

From Diagnosis to Daily Life: Challenges and Coping among Caregivers of Children with IMD in India

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ABSTRACT

Background: Inherited Metabolic Disorders (IMD) are a group of rare genetic disorders that often present in early childhood and require lifelong medical, nutritional, and psychosocial management. The intensive care demands and chronic nature of these conditions place a substantial burden on caregivers, yet their lived experiences remain underexplored, especially in low- and middle-income settings like India. **Objectives:** The aim of the study is to explore the quality of life, psychosocial challenges and coping mechanisms of caregivers of children diagnosed with various types of IMD. By understanding their experiences, the study sought to inform the development of targeted support systems and policy interventions. **Methods:** A qualitative, phenomenological design was adopted to capture the in-depth experiences of caregivers. Fifteen primary caregivers of children with IMD were recruited using purposive sampling from clinical settings in Jaipur, Rajasthan. In-depth, semi-structured interviews were conducted in Hindi and English, transcribed, and analyzed using Braun and Clarke's six-phase thematic analysis. NVivo 12 software supported the coding process. **Results:** Four major themes emerged: (1) Emotional turbulence and adaptation following diagnosis, (2) Caregiving as a life-altering commitment, (3) Navigating the healthcare and social systems, and (4) Strategies for resilience and hope. Caregivers reported significant emotional distress, social isolation, financial strain, and lack of access to specialized services. However, many also demonstrated remarkable resilience, drawing on familial support, spiritual beliefs, and advocacy communities. **Conclusion:** Caregivers of children with IMD face multifaceted challenges that impact their mental, emotional, social, and financial well-being. There is an urgent need for comprehensive caregiver support interventions, including counseling, financial assistance, community awareness, and integrated health services. Policymakers and healthcare providers must recognize caregivers as integral stakeholders in IMD management and design inclusive programs to enhance their quality of life.

Keywords: *Inherited Metabolic Disorders, caregivers, quality of life, rare diseases, qualitative research, India, thematic analysis*

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Inherited Metabolic Disorders (IMD) are a heterogeneous group of rare genetic disorders caused by defects in specific enzymes or transport proteins, leading to disrupted metabolic pathways and accumulation or deficiency of key metabolites (Saudubray et al., 2016). Although individually rare, collectively these disorders pose a significant health burden, especially in regions lacking routine newborn screening programs and specialized care services (Sanderson et al., 2006; Waters et al., 2018). Children diagnosed with IMD often require lifelong medical nutrition therapy, frequent hospital monitoring, and individualized care protocols to prevent metabolic crises and irreversible complications (O'Brien, D., 1979; Blau et al., 2018)

While much research has focused on the clinical management and biochemical outcomes of children with IMD, less attention has been given to the psychosocial and emotional experiences of their caregivers. Parents, particularly mothers, often assume the role of full-time caregivers and medical advocates, navigating complex health systems and performing extensive “invisible labor” to maintain their child’s stability ((Feeley et al., 2021). Caregiving in this context is not only physically and emotionally taxing, but is also shaped by profound uncertainty regarding prognosis, treatment continuity, and quality of life (Fekete et al., 2021).

Qualitative research on caregiving in rare diseases has emphasized the unique emotional landscape in which families operate—marked by delayed diagnoses, medical instability, limited societal understanding, and financial insecurity (Pelentsov et al., 2016). However, there remains a gap in the literature regarding how these stressors influence the day-to-day lives, identities, and coping mechanisms of caregivers managing complex, life-altering metabolic conditions. In the Indian context, where public support services and awareness around IMDs remain limited, caregivers often experience an additional layer of systemic and structural burden (Siddiq et al., 2016).

This study seeks to fill this gap by exploring the lived experiences of caregivers of children with IMD in India. Using thematic analysis, it examines how caregivers make sense of their roles, the emotional and logistical challenges they encounter, and the ways in which they reconstruct meaning, normalcy, and resilience in the face of ongoing uncertainty. The findings aim to inform more empathetic care models and policy interventions that center the voices of caregivers and address their multifaceted needs.

