From Rare Disease Insights to Complex Care Innovation

Leveraging Expert Perspectives to Redesign Caregiver Support Through Data and Technology

Ekta Jain

Vandana Yadav

Stan Kachnowski

HITLAB

Contents

01 Executive Summary

The U.S. is facing a caregiving crisis, one that is particularly acute in the rare disease community. More than 30 million people in the US live with rare disease, many of whom are unable to communicate their basic needs.[1] In their place, family caregivers step in as advocates, coordinators, and providers, managing intense emotional, physical and logistical demands on a daily basis.

While 1 in 5 Americans serve as caregivers, their essential role remains largely overlooked in healthcare and research. Although, rare diseases bring these challenges into sharp focus, they reflect a broader reality: **more than 100 million Americans live with complex medical needs.**[2] To understand these challenges, HITLAB, in partnership with MiraKare, conducted interviews with rare disease organization stakeholders to assess real-world evidence (RWE) practices and explore how digital platforms can better support caregivers.

To advance rare disease care and research, stakeholders call for centralized, interoperable platforms that capture real-time caregiver data. Digital tools that ease burden, support emotional health, and integrate into daily life can transform care and research. Broader use of RWE and stronger partnerships between advocacy groups, researchers, and tech providers like MiraKare are essential for inclusive, data-driven solutions grounded in lived experience.

KEY INSIGHTS

- **Fragmented Data:** Current RWE efforts focus on clinical events, missing everyday caregiver insights crucial for research and care.
- **Overlooked Caregivers:** Caregiver stress and observations are rarely tracked or supported.
- **Digital Potential:** Stakeholders want tools like real-time medication tracking, caregiver well-being assessments, video uploads, and symptom logs.
- **Systemic Barriers:** Language issues, provider knowledge gaps, data silos, and low research engagement limit access and inclusion.
- **Willingness to Share:** 97% of patients and caregivers are open to sharing data for better treatment and diagnosis; 95% would do so for unrelated diseases.
- **Promise of MiraKare:** A caregiver-led RWE platform was welcomed as a foundation for cohort building, natural history studies, and advocacy.
- **Caregiver Time Matters:** Caregivers are time strapped and overwhelmed; data collection must be purposeful and non-burdensome to earn their engagement.

Footnotes:

[1] Wan EL, Elkaim Y, Gao W, Yoon R. Zebras Among Us: Advocating for the 30 Million Americans Living with Rare Disease. Med Sci Educ. 2023 Aug 15;33(5):1239-1242. doi: 10.1007/s40670-023-01856-2. PMID: 37886282; PMCID: PMC10597899.

[2] Chronic Disease Prevalence in the US: Sociodemographic and Geographic Variations by Zip Code Tabulation Area. https://www.cdc.gov/pcd/issues/2024/23_0267.htm

02 Background

DEFINITIONS

The World Health Organization defines rare diseases as those diseases that affect fewer than 1 in 2000 people in any WHO region (less than 65 per 100,000 individuals) [Source: Lancet Editorial 2024]. These diseases are further characterized by being present life-long and having a significant impact on the quality of life to those affected.

The definition of rare diseases can differ across settings in the US, Europe and other parts of the world in the context of their local population and healthcare systems. For instance, US defines rare diseases affecting fewer than 200,000 patients in the country (6.4 in 10,000 people).

EU defines rare diseases as a life-threatening or chronically debilitating condition affecting no more than 5 in 10,000 people.

Japan identifies rare diseases as those diseases with fewer than 50,000 prevalent cases (0.04%) in the country.

Figure 1: Prevalence based definitions of rare-diseases in different parts of the world

(Source: Roberts Christ-Tan, 2024)

TYPES OF RARE DISEASES

Rare diseases include and are not limited to rare genetic diseases, rare cancers, rare infectious diseases, rare poisonings, rare immune-related diseases, rare idiopathic diseases, and rare undetermined conditions. Following illustrations depict some categories and examples of rare diseases.

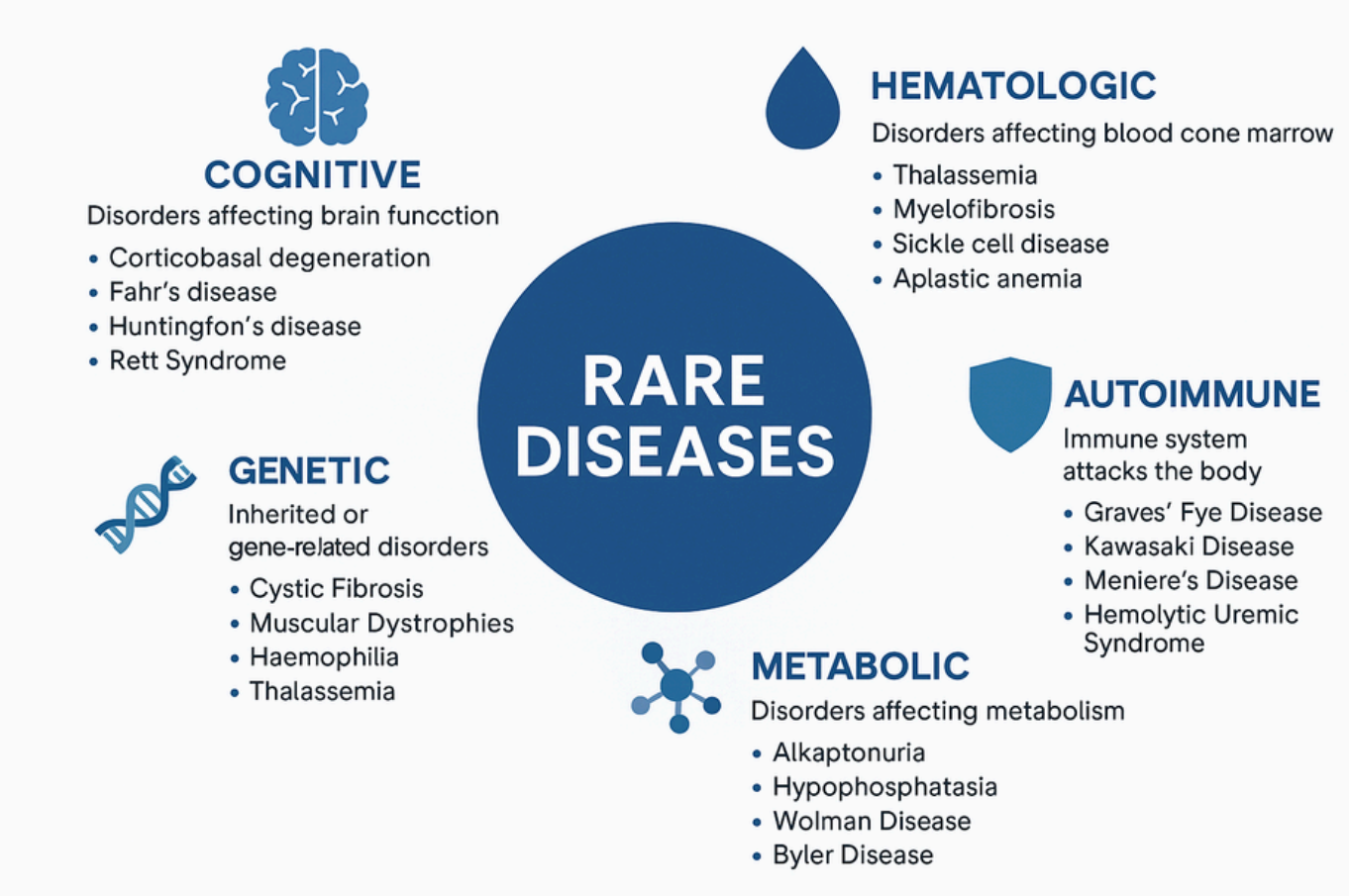


Figure 2: Different categories of rare disease and some common rare diseases for each category – Around 75% of the total rare diseases are genetic and 70% of the rare genetic disorders start in childhood.

