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Original Article

An urgent need for early diagnosis and universal health care: insights from a series of interviews with parents of children living with congenital adrenal hyperplasia in Indonesia

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Highlights

- Families of children with CAH experience significant challenges associated with late diagnosis.
- Challenges reported by parents include emotional stress, financial costs and social stigma.
- CAH communities provide significant family support including accessing medications not covered by the national health insurance.

Abstract. Congenital adrenal hyperplasia (CAH) presents significant health challenges and requires a timely diagnosis and comprehensive treatment. This qualitative study assessed the experiences of parents of children with CAH in Indonesia, and focused on the challenges associated with delayed diagnosis. In-depth interviews with 40 parents of children with CAH from 9 Indonesian provinces were conducted between December 2022 and January 2023. The results revealed parents experienced challenges due to the absence of a newborn screening program (NBS) and the minimal capacity of healthcare professionals to diagnose CAH. Parents reported having emotional stress, financial challenges, and social stigma. Fludrocortisone and 17-OHP are not covered by the national health insurance, thus financial challenges prevailed. The impact of late diagnosis was also notable in their children; parents reported that their children had tendencies to self-isolate, insecurities, temperamental behavior, and masculine behavior (for females). These findings emphasize the critical need for the NBS to implement early diagnosis, increase healthcare professionals' capacity to diagnose CAH, and ensure accessible and affordable healthcare policies for patients with CAH. Addressing these gaps is essential for improving the quality of life for children with CAH and their families in Indonesia.

Key words: congenital adrenal hyperplasia, newborn screening, delayed diagnosis, universal healthcare, challenges

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Introduction

Congenital adrenal hyperplasia (CAH) is a genetic disorder characterized by impaired cortisol synthesis due to deficiency of certain enzymes involved in adrenal steroidogenesis. Globally, the incidence of classical CAH is estimated to be 1:10,000–1:18,000 per live births, whereas the non-classical form is more common (1, 2). Classical CAH, most often attributed to a 21-hydroxylase deficiency, is further divided into 2 classifications based on the degree of enzyme deficiency: salt wasting (SW) and simple virilizing (SV). Patients without the capacity to produce aldosterone experience a salt-wasting crisis, also known as adrenal crisis, which, when left untreated, can cause mortality within the first few weeks of life. Classic simple virilizing has a residual enzymatic activity of 1–2%; therefore, sufficient aldosterone is produced, and salt wasting does not occur (3). However, hyperandrogenemia, oligoovulation, and infertility can occur in adolescence or adulthood (4, 5).

Universal newborn screening (NBS) has been the norm in high-income countries such as the United States of America, Japan, and most European countries, although developing countries such as Indonesia, do not have this screening. As Indonesia is currently still in the process of establishing an NBS program for CAH, the exact incidence of CAH in Indonesia is unknown and cases are often undiagnosed. However, a pilot study with several hospitals in Indonesia found two positive cases out of 1226 patients screened (6), which reflects the possibility of a higher incidence than that reported in many countries. Due to adrenal crisis in babies with salt-wasting CAH and sex development disorders in children with simple virilizing CAH, late diagnosis can negatively impact patients and their families because of sex misalignment or other preventable morbidity and mortality (7, 8). Therefore, NBS for CAH should be prioritized in developing countries, including Indonesia.

CAH presents unique challenges to individuals, affecting not only their endocrine system, but also their psychosocial well-being. The interplay between genetic, hormonal, and socio-environmental factors potentially contributes to various psychological aspects that deserve closer investigation. Understanding the psychological impacts of CAH is crucial for developing comprehensive healthcare approaches that address the holistic needs of affected individuals.

To our knowledge, this was the first study to report the experiences of families with patients diagnosed with CAH through a series of in-depth interviews that assessed the challenges associated with delayed CAH diagnosis in Indonesia. Although previous research has primarily concentrated on the effects of CAH on the patients themselves, this study shifted focus toward understanding parent perspectives. By elucidating the impacts of late CAH diagnosis, this study aimed to strengthen the justification for urgent development and expansion of CAH NBS as a national program, as well as actions to improve CAH diagnosis and management in Indonesia, encompassing both medical and social aspects.

Methods

Methodological approach

This study used a qualitative research design approach. The study protocol was approved by the Faculty of Medicine Ethics Committee of Universitas Indonesia-Cipto Mangunkusumo Hospital (protocol number: 22-10-1221).