METHODOLOGY

This qualitative study adopted a phenomenological research design to deeply explore the lived experiences of caregivers of children diagnosed with Inherited Metabolic Disorders (IMD). Phenomenology was chosen for its strength in capturing subjective meanings, emotional depth, and individual perceptions of complex realities, which are especially relevant in caregiving for rare, lifelong metabolic disorders. (Creswell & Poth, 2018).

The study was conducted in Jaipur, Rajasthan, in collaboration with Government Paediatric hospitals and tertiary care centres that manage children with rare metabolic conditions. Fifteen participants were recruited through purposive sampling to ensure diversity in demographic backgrounds, duration of caregiving, and IMD subtypes. To be eligible, participants had to be the primary caregiver (either parent or legal guardian) of a child diagnosed with Amino acid disorders involving lifelong dietary treatment and should have been actively involved in the child’s day-to-day care for at least six months. Written

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informed consent was obtained prior to participation. Extended family members who were not directly responsible for the child's daily care were excluded from the study. Additionally, caregivers of children who were undergoing acute metabolic crises at the time of data collection were not included, in order to avoid placing further emotional burden on families during such vulnerable periods.

Semi-structured, in-depth interviews served as the principal method of data collection. Interviews were conducted in either Hindi or English, depending on the participant's comfort, and held in private settings to ensure confidentiality and openness. Each interview lasted between 45 and 75 minutes and was audio-recorded with consent. Field notes were maintained during and after the interviews to capture relevant contextual and non-verbal cues that added depth to the narrative understanding.

The interview guide used in the study was developed by drawing on a comprehensive review of existing literature related to caregiving in chronic and rare pediatric illnesses. It was also informed by expert consultation from professionals experienced in clinical nutrition and pediatrics. Elements of the guide were adapted from previously validated tools such as the Zarit Burden Interview and WHO-QOL BREF, which were modified to suit the context of caregivers managing children with IMDs. The guide was designed to explore domains including emotional responses to diagnosis, caregiving responsibilities, psychosocial adjustment, financial burden, healthcare access, social support, and coping strategies.

All interviews were transcribed verbatim (a written record of spoken words). Transcripts in Hindi were translated into English and were subsequently back-translated to ensure linguistic and conceptual accuracy. The data were analyzed using the six-phase framework of thematic analysis proposed by Braun and Clarke (2006). This involved familiarization with the transcripts, generation of initial codes, identification of candidate themes, review and refinement of themes, theme definition and naming, and final synthesis of findings. NVivo 12 software was utilized to assist in coding and data organization.

To ensure trustworthiness, the study adhered to the criteria of credibility, transferability, dependability, and confirmability as outlined by Lincoln and Guba (1985). Credibility was enhanced through member checking, where five participants reviewed and confirmed the accuracy of their transcripts and thematic interpretations. Thick descriptions of the study setting and participant experiences were provided to facilitate transferability. Dependability was addressed by maintaining an audit trail documenting all methodological decisions and analytical steps. Confirmability was ensured through reflexive journaling by the primary researcher to minimize bias and maintain objectivity throughout the research process.

Ethical approval for the study was obtained from the Institutional Ethics Committee of the Department Research Committee (IECHR/FCSc/Ph.D/2023/10) and Medical College (2023/14217). Written informed consent was obtained from all participants prior to data collection. Participants were informed of their right to withdraw at any time without consequence.

RESULTS

This study explored the lived experiences of caregivers of children with Inherited Metabolic Disorders (IMD), focusing on the impact on their quality of life. Thematic analysis of fifteen in-depth interviews revealed a rich and complex set of experiences, which were organized

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into four major themes and corresponding subthemes. These themes reflect the emotional, social, financial, and practical challenges caregivers face, as well as the coping strategies they employ in the context of rare metabolic disorders.

The four overarching themes that emerged from the analysis were:



Each theme is presented in detail below, supported by direct quotations from participants to illustrate key findings and provide voice to their narratives.