PREVALENCE AND KEY STATISTICS

The availability of data on rare diseases is another challenge. The available epidemiology data for rare diseases is mostly from data by national registries or small cohorts on single disease or disease groups. While each rare disease affects a small number of people, their collective impact is significant.

More than 10,000 identified rare diseases[1]

~400 million people live with rare diseases[2]

Around 1 in 20 people will live with rare diseases at some point in their life[3]

80% of rare diseases are genetic[2]

50% of rare diseases affect children[2]

4-5 years average time for an accurate diagnosis[5]

30% of children diagnosed with a rare disease die before they reach the age of five[4]

FINANCIAL AND TREATMENT BURDEN OF DISEASE

Only about 5% of the known rare diseases have an FDA-approved treatment[6]

Lack of treatment is associated with a 21% increase in total costs per patient per year (PPPY)[7]

Delayed diagnosis can lead to up to $517,000 in avoidable costs per patient[8]

The burden of rare disease is ~10x higher than mass market diseases on a PPPY basis[7]

Overall burden is highest for metabolic disorders ($334,000 PPPY) and neurological disorders ($317,000 PPPY)[7]

Footnotes:

[1] Haendel M, et. al. How many rare diseases are there? Nat Rev Drug Discov. 2020 Feb;19(2):77-78. doi: 10.1038/d41573-019-00180-y. PMID: 32020066; PMCID: PMC7771654.

[2] https://www.pfizer.com/science/focus-areas/rare-disease.

[3] https://www.weforum.org/about/story/

[4] Fleming, S. (2020, February 29). One in three children with a rare disease won't live to see their fifth birthday. ThePrint.

[5] Phillips, C.,et. al. (2024). Time to diagnosis for a rare disease: managing medical uncertainty. A qualitative study. Orphanet journal of rare diseases, 19(1), 297.

[6]U.S. Department of Health and Human Services. (2023, March 21). Rare disease day at NIH 2023: Putting hope into action. National Center for Advancing Translational Sciences.

[7] https://www.outsourcedpharma.com/doc/rare-diseases-cost-burden-on-patients-0001

[8] https://everylifefoundation.org/delayed-diagnosis-study/

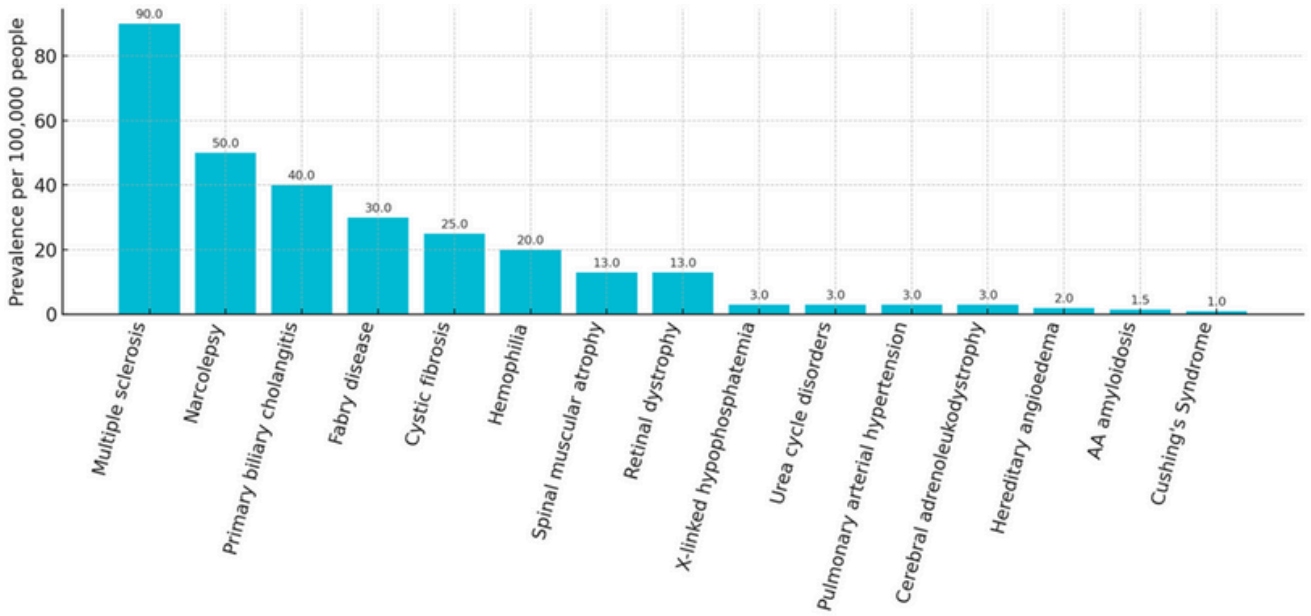


Figure 3: Prevalence of some commonly known rare diseases

(Source: World Economic Forum, 2020) – The recorded highest prevalence among rare diseases is of Multiple Sclerosis (MS). As per latest figures for MS, around 2.8 million people globally live with MS.

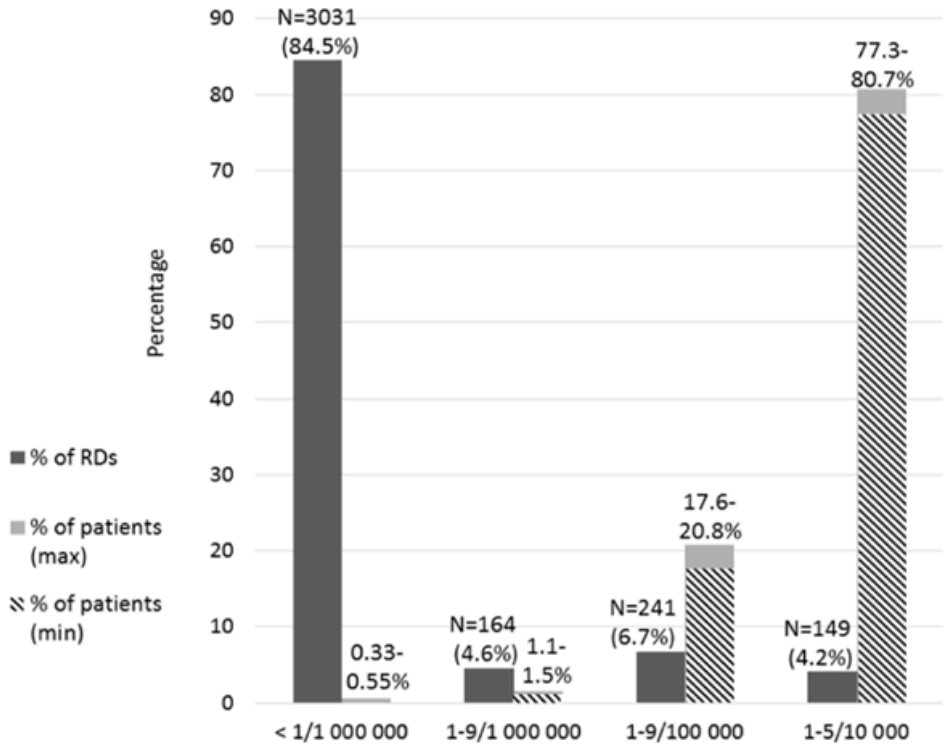


Figure 4: Distribution of rare diseases and rare disease patients according to the point prevalence class.

(Source: Nguengang Wakap et al, 2020) – This study includes 3585 rare diseases that have point prevalence data; 745 rare diseases have annotated data with a point prevalence class (20.8%),; 2496 rare diseases are described by reports of single cases (69.6%), and 344 described by reports of families (9.6%).

03 Complexities and Unmet Needs in Rare Disease Management

While rare disease journeys are significantly complex on their own, there is an added layer to the paradigm that is primarily attributed to the fact that rare diseases are characterized by delay in diagnosis and limited research. Most rare diseases are poorly understood; there is little known about their etiology. Rare diseases although genetically diverse, present with a wide range of symptoms even within the same condition. This makes diagnosis difficult and thus delayed, and further makes it less treatable.

