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Qualitative needs assessment of financial impact on caregivers of children living with rare diseases

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ABSTRACT

Purpose: Families caring for children with rare diseases face sustained financial, emotional, and systemic challenges, yet cross-disease, cross-regional data remain limited. This study investigates the financial impact of caregiving from the perspective of rare disease advocacy organisations, rather than by caregivers themselves, focusing on costs, structural barriers, and potential reforms.

Methods: Semi-structured interviews were conducted with 45 rare disease organisations (24 U.S., 21 Europe). Purposive and snowball sampling ensured breadth. Interviews were transcribed, translated where needed, and thematically coded by six analysts using a validated framework (Cohen's $\kappa \geq 0.80$). Frequency analysis quantified recurring themes; narrative synthesis integrated cross-case patterns.

Findings: Eighty-two percent of organisations reported chronic caregiver financial distress, with average household income losses of €500–€2000 per month. Out-of-pocket costs for therapies, assistive devices, home adaptations, and travel were widely cited and often unreimbursed. Structural barriers, including restrictive eligibility, administrative complexity, and geographic inequities, were reported in more than 250 references. Approximately 80 % of caregivers were women, disproportionately facing employment disruption and emotional strain. Secondary impacts included debt, housing instability, marital breakdown, and reduced educational participation for affected children.

Conclusion: The financial toll of rare disease caregiving is compounded by fragmented support systems and inequitable access to benefits. Advocacy organisations identify urgent needs for caregiver recognition and compensation, streamlined benefit processes, expanded coverage for non-clinical costs, and harmonized cross-regional standards. These findings provide a policy-relevant evidence base to address structural drivers of caregiver financial hardship.

Introduction

Rare diseases are individually uncommon but collectively affect between 300 million and 400 million people worldwide [1–3]. Defined as conditions affecting fewer than 5 per 10,000 individuals in the European Union or fewer than 200,000 individuals in the United States, there are between 6000 and 8000 recognized rare diseases [1, 4]. Approximately 70 % of persons living with a rare disease are children, and around 95 % of these diseases lack curative treatment [3]. The diagnostic journey is complex and prolonged, families often wait over a year for an accurate diagnosis, and in some cases, as long as five years [5, 6]. These challenges place enormous pressure not only on affected individuals but also on their caregivers, who are often family members providing long-term, uncompensated support.

While clinical, diagnostic, and therapeutic complexities of rare diseases have received growing attention, less research has focused on the lived experience of caregivers, particularly those caring for children. Caregivers, most often parents, play a central role in managing rare pediatric diseases, providing medical coordination and emotional support while bearing financial burdens such as lost income due to employment disruption, out-of-pocket medical costs, adaptive equipment, and housing or travel modifications [2, 6]. Caregivers frequently operate in fragmented, under-resourced systems not tailored to meet long-term needs of rare disease families [7–11].

Cross-national cost-of-illness studies have demonstrated that rare disease caregiving generates substantial non-medical expenditures, lost income, and long-term financial insecurity for families across Europe and North America, yet these analyses typically rely on caregiver self-

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report and remain limited in geographic scope [12]. Parallel literature has documented significant emotional and psychosocial burden, including chronic stress, anxiety, identity disruption [13], and deteriorating mental health among caregivers of children with complex conditions [14–16]. However, far less is known about how structural factors shape or exacerbate these burdens; factors include administrative requirements, eligibility thresholds, fragmented insurance systems, and employment constraints [7, 8]. Existing studies rarely integrate these elements or examine how organisations that support rare disease families perceive such barriers across different national systems [12]. To our knowledge, no multi-country analysis has synthesized organisational insights on financial hardship, structural barriers, and employment disruption [7, 8, 17, 18]. This gap underscores the need for a cross-regional, organisation-level assessment capable of capturing the systemic forces that influence caregiver vulnerability.

Methods

The objective of this study was to identify and characterize the financial needs of families caring for a child with a rare disease through a structured qualitative needs assessment. Forty-five organisations agreed to participate. Of these, 24 were based in the United States and 21 in the European Union, representing a diverse range of rare diseases, including both ultra-rare and more commonly recognized conditions (Fig. 1). Participating organisations varied in size, with the smallest supporting approximately 30 families and the largest serving over 50,000 registered individuals. Disease areas included metabolic, neuromuscular, genetic, neurodevelopmental, and immunological disorders.

Through grounded theory and validated coding protocols [19, 20], institutional perspectives were captured on the nature, extent, and structural determinants of financial hardship in caregiving. The study examines these issues from the vantage point of rare disease organisations, which serve as intermediaries between families and health or social systems; it does not analyze direct caregiver interview.

Recruitment of rare disease associations

The initial phase of this needs assessment was conducted between June and September 2023. The project team finalized the research protocol, reviewed the literature on caregiver financial burden in rare disease, and developed the structured interview guide. The target was to recruit 45 rare disease organisations from across the United States and European Union, ensuring representation across geographies, disease types, and organisational models.

Literature landscape scan

A targeted literature landscape scan was conducted during protocol development to identify existing evidence on caregiver financial burden [7], structural barriers to support, and the psychosocial impact of rare disease caregiving [21]. Prior cross-national studies have shown that rare disease families experience substantial non-medical expenditures, lost income, and long-term financial insecurity [13, 22]. Research on emotional and psychosocial burden consistently reports chronic stress, anxiety, identity disruption, and deteriorating mental health among caregivers of children with complex disorders [9, 13, 22]. Additional work highlights how administrative complexity, eligibility thresholds, fragmented insurance systems, and geographic inequities amplify these burdens [3, 23, 24]. Studies examining employment effects similarly document reduced working hours, workforce exit, and long-term income loss among caregivers [10, 25, 26]. However, the literature includes almost no multi-country analyses reflecting the perspectives of rare disease organisations on financial hardship and system-level barriers [12]. These findings informed development of the interview guide and ensured that domain questions targeted clearly identified gaps in the existing evidence base [27, 28].

Sampling combined purposive, convenience, and snowball strategies [29–31]

