Testicular Adrenal Rest Tumors in Congenital Adrenal Hyperplasia

Hedi L. Claahsen-van der Grinten
Paediatric Endocrinologist
Head of the Department of Paediatric Endocrinology, Radboud University Nijmegen Medical Centre, The Netherlands

Introduction
The classic (severe) form of congenital adrenal hyperplasia (CAH) is considered to be a life-threatening, chronic disease. In the last 20 years, improved diagnosis and treatment have allowed almost all children to reach adult life without serious complications. Therefore, long-term complications of CAH have become more important to address. It has been shown that some complications that appear in adulthood may have their origins in childhood and even infancy, so pediatricians have to be aware of signs of these complications.

One of the most serious long-term complications in adult men with the classic form of CAH is the presence of testicular adrenal rest tumors* (TART). These lesions were first described 40 years ago; since then, many case reports and small studies have described these lesions in greater detail. The tumors are always benign, are often present in both testes, and resemble adrenal tissue when investigated microscopically. The cells can produce typical adrenal hormones; therefore, they are called adrenal rest tumors.

In this article, I will give an overview about what is actually known about these lesions.

How can the tumors be detected?
The tumors are located within the testes rather than the surface and therefore can be easily missed by palpation. Ultrasound is a good and inexpensive method for detection of these tumors, and even small tumors of several millimeters can be detected. Some clinics use MRI, but the detection rate is no better than ultrasound, and it is much more expensive.

Are the tumors also found in childhood?
Yes, we can find these lesions already in childhood and even early infancy. In a recent study, we showed that there is an increase in number during puberty. Therefore, we recommend regular ultrasound in males with classic CAH starting at puberty.

[continued on page 3]

Table: Tumor Detection in CAH patients

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<tr>
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<tr>
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<td>17-43</td>
<td>16-40</td>
<td>5-27</td>
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<td>Palpation</td>
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<td>6/17 (35%)</td>
<td>1/9 (5%)</td>
<td>2/42 (5%)</td>
</tr>
<tr>
<td>Ultrasound</td>
<td>9/18 (50%)</td>
<td>16/17 (95%)</td>
<td>8/19 (42%)</td>
<td>12/42 (29%)</td>
</tr>
<tr>
<td>Bilateral</td>
<td>k.A</td>
<td>10/16 (63%)</td>
<td>5/8 (63%)</td>
<td>9/12 (75%)</td>
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* Tumor is the medical term for a lesion or mass that can be felt or detected by ultrasound or MRI.
A Message from the Executive Director

Dear Friends,

Welcome to the new edition of CARES Connections! We have been hard at work since our last edition hosting two regional educational conferences, a gala, three awareness walks, sponsoring medical education, supporting research, developing new tools for patients, addressing the issue of nomenclature (classification of CAH), working on EMS protocols for adrenal insufficiency, strengthening our support network, and developing a new website to better serve the needs of the CAH community.

Education

Patient and medical education continues to be a focus of CARES. Over the last year we have hosted two regional conferences aimed at educating patients and families, one at the CARES–designated “The Comprehensive Center for Congenital Adrenal Hyperplasia” at New York Presbyterian Weill Medical Center in New York and the other at Miami Children’s Hospital in Florida. We also sponsored a symposium on “Observations on Congenital Adrenal Hyperplasia” at New York Presbyterian Weill Medical Center in Chicago. With your support, we will continue to work to educate both patients and doctors to ensure that patients receive better care.

Awareness

Raising awareness about CAH is key to making progress in diagnosis, treatment and advancing research for CAH. On this front, we have increased the number of awareness walks we have hosted from one to three this year. Walks were held in New Jersey, California and Florida. These walks are a great opportunity to connect patients and families, further educate them about treatment and research, and to teach others about what it’s like to live with CAH. Similarly, our annual Gala brings together patients, families, medical professionals and supporters often for the first time.

We have witnessed firsthand the impact these events have had on patients and families. During one of the walks, a few families learned for the first time about the importance of having Solu-Cortef® on hand. It still amazes me that many doctors don’t understand that it is a life-saving tool in the event of an adrenal crisis. What doesn’t surprise me is hearing about the poor care so many patients continue to receive. Many of them think they are receiving good or adequate care until they talk to another patient or family who is indeed receiving appropriate care and realize their care is inferior at best. I am not exaggerating when I say that some of these events are “life changing” and urge you to participate in a future event.

Patient Resources

A significant amount of work has been done to improve the services offered to patients. In the last year we have strengthened our support network to include conference calls with various patient populations and started a new Facebook page to allow for the creation of secret pages in which patients can communicate with each other in a safe and secure environment. We have also completely overhauled our website. It is now easier to navigate, offering patients easier access to services and other resources such as “Ask the Expert,” CARES shop, and news on support network activities, and other events.

Our goal is to continually grow to serve you better. Let us know how we are doing and what you would like to see us do to make your life easier. Your feedback will help us help you!

CONTINUED FROM PAGE 2: ARE THESE TUMORS ALSO FOUND IN NON-CLASSIC CAH (NC CAH)?

There is discussion about this question. I have never found TART in NC CAH, but some authors describe them in the literature.

WHAT KIND OF TUMOR ARE THESE? TART consist of cells that resemble adrenal cells: they look like adrenal cells and can produce adrenal hormones. Therefore, it was thought that these tumors consist of aberrant adrenal cells. In recent studies we found that the tumors consist of more embryologic-like cells that may already be present during pregnancy.

WHY DO THESE CELLS GROW? In CAH a pituitary hormone called ACTH (adrenocorticotropic hormone) is elevated due to the lack of cortisol. ACTH stimulates the adrenal gland, leading to hyperplasia (growth of cells). It is thought that the elevated ACTH may also play a role in the development of TART. In early descriptions, patients with TART mostly had poor adrenal control and high ACTH levels. Nowadays, we know that TART can also develop in well-controlled patients, and there must be other factors besides ACTH that lead to tumor growth.

WHY DO THESE TUMORS DEVELOP? The etiology of TART is still unknown. We know that the adrenal gland and testicular cells have a common origin; in early fetal life, there are cells containing both adrenal and testicular features. During further development of the embryo, these cells differentiate and become testicular or adrenal cells, but both still in close relationship. During even further development, the testes descend to the scrotum. It can be speculated that some of these embryologic cells may persist within the testes. In healthy children, these cells diminish, but in the special situation of CAH, they are still in close relationship. During even further development, the testes descend to the scrotum and possible other factors, such as local hormonal production, may lead to infertility. Longstanding TART and obstruction can also lead to damage of the whole testis.