Importantly, there exists a lack of expertise. The number of clinicians that are trained to recognize or manage these diseases are very few. This further contributes to misdiagnosis and suboptimal care.

The small prevalence means that there are less cases to study and even more it is hard to get data on such small populations. This limits the availability of robust datasets for research or clinical trials, and the subsequent limited availability of treatment drugs.

SOCIAL AND PSYCHOLOGICAL IMPACTS

There are social and psychological complexities as well. Patients often feel isolated due to the rarity of their condition, and support networks are seldom present. Moreover, there is added burden on the caregivers and they face emotional, physical, and financial challenges in providing care.

There is also societal stigma and misunderstanding associated with rare conditions that are often poorly understood by the public.

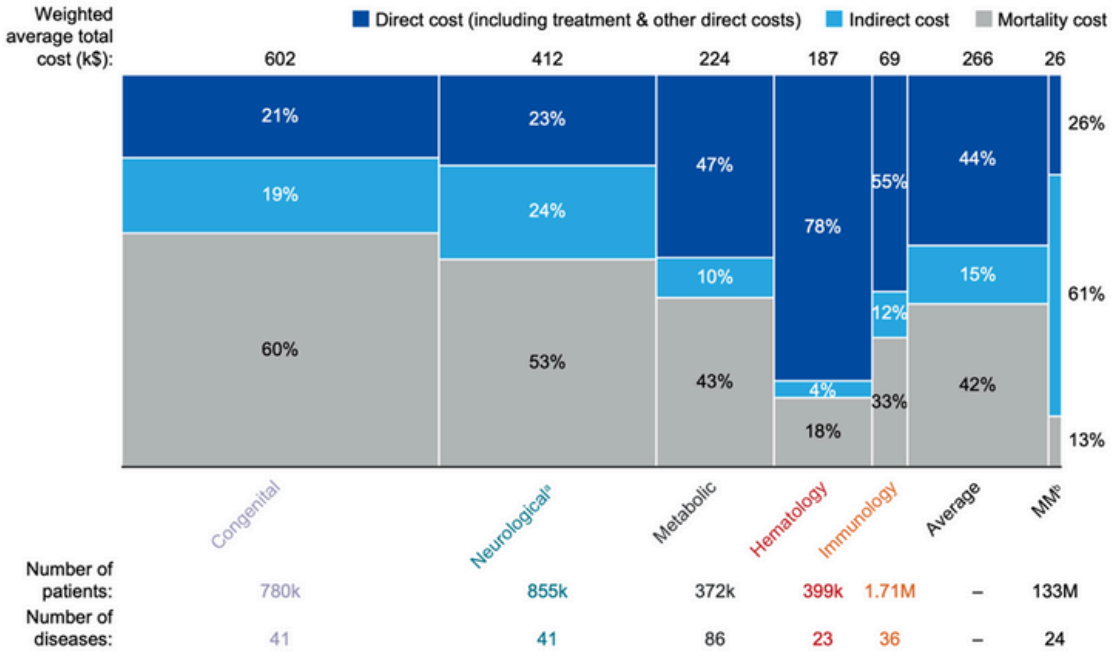


Figure 5: Extrapolated average burden of rare disease therapeutic areas on per patient per year basis across 227 rare diseases *(Source: Pedro Andreu et al, 2022)*

- Value of treatment is demonstrated by decreases in PPPY indirect costs. Access to treatment effectively shifts burden relating to indirect and mortality costs into direct costs (treatment and other direct costs).
- Providing access to rare disease treatments generally generates substantial value for society because it lowers the associated economic burden on patients and caregivers.
- Mortality cost: economic value attributable to lost productivity and finances owing to premature death.

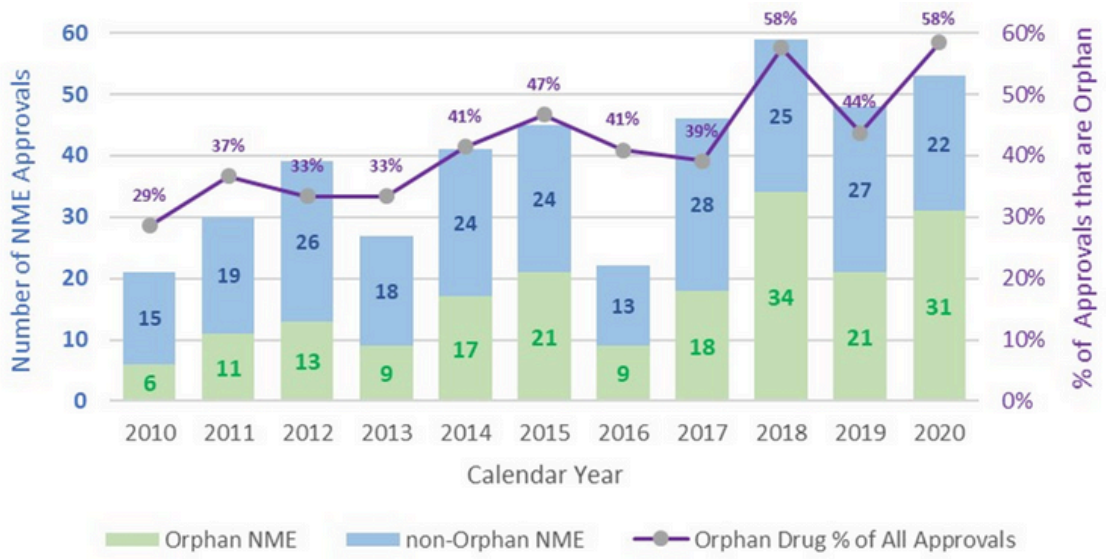


Figure 6: New Molecular Entities (NMEs) Approved by FDA between 2010 and 2020 *(Source: FDA, Center for Drug Evaluation and Research, 2021)*

CAREGIVING CRISIS IN THE US

The United States is facing a growing caregiving crisis, particularly in the context of rare diseases. In most cases, caregiving responsibilities fall on family members—currently, one in five Americans serves as a family caregiver.

- One in four individuals in the US, amounting to 28.7% have some type of disability.[1] Data from 2020 show that there are around 53 million US adults who are caregivers.[2]
- Estimates show that between 40 to 70% of caregivers have clinically significant symptoms of depression, with approximately one quarter to one half of these caregivers meeting the diagnostic criteria for major depression.[3]
- Caregivers reporting fair or poor health increases from 14% within first year to 20% after 5 years or more of providing care.[4]
- 11% of family caregivers report that caregiving has caused their physical health to deteriorate.[4]
- For rare disease patients who are unable to communicate their basic needs, caregiving becomes even more complex and demanding.
- The physical and emotional impact of dementia caregiving resulted in an estimated $9.7 billion in healthcare costs in 2014.[4]
- Despite the urgent need for over 4.6 million direct care workers,[5] this demand remains critically unmet.

NEEDS AND IMPACTS: FAMILIES AND CAREGIVERS

Caregivers—particularly family members—of individuals with rare diseases face numerous unmet needs. Limited knowledge about these conditions, combined with the emotional and social toll on both patients and their families, presents a persistent public health challenge. Common difficulties faced by these families include delayed diagnoses, limited access to specialized healthcare professionals, and challenges in obtaining appropriate medications and treatments. Even after receiving a diagnosis, clinical care and management often remain insufficient.

Footnotes:

[1] https://www.cdc.gov/disability-and-health/media/pdfs/disability-impacts-all-of-us-infographic.pdf

[2] https://www.guardianlife.com/reports/caregiving-in-america

[3] Zarit, S. (2006). Assessment of Family Caregivers: A Research Perspective. In Family Caregiver Alliance (Eds.), Caregiver Assessment: Voices and Views from the Field.Report from a National Consensus Development Conference (Vol. II) (pp. 12 – 37). San Francisco: Family Caregiver Alliance.