Purposive sampling was used to ensure representation of

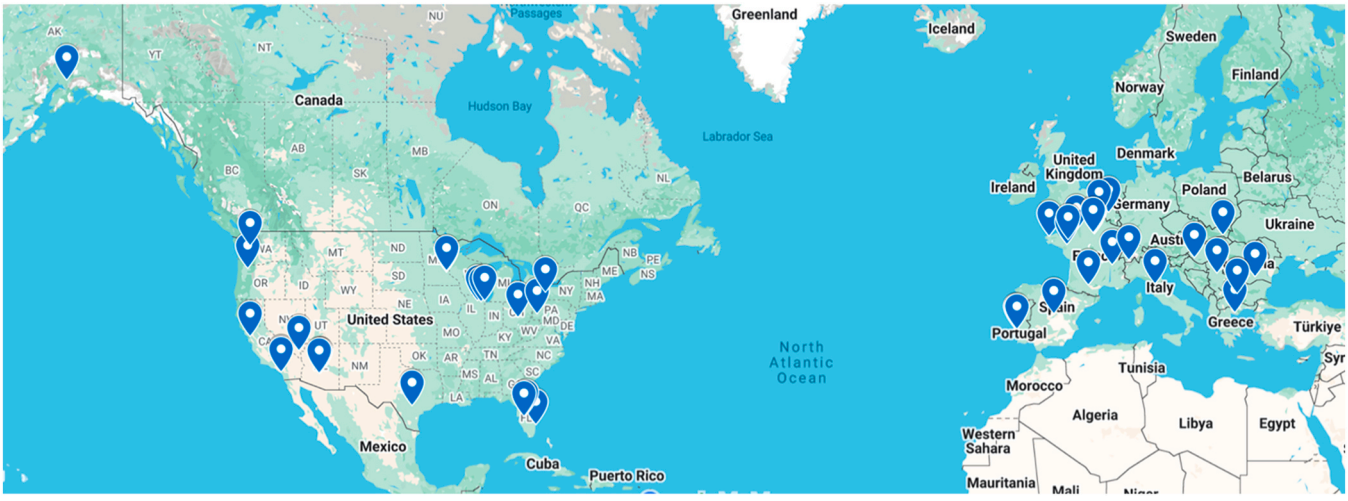

Fig. 1. Map of rare disease associations: Acid Maltase Deficiency Association, AIRG Belgique, All Things Kabuki, Angelman Syndrome Foundation, Association Bernard Pépin pour la maladie de Wilson, Association Neurofibromatoses et Recklinghausen, Association of Children with Mitochondrial Diseases, Association Smith Magenis 17 France, Association Valentin ACAP (France), Associação de Síndrome de Angelman Portugal, Batten Disease Support and Research Association (BDSRA), BCM Families Foundation, Bear Necessities Pediatric Cancer Foundation, Brylan's Feat Foundation, Bulgarian National Alliance of People with Rare Diseases, Cure 4 The Kids Foundation, Cure GM1 Foundation, Fundación Síndrome de Dravet, Fragile X France, Gaucher Community Alliance, Génération 22, Global Genes, Gopher Buddies, Hellenic Friedreich's Ataxia Association, HopeKids, Hunter's Hope Foundation, Italian Gaucher Association ODV, Kcnq2 France Développement, La vie par un fil, Les enfants du jardin, Magyar Prader-Willi Szindróma Egyesület, MaRaVal (Suisse), Metachromatic Leukodystrophy Foundation, National Association of Patients with Mitochondrial Diseases in Bulgaria, Nacionalna organizacija za retke bolesti Srbije, Northwest Rare Disease Coalition, PACER Center, Project Alive, Rare Givers (formerly Angel Aid), RD-Portugal – União de Associações das Doenças Raras de Portugal, Slovak Cystic Fibrosis Association, Sickle Cell Foundation of Minnesota, Translational Genomics Research Institute (TGen), Un défi de taille, Un sourire avec Hadrien, United Leukodystrophy Foundation, VACTERL Network, and Wishes & More.

organisations with diverse geographic coverage, operational capacities, and scopes of activity, while convenience and snowball sampling facilitated contact with eligible associations through established rare disease networks.

A primary contact list was generated from participants in a prior research [32] and expanded using publicly available directories from Rare Diseases Europe (EURORDIS), the National Organization for Rare Disorders (NORD), and Rare Diseases International (RDI). Organisational eligibility was confirmed through public review of website content, mission documents, and social media activity. Selection criteria emphasized diversity in geography, family reach, and experience supporting children with rare diseases [33, 34]. Formal invitations were distributed via email with a study overview, confidentiality terms, and scheduling options. Follow-up communications were issued at one- and two-week intervals.

Seventy-eight organisations were contacted, 38 from the United States and 40 in Europe. Forty-five organisations (24 from the United States, 21 from Europe) agreed to participate in a 60-minute structured interview representing a response rate of, 58 %. Non-participating organisations (n = 33) were distributed across the United States and Europe, including the Netherlands, Germany, Austria, Sweden, Poland, Denmark, Croatia, Luxembourg, Finland, and Ireland. Based on publicly available data such as website materials, annual reports, and organisational mission statements, no systematic differences were identifiable between participating and non-participating organisations in terms of size, disease focus, or operational model.

Structured interview methodology

Structured interviews were conducted as described previously [32, 35], from October 2023 to March 2024 in English, French, or Spanish. Interviews were led by trained interviewers using a standardized script with probing questions. All interviewees participated in their organisational and professional roles and were not asked to provide personal, identifiable, or private information about themselves or others. No health, demographic, or sensitive personal data were collected, and no individual caregiver testimony was solicited. All findings reflect organisational perspectives on service delivery and system-level challenges. Verbal consent was obtained from all participants, and data were anonymized prior to analysis.

Sessions were conducted virtually, lasted approximately 60 min (range: 52–67 min), and were recorded following verbal consent. Transcripts were produced verbatim and translated into English where required. The interview guide included 26 questions grouped into seven thematic domains:

1. Opening Questions: Organisations described their geographic reach, family base, and the forms of support they provide.
2. Gender Bias and Caregiver Profile: Participants discussed the gender distribution of caregiving, disparities in financial burden, and unequal access to financial assistance.
3. Work and School Impact: Participants addressed caregivers' employment disruption, estimated income loss, and secondary employment to cover care-related costs. School disruption for affected children was also explored.
4. Insurance and Out-of-Pocket Costs: Organisations reflected on the role of insurance and shared examples of unremitted caregiver expenses.
5. Financial Support Access: Participants identified available financial assistance and the barriers encountered when applying for or accessing such support.
6. Impact on Quality of Life: Participants discussed how financial strain affects family well-being and caregiver functioning.
7. Final Thoughts: Organisations offered views on systemic needs, the adequacy of current supports, and potential mechanisms for reform.

These domains were drawn from prior literature and refined during protocol development to accommodate cross-national variation in caregiver support structures and policy environments [3,5–8,17,18]. During qualitative analysis, these seven interview domains were consolidated and, in one case, split into eight analytic domains, which structure the Results section and Table 1.

Table 1
Highest-frequency themes across analytic domains. Responses from structured interviews from 45 rare disease organisations. The term "reference" is used to indicate the frequency with which a theme or concept appeared in the coded data. For example, the number of times "fundraising" was mentioned by organisations was analyzed to highlight the recurring focus of fundraising in how organisations establish strategies to provide financial assistance to the families they aim to serve. These counts were not meant to imply relative importance but were instead used to identify dominant themes. Further detail is provided in the supplementary material.

Domain	Highest-Frequency Theme(s)	Frequency Evidence	Summary Interpretation
Domain 1 – Opening Questions: Scope, Reach & Duration	Broad geographic reach and long-term support	40 orgs described operational scope; 25 national; 12 international; 20 orgs support 100–500 families	Most organisations provide sustained, wide-reaching support, often acting as main reference points for their disease communities.
Domain 2 – Insurance, Financial Strategies & Coping Mechanisms	Financial stressors and inadequate coverage	520 references to financial stress; 167 references to insufficient coverage	Financial vulnerability is structural and ongoing, reflecting major gaps in insurance and social security systems.
Domain 3 – Gender Roles & Disparities	Mothers as primary caregivers; employment disruption	36 orgs reported women as primary caregivers; 54 references; [13] orgs described gendered financial impact	Care labor is overwhelmingly gendered, with mothers absorbing emotional, logistical, and financial consequences.
Domain 4 – Structural Barriers & Accessing Financial Support	Administrative burden and complexity	143 references to complex procedures; 29 references to lack of information	Bureaucratic hurdles block or delay access to essential support, intensifying inequality and caregiver fatigue.
Domain 5 – Direct & Hidden Costs of Caregiving	Therapy, equipment, travel, home adaptations	95 references to therapy/equipment; 65 travel; 37 home adaptations	Families incur substantial non-reimbursed costs that grow with disease severity and persist over time.
Domain 6 – Income Loss & Employment Impact	Workforce exit and reduced hours	37 orgs reporting parent exit; 29 reduced hours; income losses €500–€2000/month	Employment disruption is widespread, long-lasting, and financially destabilizing.
Domain 7 – Changes in Family Life & Quality of Life	Mental health strain and identity collapse	487 QoL references; [26] anxiety; 34 need for psychological support	Caregiving reshapes family roles and identity, causing sustained emotional strain and social isolation.
Domain 8 – Employment Disruption, School Limitations & Systemic Support Needs	Parental workforce withdrawal & educational disruption	37 orgs: parent exit; 29 orgs: reduced hours; 21 orgs: school withdrawal	The dual burden of employment and education disruption underscores the need for systemic social and workplace reform.

