the rule that CAH doesn't define who you are or what you will become in the future; CAH is only a small fraction of what you have to deal with.

"How does it affect interactions with others – friendships and dating?"

It is always tough when it comes to interacting with others, but I know that I can do anything that my friends can – and if I can't do it, no big deal. When people ask what my medic-alert bracelet is for – I don't really want to tell them because I am afraid, but they asked and I feel they deserve an answer. Some people treat me differently but others don't put a label on me and they just continue the friendship we started with in the beginning. **CF**

Jennifer Kilmartin is a student in Calgary, Alberta, Canada.

CARES Teen Chat Group



Hi, my name is Amanda Wells. I have salt-wasting CAH. I am 13 years old. My parents thought it would be a great idea to make a group for teens with CAH so they can talk about feelings, questions, and life experiences with CAH. They spoke with Kelly Leight from CARES and she agreed. I volunteered to set this up.

To join, go to <http://health.groups.yahoo.com/group/caresteenchat/> and click on "Join This Group." I hope you are all as interested as I am and I hope to speak with you in the group. **CF**

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.

NJ Resource for Affordable Prescription Drugs

"Rx4NJ," a program aimed at making New Jersey residents aware of ways to obtain free and discounted prescription drugs, provides a list of over 300 existing patient assistance programs (PAPs) that provide free, or nearly free, prescription drugs.

Assistance is generally provided free or on a discounted basis to patients who do not have prescription drug coverage or are underinsured and cannot afford the out-of-pocket cost of their medicine. For details, go to the website:

www.rx4nj.org

Or call toll-free: (888) 793-6765

PAPs to make Rx's more affordable are also offered in the following states:

Ohio:

<http://www.rx4ohio.org>
(877) Rx4OHIO

West Virginia:

<http://www.rx4westvirginia.org>
1-877-WVA-RX4U

Illinois:

<http://www.rx4illinois.org/>
1-877-793-6745

Rhode Island:

<http://rxforri.org/>
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Washington:

<http://rxhelpforwa.org/>
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GOOD NEWS FOR CORTEF AND SOLU-CORTEF USERS:

Manufacturing Shortage Ended; Drug Added to Patient Assistance Programs

Cortef is now both more available and more affordable, due to the advocacy efforts of CARES Foundation and the support of Michael P. Wajnrajch, M.D., Pfizer's U.S. Medical Director, Pediatric Endocrinology Division.

Pfizer, the maker of Cortef® has announced an end to the shortage of this drug.

"Thanks to improved efficiencies and increased production, both Solu-Cortef and Solu-Medrol are off allocation, meaning that both are freely available," says Dr. Wajnrajch. "Additionally, following approval by the FDA, we will be increasing production further, so we do not anticipate any shortages in the future either. There are no shortages of our other corticosteroids."

Thank you, Dr. Wajnrajch, for spearheading the effort within Pfizer to eliminate the shortages of these essential medications, and having Cortef added to PAP lists. See www.pparx.org for details.

In addition, CARES was able to persuade Pfizer to add Cortef to its patient assistance program (PAP). Now, those who cannot afford to pay for Cortef can apply for assistance through Pfizer.

For more information on obtaining Cortef through this PAP, call Pfizer Prescription Medicine at (800) 879-3477, or go to its website (www.pfizer.com). **CF**

FLORINEF UPDATE

Florinef® (fludrocortisone acetate) tablets, distributed by Monarch Pharmaceuticals, Inc. (a subsidiary of King Pharmaceuticals, Inc.) is not currently available under the company's patient assistance program. But keep checking—thanks to CARES' efforts, this status may change later this year.

To inquire about whether the company has begun to accept new applicants for this program, contact King Pharmaceuticals, Customer Service and Sales, at (888) 840-5370. **CF**

Walter Futterweit, M.D., Expert in Androgens and Women's Health, Joins CARES Medical and Scientific Advisors

CARES Foundation is delighted to announce that Walter Futterweit, M.D., Chief of the Endocrine Clinic of the Mount Sinai Medical Center, has joined the CARES Scientific and Medical Advisory Board.

A clinical endocrinologist and researcher with a particular interest in androgens and women's health, he has devoted the past 25 years to the study of polycystic ovary conditions (PCOD and PCOS) and their pathophysiology, treatment, and manifestations, as well as CAH and other diseases of androgen excess.

