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"…They were just treating her Symptom by Symptom": maternal experiences of having a child with spinal muscular atrophy in Ghana

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Abstract

Background Children with Spinal Muscular Atrophy (SMA) face the challenges of a rare condition impacting their motor neurons, placing substantial caregiving burdens on their mothers. Despite being primary caregivers, mothers of children with SMA in Ghana often find their voices unheard, with restricted access to vital interventions like counselling, support groups, and respite care designed to aid them. This study aimed to explore the experiences of mothers caring for children with SMA in Ghana, where the diagnosis is often delayed and support systems are limited.

Methods We conducted an interpretative phenomenological study with a purposive sample of seven mothers whose children, aged between 8 and 24 months, were diagnosed with Spinal Muscular Atrophy (SMA) and registered with the Rare Disease Ghana Initiative (RDGI). Individual interviews were conducted between August and September 2023 using semi-structured interview guides. The data were manually analysed using a pen-and-paper method. The study adhered to ethical guidelines, including informed consent procedures and confidentiality measures to protect participants' rights and privacy.

Results This study explored mothers' experiences caring for children with SMA, revealing a spectrum of challenges that impact their physical, emotional, spiritual, and social well-being. The findings identified five dominant themes and thirteen subcategories, encompassing issues such as sleep deprivation, mental health concerns, emotional turmoil, practical difficulties, and social isolation. It was also found that mothers tend to have a deep connection to their faith and a sense of closeness to God during challenging times, which provides them with strength and comfort. This collectively identifies the difficult journey of these mothers as they seek diagnosis and treatment for their children.

Conclusion The study identified challenges faced by mothers in caring for their children with SMA. These challenges are reported to significantly impact their physical, psychological, social, and spiritual well-being. To support these mothers effectively, it is recommended that the Ministry of Health, Ghana Health Service, healthcare professionals, policymakers, and non-governmental organisations in Ghana should consider community engagement and education, integration of spiritual care, and psychosocial support programmes.

Keywords Spinal muscular atrophy, Experiences, Mothers, Biopsychosocial-spiritual model, Ghana

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Introduction

With the nature of motherhood, raising a child with a chronic or rare condition such as Spinal Muscular Atrophy (SMA) involves profound dedication and commitment encompassing caregiving duties and various challenges [1]. These challenges are not often medical; they encompass the caregiving duties and emotional, financial, and social dimensions [1]. Despite new treatments such as nusinersen, onasemnogene aberparvovec, and risdiplam being available, issues of access and affordability persist, particularly in low- and middle-income countries like Ghana, highlighting a disparity with high-income countries [2]. Specifically, SMA is a rare genetic disease causing motor neuron degeneration, muscle weakness, potential loss of function, and various other symptoms. These include feeding and swallowing difficulties, respiratory problems, contractures, and osteoporosis. SMA is a major genetic cause of childhood mortality and a common hereditary condition [3]. In terms of classification, SMA is divided into five clinical subtypes based on the highest level of motor function achieved, such as sitting or walking [4]. Type 0 is the most severe subtype that onsets during the prenatal period, and the child survives less than one month after birth—type I manifests within the first few months of life. Affected children do not survive beyond their second year of life. Type II is typically noticed between 6 and 18 months of age. Type III is observed after 18 months. Approximately 10% of SMA cases are classified as SMA Type III, with milder symptoms than types I and II. The last type, type IV, which is the rarest, has the lowest morbidity and mortality and occurs after 20 years of age. Although the global incidence of SMA varies across regions and studies, the life span of type IV patients is like that of type III patients [5–10].

The global incidence of SMA ranges from approximately 1 in 6,000 births to 1 in 100,000 births. In the United States, the prevalence of SMA is estimated to be around 1 in 10,000 individuals, with approximately 25,000 individuals living with SMA [11]. In Europe, SMA ranges from 1 in 6,000 to 11,000 births affected by SMA [12]. Estimating the prevalence of SMA in Africa is a significant challenge. In the case of West Africa and Ghana, for instance, the prevalence of SMA is not well-established due to limited studies on the epidemiology of the disease in the country [13]. However, Hammond et al. [14] reported that cases of SMA in Ghana revealed six cases between 1992 and 2016, and a total of 13 suspected SMA cases were seen in the Pediatric Neurology Unit of Komfo Anokye Teaching Hospital from January 2018 to August 2021 [14]. While SMA impacts individuals worldwide, limited studies have focused on the experiences of mothers caring for children diagnosed with SMA in Ghana [15].