WHAT ARE THE TREATMENT OPTIONS OF TART? A clear treatment protocol does not yet exist since the origin and the factors that contribute to tumor growth are still unclear. Because of the relation between tumor growth and initial TART and obstruction, the first step of treatment is optimizing glucocorticoid therapy, for example by using prednisone or dexamethasone. This is used in growing children to optimize glucocorticoid therapy on both sides who want to achieve pregnancy. By using high dosages of dexamethasone, some doctors describe shrinkage of the tumors and successful pregnancy.

In our center we used tests–sparing surgery in 8 adult patients with longstanding bilateral TART and infertility in an attempt to improve treatment options. After end-stage damage of the testes in all patients during surgery and not surprisingly, testicular function did not improve. We concluded that with end-stage damage, surgery is not recommended unless there is pain or discomfort.

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To describe the progression of TART during life, we introduced a classification of TART in 5 stages (see Figure 1).

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ARE THERE OTHER POSSIBILITIES TO IMPROVE FERTILITY? As long as there are no clear treatment options, we offer cryopreservation to all young adults with bilateral TART.

WHAT ARE THE FUTURE DIRECTIONS OF OUR RESEARCH? We now focus on the origin of these lesions. We want to know the features and factors that contribute to tumor growth. With this knowledge we hope to develop treatment strategies to diminish or even prevent tumor growth.

IS TART A SPECIFIC FEATURE OF CAH? TART is mostly described in CAH, but there are other conditions with elevated ACTH that have been described very sporadically to have TART.

Do females also develop adrenal rests in their ovaries? Some female patients with adrenal rests in the ovaries (OART) are described in the literature, but the presence is rare. Some systematic studies exist. One study in our center searched for OART with ultrasound and MRI, but did not find these lesions.

Figure 1 Claasen-van der Grinten HL, et al. Best Practice and Research Clinical Endocrinology and Metabolism, 2009
and Metabolism with Professor Dennis Lo, who discovered New MI, Tong YK, Yuen T, Jiang PY, Pina... future research to validate this new expectation for families interested in participating in noninvasive prenatal diagnosis of CAH to... affected female fetuses will receive prenatal treatment with low-dose dexamethasone. On the other hand, in the noninvasive prenatal invasive method does not arrive in time to... procedures of amniocentesis carried out at 14... any damage to the normal cortisol circadian rhythm. Current available regimens for insulin resistance... treatment of our patients and their relatives, who make it possible for us to study CAH and develop new treatment approaches. References 1. Finkielstain GP, Kim MS, Sinaii N, Nishitani M, Van Ryzin C, Hill SC, Reynolds J... increased risk for the non-steroidal antidepressant... disease. Findings from this study are ongoing. We are very excited to report expansion of our research team. Dr. Rachel Morissette, Ph.D., completed her post-doctoral fellowship at the National Institute on Aging, National Institute of Health in Bethesda, Maryland, and joined our team in June 2013. Dr. Morissette is using biochemical and molecular methods to study biomarkers of disease. Ms. Teri McHugh, RN, joined our team this year. She has extensive nursing experience caring for children and adults with CAH in both inpatient and outpatient settings. She works closely with our research nurse on our team. We continue to recruit patients of all ages with CAH (both classic and nonclassic) to the Natural History Study. The current wait time for an appointment is 4 to 6 months. Participants will be seen as outpatients at the NIH Clinical Center in Bethesda, Maryland, for a full evaluation and up to four visits. Patients will have a full report sent to their private endocrinologist. All testing is free of charge and will include: • Genotyping • Hormonal evaluation including evaluation for insulin resistance • Bone age (growing children), ultrasound • Bone density (patients 8 years and older) For more information, please contact our research nurse Teri McHugh, RN, at codm-patient@mail.nih.gov. We gratefully acknowledge the enthusiastic participation of our patients and their relatives... through the sponsorship process. We are grateful to them for sharing their expertise with approximately 2,000 physicians from across the globe. We owe a debt of gratitude to Dr. Ellen Seely for chairing the session and to Dr. Richard Auchus for guiding us through the sponsorship process. None of this would be possible without the financial support of our community. Thank you! 2015 Conferences We are working on our 2015 conference schedule. Stay tuned to our website and your email for more information.
THE DOCTOR’S IN

Growth Hormone Therapy to Improve Adult Height in Patients with Congenital Adrenal Hyperplasia

Karen Lin Su, M.D.

Many patients with CAH do not reach an adult height within their mid-parental target height range. In a meta-analysis of 18 studies published between 1977 and 1997, the overall average adult height was 11.0 cm (4.3 inches) below mid-parental target height for females and 8.7 cm (3.4 inches) below target height for females [1]. Another meta-analysis of 36 studies published between 1977 and 2008 reported an overall average final height of 9.7 cm (3.8 inches) below the population mean [2].

Factors Affecting Adult Height in CAH

There are several factors contributing to subtotal adult height in CAH patients. CAH excess adrenal androgens result in rapid growth during childhood; however, the estrogens produced from these androgens result in premature fusion of the growth plates, ultimately compromising adult height. Long-term glucocorticoid treatment with or without LHRHa was equally effective in improving height in all CAH patients. However, for those patients who were headed for an adult height below the population mean, glucocorticoid therapy may be a viable option.

Studies with Growth Hormone Treatment to Improve Adult Height in CAH

For CAH patients who develop central precocious puberty, LHRHa, can effectively delay bone age by 2-4 years, unless estrogen therapy is used. Estrogen administration in combination with LHRHa appears to be effective for improving adult height in CAH patients. In the 2010 Endocrine Society CAH Clinical Practice Guideline, GH treatment was not recommended as standard treatment for CAH patients due to limited evidence; however, because not all patients require intervention to reach an adequate final adult height [3]. The recent GH treatment refer only to subjects with significantly compromised predicted adult heights due to advanced bone ages compared with mid-parental target values. With new screening now in place in all 50 states in the U.S., early diagnosis and treatment may help in preventing this in the future. GH outcome for all CAH patients, however, for those patients who were headed for an adult height below the population mean, glucocorticoid therapy may be a viable option.

References

ADVOCACY

CAH Nomenclature

Thank you to all who participated in our survey about current nomenclature (classifi cation) describing a subset of CAH. An umbrella term called Disorders of Sex Development was developed in 2006 to classify phenotypic sex at birth. There was a consensus group to classify conditions in which there is an issue during fetal development of the X or Y chromosome, ovaries, testes, or genitilia. The use of this term is being looked at critically by the CAH community along with other patient advocacy groups because it is not a complete description of CAH and may be perceived negatively by patients. The CARES Focus group discussions, parents and patients have expressed their concerns about the potential misconceptions that might arise from having CAH linked to this nomenclature. In order to get a better sense of patient and parent perception of this nomenclature, CARES Foundation conducted a survey of the CAH community regarding their thoughts on the issue. Close to 600 responses were received, and the results showed that the majority disliked the term and did not identify with it. The majority also felt that the term has a negative effect on the CAH community. The majority of responses have come from parents.