Dr. Futterweit is currently a Clinical Professor of Medicine at the Mt. Sinai Medical Center, and has published more than 40 articles and abstracts on PCOS, as well as a 1984 textbook.

For the past five years, Dr. Futterweit has been active in national and international meetings in developing proposed guidelines for endocrinologists who treat PCOS.

He has been elected to the Advisory Board of Polycystic Ovarian Syndrome Association (PCOSA), a national organization of women with PCOS, as well as to the board of the Androgen Excess Society, and often lectures at their meetings.

CARES Foundation Welcomes Ellen Seely, M.D., to its Medical Advisory Board

Ellen W. Seely, M.D. is the Director of Clinical Research in the Division of Endocrinology, Diabetes and Hypertension at the Brigham and Women's Hospital in Boston, Massachusetts and Associate Professor of Medicine at Harvard Medical School.

A leader in patient-oriented research of the physiology of blood pressure regulation in women during pregnancy and following menopause, Dr. Seely's research has focused on understanding cardiovascular risk factors in women. She is the principal investigator on three NIH projects. Clinically, she sees women with endocrine problems complicating pregnancy, and has expertise in assisting women with classical CAH who wish to become pregnant.

Dr. Seely is also a Fellow of the Council for High Blood Pressure Research of the American Heart Association; has been designated a Clinical Specialist in Hypertension by the American Society of Hypertension; and has been selected for inclusion in *Who's Who in Medicine and Healthcare*, and *Who's Who in America*. She was named one of Boston's Top Doctors for Women in Endocrinology by *Boston* magazine in 2001.

Renata Blumberg Joins CARES Staff

Renata Blumberg, our new administrative assistant, received a B.A. from Columbia University and a Master's degree from the University of California, Davis. As a student in New York City, Renata worked for a variety of non-profit organizations. Prior to joining the CARES Foundation staff, she worked at the Women's Resources and Research Center at UC Davis.

Renata recalls that when she first visited our office, she was "impressed with how much CARES has accomplished in such a short time," and "knew she wanted to be a part of this inspirational team."

Renata also has extensive international experience as a Fulbright student in Latvia and a researcher in Lithuania, where she assisted rural non-profits. Renata is enthusiastic about being a part of an actively growing organization, and hopes to learn more about non-profit management.

She brings to CARES her considerable organizational, linguistic, and computer skills, as well her interest in serving children and families affected by chronic illness. You can reach her at (866) 227-3737, or e-mail her at: renata@caresfoundation.org.

U.S. LABORATORIES OFFERING DNA TESTING FOR CAH

NAME	CENTER FOR GENETIC TESTING	COMPGENE COMPREHENSIVE GENETIC SERVICES	
DIRECTOR	Frederick V. Schaefer, Ph.D., Director of Molecular Genetics	Anthony T. Garber, Ph.D., Head of Molecular Genetics	
CONTACT INFORMATION	6465 S. Yale Ave. Tulsa, OK 74136 Toll Free: (866) 846-0315 (918) 502-1725 www.saintfrancisgenetics.com	3720 N. 24th St. Milwaukee, WI 53222 Toll free: (877) COMPGENE (414) 393-1000 www.compgene.com	
HOW LONG OFFERING CAH DNA TESTING	10 years	6 years	
MUTATION DETECTION RATE	94%	90-95%	
HOW MANY TESTS DONE TO DATE?	Over 500	Over 500	
RESULTS INTERPRETATION / GENETIC COUNSELING INCLUDED	Informally	Available on site, not included in testing fee	
CLIENT PRICE	\$389	\$324	
ACCEPT INSURANCE?	Only from Oklahoma insurers	Only from Wisconsin insurers	
DRAW SAMPLES AT THE LAB	Yes	Yes	
ACCEPT SELF-REFERRALS	Yes	No	
USUAL PROCEDURE	First, sequence most of latter half of gene; then mutation analysis by PCR; their method finds site-specific mutations, deletions and rearrangements. No Southern blot.	Standard mutation panel, direct mutation analysis. PCR and Southern blot. No sequencing. Linkage analysis.	
TURN-AROUND TIME	3 weeks	Linkage analysis: 72 hrs. DNA analysis: 7-10 days	
PRENATAL DIAGNOSIS	Yes	Yes	
TYPES OF SAMPLES TESTED	Blood, cells from amniotic fluid (supernatant), cultured cells from amniocentesis or CVS; preserved tissue samples	Blood, cells, prenatal cells, buccal cells, preserved tissue samples	
FOR PRENATAL DIAGNOSIS, REQUIRE PARENT/S AS WELL AS OFFSPRING BE TESTED?	Not required, often helpful; do as needed/desired	Very strongly preferred, not absolutely required	
COMMENTS	Integrate sequencing very early in test procedure; can test DNA directly from amniotic fluid; this gives fast results; also test cell cultures for quality control and maternal cell contamination studies	Can test DNA directly from amniotic fluid; this gives fast results, also test cell cultures for quality control and maternal cell contamination studies	

