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WORLD ECONOMIC FORUM

Making Rare Diseases Count: How Better Data Can Unlock a Multitrillion-Dollar Opportunity

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Images: Getty Images

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Foreword

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Rare diseases are one of the largest areas of unmet need in global health. While individually uncommon, they collectively affect more than 300 million patients around the world. When families and caregivers are included, the circle of direct impact expands to more than 1 billion people,[1] resulting in immense human and economic costs across societies. By some estimates, the total global cost exceeds $7 trillion each year.[2]

Yet the story of rare diseases is also one of extraordinary progress. The innovations incubated in this space – from diagnostic platforms to advanced therapeutic modalities, digital health tools and data-driven care models – have not only delivered life-changing solutions for people living with rare conditions but also underpinned scientific insights and healthcare innovations that have transformed medicine itself.

The benefits extend to other stakeholders, too. Rare disease investments reduce pressure on health systems, enhance workforce productivity, improve the efficiency of insurance systems and strengthen fiscal sustainability for governments. The challenge now is to make these gains visible, measurable and replicable across societies.

Better data is the foundation that makes this possible. Data illuminates unmet needs, guides investment and enables collaboration across sectors and borders. When used responsibly, data helps turn a multitrillion-dollar challenge into a shared opportunity for innovation and impact.

This paper offers a roadmap for achieving this: five strategies to strengthen rare disease data systems worldwide and unlock the full potential of these conditions to drive progress for all.

Executive summary

Investment in rare disease data systems will benefit society and the economy, drive medical progress and improve the health and life outcomes of more than 300 million people.

Investment in rare diseases represents a multitrillion-dollar opportunity to improve lives, strengthen economies and advance science. However, around 95% of rare diseases have no treatment authorized by a major regulatory agency, leaving most patients without effective treatment and so placing a heavy burden on families and caregivers.

This paper explores the case for greater societal investment in rare diseases and the central role of data in assessing need, measuring impact and enabling progress. These investments generate impact across three key dimensions:

- **Human health:** Improving diagnosis, treatment and understanding of rare diseases can directly enhance quality of life and outcomes for more than 300 million people worldwide.
- **Socioeconomic resilience:** Reducing the impact of rare diseases can lower healthcare costs and strengthens economic productivity, benefiting the wider circle of more than 1 billion people whose lives are touched by these conditions, including families and caregivers.
- **Scientific and medical progress:** Insights and technologies pioneered in rare diseases often spill over into more common conditions, driving scientific discovery and innovation across healthcare.

The paper also provides a practical roadmap to strengthen rare disease data systems worldwide, with five key recommendations:

- **Define and track a minimum dataset across countries** to standardize data collection processes and expand the global knowledge base on rare diseases
- **Strengthen patient engagement in data collection** to maximize the quality, breadth and utility of patient registries and other key datasets
- **Improve newborn screening and diagnostic capacity** to enable earlier detection, better prevalence estimates and more efficient care
- **Enable trusted data sharing across health systems** by aligning standards, governance and safeguards to make data more usable while protecting privacy
- **Use AI and digital tools to address evidence gaps**, turning scattered information into structured, actionable insights

These strategies are broadly applicable across healthcare systems, although implementation will vary based on socioeconomic and cultural factors. In all contexts, success depends on tailoring the roadmap to local realities while ensuring alignment with international standards.

Public, private and philanthropic actors all have roles to play, independently or in partnership. Those who benefit – patients, families, health systems, employers, payers, governments and industry – are the same stakeholders who can invest, and each realizes gains in health, productivity and innovation.

Landmark policy actions, including the recent resolutions on rare diseases from the United Nations (2021)[3] and World Health Assembly (2025),[4] have placed rare diseases firmly on the global health agenda. There is an opportunity for countries and health systems to act decisively, using this roadmap to convert global recognition into national and local progress.

Rare diseases matter to all of us. While individually rare, they are collectively common, and their impacts are felt across every part of society. The call to action is clear: invest in rare diseases, with data as the foundation. This will improve lives, strengthen economies and accelerate innovation across healthcare.

1 Why investing in rare diseases matters

Data-driven investments in rare diseases improve lives, cut costs and accelerate health innovation.

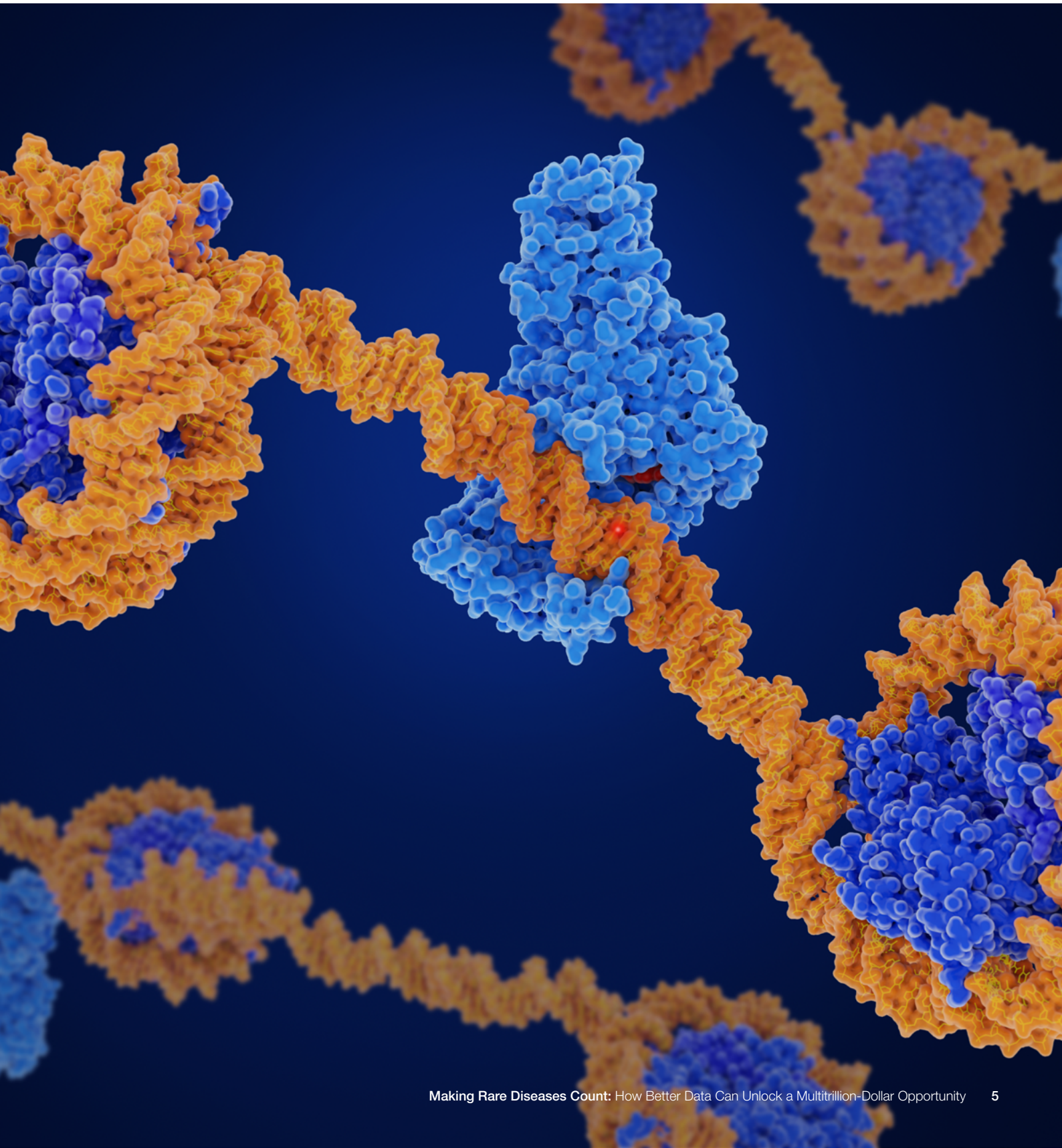


FIGURE 1 | **The case for investing in rare diseases**

	The challenge	Rare disease opportunity	Broader systemic impact
Human health	**7,000+ rare diseases affect 300 million+ globally,** with a disproportionate impact on children	**Reduced patient suffering** through earlier diagnosis and coordinated, data-driven care	**Greater socioeconomic participation** for patients, caregivers and families, strengthening overall economic productivity
Economic impact	**>$7T in annual global impact,** largely from hidden, indirect and avoidable costs	**Lower avoidable costs** for rare disease patients, healthcare systems, employers, payers and governments	**Resources liberated and redirected** towards prevention, innovation and other socioeconomic priorities
Science and technology	**~95% of rare diseases lack treatments** approved by a major regulatory agency	**New treatments and care models** enabled by data-driven research and development	**Innovation spillovers** that accelerate progress across healthcare, industry and society

Source: Rare Diseases International; IQVIA and Chiesi Global Rare Diseases

Rare diseases are under-prioritized in many health systems, even though a growing evidence base indicates that well-targeted investments deliver significant returns. The benefits extend far beyond the patients, families and communities most directly affected; they also accrue to healthcare systems, employers, payers, governments, industry and investors (see Figure 2).

Rare disease investments can take many different forms: funding research and therapy development; scaling screening and diagnostics; investing in rare disease medicines; deploying digital tools and AI; and strengthening system capacity. Across all of these areas, investment in data serves as both an enabler and an amplifier.

While the opportunity for immediate financial return varies across contexts, targeted and low-cost investments in data can yield substantial and compounding benefits, including in low- and middle-income countries (LMICs) with constrained health budgets. Even simple actions such as establishing a patient registry or improving diagnostic reporting can enhance visibility, coordination and efficiency across a given health system.

The sections that follow first describe the global challenge of rare diseases, then explore how sustained investment in rare diseases can reshape economies and healthcare systems.

