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CONGENITAL
ADRENAL
HYPERPLASIA FOUNDATION
www.CARESFoundation.org

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:

Classic CAH

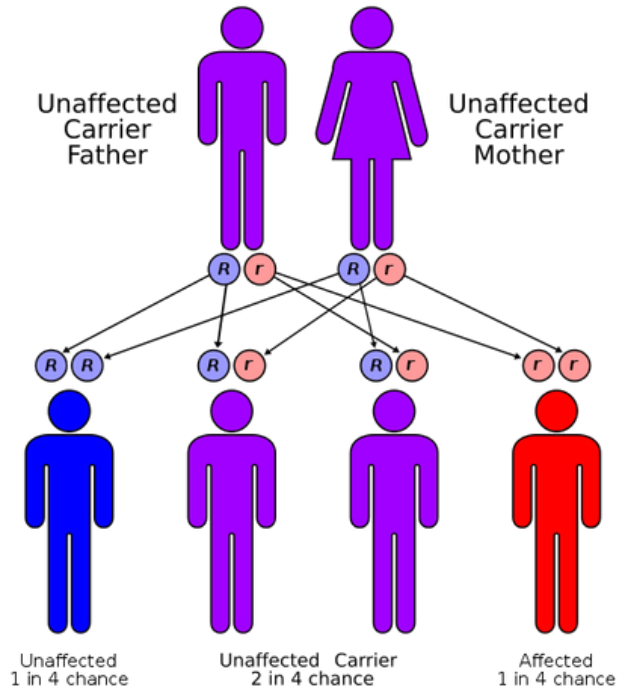
The severe form of CAH or classic CAH can result in life-threatening imbalances in salt and hormone levels. If undetected at birth, classic CAH can lead to adrenal crisis and death. Frequently, newborn babies show no outward signs of the disorder and are sent home only to return a few days later for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

Non-Classic CAH (Late Onset)

The mild form of CAH may cause symptoms at any time 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 classic CAH is approximately 1 in 10-15,000 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 mutation for the disorder.

Treatment

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

Long-term Effects

If not detected or treated, classic CAH may lead to adrenal crisis and death within a few weeks of life. Non-classic CAH can result in long-term quality of life issues, as well as growth problems and early puberty in childhood. However, despite the fact that there is currently no cure for CAH, gene therapy, that may result in a cure, is in clinical trials. With proper treatment those affected by CAH can expect to live normal lives.



Our 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.

CARES Foundation is a nonprofit organization based in the U.S. representing nearly 8,000 affected individuals, families, and health care professionals in the United States and more than 70 countries. CARES provides access to expert care in CAH. We offer a myriad of services and expert care at CARES-designated centers of excellence across the United States.

We offer many types of patient, caregiver, and family support, as well as many educational opportunities. We strive to advocate for treatment options, patient rights and awareness programs for CAH. CARES supports research for quality treatment, medication, and ultimately a cure. As we enter our 3rd decade of supporting the CAH community, we look forward to even better care, better support, and a better quality of life for CAH patients and families.

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 for direct email communication with our Medical Director
- 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
- Research grants and participant recruiting for qualifying researchers studying treatments and searching for a cure
- Connections with support groups throughout the U.S. and several other countries
- Information on current CAH research
- Advocacy on important issues facing the CAH community
- Bi-annual newsletters
- Monthly email updates
- 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.

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:

care

CARES now represents nearly 8,000+ affected individuals, families, and health care professionals in all 50 states and 70 nations.

advocacy

The advocacy efforts of CARES, member families, and professionals have led to every state in the U.S., Washington D.C., provinces in Canada, and states in Brazil, adopting newborn screening programs, thereby saving the lives of thousands of babies with CAH. Having achieved the goal of comprehensive newborn screening in every state, CARES continues to support efforts internationally.

education

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.

support

There are more than 70 family support leaders across the United States and in eleven other countries. These leaders provide individuals and families an opportunity to network with other affected individuals in their local communities. Regularly scheduled support group Zoom meetings 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.

research

CARES has awarded hundreds of thousands of dollars in grants for CAH-related research. We have partnered with Neurocrine Biosciences to create CAHtalog™, a patient registry that will advance CAH research.

