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Classic Congenital Adrenal Hyperplasia (CAH)

VOICE OF THE PATIENT REPORT

Externally Led Patient-Focused Drug Development Meeting (EL-PFDD)

Public meeting: October 1, 2024
Report published: April 17, 2025
Hosted by: CARES Foundation

Voice of the Patient:
Classic Congenital Adrenal Hyperplasia (CAH)

AUTHORS: This report was submitted and prepared on behalf of CARES Foundation with input provided by Dina Matos (Executive Director) and Dr. Karen Lin Su (Medical Director). Consultant Trista Dawson Rutledge assisted in writing this report.

CONSULTING ADVISORS: Meeting support was provided by Larry Bauer, RN, MA, and James Valentine, JD, MHS, from Hyman, Phelps & McNamara, P.C., a law firm that represents sponsors focused on developing drugs for rare diseases as well as patient advocacy organizations.

ACKNOWLEDGMENT: Thank you to all the patients and families who participated in the meeting and shared their personal experiences. Special thanks to Dr. Theresa Kehoe (FDA), Dr. Karen Lin Su (Weill Cornell Medicine/CARES Foundation) and Dr. Richard Auchus (University of Michigan/Ann Arbor VA Medical Center) for presenting during the meeting.

TECHNICAL SERVICES: Thank you to Dudley Digital Works for providing technical support.

DISCLOSURES: Financial support for the meeting was generously provided by Crinetics Pharmaceuticals, Eton Pharmaceuticals and Neurocrine Biosciences. No pharmaceutical company was involved in the planning and coordination of the meeting.

PUBLIC MEETING: October 1, 2024

PUBLICATION DATE: April 17, 2025

SUBMITTED TO:

- Center for Drug Evaluation and Research (CDER)
- Center for Biologics Evaluation and Research (CBER)
- U.S. Food and Drug Administration (FDA)

VERSIONS AND MODIFICATIONS: This document has not been revised and/or modified in any way after the publication date. The submitters have all necessary permissions to submit this external resource to the FDA and linking to this resource from the FDA website does not violate the proprietary rights of others.

POINT OF CONTACT: Dina Matos, CARES Executive Director, dina@caresfoundation.org

CAH RESOURCES:

- CAH EL-PFDD Meeting Recording
- CARES Foundation
- Clinical Practice Guidelines
- CAH Guide for Families

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EXECUTIVE SUMMARY

Classic Congenital Adrenal Hyperplasia (CAH) is a genetic disorder affecting the adrenal glands, which are responsible for producing important hormones necessary for life, such as cortisol, aldosterone, and androgens. Classic CAH must be detected at birth (via newborn screening) to avoid an adrenal crisis, which can lead to death. There have been no new FDA-approved treatments for classic CAH in more than 70 years.

On Oct 1, 2024, CARES Foundation convened an Externally Led Patient-Focused Drug Development (EL-PFDD) meeting on classic CAH. The goal of this meeting was to enable the FDA, drug developers, and other stakeholders to hear from those directly impacted by the condition.

The meeting presentations, video stories, live panelist discussions, caller remarks, meeting polling data and comments submitted online (before and after the session) provided the content for this “Voice of the Patient” report. The key themes from these discussions are noted on the next page and explored in greater detail throughout the report.

KEY MEETING THEMES

1. **Classic CAH brings a range of debilitating physical symptoms, from the condition itself as well as from lifelong steroid treatment. These can include fatigue, weight gain, poor growth, muscle weakness, benign testicular tumors, and more.**
   - Participants also highlighted salt-related issues, such as dehydration, dizziness, excess urination, and salt-wasting, with many patients requiring salt supplements to maintain proper balance and prevent dangerous complications.

2. **Classic CAH can carry a profound emotional toll as patients face deeply sensitive, life-altering challenges around body image, obesity, atypical genitalia (females), early-onset puberty, infertility, and more.**
   - Patients and families carry a heavy mental load from navigating not only daily health complexities but also accepting heartbreaking realities such as the potential inability to have children.

3. **The treatment of CAH has been called "an art, not a science" due to complexities of managing the condition which can create a significant burden for patients. This includes ensuring precise dosing, managing multiple daily treatments, dealing with unpredictable symptoms, and ongoing monitoring to prevent dangerous imbalances.**
   - The transition from childhood to adulthood intensifies these challenges, as young adults take on the responsibility of managing their complex treatment plans, often made harder by the lack of experienced CAH specialists.

4. **An adrenal crisis, triggered by illness, stress or injury, demands vigilant emergency preparedness as a lifesaving yet cumbersome treatment must be administered at a moment's notice to avoid potentially fatal consequences.**
   - Injectable hydrocortisone (Solu-Cortef), used to treat adrenal crises, requires a complex, multi-step preparation process that can be overwhelming in emergencies - especially since many healthcare providers (including emergency response staff) are unfamiliar with it.

5. **Patients and parents urgently need new treatment options that not only reduce the exposure to glucocorticoids and side effects but also provide easier, more reliable ways to monitor hormone levels and circumvent adrenal crises.**
   - Major concerns include the lack of timely methods to assess current hormone levels for precision dosing (blood tests results can take weeks), the long-term impact and side effects of lifelong steroid use, the strict daily medication management needs, the complex administration of life-saving adrenal crisis medication, and more.

INTRODUCTION

The Externally Led Patient-Focused Drug Development (EL-PFDD) meeting on classic congenital adrenal hyperplasia, or CAH, was held on October 1, 2024. The meeting was organized by CARES Foundation, an organization that aims to improve the lives of the CAH community and advance healthcare through advocacy, education, research and support.

The FDA's PFDD initiative was created to recognize the unique insights that patients and their families bring to drug development and evaluation. The PFDD meetings provide an opportunity for the FDA and other stakeholders to hear directly from patients and families about the symptoms that matter most to them and the disease's impact on daily life, as well as experiences with current treatments and hopes for the future.

What is CAH?

Congenital adrenal hyperplasia, or CAH, is a genetic disorder present at birth that affects the adrenal glands, which produce hormones such as cortisol, aldosterone, and androgens (type of sex hormones). In most people with CAH, an enzyme called 21-hydroxylase, essential for hormone production, is deficient due to a pathogenic variant in the *CYP21A2* gene. This leads to a hormone imbalance, with reduced cortisol and aldosterone and excess androgen production, which can result in a range of debilitating symptoms depending on the severity of the condition.

What are the types of CAH?

There are two types of CAH which vary in severity and the typical age of diagnosis:

- **Classic CAH** is the more severe form and the focus of the EL-PFDD meeting. The condition is typically diagnosed in infancy and characterized by significantly reduced production of cortisol and aldosterone, leading to hormonal imbalances.
    - The **salt-wasting form,** affecting about 75% of those with classic CAH, is the most severe form. The body cannot produce enough cortisol or aldosterone, necessary for maintaining normal fluid volume of sodium and potassium, which among other functions, stabilizes the heart. This can lead to a dangerous loss of salt and fluids, which can be life-threatening without treatment.
    - In contrast, the **simple-virilizing form**, affecting around 25% of people with classic CAH, does not cause significant salt loss but still results in a shortage of cortisol, affecting physical development and growth.
- **Non-classic CAH** is milder, often diagnosed later in childhood or adulthood (also called "late-onset" CAH), and involves a partial enzyme deficiency, allowing for more normal hormone production, though some hormonal imbalance can still occur.

INTRODUCTION

What are the symptoms of classic CAH?

The signs and symptoms of classic CAH include premature signs of puberty, poor growth, abnormal periods, infertility, benign testicular tumors, and more. Classic CAH often causes females to be born with atypical genitalia. Hormonal imbalances caused by salt-wasting CAH may also result in severe adrenal symptoms that include dehydration, weight loss, hypotension, hypoglycemia and seizures.

Who is most affected?

The condition can affect females and males equally. Classic CAH affects anywhere from 1 in 9,000 to 1 in 30,000 births worldwide or ~1 in 16,000 births in the U.S. One in 60 people are carriers for classic CAH. Non-classic CAH is more common, affecting ~1 in 1,000 births worldwide.

How is CAH detected?

Prior to having newborn screening available, males with salt-wasting CAH often died in infancy. As of 2010, newborn screening is available in all 50 U.S. states.

What causes CAH?

CAH is inherited in an autosomal recessive pattern, which means that a child must inherit a pathogenic variant from both parents to develop the condition. If both parents are carriers with each having one copy of the mutated gene, there is a 25% chance with each pregnancy that a child will have CAH.

How is the condition treated?

Classic CAH is primarily treated with hormone replacement therapy to correct cortisol and aldosterone deficiencies. Patients typically take glucocorticoids, like hydrocortisone, to manage cortisol levels and mineralocorticoids, such as fludrocortisone, to regulate sodium and potassium balance. This can lead to side effects from those medications, including short stature in children, as well as obesity, hypertension, and osteoporosis.

In addition, hydrocortisone is dosed two to three times a day to mimic the adrenal gland's normal pattern of cortisol release.

During times of illness, injury, or stress, patients may need higher doses of glucocorticoids to prevent adrenal crisis. Additionally, some individuals may require salt supplements, especially in the early years of life.

Patients with CAH need frequent monitoring with routine lab tests to manage their condition, their medication dose and to avoid complications.

There have been no new FDA-approved treatments for CAH in more than 70 years.

MEETING OVERVIEW

This EL-PFDD meeting was designed to highlight the symptoms and impact of CAH on daily life while also addressing current treatment approaches and hopes for future therapies (see agenda in Appendix 1).

Both morning and afternoon sessions featured pre-recorded videos from patients and caregivers, followed by panel discussions. Viewers could share their perspectives through phone calls or online written comments. Participants were also invited to answer polling questions online to gather demographic data and experiences (see polling results in Appendix 2). Caregivers were asked to respond on behalf of patients they currently care for, or those who have passed away from CAH complications.

In addition to patients and families, speakers included:

- **Dina Matos, Executive Director at CARES Foundation**, served as meeting co-moderator and expressed gratitude at the opportunity for the CAH community to share their experiences.
- **James Valentine, JD MHS, Hyman, Phelps & McNamara, P.C,** who helped launch the PFDD program at the FDA, served as co-moderator and outlined the meeting format and guidelines.
- **Dr. Theresa Kehoe, Director, Division of General Endocrinology, Center for Drug Evaluation and Research (CDER) at FDA,** provided a brief overview of CAH and reinforced the importance of this meeting for FDA reviewers to hear directly from patients and caregivers.
- **Dr. Karen Lin Su, Co-Medical Director of the CAH Comprehensive Center at New York-Presbyterian Weill Cornell Medicine and Medical Director at CARES Foundation** presented a clinical overview of CAH.
- **Dr. Richard Auchus, Division of Metabolism, Endocrinology and Diabetes at the University of Michigan and Ann Arbor VA Medical Center,** shared a clinical treatment overview.
- **Larry Bauer, RN, MA, from Hyman, Phelps & McNamara, P.C,** who is a former member of the FDA Rare Diseases Program, summarized the meeting.