FIGURE 2 | **How rare disease investments benefit everyone**

Patients and caregivers	Healthcare systems	Employers
Better care	Significant cost savings	Lower insurance premiums
Lower out-of-pocket costs	Accelerated research translation	Stronger employee retention
Greater workforce participation	More innovative care	Enhanced corporate reputation
Improved quality of life		

Payers	Governments	Investors and industry
Lower long-term costs	Higher economic productivity	New market opportunities
Stronger evidence for coverage and reimbursement	Better population health	De-risked R&D
Improved member outcomes	Less strain on social services	Faster time to market
	Greater equity	Improved return on investment

Source: World Economic Forum, Rare Disease Community analysis

1.1 The human impact of rare diseases

More than 7,000 rare diseases are recognized worldwide,[5] most of them genetic in origin. These conditions vary widely in presentation and progression, but many are severe and complex, placing extraordinary stress on families, communities and economies. Many require round-the-clock care, generate high out-of-pocket expenses and limit participation in education and employment.

Children are disproportionately affected. Around 70% of rare diseases manifest in childhood, and many are life-limiting or fatal. As deaths from preventable illnesses decline, rare diseases are emerging as a leading cause of childhood mortality and disability, including in LMICs.

For the purposes of this paper, a rare disease is defined as a condition affecting fewer than one in 2,000 people in any World Health Organization (WHO)-defined region (see Figure 3). This aligns with the *Operational Description of Rare Diseases* developed by Rare Diseases International and global partners,[6] although national thresholds vary worldwide.

FIGURE 3 **What is a rare disease?**

The *Operational Description of Rare Diseases* defines a rare condition as one affecting ≤1 in 2,000 people in any WHO-defined region, although national thresholds remain in place and differ globally

USA	Europe	Japan	China	India
< 200,000 people (≈ 1 in 1,650)	< 1 in 2,000 people	< 50,000 people (≈ 1 in 2,500)	Based on national rare disease list	Based on policy-linked rare disease lists

Source: Rare Diseases International; national policy documents

Barriers to diagnosis and care

Rare diseases are often under-recognized, even among skilled healthcare professionals. A 2021 cross-national survey found that only 19% of medical practitioners felt confident diagnosing rare diseases.[7] This helps explain why the average time to diagnosis in Europe is four to five years,[8] and often longer elsewhere.

Even when a condition is identified, providers often lack the expertise or resources to guide the next steps in management. Without coordinated systems of care, diagnosis can remain a label rather than the beginning of effective treatment and support. Many patients thus cycle through multiple tests and providers, driving up costs for health systems and delaying timely intervention.

Approved therapies also remain scarce: around 95% of rare diseases have no treatment authorized by a major regulatory agency. Drug development continues to be focused on a relatively small number of higher-prevalence rare diseases, while thousands of others still lack effective options.

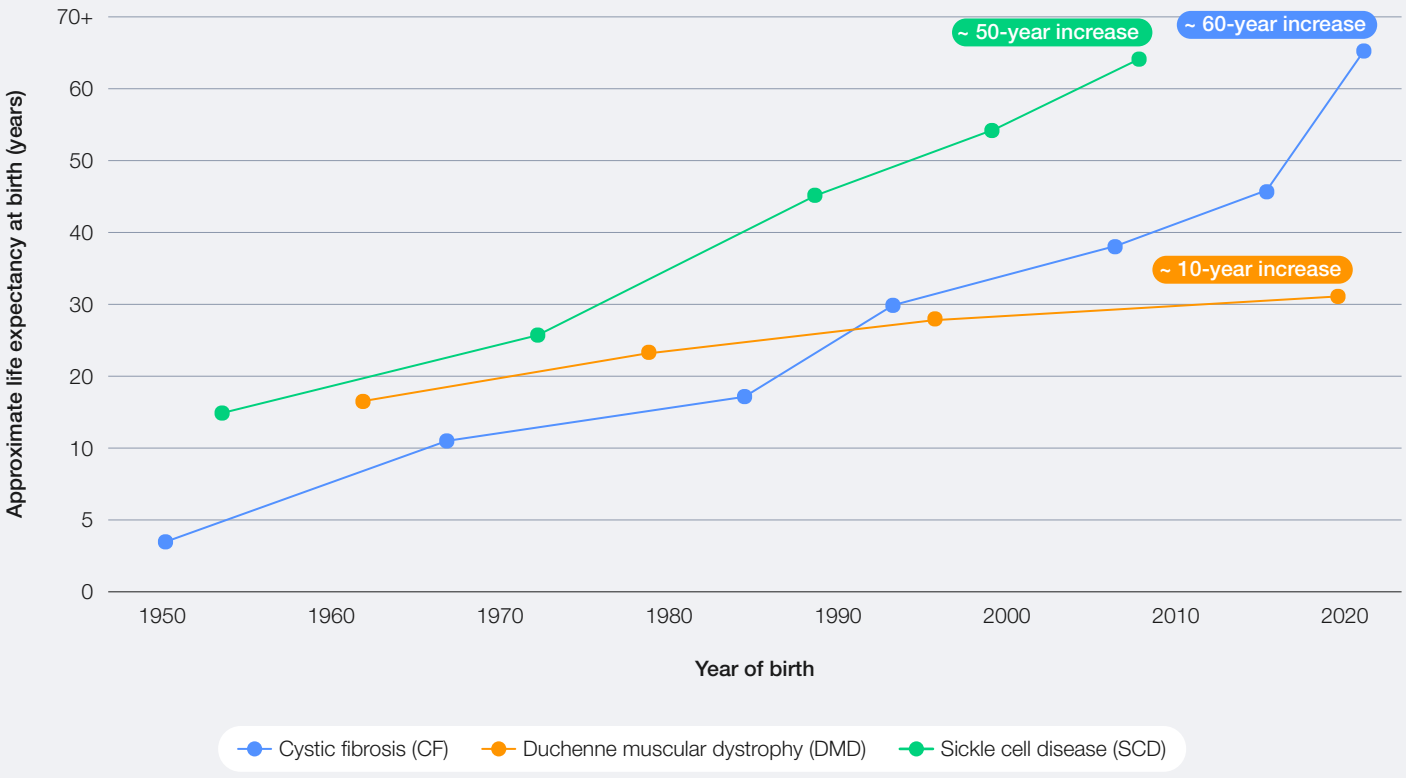
Even when treatments exist, many patients are diagnosed too late to benefit from disease-modifying care, remain undiagnosed or face persistent barriers to coverage and access. These barriers often stem from pricing and reimbursement frameworks that are not well suited to assessing rare diseases, where evidence is often limited. More pragmatic approaches to managing uncertainty could help ensure that effective therapies reach patients sooner.

Reducing suffering and empowering families

Closing these gaps means patients, families and caregivers can live healthier, more productive lives. They can participate more fully in education, work and community life. This not only reduces suffering but also restores productivity, strengthens family stability and eases pressure on health and social systems.

Progress in rare diseases such as cystic fibrosis, sickle cell disease and Duchenne muscular dystrophy demonstrates what is possible when investment aligns with science and coordinated care (see Figure 4). These improvements reflect decades of collaboration and data-driven care, showing how targeted innovation can transform lives across generations.

FIGURE 4 | **Advances in life expectancy for patients with treatable rare diseases**

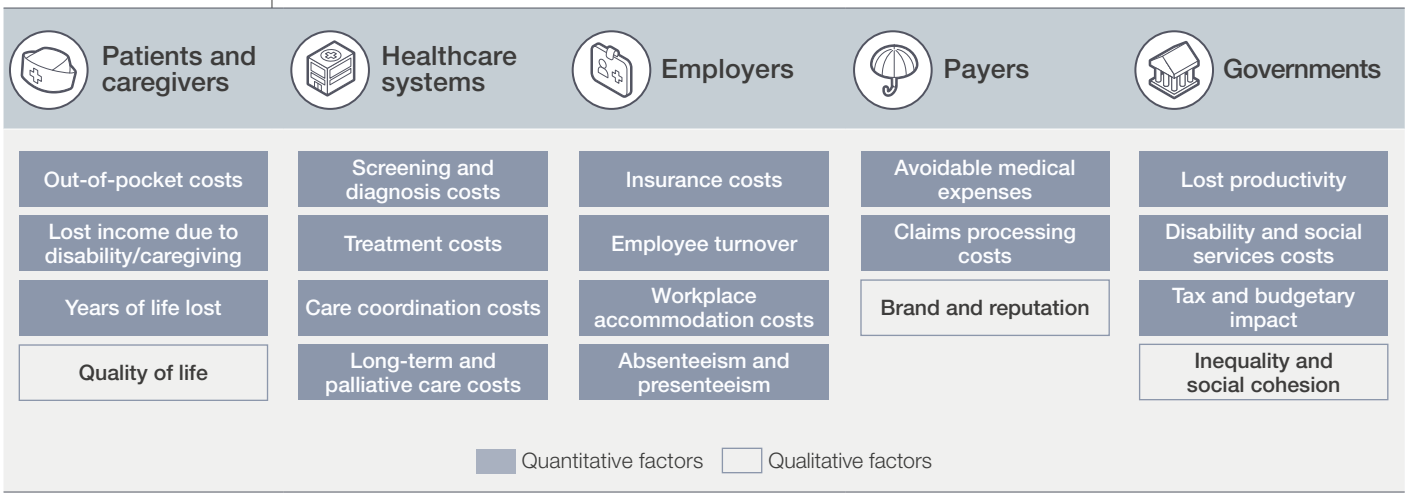


Note: Data represents approximate averages from multiple published studies and sources
Source: Deloitte analysis

1.2 Reducing costs and unlocking economic gains

Rare diseases generate financial strain across every part of society: families face high out-of-pocket expenses and lost income; health systems absorb the costs of diagnosis, treatment and long-term care; employers and payers contend with productivity losses and high claims; and governments bear the weight of social services and reduced tax revenues (see Figure 5).

