# CAH Global View

A publication of CAH International
voices of congenital adrenal hyperplasia

FALL 2023 | VOL. 1

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Dear friends of CAH International,

It has been a long road already! We have created connections between CAH support groups around the world, gotten to know each other in our kick off sessions and follow up meetings online, and have gained a better understanding of what our communities around the world are struggling with.

Together, with our wonderful volunteers, we have been discussing ways forward in our work groups: Standard of Care and Surgery & Research, and have slowly but steadily been achieving milestones.

Our focus now is being more in touch with all of you on a regular basis. We plan on sending out various newsletters throughout the year that will keep you informed with what is happening at CAH International, and we also anticipate offering new online sessions starting 2024 that will revolve around topics that are most critical to our community.

CAH International was born out of the conviction that in order to make positive changes for our CAH community, we must focus on connecting internationally by helping each other out, sharing critical information, and working together internationally on critical issues.

CAH might be a rare condition, but together we are many! Together we can be an important voice for our community.

Let's continue this journey!
Your CAH International Org-Team

# Events

We are in the process of planning our next meetings, and your perspective is crucial in shaping the content and discussions. We want to ensure that the meetings address topics that matter most to you.

*AdobeStock_598784902*

**SAVE THESE DATES!**

February 24, 2024
June 22, 2024
November 16, 2024

***Online Meetings will be held via Zoom***

To make these meetings as valuable and relevant as possible, we invite you to suggest topics that you'd like to see on the agenda. Whether it's a specific challenge you're facing, a subject you're passionate about, or an area where you seek insights, your input will guide our conversation.

Here are some categories to consider when suggesting topics:

- Best Practices: Sharing and learning from one another about successful strategies, methods, or techniques.
- Challenges and Solutions: Identify common challenges and explore potential solutions collaboratively.
- Guest Speakers or Experts: Suggesting renowned experts or speakers who can provide valuable insights on a particular subject.
- Future Initiatives: Conversations about upcoming projects, goals, and opportunities for collaboration.
- Open Forum: A general discussion where attendees can bring up any topics of interest or concern.
- Other: If you have any other topics or concerns you'd like to discuss, please don't hesitate to share them.

Your feedback will help us tailor the agenda to ensure that the meeting is both informative and engaging for everyone. We want this to be a productive session that addresses your needs and interests.

Please send your preferred topics or other suggestions by email to info@cah-international.org by 15.12.2023. We will do our best to incorporate as many of your ideas as possible into our meeting agenda.

Your active participation in shaping this meeting is highly appreciated, and we look forward to your contributions. Should you have any questions or require further information, please do not hesitate to contact us.

# Focus on

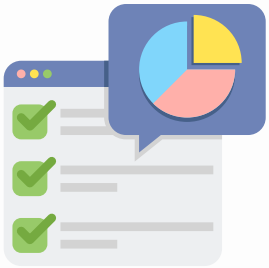

In this part of our newsletter we would like to take a look at one of the impressive groups/organizations that is a part of our network: CLAN (Caring & Living As Neighbours). The organization's founder, Dr. Kate Armstrong is a medical doctor and public health physician, that is also the mother of a young person living with CAH.

CLAN is an Australian NGO committed to equity for children living with chronic health conditions (also called non communicable diseases - NCDs) in resource poor settings. CLAN supports CAH Communities in different countries and recently founded an international movement called @MATES4Kids (Maximizing Access To Essential Supplies for Children), a coalition of individuals and organizations working to reducing the preventable mortality associated with CAH by 30% by 2030.

Check out CLAN's homepage Home (clanchildhealth.org), read "We all have a role to play" IJNS | Free Full-Text | We All Have a Role to Play: Redressing Inequities for Children Living with CAH and Other Chronic Health Conditions of Childhood in Resource-Poor Settings (mdpi.com) or visit @MATES4Kids (https://www.knowledge-action-portal.com/en/cop-categories/mates4kids-–-maximising-access-essential-supplies-children-living-ncds) to learn more!

# *Survey to identify the lifestyle of patients and caregivers living with CAH*

We are thrilled to announce the successful completion of our surveys designed to identify the lifestyle of patients and caregivers living with Congenital Adrenal Hyperplasia (CAH). A total of 498 responses were received from more than 20 nations. We extend our sincere thanks to each and every one of you who took the time to participate and share your valuable insights.

The analysis of the survey results has begun and we will be pleased to share the results with you in the near future.