Emergency Medical Service Protocols for Adrenal Insufficiency

We extend our appreciation to everyone who works on EMT protocols for adrenal insufficiency on the grassroots level. While state-based protocols are being developed, we are meeting with members of Congress to develop legislation recommending the adoption of national protocols. Special thanks go to Congresswoman Leonard Lance for New Jersey for his support.

If you have experienced EMIS or EMT personnel lacking knowledge in the diagnosis of CAH, please be sure to give them this resource. It contains information about CAH, the symptoms, signs, diagnosis, treatment options, and references. It is also available in Spanish.

How to Diagnose CAH

A new blood test, drawn in the morning prior to any dietary intake at adrenal insufficiency (17-Hydroxyprogesterone (17OHP)), anandrostenedione and testosterone), may be sufficient to make the diagnosis of CAH, as long as the subject is not preg nant, is not receiving 17OHP, or taking with CAH. The ACTH stimulation test is the gold standard to make the diagnosis if the lab values from the single blood test are equivocal. A dose of ACTH (adrenocorticotropic hormone) is given intravenously. Blood samples are then taken before the medication is given and again an hour later.

Normal response to ACTH stimulation is to release cortisol into the bloodstream. In CAH blood samples taken after the dose of ACTH show large amounts of 17-OHP, the “raw material” from which cortisol is normally made. The results of the ACTH stimulating test are plotted on a “Nomogram,” figure at see page 245-291, to determine whether the values indicate a diagnosis of NCAH or classic CAH. Nomogram for comparing 17-OHP lev els before and 60 minutes after a 0.25 mg bolus of cosyntropin in subjects with or without 21-hydroxylase de fi ciency. Note that the values for normals and heterozygotes (carriers) overlap.

DNA testing and certain other hormone tests are also meeting with members of Congress to develop legislation recommending the adoption of national protocols. More information about this is available at the website of the CARES (Congenital Adrenal Hyperplasia Network) and can be found in the references.

Table 1: Longer-acting synthetic glucocorticoids (prednisone, prednisolone, methylprednisolone, and desamethasone) are more growth-suppressive than cortisone and hydrocortisone even at equivalent doses.

<table>
<thead>
<tr>
<th>Medication</th>
<th>Growth-Suppressing Effect Relative to Hydrocortisone</th>
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<tr>
<td>Cortisone acetate</td>
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<tr>
<td>Hydrocortisone</td>
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<tr>
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<td>Methylprednisolone</td>
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<td>Dexamethasone</td>
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<td>Prednisolone</td>
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</table>

Table 1: Longer-acting synthetic glucocorticoids (prednisone, prednisolone, methylprednisolone, and desamethasone) are more growth-suppressive than cortisone and hydrocortisone even at equivalent doses.
and treatment of adrenal crisis, please email your story to Karen at karen@caresfoundation.org. We are sharing stories from those with CAH and other forms of adrenal insufficiency with legislators to demonstrate the need for national protocols.

Updates
• Alabama has an adrenal insufficiency protocol. EMS is allowed to administer patient-carried medications.
• Montana has adopted patient-carried medication protocols.
• New Jersey passed full protocols. Individuals must visit their local EMS to request emergency medical care/

• Comprehensive Care Centers of the protocols found at http://www.caresfoundation.org.

• Important medication with a completed Individualized Care Plan.
• Advanced Life Support (ALS) Individuals must visit their local EMS to request emergency medical care.

• Family activities. And now, we have created the CAH Champions page with secret groups which offer a closed and safe environment in which to have conversations with others with similar experiences. Our secret pages include:

• 3 Beta Women
• 17 Hydroxylase
• Classic CAH Women Support Group
• Men with CAH
• NCAH Women Support Group

• Newborn Support Group
• Parents of Kids with CAH
• Parents of Teens/YA
• Support Group
• SWCAH Women
• Teens/Young Adults Support Group

• Also we have pages for those who live in the following states: Alabama, Arizona, Arkansas, California, Colorado, Connecticut, Florida, Georgia, Idaho, Illinois, Indiana, Kansas, Kentucky, Louisiana, Massachusetts, Minnesota, Montana, Nebraska, Nevada, New Jersey, New York, Ohio, Oklahoma, Pennsylvania, Tennessee, Texas, Utah, Virginia, Washington, West Virginia, Wisconsin.

Facebook
Our presence on Facebook continues to expand. The CARES Facebook page keeps our community connected on the latest events and news about CAH and CARES activities. And now, we have created the CAH Champions page with secret groups which offer a closed and safe environment in which to have conversations with others with similar experiences. Our secret pages include:

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Moving On
We are very grateful to Stephanie Gruber for her many years as a dedicated support group leader in Connecticut. Thank you, Stephanie! We greatly appreciate all those who have served as support group leaders and have had to step down. You have made an important difference!

Telephone Meetings
We continue to hold our Newborn Support Group meetings on the second Thursday of the month at 5pm EST. Every week our leaders answer questions from curious and sometimes anxious parents about caring for an infant with CAH. We have participants from across the U.S. and as far away as India and Estonia.

Our meetings have expanded to include Women with CAH and Parents of Children, Teens and Young Adults. These calls will be alternating every other month, usually on the second Wednesday at 8:30-10:00pm EST. Please check the calendar section of our website for meeting information, or contact Karen at karen@caresfoundation.org.

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For more information on these pages, contact Karen at karen@caresfoundation.org.

Events
• Massachusetts
On September 3-4, Al DeObion, MA support group leader and CARES trustee, gathered families from the Boston area. One of our CAH families, the Gleasono’s, hosted the event at their athletic facility, Athletic Revolution. The five families who attended, each with visits with CAH, had a great time utilizing the facility and enjoying each other’s company.

• Ohio
Sara Teed, one of our Ohio leaders, once again held an outing for CAH families at the Cincinnati Zoo on September 20. Ohio, Kentucky and Indiana families were invited. The event was great fun for the children and parents!

• Pennsylvania
Debbie Cullen, our PA, MD and DE support group leader, held her 3rd annual bowling fundraiser. Pennsylvania friends and families enjoyed an afternoon of bowling and tricky tray items.

A huge thank you to all of our support group leaders who respond to those wanting to connect, hold meetings or events, dial-in to specialized support group calls, moderate secret Facebook pages and more. Your work is invaluable and the CARES community is so fortunate to have your help!

In-person Support Group Meeting
Mark your calendar and plan to join us for an in-person Support Group meeting Saturday, December 6th, 11 am.