FIGURE 5 **Direct and indirect impact of rare diseases across key stakeholder groups**



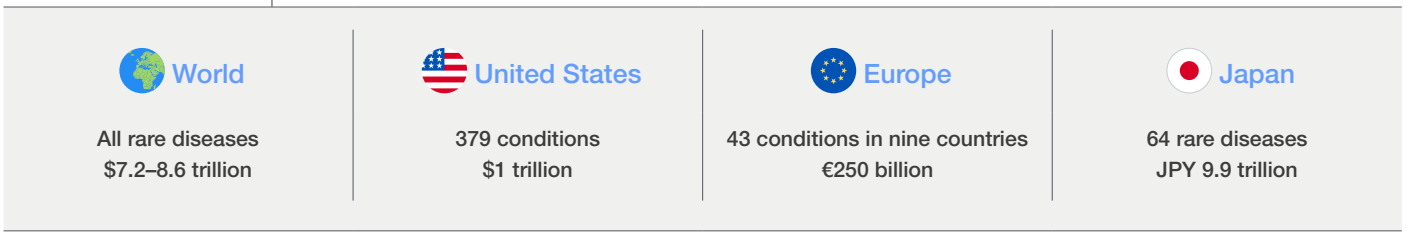
Source: Deloitte

Although comprehensive estimates remain limited, available data places the global collective cost of rare diseases in the trillions of dollars. A 2022 analysis estimated an annual cost of $7.2–8.6 trillion across all known rare diseases when accounting for direct medical expenses, mortality and indirect losses such as reduced workforce participation and disability accommodations.[9]

National and regional studies corroborate these figures (see Figure 6). In the United States, a 2021 study found that just 379 rare conditions generated nearly $1 trillion in annual costs.[10] In Europe, a 2024 study estimated that 43 rare conditions imposed a combined impact of approximately €250 billion ($290 billion) annually across nine countries.[11]

Beyond North America and Europe, data is scarce. In Japan, a 2025 study found that 64 rare diseases imposed a total cost of JPY 9.9 trillion annually (approximately $62 billion).[12] For most LMICs, no comparable estimates exist.

FIGURE 6 **Estimated annual economic impact of rare diseases**



Source: IQVIA and Chiesi Global Rare Diseases; EveryLife Foundation for Rare Diseases; Charles River Associates (*The economic cost of living with a rare disease across Europe*; *The economic cost of living with a rare disease in Japan*)

These figures underscore both the cost of inaction and the scale of potential returns. Closing data gaps will not only quantify the burden more accurately but also enable key stakeholders to target resources more effectively and track the value of intervention over time.

Why costs are overlooked

Despite the staggering numbers, most stakeholders in healthcare cannot see the full economic impact of rare diseases in ways that are clear or actionable. The root cause is data fragmentation.

Rare disease patients often move among multiple specialists, facilities, jurisdictions and insurance systems. Indirect costs are rarely tracked. Health systems' measurement of activity is compartmentalized, making it difficult to capture the cumulative impact. Insurers face similar challenges: expenses are dispersed across providers and coded under multiple diagnoses, obscuring the total cost.

This fragmentation has profound consequences: health systems struggle to identify high-cost patients for early intervention; researchers cannot link genomic and clinical data with real-world outcomes; regulators lack the evidence to approve therapies efficiently; payers cannot assess the return on investment of early diagnosis and coordinated care; and industry faces higher development costs, longer timelines and greater uncertainty in demonstrating value.

The result is that visible spending tells only a fraction of the story. A 2025 study found that rare disease medicines account for roughly 6.6% of total pharmaceutical budgets and just 1.2% of overall public healthcare expenditure across Europe.[13] Such numbers are relatively easy to track, but they risk being misinterpreted without the broader context of total economic impact.

1.3 Advancing scientific discovery and healthcare innovation

Rare disease research and innovation have been powerful catalysts of global health progress. Breakthroughs pioneered in rare disease settings extend far beyond their immediate community, reshaping therapeutics, regulation and the culture of medical innovation more widely. They show how investments in rare diseases generate benefits for all of society – provided they are informed and amplified by data.

Driving scientific and therapeutic progress

With most rare diseases having a clear genetic origin, they have been central to advancing genetic medicine. Early applications of sequencing technologies focused on rare disorders well before they became routine in oncology and infectious diseases. Discoveries in rare cardiac conditions paved the way for cholesterol-lowering therapies such as PCSK9 inhibitors, which have expanded treatment options for patients at highest risk.[14]

Rare diseases have also helped incubate new therapeutic platforms: messenger RNA (mRNA) technologies, first explored in rare diseases and select cancers, provided the foundation for the rapid development of COVID-19 vaccines; gene therapies, RNA interference (RNAi), antisense oligonucleotides (ASOs) and base editing all saw their earliest clinical successes in rare indications. Early milestones, including the first personalized base-editing treatment for CPS1 deficiency in 2025, further underscore how frontier technologies are often pioneered in rare disease settings.[15]

Rare diseases are also redefining how advanced therapies are manufactured and delivered. Point-of-care models, such as those developed by US non-profit Caring Cross, bring cell and gene therapies directly into hospital settings rather than relying on centralized facilities, making production viable in both high- and low-resource environments.

Rare disease research has reshaped scientific thinking. Fields such as pharmacogenomics and precision oncology draw directly from rare disease research. Some rare diseases even serve as models of resilience or physiological extremes – mimicking extreme physiological states such as altered oxygen use or heightened immune sensitivity – offering insights relevant to space exploration, environmental adaptation and pandemic preparedness.[16]

Advancing regulation, policy and governance

Rare diseases have helped reshape how health systems enable innovation, spurring reforms in regulation, policy and governance. Their complexity has forced institutions to rethink how new therapies are brought into use, setting precedents now applied far beyond the rare disease community.

– **Regulatory reform:** Rare diseases have driven major advances in how medicines are evaluated and approved. They have prompted flexible clinical trial designs and greater use of alternative data sources.[17] They have also led to new regulatory pathways such as accelerated approvals that bring promising therapies to patients more quickly.[18]

– **Policy innovation:** The need to coordinate action across research, care and reimbursement has inspired new policy tools to link these efforts. Governments have introduced rare disease strategies and national frameworks that align incentives and promote early diagnosis and access.

– **Governance and accountability:** Sustaining momentum requires strong governance to ensure accountability. National frameworks such as the UK Rare Diseases Framework and Action Plan illustrate how structured oversight, transparent reporting and cross-agency coordination can translate policy ambition into lasting action.

Patient advocacy groups have been central to these shifts by helping communities prepare for participation in clinical trials, contributing to protocol design, pressing for new regulatory and policy frameworks and building capacity to engage in access and reimbursement decisions. Together, these efforts are shaping how new therapies are developed and delivered across healthcare.

TABLE 1 **The spillover effects of rare disease research and innovation**

Technologies and therapeutic platforms		
Rare disease focus area	**Direct impact on rare disease**	**Broader impact on healthcare and medicine**
Sequencing technologies	Sequencing platforms enabled diagnosis, natural history studies and drug development for rare genetic diseases and are now embedded in routine rare disease screening and diagnostic pathways.	Next-generation sequencing platforms developed through rare disease research are now widely applied across oncology, infectious diseases and other areas of medicine.
Genetic medicine	Therapeutic platforms – including gene therapy, ASOs, mRNA, RNAi and base editing – achieved early clinical successes in rare diseases, improving outcomes in previously untreatable conditions and de-risking novel modalities.	Platform technologies pioneered in rare diseases now underpin treatments for more common conditions, including mRNA vaccines and expanding gene- and RNA-based therapies.
Cardiometabolic medicine	Discovery of rare inherited cardiac conditions informed targeted interventions and preventive strategies for affected patients and families.	Insights from rare genetic heart conditions advanced understanding of lipid metabolism, contributing to the development of widely used cholesterol-lowering therapies.
Precision oncology	Study of rare inherited cancer syndromes and molecularly defined tumours enabled genotype-driven diagnosis, stratification and treatment selection.	Genomic stratification approaches developed in rare and molecularly defined cancers are now central to precision oncology and biomarker-guided cancer care.
Approaches and methodologies		
Rare disease focus area	**Direct impact on rare disease**	**Broader impact on healthcare and medicine**
Registries and data standards	The need to aggregate small, geographically dispersed patient populations drove early attention to consistent data definitions, classification and cross-system comparability.	Approaches to data collection and standardization developed in rare disease contexts informed wider efforts to connect health data across institutions and borders.
Regulatory flexibility	Rare disease development helped establish early precedents for small trials, adaptive designs, proxy measures of clinical outcomes and conditional approvals in areas of high unmet need.	Regulatory approaches first normalized in rare diseases informed broader accelerated approval pathways and evidence frameworks used in oncology, infectious diseases and other serious conditions.
Patient engagement	Rare disease communities pioneered structured patient engagement, patient-reported outcomes and the systematic inclusion of lived experience in research and regulatory decision-making.	Patient-focused regulatory initiatives and wider acceptance of patient-reported evidence across medicine were shaped in part by practices established in rare disease contexts.
Screening and early detection	Rare disease contexts established early approaches to genetic and biomarker-based screening, including ethical, clinical and data frameworks for identifying risk before symptom onset.	Screening methodologies and governance approaches developed in rare diseases informed wider applications in inherited cancer screening, carrier screening and other population health contexts.

Source: World Economic Forum, Rare Disease Community analysis

1.4 The virtuous cycle: How rare disease investments compound

When informed by high-quality data, investments in rare diseases thus create reinforcing benefits that extend throughout society. This virtuous cycle operates across four interconnected domains, with progress in one area fuelling gains in the next.

1. **Clinical impact → system efficiency:** Better data, informed by robust screening and diagnostic systems, enables earlier identification of rare diseases and feeds back into improved research and care. These improvements translate directly into lower costs through fewer emergency visits, shorter hospital stays and more efficient resource use.

2. **System efficiency → research capital:** When health systems, payers and governments can measure and manage rare disease costs, they unlock resources for reinvestment. At the same time, better data reduces the time and expense of R&D, making rare disease innovation more attractive to industry and investors.

3. **Research capital → rare disease innovation:** Increased investment accelerates the development of new healthcare technologies, from diagnostics and therapeutics to digital health tools and data-driven care models. Rare diseases often serve as the proving ground for these innovations.

4. **Rare disease innovation → spillover effects:** The scientific insights and healthcare technologies established in rare disease research consistently spill over into more common conditions. Advances first tested or refined in rare conditions, such as gene and RNA-based therapies or genetic sequencing platforms, are now driving progress across many common diseases.