Theme 1: Emotional Turbulence and Adaptation following Diagnosis

1. Living with Uncertainty

Caregivers of children with Inherited Metabolic Disorders (IMD) inhabit a world dominated by pervasive uncertainty—a relentless backdrop shaping every decision, emotion, and plan. This uncertainty stems from several interrelated sources: the medical complexity and unpredictability of IMDs, inconsistent health trajectories, lack of clear prognostic indicators, and the fluid nature of care requirements that change with each developmental stage or health event. The burden of not knowing permeates the caregiver experience, influencing mental health, family dynamics, and the capacity to hope.

Diagnostic Delay and Emotional Turmoil

The journey to diagnosis often resembles a maze with multiple dead ends. Long stretches of uncertainty, characterised by frequent physician appointments, misinterpretation, and symptom minimisation, were described by caregivers. A lack of knowledge of uncommon metabolic disorders among primary care physicians was evident in the early medical reactions, which usually comprised assigning symptoms to common illnesses or behavioural problems. In addition to delaying necessary medical care, this delay exacerbated the mental

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distress of the caregivers, who began to feel more helpless, insecure, and afraid as the child's condition worsened without warning.

"We saw five doctors over six months. Everyone treated the symptoms but missed the root cause. By the time the diagnosis came, we were already deep into complications." (Participant 1)

The moment of diagnosis was immensely paradoxical: parents experienced intense sadness over the loss of their child's "normal" and healthy future, but they also felt a mixed sense of relief at finally having an explanation for the condition and a road forward. The enormous amount of new information, complicated language, and the knowledge that taking care for their child would become a lifelong journey requiring continual vigilance all contributing to this emotional ambivalence.

"I finally had an answer, but no roadmap. I had to learn everything like how to read lab results, what foods to give, when to panic." (Participant 4)

Constant State of Alert: The Crisis Mentality

For caregivers managing disorders prone to sudden metabolic crises (such as IVA, MSUD, and MMA), life becomes an ongoing emergency drill. The fine line between stable health and acute crisis demands a hypervigilant state where every minor symptom—a slight fever, vomiting, or reduced appetite—is scrutinized as a potential trigger for hospitalization or worse. This "crisis mentality" leads to chronic anxiety and exhaustion, impacting sleep quality and mental well-being.

"Any fever or vomiting is a red flag. We don't wait to see how it goes—we go straight to the emergency." (Participant 2)

Caregivers often reported the psychological strain of living in this heightened alertness, describing it as "wearing armor" continuously. The need to always be prepared, like packing emergency supplies, maintaining detailed medical records, and having rapid access to healthcare, created a lifestyle defined by urgency and limited flexibility. Spontaneous family activities or travel became fraught with risk, isolating families socially and emotionally.

Uncertain Prognosis and Restricted Planning

Uncertainty extends beyond the immediate medical crises into long-term considerations—education, independence, relationships, and lifespan remain unknown variables. Caregivers grieved not only present hardships but also the "unknown future," which prevented meaningful long-term planning. The ambiguity around developmental potential or quality of life created an emotional limbo, compounded by a societal lack of understanding or acknowledgment of this unique form of grief.

"The future is a big question mark. We celebrate every small achievement because we don't know what lies ahead." (Participant 5)

This uncertain future often caused caregivers to live "one day at a time," focusing on immediate health but struggling with existential worries about their child's ability to thrive independently or achieve milestones that other children might reach effortlessly. This ambiguity fueled a persistent emotional tension between hope and despair.

Fear of Loss of Access and Continuity of Care

The uncertainty was magnified by practical fears around access to care, especially regarding specialized medical foods, formulas, and expert healthcare providers. Families in rural or underserved areas faced additional logistical challenges and were acutely aware of systemic vulnerabilities such as supply chain disruptions or loss of insurance coverage. This created an omnipresent anxiety about possible interruptions in critical treatment.