The results of this survey will also be a valuable help for healthcare professionals, patients and caregivers who want to improve the quality of life of people with CAH.

Should you have any questions or wish to stay updated on our progress, please feel free to contact us:

# CAH and Surgery/DSD/Intersex

Our working group "CAH and Surgery/DSD/Intersex" is currently dealing with the topic very much from the perspective of European countries and the USA. In many countries of the northern hemisphere there are strong discussions about the assignment of CAH to the topic of intersex and surgery in females with CAH in childhood. In quite a few countries there are initiatives to ban or legislate surgery on females with CAH. In our working group we are dealing with the different views on this and we are working on finding a common position for this.

At the moment we are working on a document in which we deal with the frequency of intersex, which is often stated as 1.7% of the population (for example on the page of the UN Intersex people | OHCHR ). This figure is so high because non-classical CAH is included at 1.5%, which goes back to a survey study from 2000 by Blackless et al. (How sexually dimorphic are we? Review and synthesis - Blackless - 2000 - American Journal of Human Biology - Wiley Online Library) From our point of view parts of this study, which deal with non classical CAH, are not correct. The study with this incorrect figure of 1.7% has great impact, as we can see from the mention on the UN's homepage.

After that we will work on a statement for the Council of Europe. The Council of Europe has held events on intersex in 2022 and 2023 and is working on recommendations for its member countries. Human rights of intersex people: work launched on new Council of Europe recommendation - Portal (coe.int)

We are aware that at this time we are primarily addressing the issue from the perspective of countries where early diagnosis, adequate supply of medication and access to surgery make the issues of girls and women with CAH different issues than in countries with no or less comprehensive treatment. Our long-term goal is to cover the issue in its global diversity, but many more steps are needed to get there. We are only at the beginning of this journey together.

# Updates and Success Stories from our Community

KAHAKI
Komunitas Keluarga HAK Indonesia

## Medicine

On August 2023, a brand new medicine called Sydnacort has launched with the main ingredients is Fludrocortisone. This Sydnacort basically was requested from some doctors because they know how much this medicine can help some patients.

As CAH kids that are diagnosed with salt-wasting or salt-losing CAH they need this fludrocortisone to help them for doing their activity. We usually bought the medicine from Singapore with expensive price. But now, patients can buy in drugstores with cheaper price and receipt.

## Patient's Story

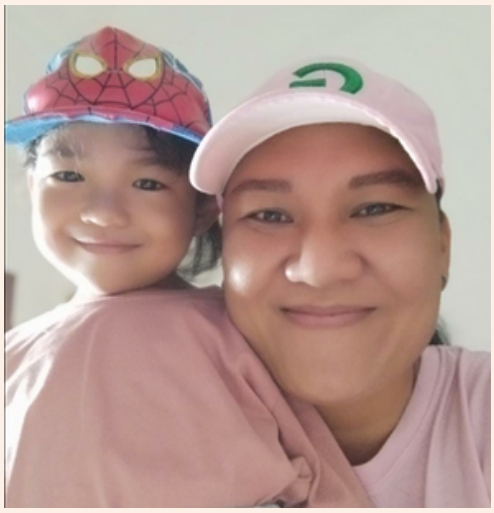
Last year, KAHAKI started to interview parents who are living with CAH kids to share their story and how they're struggling.

Why does this matter? As a rare disease, CAH is not well-known. There are a limited number of endocrinologists and there is no full healthcare or health service that are available in Indonesia.

So many parents are feeling lost, having nowhere to ask and to share. We do this so they can share with other CAH parents who are struggling too and feeling alone.


*August, 2023 - When the Sydna Farma Company presented about their Fludrocortisone medicine (called Sydnacort) in front of specialist doctors in Solo, Indonesia.*