The meeting will be held at the Comprehensive Center for Congenital Adrenal Hyperplasia at New York Presbyterian/Weill Cornell Medical Center (525 East 68th Street, New York, NY). Please RSVP to karen@caresfoundation.org.

Doctors from the Center will be on hand to answer your questions. It’s also a great opportunity to meet other patients and families. You do not have to be a patient at the Center to attend.

Year-Round Camp Opportunities
We continue to connect children with medically safe summer camp opportunities in different parts of the U.S. Most of these programs are free and some assist with transportation. This year’s camps welcomed our CAH kids in Arizona, California, Kentucky, Upstate New York, North Carolina, and Oklahoma and Washington. You don’t have to live near the camp to attend. Children have traveled from Colorado, New Mexico and Utah to the New York camp.

This past spring, The Painted Turtle Camp in Lake Hughes, California, hosted a Special Diagnosis Family Weekend, March 14-16. Families enjoyed boating and fishing, arts and crafts, woodshop, music, and much more.

D. Mimi Kim, pediatric endocrinologist from CHLA, was one of the volunteer doctors that weekend.

Center for Courageous Kids in Scottsville, Kentucky, held a Spring Weekend for CAH, April 11-13. Families came from far and wide to be with other CAH families in a relaxed and welcoming atmosphere. As one mom said, “How very much we enjoyed the Courageous Kids Camp in Kentucky. This facility was top-notch and very organized so was nice to attend such a well-organized event. It was a very nice retreat for our entire family.”

SUCCEED Clinic held their annual camp at Schaeffer Farm in Noble, Oklahoma, on May 3-4. Patients and their siblings experienced a fun-filled weekend of meeting new people, outdoor cooking, archery, crafts, campfires, songs, games, and more. Thank you, Schaeffer, Oklahoma support group leader and SUCCEED Clinic pediatric nurse, coordinated the event.

Staying Connected Online – Our New Website
This fall, we unveiled our new website at www.caresfoundation.org. For more than 10 years, the site has been an invaluable tool for both our patient and professional communities, but it was time for a much needed makeover. With rapid changes in how we receive and share information, the site was redesigned to better meet the needs of the community by making it more functional, intuitive, and aesthetically pleasing.

The new site has a clean, fresh look with easy to follow navigation. Other features include the ability for visitors to comment directly on articles, allowing for greater engagement within the community, sharing thoughts and advice, as well as the ability to quickly and easily share information with their personal and professional contacts via social media buttons (Facebook, Twitter, etc). We would love to know what you think about the new site. Please check it out and send your feedback to Dina@caresfoundation.org.
Once again Double H Ranch in Lake Luzerne, New York, held a CAH session during their summer program. Wendy Thornley, our Criminal support group leader, was one of the volunteer nurses that week. “Sebastian had such an amazing time at camp,” sum up how most campers felt.

Double H Ranch offers Winter Adaptive Sports Weekends where families come to learn how to ski, snowboard, snow tube, ice fish, play games, do crafts and more. The application deadline for Winter 2015 is December 1. “We had so much fun. It was such a great oppor-
tuation – no laundry, no cooking, plus skiing. I can’t thank you enough for bringing Double H Ranch to the Cares connections.”

Look for emails announcing camp opportu-
nities or contact Karen at karen@caresfoun-
dation.org for more information.

International Update

Australia and Beyond

Congratulations to CLAN (Caring & Living As Neighbours) for the development of the Leave No Child Behind booklet launched at the recent UNPDF/NGO Conference in NYC! CLAN is an Australian-based, not-for-

profit, non-governmental organization (NGO), approved by AusAID for Overseas Aid Gifted Stewardship (OAGDS) and the Australian Taxation Office for Tax Deductibility Status. CLAN is dedicated to the dream that all children living with chronic health conditions in resource-poor countries of the world will enjoy a quality of life on par with that of their neighbors’ children in wealthier nations.

France

Two organizations in France are currently dedicated to CAH: Association Surrénales and IFCAH. The former helps to maintain relationships between patients and physicians, the latter on funding medical research.

Association Surrénales organizes a yearly international seminar dedicated to develop-
ing health professionals’ knowledge about adrenal diseases and updating them on current research projects. In May 2014, the seminar was held in Paris and brought together 94 participants from western European countries.

For more information go to: www.surre-
nales.com.

IFCAH (International Foundation for CAH) was established in 2010. Each year, IFCAH awards 350,000€ to the most interesting pro-
jects as selected by its Scientific Committee in collaboration with the European Society for Pediatric Endocrinology. Projects funded in 2013 include:

- Project 1: Characterization of precursors of Testicular Adrenal Rest Tumors (TARTs) found in CAH patients. Investigator: Dr. A. Swan, London, UK. Funding: 115,000€.
- Project 2: CAH beyond the age of 40-years: What challenges do we face and what do we need to monitor? Investigator: Dr. N. Reisch, Munich, Germany. Funding: 85,000€.
- Project 3: Development of non-invasive prenatal diagnosis of CAH using cell-
free fetal DNA in the maternal circulation. Investigator: Dr. L. Chitty, London, UK. Funding: 50,000€.
- Project 4: Cell therapy for the treatment of congenital adrenal hyperplasia (CAH) using an implantable medical device. Investigator: Dr. S. Bornstein, Dresden, Germany. Funding: 73,000€.

For more information go to: www.ifcah.org.

Netherlands

The Dutch Adrenal Society NVACP was founded in 1988 and has at present just over 1,300 members, representing the following adrenal diseases/disorders:

- Addison primary (incl. ex. Cushining without adrenals): 665 (50.9%)
- Addison secondary: 106 (8.1%)
- Addison tertiary: (using corticosteroids for other diseases): 27 (2.1%)
- Cushing: 249 (19.0%)
- CAH/AGS: 196 (15.0%)
- Conn syndrome: 47 (3.6%)
- Miscellaneous: 19 (1.5%)

Our mission is to support and educate our members, their families, school employee-
es, employers, and general practitioners, in an effort to help improve quality of life after diagnosis.

Often a diagnosis comes too late, result-
ing in significant complications and poor quality of life. However, there is one excep-
tion! Nowadays CAH is included in newborn screening tests. All babies in the Netherlands receive a screening test also called the “heel prick test,” in which blood is drawn in the first ten days of life, irrespective of where they are born – at home or in the clinic.

For our young members, we have recently developed some animations to help them better understand their disease and to help them explain it to family members, at school, etc. Adults also use the animations to explain their disease. You can view these animations here: www.youtube.com/user/adrenalchannel.