Data collection and procedures

The target population was identified from the Komunitas Keluarga Hiperplasia Adrenal Kongenital (KAHAKI) database, Dr. Cipto Mangunkusumo National Central Public Hospital, Jakarta, and National Diponegoro Hospital, Semarang, Indonesia. The recruitment process was conducted in December 2022. The inclusion criteria were: parents or caregivers of children with CAH; age ≥ 18 yr; and willingness to participate in the study. The research team sent an invitation to participate in the study via a telephone message. Candidates were allowed to ask questions about the study before deciding to participate. Eligible participants interested in the study were scheduled for an interview. Participants could opt in or out of the study at any point with reassurance that this would not impact their future care. Participants were informed of the study objectives and risks prior to participation and signed a consent form. After signing the consent form, each participant was interviewed using a Zoom or WhatsApp video call.

All the interviews were conducted between December 2022 and January 2023. The duration of each interview was 45–60 min; they were digitally recorded with the participants' consent. After questions about participant demographics, 5 key topics comprised the interview questions: challenges, daily life adjustment to incorporate CAH management, emotions, CAH management resources, and support. A semi-structured approach was used during the interviews to explore key topics and allow novel responses to emerge. Participants were given a copy of the "CAH in Indonesia" booklet after the interview.

Data analysis

The participant demographics are displayed using descriptive statistics, and data are presented as numbers and percentages. A thematic analysis method was used to identify, organize, and analyze the similarities, differences, and outliers of the dataset based on the generated themes.

Results

Participant demographic profiles

Of the 40 participants in this study, 36 were mothers and 4 were fathers of at least 1 child with CAH. They were from 9 provinces, the majority from the Central Java Province (60%). The participants' highest education levels were senior high school (37.5%) and undergraduate (32.5%) (**Table 1**).

One mother had 2 children with CAH; thus, there were 41 children with CAH (33 girls, 8 boys) of the 40 participants. The majority (23/41; 56%) of the children, both males and females, were diagnosed with CAH when older than 1 mo. All the children were diagnosed with 21-hydroxylase deficiency CAH based on laboratory and clinical findings (**Table 2**). A detailed overview of themes, categories, and key quotes from the parent interviews is provided in **Table 3**.

Biological challenges

Physical challenges: Children with CAH experience physical changes including early puberty, menstruation delays, incorrect sex assignment at birth, dark skin, testosterone increase in girls, and developmental delay mostly as a result of late diagnosis (diagnosis at ≥ 1 mo of age) and treatment.

Challenges due to late diagnosis: Among the 41 children, the average age at initial diagnosis was 21 d for girls and 31 d for boys. Only 1 girl underwent newborn screening paid by the parents. The remaining were diagnosed with ambiguous genitalia, persistent vomiting, or dehydration.

The major challenges expressed due to late diagnosis included parents having to endure confusion about their child's illness without a clear diagnosis. This problem was exacerbated by the minimal capacity of primary healthcare centers to diagnose CAH early. Even after the diagnosis, many parents felt it was difficult to find resources and acquire adequate treatment. When asked whether early diagnosis would have changed their child's current condition, 17.5% answered yes, 17.5% answered maybe, 12.5% answered no, and 52.5% answered not applicable because their children were diagnosed early.

Psycho-social challenges

A notable percentage of the participants (20%) chose to discontinue their current employment, diverging from their initial career paths or professional ambitions when their child was diagnosed with CAH. The same percentage of participants struggled financially because of CAH-related expenses.

Table 1. Participant characteristics

Characteristics	N (%)
Participant relationship with the patient(s) with CAH (n = 40)	
Mother	36 (90)
Father	4 (10)
Province of participant home address (n = 40)	
Central Java	24 (60)
West Java	5 (12.5)
Banten	2 (5)
South Sumatra	2 (5)
Special Capital Region of Jakarta	2 (5)
Special Region of Yogyakarta	2 (5)
Central Kalimantan	1 (2.5)
Jambi	1 (2.5)
Lampung	1 (2.5)
Participant highest level of education (n = 40)	
Undergraduate	13 (32.5)
Senior high school	15 (37.5)
Junior high school	5 (12.5)
Primary school	7 (17.5)

Table 2. Children demographics

		Sex		Total N (%)
		Females N (%)	Males N (%)	
Age at diagnosis	< 1 mo	15 (45.5)	3 (37.5)	18 (43.9)
	≥ 1 mo	18 (54.5)	5 (62.5)	23 (56.1)