Interview framing and respondent instructions

At the start of every interview, participants were explicitly instructed to respond *in their organisational role* rather than as private individuals. Several respondents voluntarily disclosed that they were themselves caregivers, parents, or persons living with a rare disease; however, this personal information was not solicited, coded, or analyzed as lived-experience testimony. Following a reviewer request, we re-analyzed the interview dataset and confirmed that 32 of 45 interviewees reported having personal proximity to rare disease through family or caregiving experience. All 45 interviewees indicated direct and regular contact with families or individuals living with a rare disease as part of their organisational responsibilities. Nonetheless, all analytic interpretations in this study derive exclusively from *organisational* perspectives.

Data collection and analysis

Interview transcripts were imported into NVivo (QSR International, Burlington, MA, USA) for coding and analysis as described previously [32, 35]. A multi-level codebook was developed using grounded theory. Deductive codes were informed by the interview domains, while inductive codes emerged from iterative transcript review. The final codebook comprised three tiers: primary domains, subthemes, and analytic codes. Financial ranges reflect organisational recollection and should not be interpreted as verified household economic data

Procedures included:

a) Coding and validation: Six coders, working in teams of two, double-coded each transcript for a total of 90 passes across 45 interviews. Inter-rater reliability was assessed using Cohen's kappa coefficient; all teams maintained $\kappa \geq 0.80$. Regular team meetings were held to refine code definitions, discuss emerging patterns, and resolve discrepancies.
b) Frequency of themes: The term "referenced" was used to indicate the number of times a given theme or concept appeared in the coded data. For instance, references to "awareness" were analyzed to capture recurring non-financial support mechanisms reported by organisations. These counts were not intended to imply relative importance but served to identify dominant and recurrent themes.
c) Thematic analysis: Beyond frequency counts, the analysis focused on the contextual and qualitative significance of emergent themes. Attention was paid to the framing, language, and strategic approaches described by organisations, enabling a nuanced understanding of the financial burdens experienced by caregivers.
d) Team calibration: Four structured team meetings were held during the coding period to ensure data quality and analytic consistency. Feedback from coders was used to revise the codebook iteratively and standardize interpretation across transcripts.

The final dataset yielded 2817 discrete coded references across 74 thematic subcodes nested within eight major analytic domains, derived from the seven interview domains described above (Table 1 and Supplementary Online Material). Themes were analyzed for recurrence, co-occurrence, and salience. Geographic cross-tabulations were used to compare U.S. and EU responses. Quotes were selected to illustrate key findings; they were anonymized and, where necessary, translated into English with minor edits for clarity.

Results

Interpretive framing

All results presented below reflect what *organisations reported* during structured interviews. No statements in this section represent individual caregiver testimony. The findings are derived from organisational perspectives rather than from personal lived experience. All financial amounts described in the Results represent estimates conveyed by participating organisations and should not be interpreted as audited, verified, or caregiver-reported financial data.

Participants

Forty-five rare disease associations participated in structured interviews between October 2023, and March 2024 recruited by combined purposive, convenience, and snowball strategies [29–31]. All organisations provided direct support to caregivers of children living with rare diseases, either as their sole focus or as part of a broader population. Participants included executive directors (n = 18), program managers (n = 14), family liaisons or advocates (n = 9), and board-level representatives (n = 4). The associations reflected a heterogeneous but broadly comparable landscape between the United States (24 associations) and Europe (21 associations). Their size extended from, volunteer-led groups supporting fewer than 30 families to national or multinational associations representing more than 50,000 members. Staffing patterns showed regional contrasts with European associations showing almost total reliance on volunteers. By contrast, U.S. associations averaged 32 ± (SD) 82 employees (range 0–350), several maintaining professional teams. Financial capacity varied widely. European associations reported mean annual income of $137,091 ± $215,387 (range: $1486–$727,276) (Conversion Euro to USD=1.17), with nearly all operating below $1 million. U.S. associations reported mean annual income of $7089,130 ± $14,254,558 (range: $30,163–$49,600,000). Most US associations combined philanthropic campaigns, grants, and donations, while European associations relied more heavily on public subsidies and community fundraising.

Despite the differences in resources, the missions of these associations converged. U.S. organisations more often provided direct financial grants, typically $500–$1000, targeted at urgent needs such as transportation, medical bills, or equipment. European associations prioritized support for equipment, therapy reimbursement, and medical travel, aligning with national health frameworks yet constrained by regional disparities. Across both regions, non-financial services were central, including peer groups, counseling, hotlines, awareness campaigns, and advocacy.

All interviews were completed in full and met transcription quality thresholds. Interviewees represented a range of positions within their organisations, including executive leadership, program managers, family liaisons, and advocacy or board representatives. These roles differ substantially in their day-to-day proximity to families. For example, family liaisons often engage directly with caregivers seeking support, whereas executive directors may interact primarily through policy, governance, or high-level program oversight. Because of this heterogeneity, organisational perspectives in this study are shaped by the respondents' professional responsibilities rather than by personal caregiving experiences. All findings therefore reflect institutional vantage points filtered through role-specific mandates. The final dataset yielded 2817 discrete coded references across 74 thematic subcodes nested within eight major analytic domains, reflecting refinement of the seven interview domains during coding (Table 1 and Supplementary Online Material).

Domain 1: opening questions – financial support, scope, reach, and duration of organisational work

Organisations described their geographic reach, the families they serve, and the services they provide to children with rare diseases. Across all interviews, financial assistance was the most frequently referenced form of support, appearing 110 times and identified as a priority activity by 37 of 45 organisations (19 U.S.; 18 Europe). Respondents consistently framed financial aid not only as a practical service but as central to their mission and identity within the rare disease

community.

Despite its importance, providing sustained financial support was widely described as challenging, particularly for small or volunteer-run groups. Limited funding and infrastructure forced many to rely heavily on fundraising, explicitly mentioned in 19 interviews (12 Europe; 7 U.S.), often through community events or targeted campaigns. As one European participant noted, "We want to help every family, but sometimes must prioritize based on urgency and available funds."

The nature of financial assistance varied. Some organisations offered recurring stipends, while others provided ad hoc grants or reimbursement schemes. Most support targeted medical costs and essential equipment, including adaptive strollers, feeding devices, AAC tools, and mobility or vehicle modifications, reflected in [26] references to "Medical equipment and home improvements" (19 U.S.; 7 Europe). Many described this work as "filling the gap left by government or insurance coverage."

In the U.S., eight organisations offered direct grants (nine references), typically $500–$1000. Although modest relative to monthly costs, these grants were considered vital lifelines for emergencies, including travel, housing, and bereavement needs, particularly when public systems were slow or inaccessible.