U.S. LABORATORIES OFFERING DNA TESTING FOR CAH

	ESOTERIX, INC.	MOUNT SINAI MEDICAL CENTER	QUEST DIAGNOSTICS
DIRECTOR	Frank K. Fujimura, Ph.D., Executive Director, Molecular Genetics	Maria New, M.D., Professor of Pediatrics and Director, Adrenal Steroid Disorders Program	Raymond Fenwick, Ph.D., Scientific Director, Molecular Endocrinology
CONTACT INFORMATION	4301 Lost Hills Rd. Calabasas Hills, CA 91301 (800) 444-9111 www.esoterix.com	1 Gustave Levy Place, Box 1198, Annenberg, 17 th Floor, Rm. 271 New York, NY 10029 (212) 241-7962 www.marianew.com/Laboratory	33608 Ortega Highway San Juan Capistrano, CA 92690 (949) 728-4000 www.questdiagnostics.com
HOW LONG OFFERING CAH DNA TESTING	1 year	13 years	1 year
MUTATION DETECTION RATE	95% with PCR and Southern blot; almost 100% with sequencing	97% with PCR and Southern blot; almost 100% with sequencing	90%
HOW MANY TESTS DONE TO DATE?	About 200	Over 5,000	Over 350
RESULTS INTERPRETATION / GENETIC COUNSELING INCLUDED	No genetic counseling; results interpretation given to referring physician	Yes, included in testing fee. All results reviewed by Dr. Maria New; patient given interpretation in context of all clinical data	Staff genetic counselor relays results to referring physician
CLIENT PRICE	\$420	\$600*	\$495
ACCEPT INSURANCE?	Yes	Yes	Yes
DRAW SAMPLES AT THE LAB	Yes	Yes	No
ACCEPT SELF-REFERRALS	Yes	Yes	No
USUAL PROCEDURE	PCR, multiplex mini-sequencing for 12 common mutations, Southern blot; further sequencing if requested.	PCR for panel of selected mutations and Southern blot for deletions and gene conversions; sequencing when needed to make diagnosis	PCR, multiplex mini-sequencing for panel of selected mutations; full gene sequencing for rare mutations available in mid- to late 2005
TURN-AROUND TIME	2 weeks	3 weeks or less; urgent samples 2 weeks or less	1 to 2 weeks
PRENATAL DIAGNOSIS	No	Yes	Yes
TYPES OF SAMPLES TESTED	Blood	Blood, DNA, cultured cells from amniocentesis and CVS	Blood; prenatal specimens accepted only after speaking with staff genetic counselor
FOR PRENATAL DIAGNOSIS, REQUIRE PARENT/S AS WELL AS OFFSPRING BE TESTED?	Not applicable	Prefer to have parents or offspring, not both, for informative results. Not required when ultrasound findings are the indication for testing.	Yes, either a parent or previously affected child must be tested in addition to offspring
COMMENTS		*Fee includes genetic counseling and reflects laboratory's specialization in this test. Leading CAH expert Dr. Maria New personally reviews each case.	

HHS Newborn Screening Committee Update

Within the next few months, the US Department of Health and Human Services (HHS) is expected to release national guidelines detailing a minimum set of newborn screening tests recommended for inclusion in all state newborn screening programs.

A major impetus for the guidelines is the wide disparity in the number of conditions now included in state-mandated newborn screening programs. For example, Kansas, Kentucky and Arkansas test for just four or five conditions each, while Iowa tests for more than 40.

"We're concerned that there is not equity for parents across states, and we feel that we should move in that direction," says Peter Van Dyck, director of the Maternal and Child Health Bureau within the

HHS Health Resources and Services Administration (HRSA).

The guidelines will be based in large part on a report prepared for HHS by the American College of Medical Genetics (ACMG). Although the report is not yet final, the ACMG shared an early draft with the HHS Advisory Committee on Heritable Disorders & Genetic Diseases in Newborns & Children, which voted in September 2004 to "accept and recommend" the report's conclusions.