Members of the CAH community were invited to submit written comments up to 30 days after the meeting (see Appendix 3).

A recording of the CAH EL-PFDD meeting can be found here:
https://caresfoundation.org/externally-led-patient-focused-drug-development-el-pfdd/

MEETING OVERVIEW

Meeting participants and polling: By the numbers

- **Overall meeting attendance**: 160+ total, with ~60 patients or caregivers
- **Polling participation**: ~50 (40% of people living with CAH, 60% caregivers)
- 57% female, 38% male, 4% other
- **Ages represented among those with classic CAH:** < 12 years old (30%); 13-18 years old (17%); 19-35 years old (26%); 36-50 years old (17%); 51-60 years old (6%); 61-70 years old (4%)
- **How classic CAH was detected:** Clinical signs/symptoms (63%); Newborn screening (37%)
- **Majority of participants reside** in the US Eastern and Central time zones, while others joined from the Pacific and Mountain time zones, as well as Europe

Report Overview

This Voice of the Patient report intends to enhance the understanding of the FDA and other stakeholders regarding the experiences, perspectives, and needs of those living with classic CAH.

The report summarizes input from the EL-PFDD meeting and does not represent the specific views or experiences of any particular group. It may not cover all symptoms, impacts, treatments or aspects of classic CAH.

LIFE WITH CLASSIC CAH

Patients and parents described how classic CAH affects every dimension of life. Individuals living with the condition face relentless physical and mental health challenges — not only from the disease itself but also from its treatment regimen. For parents, the journey brings a constant undercurrent of worry — the threat of adrenal crises, the uncertainty of their loved one being able to live independently, worries about the future, difficulty finding experienced healthcare providers, and much more. As these families confront the day-to-day demands of managing CAH, the cumulative impact on their lives defines this condition.

"At 12 days of age, Joey was put in the pediatric ICU with severe adrenal crisis symptoms, needing advanced and intense care. We were told **he may not survive the first 24 hours** of his hospitalization. It was a **harrowing introduction to CAH**, one that etched fear into our hearts and clouded our vision of the condition for years to come," recounted Louise about her son's early days with CAH. "I began the task of researching and learning all I could about a rare condition that, **in the words of the endocrinologist, would likely be fine but, without quick action, could kill my son."**

> **"Activities like field trips and going to camps and everything that he does, we have this extra layer of things that we have to prepare for...we have to be so thoughtful about each and every one of those activities,"** shared Katherine, about her 15 year-old son.

CLASSIC CAH: SYMPTOM SUMMARY

Patients face a spectrum of debilitating symptoms

Classic CAH presents a wide range of symptoms that can profoundly impact individuals from infancy through adulthood. Physically, symptoms can include fatigue/low energy, adrenal crises, dehydration, and challenges with growth and weight, all requiring vigilant management and frequent medical intervention. Hormonal imbalances often lead to complications like early-onset puberty, testicular tumors in males and atypical genitalia in females. Over time, these physical symptoms contribute to a host of secondary complications, such as fertility issues, bone density concerns, and metabolic strain. Mentally, the disorder contributes to stress and anxiety due to the daily demands of treatment and fear of health crises. The complex array of symptoms requires constant attention.

SYMPTOM	PATIENT/CAREGIVER VOICE
Low energy and fatigue Many participants mentioned their struggle with low energy and fatigue as a common symptom, describing an overwhelming, constant sense of tiredness. Some shared that the fatigue can feel both physical and mental, making even simple tasks feel like significant challenges.	“One effect of CAH overall is being **low energy** most of the time. I can't remember when I didn't **feel tired**,” shared Nancy, 49 years old. “All day, every day, I struggle with **fatigue,** both physically and mentally,” wrote Jill.
Dehydration, dizziness, excess urination Participants discussed having issues with dehydration, caused by the body’s inability to regulate salt and water balance due to adrenal hormone imbalances. Managing these issues often involves drinking large amounts of fluids and increasing salt intake to stay hydrated and avoid serious complications.	“From a daily standpoint, **dehydration and dizziness** comes into play pretty quickly if an afternoon dose, for example, is missed,” said Erik, 33 years old. “**Dehydration** can be serious very quickly,” said Lesley, 67 years old. “If I play tennis, I'll down two 32-ounce bottles of Gatorade and then more at home and people are just kind of like, ‘You **drink a lot’** but I know that's what I need,” shared Ann.
Excessive weight gain For some individuals with CAH, weight gain can be a persistent and distressing issue often tied to hormonal imbalances and the effects of long-term steroid use. This challenge often contributes to additional struggles, including disordered eating, body dysmorphia, and emotional frustration, impacting both children and adults alike.	"As an adult female with CAH, I have struggled with **my weight**, disordered eating, and body dysmorphia for quite a while,” wrote Sabrina. **“Weight gain** has been a tremendous frustration for him,” said Michele about her 12-year-old son.
Anxiety and/or depression Patients reported dealing with anxiety and depression, which may be influenced by the chronic stress of managing this rare, lifelong condition and its physical symptoms. These mental health challenges can be further affected by feelings of isolation, body image concerns, and the emotional toll of navigating a complex treatment regimen.	“At the age of 31, I started getting **very low and depressed.** I always thought I was the only one with this health condition because it's so rare. I had a plan. I was going to take my own life, but instead, I decided to go get help in a mental institution,” said Erica, 48 years old. “I have been **hospitalized** several times, take oral **antidepressants**, talk to a therapist. I have had a therapist since I was age 15,” said Nancy, 49 years old.

SYMPTOM	PATIENT/CAREGIVER VOICE
Atypical genitalia Females with CAH can be born atypical genitalia due to excess androgen exposure during fetal development. While some affected patients reported undergoing reconstructive surgery, others noted the personal impact on their sense of identity and self-esteem.	"At my birth, my exhausted mother asked, '**Is it a boy or a girl?**' 'We're not sure,' replied the doctor," shared Lydia, 38 years old. "The way I was diagnosed was because of my **atypical genitalia**. They told my parents they had a baby boy," said Erica, 48 years old.
Poor growth Stunted growth is a challenging issue for some individuals with CAH. Parents shared stories of children not reaching their projected heights despite their best efforts. Concerns about height often go beyond the physical, significantly impacting self-esteem and overall mental well-being.	"While he is taller than both his parents, he is two inches **shorter** than projected height. He will never forgive us for not proceeding with **growth** hormone and speaks often of going to Korea for leg lengthening surgery. While this sounds extreme and he is young, his sentiment is real," wrote Gretchen about her son with CAH. "**Height** has always been a concern impacting both his physical and mental well-being. Despite our best efforts, John has **only grown a few inches** and is not expected to exceed five feet, three inches tall," said Virginia about her 16-year-old son with CAH.
Poor feeding and/or poor weight gain Experiences of "failure to thrive" during infancy were described in the meeting, highlighting the critical need for vigilant monitoring and care in early life to ensure survival and support healthy development.	"Very little was known about CAH and a few days into my life, I was **not eating and very lethargic**. My early life was a constant battle to stay alive," said Lesley, 67 years old.
High blood pressure Some individuals noted issues with high blood pressure, which can be triggered by imbalances in treatment or life circumstances.	"If you get out of balance with your medications or with what's happening in life, you can very quickly get into zones of **high blood pressure** and issues that become much greater problems," said Katherine, mom of a 15-year-old son with classic CAH.

SYMPTOM	PATIENT/CAREGIVER VOICE
Irregular menstrual periods, testicular tumors, and infertility Discussions during the meeting revealed how these complex health challenges intertwine with emotional well-being, emphasizing the broader impact of CAH on reproductive health and quality of life.	"I have definitely struggled with some gender dysphoria. **Not having regular menstrual periods**…made me feel less feminine," shared Al, 43 years old. "My doctor placed me on hormonal birth control to **regulate [my periods]**, which plunged me into a diagnosis of severe depression and anxiety," said Alana, 25 years old. "I was diagnosed with **testicular tumors** which led to fertility issues," said Mark, 57 years old.
Other symptoms Meeting participants highlighted a wide range of additional struggles faced by individuals with CAH, from bone density issues and adrenal tumors to debilitating headaches, body aches, and the effects of precocious puberty. These varied symptoms underscore the complex and evolving nature of CAH, affecting patients differently across their lifespan.	"At 30 years old, you have the **bone density** of someone 80 years old and the prospect of a fracture that could end your life at a really young age," said Tim, now 58 years old, who also dealt with a benign **adrenal tumor the size of a volleyball** which took eight hours of surgery to remove. "I know I'm in my early twenties and **my bones** shouldn't hurt yet, but they do," said Simran, a 23-year-old medical student living with classic CAH. "Her symptoms have evolved from infancy. She used to get a little **nauseous** and **extremely lethargic**. Now she gets **debilitating headaches**, **worse nausea**, and her whole body starts **aching** with **shakes**, **clammy** and hard to move." wrote Amanda about her daughter with classic CAH. "John entered **precocious puberty** at age 6. This rapid development was alarming," shared Virginia about her son.

Compiled from patient and caregiver discussions and responses to polling questions during the EL-PFDD meeting (see Appendix 2 for full polling results).

The ever-present threat of adrenal crisis: A life on high alert

Meeting participants characterized adrenal crises as the **most troublesome** symptom. Throughout the meeting, participants repeatedly expressed their deep-seated fears about facing a potentially life-threatening crisis and accessing the complex, time-sensitive treatment required to survive. This ever-present concern often prevents parents from feeling secure to leave their child in someone else's care, unsure if others would recognize the signs of an impending crisis or respond effectively in an emergency. The relentless anxiety around these emergencies impacts not only day-to-day activities but also shapes long-term decisions and plans.

"An **adrenal crisis**…it feels like a **wilting flower**. You lose your energy, you lose your ability to communicate, **you start to just truly shut down,** which is a feeling that unless you've experienced it, you really can't explain it except for that," said 33-year-old Erik.

"I'm afraid that if I was unconscious and couldn't fight for myself, that I'd die in an **adrenal crisis**," said Lydia.

"The most troublesome part of raising girls with CAH is **preventing and treating an adrenal crisis**. The risk of the crisis, since it is life-threatening, makes it necessary for training for anyone who's going to be left with my girls," said Colleen, mother to two young daughters with classic CAH.

"There was a period of time that he would go into crisis, I would say, **five, six times a year,**" said Michele about her now 12-year-old son.

> **Participants said that "having an adrenal crisis" was their biggest fear (28%), followed by being mistreated in the ER (19%), worsening symptoms (14%) and dying prematurely (11%)**

Life-saving medication is complex and challenging to administer

People with classic CAH in an adrenal crisis can die without access to certain medication, but the process of administering this life-saving treatment is anything but straightforward. Solu-Cortef treatment can involve many steps, which is especially difficult to manage under the intense pressure of an emergency.