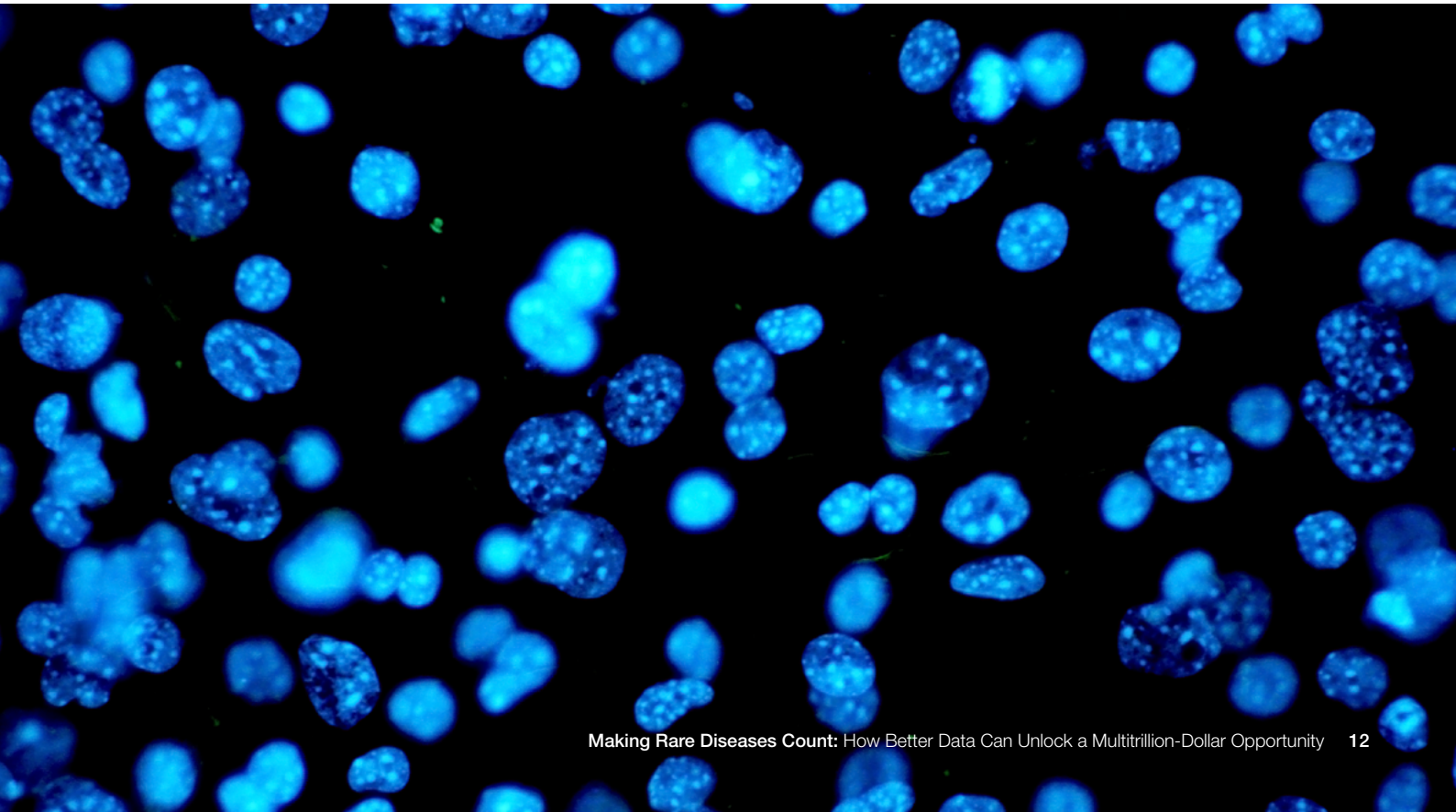
Each turn of this cycle strengthens health systems, expands economic capacity and generates resources for the next wave of rare disease investment.

Global equity and the data imperative

A key caveat remains: most rare disease breakthroughs have occurred in high-income countries. Global equity depends on targeted investments in LMIC health systems, where stronger data can help identify gaps, track progress and guide investment in ways that scale innovation fairly.

The International Rare Disease Research Consortium (IRDiRC), established in 2011, seeks to advance this kind of global coordination. By bringing together public funding agencies, industry and umbrella patient organizations, it set ambitious targets: achieving a diagnosis for undiagnosed cases within one year, catalysing 1,000 new therapies and developing methodologies to evaluate their impact.[19]

Meeting targets like these will require stronger data systems. Section 2 of this paper examines how better data can unlock even greater impact.



2

Unlocking impact with better data: Five strategies for success

Stronger data systems underpin impact in rare diseases, providing the visibility, evidence and accountability needed to drive global progress.

Improving data is one of the most powerful and practical ways to accelerate progress in treating rare diseases. This priority is gaining global traction, but major gaps remain. The challenge is not just to collect more data but to ensure it is meaningful and actionable.[20]

This section sets out a roadmap built on five strategies to address critical barriers (see Figure 7). Together they provide a theory of change: better data can generate evidence of need, guide investment and drive innovation.

FIGURE 7 **Five strategies for smarter data collection**

Focus Area	Challenge	Solution
Data collection	Many countries lack basic data	Define and track a minimum dataset across countries
Patient engagement	Patient experiences are underweighted	Strengthen patient engagement in data collection
Diagnostics	Many patients remain undiagnosed	Improve newborn screening and diagnostic capacity
Data sharing	Data silos and lack of interoperability	Enable trusted data sharing across health systems
Analytics	Data is hard to collect and analyse	Use AI and digital tools to address evidence gaps

Source: World Economic Forum, Rare Disease Community analysis

2.1 Define and track a 'minimum dataset' across countries

Countries differ widely in how they manage rare disease data. This lack of alignment creates blind spots around one of the largest cost areas in healthcare systems. An internationally harmonized minimum dataset would provide a common language for understanding rare disease impact and opportunity.

A key enabler is the adoption of international classification systems, notably ORPHAcodes and the ICD-11 rare disease extension codes. These standards make it possible to identify individual conditions in health records with greater precision. Many countries – starting with France and Germany, extending across Europe and now reaching Australia and Canada – have already embedded ORPHAcodes into public health systems.[21]

The goal is not to build a perfect system at once but to establish a practical starting point: a small set of metrics that it is feasible to collect across health systems (see Table 2).

TABLE 2 **Key metrics for a minimum and expanded dataset**

Minimum dataset (all countries)		DATA TYPE
Incidence and burden of disease	**Access to treatments and services**	**Scale and impact of rare disease investments**
Estimated overall prevalence (diagnosed and undiagnosed)	Name and number of approved rare disease therapies	Number of rare disease clinical trials (all phases)
Type of rare disease coding system in place, e.g. ORPHAcodes and/or ICD-11 extension codes.	Number of conditions covered on newborn screening systems	Rare diseases as named healthcare priority at the national level [yes/no]
	Presence of rare disease COE [yes/no]	Rare diseases as named research priority at the national level [yes/no]
	Presence of national rare disease advocacy organization [yes/no]	
	Separate reimbursement/HTA pathway for rare diseases [yes/no]	
Expanded dataset (higher-resource and more data-driven settings)		DATA TYPE
Incidence and burden of disease	**Access to treatments and services**	**Scale and impact of rare disease investments**
Detailed prevalence rates aligned to international coding standards	Quality ratings covering:	Total public-sector funding
Estimated total cost to healthcare systems, including screening/diagnostics, treatment costs, care coordination and long-term/palliative care	Availability of essential treatments, diagnostics and services	Total philanthropic funding
Socioeconomic burden of disease, rigorously quantified for key stakeholders	Healthcare professional training and awareness	Total private-sector investment, including number and estimated total market value of companies with rare disease focus
	Reimbursement	Total number of jobs tied to rare disease research, care and/or innovation
	Social services	National rare disease policy assessment (quality rating)
	Patient advocacy	

COE = centre of excellence
HTA = health technology assessment
Source: World Economic Forum, Rare Disease Community analysis

In higher-resource settings, the minimum dataset can expand into a more comprehensive dataset capturing details on prevalence patterns, socioeconomic impact, quality and coordination of care, levels of public and private investment and workforce capacity. Governments can strengthen these efforts by incorporating third-party data sources, including from payers or employers.

A globally aligned minimum dataset will not solve every problem, but it can establish a shared baseline, spotlight where action is most needed and help ensure that rare disease policy and investment are grounded in evidence.