PACE app

The PACE app is designed for adrenal insufficient patients and parents/caregivers and features a stress dose calculator, help recognizing signs of adrenal crises, and injection training. This app is available to the CAH community.

protocols

Advocacy work on the EMS front and passing protocols to include life-saving hydrocortisone sodium succinate onboard ambulances nationwide.

care centers

Since 2013, we have established a number of comprehensive care centers that have been designated as centers of excellence for CAH. Check our website www.caresfoundation.org for a current list.

website

9,000+ monthly online visitors to our website.

fundraising

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 individuals with CAH do not make enough cortisol (the hormone necessary in times of stress and for glucose reputation). 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.

meeting the needs of the community

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 Comprehensive Care Center (CCC) for CAH was designated at New York Presbyterian/Weill Cornell Medical Center in New York. Since then we have designated a number of highly specialized care centers throughout the country that offer care throughout a patient's life cycle. Please check our website, www.caresfoundation.org for a complete list of centers.

CAH Education Conferences

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

Newsletter/Monthly Updates

Our bi-annual newsletter 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 information, and upcoming CARES programs. The newsletter is available to anyone who registers with CARES. There is no charge to join the CARES community. We also publish monthly updates to keep the community informed about current topics and events.

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. We provide education at conferences, support group meetings and via webinars.

Educational Materials

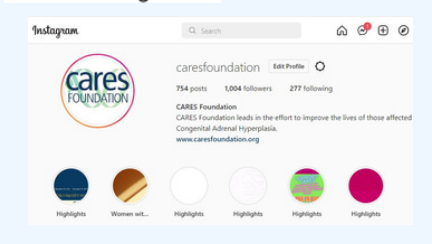
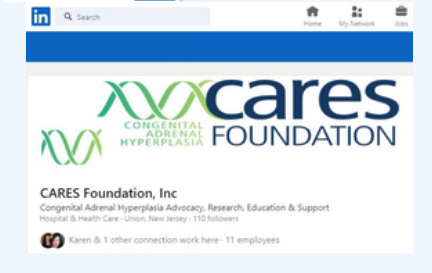
As the only organization in the United States solely dedicated to CAH, CARES' comprehensive website is often the first entry into the Foundation. Resources developed to assist individuals and families cope with the day-to-day stresses of the disorder are:

- 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

Our "Ask the Expert" service provides specific answers for needed questions. This service is available in English and Spanish.

meeting the needs of the community

CARES maintains an essential presence on several social media platforms. We encourage our community to check in for the most up-to-date CAH/CARES news.



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. Currently, there are protocols in place or under development in many states.

CAHtalog™

We have partnered with Neurocrine Biosciences to conduct a new type of registry to combine the de-identified medical histories of many volunteers into a single, CAH research database: CAHtalog™. The main purpose of the CAHtalog™ registry is to obtain information about how adults and children with CAH are managed by their doctors in a real-world setting.

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 U.S. state's newborn screening panel. This landmark accomplishment is directly due to the grass-roots advocacy efforts of CARES.

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 its establishment in 2000, 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 leaders. Support group leaders 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/Pregunta el Experto

Patients or parents can communicate with our medical director via this service for questions about treatment, testing, and other medical issues. Also available for Spanish-speaking patients and families.

I was 57 when I learned about CARES. Up to that point, I had not met or spoken to anyone about my CAH (except doctors). CARES has helped expand my knowledge of CAH through the educational programs offered, the support network and access to experts."

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



"CARES Foundation was the connection we needed to make sure our newborn had the doctors she needed, and we had the support we didn't even know we needed. Almost 10 years later, and they are still my first call!"



Upon diagnosis from the newborn screening test 13+ years ago, I can clearly remember the endocrinologist advising us to go directly to the CARES Foundation website to get accurate information and support. Here we are over a decade plus later, and we still look to CARES... to help navigate our journey."



www.CARESFoundation.org