“Solu-Cortef is a wonderful drug, but especially when you're stressed, not feeling well - **it's hard to open**. But once you get that open, you have to put some air in the syringe, put it in the vial, turn the vial upside down, have the syringe fill up with the liquid, and then inject it. **That's all really hard to do when you are not feeling well**, when you feel like you're going to pass out and you're weak and you're confused,” shared Stephanie, mother to twins with classic CAH who are now 30 years old.

“One of the things I worry most is my son will have an adrenal crisis when he's alone or around people who aren't extremely familiar with his emergency Solu-Cortef injection. It is a **really clunky, complicated process** to administer the life-saving injection. Even my son's teachers and grandparents who care for him regularly and have received training are **terrified that they'll not remember all the steps correctly** if they ever have to use it in an emergency,” wrote Chelsea.

Emergency staff may not be willing or able to provide Solu-Cortef

Accessing timely and appropriate emergency care is a critical challenge for individuals with CAH, often compounding the stress of managing the condition. From first responder restrictions on administering life-saving medications to delays in medical interventions, participants' stories highlight the urgent need for better awareness, training, and resources in emergency settings.

“In most cases, if you were to call emergency medical services, they are legally barred from administering [Solu-Cortef], and often these shots are not even carried on the truck. Imagine the stress that carries on parents**. The ones you call for help, legally cannot administer an emergency life-saving injection**,” shared Marc, 32 years old.

“I've had situations where a **doctor told me that my daughter didn't need Solu-Cortef**. He wouldn't give it to her. I asked for an attending. He wouldn't get an attending. He wouldn't get an endocrinologist. I wanted to inject her myself because I could see that she was going into an adrenal crisis and I was escorted out the hospital because of that,” said Stephanie, mother of twins who are 30 years old.

“I am a pediatric RN in Seattle and see the **impact delayed medications** for CAH have on patients. I also live in a rural area where emergency medical services can be an hour from bringing a patient to a children’s hospital. **Rapid medical interventions can change the outcomes for CAH patients**,” said Julie.

IMPACT ON DAILY LIFE

This daily burden of classic CAH is woven into the fabric of everyday life, which can be exhausting and isolating for patients and their families. The condition requires constant vigilance, from adhering to complex medication schedules to preparing for potential emergencies, making even routine activities feel unpredictable. For many, the stress of handling physical and emotional symptoms can disrupt work, school, and social interactions. Beyond the immediate impacts, classic CAH brings a range of long-term health issues that can significantly alter life trajectories. The impact extends to family members, who often share in the responsibility of managing the condition.

> **"Even though CAH does not define our son, it impacts every aspect of his life and ours,"** shared one mother.

CLASSIC CAH: IMPACT OVERVIEW

Navigating Life with CAH

Patients and caregivers vividly described how living with CAH disrupts nearly every aspect of daily life. Their stories painted a compelling picture of the challenges and pervasive impact this condition has on physical, emotional, and social well-being. From physical exhaustion and medical complexities to emotional challenges and social limitations, individuals with CAH and their families face seemingly constant challenges. These experiences illustrate the condition's far-reaching effects — encompassing both short- and long-term impacts — and emphasizing the resilience required to navigate its daily demands.

ACTIVITY	PATIENT/CAREGIVER VOICE
Restful sleeping As shared during the meeting, managing CAH can greatly disrupt sleep for both patients and caregivers, with treatment schedules often requiring nighttime doses and ongoing vigilance.	"Taking meds every 6 hours is **exhausting–** especially as a parent having to stay up until midnight daily. We never get enough **sleep**," wrote Amanda, whose daughter has CAH. "I think people with CAH are always **tired** to some degree," shared Julia, 33 years old.
Sports or other recreational activities Meeting discussions highlighted the challenges individuals with CAH can face in participating in sports and recreational activities. Concerns included access to medical support, the need for frequent hydration or salt intake, and the physical toll of the condition often limit involvement or require significant preparation and adjustments.	"There have been times that he has **not been able to do a certain sport** because there hasn't been the access to the medical teams that we'd feel comfortable with," said Carlos about his 14-year-old son. "On a softball field, I was a catcher, which is a very hot job. You might catch me in a dugout **eating a bouillon cube**. Yes, I know it sounds nasty, but I had to get my salt intake from as much sweat as I had," said Erica, 48 years old.
Loss of independence Participants expressed deep concerns about the potential loss of independence caused by the demands of managing CAH. The constant need for monitoring, medication, and readiness for emergencies can limit the ability to live alone, travel, or fully pursue personal goals.	"My biggest fear is the potential **loss of independence**. The thought of not being able to take care of myself or pursue my passions is a constant source of anxiety," said Alana, 25 years old. "The **loss of independence** has been the big one for me. I live alone, so you always have in the back of the mind what if you do have an adrenal crisis. I would probably never travel alone," said Ann, 55 years old. "The real limiting factor of living with CAH is: **He cannot live alone**," wrote Gretchen.

Attending social events with family and friends

The need for careful planning and preparation often makes attending social events challenging for individuals with CAH. Concerns about medication schedules, emergency preparedness, and ensuring caregivers or trusted individuals are present can limit participation and add stress to what should be enjoyable experiences.

“She can’t just go to anyone’s house because of her CAH. I have to know the family, trust the family, and train the family on her injection. **I can’t just drop her off with anyone or anywhere,**” shared Amanda about her daughter.

“Activities like **field trips and going to camps** and everything that he does, we have this extra layer of things that we have to prepare for… we have to be so thoughtful about each and every one of those activities,” shared Katherine, about her 15-year-old son.

“I have to **stop playing or doing activities** because of having to take medicine. And I **can't do sleepovers** because of my noon pill or my midnight pill,” said Raelie, 10 years old.

"Our five-year-old daughter frequently has symptoms of headaches and nausea, and once a week this **changes our dinner routine** or even sports attendance. During **family vacations,** this results in a day the family has to take a slow day," shared Jess.

Ability to have children

The prospect of not being able to have children was described as a profound source of grief and emotional pain.

“My family doctor at that time told me that it would be very difficult for me to conceive with CAH, and I should just forget about having children. There was no referral to a **fertility** expert. Therefore, I have **no biological children,**” said Lesley, 67 years old. “This has been a **huge emotional regret** for me to this day.”

Self-esteem and body image

Participants shared how the invisible nature of CAH can leave them feeling isolated and misunderstood, with body image concerns like severe acne and weight changes significantly impacting self-esteem and emotional well-being.

“The hardest thing for him is obviously that it's an **invisible disease** and that a lot of people do not understand what he goes through on the day to day,” said Andrea about her young son.

“As a teenager, I had acne that swallowed up and made huge sores all over my face. This added to my **low self-esteem and body image issues,**” said Nancy, 49 years old.

“They've always been **sensitive about their weight**. They look back at pictures and don't like how they looked. It has definitely affected their **body image** and how they go through life,” shared Stephanie, mother to twins with classic CAH.

Working or having a career Participants expressed how CAH can make it difficult to maintain a career, with some describing physical and mental fatigue that hinders workplace performance, while others highlighted the challenges of balancing caregiving responsibilities. Families noted the financial strain of adjusting work schedules or keeping one parent at home, creating additional stress and impacting long-term career opportunities.	"Afternoons lack motivation, at times can lead to issues with communicating my thoughts properly. This can be very **troublesome at work**, speaking to groups or just friendly conversations. It can be very **frustrating and embarrassing**," said Mark, 57 years old. "We kept **one parent at home at all times** after trying a dual worker household and found it impossible," said Gretchen. "It's been hard as a parent of a toddler with CAH to have a 'normal' career. I would always give up my career to take care of my children if it came to it, but the **income restriction** affects the whole family and I'm noticing how different it is from my other colleagues with children. We don't feel comfortable being more than five minutes away from our child while they're starting preschool, and thus we have to 'switch off' when we work which **severely impacts our income,**" wrote Corrine.
Attending school Participants recounted how CAH can make attending school or daycare a significant challenge. Some described emotional and behavioral struggles, while others noted safety concerns, feeling unequipped to trust caregivers with the medical complexities of CAH.	"**Straight-up refusals at school**, not wanting to do things. She gets very stubborn. She shuts down. She feels like she's crawling out of her skin a lot of times," shared Rita about her 5-year-old daughter with classic CAH. "We have **not felt safe** sending our 13-month-old to daycare since she was born. We are ill equipped as non-medical professionals to draw up a needle in a crisis and certainly **don't trust untrained day care staff**," wrote Diana.
Preparing meals and household tasks Fatigue from CAH can make everyday tasks, like cooking and chores, feel overwhelming and difficult to complete.	"I'll tell my husband: 'It feels like an energy vampire has sucked out my life force,' and I'll just go lay down. I **can't continue** cooking dinner," said Lydia.

Compiled from patient and caregiver discussions and responses to polling questions during the EL-PFDD meeting (see Appendix 2 for full polling results).

Mental Health Impacts can be Severe

Both women and men with CAH face significant mental health challenges, stemming from shared struggles with body image and self-esteem, as well as unique concerns tied to specific symptoms. For women, atypical genitalia can trigger additional emotional impacts, while men often contend with issues related to height, weight, and overall physical development. These combined pressures contribute to a complex mental health burden that affects many aspects of life.

Trauma due to invasive and sensitive exams

“In my 20s, I realized that my fear of men's sexual advances was so intense that it was **similar to the feelings of rape survivors**. Through talk therapy, I identified the main source of the trauma. When I was about 11, I underwent a **coercive, painful, and terrifying genital exam** performed while I was in **medical restraints** to evaluate if I had a vaginal opening. Being restrained while an adult man pushed something into my vaginal area was terrifying and painful. This was one of the worst experiences of my life,” said Lydia, 38 years old.

“Phlebotomists struggled to get labs from Joey as a child and he experienced **multiple episodes of fainting** during attempts to get his blood work, which resulted in emergency response protocols from the labs and hospitals multiple times per year. At times, they would draw his labs with a special pediatric team in the ICU and once a surgical port insertion was even considered. He eventually had to have **cognitive behavioral therapy** due to the frequent lab draws, which were rarely successful on the first attempt, along with **anxiety medication** prior to all lab work,” shared Louise who has a 20-year-old son with classic CAH.

“John, accustomed to testicular examinations during his visits, now faces regular sonograms with a parent in the room, which can be often **embarrassing**. Unfortunately, treating these tumors required a high dose of dexamethasone, again, bringing on the new challenges of acne, stretch marks, and severe weight gains, all of which affect John's **self-esteem**,” Virginia talked about the emotional and mental impacts her 16-year-old son faces.