Existing research on SMA extensively explored the clinical aspects, including genetic factors, therapeutic approaches, and the direct impact of SMA on patients, fatigue severity in patients with SMA type II, as well as survival patterns and functional status in SMA [16–22]. These studies shed light on SMA's physical, social, and psychological effects on patients and offer practical recommendations to enhance the current management of the disease, with limited studies in understanding the experiences of mothers [23]. Mothers of children with SMA often find themselves as primary caregivers due to the constraints on the healthcare system, typically in Ghana [14, 23]. Thus, as Ghana seeks to achieve the Sustainable Development Goals, especially 3,10,16, and 17, it is essential to tell the story of maternal caregivers, who are usually the most involved in family support for children with SMA.

Despite striving to provide quality care, Ghana's healthcare system faces constraints such as limited resources, inadequate specialised services, and minimal awareness of rare diseases like SMA [24]. This unfamiliarity leads to a scarcity of information about the experience of families of children with these conditions. Mothers are forced to coordinate medical appointments, from scheduling numerous appointments to managing treatments and therapies and advocating for their children's needs within and outside of the healthcare system [25]. These mothers also engage in the daily physical care of their children by supporting their children's mobility, feeding, and bathing. The responsibilities shouldered by the mothers exert a profound impact on their well-being. The constant hospital visits and physical care demands place physical, emotional, financial, social and spiritual strain on the mothers [25]. Thus, this study aims to explore the unique challenges mothers face in providing care for their children in Ghana. We looked at diagnosis processes and how caregiving impacts the physical, psychological, financial, and social lives while caring for a child with SMA.

Conceptual framework of experiences of mothers of children diagnosed with SMA

The biopsychosocial-spiritual model by Engell, 1974 serves as a conceptual framework for understanding mothers' experiences caring for children with Spinal Muscular Atrophy (SMA). This model acknowledges the multidimensional nature of the caregiving experience, encompassing biological, psychological, social, and spiritual aspects [26]. Mothers of children with SMA face significant challenges that impact their physical, emotional, and social well-being. These mothers often experience a range of emotions, including grief, anger, fear, and depression, as they cope with their child's diagnosis and the progressive nature of the disease [27]. Mothers also report feeling guilty and questioning their actions during pregnancy, sometimes associating the cause of the condition with their own perceived sins [26]. Spirituality is crucial in helping mothers find meaning and purpose in their caregiving journey [27]. Many mothers report connecting to their religious beliefs, finding hope, and feeling a sense of personal growth and connection with others. However, mothers also struggle with spiritual concerns and often express a need for professional support to navigate these challenges [27, 28]. The biopsychosocial-spiritual model emphasises the importance of addressing the complicated needs of mothers caring for children with SMA. By providing comprehensive support that addresses biological, psychological, social, and spiritual aspects, healthcare professionals can better support these mothers in their caregiving journey and improve their overall well-being.

Methods

Study design and setting

This study utilised an interpretative phenomenological design to investigate the lived experiences of mothers caring for children with Spinal Muscular Atrophy (SMA). This design enabled an in-depth examination of the mothers' subjective experiences and the meanings they attributed to their caregiving role. Guided by the Consolidated Criteria for Reporting Qualitative Research (COREQ) checklist, the study was conducted in Ghana, with participants recruited from the Rare Disease Ghana Initiative (RDGI). The participants hailed from three regions of Ghana: Greater Accra, Ashanti, and Western. Seven families participated, with three from Greater Accra, two from Ashanti, and two from Western. The study revealed significant regional variations in access to healthcare services, with families often requiring travelling to access specialist care. Of the three regions, the Greater Accra and Ashanti Regions had better access to healthcare facilities and specialists, while the Western region had limited access, necessitating travel for hospital appointments. Families from Ashanti typically access healthcare services at the Okomfo Anokye Teaching Hospital, and those in Accra also access services at the Greater Accra Regional Hospital and the Korle Bu Teaching Hospital. However, those from Western may travel to Ashanti or Accra for treatment. The geographical diversity of the study provides valuable insights into the experiences of mothers caring for children with SMA in Ghana, highlighting the need for improved access to healthcare services across regions.