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Table 3. Themes, categories, and quotes from the parent interviews

Themes	Categories (Questions)	Quotes from interviews
Biological challenges	Physical challenges for children with CAH (Can you briefly describe key challenges that your child had experienced since the diagnosis?)	*"This child…development is also different. As they get older, their skin gets darker, and their genitals also get darker and bigger."* (Participant 36)
	Challenges due to late diagnosis and how early diagnosis would have made a difference (Do you believe that if your child was diagnosed at an early age (newborn) your circumstances would be considerably different?)	*"If my child was diagnosed early, maybe she would not have to be severely sick and have confusions about what's happening to her. Early diagnosis would have helped us accept our child's conditions."* (Participant 10)
Psychosocial challenges	Parental impact (How did your child's diagnosis impact your life?)	*"When my child was first diagnosed, I had to be very careful and make sure the child was given medication really intensely. I had to leave my job, so I had to give up my dream of becoming a teacher for a long time."* (Participant 20)
	Mental or psychological problems of children with CAH (How do you think living with CAH has influenced your child's mental wellbeing?)	"*Yes"* (the participant felt that the child had a psychological problem). *"Friends often come to the house, if they are familiar, they are familiar. If you're a new friend, it's hard to make friends. Even my child often cries when forced to go out."* (Participant 9)
Emotions	Parents' prominent emotions related to CAH diagnosis (What were the prominent emotions you experienced since the CAH diagnosis?)	"*I'm worried that when my child goes to school and has to take medicine every day, many people will ask questions and not all of them will understand. How do I explain it?"* (Participant 31) *"At diagnosis, I couldn't understand. Even pediatricians in my city don't know anything about CAH, especially as a housewife, at first it was quite difficult for me to understand. But because in every check-up session I was given an explanation and a guidebook, now I understand better."* (Participant 12) *"I am worried about the child's future fate and how he will live in society."* (Participant 12)
Financial impact and adjustment to daily life	Daily life adjustment to incorporate CAH management (How have you managed to accommodate the diagnosis of your child's disorder with your daily life?)	"*My child has to drink her medicine 3 times a day, after they wake up, before sleep, and during the day. Each month I also have to accompany them for their routine consultation to the hospital.*"(Participant 2)
	Strategies to ensure effective management of children with CAH (What strategies do you have in place to ensure effective management for your child?)	"*I work together with my husband to take turns giving medicine to my child. In the morning, I give the medicine before going to work. In the afternoon, my husband would give it. We reminded each other.*" (Participant 7)
	Socio-economic status (How does your living with CAH impact your family financially?)	*"The national insurance (BPJS) does not cover fludrocortisone. So we have to obtain the medicine in Semarang, which we do not have the money to do so. During the pandemic, the price of fludrocortisone increased almost twice from "IDR 7,500.00 to IDR 12,000.00, but fortunately at that time the doctor got a donation to help parents get fludrocortisone.*" (Participant 12)
	Difficulties in making monthly hospital visits (How does routine hospital visits affect your family?)	*"Hospital visits are a big hassle for my family. I have other young children. I can't leave for too long and I don't have free time. It's far more convenient for us to pay out of pocket for additional hydrocortisone so we don't have to go through the hassle of hospital visits each month."* (Participant 18) *"Coming from Pati, a small city in East Java, it takes us 2 hours by car to reach Semarang, where my child goes for checkups. The transportation cost is huge. We are only able to go for checkups and get hydrocortisone every 2 months. We pay for the extra month of prescription out of pocket as it is not covered by the NHI."* (Participant 12)
	Difficulties in adhering to national insurance requirements (How is your experience with the national insurance system in Indonesia?)	*"Consultations with pediatric endocrinologists are covered by NHI, but as their service is only available in referral hospitals, it is very time-consuming to fulfill the requirements for the referral as we need to start at the primary healthcare center. Therefore, we chose to pay out of pocket."* (Participant 20)

Table 3. Continued.