2.2 Strengthen patient engagement in data collection

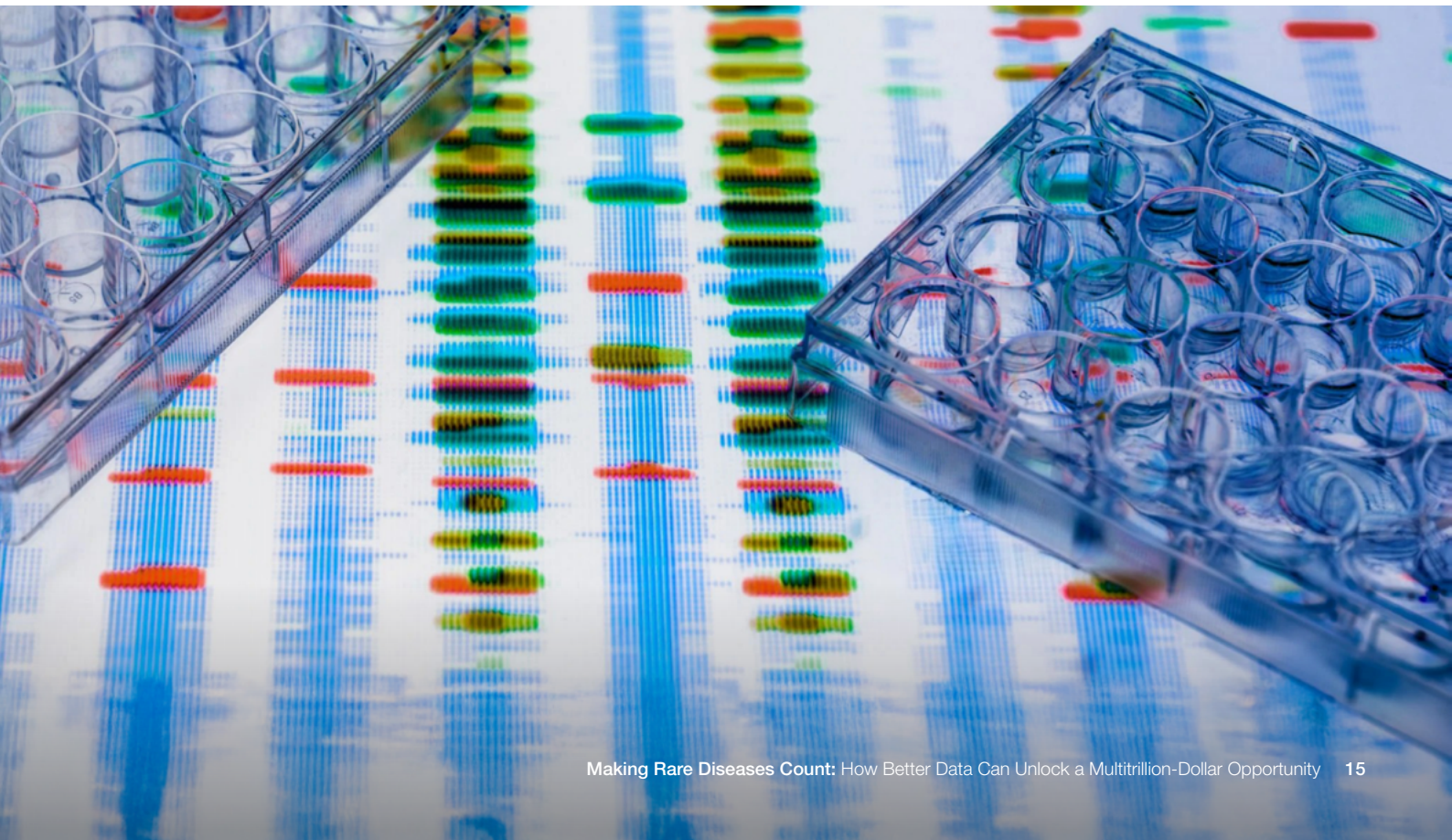
Efforts to improve the quality and comprehensiveness of rare disease data frequently overlook the individuals, families and communities directly affected. These stakeholders bring unique insights, including natural history data, social and environmental determinants of health, quality-of-life measures and day-to-day experiences with care and treatment. When these insights are captured systematically through "patient registries" – structured databases that collect and track health information from individuals living with a specific condition – they become engines of innovation.[22] Examples such as the Registry and BioBank platform operated by the Genetic Alliance (a US-based non-profit that works with patient communities to develop shared data infrastructure and diagnostic programmes to support research and care), launched in 2003, demonstrate how early adoption of these principles can create durable infrastructure for research and care.[23]

The design of these systems matters as much as their content. Registries and data platforms must be participatory and equitable to build trust and fairness. Involving patients as co-creators in governance and implementation enhances legitimacy, reduces duplication and increases the likelihood that the data generated will be used in decision-making.

A powerful early example of participatory data collection came from the online community PatientsLikeMe. When a small 2008 study suggested lithium might slow the progression of amyotrophic lateral sclerosis (ALS, also known as Lou Gehrig's disease or motor neurone disease), patients used the platform to share their experiences and outcomes in real time. Within months, hundreds had contributed structured data, enabling a rapid analysis far larger than the original trial. Although the results showed no therapeutic effect, the effort demonstrated how patient communities can generate credible evidence quickly, ethically and at scale.[24]

In LMICs, registries can be particularly powerful tools for strengthening health systems and expanding access to care. In Telangana state, India, a partnership with the World Economic Forum has introduced a haemophilia clinical registry that helps clinicians deliver more consistent care, supports better resource allocation by authorities and lays the groundwork for incorporating more patient-reported data in the future.

Given the small number of affected individuals with any single rare disease, global collaboration is essential.[25] Registries must transcend cultural and linguistic barriers to create datasets that are inclusive and representative, capturing diversity across race and ethnicity, geography, socioeconomic status and other determinants of health.



CASE STUDY 1

Harnessing registry data to advance PWS therapies

Prader-Willi syndrome (PWS) is a rare neurodevelopmental disorder that causes developmental delays, intellectual disability and a spectrum of complex medical and behavioural challenges. Among the most debilitating features is hyperphagia, a chronic and insatiable hunger that often leads to life-threatening weight gain and severe anxiety. The impact on families is profound. A 2018 study found caregiver impact in PWS exceeded that reported in dementia, Alzheimer's disease and traumatic brain injury.[26]

Building a high-impact registry for novel insights

To better understand and address the real-world challenges of PWS, the Foundation for Prader-Willi Research (FPWR) launched a patient registry in 2015. Hosted on the US National Organization for Rare Disorders (NORD) IAMRARE platform, the registry collects structured data on symptoms, medications, developmental milestones and other aspects of patient and caregiver experience.

By 2025, over 2,200 participants had completed more than 60,000 surveys. Most participants are from the United States, but the dataset covers dozens of countries. While this represents only a fraction of the estimated 350,000 individuals living with PWS globally, the dataset has proven large and diverse enough to generate meaningful, actionable insights for the community.

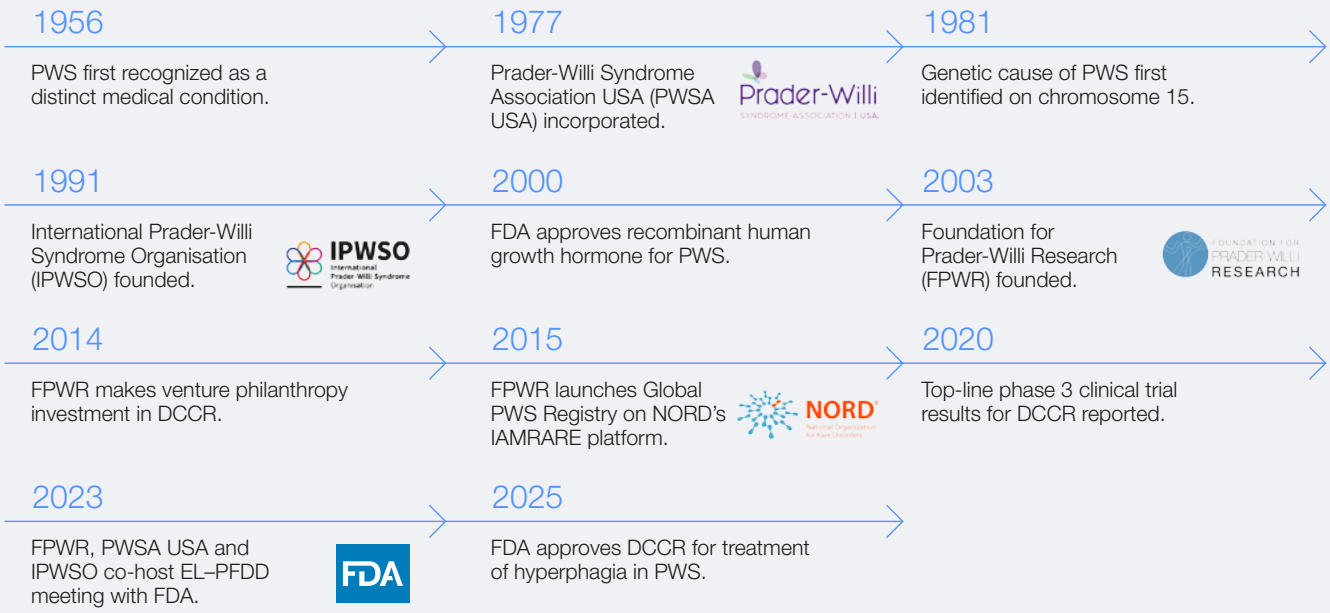
The registry has enabled peer-reviewed publications and clinical insights that have helped shape best practices.[27] These findings have supported clinician education, informed research priorities and helped guide FPWR's strategic decisions. For example, registry data revealed that many PWS cases in the US are still not diagnosed until after the first year of life, evidence that has informed FPWR's investments in newborn screening.

Supporting regulatory engagement and therapeutic breakthroughs

FPWR has also made use of registry data to strengthen its engagement with regulators and policy-makers. During meetings with the US Food and Drug Administration (FDA), structured evidence from the registry added analytical depth to caregiver and patient testimonials, helping translate lived experience into quantifiable patterns of disease impact and unmet need.

More notably, registry insights played a key role in the 2025 FDA approval of DCCR (diazoxide choline extended-release) tablets.[28] This is the first therapy approved specifically to treat hyperphagia in PWS. It represents the culmination of years of collective effort (see Figure 8) and the beginning of a new chapter in PWS care.

Figure 8 – Timeline of PWS community milestones, 1956–present



Source: Foundation for Prader-Willi Research

FPWR continues to advance its registry infrastructure. New platforms allow expert clinicians to contribute structured clinical observations to complement patient-reported data. The foundation also recently launched a registry for Schaaf-Yang syndrome, a condition with close genetic and phenotypic overlap with PWS. Additionally, FPWR is collaborating with peer organizations to broaden participation in under-represented populations, aiming to make its data even more inclusive and globally representative.

2.3 Improve newborn screening and diagnostic capacity

For many rare diseases, early diagnosis is critical to enabling timely treatment, informed care decisions and better outcomes. It also serves a broader purpose: by identifying cases early and systematically, it generates more accurate prevalence estimates that strengthen the overall evidence base for rare diseases.

The economic case for improving early diagnosis is compelling, even before factoring in the value of the data these tools produce. A 2022 study found that delays in diagnosis across seven rare conditions led to avoidable costs of up to $517,000 per patient.[29] While these figures reflect a limited set of diseases, they illustrate how prolonged diagnostic journeys can impose substantial economic and human costs that are echoed across many rare conditions.

One way to streamline early diagnosis is to expand newborn screening (NBS). In many countries, newborns are screened through state-sponsored, population-level NBS programmes. The scope and quality of these programmes vary dramatically. Some screen for more than 60 conditions, others cover only a few and some countries have yet to establish national NBS programmes.

At the leading edge, researchers are exploring the use of next-generation sequencing (NGS) for newborn screening. Numerous initiatives worldwide are now evaluating which conditions to include, how results should be returned and what ethical and operational safeguards are required. In parallel, commercial offerings are beginning to emerge from companies such as Nurture Genomics, bringing NGS-based newborn screening into clinical settings while public health frameworks continue to evolve.