Sampling and sample size

We employed purposive sampling to select participants based on the following inclusion criteria: (a) mothers with at least six weeks of experience caring for children with SMA, (b) the ability to communicate in either English, Ga, or Twi, and (c) willingness to participate in the study. The goal was to recruit participants who could provide in-depth insights and expertise that would significantly contribute to meeting the study objectives. We then excluded mothers who were pregnant, exclusively lactating, or diagnosed with a known mental illness. The exclusion was due to perceived vulnerability and the potential impact of pregnancy, breastfeeding, and mental illness on the mother's emotional and physical states during the interview process. We determined the presence of a known mental illness through self-reporting and medical history. We did not screen participants for their ability to provide information-rich insights, as our goal was to capture the authentic experiences of mothers caring for children with SMA. We stopped at seven interviews after reaching data saturation, determined by the repetition of themes and the lack of new information emerging from subsequent interviews. In the sampling process, not all mothers approached participated in the study, citing privacy concerns, fear of judgment, emotional vulnerability, the personal nature of the topic, and time constraints as reasons for declining.

Interview guide

Concerning reviewed literature [27–29], we developed a semi-structured interview guide with two main sections. Before the final interview guide was administered, the interview guide in this study was pretested in Cape Coast with two mothers having a child with Muscular Dystrophy, which shares similar characteristics with SMA and is also a rare condition. The original interview guide consisted of 6 questions and 15 probe items. During the pretesting, unclear questions were restructured, resulting in the final interview guide for the data collection containing two sections. Section A focused on gathering demographic information about the participants. At the same time, Section B explored the mothers' experiences with their child's SMA diagnosis, challenges in caregiving, support systems, daily routines, advice for other parents, and insights for healthcare providers. The guide aimed to capture a comprehensive understanding of maternal experiences in caring for children with SMA.

Data collection

Following ethical approval from the University of Cape Coast, we obtained permission from the Rare Disease Ghana Initiative's (RDGI) management to conduct a phenomenological study on Spinal Muscular Atrophy (SMA). We employed purposeful sampling, leveraging RDGI as the gatekeeper to access potential participants. The principal investigator contacted eligible participants, explained the study's purpose, and obtained their voluntary consent. Seven participants meeting our inclusion criteria were selected, and informed consent forms were obtained through face-to-face meetings ($n = 5$) and email ($n = 2$). Participants were assigned pseudonyms to maintain anonymity throughout the interview process. Participants had the autonomy to participate or decline, and feedback on the findings was provided to those who requested it. A clinical psychologist was available for support during the interviews to address any potential emotional distress. We conducted six face-to-face interviews (45–60 min) in English and one Zoom call interview (30–40 min) in Twi. Data was captured using field notes and audio recordings. The audio recordings were transcribed verbatim, and the transcripts were reviewed multiple times to ensure accuracy and completeness. This process enabled us to produce a comprehensive and detailed transcript for each interview, which was used for analysis. To mitigate potential harm during the Zoom interview, participants were informed of the availability of psychological support and allowed to pause or stop the interview at any time. Data collection occurred between August and September 2023.

Data analysis

We employed Interpretative Phenomenological Analysis (IPA) [30] to analyse the data, following seven steps: familiarisation, exploratory commenting, developing emerging themes, searching for connections across themes, analysing remaining cases, looking for patterns across cases, and reporting. These steps were applied to each interview, repeating the process for all seven participants. For the interview conducted in Twi, the audio recordings were first transcribed verbatim and then translated to English by bilingual researchers fluent in both the local languages and English. A third researcher also cross-checked the translated transcripts for accuracy.