At this moment we are also working together with several European patient organizations on a standardized emergency card, which will include crucial minimal medical information that will be recognizable to ambulance personnel. We will keep you informed on our progress.

Starting January 1st, 2015, all ambulance personnel in the Netherlands will follow the same treatment procedures due to the adop-
tion of an ’Adrenal Insufficiency Ambulance Protocol’ in the event of adrenal crisis. If someone suffers from adrenal insufficiency, the ambulance will arrive and contact the spe-
cialist/endocrinologist/hospital. If necessary, they will also contact the appropriate medication center (Solu-Cortef) and transport the patient to the hospital. Currently, patients are taken to hospital with the secondary effects of the adrenal crisis have already created significant harm. This is a huge improvement in treatment.

In the future, we would like to have a global meeting on CAH (similar to the meetings held this year in Oslo for Addison’s and in Zurich for Cushing) where those involved with CAH work across the globe could meet to discuss mutual experiences and coordinate activities for the best patient care “across borders.”

One of our recent successes was the emergency kit. We are now working on the improved safety belt clip and the emergency card. We will continue to provide updates on these initiatives.

We have lots more information on our website. Feel free to contact us or consult our website (www.nvacz.nl) or our test-site (www.addisoncrisis.info). This website is “under construction”; we need your comments, advice and criticism.

A PERSONAL STORY

Wrapped in Miracles

by Michèlle Janine Bacus

Every parent has a story. This is one … much I know.

We celebrate their miraculously from preconception dreams to postnatal bliss and beyond. My son, Maris Paul, is wrapped in tiny little miracles that have defined his life thus far. As I reflect, I celebrate his milestones, seemingly equal as important. I am reminded of just how lucky we are to have him with us today.

I like to say that I waited a lifetime for Maris plus 296 days, 5 hours, and 19 minutes. Maris took his time to meet the world. I was a week past my due date and finally my OB/GYN made the decision to induce. We checked into the hospital on Monday, May 7th, 2012, and the induction process began. Seventeen hours later, with failure to progress, my body began to go into shock. The end result was an emergency C-section and a large loss of blood for me. I was given a transfusion, and I was not able to see my angelic 8 pound, 9 ounce baby boy until the next day. He was perfect, I, however, was far from it. I was an extreme case of the C-section, loss of blood, and trauma to my body, the decision was made to keep me in the hospital for five days. I always think these facts are important as I tell Maris’s story. I feel that it is necessary to hear that I had an extended stay in the hospital because that means Maris stayed with me. He was under hospital and pediatric supervision for five days after his birth. When we were released that Saturday, he and I were given a seemingly clean bill of health. Even with those five days in the hospital, Maris was not on anyone’s radar.

We went home and began the process of settling in as a family. My sister flew in from New York to help, and my mother would follow. The first few days were a blur. The following Wednesday, Maris was eight days old and that is when I noticed things begin to change. Our morning that Wednesday started fine. I had errands to run. It was a beautiful spring day, and with Maris in tow, I went about my day. By late afternoon, I no-
ticed something started to shift in my son. He became irritable. He was only eight days old, but he hadn’t shown this behavior before. It was subtle, but it still bothered me. Then, as the evening progressed, he began to cry incessantly and was inconsolable. I tried to feed him and he wouldn’t take to any of the variety of bottles I had for him. Finally, as the crying continued and evening approached, I relented and took him on a car ride. It seemed to help a bit. He slept, but he whimpered every now and again throughout the ride. He never went into a deep good sleep that a parent hopes for when you take that magical car ride. At the end of the evening, I remember looking at my sister and asking her, “do you think he is sick?” She responded, “I think Cushing is an Aiguil” I thought. “I’ll talk to him today; you need to just stay home and take it easy.” I followed my sister’s advice and had a lazy day the next day. I will admit, he seemed better. He slept well throughout the day and didn’t seem as irritable. He still wasn’t eat-
ing well and that was concerning to me. At the hospital, he really enjoyed his 2 ounces at a time. As the evening approached, the crying began again. I wondered if perhaps I was just going to have a colicky baby. He was struggling to eat, seemed lethargic and was just inconsolable. In my heart, I just felt something wasn’t right – but what? It wasn’t anything blaring. It was subtle things: lethargic, irritable, diminished appetite, inconsolable.

I had a nagging feeling that something was not right … I had no idea what was in store for us.

I can only imagine what it was like for me crying … any of this could be attributes of any typical newborn. We got through the night, I woke up Friday, March was now 10 days old, and I had a nagging feeling that something was not right. I recall it was a Friday because I thought to myself, I can’t want to go through the weekend worrying. I thought, let me call the doctor while she has normal appointment hours. I called the pediatrician’s office. I apologized in advance, saying, “perhaps I am just being a paranoid first-time mom, but something doesn’t seem right. I was transferred to a nurse who thank-

fully said, “You know, it is probably best you just bring him in.” Two hours later, my sister and I were heading into the pediatrician’s of-

fice. I had plans with my girlfriend for lunch, I called her and said we may be a bit late, had no idea what was in store for us. When I got to the office, the pediatrician asked me something. She began to examine him and then put him on the scale. He had lost enough weight since being discharged
I am not even sure in what order it all happened. I kept hearing sodium, potassium, 17 OH-Progesterone. None of this was making sense. I was paralyzed by fear of what this all meant and praying that he would be okay. He was hooked up to machines and blood was drawn. EKGs were coming back irregular and he could barely lift his head. My little rosy-cheeked child seemed pale and gray. By the end of the night, only one thing was clear, that my little boy would not be coming home with me. He was admitted to the NICU and I realized that little miracles happen every day. Maris is proof of those miracles, and I realize that much hard work, advocacy, and education goes into making those miracles happen.

hospital staff and doctors hovered around him, I was frightened and confused. The next morning shed some light, and it also revealed that our lives would now be forever changed with blood tests, medicals, monitoring and doctors. Yet, I felt so incredibly blessed. The miracles surrounding my son appeared daily, and we woke up to new life, real life, a lifetime of questions regarding my son being with us and recovering was truly some-
Talk It or Walk It

This year’s CAREing Hearts Walks have been a huge success! The New Jersey walk had its 3rd Annual CAREing Hearts Walk on September 28th at Wild Duck Pond Park in Ridgewood, California and Virginia en-
teried the mix with their first ever CAREing Hearts Walk held on October 12th in Los Angeles and November 10th in Chicago. Hundreds of patients, families, friends and medical professionals came together to share experiences, learn about CAH, and network with each other. It was a rewarding experience to see families and supporters come together to enjoy a day of fun filled activities and education.