Since NBS is not diagnostic, programmes are expanding follow-up testing, both to confirm findings and to detect cases screening may miss. Whole genome sequencing (WGS), pioneered in critically ill children, is now applied more broadly, including to adults who have lived for years without a diagnosis. Advances in next-generation diagnostics and AI are also creating new tools to uncover elusive conditions and address inequities in diagnosis.

The Genetic Alliance's iHope programme illustrates this potential. As the largest free clinical genomic testing effort worldwide, iHope has enabled clinical WGS and whole exome sequencing (WES) for more than 3,000 families from 30 clinical sites in 14 LMICs and 5 HICs. In a recent analysis of 1,004 participants, 41.4% received a molecular diagnosis and over two-thirds of those diagnoses were associated with changes in clinical management, genetic counselling or avoidance of further testing.[30] The programme also empowers families by granting them control of their genomic data through a user-centric platform.

Collaborative initiatives such as the Undiagnosed Hackathons, founded and led by the Swedish non-profit the Wilhelm Foundation, show how cross-disciplinary teams are working together to tackle cases that remain unsolved even with today's most advanced technologies.[31] These efforts also generate data and insights into the estimated 350 million people worldwide living with undiagnosed conditions.[32]

Clinician awareness is also critical to improving early detection. Accredited continuing education programmes have shown tangible results: paediatric neurologists who participated in rare disease training on Medscape – the world's largest medical education platform – ordered 69% more genetic tests than those who did not.[33] Such findings highlight the importance of investing in professional education as a practical lever for earlier diagnosis.

CASE STUDY 2

Rady Children's Institute for Genomic Medicine (RCIGM)

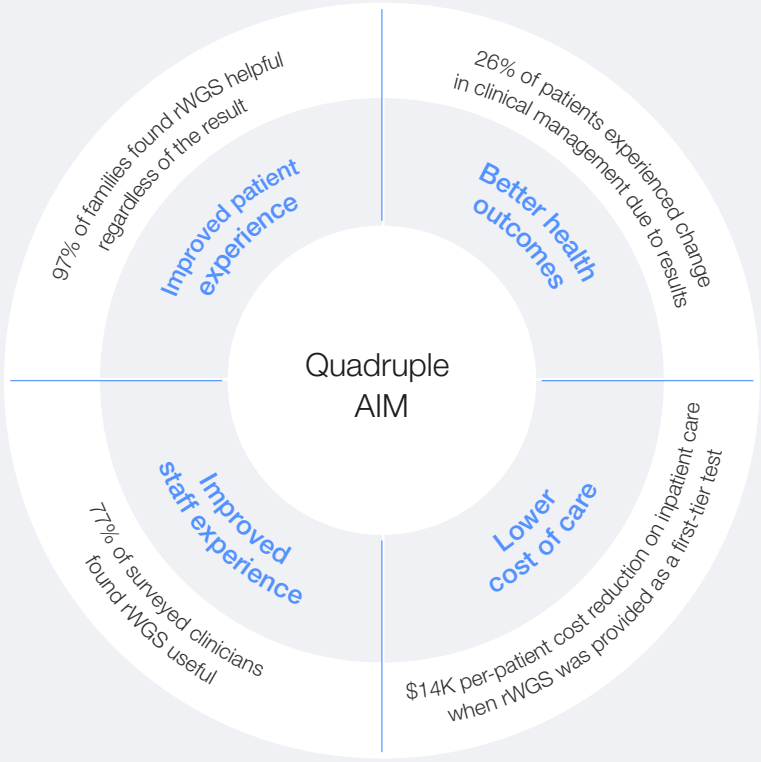
Rady Children's Institute for Genomic Medicine (RCIGM), based in San Diego, US, has emerged as a global leader in applying next-generation sequencing to rare disease diagnostics and screening. Data excellence, innovation and a focus on improving health outcomes for children are central to its model.

Scaling rWGS and achieving reimbursement

One of RCIGM's flagship achievements has been the roll-out of rapid whole genomic sequencing (rWGS) services across more than 100 clinical sites in 29 US states. Its model integrates sequencing, bioinformatics and clinical interpretation into a seamless end-to-end workflow, transforming care for some of the most vulnerable patients: critically ill children with diseases of unknown origin.

RCIGM has systematically validated rWGS, demonstrating broad acceptance by families and clinicians, as well as cost savings by reducing unnecessary interventions and shortening hospital stays (see Figure 9). Between 37% and 50% of children who undergo rWGS receive a diagnosis, and many experience immediate changes in clinical management.[34] These results helped secure US healthcare programme Medicaid coverage for rWGS in 18 US states as of 2025, alongside coverage under most major national private insurers.

Figure 9 – Impact of rWGS on patients, clinicians and health systems



Source: Rady Children's Institute for Genomic Medicine

Extending to newborn screening and AI

Another flagship initiative is BeginNGS, a research consortium applying WGS to screen newborns and infants for more than 500 rare diseases. The programme aims to demonstrate the utility and cost-effectiveness of sequencing in newborn screening, deepen understanding of rare disease incidence and prevalence and engage stakeholders in preparation for potential population-wide implementation.

RCIGM has also applied AI to genomic and clinical data, improving interpretation speed and accuracy, identifying undiagnosed patients and extending access to advanced diagnostics across diverse settings. Through Project Bridge, researchers have used AI to analyse electronic medical records from the Rady Children's Health system to flag patients likely to have undiagnosed rare conditions, including older individuals who missed early genomic testing and spent years navigating the system without answers.

These innovations highlight the central role of data excellence in rare disease progress. By generating, analysing and sharing high-quality data, RCIGM has shown how targeted investment in cutting-edge diagnostics can deliver rapid, life-changing answers for children and families, while also generating compelling savings for healthcare systems.

2.4 Enable trusted data sharing across health systems

Rare disease progress depends on how effectively data can move across institutions and borders. Yet today, most information remains scattered across electronic health records, registries and other databases that rarely communicate with one another. This fragmentation limits the usability of rare disease data, causing duplication of effort and slowing innovation.

A trusted data-sharing ecosystem allows information to flow securely between stakeholders while respecting privacy, ethics and sovereignty. Federated data systems offer a compelling model for making this possible.[35] In such systems, datasets stay where they are generated but insights travel freely. The goal is not to centralize information but to connect it while giving patients and institutions confidence in how their data is used.

Achieving this vision requires both technical and institutional alignment. On the technical side, shared standards enable data to be captured consistently and linked across systems. On the institutional side, common governance frameworks ensure that data sharing is ethical, secure and transparent. These frameworks include privacy-by-design architecture, consent management systems and clear rules for data access and secondary use.

Shared standards for data quality and validation also enable regulators and health authorities to draw on one another's evidence, reducing duplication and accelerating access to innovation. This harmonization also helps industry, academia and patient organizations to collaborate at scale, confident that their data collection efforts will be recognized and respected across systems.

Regulators are increasingly drawing on trusted counterparts to streamline reviews and reduce duplication. Collaborations such as the Access Consortium – a coalition of agencies from Canada, Switzerland, Australia, Singapore and the United Kingdom – show how aligned evidentiary standards for new medicinal products, including rare disease treatments, can speed approvals while maintaining rigour. The International Coalition of Medicines Regulatory Authorities (ICMRA) likewise called for a shared toolbox to harmonize accepted endpoints and evidence standards for rare disease therapies in its 2024 Lugano workshop recommendations.[36]

The benefits of trusted data-sharing frameworks are wide-ranging: for policy-makers, they improve efficiencies in health administration, strengthen oversight and enable evidence-based planning; for researchers and innovators, they expand access to representative datasets and diverse populations; for patients and families, they make participation in research and care pathways more transparent and impactful.

CASE STUDY 3

The European Health Data Space and TEHDAS2

The European Union is prioritizing cross-border data accessibility through the European Health Data Space (EHDS). The regulation, which came into force in March 2025,[37] will connect national health system records to ensure secure access to health data across Europe. Its implementation is being shaped through the Second Joint Action Towards the European Health Data Space (TEHDAS2), an EU-funded joint action that develops guidelines and technical specifications for EHDS operation and secondary use of health data.

A comprehensive framework

EHDS will enable electronic health records, laboratory and imaging results and other health data to be accessible for primary use across borders, allowing patients to carry their information with them when they seek care, improving continuity and avoiding unnecessary repetition of tests.

For research and innovation, EHDS provides a decentralized infrastructure called HealthData@EU for accessing and analysing anonymized and pseudonymized data in secure processing environments. This opens new opportunities for pooling rare disease data across Europe, strengthening research, improving regulatory evidence and supporting the development of new healthcare technologies.

Impact on rare diseases

For the rare disease community, EHDS represents a major opportunity. A European Organisation for Rare Diseases (EURORDIS) survey found that 97% of people living with a rare disease are willing to share their health data for research on their own condition, and 95% would do so for other conditions.[38] EHDS provides a vehicle to transform that willingness into action.

EHDS also addresses persistent challenges in data use and reuse across Europe and serves as a model globally. From a health equity perspective, it has the potential to reduce disparities across member states, giving patients in smaller or less-resourced countries a more equal opportunity to benefit from data-driven advances.

From regulation to implementation

Although the regulation has entered into force, practical implementation is still under way, with key provisions on the secondary use of health data scheduled to apply from March 2029 as part of a phased roll-out. Ongoing initiatives including TEHDAS2 are producing the guidelines and technical specifications that will guide the EHDS operation and the secondary use of health data. These will undergo public consultation; patients, providers, regulators and innovators are invited to participate.

2.5 Use AI and digital tools to address evidence gaps

Rare disease data is often unavailable, fragmented or difficult to analyse, limiting its value for all stakeholders. Advances in AI and digital tools are beginning to change this. These technologies are helping transform scattered information into structured insights and opening new possibilities for research, clinical care and health system planning.

Private AI and digital health companies play a vital role in powering these opportunities. One example is Huma, a United Kingdom-based company that has evolved from a rare disease specialist into a provider of regulated, AI-enabled digital health platforms used across healthcare systems worldwide. Its Huma Cloud Platform allows partners to build and launch AI-powered health tools quickly and securely, including for rare conditions where traditional software development approaches may be commercially unviable.

Huma's technology is actively applied in rare disease contexts, including through projects with global pharmaceutical companies UCB for myasthenia gravis and Pfizer for haemophilia, which improve symptom tracking, patient engagement and real-time data sharing. Its federated design also enables analysis across multiple data sources in small and geographically dispersed patient populations without moving or exposing sensitive information, helping protect patient privacy while expanding the reach and coordination of care.

