**Congenital Adrenal Hyperplasia (CAH)**

**Congenital Adrenal Hyperplasia (CAH)** is a family of inherited disorders affecting the adrenal gland. Over 90% of those diagnosed with CAH are affected by 21-hydroxylase deficiency. Inherited in severe, moderate, and mild forms, the major types of CAH are:

**Classical CAH** – The severe form of CAH or Classical CAH can result in life-threatening imbalances in salt and hormone levels. If undetected at birth, Classical CAH can lead to adrenal crisis and death. Frequently, newborn babies show no outward signs of the disorder and are sent home only to present a few days later for urgent medical attention at a time when they are beyond resuscitation. Classical CAH also is the most common cause of urogenital birth defects in affected females.

**Non-Classical CAH** – The mild form of CAH may cause symptoms at anytime from infancy through adulthood. While each individual presents differently, common symptoms include: premature development of body hair, body odor, rapid growth spurt (but ultimately short stature as an adult), early puberty, severe acne, anxiety, depression, mood swings, migraines and infertility.

**Frequency**

The genetic frequency of Classical CAH is approximately 1 in 10-12,000 births. NCAH affects 1 in 100 to 1 in 1000 in the general population, depending upon the ethnic composition of a given community, since its frequency varies in different ethnic groups. For example, it affects one in 27 Ashkenazi Jews, one in 40 Hispanics, one in 53 Croatians, and one in 300 Italians.

**Inheritance**

CAH affects males and females in equal numbers. For a child to be born with any form of CAH, both parents must carry a gene for the disorder.

**Treatment**

All forms of CAH are managed through oral medication that regulates hormone levels and replaces hormones not made by the body.

**Long-Term Effects**

If not detected or treated, Classical CAH may lead to adrenal crisis and death within a few weeks of life. Non-classical CAH can result in long-term quality of life issues, as well as growth problems and early puberty in childhood, and infertility. Despite the fact that there is no cure for CAH at this time, if treated properly, those affected by CAH can expect to live normal lives.
About
CARES Foundation, Inc. is a nonprofit organization that leads in the effort to improve the lives of the Congenital Adrenal Hyperplasia (CAH) community and seeks to advance quality health care through support, advocacy, education and research. We represent affected individuals, families and healthcare professionals in all 50 states and more than 80 nations.

We are the only U.S. organization solely dedicated to the CAH community.

Services
We offer the following programs and services to CAH and other adrenal insufficiency patients, as well as the medical community:

- One-on-one support for affected individuals and their families
- Referrals to expert physicians
- “Ask the Expert” service
- Educational materials and tools to improve the quality of life for patients and to educate teachers, school nurses, caregivers, and EMS professionals
- Family conferences held around the U.S.
- Educational programs for individuals, their families, and health care professionals
- Educational opportunities for medical professionals
- Research grants and participant recruiting for qualifying researchers studying treatments and searching for a cure
- Connections with support groups throughout the U.S. and twelve countries
- Information on current CAH research
- Advocacy on important issues facing the CAH community
- Bi-annual newsletters
- Monitoring of newborn screening in the U.S. and support for development internationally
- Free camp opportunities around the country at medically-safe facilities

To join the CARES community and encourage patients to join, go to https://www.caresfoundation.org. You will receive regular updates on our patient programs, services, research opportunities and more.

Mission
CARES Foundation leads in the effort to improve the lives of the Congenital Adrenal Hyperplasia community and seeks to advance quality health care through support, advocacy, education and research.
benchmarks of success

Since CARES’ founding in 2001, major strides have been made to improve health, connect people and save lives. Benchmarks of success thus far include:

- CARES now represents nearly 8,000+ affected individuals, families, and health care professionals in all 50 states and 80 nations.

  The advocacy efforts of CARES, member families, and professionals have led to every state in the U.S., Washington D.C., six provinces in Canada, and four states in Brazil, adopting newborn screening programs, thereby saving the lives of thousands of babies with CAH and other metabolic disorders every year. Having achieved the goal of comprehensive newborn screening in every state, CARES continues to support efforts internationally. CARES continues to monitor threats to treatment options available for patients.

- CARES has organized and presented numerous conferences bringing together the world’s experts in CAH with those affected by the disease and providing a wonderful opportunity for support. Hundreds of people have attended the conferences benefitting from what is often the only opportunity to meet others with the same challenges of living with CAH.

  60 family support groups have been formed across the United States and in twelve other countries. These groups provide individuals and families with opportunities to network with other affected individuals in their local communities. In 2011, CARES added specialized support groups to its network including parents of newborns, women with NCAH, and young adults. Regularly scheduled support group conference calls connect patients and families with each other, support group leaders, and medical professionals. Additionally, CARES has a Facebook page and many private Facebook groups offering further support.

- CARES has awarded hundreds of thousands of dollars in grants for CAH-related research.

  Advocacy work on the EMS front and passing protocols to include Solu-Cortef® on board ambulances nationwide.

  Since 2013, four comprehensive care centers have been designated as centers of excellence for CAH. They include New York Presbyterian/Weill Cornell Medical Center in New York City, Children’s Hospital of Los Angeles, Riley Hospital for Children in Indiana, and Cohen Children’s Medical Center at Northwell Health in New Hyde Park, New York.

- 8,000+ monthly online visitors to newly launched website which was redesigned to further engage and educate the CARES community.

  CARES counts on the generosity of patients, families and friends to support its many programs and services. An annual gala, awareness walks, and family fundraisers help fund research, support, advocacy, education and other services available to the CAH community.
CAH is a family of genetic disorders affecting the adrenal glands. The adrenal glands of people with CAH make little or no cortisol (the stress hormone). Like those with diabetes who take insulin, those with CAH take hydrocortisone every day to replace what their bodies do not make. Cortisol is a stress hormone and must be available to the body during times of injury, illness or other stressors on the body. In order for any treatments to address the injury or illness to be effective, the body must first receive hydrocortisone...