CLASSIC CAH: IMPACT OVERVIEW

Frustration at the lack of understanding

"**I do not identify as intersex. I am a woman**. I have had doctors say things to me like 'in trans patients like yourself.' I am NOT trans, and **I do not want the horrific disease I experience to be reduced to 'atypical genitalia.'** I am embarrassed to even tell people I have CAH, because the first thing that shows up on the internet (including medical websites) mentions that girls will be born with 'masculine" genitals," wrote Jill.

"As an adult living with CAH, I experience so many issues with **emergency care and physicians not understanding** how to treat CAH in an emergency situation, not taking crisis seriously, and **sometimes even gaslighting me**, implicating that I am seeking pain meds or attention due to their lack of understanding. I have learned to be very vocal and have had to advocate for myself often, but when I am in crisis sometimes I am physically unable to," wrote Morgan.

"Multiple endocrinologists who have a lack of knowledge about CAH have **written off my weight issues as being due to my laziness, overeating, incorrect diet,** and not recognizing that I struggled with weight as a side effect of PCOS, hypothyroidism, or over-suppression at times from my lifesaving and necessary steroid medication. This **medical gaslighting has created major mental health issues** for me, which I still struggle with and address currently in therapy," shared Sabrina.

> **"My family doctor at that time told me that it would be very difficult for me to conceive with CAH, and I should just forget about having children. There was no referral to a fertility expert. Therefore, I have no biological children"** said Lesley, 67 years old. **"This has been a huge emotional regret for me to this day."**

TREATMENTS TODAY AND HOPES FOR TOMORROW

Current treatments for classic CAH focus on managing symptoms and preventing life-threatening crises. However, many patients reported struggling with side effects, complex medication schedules, and incomplete symptom control and going into crisis. These challenges take a significant toll on both their short- and long-term health, as well as their overall quality of life. Patients and families expressed an urgent need for safer, more convenient and more effective options.

“It’s very frustrating to have such limited options to try, and watch your daughter struggle on a daily basis, and not be able to do a thing to help her,” shared Johnette.

Meeting Poll: What medications or medical treatments have you or your loved one used (currently or previously) to treat symptoms associated with classic CAH?
Participants were asked to select all that apply.

The treatments below are ordered from most to least frequently used, as reported by patients and caregivers in the meeting (see Appendix 2 for meeting poll results).

- Glucocorticoids (prednisone, hydrocortisone, dexamethasone)
- Mineralocorticoid (fludrocortisone)
- Emergency use Solu-Cortef
- Salt supplements
- Surgery
- Antidepressants/anti-anxiety medications
- Other medications or medical treatments
- Treatment to improve height (growth hormone, puberty blocker, aromatase inhibitor)

Meeting Poll: What are the biggest drawbacks of your or your loved one’s current treatment? Participants were asked to select up to three.

The drawbacks below are ordered from most to least frequently cited, as reported by patients and caregivers in the meeting (see Appendix 2 for meeting poll results).

- Side effects
- Frequency of administration
- Only treats some, not all symptoms
- Requires significant time and effort; limits daily activities
- Limited availability or accessibility
- High cost or co-pay, not covered by insurance
- Not very effective at treating main symptoms
- Other

Side effects are a top concern

Throughout the meeting, it was clear that side effects are a huge challenge, not only in the short term but the long-term impact of steroid use and what that could mean for patients.

“The **steroid side effects plus the CAH itself were hell**,” wrote Heather. “I'm to the point of desperation for a treatment that isn't steroids.”

“A huge frustration is the lack of concern, knowledge, or interest in evaluating and treating **side effects** of long-term steroid replacement therapy on mental health, bone health, reproductive health, etc.,” shared Nicole, who has two adult children with CAH.

Guessing game of CAH management

Medication management for CAH relies on delayed lab results and fluctuating symptoms that can change rapidly. Without real-time tools to monitor sodium levels or other critical markers, patients and families are left navigating blindly, making treatment decisions based on incomplete or outdated information.

“That's the hardest part is with the medication, it's a **guessing game** and there's no way of knowing where you're at as far as blood work. It comes in too late and your symptoms can change so drastically,” shared Andrea whose son was misdiagnosed until he was 10 years old due to a false negative on newborn screening.

“CAH medication dosing relies on lab results, which should be back in five to seven business days. That is a **long time to be unwell** and waiting on results,” said Meredith, who has a 19-year-old son with classic CAH.

“The actual dosing is kind of like a **moving target**. We take a blood test early in the morning, once every three months or so to kind of get a general idea of what it might look like. We made an **educated decision** on what my dosing should be,” shared Ryan, 32 years old with classic CAH.

TREATMENTS TODAY AND HOPES FOR TOMORROW

Strict schedules have life-altering stakes

Medication schedules require constant vigilance, with every dose playing a critical role. Adhering to a precise treatment regimen creates another burden for patients and their families.

“Height, weight, bone health, fertility, mental health, all of it is wrapped into **whether our medications were on point** and equal to that, whether we were **disciplined enough** to follow the regimen that our parents and doctors laid out for us in detail,” said Marc, 32 years old.

"Our 18-year-old follows a **circadian dosing schedule** using hydrocortisone, which has been beneficial health-wise, but it's a **huge commitment**,” said John.

“I'm **constantly vigilant** about my **medication schedule**, my diet and my stress levels, fearing that any lapse could lead to manifested symptoms, an adrenal crisis, or even death. This constant state of alertness takes a **toll on my mental and emotional well-being**,” shared Alana.

Participants also noted uses of medical alert bracelets, alarms to take medications, and counselling or psychotherapy as the most common approaches beyond medications and treatments (see Appendix 2 for polling results).

Difficulty finding experienced healthcare professionals

Finding healthcare professionals with experience in managing CAH remains a significant challenge, especially when transitioning from pediatric to adult care. Specialists with expertise in CAH are scarce, often requiring patients to travel long distances to find knowledgeable providers or manage their care with limited support and expertise nearby.

“As a parent of two CAH children, who are now adults, we have noticed that there is a **huge difference** in the kind of care they received as children versus the care they receive as adults, with **not enough endocrinologists knowledgeable about how to treat adults with CAH and the unique problems they face as a result of CAH,”** wrote Nicole.

“Switching endocrinologist is incredibly scary,” shared Dawn, whose 18-year-old son has CAH.

CLASSIC CAH: HOPES FOR FUTURE TREATMENTS

Meeting Poll: Short of a complete cure, what top three specific things would you look for in an ideal treatment for classic CAH? Participants were asked to select up to three.

The treatment attributes below are ordered from most to least frequently cited, as reported by patients and caregivers in the meeting (see Appendix 2 for meeting poll results).

- Treatment without steroid side effects
- Prevent adrenal crisis
- Improve daily functioning
- Stop disease progression
- Treat fertility issues
- Prevent testicular adrenal rest tumors
- Increase energy and stamina
- Treat emotional symptoms

> **"We urgently need options, because we are suffering,"** said Sarah who lives with classic CAH.

In Their Own Words: Family and Patient Desires for New Treatments

Reduced steroid use	“Anything at all that could be done to **reduce the steroid dose** would be a huge improvement, particularly for overall health over a lifetime. When I think about young kids today with CAH, being able to take a much lower steroid dose over decades will make a big difference in their long-term health,” said Tim. “It would be **life-changing** to have a drug that allows us to lower or eliminate the hydrocortisone dose to **minimize long-term effects of daily steroid treatment**, things like obesity, short stature, osteoporosis, heart disease, and hyperglycemia. While I know I need to give my son these steroids to save his life, I can't help but feel like I'm contributing to a whole host of other health issues every time I administer his medications,” said Chelsea. "My adult child has had many side effects already from steroid usage and would **highly benefit from reduced dosage**, better maintenance, and an EpiPen-style emergency injection,” wrote Arthur. “An ideal treatment would be anything to **avoid daily steroids**. I worry about the impact the daily steroids may have on her **long-term health**,” said Natalie who has a 7-year-old daughter with classic CAH.
Auto-injector for adrenal crises	“If the [Solu-Cortef] injection was easier, like an **auto-injector**, so many people are familiar with those. It would be a **game changer** in activities and getting to go do things with the comfort of knowing that far more people would be able to and willing to do that injection in an emergency,” said Amanda. “The development of an auto-injector would just **change our lives**,” said Colleen, mother of two girls with classic CAH. "**Something like the EpiPen** would be great to make training so much easier. Thus, the lives of our entire family would open up drastically. The assurance we would feel knowing that the life-saving treatment is so much more **accessible** would be **life-changing** for all,” wrote Karina. “Having an **auto-injectable emergency drug** would be a huge peace of mind,” wrote Dawn. “**A simplified injector or an auto-injector** would give us so much more peace of mind that she'll be ok if she suffers a medical emergency, especially when she's at daycare or with other caregivers,” shared Kate, mother to a 3-year-old with CAH.

More dosing options

"A seemingly big effort was made to create Alkindi Sprinkles, the purpose of which is to allow for smaller dose adjustments for hydrocortisone to get more accurate doses for patients, particularly for kids that can benefit from minor dose adjustments to get to the right dose for their bodies. The need for these **more accurate doses** is there, however, Alkindi Sprinkles are **hard to administe**r for kids in their current formulation. It's hard to swallow and cannot mix with liquids for babies that need these smaller dosages and still tricky for toddlers as it is most easily taken on top of food to make sure it all ends up in the toddler's mouth, which is not easy to administer on the go. Why can't we just have smaller dose of hydrocortisone in tablet format like the current generics?" asked Jessica.

“It would be fantastic for a CAH person to have a **pump** in their body that could check their cortisol levels and **provide medication** when needed rather than doing it by clock or symptom. It would be fantastic if a CAH person could check their levels in minutes rather than weeks to make sure the appropriate dose is being given. Aside from a pump to regulate CAH, it would be ideal to have a **time-release pill** for CAH, much like ADHD medication,” said Meredith.

“I'd really like to see an **injection device that could be used to deliver preloaded hydrocortisone into your subcutaneous tissue**. The device would allow the milligrams delivered to be changed,” wrote Beth.

Better monitoring

"I hope there's a future where there is a **wearable continuous glucose monitor** equivalent to test hormone and/or electrolyte levels for CAH so we can be aware of a more constant basis of how his body is responding to his daily glucocorticoid treatment and adjust more rapidly," wrote Jessica.

“The biggest struggle is constantly trying to find that adrenal balance **“blindly.”** You have **no idea** what the cortisol levels are and you are dosing a bit intuitively. A corticometer would change a lot,” wrote Adriaan.

“It would be great to have a **test that measures the adrenal function** over a three-month period such as the A1C test that diabetics have. Currently we’re being monitored based on instantaneous blood sample results, which **don’t give the full picture**,” wrote Lieve.

“There's no **on-demand test** to know are you low on sodium like there is for people with diabetes and glucose. If there was something available **so you knew you were dropping,** it'd be a huge help,” said Ann, 55 years old.