*Sydnacort PPT here*



## Martalia Ginting and her son, Joe

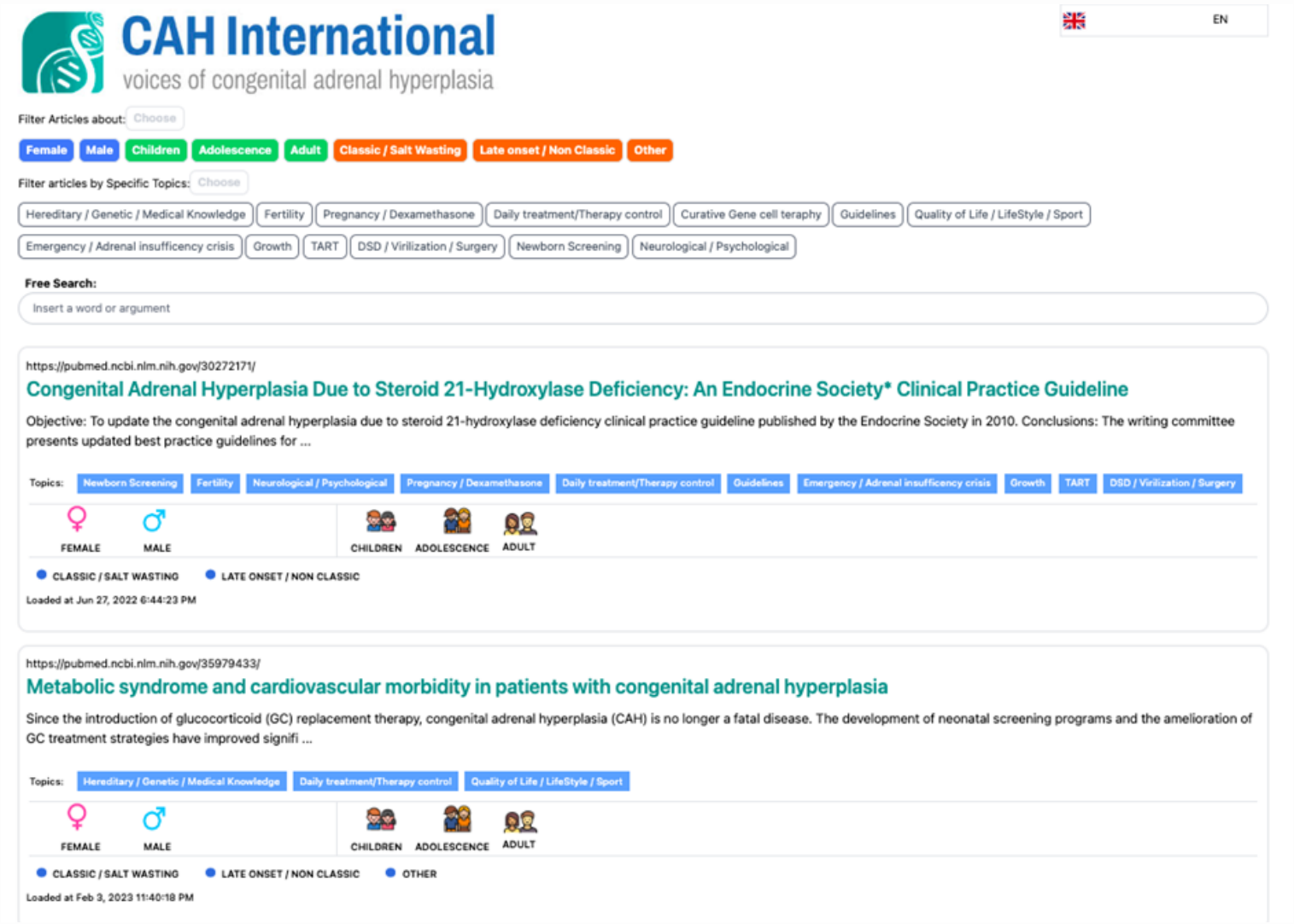
Joe was diagnosed with CAH when he was 10 months old. Like most moms out there who had heard the results of the doctor's examination about CAH, it made her sad. CAH is one of the rare diseases in Indonesia where treatment and health services are limited. But that didn't stop her as Ms. Martalia continued to force herself to learn about what CAH is, and she remained focused on taking care of Joe and making sure that he would always take his medication.

Watch video here

Thank you!

# Research Platform

Thanks to our fantastic supporters in the Research Work Group, we are currently finalizing the framework for our CAH International Research Platform. It is our intention to make research and critical information on CAH available to everyone, thus eliminating disparities in access and empowering communities to challenge the Standard of Practice in their respective regions. There are already various open source articles available on the platform, classified into categories and therefore easy to find if you are looking for a specific topic about CAH (such as fertility, emergencies, etc.). The amount of articles and research will grow over time as our research platform will eventually present an extensive resource for our community. We will share the platform with you once it is accessible to everyone. Here's a preview of what the platform looks like at the moment:



# Research

## Important Information for the CAH Community

Adrenas
a bridgebio company

### Developing an Investigational Gene Therapy for Classic CAH

Adrenas is developing an investigational gene therapy for classic congenital adrenal hyperplasia (CAH) due to 21-hydroxylase deficiency, which is the most common cause of CAH. The investigational gene therapy has the potential to enable a person living with CAH to produce cortisol and aldosterone in response to the body's rhythms, stress levels and the demands of daily living.