Themes	Categories (Questions)	Quotes from interviews
Resources for CAH management	Knowledge/ Educational Resources (Do you feel that you have adequate knowledge/ education about CAH/management?)	*"No, I feel that I have no adequate knowledge, only for the basic routine treatment (if her child gets fever, daily drugs)."* (Participant 4)
	General healthcare facilities/medicine (What resources do you wish you had access to?)	*"I wish more doctors who are competent to identify manifestations of CAH so it could be diagnosed as soon as possible."* (Participant 2) *"I hope that in the future, more primary care doctors are competent and have an understanding about CAH so no child will suffer the consequences of late diagnosis as my child did."* (Participant 2)
Social	Family or social support (Do you have adequate family support?)	*"Unlike in big cities where your problem is only your own concern, living in a village is really different. Everyone is involved in everyone else's business. I want to explain my child's CAH diagnosis to my family members, but I'm confused as to how and I'm worried of the discrimination he might face if they find out. It's hard, I'm worried about the things they will say. So, it's been 2 years since my child's diagnosis and my family are still largely unaware of the condition."* (Participant 14)
	Support from the CAH community (Are you a part of any community groups? If yes, has this helped you in coming to terms with your child's disorder?)	*"From my experience, when at the hospital outpatient clinic, everyone tends to be more closed off, therefore neither I nor my child socializes with other patients. Her main source of support in the CAH community is her pediatric endocrinologist."* (Participant 40).
	Daily life adjustment to manage children with CAH (How have you managed to accommodate the diagnosis of your child's disorder with your daily life?)	*"Take the time to remind your child when it's time to take their medicine and when your child is 8 years old, like now, your child will be independent and used to taking their own medicine."* (Participant 12)

In addition, to financial stress, some participants (15%) experienced major physical and mental stress. Some participants (8%) reported they were now more careful in taking care of their children because of previous child deaths, and some (5%) expressed they were worried about having another child because of their current experience in having a child with CAH.

A quarter (28%) of the participants' children had psychological conditions such as tendencies to self-isolate, feeling insecure, being temperamental, and becoming masculine (for girls). Parents with a late-diagnosed child indicated their daughters preferred to play with children boys. None of the parents reported changes in their children's social gender.

Emotions

The prominent emotions experienced by the participants at the time of diagnosis encompassed negative feelings such as sadness, shock, disappointment, and devastation. Additionally, some participants reported experiencing contempt from the community regarding their children's condition. Most participants (83%) found it difficult to accept the diagnosis because they had never heard of CAH and found that the healthcare professionals did not educate them sufficiently regarding CAH.

Some participants (35%) felt afraid regarding how others perceive their children's condition, primarily driven by concerns about potential exclusion. Over one-quarter of the participants (13/40) expressed feelings of fear and worry about their children's future.

Financial challenges

Participants mentioned financial issues (60%), emphasizing the high costs of routine medicine (fludrocortisone) and a blood tests (17-OHP) that are not covered by the national health insurance (NHI), as well as transportation costs to healthcare facilities. Some participants described "sick days" as a major challenge because additional medicines needed for stress dosing are often unavailable, as their insurance only provides the exact amount of maintenance medicine needed.

Maintaining children's health by ensuring they take their medicines regularly as instructed and attend regular doctor visits were mentioned as stressors by most of participants (82.6%). Out-of-pocket expenses were associated with the purchase of fludrocortisone and 17-OHP testing, which are not covered by the NHI.

Almost all of the participants (90%) relied on the NHI, whereas the others relied on private insurance (5%) or out of pocket payments (5%). Participants reported that only 1 of the 2 essential medications, oral hydrocortisone, was covered by the NHI, whereas fludrocortisone was not. Access to fludrocortisone in Indonesia was limited at the time of this study; therefore, patients had to rely on donations or purchase the drug out of pocket via connections within the CAH community.

Although oral hydrocortisone is covered by the NHI, several challenges forced participants to make additional out-of-pocket payments. As the system

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only enables pediatric endocrinologists to prescribe a 1-mo supply of medication, hospital visits needed to be scheduled monthly for patients to attain their routine prescription. Thus, 5 participants opted to purchase oral hydrocortisone out of pocket to allow for a 2–3-mo supply. In addition to transportation costs, participants mentioned difficulties in having to leave other responsibilities, such as caring for other young children, as a major challenge in making monthly hospital visits.

In addition to hindrances regarding expenses and time allocated for monthly hospital visits, 4 participants cited the complexity of the NHI system as a reason for choosing out-of-pocket payments.

CAH management resources

When parents were asked whether they had adequate knowledge regarding CAH management, 47.5% answered yes and 50% answered no. Moreover, during the time of diagnosis, 75% answered yes and 25% answered no. Encouragingly, 80% of the parents felt they had adequate awareness of what to do during their child's emergency situations.