To ensure data trustworthiness, we stored audio recordings on a password-protected portable recorder and transcribed them verbatim. Each researcher individually familiarised themselves with the complete dataset through active reading and simultaneous listening to the audio to validate the data. We manually generated initial codes, highlighting pertinent sentences and phrases using different shades of ink. Similar codes were gathered and grouped based on the frequency and then organised into categories and subcategories. Emerging themes were defined and explained, and analysis was conducted concisely. While the interpretative approach in IPA acknowledges the influence of the researcher's preconceptions and ideas in the interpretative process, we engaged in reflexivity before, during, and after data collection and analysis. This practice involved critical self-reflection to identify and bracket our assumptions, biases, and preconceptions, allowing us to stay grounded in the participants' lived experiences [31]. Member checking, as described by Lincoln and Guba [32], was conducted by returning the transcripts to participants to verify accuracy and obtain feedback on the initial interpretations. Confirmability and dependability were also ensured through audit trails, which captured the study details and processes. The analysis was guided by the study objectives, which focused on exploring the lived experiences and coping strategies of mothers caring for children with SMA. One participant experienced an emotional breakdown during the interview. Her responses were included in the study, and the emotional depth of these experiences was incorporated into the analysis. Any disagreements between the two researchers during the analysis process were resolved through discussion and consensus.

Results

Demographics

The researchers interviewed seven mothers (pseudonyms P1-P7) who were registered with the RDGI. The mothers were between the ages of 29 and 45. Six of the seven mothers were married, and one was a single mother. One was in her late twenties, two were in their early thirties, three were in their late thirties, and one was in her early forties. Five mothers had tertiary education, and two had senior high education. Six of the participants were Christians, and one was a Muslim. The researchers found that out of the seven families, five mothers had their children diagnosed with SMA type 1 and two with type 2. The ages of the children ranged between 8 and 24 months old at the time of diagnosis. At the point of the interview, three of the mothers were working. Of the four mothers remaining, two had to quit their jobs due to their child's condition. The other two never worked. Table 1 summarises the characteristics of the participants.

Table 1 Summary of demographic information of participants (mothers)

Participant	Educational status	Occupation	Age	Religion	Marital status	Type of SMA	Age of Children
P1	Tertiary	Programmes manager	29	Christian	Single	1	8 months
P2	Tertiary	Housewife	33	Christian	Married	1	1 year
P3	SHS	Seamstress	38	Christian	Married	1	1 year
P4	Tertiary	Unemployed	37	Christian	Married	2	1 year
P5	SHS	Entrepreneur	30	Christian	Married	2	2 years
P6	Tertiary	Housewife	37	Muslim	Married	1	2 years
P7	Tertiary	Unemployed	43	Christian	Married	1	9 months

Table 2 Summary of categories and subcategories

Categories	Subcategories
1. Diagnostic Experiences	I. Initial identification and II. impression
	III. Seeking answers and medical attention
	IV. Delayed diagnosis
	V. Misdiagnosis
2. Physical Experiences	I. Daily caregiving challenges
	II. Caregiver physical strain
3. Financial Challenges	I. Medical expenses
	II. Career sacrifices
4. Psychological Distress	I. Depressive behaviours
	II. Heightened tension
5. Social and spiritual Experiences	I. Stigmatisation and social isolation
	II. Social support
	III. Search for spiritual support and meaning

Emergent themes and subthemes

The results of the phenomenological analysis revealed five categories and twelve subcategories that captured the experiences of mothers caring for children with Spinal Muscular Atrophy (SMA). The five categories included: (1) diagnostic journey, (2) physical experiences, (3) financial experiences, (4) psychological distress, and (5) social experiences. The diagnostic journey theme focused on the challenges mothers faced during the diagnostic process, including initial identification and impression, seeking answers and medical attention, delayed diagnosis, and misdiagnosis. The physical experiences theme explored the physical challenges mothers faced while caring for their children, including daily caregiving challenges and caregiver physical strain. The financial experiences theme addressed the financial challenges mothers faced, including medical expenses and career sacrifices. Additionally, the psychological distress theme explored the emotional challenges mothers faced, including depressive behaviours and heightened tension. Finally, the social experiences theme addressed the social challenges mothers faced, including stigmatisation, social isolation, and the need for social support. These themes and subthemes captured the complex experiences of mothers caring for children with SMA, highlighting the need for comprehensive support and care. A summary of the categories with the corresponding subcategories is presented in Table 2.