ACMG Executive Director Michael Watson said that the ACMG considered two alternate recommendations for the core panel: "to argue that (government) mandated newborn screening is the only way or to say that . . . it should be the standard of care that all babies be screened, basically putting responsibility on the pediatrician." "We opted for the first choice," he said.

Earlier this year, HHS funded seven regional newborn screening centers to share best practices among states and develop regional strategies to optimize newborn screening services.

In the meantime, the HHS genetics committee has met only twice. Future recommendations will likely deal with funding issues as well as consideration of a national newborn screening process.

CARES Executive Director Kelly Leight sits on the Newborn Screening and Follow-Up and Diagnosis workgroup for the ACMG, representing the interests of the CAH community.

Adapted and abridged from the Association for Public Health Laboratories web site: www.aphl.org

VOLUNTEERS FOR RESEARCH STUDY NEEDED!

Women's Hormones and Impact on Mood Study (WHIMS)

If you are between the ages of 18 and 40 and are premenopausal, you may be interested in a study of the effects of excess androgens (such as testosterone) on women's mood. Dr. Gail Schoen Lemaire, a member of CARES, and a nurse and researcher at the University of Maryland School of Nursing in Baltimore, is recruiting volunteers for the WHIMS study of how symptoms of androgen excess influence women's mood and overall well-being.

In order to take part in the study, participants must be premenopausal, between the ages of 18 and 40, and diagnosed with NC-CAH. Study participation involves completing a one-time, written survey. For more information or to request a survey, please email Dr. Lemaire at the WHIMS Study Center at lemaire@son.umaryland.edu or call her at (410) 706-4914.

CF

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New

New

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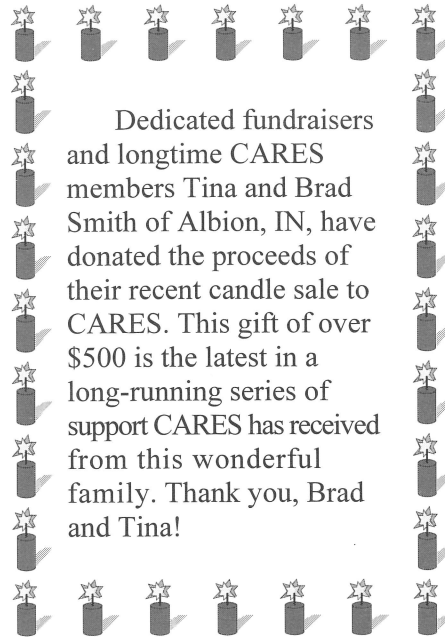
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Fundrai\$ing Corner

Thank You... Thank You!!!



Dedicated fundraisers and longtime CARES members Tina and Brad Smith of Albion, IN, have donated the proceeds of their recent candle sale to CARES. This gift of over \$500 is the latest in a long-running series of support CARES has received from this wonderful family. Thank you, Brad and Tina!

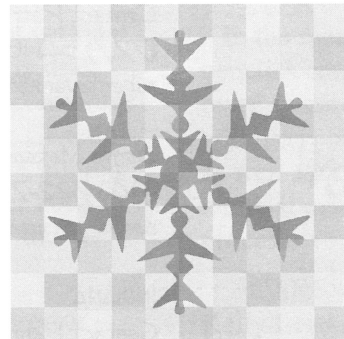
Many thanks are due to Louise Fleming, our South Carolina support group leader who dreamed up our very successful holiday card fundraiser.

Those blue-and-white snowflake cards, suitable for all December holidays, were the inspiration of Louise and her friends, Stephen and Michelle Keith and Barry Stanley, co-owners of a commercial graphics company in Greenville, SC. called Visual Graphics.