"If the pharmacy runs out of her formula, we have no backup. It is not something you can find in local stores." (Participant 10)

Caregivers expressed a profound fear that any disruption could precipitate a rapid health decline, making stability precarious and highlighting the fragility of their support systems. This insecurity added a significant emotional and financial burden, further exacerbating caregiver stress.

Theme 2: Caregiving as a Life-Altering Commitment

1. The Invisible Labor of Care

The “invisible labor” of caregiving refers to the vast and multifaceted work performed by caregivers that often goes unrecognized both socially and institutionally. This labor spans a broad spectrum—from the practical to the emotional—and encompasses ongoing efforts that are physically exhausting, cognitively demanding, and emotionally draining. The theme underscores how caregiving is not just about attending to immediate physical needs but involves sustained, complex management across multiple domains.

Nutritional Workload: A Full-Time Role

Dietary management is central to caring for children with IMD, requiring caregivers—mostly mothers—to master detailed nutritional calculations, ingredient sourcing, meal preparation, and constant monitoring. This role is intensely demanding, involving skills in nutrition science, arithmetic, and pharmacology to ensure protein restriction compliance while maintaining adequate growth and development.

"I spend more time planning her meals than anything else in the day. It's not just food—it's medicine in the form of meals." (Participant 3)

The work extends beyond typical cooking to include planning for special occasions, negotiating social situations, and mitigating the emotional impact on the child. Caregivers described the emotional weight of balancing strict dietary compliance against the child's desire for normalcy and participation in family or cultural events. This labor is unrelenting and often without respite.

Bureaucratic Navigation and System Gaps

Caregivers faced numerous bureaucratic hurdles—applying for government schemes, securing insurance reimbursements, obtaining dietary charts, and scheduling specialist consultations. These administrative tasks required significant time, energy, and knowledge often absent in low-resource or rural settings. The healthcare system's lack of transparency and guidance compounded caregivers' burden, forcing them to become self-taught experts in paperwork and system navigation.

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"There's a new form every time. One small mistake, and the claim gets rejected. It's like they're testing our patience." (Participant 11)

Educational Advocacy: Fighting the System

Schools often lacked awareness or readiness to accommodate children with IMD, compelling caregivers to assume the role of educators and advocates. This included educating teachers about the child's condition, dietary needs, emergency protocols, and cognitive challenges. Caregivers frequently described this advocacy as a continuous battle, with each new school year or teacher requiring renewed efforts to secure understanding and support.

"I had to write letters and make calls just to get the school to stop giving him chocolates and cakes from other students. They thought I was just being overprotective." (Participant 2)

Some caregivers were forced to remove their children from mainstream education due to inadequate accommodations or bullying, leading to social isolation and additional educational challenges. This advocacy role consumed emotional and physical resources and underscored the systemic gaps in inclusive education for children with chronic health conditions.

Gendered Expectations and Emotional Cost

The caregiving burden was strongly gendered, with women disproportionately responsible for the extensive labor involved. Even in two-parent households, fathers often contributed primarily in logistical or financial capacities, while mothers managed the nuanced details of care. This unequal division entrenched professional sacrifices, interrupted career trajectories, and fostered a sense of isolation among female caregivers.

"I had a career once. But no employer can accommodate the unpredictability of this condition." (Participant 1)

"He's a great father, but I'm the one who keeps track of protein intake and fills out medical forms. He doesn't know the details." (Participant 10)

Women expressed feelings of invisibility despite the expertise they developed, often internalizing responsibility for their child's survival and outcomes. The emotional toll of this "sole caregiver" identity included burnout, stress, and loss of personal identity beyond caregiving roles.

Theme 3: Strategies for Resilience and Hope.

1. The Transformation of the Self

Caregiving precipitated profound internal changes in caregivers' emotional, cognitive, and existential frameworks. These transformations were complex and ambivalent, reflecting both coping mechanisms for survival and emergent strengths. Caregivers navigated evolving identities, shifting priorities, and new understandings of success and purpose.