Organisations also reported supporting caregiver health and well-being, including doctor visits and therapy ([16] references: 10 Europe; 6 U.S.). Further assistance included medical travel support ([10] references: 7 Europe; 3 U.S.) and education-related aid, scholarships, adaptive learning supports or required accommodations ([7] references: 3 Europe; 4 U.S.). These services were described as critical to maintaining both physical stability and educational continuity.

Across interviews, organisations stressed that even modest financial assistance had outsized impact on family well-being. As one respondent noted, "It's not just paying a bill, it's restoring a sense of control." Many hoped to expand such programs but cited structural constraints, funding availability, staffing, and policy limitations, that restricted their ability to meet high and increasing demand.

Domain 2: opening questions: scope, reach, and duration of organisational work

In response to opening questions, organisations described the scale and structure of their services. Most positioned themselves as long-term partners to families, operating with broad geographic reach and inclusive models that serve families across income levels. Although low-income households often face the greatest need, few organisations apply income-based eligibility criteria.

Forty organisations (185 references) detailed their operational scope. As shown in Fig. 1, 25 organisations (18 Europe; 7 U.S.) operated nationally, serving as primary reference points for their condition within their country. Twelve organisations (5 Europe; 7 U.S.) described international reach, typically enabled by collaborations or online platforms. Five U.S. groups reported state-level focus, reflecting the structure of localized care and insurance systems. Respondents emphasized that, in practice, rare disease support often extends beyond formal jurisdictional boundaries, especially where expertise is concentrated or public support is fragmented.

Reported reach varied by disease prevalence and organisational capacity. Twenty organisations (10 Europe; 10 U.S.) supported 100–500 families, the most common range. Smaller groups served fewer than 50 families, often for ultra-rare conditions. A minority supported more than 1000 families, typically those with broader missions or strong digital visibility. This variability reflects substantial sectoral heterogeneity ranging from small volunteer-led associations to established NGOs with national or international infrastructure.

The duration of support also varied. Twenty organisations (13 U.S.; 7 Europe) provided long-term assistance, often beginning at diagnosis and continuing throughout childhood. Fourteen (7 U.S.; 7 Europe) offered episodic or as-needed support, while nine reported hybrid models combining sustained and intermittent engagement. Across models, flexibility was emphasized: families' needs fluctuate during crises, transitions, or disease progression, requiring organisations to adapt accordingly.

Thirty-four organisations reported serving families across socioeconomic backgrounds. Eight (4 Europe; 4 U.S.) noted that most of their beneficiaries were low-income, largely due to broader systemic inequities. Although income rarely determined eligibility, economic hardship strongly influenced service intensity, responsiveness, and the types of support families requested.

Organisations collectively described a commitment to accessible, relationship-centered support for all families affected by rare diseases. At the same time, many acknowledged the tension between aspiration and capacity: while most aim to serve their entire disease community, limited resources often require prioritizing the most urgent needs. Even within these constraints, participants emphasized their shared goal of broad, inclusive, and enduring support.

Domain 3: gender roles and disparities in caregiving

Organisations reported strikingly consistent gendered caregiving patterns. Thirty-six organisations (18 Europe; 18 U.S.) stated unequivocally that women, specifically mothers, are the primary caregivers, a distribution reflected in Fig. 2 and supported by 54 references. Several estimated that "~90 % of caregivers are women," underscoring the deep social and structural norms shaping responsibility for coordinating medical care, managing appointments, and providing day-to-day support. Only five organisations described gender-balanced arrangements, and none reported fathers as the caregiving majority. Respondents frequently linked this pattern to the gender pay gap, noting that the lower-earning parent, typically the mother, was more likely to leave the workforce.

Participants also noted that the definition of "caregiver" can diverge from household reality. In some families, the full-time working parent (often the father) was still considered a caregiver due to financial contribution and shared decision-making, reinforcing a "team-based" model even when daily tasks were unequally distributed.

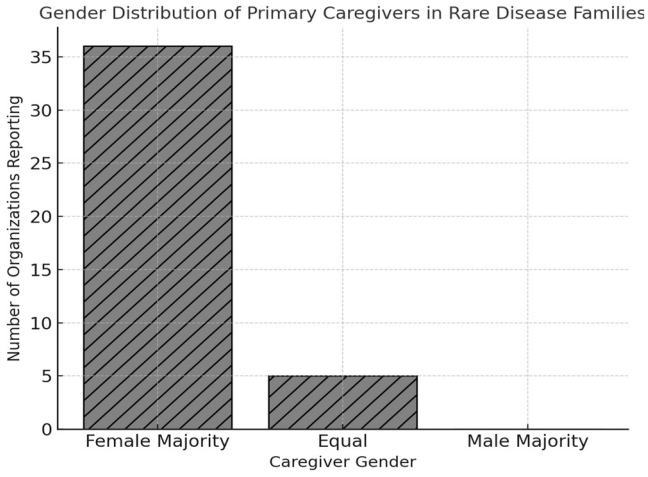

Fig. 2. Gender distribution of primary caregivers among rare disease families (organisational reports). Bar chart showing the number of organisations (N = 45) reporting each caregiving pattern: female majority, gender-equal, male majority. Counts reflect organisations' characterizations during structured interviews analyzed with a multi-coder grounded-theory approach (NVivo; six coders; $\kappa \geq 0.80$). Bars represent organisation counts, not population-weighted estimates; no error bars are shown because this is a qualitative frequency summary. "Female majority" indicates respondents identified mothers as the primary coordinators of care and logistics; this pattern was also reinforced by multiple coded references noting mothers' predominance.

Of the 28 organisations able to comment on gender-based access to assistance, 25 reported no formal gender differences in eligibility, as benefits were tied to the child's diagnosis rather than the caregiver's gender. However, four organisations (3 Europe; 1 U.S.) described indirect disparities, including fathers' ineligibility for certain leave policies and social norms discouraging men from seeking help. These examples illustrate how structural and cultural factors shape caregiving roles even in the absence of explicit policy barriers.

Among the 24 organisations addressing this question, 13 reported that financial burden was shared equally, while eight described disproportionate impact on mothers, largely due to reduced employment or complete workforce exit. Respondents emphasized that financial strain reinforces gendered caregiving patterns over time.

Several organisations highlighted the precarious position of single parents, particularly single mothers, who shoulder both caregiving and income generation. A smaller group (five organisations: 3 Europe; 2 U.S.) noted that fathers who serve as sole earners may experience distinct financial stress, balancing employment demands with caregiving responsibilities.

Overall, these findings reveal persistent gender disparities in caregiving roles and financial strain. While widely recognized, more nuanced inequities, such as job flexibility, language around entitlement to benefits, and cultural norms shaping help-seeking, continue to influence the lived experiences of families.

Domain 4: insurance, financial strategies, and coping mechanisms

Organisations described substantial financial strain resulting from gaps in insurance coverage, geographic inequities, and the fragile coping strategies families rely upon. Across the 45 interviews, 520 references concerned financial stressors and adaptation.

A dominant theme across both regions was incomplete or inconsistent insurance and social security support. Fig. 3 summarizes these patterns. In total, 167 references described insufficient or absent coverage for essential expenses. Of these, 98 references highlighted uneven coverage, distributed similarly between Europe (51) and the U.S. (47). Families were commonly reimbursed for core medical care but not for therapies, assistive technologies, nutritional supplements, or rehabilitation, services viewed as essential to maintaining quality of life.

Only a small minority ([10] references; 5 Europe, 5 U.S.) reported experiences of near-complete coverage. By contrast, [18] references (10 Europe; 8 U.S.) described families receiving no support, often due to rarity of the condition, absence of formal diagnosis, or policy exclusions.