For information: www.cahgenetherapy.com and https://clinicaltrials.gov/study/NCT04783181

Crinetics
Pharmaceuticals

**Crinetics is a pharmaceutical company that develops much-needed therapies for people with rare endocrine diseases. We're here for patients who are eager to find therapies that provide effective disease control and more simplicity in their lives. One important condition we are currently exploring is congenital adrenal hyperplasia (CAH). We are developing a once-daily oral investigational medication called CRN04894, that acts directly on the adrenal gland to block adrenocorticotropic hormone (ACTH) action. Through this action, CRN04894 has the potential to lower adrenal androgen levels and ultimately may decrease and simplify glucocorticoid replacement therapy. Crinetics is excited to partner with the global CAH community to ensure patients' needs are addressed. We will continue to keep the global CAH community updated as clinical trials progress.**

# Neurocrine Biosciences and Diurnal Share Continued Commitment to CAH Community

*By Eiry W. Roberts, M.D., Chief Medical Officer at Neurocrine Biosciences, Inc. and John Porter, M.D., Chief Medical Officer at Diurnal Ltd., a Neurocrine Biosciences Company.*

Since the company's founding over 30 years ago, Neurocrine Biosciences has been dedicated to developing potential life-changing treatments for patients with unmet medical needs, including congenital adrenal hyperplasia (CAH). Our commitment can be traced back to the late Wylie Vale, PhD, a professor at The Salk Institute and one of Neurocrine's co-founders, who contributed to key Nobel Prize-winning work in endocrinology, specifically around the pathway impacted in CAH. This early discovery paved the way for current clinical trials that leverage our understanding of this pathway and formed the foundation of our driving purpose which remains true today: to meet the needs of under-addressed patients.

Last year, Neurocrine acquired Diurnal, a global pharmaceutical company focused on chronic endocrine conditions. Diurnal was also founded by a renowned endocrinologist, Professor Richard Ross, M.D. in 2004 as a spinoff company from the University of Sheffield in the U.K. to develop and commercialize pioneering endocrine replacement therapy research. It has three approved products for adrenal disorders: Alkindi® in the EU, Alkindi Sprinkle® in the U.S. (where it is marketed by Eton Pharmaceuticals), and Efmody® in the U.K. and the EU. Neurocrine and Diurnal are proud to unite our combined deep endocrinology knowledge and proven clinical development expertise to advance new therapies for endocrine disorders.

Neurocrine continues to investigate crinecerfont, a corticotropin-releasing hormone (CRF) receptor antagonist in Phase 3 studies for the potential treatment of adult and pediatric patients with CAH. There are currently no FDA-approved treatments for classic CAH besides glucocorticoids. For more than 60 years, glucocorticoids (and mineralocorticoids) have been the standard of care in treating classic CAH. However, glucocorticoids at doses to treat the cortisol deficiency alone are typically not enough to address the high adrenocorticotropic hormone (ACTH) and high androgen levels commonly found in patients with classic CAH. Thus, glucocorticoids serve a dual purpose in patients with CAH, to not only to treat the cortisol deficiency, but typically at higher doses to also reduce high ACTH and androgen levels. Long-term exposure to glucocorticoids at higher doses can cause metabolic issues, bone loss, growth impairment, and other health issues. This creates an undesirable trade-off of trying to balance the negative effects of too much glucocorticoid with the negative symptoms of too much androgen.

Crinecerfont was granted Orphan Drug Designation in the US and EU in 2019. In the second half of 2023, we plan to report results from our Phase 3 CAHtalyst Adult and CAHtalyst Pediatric clinical studies evaluating crinecerfont, an investigational medication that prevents CRF from binding to its receptor and may decrease the high ACTH and androgen levels seen in patients with classic CAH.

As Neurocrine and Diurnal unite, we all look forward to continuing to work closely with the CARES Foundation to keep this community informed about our investigational treatments in development for CAH and commercial offerings for other endocrine disorders. We are sincerely grateful for your active participation and partnership in this community.