Emotional Maturation and Resilience

Over time, caregivers described developing greater emotional strength and self-regulation. While initial reactions included panic and despair, many learned to channel their energy

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toward problem-solving and managing crises with composure. This emotional maturation was critical for sustaining caregiving over years of uncertainty.

"I never thought I'd be able to explain lab reports or challenge a doctor's advice. But I've become that parent." (Participant 3)

Caregivers maintained compassion and tenderness, balancing resilience with vulnerability. This growth enabled them to navigate burnout cycles and re-engage with social supports, underscoring a dynamic process of emotional adaptation rather than static endurance.

Reframing Milestones and Gratitude

Caregivers' definitions of "success" and developmental milestones shifted to more individualized, attainable goals. Conventional societal benchmarks were replaced with markers meaningful within the context of the child's condition—such as biochemical stability, crisis-free school terms, or mastering self-feeding.

"We don't celebrate birthdays with cake. We celebrate when she drinks her formula without tantrums." (Participant 5)

This reframing fostered gratitude for small victories and everyday moments unmarked by medical intervention. Celebrating these milestones created emotional relief and a positive outlook, essential for sustaining hope.

Identity Shift and Caregiver Solidarity

Caregivers described evolving self-identities that transcended traditional roles, becoming experts, advocates, and community leaders. This identity shift was empowering, giving caregivers a sense of agency in an otherwise unpredictable landscape. Online support groups, peer networks, and advocacy communities provided crucial affirmation and a platform for collective action.

"Finding other mothers going through the same thing saved me. I no longer feel like I'm doing this alone." (Participant 4)

However, this transformation also carried a risk of isolation from those outside the caregiving community, who might not understand the depth of expertise or emotional labor involved. Nonetheless, caregiver solidarity emerged as a vital source of strength and knowledge exchange.

"I used to be a banker. Now I'm a dietician, nurse, and therapist rolled into one. I've lost a lot, but I've also grown." (Participant 13)

Theme 4: Strategies for Resilience and Hope.

1. Overarching Theme: Redefining Normalcy

The intersection of uncertainty, invisible labor, and self-transformation culminates in caregivers' redefinition of what constitutes "normal." This new normal is a negotiated, personalized equilibrium that incorporates the constraints and challenges of caregiving while embracing moments of joy, stability, and meaning.

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"Our life doesn't look like others. But it's ours, and we've found our peace in it."
(Participant 9)

Caregivers no longer measure their lives against societal norms but instead construct their own rhythms—governed by medical regimes and emergency preparedness, yet imbued with a sense of peace and acceptance.

"Normal for us is lab visits, formula preparations, and early bedtimes. It's not glamorous, but it's ours." (Participant 10)

This redefinition represents both an adaptive coping strategy and a form of empowerment, positioning caregivers as experts within a healthcare system that often overlooks their critical role.

DISCUSSION

This study offers a nuanced exploration of the lived experiences of caregivers of children diagnosed with Inherited Metabolic Disorders (IMDs), a group of rare genetic conditions requiring lifelong management. Using thematic analysis of in-depth interviews, the study identified key themes that capture the complex emotional, social, economic, and healthcare challenges that caregivers encounter, as well as the resilience they exhibit in navigating these difficulties.

The first major theme, **"Emotional Turbulence and Adaptation Following Diagnosis,"** captures the shock, grief, and anxiety that accompany the initial diagnosis. Caregivers described the period immediately following diagnosis as emotionally destabilizing, characterized by fear of the unknown and a loss of control. Many experienced a "crisis of meaning" as they struggled to comprehend the nature of the disorder and its implications for their child's life. This emotional response aligns with findings from Pelentsov et al. (2016), who reported that parents of children with rare diseases frequently go through a grieving process akin to bereavement. Social stigma associated with handicap or genetic illness, as well as inadequate access to trustworthy medical information in regional languages, exacerbate the emotional load on Indian parents. Many parents said that members of the community or extended family had directly or indirectly placed the responsibility on them, which made them feel even more guilty and helpless.