“It's a **constant balancing act,** and the **more tools** that we have to control that are going to make the biggest difference in people's lives,” said Ben

INCORPORATING PATIENT INPUT INTO A BENEFIT-RISK FRAMEWORK

Benefit-risk assessment is the foundation for FDA's regulatory review of potential treatments. This sample framework, assembled based on input from the Classic CAH EL-PFDD meeting held on Oct 1, 2024, may help inform the FDA's benefit-risk assessment for urgently needed new treatments for CAH.

	Evidence and Uncertainties	Conclusions and Reasons
Analysis of Condition	Classic CAH is a complex and lifelong condition that affects nearly every part of a person's life. Beyond hormonal imbalances, it leads to significant physical and mental challenges, including fatigue, poor growth, dehydration, weight gain, anxiety/depression and a wide range of other debilitating symptoms. Living with classic CAH requires constant vigilance to adhere to a strict medication schedule and ward off potential adrenal crises.	Classic CAH can severely impact individuals' lives, often disrupting careers, limiting daily enjoyment, and reshaping lifestyles. Strict medication schedules and the threats of an adrenal crisis can severely restrict independence. The psychological effects of CAH, which can stem from issues such as a lack of understanding about the disease, medical gaslighting, and trauma from sensitive exams, are often as profound as the physical challenges, leading to stress, isolation, and emotional strain.
Current Treatment Options	While some treatments for CAH exist, there has been limited innovation in decades. Current options, including corticosteroids and mineralocorticoids, often provide only partial relief and come with significant short-term and long-term side effects.	Patients urgently want non-steroid treatment options, along with tools for more precise dosing and improved monitoring. Overwhelmingly, patients and parents wish for an auto-injector for treating adrenal crises.

CONCLUSION

CARES Foundation extends heartfelt gratitude to the patients and families who courageously shared their personal experiences with classic CAH during the EL-PFDD meeting, as well as to those who face the complexities of this challenging condition every day. Living with CAH impacts every aspect of life, presenting significant hurdles from short-term side effects to long-term health issues to the immense stress of emergency interventions. The mental and emotional toll of managing CAH is equally profound.

This community urgently needs better treatment options to ease these burdens and improve quality of life.

We hope the powerful stories shared during this meeting highlight the pressing need for change and serve as a call to action for advancing research and treatment development in CAH.

EL-PFDD Meeting Agenda

10:00AM – 10:05AM **Opening Remarks**
Dina Matos | *Executive Director; CARES Foundation*

10:05AM – 10:15AM **FDA Opening Remarks**
Theresa Kehoe, MD | *Director; Division of General Endocrinology*

10:15AM – 10:30AM **Clinical Overview**
Karen Lin Su, MD | *Pediatric Endocrinologist; New York Presbyterian-Weill Cornell Medical Director, CARES Foundation*

10:30AM – 10:35AM **Introduction and Meeting Overview**
James Valentine; Meeting Moderator | *Hyman, Phelps, and McNamara*

10:35AM – 10:45AM **Demographic Polling**
James Valentine; Meeting Moderator | *Hyman, Phelps, and McNamara*

Session #1: Symptoms and Impacts of Living with CAH

10:45AM – 11:15AM **Patient/Caregiver Panel #1**
A panel of patients and caregivers will provide comments to start the discussion on health effects and daily impacts of classic congenital adrenal hyperplasia.

11:15AM – 12:30PM **Audience Polling & Moderated Discussion**
Larry Bauer & James Valentine; Meeting Moderators | *Hyman, Phelps, and McNamara*
Patients and patient representatives are invited to add to the dialogue.

12:30PM – 1:00PM **Lunch**

Session #2: CAH Treatment and Hopes for the Future

1:00PM – 1:10PM **Treatment Overview**
Richard Auchus, MD, PhD, FACE | *Departments of Internal Medicine and Pharmacology; University of Michigan & Ann Arbor VA Medical Center*

1:10PM – 1:35PM **Patient/Caregiver Panel #2**
A panel of patients and caregivers will provide comments to start the discussion on health effects and daily impacts of classic congenital adrenal hyperplasia.

1:35PM – 2:45PM **Audience Polling & Moderated Discussion**
Larry Bauer & James Valentine; Meeting Moderators | *Hyman, Phelps, and McNamara*
Patients and patient representatives are invited to add to the dialogue.

2:45PM – 2:55PM **Meeting Summary**
Larry Bauer; Meeting Moderator | *Hyman, Phelps, and McNamara*

2:55PM – 3:00PM **Closing Remarks**
Dina Matos | *Executive Director; CARES Foundation*

APPENDIX 2 - POLLING RESULTS

DEMOGRAPHICS:

1: Are You:

Response options	Count	Percentage
A. Someone living with CAH	17	40%
B. A caregiver of someone with CAH	**25**	**60%**

2: Where do you currently reside?

Response options	Count	Percentage
A. US Pacific time zone	8	16%
B. US Mountain time zone	1	2%
C. US Central time zone	9	18%
D. US Eastern time zone	**32**	**63%**
E. US Alaska time zone	0	0%
F. US Hawaii time zone	0	0%
G. Europe	1	2%
H. Middle East	0	0%
I. Asia	0	0%
J. Canada	0	0%
K. Mexico	0	0%
L. Other	0	0%

3: What is your gender or the gender of your loved one with Classic CAH?

Response options	Count	Percentage
A. Female	**27**	**57%**
B. Male	18	38%
C. Other	2	4%

4: How old are you or your loved one with Classic CAH?

Response options	Count	Percentage
A. Less than 12 years of age	**14**	**30%**
B. 13-18 years of age	8	17%
C. 19-35 years of age	12	26%
D. 36-50 years of age	8	17%
E. 51-60 years of age	3	6%
F. 61-70 years of age	2	4%
G. 71 years of age or older	0	0%

5: How was Classic CAH detected in you or your loved one with Classic CAH?

Response options	Count	Percentage
A. Clinical signs/symptoms	**34**	**63%**
B. Newborn screening	20	37%

LIVING WITH CLASSIC CAH: SYMPTOMS AND DAILY IMPACT

1: Which of the following Classic CAH-related health concerns have you or your loved one ever had? Please select ALL that apply.

Response options	Count	Percentage
A. Atypical genitalia	26	10%
B. Dehydration/dizziness/excess urination	31	12%
C. Low energy/fatigue	**35**	**14%**
D. Adrenal crisis	34	13%
E. Poor feeding/poor weight gain	17	7%
F. Excessive weight gain	28	11%
G. Poor growth	22	9%
H. High blood pressure	15	6%
I. Irregular menstrual periods/infertility	12	5%
J. Testicular tumors	4	2%
K. Anxiety/depression	25	10%
L. Other	9	3%

2: Select the top 3 most troublesome Classic CAH-related heath concerns that you or your loved one has ever had.

Response options	Count	Percentage
A. Atypical genitalia	16	12%
B. Dehydration/dizziness/excess urination	13	9%
C. Low energy/fatigue	21	15%
D. Adrenal crisis	**32**	**23%**
E. Poor feeding/poor weight gain	3	2%
F. Excessive weight gain	20	14%
G. Poor growth	6	4%
H. High blood pressure	9	6%
I. Irregular menstrual periods/infertility	2	1%
J. Testicular tumors	3	2%
K. Anxiety/depression	9	6%
L. Other	5	4%

3: What specific activities of daily life that are important to you or your loved one are you/they NOT able to do or struggle with due to CAH? Select TOP 3.

Response options	Count	Percentage
A. Preparing meals/household tasks	2	2%
B. Loss of independence	14	14%
C. Sports or other recreational activities	16	16%
D. Restful sleeping	**18**	**18%**
E. Attending school	8	8%
F. Working or having a career	9	9%
G. Attending social events with family/friends	12	12%
H. Ability to have children	10	10%
I. Other	10	10%

4: What worries you most about your or your loved one's condition in the future? Select TOP 3.

Response options	Count	Percentage
A. Having an adrenal crisis	**30**	**28%**
B. Being mistreated in the ER	20	19%
C. Impacts on social/family life	11	10%
D. Inability to live alone	6	6%
E. Worsening symptoms	15	14%
F. Dying prematurely	13	12%
G. Ability to start their/my own family	12	11%
H. Other	1	1%

CURRENT AND FUTURE APPROACHES TO TREATMENT

1: What medications or treatments have you or your loved one used (currently or previously) to treat symptoms associated with Classic CAH? Select ALL that apply.

Response options	Count	Percentage
A. Glucocorticoids (prednisone, hydrocortisone, dexamethasone)	**35**	**21%**
B. Mineralocorticoids (fludrocortisone)	32	19%
C. Emergency use Solucortef	30	18%
D. Salt supplements	22	13%
E. Treatment to improve height (growth hormone, puberty blocker, aromatase inhibitor)	7	4%
F. Surgery	21	12%
G. Antidepressants or anti-anxiety medications	14	8%
H. Other medications or medical treatments	8	5%
I. Have not used medications or medical treatments	0	0%

2: Besides medications and treatments, what have you or your loved one used (currently or previously) to help manage Classic CAH? Select ALL that apply.

Response options	Count	Percentage
A. Physical/occupational therapy	3	3%
B. Medical alert bracelet	**29**	**31%**
C. Alarm to remember to take medications	25	27%
D. Acupuncture	3	3%
E. Massage	4	4%
F. Herbal supplements	3	3%
G. CBD	2	2%
H. Counseling or psychotherapy	17	18%
I. Other management approaches	7	8%
J. We are not doing anything to help manage CAH	0	0%

3: How well does your or your loved one's current treatment regimen treat the most significant symptoms of Classic CAH?

Response options	Count	Percentage
A. Not at all	1	3%
B. Very little	0	0%
C. Somewhat	**24**	**62%**
D. To a great extent	14	36%
E. Not applicable- not using anything	0	0%

4: What are the biggest drawbacks of your or your loved one's current treatment approaches? Select up to 3.

Response options	Count	Percentage
A. Not very effective at treating main symptoms	4	4%
B. Only treats some, not all symptoms	18	18%
C. High cost or co-pay, not covered by insurance	4	4%
D. Limited availability or accessibility	5	5%
E. Side effects	**36**	**36%**
F. Frequency of administration	23	23%
G. Requires significant time and effort; limits daily activities	6	6%
H. Other	3	3%
I. Not applicable as I am not using any treatments	0	0%

5: Short of a complete cure, what Top 3 specific things would you look for in an ideal treatment for Classic CAH?

Response options	Count	Percentage
A. Stop disease progression	8	7%
B. Prevent adrenal crisis	31	28%
C. Treat fertility issues	8	7%
D. Prevent testicular adrenal rest tumors	7	6%
E. Treatment without steroid side effects	**33**	**29%**
F. Treat emotional symptoms	4	4%
G. Increase energy and stamina	7	6%
H. Improve daily functioning	14	13%
I. Other	0	0%

APPENDIX 3 - SUBMITTED COMMENTS ONLINE

Organized alphabetically by first name

The following quotes are presented verbatim, as received, and may contain spelling or punctuation mistakes.