You can learn more about crinecerfont, our investigational treatment in development, here, and more about Diurnal's approved treatments here.

# NEUROCRINE BIOSCIENCES UPDATES

**The CAHtalyst study was designed by Neurocrine Biosciences to evaluate the safety, efficacy, and tolerability of their novel drug crinecerfont in children (2-17 years) and adults with classic CAH.** **Please see the next page for trial site locations.**

**Crinecerfont is an oral, selective corticotropin-releasing factor type 1 (CRF1) receptor antagonist**

**September 12, 2023**

Neurocrine Biosciences announced positive data from the Phase 3 study of crinecerfont in adults with classic CAH. After 24 weeks of treatment, there was a statistically significant percent reduction in daily glucocorticoid (GC) dose versus placebo while maintaining androgen control ($p < 0.0001$). Approximately 63% of patients on crinecerfont were able to lower their daily GC to a physiologic dose in contrast to 18% on placebo.

They also reported a decrease in androstenedione after 4 weeks of treatment vs placebo ($p < 0.0001$).

Crinecerfont was generally well-tolerated. The most common adverse events were fatigue, headache, and coronavirus infection. No serious adverse events were assessed to be related to crinecerfont.

**October 5, 2023**

Neurocrine Biosciences announced Phase 3 pediatric study results of crinecerfont in children and adolescents (ages 2-17 years) with classic CAH. There was a significant decrease in androstenedione at Week 4 compared to baseline ($p = 0.0002$). There was also a significant percent reduction in daily GC dose while maintaining androgen control after 28 weeks of crinecerfont versus placebo ($p < 0.0001$). Approximately 30% of treated participants achieved reduction to physiologic GC dose compared to 0% of the placebo group.

The most common adverse events were headache, fever, vomiting, upper respiratory tract infection, and nasopharyngitis. No serious adverse events were assessed to be related to crinecerfont.

NEUROCRINE® BIOSCIENCES

# CAHtalyst Adult Study

## Trial Site Locations

### United States
Ann Arbor, MI
Atlanta, GA
Aurora, CO
Bethesda, MD
Boston, MA
Dallas, TX
Great Neck, NY
Indianapolis, IN
Los Angeles, CA
Minneapolis, MN
New York, NY
Philadelphia, PA
Pittsburg, PA
Rochester, MN
San Diego, CA
San Francisco, CA
Seattle, WA
St. Louis, MO
Tulsa, OK
Winston-Salem, NC

### Austria
Graz, Austria
Vienna, Austria

### Belgium
Brussels, Belgium
Leuven, Belgium

### Bulgaria
Sofia, Bulgaria
Sofia, Bulgaria

### Canada
Halifax, Nova Scotia, Canada

### Czechia
Hradec Králové, Czechia

### France
Angers, France
Grenoble, France
Le Kremlin-Bicêtre, France
Nantes, France
Paris, France
Paris France

### Germany
Dresden, Germany
Essen, Germany
Frankfurt, Germany
Leipzig, Germany
Munich, Germany

### Israel
Afula, Israel
Beer Sheva, Israel
Petah Tikva, Israel
Tel Aviv, Israel

### Italy
Ancona, Italy
Bologna, Italy
Florence, Italy
Messina, Italy
Milan, Italy
Milan, Italy
Naples, Italy
Padova, Italy
Roma, Italy

### Netherlands
Leiden, Netherlands

### Poland
Kraków, Poland
Poznań, Poland
Warszawa, Poland

### Portugal
Porto, Portugal

### Serbia
Belgrade, Serbia

### Spain
Madrid, Spain
Neurocrine Clinical Site-
Sevilla, Spain

### Sweden
Gothenburg, Sweden
Stockholm, Sweden

### United Kingdom
Cardifff, UK
London, UK
Manchester, UK
Salford, UK

CAH International
voices of congenital adrenal hyperplasia

# IS BUILDING STRONGER GLOBAL CONNECTIONS AND MAKING A DIFFERENCE

**A mom who was planning a trip to Italy this past summer reached out to us about resources to help her CAH child in the event of an illness or emergency. We connected her to one of our members who was able to provide her resources, guidance and most importantly peace of mind knowing that she was prepared in the event of an emergency.**

**Thank you for your support of the CAH Global View newsletter, a publication of CAH International!**

**We would appreciate your thoughts on this newsletter! If you have ideas for what to include in future editions or if you would like to contribute something, please email: info@cah-international.org**