Geographic disparities intensified inequity. Twenty-nine references (20 U.S.; 9 Europe) described how state or regional differences produced a patchwork of benefits. In several instances, families relocated across states or within countries, to obtain more favorable insurance or social security provisions. These examples underscore the extent to which financial protection depends on location rather than need.

Families employed a wide range of coping mechanisms in response to inadequate coverage (Fig. 4). These strategies, captured in 316 references and summarized in Fig. 4, illustrate both creativity and vulnerability.

The most common response was reduction in parental employment, predominantly among mothers (85 references: 41 Europe; 44 U.S.), including reduced hours or full workforce exit. This frequently required the other parent to increase working hours or take multiple jobs, compounding emotional and financial strain.

Families also relied on formal social assistance (75 references: 32 Europe; 43 U.S.), though availability and eligibility were inconsistent. Community fundraising was widespread (48 references: 20 Europe; 28 U.S.), with platforms such as GoFundMe or condition-specific campaigns functioning as essential safety nets.

Extended family support was another major resource (45 references: 27 Europe; 18 U.S.), providing financial help, childcare, or respite. Patient organisations helped navigate complex systems ([36] references: 29 U.S.; 7 Europe**), especially when formal pathways were inaccessible or unclear.

More affluent families occasionally drew on savings or investments ([12] references: 9 Europe; 3 U.S.), though participants stressed that this was viable only for high-income households. By contrast, many families described depleting savings ([19] references: 14 U.S.; 5 Europe**), selling property, or incurring debt through loans or credit ([14] references, evenly divided).

Taken together, these findings depict a fragmented financial landscape in which resilience depends more on personal networks, improvisation, and sacrifice than on reliable structural support. The economic

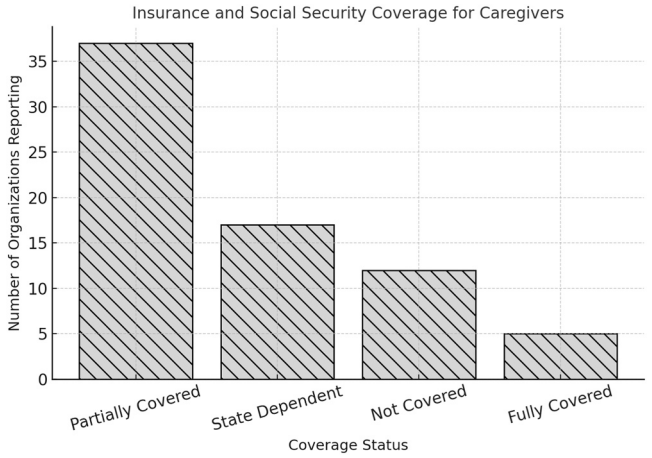

Fig. 3. Insurance/social-security coverage patterns reported by organisations (N = 45) supporting families of children with rare diseases. Bar chart showing the number of organisations reporting each pattern of coverage for the families they serve: partially covered, state/region-dependent, not covered, fully covered. Categories reflect how organisations described typical experiences within their context; the "state/region-dependent" category denotes geographic variability within a country. Bars represent organisation counts; they summarize qualitative reports rather than claims data. These distributions align with broader interview themes highlighting uneven benefits and a patchwork of provisions across jurisdictions.

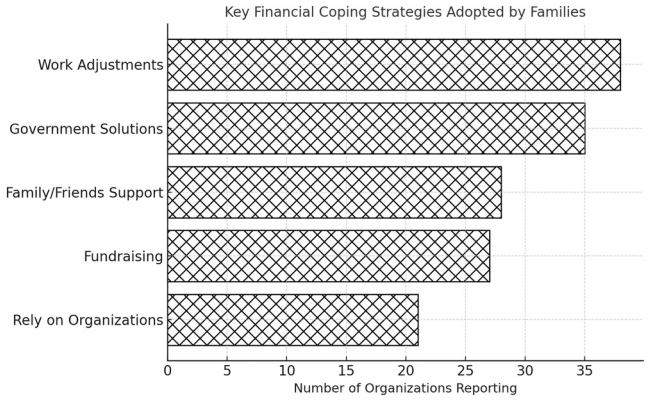

Fig. 4. Financial coping strategies used by families (organisational observations, N = 45). Horizontal bar chart indicating the number of organisations that observed families using each strategy: work adjustments (e.g., reduced hours, exit from workforce), government assistance support from family/friends, community fundraising, and reliance on patient organisations for navigation/support. Where available, organisations also provided multiple interview references per strategy (e.g., work adjustments 85 references), underscoring salience; however, bars show organisation-level counts, not reference totals. Categories are not mutually exclusive; a single organisation may report multiple strategies among different families. Findings derive from semi-structured interviews coded using a validated codebook (NVivo; $\kappa \geq 0.80$).

burden of rare disease caregiving is substantial and ongoing, often forcing families to make unsustainable decisions to compensate for gaps in insurance and public programs.

Domain 5: structural barriers and the complexity of accessing financial support

Organisations described significant structural obstacles that hinder families' ability to obtain financial assistance. Rather than reducing the burden of rare disease caregiving, many systems were perceived as amplifying inequity, emotional strain, and administrative fatigue. Across interviews, 42 organisations contributed 397 references to this domain.

Eligibility criteria were the most consistently cited obstacle (111 references). Respondents described requirements as rigid, opaque, and discouraging, with families frequently abandoning applications due to extensive documentation demands or unclear approval pathways. A recurrent concern was the exclusion of middle-income families, noted in [20] references (4 Europe; 16 U.S.), who earned above formal thresholds but could not absorb substantial out-of-pocket costs, leaving them in a systemic "blind spot."

Geographic disparities further contributed to inequity ([24] references: 11 Europe; 13 U.S.). In the United States, access to reimbursed benefits varied widely by state. In Europe, decentralization resulted in significant differences between and within countries, with local authorities determining eligibility and benefit levels. Organisations emphasized that this uneven governance created confusion and unequal access to support.

The application process itself constituted a major barrier (143 references). Families reported complex, lengthy procedures (54 references, evenly split across regions), often resulting in delays or withdrawal from the process. Respondents highlighted that the emotional cost stress, fatigue, and resignation, which was experienced at a time when families were already vulnerable. Lack of clear information was another prominent issue. Twenty-nine references (20 Europe; 9 U.S.) noted that families were often unaware of available benefits or how to apply, especially at diagnosis, reflecting gaps in communication and inter-agency coordination.

Secondary exclusion factors included language barriers ([5] references: 1 Europe; 4 U.S.), fear of judgment or mistrust of institutions ([15] references: 10 Europe; 5 U.S.), and stress-related withdrawal ([8] references: 2 Europe; 6 U.S.). Three organisations reported applications denied without explanation, raising concerns about discretionary decision-making and lack of transparency.

Although six organisations (4 Europe; 2 U.S.) expressed moderate satisfaction with centralized government portals, these remained exceptions. The dominant view was that systems were inaccessible, insufficiently flexible, and poorly aligned with caregivers' lived realities, particularly when one parent had already exited the workforce.

Taken together, these findings position structural and procedural barriers as central drivers of inequity in rare disease caregiving. These obstacles delay or block access to essential aid, exacerbate caregiver exhaustion, and disproportionately affect families already experiencing financial strain. In the absence of reliable formal support, many are pushed toward informal networks, personal debt, or unsustainable sacrifices, further entrenching inequality.