[4] https://www.caregiver.org/resource/caregiver-statistics-health-technology-and-caregiving-resources/

[5] https://solve.mit.edu/solutions/57874

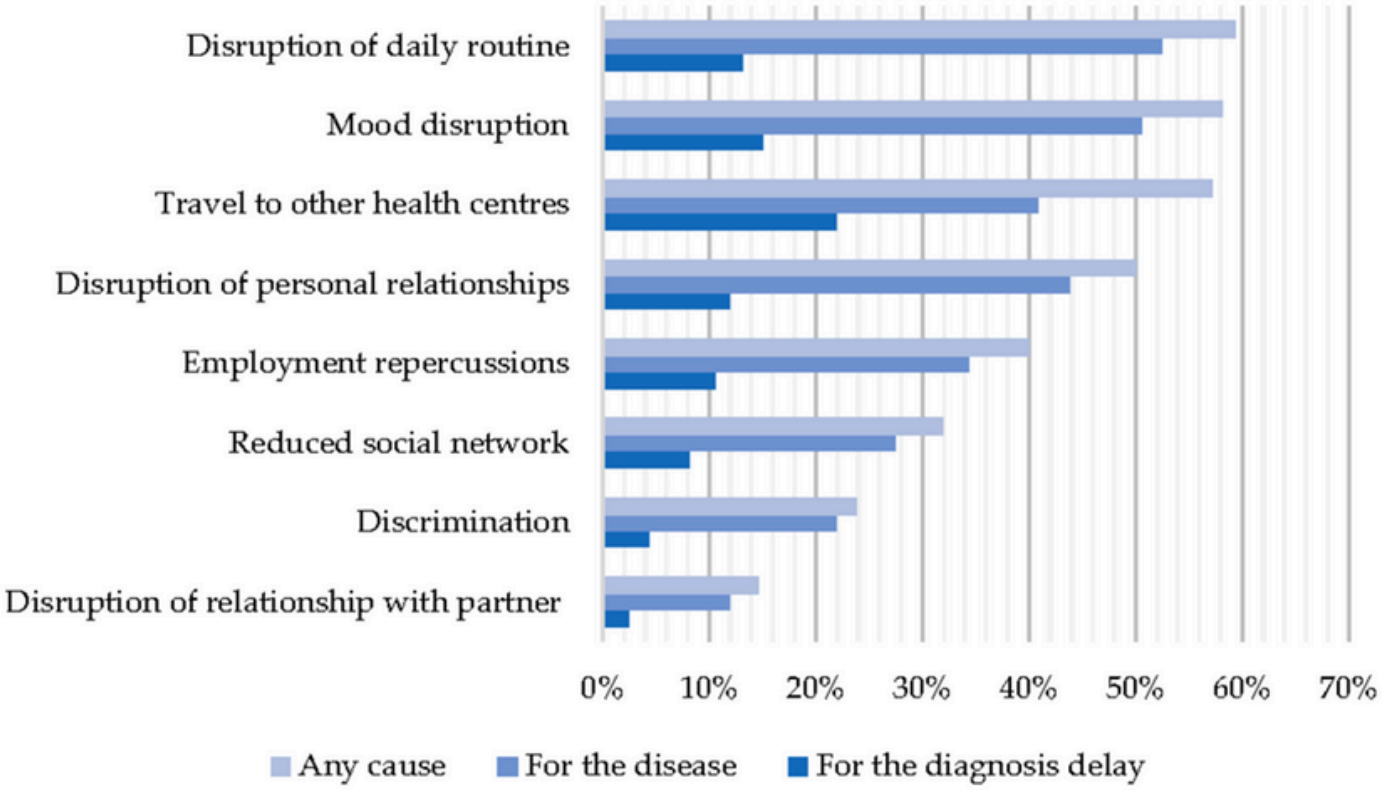


Figure 7: Survey on impact on the emotional and social sphere caused by the disease or delay in diagnosis *(Source: Gimenez-Lozano C. et al, 2022)*

When the patient is a child, families tend to experience the greatest strain. The financial burden can be significant, with medication costs making up most of the expenses. In addition, families are often responsible for managing complex care regimens, requiring extensive support and guidance.

Frustration

Stress

Importantly, these demands leave caregivers with little time for themselves. The constant need to stay engaged in every aspect of caregiving comes at the expense of their mental health, personal relationships, and career advancement. Research shows that the physical and emotional burden on caregivers of individuals with rare diseases can rival that of those caring for patients with cancer or stroke. Yet, they frequently do so without the resources or recognition afforded to other conditions.

Care is impacted

Lost time

The challenges associated with rare diseases are complex and often deeply personal. Delayed diagnosis, a lack of robust clinical evidence, the absence of standardized treatment guidelines, and the difficulty in locating appropriate specialists all contribute to fragmented and frustrating care experiences. For both patients and caregivers, the journey is often marked by profound emotional strain.

Feelings of isolation and loneliness are common, stemming not only from the rarity of the condition but also from limited public awareness and understanding. Beyond physical symptoms, patients frequently face psychosocial hardships—including stigma, exclusion, and discrimination—which further erode quality of life.

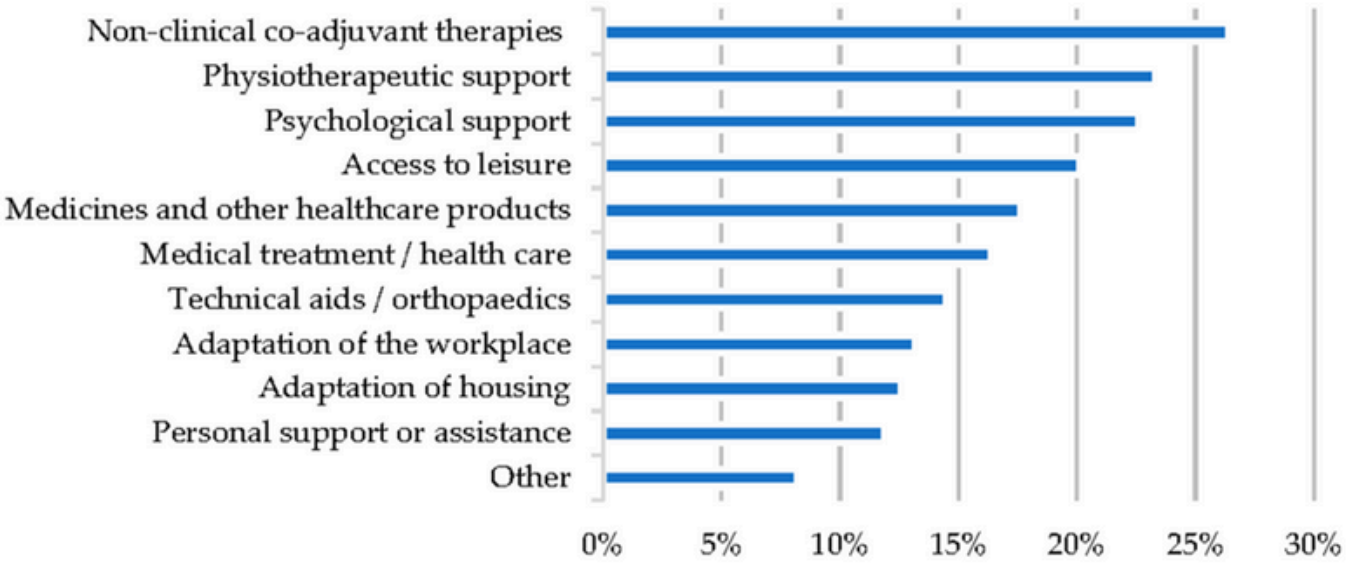


Figure 8: Survey results for unmet needs of rare disease patients and their caregivers *(Source: Gimenez-Lozano C. et al, 2022)*

Systematic review and meta-analysis of prevalence of anxiety disorders in adult rare disease patients reported higher levels as compared to the general population (Dieris-Hirche et al, 2025). Multi-center studies reported 54.4% patients of rare diseases having a mental disorder (Uhlenbusch N et al, 2021). In another recent study, 90% of 1231 individuals living with rare disease had felt anxious, stressed, depressed and around 19% of the 564 caregivers had suicidal thoughts (Spencer-Tansley et al, 2022). Importantly, both patients and caregivers experience a high and persistent emotional burden, with the majority reporting feelings of anxiety, stress, emotional exhaustion, and depression — often or all of the time. (Terry Richardson et al, 2022)

04 Current Digital Strategies And Gaps

HOW DIGITAL HEALTH TOOLS CAN HELP

Evidence shows that individuals in the rare disease community have the inclination to use digital tools for their informational, emotional, and logistical needs especially those providing care to an individual with rare disease and the patient themselves. Caregivers end up spending a lot of time researching information, such as potential diagnoses or new therapeutics, speaking with multiple providers for appointments or paperwork or with insurance companies. Caregivers often express the need to have digital tools that help them save their energies and time.