Other collaborations between industry and technology firms are expanding the use of AI to detect and manage rare diseases. Pangaea Data and Alexion, AstraZeneca Rare Disease have partnered to develop an AI clinical decision support system to detect hypophosphatasia in adults, a rare metabolic disorder that is often missed due to its diverse, non-specific symptoms.

Complementing this effort, Alexion, AstraZeneca Rare Disease has developed deciphEHR™, a suite of educational resources and toolkits that help healthcare organizations make better use of their EHR systems to identify patients potentially affected by rare diseases. By using relevant patient history, disease codes and suspect patient lists, deciphEHR™ supports clinicians in triaging patients for further evaluation.

Public-sector organizations are also applying AI to strengthen rare disease data and improve health system intelligence. Genomics England's Clinical Variant Ark, a secure knowledge base integrating genomic and clinical data from tens of thousands of rare disease families, has already facilitated more than 100 new diagnoses by enabling experts to build on prior evaluations.[39] Foresight, a generative AI trained on de-identified records from 57 million people in England's National Health Service Secure Data Environment, uses national-scale data to forecast major health events across all demographics, including those living with rare conditions.[40]

While private and public organizations are making important progress independently, some of the most powerful opportunities emerge through public–private partnerships that combine the reach and governance of public systems with the innovation capacity of private actors. These collaborations strengthen data infrastructures, accelerate responsible use of AI and ensure that new tools are designed with patients at the centre.

CASE STUDY 4

Sanofi's OSCAR AI algorithm and Project Saturn

Rare diseases remain difficult to identify early because data is fragmented across institutional silos. Traditional AI models rely on integrated data warehouses that are expensive to build, difficult to govern and often unviable in many health systems. What is needed is a fundamentally new architecture that enables analytics across distributed data sources without moving data or compromising privacy.

The OSCAR model

Global biopharmaceutical company Sanofi's OSCAR (multimOdal tranSformer foundation model for Clinical Analysis of Rare diseases) algorithm, developed through the cross-industry partnership Project Saturn, demonstrates how multimodal, privacy-preserving AI can address this challenge. In its initial clinical evaluation shared through the American Diabetes Association, OSCAR increased detection efficiency for type 1 diabetes more than 18-fold and corrected diagnostic misclassification for nearly one-third of affected individuals, reducing delays to appropriate treatment.[41] The model has been tested computationally and is planned for subsequent validation in clinical practice. The same architecture allows deployment across diverse health systems, from tertiary research networks to emerging national infrastructures in LMICs, contingent upon alignment with local regulations.

Continued refinement of OSCAR uses a federated learning architecture, training models across decentralized datasets that remain securely within each participating institution. This enables advanced analysis of clinical, genomic and other data while maintaining data sovereignty and compliance with local privacy standards.

Extending to rare diseases

The OSCAR model is now being adapted for rare disease detection, starting with Pompe and Fabry diseases, two lysosomal storage disorders with subtle, multisystemic presentations that make them ideal test cases for multimodal AI approaches. By fine-tuning the architecture on rare disease datasets that combine genomic, clinical and laboratory signals, researchers aim to identify early markers that could significantly shorten diagnostic journeys.

Deployment pilots are planned for 2026, expanding collaborations with health ministries, patient organizations, research institutions, payers and industry partners. The goal is to create a global federated learning network by 2030, enabling privacy-preserving AI to improve early detection, diagnosis and coordinated care for rare diseases worldwide.

Integrating AI with coordinated care

Data infrastructure and AI can reveal new insights, but their full potential is realized only when linked to care models that provide practical benefits for patients, caregivers and communities. Coordinated care connects data-driven insights with human support networks, allowing interventions to extend beyond the clinic into daily life.

Programmes developed under Project Saturn show how digital tools can empower families to manage health collaboratively, improving adherence, engagement and continuity of care. For rare diseases, where small populations and complex care needs make coordination difficult, such models demonstrate how combining federated AI with relational support can improve outcomes, creating a virtuous cycle of insight and intervention.

3 Tailoring strategies to local contexts

Adapting rare disease data strategies to local needs ensures that global ambitions translate into equitable, lasting progress.

While the value of better rare disease data is universal, the pathways to achieving it vary greatly. Health systems differ widely in their resources, governance, digital infrastructure and political priorities. Strategies that succeed in one setting may fail in another if they ignore these realities.

Tailoring approaches to each context requires an understanding of technical readiness alongside cultural, economic and political dynamics. This means engaging local stakeholders, building on existing capabilities and aligning with broader health and social priorities. This section outlines practical frameworks for adapting rare disease data strategies to diverse contexts.

3.1 Building healthcare system capacity in LMICs

In LMICs, basic capacity constraints are often the most immediate challenge to rare disease data collection. Health systems may be under-resourced and overstretched, with shortages of trained professionals, fragmented or paper-based records and limited governance frameworks for data use. Affordability is a constant pressure, and inequitable digital access further limits reach.

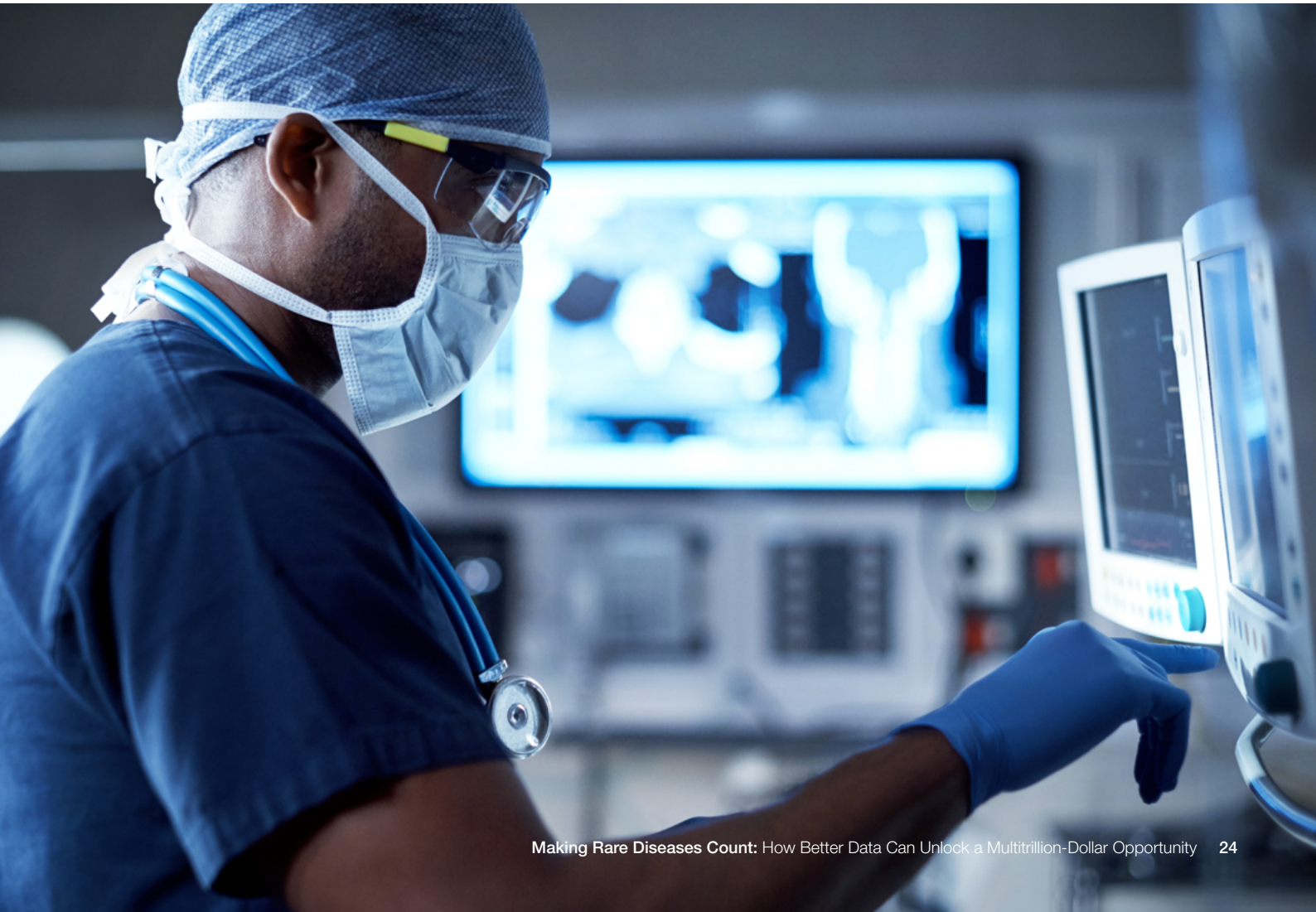
These barriers are real, but they are not insurmountable. With appropriate tools, training and support, individuals and communities can play an active role in data initiatives. Local patient groups – both formal and informal – are often pivotal, expanding registries, mobilizing participation and ensuring that systems reflect community priorities.

Strengthening the healthcare workforce is critical. This includes training clinicians to recognize and accurately record rare diseases, equipping data managers to handle sensitive information securely and enabling regulatory authorities to oversee quality, privacy and ethical use. Primary care networks can be particularly powerful levers,[42] serving as gateways for registry enrolment and early case identification.[43]

Directly transplanting strategies from high-income settings is rarely effective. Yet approaches that prioritize interoperability, patient engagement and low-cost, adaptable tools can succeed. Many LMICs are already innovating in ways that are more cost-effective and sustainable than those in wealthier nations. Mobile health platforms, for example, have enabled the collection of patient-reported outcomes in areas with limited broadband but broad mobile coverage.

Some countries are also leapfrogging traditional barriers. China's nationally networked hospital and rare disease data ecosystem was built rapidly, avoiding many of the legacy integration problems faced elsewhere.[44] Economic incentives can also accelerate progress – India, for instance, is positioning rare diseases as a growth sector, leveraging its manufacturing capacity to supply therapies, including generic medicines, for global markets.