Domain 6: direct and hidden costs of caregiving

Organisations identified a broad set of visible and less apparent expenses associated with rare-disease caregiving (Fig. 5). Their responses depict a persistent, multi-layered financial burden comprising direct medical costs, indirect losses, and structural economic vulnerability. These expenses shift over time with disease progression, creating a dynamic and often unpredictable financial environment for families.

The most frequently cited costs related to medical care, therapies, and equipment (95 references: 46 Europe; 49 U.S.). While some services receive partial reimbursement, many essential therapies, speech, physical, psychomotor, fall outside standard coverage. Families also regularly pay for assistive technologies such as wheelchairs, stair lifts, or bathing chairs (64 references: 23 Europe; 41 U.S.), which are critical for daily living but commonly excluded from insurance schemes.

The centralization of rare-disease expertise requires frequent travel to specialist centers, mentioned in 65 references (25 Europe; 40 U.S.). Associated costs, transport, lodging, meals, represent ongoing out-of-pocket expenditures. Families also reported the need for substantial home adaptations ([37] references: 12 Europe; 25 U.S.), including widened doorways, ramps, and accessible bathrooms, to ensure safety and functionality.

Vehicle adaptations, or purchasing pre-adapted vehicles, were noted in [27] references (5 Europe; 22 U.S.). These costs are typically immediate and unavoidable, especially for children requiring frequent therapy or medical appointments. Several families reported monthly care-related expenses exceeding €3000, reflecting cumulative and

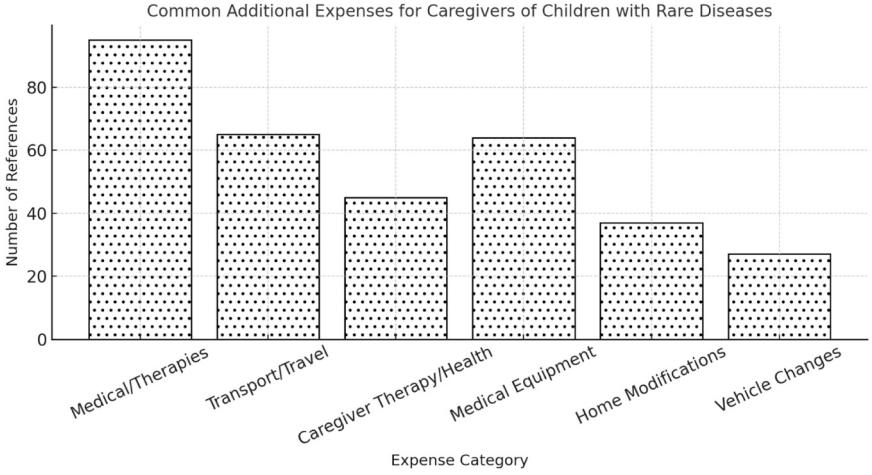

Fig. 5. Common additional expenses reported by organisations (N = 45) for families caring for a child with a rare disease. Bar chart depicting the number of coded references across interviews for major expense categories: medical care/therapies, transport and travel for care, medical equipment and assistive devices, home modifications for accessibility/safety, and vehicle adaptations/changes. Bars represent reference frequencies (indicator of how often a theme appeared in coded data), not the number of organisations or the magnitude of spending. Categories are not mutually exclusive and reflect the breadth of recurrent costs; exemplar narratives noted substantial out-of-pocket expenditures for equipment and travel, and occasionally very high vehicle-adaptation costs (NVivo; $\kappa \geq 0.80$).

intensive needs.

Beyond clinical care, families incur expenses for siblings' activities, adapted holidays, and recreational opportunities that preserve household cohesion. These secondary costs, although rarely covered, were described as essential to family well-being and an accurate reflection of how caregiving permeates everyday life.

Income reduction was a recurrent theme ([40] references: 16 Europe; 24 U.S.). One parent, most often the mother, commonly reduces working hours or leaves employment entirely, with long-term implications for earnings, career progression, and retirement security. Administrative demands also create financial strain: [15] references (8 Europe; 7 U.S.) described productivity loss due to time-consuming paperwork and appeals. Three organisations (all Europe) noted property damage attributable to the child's condition or behavioral challenges. Organisations reported that families commonly describe household income losses in the range of €500–€2000 per month. The combined effect is a high-risk financial profile in which increased costs overlay declining income, creating chronic vulnerability. Without sustained structural support, families risk debt, social exclusion, and reliance on unstable informal assistance.

Domain 7: changes in family life and impact on quality of life

Organisations consistently described caregiving for a child with a rare disease as transformative, isolating, and emotionally taxing. Impacts extended well beyond clinical logistics or financial strain, shaping daily life, family roles, and mental health. Across 487 references, participants depicted profound lifestyle, relational, and psychological disruption.

Mental health deterioration was a central concern. Caregivers were frequently described as having no personal time ([19] references: 10 Europe; 9 U.S.), with lives organized entirely around caregiving routines. Anxiety about the future appeared in [26] references, and the need for professional psychological support in [34] references (evenly split across regions).

Respondents described an "identity collapse," in which individuals ceased identifying as parents, partners, or professionals and instead saw themselves solely as caregivers [12], an intense, often isolating role with little emotional reciprocity. A persistent, unspoken fear about what will happen to the child after the caregiver's death compounded this strain.

The interaction of financial insecurity and emotional exhaustion was noted in [47] references (19 Europe; 28 U.S.). As caregiving demands intensified, so did financial pressure, eroding coping capacity and narrowing opportunities for rest or self-care. These dual burdens amplified stress throughout the household.

Siblings were also affected: [15] references (7 Europe; 8 U.S.) described feelings of neglect or emotional displacement as family life increasingly centered around the affected child.

Organisations reported widespread erosion of leisure and family activities. Families often stopped vacationing ([21] references: 15 Europe; 6 U.S.) and withdrew from leisure activities ([12] references: 7 Europe; 5 U.S.). In some cases, caregivers sacrificed their own health or nutrition to meet care demands ([10] references: 8 Europe; 2 U.S.).

Relocation was another common consequence. Twenty-two references (12 Europe; 10 U.S.) described families moving to be closer to specialized care; [10] references (6 Europe; 4 U.S.) described moves driven by financial necessity. These relocations often disrupted social networks and employment stability.

Financial deterioration was a recurrent theme. Families reported debt ([12] references), insolvency ([9] references: 4 Europe; 5 U.S.), and poverty ([10] references: 8 Europe; 2 U.S.). In contexts with limited caregiver recognition, families also struggled to access credit, benefits, or long-term planning tools, trapping them in short-term survival cycles.

These pressures had marked relational consequences. Eighteen references (8 Europe; 10 U.S.) noted higher divorce rates among caregivers, with chronic stress, financial hardship, and fractured identities undermining family stability.

Overall, organisations described caregiving as a pervasive, multi-domain burden that reshapes identity, erodes well-being, destabilizes relationships, and compounds financial hardship which reinforced the need for comprehensive, family-centered support systems.

Domain 8: employment disruption, school limitations, and needed systemic support

Participants described how caregiving for a child with a rare disease profoundly disrupts caregivers' professional trajectories and children's educational opportunities. These disruptions are sustained rather than episodic, driven by inadequate systemic support and inflexible workplace and school policies. Across 198 references, nearly all emphasized the extensive sacrifices families make in the absence of institutional adaptation.