The interviewed parents received medication and consultation from 10 hospitals and 2 private pediatric clinics in 8 provinces of Indonesia. The majority of the participants attended routine medical consultations every month (60%), followed by 17.5% once every 2 months.

When asked about what they hope to improve in the future, the participants expressed their hope that primary health care doctors would be more aware of CAH so similar conditions would be detected earlier in other children. Moreover, they hoped for an integrated system to help support the financial needs of patients with CAH, especially for routine medical checkups and treatments.

Most participants (80%) felt that currently they could easily access educational information regarding CAH from the Indonesian CAH community, KAHAKI, and CAH webinars.

Social

When asked about support from their families, most participants (85%) felt they had adequate support. Families provided support in the form of encouragement and assistance in taking over household responsibilities such as taking care of other children when away for hospital visits and financial support. However, several participants expressed difficulty explaining their child's condition to other family members. Therefore, some chose to conceal their children's CAH diagnoses from their families and close community members. Others expressed that although their families were aware of the CAH diagnosis, they did not feel they were given support.

In addition to support from family members, participants were asked about other aspects of their children's support system. All the participants whose children attended school expressed that they had communicated with their children's teachers, who showed empathy and understanding without discrimination. As for their children's friends at school, participants generally stated that because they were still children, they did not understand their child's condition, hence treated them as other peers. Generally, the participants agreed that the strongest support system for their children was their own immediate family.

Most participants (90%) were members of a CAH support group. The benefits of being part of a CAH community mentioned by the participants included the ability to ask questions, the feeling of being supported and not alone, and the opportunity to share experiences. Furthermore, the CAH community played an important role in helping patients gain access to oral fludrocortisone.

Most participants (80%) explained that they allocated time to remind their children to take their medicines regularly and visit pediatricians/pediatric endocrinologists as scheduled.

The participants also cooperated with other family members (older siblings and grandparents) and the school to encourage the children to take their medicines. Maintaining good communication with their children was also frequently referred to given the routine administration of medicines. Some participants stated they were already getting used to their routine and felt just like other people on ordinary days.

Discussion

This study revealed significant challenges faced by children diagnosed with CAH, notably in clinical conditions such as physical changes, early puberty, menstruation delay, incorrect sex assignment, darkened skin, heightened testosterone levels in girls, mood swings, and developmental delays due to late diagnosis and treatment. At the time the study was conducted, Indonesia had not implemented a universal NBS for CAH. Therefore, all patients, with the exception of 1, were not screened and diagnosed based on clinical presentations of ambiguous genitalia, persistent vomiting, and dehydration. A preliminary study in Indonesia involving 1,188 newborns aged 2–5 d with a gestational age ≥ 36 wk determined 2 babies screened were confirmed positive for CAH (6). Although the results were obtained from a small sample size, they imply the need to integrate CAH screening into national health programs because it provides the opportunity for early management. As described by Yu *et al.*, compared with those diagnosed earlier, patients with CAH diagnosed aged > 3 yr had significantly lower emotional social, and psychological functions, school performance, and total HRQOL scores (9).

Parental sacrifices, emotional toll, and the financial burden of managing CAH have accentuated its multidimensional impact on families. The psychological challenges of the children, including self-isolation and

gender-related concerns, underscore the intricate dynamics. Socioeconomic factors, financial hurdles, and complexities of monthly hospital visits contribute to the overall complexity of CAH management. These findings are consistent with those of previous studies that identified ambiguous genitalia, premature pubarche, primary amenorrhea or infertility, and failure to thrive as the main complaints during initial CAH patient evaluations (10, 11). These complaints can cause a reduction in the quality of life of patients with CAH, especially in the psychological domains (11). Early diagnosis, timely initiation of glucocorticoid treatment, affordable access to medicines, and quality care are essential for addressing these challenges (10). Ekbom *et al*. highlighted that good adherence to treatments and early initiation can enhance the quality of life of patients with CAH (12).

A total of 20% of the participants in this study chose to discontinue their current employment, diverging from their career paths or professional ambitions, to take care of their children with CAH after diagnosis. A previous study by Parish *et al*. mentioned that owing to the time-intensive care of children with chronic health problems, parents put aside their interests, and mothers often have to give up their employment because they are the primary caregivers. This can reduce paid work and exacerbate financial problems within families (13).