Name, country	Comment
Adriaan, The Netherlands	The biggest struggle is constantly trying to find that adrenal balance “blindly”. What I mean by blindly is that you have no idea what the cortisol levels are and you are just dosing a bit intuitively. A corticometer would change a lot. But, also a method that enables us to better manage that balance and get rid of the constant paranoia which is part of our lives.
Amanda, USA	Topic 1: Living with CAH Of all the symptoms of CAH which 1-2 symptoms have the most significant impact on your life? Nausea and lethargy How does CAH affect you on best and worst days? Describe your best days and your worst days. • Best- we don’t even know she has CAH aside from taking her meds every 6h. • Worst- high anxiety- she gets unbearable headaches that leave her barely able to lift her head. Nausea that causes vomiting, that we then have to treat and pray she keeps her meds down. And then the constant checking her for fever, treating the fever, making sure she’s drinking enough liquids to not get dehydrated. Are there specific activities that are important to you that you cannot do at all or as fully as you would like because of CAH? • She can’t just go to anyone’s house because of her CAH- I have to know the family, trust the family, and train the family on her injection. • I can’t just drop her off with anyone or anywhere. What is one thing that you would like people to know about CAH that is often misunderstood? • It’s a very serious condition- just because you wouldn’t know she had a medical condition 90% of the time doesn’t mean we’re not on pins and needles waiting for the ball to drop. How have your symptoms changed over time? • Her symptoms have evolved from infancy- she used to get a little nauseous and extremely lethargic. Now she gets debilitating headaches, worse nausea, and her whole body starts reacting with shakes, clammy, and hard to move. How has the ability to cope with the symptoms changed over time? • As a child, I think her ability to cope has stayed the same- except now she can verbalize what she’s feeling and we can treat her more easily based on specific symptoms.

Name, country	Comment
Amanda, USA	What do you fear the most as you get older? • Independence. Going out into the world without me and without people who know what to do surrounding her. What worries you most about your condition? • Changing symptoms as she starts puberty- her struggling more often because her levels are all over the place. What frustrates you most about your condition? • Lack of education in doctors and having to explain things all the time. Topic 2: Current Challenges to Treating CAH What are you currently doing to manage CAH symptoms? • Cortef 4x daily, florinef once daily. Carry the emergency injection everywhere we go. How well do these treatments treat the most significant symptoms of CAH? • Her body responds very well to cortef and we have been able to manage symptoms without using the injection more than once. What are the most significant downsides to your current treatments and how do they affect your daily life? • Taking meds every 6 hours is exhausting- especially as a parent having to stay up until midnight daily. We never get enough sleep. Short of complete cure, what specific things would you look for in an ideal treatment for CAH? • Delay-release meds to help get through the night would be amazing. And better research and access to the pump would be awesome. • Injection - we desperately need access to an auto-injector to train more people and make it easier for anyone to give an injection.
Arthur, USA	How does one become a participant in a study. My adult child has many side effects already from steroid usage and would highly benefit from reduced dosage, better maintenance and an epipen style emergency injection.
Beth, USA	I'd really like to see an injection device that could be used to deliver preloaded hydrocortisone into your subcutaneous tissue. The device would allow the milligrams delivered to be changed. In the UK prescribing a cortisone pump is routine and the quality of life is significantly improved. Millions of diabetics are using them and the idea that CAH patients would find them burdensome is ridiculous...

Name, country	Comment
Chelsey, USA	I have a 2-year-old son with salt-wasting classical CAH, my husband and I are both carriers. We recently became pregnant with a baby girl, and I (pregnant mom) did a dexamethasone treatment to try to prevent or minimize atypical genitalia at birth. I stopped treatment midway through pregnancy when we got her chorionic villus sampling genetic results back that she was unaffected by CAH. The dexamethasone treatment was considered highly experimental based on minimal data. The treatment was very stressful (not ideal for a pregnancy!) and had many side effects for me (mom). For future pregnancies, it would be really amazing if there were a more tested drug that you could take to minimize or eliminate the concern of atypical genitalia in carriers who are pregnant with baby girls.
	It would be LIFE-CHANGING to have a drug that allows us to lower or eliminate the hydrocortisone dose, to minimize the long-term effects of daily steroid treatment: things like obesity, short stature, osteoporosis, heart disease, and hyperglycemia. While I know I need to give my son these steroids to save his life, I can't help but feel like I'm contributing to a whole host of other health issues every time I administer his medications.
	It would be so much easier to manage CAH if there were a drug that we were able to administer twice daily, rather than three times daily. (Especially with younger kids who sleep 10+ hours at night, it's painful to have to wake them up in the middle of the night to administer their medications.)
	One of the things I worry about the most is that my son will have an adrenal crisis when he's alone or around people who aren't extremely familiar with his emergency Solu-Cortef injection. It's a really clunky, complicated process to administer the life-saving injection. It would be made SO MUCH easier in an auto-injection form, similar to an epipen, that most people are comfortable using in an emergency. Even my son's teachers and grandparents (who care for him regularly and have received training on the Solu-Cortef injection) are terrified that they'll not remember all the steps correctly if they ever have to use it in an emergency.
Corrine, USA	It's been hard as a parent of a toddler with CAH to have a "normal" career- I would always give up my career in order to take of my children if it came to it, but the income restriction affects the whole family and I'm noticing how different it is from my other colleagues with children. We don't feel comfortable being more than 5 minutes away from our child while they're starting pre-school, and thus we have to "switch off" when we work which severely impacts our income. As Colleen mentioned, after-school programs and YMCAs in our case don't have adequate or consistent medical oversight, so further limits our ability to have our affected child in daycare and after school programs. The injection training is complex, and we worry we will struggle with it when the time comes nevermind trusting others to do it with ease. Something like the epi-pen would make the training so much easier. Thus, the lives of the entire family would open up drastically. The assurance we would all feel that the life saving treatment is so much more accessible would be life changing for all.

Name, country	Comment
Dawn, USA	Thank you for hosting this session, it is so important for the CAH community and individuals living with CAH. I have many concerns around the current treatment of CAH and how it currently, and will, affect our 18-year-old son…Blood pressure, weight gain, foggy brain, bones, fertility, finding a mate who will accept you for having CAH, are just a few. As he is in a transitional stage of life, leaving the security of home and pediatric endocrinologist, and venturing into the world on his own, that brings a whole new round of fears. Finding a proper caregiver is extremely difficult, one that will work with you to be creative for an individual approach, and one that is well informed and resourceful about current and future treatments is difficult. Switching endocrinologist is incredibly scary. Also, the transition to college where he is now living on his own and is responsible for advocating for himself and caring for himself. We've given him everything possible to be able to enter this next stage of life, and it comes down to us having to trust him. But it's more about the trust of his community and healthcare providers where he is now going to school. Will they be able to respond properly during the crisis? Will they accept him for who he is and support him when needed? Having an auto injectable emergency drug would be a huge piece of mind. Also, because our son follows the circadian dosing and takes medication five times a day, having a proper time released drug that is not prednisone would be amazing. I'm so hopeful about the new drugs Neurocrine Is working on, lowering the androgens would help with several of the long-term worries that we have for our son. Even though CAH does not define our son, it does impact every aspect of his life, and ours. Thankful to the foundation for their support and advocacy. Along with the other pharmacist, they are willing to work to find better treatments for those living with CAH.
	Our 18-year-old follows the circadian dosing schedule using hydrocortisone, which has been beneficial health wise, but it's a huge commitment. He switched to prednisone before bed to cut out the 2:30 am dosing. It's helped sleep wise, but the prednisone isn't a great match for him health wise. He notices a difference in energy with the prednisone.
Diana, USA	Body The thing I fear most is my child having an adrenal crisis and not being able to get her the life saving shot. Children with CAH need to have an auto injector immediately. We have not felt safe sending our 13-month-old to daycare since she was born. We are ill equipped as non-medical professionals to draw up a needle in a crisis and certainly don't trust untrained day care staff. As a result, we have made the decision to have one parent leave the workforce. I have significant concern over our ability to get accurate treatment even with the best CAH Drs on our team. I hope drugs to treat this rare disease are prioritized.

Name, country	Comment
Dinara, South Korea	Hello, Thank you very much for sharing this video for those who could not participate live due to time zone. I am 43 years old, with classic virilizing CAH, not salt-wasting. I am on prednisolone for all my life. Now I take 5 mg divided to morning and evening. I think I did not have any side effects except bone density problems. I had minus 4 and had treatment. For about two years I start to experience hard fatigue and I find out it was connected to my periods. Here doctors do not know anything about CAH...and they even do not control my dosage. So the doctor said my fatigue was due to perimenopause or anxiety. I had several panic attacks finished in ER. So now I am approaching menopause, I had bleeding between periods because of adenomiosys so was put on Mirena. Now I have heavy fatigue days several times in a month and do not understand is it my periods, anxiety or adrenal crisis. I had adrenal crisis when I was undiagnosed at age 1. Can CAH be worsened when you age? Thank God I have children. I do not have emergency kit, doctors said they don't have it. How to differentiate adrenal crisis symptoms with menopause symptoms or with anxiety? Thank you very much!
Gino, Canada	Dear FDA Contact, Significant improvement in patient outcomes (adrenal diseases) will result from: Fast-tracking pediatric and parenteral auto-injector formulations of hydrocortisone. Lives are at stake. What is the FDA's marketing timeline and current update? Kindest regards and thanks
	The FDA must support the request to the US Senate Finance Committee to fund the stocking of injectable rescue glucocorticoid (hydrocortisone) to treat suspected adrenal crisis (diagnosed/undiagnosed patients). The FDA's review of auto-injector products currently under review must affirm the "minutes count"/"time critical" nature of PRN hydrocortisone. The availability of PRN hydrocortisone on ambulances and in the pre-hospital/hospital community setting will save lives. What action has the FDA taken to support the availability of this declared "essential medicine" in the community setting? Lives are at stake. Please endorse/support patient-advocacy groups to get EMS scope of practice directives to administer this life-saving medication (from ambulance or patient-carried supply). Please fast-track (with rigorous safety data) the availability of a hydrocortisone auto-injector to safeguard the lives of all adrenal disease patients at risk of death. The FDA must work collaboratively with ISMP to document all cases of delayed/omitted treatment of adrenal crises which have led to multiple deaths in the United States. The FDA is able to play a pivotal role as both industry advocate and regulator. We must work with pharma and patient advocacy groups to expedite policy wrt to new formulations, especially an auto-injector, to safeguard lives. Pre-hospital/hospital healthcare providers must align policy and recognize that all patients who present in suspected adrenal crisis must be treated without delay. Emergency treatment for adrenal crisis with a first-line medication affords a fundamental right to life.