Special thanks to Chad Lapp, Karen Bogaard, Cynthia Winne and Laurie Zehner for hosting the walks. We are also grateful to all the walkers, sponsors and supporters who helped make a difference in the lives of CAH patients. Thank you to the Division of Pediatric Endocrinology at Children’s Hospital Los Angeles for providing injection training!

If you participated in one of the walks this year, let us know what you thought of the experience. If you didn’t have the opportunity to join us, we hope you will join us next year.

Walking Together

Here’s what one mom had to say about par-
ticipating in a CAREing Hearts walk: “…my
mothers find solace in each other’s com-
pany. We share stories and strategies; we
walk together, commiserate over copic, and
collectively mourn loss of sleep. It makes us
feel like we’re not alone, like maybe we’re not
messing this whole parenting thing up and
that at the end of the day, at the very least,
someone else gets it. So, when you’re the
mother of a child with a rare, life threatening medical condition that few can relate to, it
can leave you feeling isolated on top of the
emotional roller coaster you’re already on.
Family and friends may not be able to understand the day to day struggles we
face, but they love and support us with a
vengence. They put their support into action at that annual Walk for a Cure (CAYA’s
CAREing Hearts Walk for CAH Awareness) in honor of our daughter, raising money for our cause. On this one day every year, I don’t feel quite
so alone in this fight as I walk with families
who know our struggle intimately as they
battle it everyday with their own child. And,
when I glanced around the crowd walking with us and when the donations kept rolling
in, I felt such love and gratitude that I know
will see me through those bleak days. My
daughter may not understand it just yet, but
imagine how incredibly loved she will feel
when she looks around and sees all those
people who came out just for her.

Wouldn’t it be awesome for every man,
woman and child with CAH to feel such sup-
pport? We may not be able to cure anyone just
yet, but wouldn’t it be great to let people know they are not alone, that someone does care?
Just knowing you came out in support of them
would be so rewarding for them and for us.
Imagine instilling this value in our children
now—what a compassionate world this
world to us, and we thank you!”

Chad Lapp Runs 24 Hour Marathon for 4th year in support of Andrew

We want to thank our very own Chad Lapp, Vice President of the Board of Trustees, for an amazing display of endurance and determi-
nation in completing another North Coast 24 Hour Run in Cleveland. This year marked his fourth run in support of five-year-old Andrew who served as his motivation as he ran a
grueling 107.35 miles. Chad finished second in
his age group and 13th overall while raising awareness and funds for Andrew who has
cell-wasting CAH and others with CAH.

Over the past four years, Chad has raised
more than $40,000 for CARES. Thank you
Chad for your dedication and commitment!

Talk It!

Our Talk It campaign has come to a close with great success. The Talk It campaign promotes
an opportunity for those unable to attend our
CAREing Hearts Walks or unable to host their
own event, a simple approach to supporting
CARES and raising funds for CAH.

Talk It is a virtual fundraising campaign that
allows participants to set up their very own web page to fundraise. From the comfort of their own home, they can make
personal donations, receive donations from
friends and peers, and track their team’s progress as donations grow. It’s also an op-
portunity for an enormous awareness about CAH.

Each year, participants in our Talk It or Walk It
campaign are entered in a drawing for one of
three prizes. For each $600 raised, they get
one entry. Congratulations to our winners!

This year’s Talk It or Walk It campaign raised
more than $75,000. Thank you for your support! We look forward to having you
join us again next year.

Family Fundraising

No event is too small when it comes to sup-
porting CARES! Oftentimes small, family
hosted fundraisers are a great way to raise
awareness of CAH while also providing a fun
and enjoyable experience for our CAH com-
munity. We report some examples of family
hosted fundraisers held this year.

Party with a Purpose — California

Thanks to Sue Shryer for hosting Party with a
Purpose, bringing together CAH families and
supporters in Southern California for a night of
music, dancing and raffles. The event raised
more than $10,000 for CARES Foundation.

Girls Night Out – New Jersey

Our Girls Night Out event, hosted by Sari
Lee, helped raise more than $2,000 as sup-
porters met at CAH DO Fitness Club in Short
Hills for a night of food, fun and shopping.

Family Fun Day - Illinois

The Quarter Auction, Buffalo Wild Wings and
Family Fun Day fundraisers in Galesburg
planned by Meo Kyser, raised over $1,000.

A Big Thank You

We also want to extend our gratitude to Fred
and Whitney Thomey for raising funds for CARES in celebration of Fred’s birthday, and
to Michelle Deearie for hosting a Christmas Stays at Home CARES. Thank you for
bringing patients and families together!

If you would like to plan an event to benefit
CARES, contact Dina at Dina@caresfounda-
tion.org. Stay connected via our website and
email to receive information on how to connect with individuals who are hosting events.

AmazonSmile

Help put more smiles on the faces of CAH
patients by using AmazonSmile for your
holiday shopping! AmazonSmile is a simple and automatic way for you to support CARES Foundation
every time you shop at no cost to you! When
you shop at smile.amazon.com, you’ll find
the exact same low prices, vast selection
and convenient shopping experience as you
do on Amazon.com. The only difference is
that Amazon will donate a portion (0.5%) of your purchase price to CARES.

How to support CARES with AmazonSmile

On your first visit to AmazonSmile (www.
smile.amazon.com), go to the bottom of the page, and in the box under “pick your
own organization,” type in Congenital
Adrenal Hyperplasia Research, Education
and Support. Once you select CARES
Foundation, your account will be set to
CARES for all future purchases. If you
already shop with Amazon, do not need to
create a new account. Your account informa-
tion is the same for AmazonSmile.

So go ahead and put a gift under the tree and
a smile on many faces. Happy shopping!

Good Search

Raise money for CARES Foundation just by
searching the web and shopping online!
Here’s a great way to raise money for CARES! Use Yahoo! powered GoodSearch.com as your default search engine and it will
donate a percentage of your voluntary contribution any time you do a search!

In addition to their online shopping mall,
GoodShop.com —where you can choose from more than 100,000 online retailers and
a percentage of your purchases will go to
the charity of your choice.

Visit the chart below for an example of how much we can earn. The sky’s the limit! Searching has never been better, it’s free and a great way to
support. Once you select CARES
Foundation, your account will be set to
CARES for all future purchases.

The Deena Jo Heidi-Diesslin Foundation

CAREing Hearts Society

We are pleased to introduce CARES
Foundation to the CAREing Hearts Soci-
ey launched in 2013 to recognize the outstanding generosity of our donors without whom CARES would not exist.

The impact this group of donors makes on CARES is significant. The development of Comprehensive Care Centers, research, and education, along with our other initiatives and services, is directly related to support we have received over time from these individuals.

We thank our Society members for playing an integral role in the success of CARES.