Some participants experienced significant physical and mental distress and were worried about having another child with CAH. A previous study performed in Central Java, Indonesia showed that the proportion of parental depression among those with girls who had CAH was approximately 24.4%, anxiety 45.1%, and stress 21.8% (14). Another study conducted in Sri Lanka reported that parents of children with CAH had symptoms of depression that did not improve over time (15). The mental health system in Indonesia is still considered underdeveloped, as reflected by low government funding for mental health and the availability of mental health professionals (16). Thus, there is a need to address mental health disparity issues in Indonesia to better accommodate caregivers' mental health issues.

Some children with CAH also exhibit psychological or emotional issues, including tendencies toward social isolation, feelings of insecurity, increased temperamental behavior, and the development of masculine traits in girls. A study conducted by Doktur *et al*. mentioned that some participants had social phobias due to social stigma (17). The study also found that many female participants displayed masculine behaviors concerning their choice of playmates, toys, and activities (17). Some even experienced gender dysphoria, a condition characterized by incongruence between an individual's gender identity and their assigned gender, accompanied by distress or impairment (17, 18). This study suggests that these masculine behaviors may be linked to prenatal and postnatal androgen exposure, and the latter (when prolonged) has a particular influence on gender roles (17). Our study did not investigate whether parents sought professional healthcare assistance for their child's psychological issues. A thorough assessment of psychiatric disorders onset in children with CAH, particularly during adolescence, is crucial (17).

In this study, the parents of children diagnosed with CAH experienced a range of negative emotions, including sadness, shock, disappointment, and devastation. These results align with those of a previous study highlighting that parents grapple with overwhelming emotions. An earlier study suggested that these emotions were due to the parents having difficulties in processing the information provided (19). In our study, the participants expressed that their negative emotions occurred because they felt healthcare professionals did not adequately educate them about CAH. Some parents also felt that the doctors had a limited understanding of CAH. Therefore, it is crucial that healthcare professionals are better equipped to educate parents regarding the condition and essential aspects, such as daily care requirements and the potential consequences of their child's diagnosis, to help alleviate the emotional challenges parents experience upon diagnosis (19).

Another component discussed in this study was the financial challenges experienced by families. The ensuing financial burden associated with chronic illnesses, especially in developing countries such as Indonesia, tends to be exacerbated by myriad challenges in prevention, diagnosis, and management (20). Healthcare spending is significantly higher for patients with chronic illnesses, with recent estimates of up to 6 times that of patients who do not have chronic illnesses (21). Without universal health coverage, patients cannot access essential quality health services without suffering from financial hardships. Indonesia introduced an NHI scheme, *Jaminan Kesehatan Nasional* (JKN), in 2014. By 2023, 255.89 million people had signed up for the NHI scheme, making JKN 1 of the world's largest single-payer health insurance schemes (22–24). However, owing to the limitations in the medications and services covered, progress made in the NHI does not necessarily translate to adequate coverage for patients with CAH.

Although 90% of the patients in this study relied on NHI for CAH management, their parents unanimously agreed that the insurance scheme does not cover all the required medications and investigations. At the time of this study, oral fludrocortisone and 17-OHP testing were not covered by the JKN. Oral fludrocortisone costs approximately Rp 5.700 (0.36 USD)/per tablet, whereas oral hydrocortisone costs Rp 10.000 (0.64 USD)/per tablet. Even though the needs of patients vary, it is estimated that their costs are approximately Rp 400.000–500.000 (25–32 USD) per month for CAH medications. As of 2024, the monthly minimum wage in Java, where most patients in the study were based, ranges from approximately Rp 3.200.000 (205 USD) in Semarang to Rp 5.000.000 (321 USD) in Jakarta. Therefore, CAH-related expenditures cause added financial burdens to families. Furthermore, the findings

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of this study highlight the difficulties in accessing CAH treatment and medications, which exacerbated by complex financial, social, and practical barriers such as difficulties in sparing time for monthly check-ups to obtain their prescribed medications due to familial obligations (leaving other children) and transportation costs. Challenges in fulfilling the requirements to obtain referral letters from different levels of healthcare facilities in Indonesia also worsen the burden on patients with CAH and their families.