Theme one: diagnostic journey

The diagnostic journey emerged as a significant theme, with mothers describing various aspects of their experiences. Mothers indicated noticing changes in their children's movements and development, such as decreased foetal kicking during pregnancy or struggling to lift their hands after birth. As one mother noted,

"*While I was pregnant, I realised my child wasn't kicking*" (P2). Another mother said, *"I noticed a change in my baby a month and some weeks after birth. She is a playful child, so one afternoon, I was playing with her as usual, and I realised she was struggling to lift her hand*" (P4).

Upon noticing the changes, these mothers actively sought medical care, desperately seeking answers for their children's condition, but often encountered healthcare providers who lacked knowledge about SMA. This was evident by what some of the mothers had to say:

"*Anytime I notice there is something wrong, I rush her to the hospital; I just want to know what's wrong*" (P3). Likewise, another mother added, *"I was desperately seeking answers, but it felt like no one knew what was wrong with him"* (P1). In seeking answers, the mothers faced prolonged periods without a definitive diagnosis, with some children passing away before the SMA diagnosis was confirmed.

"*For over six months, they couldn't tell us what was wrong with my child"* (P7).

In addition, "*The doctors were wasting our time. They had no answers, so I just asked them to discharge us"* (P1).

The health professionals, due to a lack of knowledge about SMA, misdiagnosed some of the children with developmental conditions such as cerebral palsy and autism, further delaying appropriate treatment and support. Some mothers had this to say.

"*We took her for some check-ups; nobody knew what was going on with my child. One of the doctors said it was just a developmental delay; she would move when her motors were ready. She died a month after the actual diagnosis was known"* (P4).

"*Before we took her to Australia, the doctors here were treating her for cerebral palsy"* (P2).

Theme two: physical experiences

The physical experiences of mothers were characterised by the daily challenges of caregiving and the strain it placed on their well-being. Mothers described the immense physical demands of providing round-the-clock care for their children, who were often unable to perform basic functions independently. Some mothers had this to say.

"*I do everything for him because he cannot use his muscles. I will have to help him sit and stand; the water bottle, too, unless it is being held for him. Can you imagine he is unable to hold my breast whiles sucking it?"* (P2). "I *go everywhere with her, washroom, like I mean everywhere. When she is asleep, I have to put her in her bed closer to me, especially when in the kitchen"* (P4).

These constant demands of caregiving took a toll on the mother's physical health, leading to neglect of their own needs, sleep deprivation, and physical pain.

"If you see me lean now, it is because I do not eat properly, have no proper sleep, and mostly have poor hygiene. I keep having this constant waist pain. My husband mostly massages me before he goes to bed." (P1).

"Madam, I have not slept properly. I cannot close my eyes in peace. I try to sleep small, and I will have wild dreams about him falling" (P5).

Theme three: financial challenges

The financial challenges faced by families were significant, with mothers reporting the high costs of medical expenses and the need to make career sacrifices to prioritise their child's care. The cost of diagnosis, treatment, and ongoing care placed a heavy financial burden on families, with some mothers paying thousands for tests that came after their child's passing. Some mothers shared their experiences by saying:

"The medical bills were just outrageous. I paid thousands for diagnosis test results that came after he died. I wonder how things may be now if he is still alive" (P1).

"Insurance does not cover anything regarding his treatment. We have to bear everything. I'm just a seamstress, and my husband doesn't do any work that fetches plenty of money. The little money we get goes into her hospital bills" (P7).