Pioneer

Jessica & Matthew Upchurch

Visionary

Kelly & Adam Leight

Merith & Daniel Taylor

Vicotrina Charitable Trust

Anonymous

Champion

Cindy & Alan Macy

Marc & Marjorie McDonald

Elsie & Susan Wise

Drs. Karen & Edwin Stu

The Deena Jo Heidi-Diesslin Foundation

Advocate

Mitz & Bill Davis

Alexandra Dubas

Doug Zehner & Katherine Fawler

Rhonda & Gregory Kraft

Chad & Sandra Lapp

Richard & Deborah Pendino

Sandra & Mack Rose

Dr. Diane Snyder & Albert Stener

Anonymous

Friend

Rodrigu Quintanilla & Vivian Allman

Susan & Carl Aycock

Sondra & Michael Brunone

Pamela Chiles

Jeffery & Leah Krontahl

Alex & Sari Lee

Art & Nancy Lee

Anna Pinto

Hope & James Raphalian

Dr. Richard Rink

Dr. Peter Schlegel

Kenneth & Vicki Upchurch

Matt & Barbara Wilson

Stephanie Rose

Anonymous
New Additions to the Board of Trustees

We want to extend a warm welcome to our new members of CARES Board of Trustees – Kathryn Ashenfelter, Karen Bogaard, Carlos Da Silva, Anthony Fine and Cynthia Winze.

Kathryn Ashenfelter
Kathryn Ashenfelter of Denver, Colorado, comes to CARES with 30 years of finance and progressive leadership experience. She serves as a Senior Trust Officer for Swedish Medical Center, a 368-bed acute care, full-service hospital in Englewood, Colorado, which is part of the HealthONE hospital system based in Denver. In addition to CARES, she is active in a number of other organizations including the Shirley Foundation of Colorado, having served as President and Treasurer, and is the current Treasurer of Doctors Care of Colorado In the Colorado Restaurant Society. Ms. Ashenfelter received a Bachelor of Science Degree in Accounting from the University of Nebraska and is a Certified Public Accountant.

Karen Bogaard
Karen Bogaard, a licensed physical therapist since 1980, Karen Bogaard provides Home Health services for patients who are homebound. She specializes in geriatrics and joint replacement rehabilitation services.

Anthony Fine
Anthony Fine is Head of Finance at England, Anthony Finance is Head of Finance and Planning Systems for Thomson Reuters Group, prior to joining Thomson Reuters, he was President of Price Waterhouse Coopers in Sydney and London. A graduate of Lehigh University, Mr. Da Silva lives in California with his wife Sonya and their two sons – one of whom has CAH.

Carlos Da Silva
Carlos Da Silva As President of LB Industries, Inc. of Pennsylvania, Carlos Da Silva’s responsibilities include general administration with special emphasis on finance, bonding and insurance, as well as day-to-day management and supervision of construction operations. Mr. Da Silva founded LB Industries in 2004. The company established itself as a premier niche contractor for water and wastewater treatment authorities in Central New Jersey, Eastern and Central Pennsylvania.

Prior to LB Industries, Mr. Da Silva was Director of Financial Operations for EIC Associates, Inc., a diversified heavy civil construction firm with annual sales in excess of $50 million. He also served as a Senior Consultant at Arthur Andersen LLP, where he began his professional career.

A graduate of Lehigh University, Mr. Da Silva lives in California with his wife Sonya and their two sons – one of whom has CAH.

Cynthia Winze
Cynthia Winze, President and Owner of PSF Manufacturing Company, is a leader in the field of interior plantscapes. Cynthia and her team of artisans passionately create signature silk florals, accents, and custom plantscapes that captivate the senses and elevate the environments in which they reside. For the past 10 years, Cynthia’s driving passion is to make the lives of others better by making their surroundings beautiful. That passion has transformed hundreds of successful and fulfilling partnerships, collaborations, and projects – partners include furniture stores, restaurants, corporate office spaces, historic architectural landmarks, residential developments, hospitality corporations, as well as collaborations for special events.

Prior to starting her own business, Cynthia worked as a regional asset manager for one of the nation’s largest and most respected real estate development and investment firms. For 18 years, Cynthia managed a 15 million square feet of class A, multi-use, industrial and retail estate for Trammel Crow and Cushman & Wakefield. Cynthia also worked as a successful consultant for international clients, managing and expanding their domestic portfolio of commercial and residential buildings to 250 million dollars in investments.

Cynthia graduated from the University of Southern California with a degree in business. She is a cancer survivor and married with two children. She and her husband are also family of a beautiful five-year-old granddaughter who was diagnosed with CAH at birth. Cynthia resides in Southern California with her family.

New Members of Our Team

Chris Gerena, Intern
We are pleased to announce the appointment of Chris Gerena to our staff. As an intern, Chris handles electronic communications for the CARES community, as well as maintaining web tools for fundraising events.

Chris Gerena completed an internship in Pediatrics at the Robert Wood Johnson Medical Center in New Brunswick, NJ and residency in Pediatrics at the Tufts New England Medical Center in Boston, MA. She went on to complete a fellowship in Pediatric Endocrinology and Diabetes at the Mount Sinai School of Medicine in New York, NY. Her research and much of her academic work during her fellowship was on the subject of Congenital Adrenal Hyperplasia, conducted under the guidance of Dr. Maria New. She has published on the subject of CAH and other adrenal disorders in a number of peer-reviewed journals and textbooks.

Dr. Lekarev is excited to be involved with the CARES Foundation and its families and to be an integral part of The Comprehensive Children’s Care Center at Well-Cornell.

Dr. Oksana Lekarev has been appointed to the CARES Foundation’s Scientific and Medical Advisory Board. Dr. Lekarev is an Assistant Professor of Pediatrics in the Division of Pediatric Endocrinology and Metabolism at Weill Cornell Medical College. She is also the Assistant Medical Director of The Comprehensive Care Center for Congenital Adrenal Hyperplasia at New York Presbyterian Hospital at Well-Cornell.

Dr. Lekarev graduated from the University of Michigan. Prior to joining the University of Washington and Downtown Saint Paul, Minnesota. She plans to attend law school and become a JAG officer.

CARES in 2009, Ms. Fowler has served as Vice President and Chair of the Fund Development Committee. Her responsibilities for her presidency includes expanding access to clinical information and expertise for all CAH patient and families. She is also leading the effort to develop a long-term strategic plan for CARES.

The mother of twins, she resides in New Jersey with her husband.