EURORDIS Rare Barometer Survey 2020

The EURORDIS Rare Barometer 2020 surveys on 2013 participants (patients and caregivers) concluded that 97% are willing to share data for research, to develop new treatments and to improve diagnosis. Around 95% were willing to share data to support research on diseases other than their own

There are a number of unmet needs that digital tools can help address. These include:

- **Coordinating care** through tele-health appointments and follow ups.
- **Centralizing medical records** to reduce silos, avoid conflicting advice, and ease the caregiver's burden of reconciling fragmented information.
- **Connecting to peers** to foster community, build trust, promote learning from others' experiences.
- **Removing the feeling of isolation** by creating avenues for emotional support, shared experiences, and ongoing connection with others in similar situations.
- **Connecting with specialists** to enable timely access to expert guidance, second opinions, and personalized care plans regardless of geographic barriers.
- **Informing with reliable insights** to empower caregivers and patients with trustworthy, up-to-date information that supports informed decision-making.

One of the most critical requirements for rare disease research and management is information gathering. Medical, patient's routine and other observational data is scarce for rare disease patients.

The current scenario for digital tools for rare diseases data collection includes primarily self-management e-resources for diseases such as haemophilia, lymphangioleiomyomatosis (LAM), congenital anemia, genetic eye disorders, cystic fibrosis, rare multisystemic vascular diseases, menstruation related diseases, complex regional pain syndrome, thoracic outlet syndrome, Addison's disease, Cushing's syndrome, acute intermittent porphyria, phenylketonuria, osteogenesis imperfecta etc.

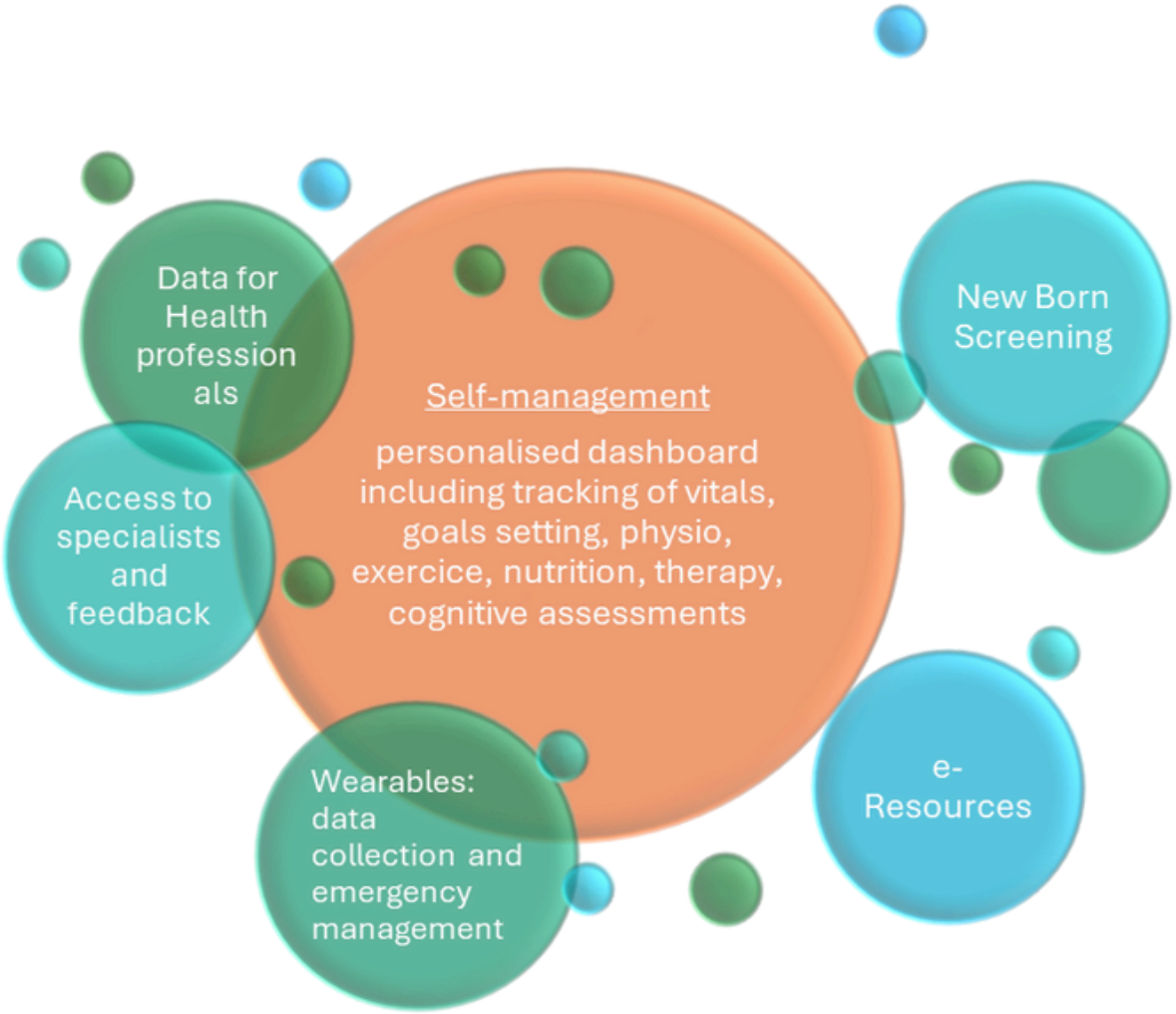


Figure 10: Needs of people with a rare disease that can be supported by health tech *(created by HITLAB)* – Self-management digital health tools make-up the majority as compared to other categories.

Some digital online tools and how they can support the needs of individuals with a rare disease are described below:

Self-management Tools

Digital self-management tools help patients track symptoms, treatments, and lifestyle factors. Apps like **Metabolic DietAppSuite** support dietary management for metabolic disorders, while **MyCBDR** tracks infusions for bleeding disorders and connects with treatment centers. Tools like **Faccio Centro** and **MyCyFAPP** guide young patients with CF in self-care, therapy management, and enzyme dosing. The **LAM** Exercise App offers tailored physiotherapy based on wearable data. **EMO.TI.ON** enables parents to share ultrasound images from home to diagnose bleeds in children with haemophilia, reducing the need for emergency visits.

Resource of High-quality Information

Trusted digital platforms provide accessible, condition-specific health information. **Mental health apps** like eHealth CF-CBT offer therapist-guided support for anxiety and depression in chronic illness. **NBS Connect** educates parents post-newborn screening, and multilingual materials aid understanding of rare conditions like hypogonadism. Sites like **Gene.Vision** offer content tailored to visually impaired users. The **NIH RDCRN** notifies patients about clinical trials. Online groups such as **MyGirlsBlood** and webcast series during COVID-19 help patients navigate their conditions with peer support and expert guidance.

Finding Specialist Centers

Tools like the **VASCERN** app help patients locate expert centers or advocacy groups for rare diseases. Innovations such as **virtual reality rehab modules** support home-based care for conditions like complex regional pain syndrome. **QR-coded emergency bracelet**s give first responders quick access to crisis protocols for conditions like adrenal insufficiency. Community tools, such as the **TOS Awareness Facebook group**, offer advice and emotional support, and can be replicated for other rare childhood diseases needing multidisciplinary care.