If someone with CAH is ill or injured and does not receive an injection of hydrocortisone, adrenal crisis may ensue. This can lead to shock or death.

Did you know many physicians and the general population are not aware of the symptoms of this inherited disorder and how common it is? Health care practitioners often fail to identify CAH and treat the symptoms rather than the underlying cause. This leaves patients to suffer years of visiting doctor after doctor in search of answers to their health problems.
COMPREHENSIVE CARE CENTERS FOR CAH

In 2009, CARES Foundation brought together the world’s leading clinicians in CAH, as well as affected individuals and families to create guidelines for the establishment of centers of excellence for the treatment and care of CAH throughout the lifecycle, using a medical home model which includes multidisciplinary specialists.

In April 2013, the first CARES-designated Comprehensive Care Center (CCC) for CAH opened at New York Presbyterian/Weill Cornell Medical Center in New York. Plans are in place to open more centers across the United States. In early 2015, CCCs were designated at CHLA in Los Angeles, Riley’s Children’s Hospital in Indiana, and Cohen Children’s Hospital in New Hyde Park, NY. Highly specialized care centers encompass care throughout a patient’s life cycle.

CAH FAMILY CONFERENCES

Each year, CARES provides conferences for individuals, families affected by CAH, as well as health care providers. Currently, conferences are held in various regions of the country and coordinated largely through the centers of excellence.

CAH NEWSLETTER

This bi-annual publication targets individuals, families and physicians who are impacted by CAH. Each newsletter contains information on CAH treatment methods, research and clinical trials, tips on living with CAH, advocacy efforts, support network, and upcoming CARES programs. The newsletter is available to anyone who registers with CARES. There is no charge to join the CARES community.

EDUCATION

Many physicians and the general population are not aware of the symptoms of this inherited disorder and how common it is. Health care practitioners often fail to identify CAH and treat the symptoms rather than the underlying cause. This leaves patients to suffer years of visiting doctor after doctor in search of answers to their health problems. CARES raises awareness of CAH, its symptoms, diagnostic protocols, treatment, genetic frequency, the necessity for early intervention and benefits of newborn screening in order to reduce suffering and save lives.

EDUCATIONAL MATERIALS

As the only organization in the United States solely dedicated to CAH, CARES’ comprehensive website is often the first entry into the Foundation. “Ask the Expert” and blog service on the website offer medical information. Resources developed to assist individuals and families cope with the day to day stresses of the disorder are:

- emergency materials on how to respond in a medical crisis
- emergency medical card to give to medical professionals to ensure the correct medication and dosages are administered
- tools for living with CAH such as school/camp packet and traveling with CAH
- educational videos
EMERGENCY MEDICAL SERVICES PROTOCOLS

In 2009, CARES embarked on a grass-roots campaign to establish protocols for emergency medical personnel to administer life-saving medications to people in adrenal crisis and for ambulances to carry the medication. When the campaign began, Rhode Island was the only state in the country with an EMS protocol for emergency medical treatment of adrenal insufficiency.

Currently, there are protocols in place or under development in a number of states. There is an interactive map on our website identifying states with protocols.

NEWBORN SCREENING ADVOCACY

Identifying CAH at birth can save a baby’s life. CARES has advocated throughout the U.S. for newborn screening. As of 2008, CAH is included on every state’s newborn screening panel in the United States. This landmark accomplishment is directly due to the grass-roots advocacy efforts of CARES.

In collaboration with other stakeholders and families, CARES continues to monitor states’ newborn screening programs and how budget cuts may adversely impact universal screening.

PHYSICIAN REFERRAL SERVICE

Affected individuals and their families can contact CARES for help locating a physician with knowledge of and experience with CAH.

RESEARCH FOR A CURE

CAH has no cure, but with continued research we may discover one. Federal funding for CAH-related research is extremely limited, therefore, the CAH community and its supporters fund research. Since inception in 2001, CARES has granted hundreds of thousands of dollars towards medical research and recruited hundreds of research participants. The goal is to seek out better treatment methods for those affected by CAH and to ultimately find a cure.

SUPPORT NETWORK

CARES supports the CAH community through support groups and one-on-one counseling. Support groups across the United States and abroad enable individuals and families to meet with each other, share stories and support each other’s medical concerns. Specialized support groups for specific topics (e.g. parents of kids with CAH, NCAH women, spouses and partners) are also available.

ASK THE EXPERT

Patients or parents can communicate with our medical director via this service and have their questions about treatment, testing, and other issues answered.
“Thank you...I don’t even think you can grasp the concept of how relieved I was as a parent of two CAH children to talk to someone who is so knowledgeable about the disorder and works so hard towards awareness! I’m grateful.”

“As a parent of a child with CAH, I love this program. CARES has been instrumental in our daughter’s health, well-being and growth over the years. So happy to support the work. Best and thanks!!”

“I’d like you and everyone at CARES to know that all your efforts are very much appreciated and invaluable for me and my wife! I remember getting downright scared in the first week, and hearing from you was assuring and encouraging. So big THANK YOU to you and your co-workers!”

“I don’t know where we would be today without CARES. You’ve been a life-saver!”

“I am excited. It will be the first time in 59 years I will be around people who understand or have CAH.”

“Thanks so much, I am glad I came across your organization on Twitter. The phone call the other night was very helpful and informative. As far as we know there are no other children in our area with CAH. Talking to actual parents was very refreshing. Thanks for all you do.”