Organisations reported widespread workforce withdrawal, particularly among mothers. According to organisational reports, in 37 (19 Europe; 18 U.S.), of the interviewed associations, families commonly include one parent who has permanently left employment to provide full-time care. This information reflects staff descriptions of the families they support and does not represent personal employment disclosures by the interviewees themselves. In 29 organisations (18 Europe; 11 U.S.), a parent significantly reduced working hours, often without adequate financial protection or workplace accommodation. Additional adaptations included shifting to lower-paid flexible work, taking unpaid leave, or continuing in positions under untenable conditions. Respondents noted that employers frequently misunderstood caregiving demands, and repeated absences sometimes resulted in job loss.

Financial strain also drove families to seek supplemental income. Twenty-seven references (17 Europe; 10 U.S.) described parents working multiple jobs or overtime. A smaller number attempted home-based work ([5] references: 1 Europe; 4 U.S.), though these arrangements were often precarious and insufficient to offset care-related income loss.

Children's education was similarly affected. School withdrawal was reported by 21 organisations (17 Europe; 4 U.S.), while frequent absences were noted in [14] references (9 Europe; 5 U.S.). Participation in school activities was commonly limited due to medical appointments, fatigue, or inadequate school accommodations. Some families turned to homeschooling ([4] references: 1 Europe; 3 U.S.), typically as a necessity rather than preference. Although a few organisations cited positive examples of inclusive schooling, most described education systems as insufficiently equipped to meet the needs of children with rare conditions.

Across 106 references, organisations called for wide-ranging reforms: more consistent health and social security coverage; legal recognition and financial compensation for caregivers; streamlined administrative processes; and improved educational supports. Additional priorities included expanded caregiver leave, enhanced mental health services, and greater availability of respite care.

Taken together, findings reveal a core contradiction: families adapt with extraordinary flexibility, yet the systems that should support them remain largely static. In this environment, stable employment and consistent schooling become fragile privileges, accessible only to those able to absorb significant personal and economic strain. Without policy reform, families will continue to sacrifice long-term stability in exchange for survival.

Discussion

This study offers insight into the economic and psychosocial burden experienced by families of children with rare diseases, as reported by advocacy organisations [3, 7, 8, 17]. Across national contexts, caregiving was consistently described as a disruptive force that is inadequately buffered by current health and social systems.

Our findings reinforce prior research documenting that the burden of

rare diseases extends well beyond the clinical setting [3]. Non-medical expenditures and lost income represent a major share of total disease burden, often accounting for 30–45 % of overall costs borne by families in Europe and North America (13. In our interviews, expenses related to complementary therapies, home adaptations, specialized nutrition, and assistive technologies, most of which are not reimbursed, emerged as dominant and recurring themes. This aligns with data from multi-country cost-of-illness studies that consistently show families absorbing extensive out-of-pocket costs over prolonged periods {Angelis, 2015 #5231, 14–16, 36, 37). These costs are unpredictable, increase with disease progression, and can permanently alter the economic trajectory of households.

Gendered caregiving dynamics were also strongly affirmed. In 80 % of organisations, women, typically mothers, were identified as the primary caregivers. This aligns with extensive literature documenting that female caregivers report higher levels of emotional and financial strain, more severe employment disruption, and reduced access to psychological support [6, 19, 22, 26, 37]. Although some research suggests gender parity in perceived burden [36], the actual distribution of care labor, time, and opportunity cost remains profoundly unequal. These disparities may also reflect persistent systemic gender roles and insufficient support for family leave and flexible employment, particularly in countries without paid caregiver protections [36].

A central finding across both continents was the failure of existing support systems to respond to the real-life needs of rare disease families [3, 17]. Eligibility thresholds, administrative burdens, and local policy inconsistencies create a fragmented safety net. Families reported being denied assistance due to minor income differences, misunderstanding of documentation, or territorial discrepancies. These themes echo prior research showing that even in countries with comprehensive healthcare, structural barriers and bureaucratic opacity severely limit access [33, 38–40]. Such mechanisms not only deter families from seeking help but actively widen inequalities between those who can navigate systems and those who cannot.

The mental health toll of caregiving emerged as a consistent theme [41]. Participants described identity erosion [12], social withdrawal, chronic anxiety, and emotional exhaustion [9, 18, 41]. These experiences are consistent with previous studies that demonstrate a heightened risk of psychological distress and depressive symptoms among caregivers in rare disease contexts Fröhlich [22]. Notably, caregiver well-being is rarely monitored formally within health systems, despite strong evidence linking caregiver strain to poorer patient outcomes and increased system costs [8].

Additional literature highlights the profound emotional burden reported by caregivers, with sustained stress and psychological fatigue commonly arising from prolonged care responsibilities [13]. Studies also describe marked shifts in caregiver identity, noting that many individuals experience a redefinition of self as caregiving demands increasingly dominate daily life [12]. Caregiver-reported studies similarly document substantial financial strain, including out-of-pocket expenditures, loss of income, and reduced long-term financial stability, which align closely with the organisational estimates described in this study [9]. Cross-national cost-of-illness analyses further demonstrate that rare disease caregiving imposes high and persistent economic burden across diverse health systems, reinforcing the international relevance of these findings [10].

We found employment disruption to be near-universal [42]. Parents, particularly women, left the labor force, reduced their hours, or accepted lower-wage, flexible work. Over time, this diminishes retirement security, depletes household savings, and curtails career advancement opportunities [26, 36, 39, 43]. These long-term impacts were compounded in single-parent households, or when the cost of care necessitated multiple jobs or home relocation. Children's education was also disrupted, with absenteeism, social exclusion, and informal homeschooling strategies substituting for accessible educational accommodations.

Importantly, advocacy organisations articulated clear, actionable reforms [37, 42]. Families called for, (a) Legal recognition and compensation for caregivers, particularly where they replace public services; (b) Simplification of aid processes, including transparent eligibility criteria and application navigation support; (c) Expanded public coverage for assistive technologies, mental health services, respite care, and non-clinical therapies; (d) Tailored employment protections, such as paid caregiver leave and job retention policies and (e) Improved access to inclusive education for children with complex health needs. These recommendations are consistent with calls from EU-level strategy documents, recent caregiver white papers, and academic analyses advocating for better alignment between caregiver burden and state support [5, 23, 44].

This study is the first to apply a structured, multi-coder thematic analysis to synthesize advocacy organisations' perspectives on the financial burden of caregiving for children with rare diseases across both the United States and Europe. While this approach allowed for the inclusion of diverse disease types and organisational structures, several limitations must be acknowledged [32].

First, a central limitation of this study is the absence of direct interviews with caregivers themselves. All data derive from organisational representatives, whose insights, while extensive, may either underestimate or overestimate the emotional, psychological, and financial burden experienced at the household level. Financial estimates provided during interviews represent organisational observations rather than verified income or expenditure data and may therefore differ from actual family-level costs. Similarly, emotional and psychosocial themes reflect institutional understanding of caregiver challenges rather than lived-experience accounts. These constraints must be considered when interpreting the findings. Additionally, all financial figures provided by organisations are inherently approximate and subject to organisational perception bias. These estimates do not constitute verified household expenditure or income-loss data and should be triangulated with direct caregiver-level financial research in future studies.

Second, the recruitment process relied primarily on purposive sampling [31] from publicly available directories and prior research networks. This ensured inclusion of organisations with established operations, geographic reach, and documented caregiver engagement, but may have inadvertently favored well-resourced, digitally visible organisations. Smaller or emerging associations, particularly those without a strong online presence, may have been excluded, resulting in findings that overrepresent the perspectives of organisations with greater infrastructure and capacity to articulate their communication and financial support strategies.