Some mothers had to resign from their jobs to provide full-time care for their children, and others had to exert pressure on their husbands, taking on additional work to cover the mounting expenses. They have these to say.

"I used to be a bank teller oo, madam; because of this, I have to resign from my job and take care of him. Nobody would take care of your child better than you will do. It was a difficult decision, but hey, I had to, my child's life first" (P2).

"My husband is working as a banker and an Uber driver. He mostly did not come home early because he had added the driving to fetch extra money" (P6).

Theme four: psychological distress

Psychological distress was a prominent theme, with mothers describing depressive behaviours, heightened anxiety, and the emotional toll of caring for a child with a life-limiting condition: Mothers expressed feelings of self-blame, hopelessness, and regret, often going to bed crying and questioning their abilities as caregivers. One mother expressed.

"I used to go to bed crying and blaming myself for his situation oo. It was like I failed as a mother in attending to his needs at the time I was supposed to. Now he gets to the hospital too, and he wasn't responding to treatment; I was just hopeless" (P2). Another mother shared:

"I regret giving birth; I should have just stayed without a child like I`m so tired and devastated" (P4).

The uncertainty surrounding their children's prognosis and the fear of losing them led to constant anxiety and worry about the future. *"Every little cough or change in breathing makes my heart beat so fast. I am always anxious about what might happen next"* (P3).

"The future was so uncertain, and so my husband and I get anxious about what lies ahead for our child. Will he be able to live a fulfilling life? It keeps us up at night" (P7).

Theme Five: social and spiritual experiences

Stigmatisation, social isolation, the role of support systems and the search for spiritual support and meaning in their caregiving journey were the main reports on the social and spiritual experiences. Mothers described being disliked by neighbours and even close friends who lacked understanding about SMA, with some being accused of trying to abort their pregnancies. These mothers had these to say.

"I've heard conversations among our neighbours and some church members about her condition. Someone asked if I tried aborting?" (P2).

"We've had people distance themselves from us, even close friends, ever since they realised our child's condition. Some people think SMA is contagious and infectious, so they wouldn't get close to us. It is painful, you know" (P3).

While some mothers felt alone in their struggles, others found support and solace from families and organisations like the Rare Diseases Ghana Initiative. Some mothers said.

"There is no help from anywhere oo; we are dealing with everything on our own" (P2).

"I will say, my son's condition was a blessing in disguise, COVID too helped. We were all home, so I got the support I needed, and oh, I cannot forget the Rare Disease Ghana Initiative. They have been supportive as well, even though I met them a few weeks before my son's death" (P1).

Few Mothers had their spirituality strengthened with the challenges of caring for children with SMA. However, for some, their faith was negatively challenged by their child's condition. These mothers had these to say.

"I pray every day for God's strength and guidance. My faith is the only thing that keeps me going" (P5).

"When I feel overwhelmed, I turn to prayer. It gives me a sense of peace and purpose in this difficult journey" (P7).

"I used to be a strong believer, but after seeing my child suffer, I question God's plan. Why would he allow this to happen to such an innocent child? The zeal to pray is not even there anymore" (P4).

Discussion

The experiences of mothers caring for children diagnosed with SMA are complex and challenging and were explored using IPA in conjunction with reviewed literature and the Biopsychosocial-Spiritual Model underpinning the study.

The diagnostic journey emerged as a significant theme, with mothers recounting the early signs they noticed in their children's movements and development, such as decreased foetal kicking during pregnancy or struggling with basic motor skills after birth. Seeking medical care, these mothers expressed frustration at encountering healthcare providers lacking knowledge about SMA, leading to prolonged periods without a definitive diagnosis and, tragically, some children passing away before receiving a confirmed diagnosis. Misdiagnoses, such as labelling children with developmental conditions like cerebral palsy or autism, further delayed appropriate treatment and support, underscoring the critical need for timely and accurate diagnoses in SMA cases. In line with these findings, other studies confirmed that many parents said they had a gut feeling something was wrong with their children during pregnancy and after birth [33–35]. These mothers reported noticing symptoms of changes in the children's development. It was also found in their study that parents consulted various health professionals about symptoms they noticed throughout their journey to diagnosis and often visited individual health professionals several times. Many participants reported multiple health visits before receiving a definitive diagnosis, and 67% of respondents felt that their child could have been diagnosed sooner.