Connecting with Peers and Advice Networks

Digital platforms foster community and emotional support for patients and caregivers. Moderated forums, like those from the **Dutch Adrenal Society**, encourage shared learning. Programs such as **BreatheCon** and Skype groups help isolated individuals connect safely. Social media initiatives raise awareness and build community around conditions like Hirschsprung's. Youth platforms like **hiFive** and **CFOne** support teens with chronic illnesses in developing independence and resilience through peer mentorship.

DESPITE INNOVATION, GAPS REMAIN

While many promising tools exist, most are **narrow in scope**, condition-specific, or lack integration with broader care management systems. Caregivers — often the linchpin in navigating rare disease care — remain underserved by most existing digital solutions. As a result, the burden remains high, and the potential for digital tools to reduce fragmentation and enhance coordination remains largely untapped.

> *"Engage caregivers — they are the missing link in understanding the day-to-day lived experience that clinical trials miss."*
>
> ***– Interviewee Quote***

> *"Families have a treasure trove of daily information — but today, most of it's lost because there's no easy way to capture it."*
>
> ***– Interviewee Quote***

05 MiraKare

ENABLING THE FUTURE OF CARE AND RESEARCH

MiraKare is a caregiver-centered health tech platform transforming how families manage care for individuals with rare diseases and complex medical needs. These conditions often involve individuals who are non-verbal, exhibit unpredictable symptoms, and present substantial emotional and logistical demands.

By integrating data from wearables, caregiver journals, and Internet of Things (IoT) devices, MiraKare provides actionable insights that enable caregivers to identify trends, respond proactively, and make informed decisions.

Starting with the rare disease community, where care challenges are most acute, MiraKare is creating a scalable model for caregiver-informed real-world evidence that applies across complex care needs.

MiraKare recognizes that caregivers are the constant in both rare and complex care journeys. Their voice is essential to improving outcomes, easing the burden, and advancing research grounded in real life.

MIRAKARE - HITLAB PARTNERSHIP

MiraKare has partnered with HITLAB to conduct a qualitative research study with rare disease researchers, executives, and decision-makers to deepen its impact. The study aimed to uncover current practices, challenges, and unmet needs in data collection for rare diseases affecting cognition.

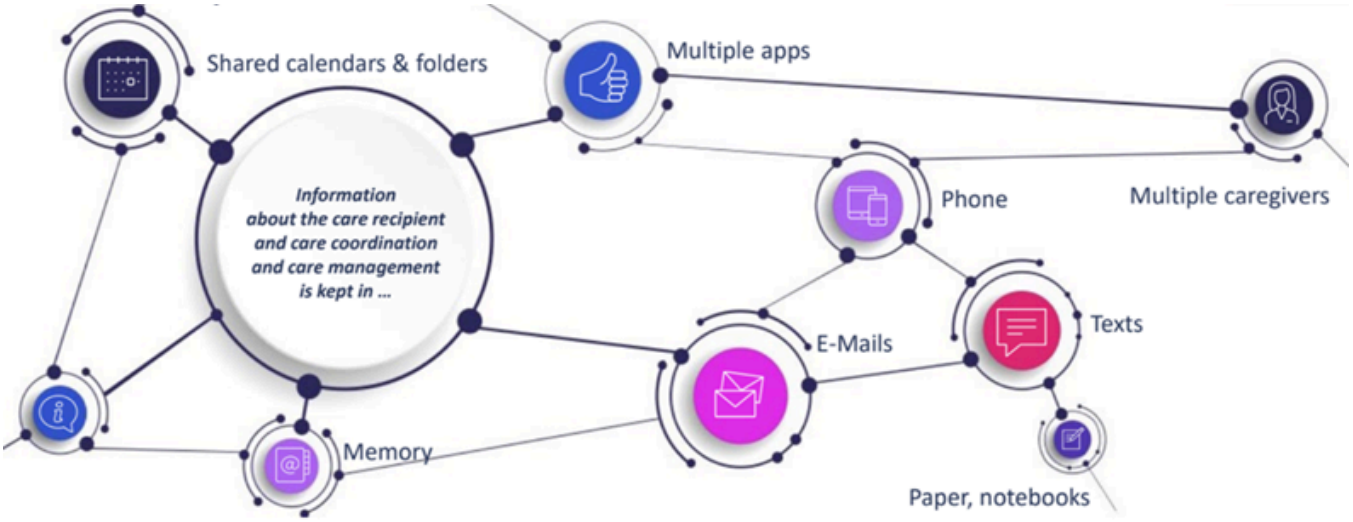


Figure 11: A pathway depicting the different digital aspects of rare disease management.

Figure 12: The information flow of MiraKare's technology.

06 HITLAB Study

AIM

The aim of this study is to gather insights about various aspects of rare disease treatment and management from organisations that pioneer rare disease awareness and research. These include data collection, analysis of available or collected data, support to caregivers and family members.

OBJECTIVES

- Gather insights from leading rare disease organizations on key aspects of treatment and care—including data collection, analysis, and caregiver support.
- Specifically, understand current practices and challenges in real-world evidence (RWE) data collection from the perspectives of patients and caregivers.
- Explore how digital technologies can empower caregivers to contribute meaningful data and enhance research and care outcomes

METHODOLOGY

HITLAB conducted a series of one-on-one, in-depth interviews with key stakeholders from rare disease organizations to better understand their data collection practices, challenges, and opportunities.

Each interview followed a mixed-structured format—primarily semi-structured with some structured components—centered around a core set of 21 main questions. Depending on the interviewee's background and experience, additional follow-up questions were posed to explore specific perspectives or processes in greater depth. Interviews lasted approximately 60 to 90 minutes, with follow-up conversations conducted as needed based on the subject matter expertise of the participants.

DATA ANALYSIS

Data from the interviews were analyzed using a combination of narrative, thematic, and quantitative methods where appropriate. Thematic analysis, in particular, surfaced key insights from the qualitative responses, offering a nuanced understanding of stakeholder needs and priorities in rare disease data collection.

Interview topics spanned a wide range of themes, including data quality, advocacy efforts, technology adoption, integration of wearable data, patient-caregiver engagement, and the perceived value of the MiraKare platform.

S.No.	Topic Summary	Theme
1	Goals of data collection	Strategy and planning
2	Importance of robust collection systems	
3	Collection process & challenges	Operations
4	Data quality assurance	Data integrity
5	Variability in data	
6	Data analysis methods	Data use
7	Patient registries and longitudinal data	Infrastructure
8	Advocacy and research priority setting	Advocacy
9	Specific data needs & barriers	Data gaps
10	Integrating multiple data types	Interoperatability
11	Use of digital/electronic tools	Technology
12	Rare disease data platforms	
13	Wearable data capture	
14	Central caregiver info tools	Caregiver support
15	Caregiver role in data collection	
16	Tech adoption: barriers/enablers	Organizational change
17	AI-generated insights	Advanced analytics
18	Org-caregiver-patient interactions	Engagement
19	MiraKare platform feedback	MiraKare-specific
20	Expected benefits of MiraKare	
21	Piloting MiraKare	

07 Results

STAKEHOLDER ENGAGEMENT OVERVIEW

HITLAB conducted in-depth interviews with eight stakeholders representing a diverse range of rare disease organizations. Participants included representatives from:

These participants brought forward insights from patient advocacy, caregiving, technology enablement, and clinical research. The interviews were carried out in partnership with MiraKare's CEO, with all data securely stored on encrypted HITLAB servers.