Name, country	Comment
Gino, Canada	The restriction of CAH in some pre-hospital/hospital protocols DISADVANTAGES other patients with adrenal disorders who need emergency treatment. We must EXTENDED pre-hospital/hospital protocols to ALL patients with adrenal disorders needing emergency rescue injectable hydrocortisone. We must be vigilant in treating ALL patients at risk for suspected adrenal crisis (adrenal failure) not just those with CAH.
Gretchen, USA	Symptoms My son was born in a state without newborn screening; so, diagnosis came after weeks of medical sleuthing that came to a head in full blown adrenal crisis that was diagnosed in the ER pre- confirmatory lab results because by chance the gastroenterologist thought to engage an endocrinologist who happened to be an expert in CAH. The sign that something was wrong was vomiting. This led us to the gastro, tons of tests, and no solutions until that moment in the ER. With universal, comprehensive newborn screening, no family should ever have to go through that again. The symptoms of living with diagnosed CAH, however, remain very real and challenging. As a parent, you get to know your child. You notice when they seem off - not quite as sharp as usual, a little pale, overly sleepy - or smell a little funny. Experience with what could happen if these signs are ignored, means you ask how they feel, start hydrating, and consider the myriad possible outcomes. You try to balance their medical safety with not overreacting. You send them off to school or activities, but you ask those around them to keep an eye on them, or you go along (we kept one parent at home at all times to allow for this after trying a dual worker household and found it impossible). If symptoms progress to fever, pronounced lethargy, diarrhea, or any other textbook signs, you start stress dosing and planning for invoking emergency procedures. For my son, I believe the two most troubling effects of medication (not symptoms of the disease but rather results of the treatment), are weight gain and stunted stature. He fought his weight throughout childhood. In young adulthood, with change in medication (moved from HC to Dex to Pred) and control of his lifestyle, he is looking and feeling good for the first time ever. While he is taller than both his parents, he is two inches shorter than projected height. He will never forgive us for not proceeding with growth hormone and speaks often of going to Korea for leg lengthening surgery. While this sounds extreme and he is young, his sentiment is real. Activities There has never been anything my son could not do from an activity standpoint. There are things however he cannot do. He cannot join the military. He likely cannot fly a plane or take on a job where if he were to into adrenal crisis he would place others in harms way. In our experience thus far, the real limiting factor of living with CAH is he cannot live alone. While he is very self-aware and understands his medical needs well, he can readily go into depression, lethargy, or illness and without someone to cheer him up, get him to turn off his alarm and get out of bed, advocate for him in times of physical stress, or sense is he will not survive long.

Name, country	Comment
Heather, USA	1. Which CAH symptoms have the most significant impact on your life? Weight, sleep/hot flashes, acne, period regularity/lack of it. Prior to the clinical trial I'm in, I would also add energy level or the lack there of; mood/affect either from the meds or the CAH (I really don't know)
	2. How does CAH affect you on best and on worst days? Describe your best and worst days? Best days – there are some issues with anxiety and focus. My appetite wouldn't be out of control. And I'm awake/alert/have appropriate energy. Worst days - Certain times of month, I would get severe hot flashes at night like I'm going through menopause (and I'm not that old). The hot flashes started in my teens. I would also say that this impacts sleep. Hirsutism and weight were also bigger issues. And I struggle/d with mood/affect either because of (I don't know why?) Therapy and the clinical trial I'm in help a TON. But if it was just steroids I was taking at those very high doses, aggression, anxiety, and other things would be an issue. My appetite would be out of control. And I wouldn't have any energy. Also, regardless of the clinical trial, my hair loss can increase excessively, and the doctors say that is hormone related.
	3. What specific activities, that are important to you, that you cannot do at all or as fully as you would like because of CAH? I love going for walks outside. I have to be VERY careful with that with Texas in the summer. The heat seems to get to me more as I get older (severely cramping legs and other things if not managed). And I do have to be more aware of my salt intake especially in the warm weather. I miss very much not being able to go outside or having to be hypervigilant with the hot weather. I do go mall-walk and regularly go to the gym, but I really miss the outdoors.
	4. How have your symptoms changed over time? How has your ability to cope with the symptoms changed over time? My symptoms have changed over time. I was poorly managed prior to entering the clinical trial. Comparing before and after that clinical trial though, I would say that my mental health pre-trial was struggling. I had a mix of anxiety plus aggression going on and my energy was either quite high or sleepy. After the clinical trial, those symptoms have only been a mild issue for me. If I was still just on the steroid-only treatment, my ability to cope with the symptoms would be MUCH worse. (I wasn't on a good trajectory prior). I would also add that I tolerate heat/salt fluctuations less well though now.
	5. What do you fear the most as you get older? What worries and frustrates you most about your condition? That steroids are currently the only FDA-approved treatment option, and the steroids are NOT working for me. Also, the damage and long-term impact on my body of the steroids as well as the cost of new treatments. What worries and frustrates me the most? My body is just worn out, and I'm not that old. With all my other comorbidities and the CAH, I can't imagine what 50 or 60 is going to look like with the worn-out body. I'm now struggling with keeping track of all my medications from the poly-pharmacy, and that scares me. Another frustrating thing about my condition is that quality of care I receive from everyone except my endocrinologist is not wanting to factor in my CAH or won't consult with my endocrinologist when issues arise.

Name, country	Comment
Heather, USA	6. What are you currently doing to manage your CAH symptoms? I'm doing glucocorticoids plus a phase 3 CAH clinical trial drug. I also see my doctors regularly, exercise, and work on my anxiety/ stress (not sure if that last one helps but I'm listing it).
	7. How well do these treatments address the most significant symptoms of your CAH? These treatments address my symptoms quite well. I wish I could stop the steroids entirely. I will add that I struggle with acne now and didn't before, but that isn't a deal breaker (and working on treating the acne).
	8. What are the most significant downsides to your current treatments and how do they affect your daily life? Downside of current treatment? I'm in a clinical trial right now, and it has been a godsend. My steroids have been reduced, and I've noticed improvement in weight, mental health, energy, and hirsutism. Prior to that trial, my body just kept requiring higher and higher doses of steroids. And the steroid side effects plus the CAH itself were hell. I would gain weight constantly, have a hard time with sleep due to hot flashes, be ALWAYS hungry, have energy levels that varied, and had no periods.
	9. Short of a complete cure, what specific things would you look for in an ideal treatment for CAH? An affordable and effective treatment that would allow me to severely reduce or use something besides glucocorticoids. I think I'm to the point of desperate for a treatment that isn't steroids.
JB, USA	So many of the participants meeting seems to me to they have been under poor or not ideal endo tx in their lives. Many adult endo's have no experience treating CCAH patients and they don't seem to ck in with the endo's that study CAH the experts ie what are the ideal labs to be drawn and what are the levels the experts recommend. Even today on FB pages it seems parents are not being given proper info for their child ie stress dosing and its 2024. It's too bad the same drugs are being used as they have been for the last 70 yrs. You don't see this with other med conditions i.e., thyroid, diabetes, heart disease. Why is this? Because there are not enough CAH patients for drug companies to develop new drugs which is costly and if they can't make money on a drug why develop one.
Jes, USA	Pill splitting is a key characteristic to consider for hydrocortisone manufacturers. Young children need smaller doses than 5mg which is the smallest pill size. Some manufacturers produce pills that are too small to split, explode when split, or do not split cleanly creating unequal dosing. The brand Cortef has tended to be the best for us, but costs more because of the name. The sprinkle is an alternative solution, but pursuing the sprinkle would incur more unwanted run-arounds with insurance and bills. I'd like to add a few thoughts on health concerns that were not mentioned or listed. Our 5 year-old daughter frequently has symptoms of headaches and nausea. About once a week this changes our dinner routine or evening sports attendance. During family vacations, this results in a day the family has to take a slow day.

Name, country	Comment
Jessica, USA	As the parent of a child with CAH, I worry about my child being in a situation without me where they need emergency medication (for example, a broken bone or concussion from sports activity) and no one around them can figure out how to use it... Our "epipen" equivalent for life or death emergency situations (called Sol-u-cortef actovial) is complex to set up for use. There has to be an easier delivery method for emergency medications.
	One of the things I worry most about as a parent is the fact that we can only check indicators of how my son's body is performing when we go in for blood tests in a lab every few months. I hope there is a future where there is some wearable "continuous glucose monitor" equivalent to test hormone and/or electrolyte levels for CAH so we can be aware on a more constant basis of how his body is responding to his daily glucocorticoid treatment, and adjust more rapidly.
	A seemingly big effort was made to create Alkindi Sprinkles, the purpose of which is to allow for smaller dose adjustments for hydrocortisone to get a more accurate dose for patients, particularly for kids that can benefit from minor dose adjustments to get to the right dosage for their bodies. The need for these more accurate doses are there, however, Alkindi Sprinkles are hard to administer for kids in their current formulation. It's hard to swallow and cannot mix with liquids for babies that need these smaller dosages, and still tricky for toddlers as it is most easily taken on top of food to make sure it all ends up in the toddler's mouth, which is not easy to administer on the go. Why can't we just have a smaller dosage of hydrocortisone in tablet format like the current generics?
Jill, USA	Please be cognizant of the language you use. I do not identify as intersex. I am a woman, and I am tired of my feminine being reduced to the size of clitoris. I have had doctors say things to me like “in trans patients like yourself.” I am NOT trans, and I do not want the horrific disease I experience to be reduced to “atypical genitalia.” I am embarrassed to even tell people I have CAH, because the first thing that shows up on the internet (including medical websites) mentions that girls will be born with “masculine” genitals. My entire life experience is then swallowed up by people wondering if my clitoris is big. Please be cognizant in the way you discuss and educate people. All day, every day, I struggle with fatigue, both physically and mentally. I routinely have headaches and GI distress. I am under 35 and I have severe osteoporosis. I have had joint surgeries. I live in almost constant pain. I would like to be treated with dignity and respect.
Johnette, USA	My daughter has had significant issues with being unable to obtain brand name Millipred 5mg. This was discontinued and she has had to switch to Prednisone 5mg. She has noticed significant decrease with her energy level, experienced more frequent dizziness, and overall hasn’t felt as good as she did while taking Millipred. It’s very frustrating to have such limited options to try, and watch your daughter struggle on a daily basis, and not be able to do a thing to help her. Things NOT quality healthcare for my SW CAH daughter.