The physical experiences of mothers caring for children with SMA were characterised by the relentless demands of caregiving, where mothers described assisting their children with basic functions and providing round-the-clock care. The strain of caregiving took a toll on the mother's physical health, leading to neglect of their own well-being, sleep deprivation, and physical pain. The physical burden of caregiving was palpable in their accounts, with mothers expressing the challenges of attending to their child's needs while grappling with their own health issues and exhaustion. This aligns with studies that have documented the significant physical burden on caregivers of children with SMA [18, 37, 38]. It is stated in their studies that the mothers often found themselves hyper-alert, resulting in poor sleep patterns and increased stress levels. Other studies confirmed that the impact of physical challenges has a significant effect on family members, causing chronic pain, fatigue, poor sleep, poor nutrition, decreased physical activity, and increased risk for injury.

Furthermore, the continued caregiving and hospital visits left most mothers and families with strains on their finances. Mothers reported making career sacrifices, resigning from jobs, or pressuring partners to take on additional work to cover mounting expenses. The financial strain was exacerbated by the lack of insurance coverage for treatments, leading to substantial out-of-pocket expenses and financial hardships for these families. The sacrifices made by mothers to prioritise their child's care underscored the immense financial challenges. Similarly, mothers were reported to have adjusted their careers and professions. They noted that mothers experience disruptions in their daily routines and jobs and an increased focus on the challenges associated with their responsibilities [37–39]. All mothers in this current study agreed that caring for these children has caused them to adjust their career goals and professions to attend to the child's needs. They described having faced the need to reduce working hours, take extended leaves of absence, or even resign from their jobs to provide full-time care for their children. These career sacrifices impact their earnings, long-term financial stability, and goals.

Also, psychological distress was a prevalent theme among mothers, with feelings of self-blame, hopelessness, and regret permeating their experiences. The uncertainty surrounding their children's prognosis, coupled with the fear of losing them, led to constant anxiety and worry about the future. These mothers expressed deep emotional turmoil, going to bed in tears, questioning their abilities as caregivers, and grappling with the overwhelming weight of caring for a child with a life-limiting condition. The psychological toll of caregiving for children with SMA was profound, highlighting the need for comprehensive support for mothers facing such challenges. In support of our findings, similar studies reported that the mothers experienced emotional states upon first being aware of symptoms in their child. These were primarily described as feelings of denial, stress, depression, anxiety, worry, and being unsure of what was happening in their child [33, 40, 41].

The social and spiritual dimensions of the caregiving experience added a complex layer to the mothers' journeys, characterised by experiences of stigmatisation, social isolation, and a quest for support and meaning. While some mothers encountered judgment and isolation from their communities, others found solace in support networks and organisations. The role of spirituality varied among mothers, with some deriving strength and comfort from their faith, while others grappled with doubts and questions about their beliefs in the face of their child's condition. The social and spiritual aspects of their experiences highlighted the importance of comprehensive support systems and understanding within communities to mitigate the isolation and stigma faced by mothers caring for children with SMA. Similarly, this study confirmed findings from earlier research that mothers face social challenges, including the fear of premature death, sleep loss, stigmatisation, isolation, limited social activities, and uncertainty [42, 43]. Despite these challenges, some mothers received positive emotional, practical, and financial support from their networks, which alleviated some of the burdens associated with caring for a child with SMA. Consistent with previous research, this study found that mothers with social support exhibit better psychosocial well-being compared to those without such support [26, 44–46].

Implications and future directions

The findings from this study have several implications for clinical practice, policy development, and future research. Firstly, healthcare providers must receive specialised training in promptly recognising and diagnosing rare conditions like SMA. This training is crucial in reducing diagnostic delays and improving access to early interventions. Policy initiatives should focus on improving insurance coverage for SMA treatments and providing financial support to alleviate the financial burdens faced by affected families. Future research should explore interventions aimed at enhancing support systems for caregivers, improving access to healthcare services, and addressing the holistic needs of families affected by SMA. Additionally, there is a need for studies that examine the long-term psychosocial outcomes of mothers and explore strategies to enhance their resilience and well-being.