Third, although geographic diversity was a stated goal, recruitment yielded limited representation from low- and middle-income countries (LMICs). Structural barriers such as language, internet connectivity, and differences in organisational priorities likely constrained participation from these regions. As a result, the study is underpowered to compare challenges in LMICs with those in high-income countries. Given the potential for more severe resource constraints in LMIC contexts, future research should purposively oversample such organisations and assess whether the magnitude and nature of caregiver financial barriers differ substantially by national income setting.

Fourth, the purposive [31] sampling approach itself introduces selection bias. By prioritizing organisations perceived to have the capacity to provide rich, detailed accounts, the study excluded the possibility of random or stratified sampling that might have yielded a more representative cross-section of the rare disease advocacy landscape. Associations with no formal financial aid programs, nascent governance structures, or minimal staff support may face distinct operational and caregiver challenges that remain underexplored here.

Fifth, participation was voluntary, introducing self-selection bias. Organisations agreeing to participate may have been those already engaged in advocacy for improved caregiver support, potentially skewing findings toward more proactive or well-organized entities.

Conversely, organisations declining participation or failing to respond may include those with the most limited capacity, whose perspectives could shed light on barriers preventing even basic engagement with financial assistance or systemic advocacy.

Sixth, linguistic constraints limited interviews to English, French, or Spanish, potentially excluding organisations operating primarily in other languages. This restriction may have narrowed the cultural and geographic diversity of the sample, particularly from regions with active rare disease networks but limited bilingual capacity among leadership or staff.

Finally, inclusion criteria required at least three years of operational history to ensure that organisations had established service delivery and caregiver engagement practices. While this criterion improved comparability, it also excluded newer associations whose formative-stage challenges, such as building financial assistance programs from scratch or establishing credibility with funders, are important to understanding the full rare disease advocacy ecosystem.

These recruitment and methodological limitations mean that the findings may disproportionately reflect the experiences of established, resource-rich, and high-income–based organisations. The consistency of themes across these entities suggests that the challenges identified are widespread; however, the severity and nature of these challenges may be even greater among underrepresented groups, including smaller organisations, newer associations, and those operating in LMICs.

Future studies should adopt more inclusive and stratified recruitment strategies, extend language accessibility, and consider mixed-method designs incorporating both advocacy organisation and direct caregiver perspectives. This would enable triangulation of systems-level and individual-level experiences, providing a more complete evidence base for policy and programmatic reform. A conceptual interpretation of these findings is presented in Fig. 6, which illustrates how structural barriers, unreimbursed costs, income disruption, and longer-term economic instability intersect to form a self-reinforcing cycle of vulnerability. This framework reflects the way organisations described caregiver burden not as a sequence of isolated pressures but as an interconnected system in which administrative complexity, financial strain, and psychosocial exhaustion compound one another over time. The central convergence shown in the figure underscores that caregiver hardship emerges from the interaction of multiple structural forces, reinforcing the need for integrated policy approaches rather than piecemeal solutions.

Despite the limitations, the sample size, geographic breadth, and methodological rigor of this study provide robust, cross-context insights into the systemic financial challenges facing rare disease caregivers. The findings underscore the urgent need for equitable, well-resourced, and sustainable support structures, irrespective of an organisation's size, location, or resource base.

In conclusion, this study demonstrates that caregiving in the rare disease context is not merely an individual challenge, it is a structural issue demanding policy-level attention. The costs, both financial and emotional, are cumulative and compounding, shaped not only by the condition itself but by the presence or absence of institutional support. Without deliberate action, families will continue to rely on fragile coping mechanisms to survive in systems not designed for them. A cycle of vulnerability (Fig. 6) highlights why piecemeal interventions are insufficient and underscores the need for integrated policy responses that simultaneously address financial support, administrative access, and caregiver recognition. This research calls on policymakers, healthcare leaders, and funders to move beyond symbolic recognition toward targeted investments in caregiver support, financial equity, and inclusive infrastructure.

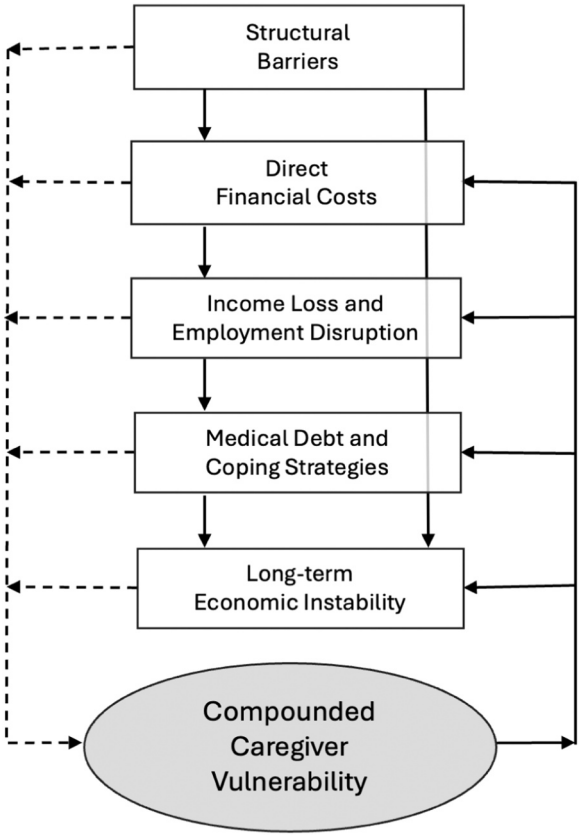

Fig. 6. Conceptual framework of financial and structural drivers of caregiver vulnerability. Self-reinforcing *cycle of vulnerability* experienced by caregivers of children living with rare diseases, as described by participating organisations. Structural barriers give rise to direct financial costs, which contribute to income loss and employment disruption, leading to medical debt and reliance on coping strategies and ultimately resulting in long-term economic instability. Solid arrows indicate predominant directional pathways through which pressures accumulate over time, while dashed arrows illustrate overlapping and compounding effects across domains. The shaded core represents *compounded caregiver vulnerability*, reflecting the cumulative financial, administrative, and psychosocial burden that emerges from the interaction of these systemic forces rather than from any single stressor in isolation.

CRediT authorship contribution statement

Levine James Andrew: Writing – review & editing, Writing – original draft, Validation, Supervision, Methodology, Investigation, Funding acquisition, Formal analysis, Data curation, Conceptualization. **Florian Delval:** Writing – review & editing, Writing – original draft, Resources, Project administration, Methodology, Investigation, Formal analysis, Data curation, Conceptualization.

Consent for publication

All authors consent to this article's publication.

Ethics approval and consent to participate

Interviews from associations convey no risk and are IRB exempt.

Ethics

The Study was approved by the York St John University ethics board. Files were stored in a secure password-protected vault using de-identified filenames.

Declaration of Generative AI and AI-assisted technologies in the writing process

The original hand-written version of the manuscript was 8400

words. To shorten it OpenAI. (2025). ChatGPT (Feb 2025 version) [Large language model] (https://chat.openai.com/_ was used on July 7, 2025, with the following prompt. "Shorten the manuscript by 20 % BUT do not generate any new language and ONLY use existing language in the shortened version." After this, the entire manuscript was verified and edited by both authors.

Declaration of Competing Interest

The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

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Appendix A. Supporting information

Supplementary data associated with this article can be found in the online version at doi:10.1016/j.rare.2026.100119.

Data availability

Data will be made available on request.

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