Thank you, Louise and Michelle, for designing and producing these cute cards, and to

Holiday Cards

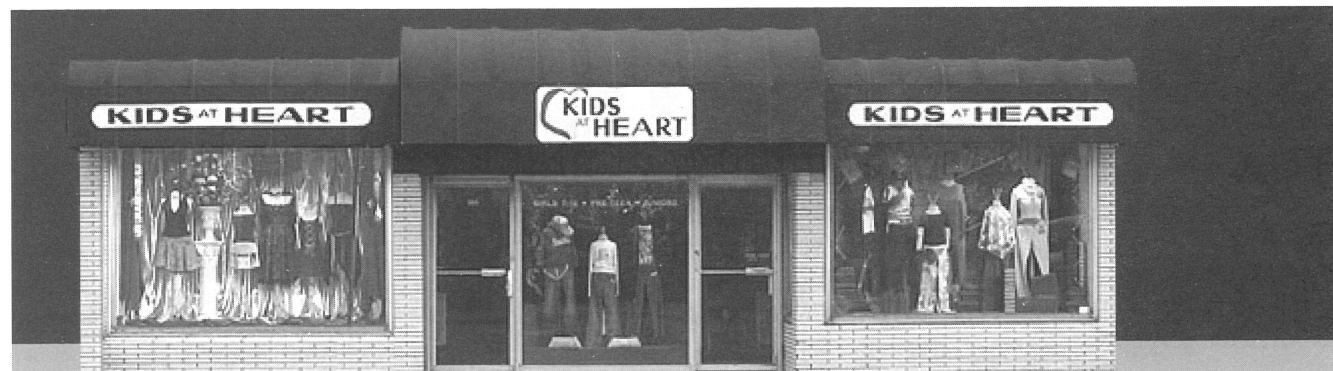
Visual Graphics for generously printing them at no extra cost.



We could not have gotten the cards ready in time without our wonderful website manager Sue Bianchi's efforts in getting the CARES web site order page up in record time.

This volunteer effort had practically no direct cost to CARES—Louise donated mailing fees—and netted us nearly \$3,000!

Thanks also to all who purchased and gave out these cards for helping to make this a success and helping to raise awareness of CAH.



Lisa Baranker, Jill Cohn, and Beth Levine of Kids At Heart at 565 S. Livingston Ave., Livingston, New Jersey have been kind enough to sell our EVERYICARES bracelets in their store for girls 7-16, pre-teen and juniors. Kids At Heart is a fun place to shop, offering electronic gift cards and beautiful gift wrapping for all your gift-giving needs. If you are looking for the latest in street wear or dress wear, visit Kids At Heart or call them at (973) 992-2440.



SAVE THE DATE

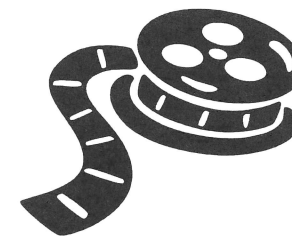
Saturday, November 12, 2005

Fifth Annual CARES Family Conference

Riley Children's Hospital Indianapolis, Indiana

NOW FILMING: THE CARES VIDEO

Filming is almost complete for our CARES video, which we hope will be an effective way to spread the word about CAH. Many thanks to Dr. Phyllis Speiser, to Altered Image and Crazy Duck Productions, and to all our CARES members who participated in the filming.



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Cody Cares Medical ID Jewelry

4155 Carson Avenue
Indianapolis, IN 46227
317-783-7702 • contact@codycaresid.com
<http://www.codycaresid.com/> (new website!!)

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

- ✓ ID Bracelets
 - ✓ Shoetags
 - ✓ ID Wristbands (*perfect for infants & sports*)
- New items added all the time... check the website!!*

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

CHAT GROUP FOR WOMEN WITH LATE-ONSET CAH

CAHSISTERS2 is a listserv for adult women with late-onset CAH.

You can subscribe to this listserv through Yahoo! Groups, a free, easy-to-use email group service.

To learn more about the CAHSISTERS2 group, go to:

<http://groups.yahoo.com/group/CAHSISTERS2>

RUNNERS WANTED!

Need a little extra motivation to train for the New York Marathon?

Let's start a team of runners to come together and run the Marathon to raise money and awareness for CARES Foundation, Inc.



I look on this as a long-term idea, which will gain more and more interest over the next few years. Any ideas or interest please marathoncontact me, John Rollo, at jrollo@comus-intl.com

Thanks to Our Wonderful Volunteers



Pictured is Donald Kauffman, who comes in each week to help with whatever we need done, from filling orders for EVERY1CARES bracelets to various mailings to our members. Thanks Donald!

HOW DO YOU LIKE OUR NEW LOOK?



Thanks to all our artists who contributed ideas for the new CARES logo.

The heart design chosen was created by Judy Bleyl of Istanbul, Turkey. The finished logo was created by Christon Holtzman of Studio 56 in New Mexico.

Christon also donated all design services for our new brochures.