Strengths

The study's use of IPA provided an in-depth exploration of the lived experiences of mothers caring for children with SMA, giving voice to their challenges and perspectives. The findings were analysed in conjunction with a review of relevant literature and the Biopsychosocial-Spiritual Model, providing a comprehensive framework for understanding the multifaceted experiences of these mothers. The study highlighted the critical need for timely and accurate diagnoses in SMA cases and the profound physical, emotional, social, and spiritual toll of caregiving on mothers. The findings are consistent with those of other studies, strengthening the validity and generalisability of the results.

Limitations

The study's focus on a specific population, mothers caring for children with SMA, may not be generalisable to other caregiving contexts. The study did not explore the perspectives of other family members or healthcare providers, which could provide additional insights into the caregiving experience. Despite these limitations, the study offers valuable insights into the complex and challenging experiences of mothers caring for children with SMA, highlighting the need for comprehensive support systems and further research to improve the quality of life for these families.

Conclusion

This qualitative study employed IPA to explore the complex experiences of mothers caring for children diagnosed with SMA in Ghana. The findings revealed complex dimensions encompassing physical, emotional, social, and spiritual aspects of caregiving.

The diagnostic journey, characterised by frustration and delay, sets the stage for the relentless demands of caregiving that follow. Mothers described the physical burden of caregiving, including assisting with basic functions, providing round-the-clock care, and neglecting their well-being. This burden is exacerbated by financial strain, as mothers make significant career sacrifices to prioritise their child's care. The psychological toll of caregiving is profound, with feelings of self-blame, hopelessness, and regret permeating their experiences. The social and spiritual dimensions of caregiving add a complex layer, with mothers facing stigmatisation, social isolation, and a quest for support and meaning. The findings of this study are consistent with those of other research, highlighting the critical need for timely and accurate diagnoses, comprehensive support systems, and understanding within communities to alleviate the isolation and stigma faced by mothers caring for children with SMA. The study underscores the importance of addressing the physical, emotional, social, and spiritual needs of these mothers, ensuring they receive the support necessary to navigate the challenges associated with caring for a child with a life-limiting condition.

Recommendation

To gain insight into the experiences of mothers caring for children with rare conditions like SMA, the Ministry of Health, Ghana Health Service, healthcare professionals, policymakers, and non-governmental organisations in Ghana should engage in more educative programmes and awareness creation. This includes collaborating with patient advocacy groups, participating in conferences and workshops, using social media and online communities, consulting with international organisations, supporting awareness-raising events, encouraging open communication between stakeholders, and fostering a culture of shared decision-making. By adopting these strategies, a more comprehensive understanding of the challenges faced by these mothers and their families will be established. Hence, targeted interventions and support systems should be created to address their specific needs effectively.

Supplementary Information

The online version contains supplementary material available at https://doi.org/10.1186/s12904-025-01651-3.

Supplementary Material 1

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Author contributions
ED-YT and JOS conceptualised and wrote the initial draft. ED-YT collected the data while both authors analysed the data. All authors read through the final script and approved for submission.

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Data availability
Data and recordings are available upon request and fulfilling requirements set out by the Ethical Review Board.

Declarations

Ethics approval and consent to participate
We conducted the study in accordance with the Declaration of Helsinki. Ethical Approval (CES/ERB/UCC/EDU/V8-23/65) was obtained from the Ethical Review Board of the College of Education Studies, University of Cape Coast on 1 August 2023. Furthermore, between the 10th and 18th of August, each participant confirmed and signed an informed consent form outlining all the facts about the study and detailed information about their rights as participants. Due to the sensitive nature of the study, we provided on-site psychologists to provide support for those experiencing any emotional or psychological trauma.

Consent for publication
Not applicable.

Competing interests
The authors declare no competing interests.

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