One participant mentioned that if her child were diagnosed early, it would have been easier for her to comprehend and ultimately gain knowledge to manage her child's condition. In an observational study conducted by Messina *et al*. regarding the impact of early diagnosis through neonatal screening for CAH on cognitive function in children and adolescents with CAH, the children and adolescents with CAH who were diagnosed early via neonatal screening programs and treated with hydrocortisone had normal psychometric intelligence and executive function (25). Moreover, Brown *et al*. demonstrated that children late-diagnosed with CAH had impairment in their working verbal memory at an early age (7–11 yr) (26). In a study conducted by Fleming *et al*. regarding the knowledge and self-confidence of CAH caregivers in managing adrenal crises, caregivers who had received education on managing adrenal crises had a better understanding and confidence in handling their child's crisis in an emergency. Furthermore, the study highlighted that parents who received adrenal crisis management training had high self-efficacy scores on how to administer oral and injectable drugs to their children (27). Rautman *et al*. presented data about how most parents of children with CAH do not contact family members regarding their child's diagnosis. These parents prefer to contact self-help groups, where they can receive active contact and direct help from parents who are going through the same situation. Therefore, the data suggest that early involvement with these self-help groups or communities, supported by an early diagnosis of CAH, allow parents to gain early access to helpful communities that support their child's CAH management and aid in their emotional needs (28). In the study by Fleming *et al*. parents who were well informed and received clarity regarding their child's CAH diagnosis had higher perceived management abilities and higher HRQOL scores (29).

When the participants were asked whether they felt they had obtained adequate knowledge regarding CAH management, 47.5% answered they did, including what to do during emergencies. The majority of participants (60%) had routine medical consultations every month, followed by 17.5% once every 2 months. Frequent medical consultations require the adjustment and integration of caregivers' daily lives to adhere to their child's lifelong medical needs. Mitchelhill *et al*. reported that caregivers or families with adequate knowledge regarding CAH management have a better opportunity to adjust their daily lives and make CAH routines part of their lives (30). This contributes to decreasing psychological burdens because they have the knowledge and understanding to adapt to their situations (30).

Since CAH is a lifelong condition, patients with CAH and their families must rely on social support from their communities. Family members are 1 of the most potent sources of emotional and tangible social support for parents of children with chronic illnesses, and service providers act as the greatest source of informational support (31). Another primary source of support is a CAH community of patients and their families, which provides participants with companionship and informational support. Regarding CAH communities in Indonesia, patient organizations also act as sources of tangible support by providing assistance in accessing oral fludrocortisone. It is difficult to find companionship support outside established CAH communities, even in hospital outpatient clinic meetings, as explained by 1 of our participants. This highlights the importance of patient and family communities, and the role of healthcare professionals in connecting individual patients with such organizations.

Prior studies have established the importance of developing social support for patients and families with chronic diseases, such as CAH, because low social support has been linked to physiological and neuroendocrine indices of heightened stress reactivity and overall increased morbidity and mortality among diverse populations (32, 33). However, the ability of members of the external community to develop empathy and ultimately act as sources of social support is highly dependent on their ability to understand the complexity of the conditions that patients and their families face. In Indonesia, disparities in education and health awareness remain as significant barriers. When someone is diagnosed with a condition infrequently heard of in the community, negative perceptions fueled by myths and a lack of understanding often occur. Several participants in this study shared that they conceal their children's diagnosis from extended family members and their communities for fear of what those people might say. For chronic and lifelong illnesses such as CAH, it is important to develop a greater understanding among the general population to prevent social isolation because the social isolation levels in individuals with chronic illnesses are worse than those of the healthy populations (34).

Study limitations

This study has several limitations. First, a selection bias might have occurred because the sample was limited to patients from major hospitals in Indonesia who regularly sought medical care. Therefore, the results may not accurately reflect the situations of all Indonesian patients. Second, the study did not categorize the participants according to early or late diagnosis, making it difficult to assess the impact of early detection on disease management. Finally, the study did not

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include the experiences of families who lost a child to CAH. Therefore, it is reasonable to assume that the true burden of late CAH diagnosis in Indonesia may be underreported.

Conclusion

The impact of CAH, particularly when diagnosed late in countries without a universal newborn screening program, presents significant challenges for both patients and their families. In countries with a lack of political will, limited infrastructural support, and varying public health priorities, the establishment and expansion of CAH NBS are often hindered. This study elucidated the potential advantages of implementing universal NBS programs to mitigate the adverse consequences of delayed CAH diagnosis and death. Furthermore, enhancing the accessibility and availability of CAH medications in Indonesia holds the promise of easing the financial strains and unnecessary hardships encountered by families of patients with CAH. Promoting greater awareness about CAH is equally pivotal in alleviating the psychological, emotional, and social burdens of the numerous families and patients affected by this condition.

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