New President of the Pediatric Endocrine Society

Dr. Mitchell Geffner
Dr. Mitchell Geffner, Division Chief of the Center for Endocrinology, Diabetes, and Metabolism at Children’s Hospital Los Angeles, and Professor of Pediatrics at the Keck School of Medicine of the University of Southern California, was recently elected President of the Pediatric Endocrine Society. The mission of PES is to provide advocates and educators for children and adolescents with endocrine disorders. With more than 1,300 members representing the multiple discipline of Pediatric Endocrinology, PES members are dedicated to research and treatment of children with endocrine disorders in such areas as growth, bone, thyroid, diabetes, obesity, growth, pituitary and adrenal. The Society works to promote the continuing education of members, its patients, their families and the general public.

Dr. Geffner’s goals for his presidency include improving PES’s relationship with other organizations, gaining an increased focus on the Endocrine Society, ESPE, and PENS (among other groups); maximizing and shaping numbers through heightened recruitment and retention efforts; increasing corporate support with attention to transitioning, moving forward on previously established initiatives related to journal affiliation and international outreach in Central America and the Caribbean; and creating a visiting fellowship program. We wish him much success in his new role!

Prestigious Honor

Dr. Maria New
Dr. Maria New, Director of the Adrenal Steroid Disorders Division at the Mount Sinai School of Medicine in New York, was awarded the prestegious Fellowship IPSIE 2014 Endocrine Regulations Prize during The 10th International Congress of Endocrinology jointly with The Endocrine Society 96th Annual Meeting and Expo in Chicago. She presented an award lecture titled “Non-Invasive Genital Diagnosis of Congenital Adrenal Hyperplasia” at the Chicago meeting on June 23rd. This prize is awarded annually to a researcher or a physician who has carried out work essential to a better understanding of the role of neuroendocrine interactions in regulating the body’s major metabolic functions and who has made a significant body of work rather than a single discovery. Dr. New joins a distinguished list of previous honorees, many of whom are Nobel Prize laureates. Congratulations on the well-deserved honor!

Comprehensive Care Centers for CAH

The Comprehensive Center for Congenital Adrenal Hyperplasia at New York Presbyterian/Weill Cornell Medical College

Individuals with CAH may have a variety of medical needs that require access to a team of health care providers and multiple subspecialists. Furthermore, not all physicians are familiar with CAH and its management. For these reasons, having a “medical home” that provides a comprehensive approach to care for CAH patients and their families. In September 2009, CARES hosted a meeting of medical professionals that included representatives from multiple medical specialties, as well as patients and family caregivers, to develop guidelines for Comprehensive Care Centers (CCC) for CAH. A summary of those guidelines was published in the International Journal of Pediatric Endocrinology. Based on those guidelines, CARES solicited applications for a pilot CCC at WMC in 2013. The New York-Presbyterian/Weill Cornell Medical College (WMC) was awarded the title of the first CARES-designated CCC for CAH in the United States. The CCC at WMC brings together health care professionals with multidisciplinary expertise and with expertise in CAH. This team is able to provide expert care at all phases of a life of an individual with CAH, from newborn through childhood, adolescence and adult life. The Center includes a medical and surgical
problems with high blood pressure, has obesity issues, repeatedly has adrenal crises, or anything else that bothers you and it is not getting solved, it is time to find a CAH expert. What does that mean? It means a doctor who aids many kids with CAH – not three … not even ten …more than that … and usually they are found in more metropolitan areas. Ask CARES to help you find one. Luckily, due to technology, bloodwork and files can be transferred to an expert and they can offer their opinions about dosing methods or testing that may need to be done. Ideally, if you can make a one time trip to visit an expert, that would be great, but a phone consult with exchange of your kid’s information can also be really helpful.

Many parents are concerned that their home endocrinologist will feel slighted or angry at having a CAH expert check on your child, their patient. My simple answer to that is, Tough! A good doctor will want to know how to treat a CAH kid the best that they possibly can.

My own experience is that although I had a very well-respected endocrinologist, my child was not only ill a lot, but her bone growth was not being checked regularly and it was getting out of hand. If her bone age is not monitored properly, I learned that my child could lose many inches off of her height with very little effort. I felt that she was not being carefully monitored, and I eventually listened to my instincts and found a doctor who treats many more CAH patients. If you cannot leave your child’s endocrinologist like I did, at the very least you need to keep it to the point, avoid the whole “slow down” discussion, tell her how to dose and test a CAH child more properly by enlisting the help of an expert. My suggestion would never be to fiddle with your child’s dose or do anything without the expert chiming in. Finding the perfect dose of Cortisol and Plomin (and yes, the stronger meds are not advised in little ones, and circadian dosing is also well accepted) is an intricate thing. You are attempting to replace the hormones as closely as possible to what the child would normally make if they could, in order to deal with daily and seasonal bloodwork (3 or 4 times a year), bone age X-rays, and exams by your child’s endocrinologist are imperative, especially in the early years when your child is growing quickly. If your endocrinologist is not handling these things closely, it may be time to get a second opinion. And if your child is having repeated adrenal crises, it is most definitely time to bring in the experts. In the meantime, read what you can, talk to CARES, and let your child’s endocrinologist know that you would like to explore new solutions to manage your child’s CAH.

I recently took part in the CARES’s walk in LA and got “re-schooled” on how to give the emergency shot. I am so glad I did, because I had forgotten exactly how to do it – it had been so long since I had even had to think about it! Please also make sure you feel comfortable with that process, as the ambulance and emergency room staff may not have a clue what CAH is or how to treat it. It is not your job to hurt your child by giving them the shot – it is an onetime dose. But overdosing regular meds over a long period of time is another story, so if you feel something is wrong, get it checked out.

And you are not alone. I guarantee you each parent living with a child who has CAH has their own anxieties about doing the right thing, including whether to get the shot. And don’t get me started about school and babysitters, etc. – well that is a later article. The more we demand that doctors know about CAH, the better it will be for us all. CARES has been instrumental in trying to increase international knowledge of CAH and its treatment, and my hope is that someday those doctors who really “have it down” can teach pediatric endocrinologists all over the world.

TIDBITS

CAH Parents as Patient Advocates

by Stephanie Erb

Having raised a SWCAH child who had many early complications, I gathered a great deal of knowledge about the trials and tribulations of raising a CAH kid, the hard way. Also, having worked as a consultant for CARES, I learned of the issues that other parents have all over the country. As we all know, each kid is different, and depending on what geographical area your family lives in, the methods of treatment vary on a daily deal. My hope is that someday in the not-so-distant future, a regular protocol for CAH kids will be well known, and implemented worldwide. For right now, though, a parent must educate themselves and seek out the CAH experts where they can.

So, my first advice is always listen to your instincts. If your kid is not growing, growing too fast, sick a lot, having

Left: The New York Presbyterian Hospital/Weill Cornell Medical Center in New York City is the first CARES-designated Comprehensive Care Center for CAH in the United States.
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