The new logo will be appearing on all our printed materials and eventually on the CARES website, so we hope you like it! **CF**

The Basics of Genetic Testing

(Continued from page 5)

The cells obtained are taken to a laboratory and grown so that the DNA can ultimately be obtained and tested.

Amniocentesis is another technique that can be used to obtain a sample of fetal cells for genetic testing. This method is typically performed at 15-20 weeks from the last menstrual period. This procedure requires that a thin needle is passed through the abdomen, under ultrasound guidance, into the fluid filled sac that surrounds the fetus. A few tablespoons of the fluid are taken from the sac. This fluid contains cells from the baby that are then grown in a laboratory. As is the case with CVS, amniocentesis is not a risk-free procedure. With amniocentesis, there is a small risk of about 1 in 200 that the procedure will cause a miscarriage and loss of the pregnancy. The risks and benefits must be taken into consideration when considering prenatal testing. Your doctor and genetic counselor are good resources for additional information regarding these procedures.

If prenatal genetic testing for CAH is to be accurate and reliable, it is very important to think about genetic testing in advance. Due to the complexities in testing previously described, it is recommended that both parents as well as the affected child be tested prior to any prenatal testing of a subsequent child. If the fetus alone is tested first, it may not be possible for a lab to issue a final

report without requesting blood specimens from the parents and the previously affected child. This may delay the results by a few weeks while the lab compares the genetic pattern present in the fetus with that of the rest of the family.

Some laboratories recommend that another genetic test called maternal cell contamination studies be ordered at the same time as the prenatal test. Maternal cell contamination studies ensure that the fetal DNA being tested is in fact from the fetus and it is not contaminated with mom's DNA, thus ensuring an accurate analysis and result. For example, if the DNA being tested were actually the mother's DNA and not the baby's, then the result would be incorrectly reported that the baby was a carrier just like mom, when in fact the baby could be affected, not a carrier at all, or a carrier of dad's mutation.

Anyone considering genetic testing for CAH, for whatever reason, may benefit from meeting with a genetic counselor to discuss the various options available. You can find a genetic counselor near you at www.nsgc.org. A genetic counselor can also assist in coordinating your testing and can help explain and interpret your results. **CF**

Genetic Nondiscrimination Bill Stalled In Congress

The 108th Congress failed to pass legislation protecting against genetic discrimination. Although the Senate passed a bill (S.1053) 95-0 in October 2003, and President Bush supported such legislation, the House has not moved any such bill to the floor for consideration.

Without protective legislation in place, some patients fear having their conditions known to insurers, who might use it as a reason to deny coverage. As a result, these patients might forego testing that would be useful in making decisions related to their health and family planning. Other may seek anonymous genetic testing, according to a recent article in the San Jose *Mercury News*, which reported that some patients are being tested under assumed names, with their doctor's help.

To call your Senator or Representative to urge their support of this legislation, you can call the Capitol Switchboard at (202) 224-3121. Ask to be transferred to your Representative or Senator's office. If you do not know who your representative is, you can go to www.congress.org, enter your zip code, and find a list of your federally elected officials.

DID YOU KNOW?

All of our newsletter articles are archived on our website. If you miss an issue or misplace it, you can always find it at: www.caresfoundation.org/newsletter/ or click on **Newsletter... Archives** on the home page.

BE A CONSUMER ADVOCATE

Rules for the Road: A Handbook for Consumers in Leadership Roles is a compendium of wisdom from experienced consumers who have wrestled with the daily dilemmas that accompany leadership.

If you are thinking of becoming more involved in advocating for CARES or another human services organization, this new handbook from the Genetics Services Branch, Division of Services for Children with Special Health Care Needs, Maternal and Child Health Bureau, U.S. Health Resources and Services Administration may help.

For further information about the handbook, call (800) 396-5694.

BRACELET SALE RAISES MONEY, AWARENESS

CARES Foundation is extremely thankful to member Jami Abell Patterson for organizing our wildly popular bracelet sale fundraiser and awareness campaign.

So far, we have distributed a total of 4,646 EVERY1CARES bracelets, resulting in \$14,319 in sales.

Bracelets are still available at a cost of \$3 each (minimum order 5, plus shipping and handling.)



To order, call
(973) 227-3737 or order online at
www.caresfoundation.org

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



CARES FOUNDATION, Inc.
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Millburn, NJ 07041

**DO NOT DELAY
MEETING NOTICE**

Address Service Requested