> *"Rare disease organizations are diverse — one size does not fit all. Platforms must adapt to different diseases, family structures, and needs."*
>
> ***- Interviewee Quote***

The discussions illuminated key challenges and opportunities in the rare disease landscape, especially regarding the collection and use of real-world evidence (RWE), caregiver needs, and digital enablement. Insights are organized into three core themes:

- **Current Practices in RWE Collection**
- **Digital Enablement and Measurement**
- **Systemic Challenges and Gaps**

CURRENT PRACTICES IN RWE COLLECTION

Fragmented and Episodic Data:
Most organizations currently rely on patient registries, but data collection is sporadic—typically based on clinical visits or intermittent surveys. Longitudinal, continuous insights into the patient journey remain limited, particularly those capturing daily lived experiences.

Existing Data Collection Practices
As shown in Figure 12 (Types of Data Collected by Different Rare Disease Organizations), the current data landscape spans a wide range—from demographics and housing needs to sleep patterns collected via wearable devices. However, data types such as real-time adherence are still emerging and underutilized.

Caregiver Perspectives Overlooked
Despite their central role in care, caregiver insights—such as stress levels, observations of patient behavior, and perceived symptom changes—are rarely captured or analyzed.

Low Engagement with Existing Tools
Traditional methods like long surveys and infrequent check-ins do not align with caregiver routines. This results in disengagement and incomplete datasets.

Growing Interest in Digital Platforms
Stakeholders emphasized the potential of digital platforms—especially those tailored to caregivers—for longitudinal tracking, natural history studies, and more meaningful RWE generation.

DIGITAL ENABLEMENT AND MEASUREMENT

Incorporating Caregiver-Centric Metrics

There is strong stakeholder support for integrating validated tools that assess caregiver burden, emotional well-being, and workload, alongside clinical data

Real-Time Medication Adherence

Time-stamped medication tracking—such as differentiating between morning and evening doses—was seen as highly valuable for both clinical coordination and research fidelity.

Multimedia System Capture

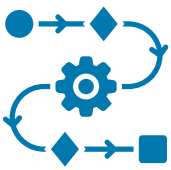
Families have expressed interest in uploading videos to visually document symptoms or behaviors, offering richer clinical context than text-based inputs alone.

Integrated, Correlated Insights

Stakeholders emphasized the importance of connecting caregiver-entered data (e.g., diet, sleep, symptom logs) with clinical records to enable more personalized and responsive care.

Accessible Health Information

Caregivers want trusted, easy-to-use resources embedded within digital platforms (e.g., via QR codes or contextual help tools).



Automation for Efficiency

Digital solutions that automate reminders, scheduling, and follow-ups can reduce caregiver burden and improve adherence to care plans.

Safe Peer Support Communities

Stakeholders noted that dedicated digital spaces for caregivers and patients to exchange experiences and emotional support are vital for long-term resilience and engagement

SYSTEMIC CHALLENGES AND GAPS

HEAVY CAREGIVER BURDEN	Emotional, physical, and financial stress affects caregivers' well-being and limits their ability to participate in research or sustain consistent care practices.
LANGUAGE AND CULTURAL BARRIERS	A lack of culturally relevant, multilingual resources reduces trust and access—particularly among marginalized populations.
HEALTHCARE PROVIDER CONSTRAINTS	Limited time, expertise, and availability among providers contribute to fragmented care and missed opportunities for early intervention.
BARRIERS TO RESEARCH PARTICIPATION	Families face a range of challenges—including logistics, lack of awareness, and motivational fatigue—that limit participation in registries and trials.
DISCONNECTED DATA SYSTEMS	Siloed platforms and incompatible infrastructure hinder coordinated care and reduce the utility of patient and caregiver data.
CAUTION AROUND AI	While stakeholders see promise in AI, they also voiced concerns about ethics, data privacy, and equitable implementation across diverse populations.

"The caregivers even passed before their loved one because of the chronic stress of caregiving"

– Interviewee Quote

STAKEHOLDER PRIORITIES: STRATEGIC FOCUS AREAS

Across all interviews, stakeholder priorities consistently aligned with four strategic pillars. These focus areas will guide the development of caregiver-centered digital solutions, driving progress toward more inclusive, accurate, and impactful rare disease care.

SUPPORT FOR PATIENTS AND FAMILIES	• Deliver targeted support programs based on need and disease stage. • Enable peer support by connecting families undergoing similar experiences. • Promote regular pediatric screenings and early detection—especially given the high prevalence of rare diseases in children.
ADVOCACY AND EQUITY	• Advocate at the federal, state, and local levels for better rare disease care and resources. • Promote health equity by addressing underrepresentation of marginalized populations. • Support collective advocacy by fostering disease coalitions.
RESEARCH AND CLINICAL TRIAL ACCESS	• Facilitate caregiver and patient enrollment in clinical trials and natural history studies. • Encourage deeper caregiver engagement in research processes. • Build strong networks between caregivers, researchers, and pharmaceutical companies. • Develop clinical care center networks tied to major research institutions.
A COMPREHENSIVE VIEW OF CARE	• Set up robust patient registries that are interoperable and inclusive. • Leverage AI to accelerate insights, lower costs, and create dynamic, adaptive care models. • Address the fragmentation of data across systems to provide caregivers and clinicians with a unified, actionable picture of the patient's health journey.

08 Recommendations

Based on stakeholder insights, HITLAB proposes the following key recommendations to advance rare disease care and research:

1. Develop Centralized, Interoperable Digital Platforms

Specific Stakeholders: Data or Study Managers.

Desired Outcomes: More scalable and secure systems capable of automated and semi-automated data collection from both patients and caregivers to improve longitudinal insights and reduce manual input burden.

2. Invest in Caregiver-Centric Technologies

Specific Stakeholders: Digital health tech developers and designers.

Desired Outcomes: Prioritization of tools and features that measure and support caregiver mental health, reduce time-intensive tasks, and facilitate peer-to-peer connection and resource access.

3. Incentivize Real-World Evidence Adoption

Specific Stakeholders: Rare disease organizations, policy makers.

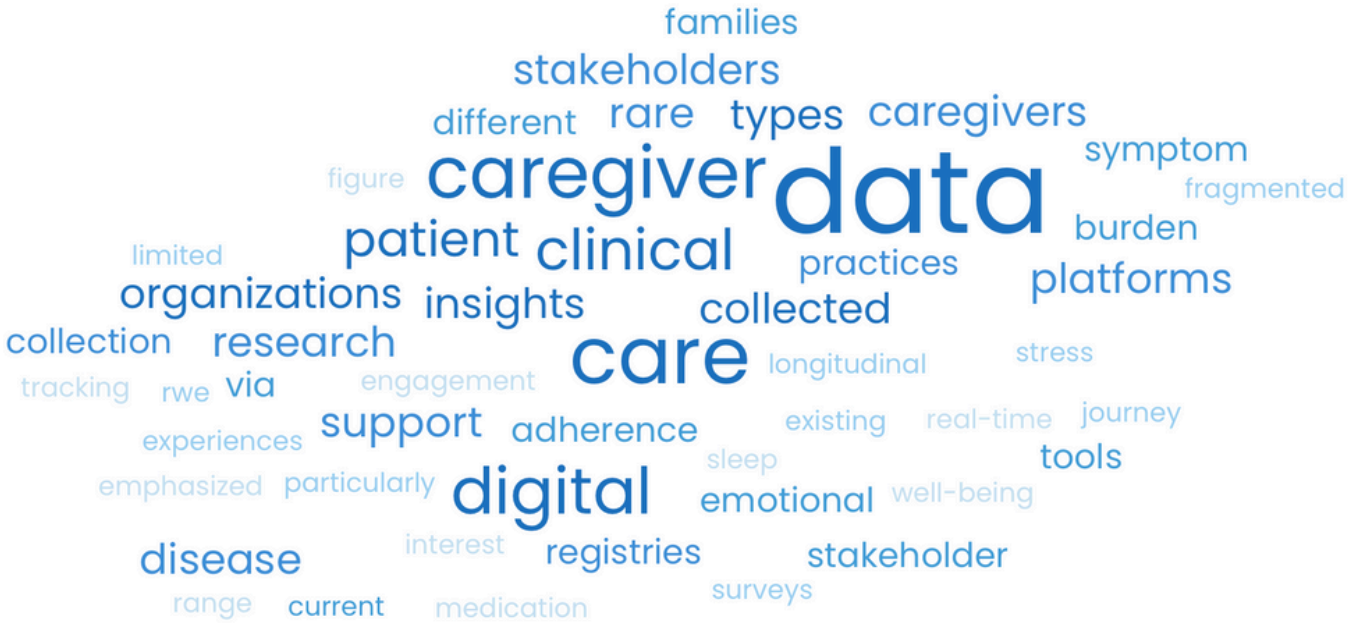
Desired Outcomes: Rare disease organizations, research institutions, and technology companies adopt and reward the use of RWE methodologies in research design, funding, and regulatory submissions.