"Caregiving as a Life-Altering Commitment," the second subject, captures the significant changes that parents experience on a daily basis. The majority of respondents, mothers in particular took on the role of primary caretaker and frequently had to give up their careers or academic goals in order to care for the child. As a result, their household income decreased, they became more reliant on their husbands or in-laws, and they were cut off from previous social networks.

The third theme, **"Navigating the Healthcare and Social Systems,"** highlighted systemic gaps that caregivers must constantly negotiate. Several participants recounted years of misdiagnoses, long diagnostic odysseys, and frequent travel to tertiary centers for confirmatory testing and consultations. Once diagnosed, many struggled to obtain medical foods such as low-protein formulas, amino acid supplements, or specialized enzymes, which are either unavailable locally or unaffordable. This mirrors the findings of Bhattacharya et

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al. (2020), who documented the fragmented care pathways and lack of policy coverage for rare disease management in India. Caregivers also reported that healthcare professionals were often unfamiliar with the child's specific IMD, leading to errors in medication or dietary advice. Furthermore, there was a widespread absence of genetic counseling, psychological support, or long-term care planning—services that are standard in high-income countries managing rare diseases.

Despite these adversities, the theme "**Strategies for Resilience and Hope**" demonstrated the capacity of caregivers to adapt and persevere. Several participants spoke of drawing strength from religious faith, online support groups, or the solidarity of other parents navigating similar challenges. Some learned to interpret biochemical reports, track symptoms meticulously, or manage emergency episodes with confidence—demonstrating an evolution from initial helplessness to skilled caregiving. These accounts echo the resilience model discussed by Raina et al. (2005), wherein caregivers with strong coping mechanisms, family support, and access to information are better equipped to sustain caregiving over the long term. However, it is important to note that resilience should not be romanticized or seen as a substitute for systemic support. Many caregivers in this study described "surviving" more than "thriving."

The **cultural context** plays a critical role in shaping caregiver experiences. Indian families often operate within hierarchical and gendered structures, where mothers are disproportionately burdened with caregiving responsibilities. Few fathers in this study were actively involved in day-to-day care, and extended family support was variable. This gender disparity is consistent with previous Indian research in chronic pediatric illness caregiving (Chadda & Deb, 2013). Additionally, the absence of rare disease awareness among school staff, employers, and community organizations contributes to the social isolation of caregivers, making it difficult for them to integrate into mainstream life.

The **implications** of this study are wide-ranging. Firstly, it underscores the urgent need to strengthen rare disease policy implementation in India, particularly in ensuring timely diagnosis, equitable access to treatment, and provision of lifelong support services. Secondly, healthcare systems must be trained to adopt a family-centered approach that includes psychosocial support for caregivers. Thirdly, there is a need for caregiver education modules that are language- and culture-specific, focusing not only on medical management but also on emotional coping, legal rights, and social welfare access. Finally, investment in community-based support mechanisms such as parent groups, helplines, and respite services could significantly improve the quality of life for caregivers.

Limitations of this study include a relatively small and geographically confined sample, which may not represent the full diversity of experiences across India. The voices of fathers, siblings, and other family members are underrepresented. Future research could expand to include longitudinal data, a wider regional sample, and a comparative perspective between caregivers of different types of IMDs or chronic disorders.

CONCLUSION

This qualitative study provides valuable insights into the multifaceted challenges faced by caregivers of children diagnosed with Inherited Metabolic Disorders (IMDs). Through in-depth thematic analysis, it is evident that the caregiving journey is shaped by emotional upheaval, demanding routines, systemic healthcare gaps, financial strain, and social

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isolation. Despite these overwhelming challenges, caregivers exhibit resilience, adaptability, and commitment that often go unrecognized in both medical and policy frameworks.

The findings underscore the urgent need to shift from a solely patient-centered model of care to a family-centered approach, where caregivers are acknowledged as key stakeholders in the child's well-being. There is a critical requirement for structured psychosocial support, improved access to medical nutrition therapy, culturally appropriate education, and caregiver-friendly health policies. Initiatives such as caregiver counseling, community support networks, financial aid programs, and rare disease advocacy must be strengthened to ensure a holistic care environment.