Name, country	Comment
Julie, USA	I am a pediatric RN in Seattle and see the impact delayed medications for CAH have on patients. I also live in a rural area where EMS can be an hour from bringing a patient to a children's hospital. Rapid medical interventions can change the outcomes for CAH patients. Funding is needed for further development of medications and treatments to support individuals with CAH to living healthy lives within our communities.
Kate, USA	As the parent of a 3-year-old with CAH, I want to acknowledge that Alkinki Sprinkle has made our lives markedly easier. It's so much simpler to make sure she's getting the proper hydrocortisone dose both at home and at daycare vs. trying to cut up crumbling pills. Now we're hoping for a simpler alternative to Solu-Cortef. The current process is complicated and extremely intimidating to people not familiar with it. It's not ideal under the best of circumstances, let alone with the stress of an emergency situation. A simplified injector or an auto injector would give us so much more peace of mind that she'll be ok if she's suffers a medical emergency, especially when she's at daycare or with other caregivers.
Lieve, USA	Better lab testing would help. It would be great to have a test that measures the adrenal function over a 3-month period such as the A1C test that diabetics have. Currently we're being monitored based on instantaneous blood sample results, which don't give the full picture.
Lydia, USA	I have salt wasting classic CAH. When I am in an adrenal crisis and go to the hospital I am worried that I won't receive the mineralcorticoid support that I need because it seems like the doctors focus on cortisol support. I worry that if I am unconscious and can't advocate for myself that I won't get fludrocortisone and will die of heart problems.
	A finer division of dosage options for hydrocortisone and fludrocortisone would really help fine tune the treatment for patients, especially children. If levothyroxone can be given at microgram level precision then fludro and hydro should be able to be delivered at a much wider, more finely divided range of dosages.
	Many videos and callers have mentioned keeping extra CAH medication with them. I also keep extra pills with me. I keep three whole days worth of meds with me so that even if I was stranded somewhere without any meds in my normal week's pill case I'll still have three days of medicine.
	The challenges I have in accessing solucortef is a worry for me. It takes weeks, sometimes more than a month for the ordering pharmacy to receive the solucortef. Additionally, I often worry that sterile paper package that the needle is in will have broken open during storage in my purse but I'll have to use it anyway in an adrenal crisis and risk an infection after injection.
	A major improvement in CAH treatment would be a better understanding of the causes of adrenal crisis in the life of a CAH patient. In my experience, adrenal crisis can arise seemingly out of nowhere even if I've been taking all my doses. It's unclear to me what can cause adrenal crisis onset aside from major illness or injury. Something that could minimize the risk of adrenal crisis without taking more glucocorticoids or mineralcorticoids would be helpful.

Name, country	Comment
Morgan, USA	As an adult living with CAH, I experience so many issues with emergency care and physicians not understanding how to treat CAH in an emergency situation, not taking crisis seriously, and sometimes even gaslighting me implicating that I am seeking pain meds or attention due to their lack of understanding. I have learned to be very vocal and have had to advocate for myself often, but when I am in crisis sometimes I am physically unable to.
Natalie, USA	CAH symptom with greatest impact: my daughter (7 years old) has a very hard time with stomach bugs. She gets stuck in a cycle of vomiting and having trouble keeping oral hydrocortisone down. An injection of solu-cortef usually is enough to get her out of it but I worry about it quite a bit (coming on too quickly, when I'm not around what if the responsible adult panics, forgets instructions, etc.). An ideal treatment would be anything to avoid daily steroids. I worry about the impact the daily steroids may have on her long term health. Biggest concern for future: fertility
Nicole, USA	As a parent of two CAH children, who are now adults, we have noticed that there is a huge difference in the kind of care they received as children versus the care they receive as adults, with not enough endocrinologists knowledgeable about how to treat adults with CAH and the unique problems they face as a result of CAH. A huge frustration is the lack of concern, knowledge, or interest in evaluating and treating side effects of long term steroid replacement therapy on mental health, bone health, reproductive health, etc. Also the lack of development and accessibility of potential new therapies/treatments/medications coming to market for patients.
Raelie, USA	CAH PFDD Meeting: Raelie's Answers (10yo- SWCAH) Topic 1: Living with CAH Of all the symptoms of CAH which 1-2 symptoms have the most significant impact on your life? • Nausea and headaches How does CAH affect you on best and worst days? Describe your best days and your worst days. • Best Day: I need to take medicine. • Worst Day: My head was pounding. I felt like I was going to throw up. I was really hot. I was shaky. It was kind of hard to get up because I was like shaky and my legs were not working right. My whole body hurt and it was hard to move. I had to crawl over to my parents because I felt like I was going to throw up so much. It went away after taking triple doses. Are there specific activities that are important to you that you cannot do at all or as fully as you would like because of CAH? • Sleepovers

Name, country	Comment
Raelie, USA	What is one thing that you would like people to know about CAH that is often misunderstood? • It is very life threatening if I don't take my medicine. It's my #1 because it is one of the most important things about it. (And also because I live my life and no one would know that I have CAH so it doesn't seem like I'm that sick) How have your symptoms changed over time? • My headaches and nausea have gotten a lot worse. How has the ability to cope with the symptoms changed over time? • I've gotten better with dealing with headaches even though they've gotten worse so it's kind of stayed the same. What do you fear the most as you gets older? • Going into a crisis. What worries you most about your condition? • Going into a crisis. What frustrates you most about your condition? • Having to stop things to take medicine. Constant bloodwork. Topic 2: Current Challenges to Treating CAH What are you currently doing to manage CAH symptoms? • Taking medicine- cortef and florinef How well do these treatments treat the most significant symptoms of CAH? • Really well. What are the most significant downsides to your current treatments and how do they affect your daily life? • Having to take it so often is a downside. Having to stop playing and doing activities because of it. • Injection: how long it takes to prep it and put it together. It's hard to do. Short of complete cure, what specific things would you look for in an ideal treatment for CAH? • Having only to take medicine once a day. Better access to a cortisol pump.

Name, country	Comment
Sabrina, USA	As an adult female with CAH, I have struggled with my weight, disordered eating, and body dysmorphia for quite a while. Most of this is due to multiple endocrinologists in my adult years who have a lack of knowledge about CAH and have written off my weight issues as being due to my "laziness", over-eating, incorrect diet, and not recognizing that I struggle with weight as a side effect of PCOS, hypothyroidism, or oversuppression at times from my life saving and necessary steroid medication. This medical gaslighting has created major mental health issues for me, which I still struggle with and address currently in therapy. I do feel like treating CAH includes treating the whole person-mentally and physically, and not being so quick to write off symptoms and side effects as poor lifestyle choices.
Sarah, USA	I was born in Canada in 1993 (Toronto) and there was no standard newborn screen. I was born with pubic hair, but apparently they didn't find that to be abnormal. At age 2, I had an adrenal crisis and was diagnosed with SV CAH. I had a very tumultuous adolescence as my CAH was poorly managed, and I was overweight and didn't develop similarly to my peers. I started birth control very early on to manage my inconsistent menstruation. I ended up developing vulvodynia from OCPs and required surgery. In order to become pregnant I needed to take high dose steroids, which has left me with major bone health issues. My child is healthy and I'm thankful I was able to successfully conceive and breastfeed, but it has left major complications in my health and made me gravely concerned for my future. More than anything, I am angry. There are so many options available to those living in different parts of the globe (cortisol cycles, ER hydrocortisone, pumps, etc.) I am so mad that I am held back from treatments that could improve my quality of life drastically, because the system doesn't see my life as a priority. We urgently need options, because we are suffering.
Stephanie, USA	As a parent of a young adult with SWCAH, I am very concerned about the long-term issues with steroids... including hard-to-control weight gain, bone issues, etc.
Stephanie, (Boston) USA	My family has had several times when pharmacies have mistakenly given vials of Solu-Cortef instead of Solu-Cortef Act-O-Vials, even though the prescription was written correctly. This mistake can be deadly. The problem is with the packaging being exactly alike. Solu-Cortef Act-O-Vials have a liquid and powder while the Solu-Cortef has only the powder. The powder alone is useless in an emergency situation and a patient could die if unable to inject the medication. Unless a patient opens the box when the prescription is filled, as my family does now, they may not have the medication needed in an emergency. The NDC for Act-O-Vial is 0009-0011-03 and the NDC for Solu-Cortef is 0009-0825-01. The boxes' only difference is the very small printed "Act-O-Vial." Please make sure the boxes are changed and have a bigger difference. Thank you!

Name, country	Comment
Stephanie, (Fairhaven), USA	My 13-year-old daughter has salt wasting CAH. The greatest hurdle we've had since she was born is related to her IM solu-cortef stress dose. She was unable to attend camp during the summer, a number of day cares, some friend's houses,etc. all because her medication needs to be drawn up, rather than having an auto-injector like an epi-pen. She older now, but it's stressful as a parent when administering her IM injection independently would be so difficult for her when she out with friends. Her friends wouldn't be able to help and she may have difficulty. If it was an auto-injector, she could easily do it, her friends could do it, most people in the vicinity could assist... please consider this and help give these kids more independence and the caregivers less worry. Thank you.
Teri, USA	Which CAH symptoms have the most significant impact on your life? My cortisol levels when I am stressed or feeling sick. This isn't a day-to-day concern though, which is good. How have your symptoms changed over time? Not really, my levels have been controlled for most of my life, so the only time I realize I have CAH is when I need to get bloodwork, see my endocrinologist or need stress dosing when I am sick How has your ability to cope with the symptoms changed over time? As I have gotten older, I can tell more easily and quickly that I need an extra dose or two, which allows me to stop the symptoms before they become bad. What do you fear the most as you get older? I am not sure how my lifelong usage will affect my health as I get older. What worries and frustrates you most about your condition? The lack of knowledge among health care professionals about CAH. I have had to explain CAH to most if not all my providers throughout my life when I first meet them. What are you currently doing to manage your CAH symptoms? Take Cortisol and Florinef daily, bloodwork and visits with endocrinologist every 6 months. How well do these treatments address the most significant symptoms of your CAH? They treat all of my symptoms as of this time Short of a complete cure, what specific things would you look for in an ideal treatment for CAH? Longer half-life cortisol so only need to take once a day.

Name, country	Comment
Thyra, USA	My son's best friend has CAH and so we have all been trained on administering his life-saving shot. He has been on many medications over the years and is currently one that is really helping with much less side effects like mood swings, listlessness and weight gain, so his quality of life is the best it has ever been thanks to the drug advancements. We encourage funding for continued research into both the cure and better medications.
Valerie, USA	Treatment helps somewhat, mostly in the day-to-day. But long term effects, such as lack of growth, symptoms of excess androgens, aren't helped as much with current treatments.
	One of my CAH sons is currently on hydrocortisone, 3x per day, fludrocortisone 1x per day, growth hormone shots, and we've just had to add Arimidex for advancing bone age, even though he has been very well managed. So because we are dealing with side effects or issues from one medication, we have to add others to treat the symptoms and side effects.
Wyatt, USA	I was born with SWCAH. When I was in first grade I went into adrenal crisis and received frontal brain damaged from the seizures. I have fully recovered from the adrenal crisis and gotten off of seizure medication. But CAH still affects my life. I take daily medication every day for SWCAH. With the development of this new drug if I was allowed to use it I could decrease the amount of medication I take resulting in a better long-time health for me.