4. Foster Cross-Sector Collaboration

Specific Stakeholders: Rare disease organizations, policy makers.

Desired Outcomes: Rare disease organizations, research institutions, and technology companies adopt and reward the use of RWE methodologies in research design, funding, and regulatory submissions.

09 Conclusion



HITLAB's interviews reveal a critical need to transform the rare disease caregiving landscape through smarter data, stronger support systems, and more inclusive technology. Caregivers and stakeholders alike voiced a shared vision: tools that capture real-time, lived experiences; platforms that offer emotional and peer support; and digital solutions that unify care delivery while upholding ethical standards.

MiraKare is turning these insights into action. By building a connected, caregiver-centered platform, MiraKare is not only addressing gaps in rare disease care but also laying the foundation for a more compassionate, data-informed ecosystem. Families caring for individuals with rare diseases, dementia, or neurodevelopmental conditions often face the same overwhelming challenges: fragmented care, delayed answers, and emotional strain. MiraKare bridges these gaps through a caregiver-first platform that transforms daily experiences into actionable insights, supporting families in real time and equipping researchers with the data needed to drive meaningful improvements in complex care.

10 References

- AARP Family Care Giving. Caregiving in the US (2020). https://www.aarp.org/content/dam/aarp/ppi/2020/05/executive-summary-caregiving-in-the-united-states.doi.10.26419-2Fppi.00103.003.pdf
- Chang A, Huang SD, Benjamin DJ, Schmidt JL, Palmer CGS, Garrison NA. (2025). Exploring the role of digital tools in rare disease management: An interview-based study. J Genet Couns. 34(1):e1908. doi: 10.1002/jgc4.1908. https://pmc.ncbi.nlm.nih.gov/articles/PMC11735183/
- Chronic Disease Prevalence in the US: Sociodemographic and Geographic Variations by Zip Code Tabulation Area. https://www.cdc.gov/pcd/issues/2024/23_0267.htm
- Gabriel A. Benavidez, Whitney E. Zahnd, Peiyin Hung and Jan M. Eberth. Chronic Disease Prevalence in the US: Sociodemographic and Geographic Variations by Zip Code Tabulation Area. February 29, 2024. Preventing Chronic Disease, Public Health Research, Practice and Policy, ORIGINAL RESEARCH, Volume 21. https://www.cdc.gov/pcd/issues/2024/23_0267.htm
- Dieris-Hirche, J., Herpertz, S., Hebestreit, H., et al. (2025). Mental disorders in people with undiagnosed diseases presenting to German centres for rare diseases – prevalence, type and relevance. Psychiatry Research, 349, 116509. https://doi.org/10.1016/j.psychres.2025.116509
- Editorial. (2024). The landscape for rare diseases in 2024. The Lancet Global Health. 12(3):e341. doi: 10.1016/S2214-109X(24)00056-1
- EURORDIS. (2020). Share and protect our health data! Rare Barometer. https://download2.eurordis.org/rbv/20-01-24%20RB%20Data%20Survey%20Fact%20Sheet_Final.pdf
- Family Caregiver Alliance. Caregiver Statistics: Health, Technology, and Caregiving Resources (2016). https://www.caregiver.org/resource/caregiver-statistics-health-technology-and-caregiving-resources/
- FDA, Center for Drug Evaluation and Research. (2021). CDER's Progress in Rare Diseases.
- Gimenez-Lozano, C., Páramo-Rodríguez, L., Cavero-Carbonell, C., Corpas-Burgos, F., López-Maside, A., Guardiola-Vilarroig, S., & Zurriaga, O. (2022). Rare Diseases: Needs and Impact for Patients and Families: A Cross-Sectional Study in the Valencian Region, Spain. Int J Environ Res Public Health, 19(16):10366. https://doi.org/10.3390/ijerph191610366
- Ibrahim, Nagwa. (2023). Navigating the Complexity of Rare Diseases: Challenges, Innovations, and Future Directions.Global Journal of Medical Therapeutics, 5. https://doi.org/10.46982/gjmt.2023.108
- Long JC, Best S, Nic Giolla Easpaig B, et al. (2022). Needs of people with rare diseases that can be supported by electronic resources: a scoping review. BMJ Open 12:e060394. https://bmjopen.bmj.com/content/12/9/e060394.citation-tools working to help others. Feb 26, 2020
- National Center on Birth Defects and Developmental Disabilities (NCBDDD). Disability Impacts All of Us Infographic. 14 April 2025. Disability and Health.
- National Institutes of Health. Rare Diseases. 01 Jan 2025. https://www.nih.gov/about-nih/nih-turning-discovery-into-health/promise-precision-medicine/rare-diseases

- Nguengang Wakap, S., Lambert, D.M., Olry, A., et al. (2020). Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database. Eur J Hum Genet 28, 165–173. https://doi.org/10.1038/s41431-019-0508-0
- Nunn, R. (2017). "It's not all in my head!" - The complex relationship between rare diseases and mental health problems. Orphanet J Rare Dis 12, 29. https://doi.org/10.1186/s13023-017-0591-7
- Pedro Andreu, Jenny Karam, Caroline Child, Giacomo Chiesi, Gina Cioffi. (2022). The burden of rare diseases: an economic evaluation. Chiesi Global Rare Diseases. https://chiesirarediseases.com/assets/pdf/chiesiglobalrarediseases.whitepaper-feb.-2022_production-proof.pdf
- Roberts, Chris-Tiann & Arezoumand, Khatereh & Kadar Shahib, Ashraf & Davie, James & Rastegar, Mojgan. (2024). Epigenetics in rare neurological diseases. Frontiers in Cell and Developmental Biology. 12. 10.3389/fcell.2024.1413248.
- Rare Diseases International Organisation. (n.d.). https://www.rarediseasesinternational.org/description-for-rd/
- Sheth, J., Nair, A., Sheth, F., et al. (2024). Burden of rare genetic disorders in India: twenty-two years' experience of a tertiary centre. Orphanet J Rare Dis 19, 295. https://doi.org/10.1186/s13023-024-03300-z
- Spencer-Tansley R, Meade N, Ali F, Simpson A, Hunter A. (2022). Mental health care for rare disease in the UK - recommendations from a quantitative survey and multi-stakeholder workshop. BMC Health Serv Res. 22(1):648. https://doi.org/10.1186/s12913-022-08060-9
- Terry Richardson, Michelle Rice, Michael Kobernick and Lesa Brackbill. (2024). Impact of mental health in persons living with rare disease: Findings from the AMCP Market Insights Program. Journal of Managed Care & Specialty Pharmacy, 30(7-b Suppl). https://doi.org/10.18553/jmcp.2024.30.7-b.s1
- Uhlenbusch N, Swaydan J, Höller A, Löwe B, Depping MK. (2021). Affective and anxiety disorders in patients with different rare chronic diseases: A systematic review and meta-analysis. Psychol Med. 51(16):2731–41.
- Wan EL, Elkaim Y, Gao W, Yoon R. (2023). Zebras among us: Advocating for the 30 million Americans living with rare disease. Med Sci Educ. 33(5):1239–42.
- World Economic Forum. Health And Healthcare Systems. A rare disease stopped this woman from standing until she was 15 - and now she's working to help others. Feb 26, 2020 https://www.weforum.org/stories/2020/02/how-rare-disease-stopped-this-woman-from-standing/