Moreover, the study highlights the lack of awareness, diagnosis delays, and poor policy implementation, calling for systemic reforms in rare disease management. Strengthening early screening programs, training healthcare professionals, and ensuring affordable treatment options can substantially alleviate caregiver burden.

In conclusion, while caregivers continue to act as pillars of strength for children with IMDs, their own quality of life must be protected and prioritized. Recognizing, supporting, and empowering caregivers through comprehensive and compassionate healthcare, policy intervention, and societal awareness is not only ethical, it is essential for improving long-term outcomes for children with rare metabolic conditions.

REFERENCES

- Bhattacharya, S. (2020). *Rare diseases in India: Time for cure-driven policy initiatives and action*. *Current Science*, 118(10), 1500–1506.
- Blau, N., Duran, M., Gibson, K. M., & Dionisi-Vici, C. (Eds.). (2018). *Physician's Guide to the Diagnosis, Treatment, and Follow-Up of Inherited Metabolic Diseases*. Springer
- Braun, V., & Clarke, V. (2006). *Using thematic analysis in psychology*. *Qualitative Research in Psychology*, 3(2), 77–101.
- Chadda, R. K., & Deb, K. S. (2013). Indian family systems, collectivistic society and psychotherapy. *Indian Journal of Psychiatry*, 55(Suppl. 2), S299–S309.
- Creswell, J. W., & Poth, C. N. (2018). *Qualitative Inquiry and Research Design: Choosing Among Five Approaches* (4th ed.). Sage
- Feeley, C. A., Sereika, S. M., Chasens, E. R., Siminerio, L., Charron-Prochownik, D., Muzumdar, R. H., & Viswanathan, P. (2021). Sleep in parental caregivers and children with Type 1 diabetes. *Journal of School Nursing*, 37(4), 259–269.
- Fekete, U., Thomas, D. S., Y. D. K., & Arulappan, J. (2021). Health-related quality of life of caregivers of children and adolescents with phenylketonuria: A systematic review. *Global Pediatric Health*, 8.
- Lincoln, Y. S., & Guba, E. G. (1985). *Naturalistic Inquiry*. Sage.
- O'Brien, D. (1979). Dietary management of inborn errors of amino acid metabolism. *Postgraduate Medicine*, 65(4), 133–138.
- Pelentsov, L. J., Fielder, A. L., Laws, T. A., & Esterman, A. J. (2016). The supportive care needs of parents with a child with a rare disease: A qualitative descriptive study. *Journal of Pediatric Nursing*, 31(3), e207–e218.
- Raina, P., O'Donnell, M., Rosenbaum, P., Brehaut, J., Walter, S. D., Russell, D., ... Wood, E. (2005). The health and well-being of caregivers of children with cerebral palsy. *Pediatrics*, 115(6), e626–e636.

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- Sanderson, S., Green, A., Preece, M. A., & Burton, H. (2006). The incidence of inherited metabolic disorders in the West Midlands, UK. *Archives of Disease in Childhood*, 91(11), 896–899.
- Saudubray, J.-M., Baumgartner, M. R., & Walter, J. H. (Eds.). (2016). *Inborn Metabolic Diseases: Diagnosis and Treatment* (6th ed.). Springer.
- Siddiq, S., Wilson, B. J., Graham, I. D., Lamoureux, M., Khangura, S. D., & Tingley, K. (2016). Experiences of caregivers of children with inherited metabolic diseases: A qualitative study. *Orphanet Journal of Rare Diseases*, 11, 10.
- Waters, D., Adeloye, D., Woolham, D., Wastnedge, E., Patel, S., & Rudan, I. (2018). Global birth prevalence and mortality from Inherited Metabolic Disorders: a systematic analysis of the evidence. *Journal of Global Health*, 8(2), 021102

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Conflict of Interest

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