Sustainable financing models are essential for embedding rare disease data initiatives into national health strategies. Approaches such as blended finance, pooled regional resources and public–private partnerships are increasingly helping LMICs move beyond short-term donor dependency towards self-sustaining rare disease programmes.



CASE STUDY 5

How Egypt is building a data backbone for rare diseases

Egypt is showing how a resource-constrained health system can become a regional leader in rare disease innovation by making data visible and actionable. Over the past two decades, it has built one of the most comprehensive systems of rare disease recognition and response in the Middle East and North Africa (MENA). Its progress and potential are driven by a combination of data-driven innovation and sustained political commitment to the rare disease community.

From screening data to systemic reform

Data-driven screening and diagnostics have been central to Egypt's progress. According to Egypt's Ministry of Health, a digitalized newborn screening programme now covers more than 90% of neonates. Pre-marital screening has reached 3 million citizens, providing valuable population-level insights into haemoglobinopathies and other conditions.

Evidence from these programmes has directly shaped policy. Early findings from newborn screening for congenital hypothyroidism and phenylketonuria (PKU) demonstrated cost-effectiveness, convincing policy-makers to expand coverage nationally. The results led to the creation of specialized treatment centres, provider training and standardized protocols that have embedded rare disease management across the health system.

Data continues to drive reform. Research from Ain Shams University showing a 4.35% prevalence of genetic disorders in paediatrics has influenced medical curricula and spurred the establishment of new genetics units across universities. Egypt is also piloting AI-based diagnostic systems to shorten diagnostic delays and guide resource allocation.

Building a national registry

Egypt's wider digital health transformation is creating new opportunities for rare disease care. The Digital Egypt 2030 strategy and Universal Health Insurance System already hold more than 4.5 million electronic health records and 42 million digital prescriptions. These assets provide the backbone for a planned national rare disease registry.

A dedicated scientific committee is reviewing registry data fields to ensure relevance for rare diseases. Universities, research centres and international partners are collaborating with the government to build capacity and secure funding. The Egyptian Healthcare Authority is also expanding "virtual hospitals" through telemedicine to reach underserved communities.

Sustained political commitment

Egypt's government has reinforced and accelerated these efforts. Health is enshrined as a constitutional right, providing a strong legal foundation for rare disease care. This principle has been translated into practice through the passage of a rare disease law that secures governance and sustainable funding.

More recently, rare diseases were elevated to national priority status alongside cancer and cardiovascular disease in the National Health Strategy 2024–2030. On the global stage, Egypt co-sponsored the 2025 World Health Assembly resolution, signalling intent to help shape the forthcoming Global Action Plan on Rare Diseases.

The prioritization of rare diseases in Egypt reflects both scientific and societal realities, notably a high prevalence of genetic disorders linked to consanguinity, which prompted the early adoption of newborn screening and research focused on locally prevalent conditions. Comparable initiatives are emerging across the MENA region, where similar issues have spurred investment in national screening and rare disease initiatives.

With strong political commitment, a rapidly maturing digital infrastructure and growing international partnerships, Egypt is transforming invisible conditions into measurable health gains – and showing that even resource-constrained systems can lead in rare disease innovation.

3.2 Maximizing impact in high-income countries

High-income countries often have advanced health and data infrastructure but face different challenges. Systems are frequently siloed, fragmented or misaligned across agencies, making it difficult to link and analyse data at scale. In this context, the opportunity lies in expanding and integrating what already exists rather than building from scratch.

Priorities may include stimulating and harmonizing research with specialist care centres, embedding rare diseases into national biobanks and genotype–phenotype studies and ensuring interoperability across secure data environments. Some countries are already moving in this direction – a United Kingdom government and Wellcome Trust investment of up to £600 million ($808 million) is building a federated health data research service that will enhance the National Disease Registration Service, improving access and linkage to rare disease data for research and trial recruitment.[45]

For high-income countries, the challenge is to align technical capability with policy, governance and incentives that encourage data sharing and coordinated action. When these elements are in place, mature systems can generate high-quality data that not only benefits their own populations but also strengthens the global rare disease ecosystem through shared insights and innovations.

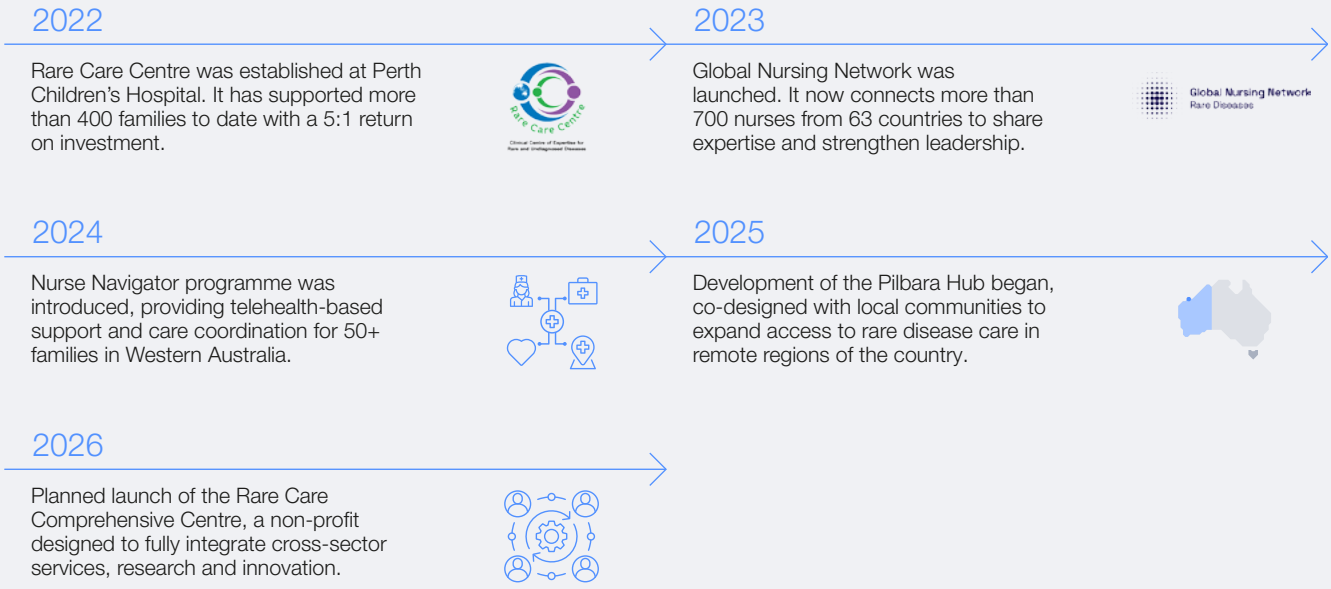
CASE STUDY 6

Rare Care Centre at Perth Children's Hospital in Western Australia

The Rare Care Centre (RCC) was established at Perth Children's Hospital in February 2022 to provide a new model of support for children and families affected by rare and undiagnosed diseases. It operates through a statewide, cross-sector, inter-agency care coordination programme that has already demonstrated value in improving outcomes and reducing costs to the health system.

Rather than focusing solely on the hospital setting, the RCC coordinates and advocates across health, education, disability and social care systems. A nurse navigator programme extends reach via telehealth for families with lower levels of complexity, while a hub in northwestern Western Australia is expanding access in one of the country's most remote regions (see Figure 10).

Figure 10 – Rare Care Centre: 2022–present



Source: Rare Care Centre, Perth Children's Hospital

Improving outcomes and delivering value

Analysis of discharged patients from the RCC highlights significant benefits. Coordinated care reduces hospital visits, travel requirements and stress for families while improving access to disability and welfare entitlements. Families living outside metropolitan Perth save an average of AUD $5,200 (US $3,500) annually in travel-related costs, and disability support funding increases have enabled greater access to therapies and equipment.[46]

The health system also benefits. Outpatient appointments fell by 33%, hospital bed days decreased by 33% and emergency department presentations went down by 40%. After programme costs, this equates to net savings of AUD $13,400 ($8,970) per child – a return of nearly AUS $5 for every dollar invested.

Building capacity and advancing innovation

Beyond clinical service delivery, the RCC invests in education, workforce development, research, peer support and system reform. It hosts the Global Nursing Network for Rare Disease, connecting nurses worldwide to share expertise and strengthen leadership. It has launched educational tools for schools, including animations and e-learning resources to improve understanding and support for children in classrooms.

The RCC is also advancing digital innovation through platforms such as Unlocking Treatment Options Personalized In-Time Access (UTOPIA), which applies AI and digital tools to generate care summaries and computational natural histories for children with rare and undiagnosed diseases. Early results show it can halve the time needed to create personalized care plans, and it is now being adopted nationally in Singapore.[47]

The RCC is now evolving into a Comprehensive Rare Care Centre, integrating clinical and cross-sector services, research, innovation and a dedicated not-for-profit entity. This next phase aims to transform how rare and undiagnosed diseases are diagnosed, treated and supported, while ensuring that children and families remain at the centre of care.

Conclusion

Better data is the foundation for smarter action on rare diseases and for unlocking the broader health, economic and scientific benefits it can deliver. This paper has outlined five strategies to strengthen rare disease data systems worldwide – defining a minimum dataset, engaging patients, expanding diagnostics, enabling trusted data sharing and applying AI and digital tools – each linked to clear objectives, implementation pathways and stakeholder roles (see Table 3).

This recommended call to action in this white paper now aligns with growing global momentum. Rare diseases are increasingly recognized as a public health priority at national and international levels. The World Health Assembly's 2025 resolution on rare diseases and the forthcoming Global Action Plan for Rare Diseases create a shared framework for progress.

Delivering on these ambitions will require collaboration across all sectors, with each stakeholder group playing a distinct but complementary role:

- **Patient organizations and caregivers** can ensure that data systems are participatory, equitable and relevant, bringing lived experience to data design and governance while improving care quality and quality of life.
- **Healthcare systems** can strengthen diagnostic and data-collection capacity, expand registry participation and promote consistent data sharing to accelerate research translation and more innovative care.
- **Employers** can support workforce participation for those affected by rare conditions, benefit from lower absenteeism and turnover and contribute data and insights that help quantify productivity gains.
- **Payers** can use better data to generate stronger evidence for coverage and reimbursement, reduce long-term costs and improve member outcomes.
- **Governments and health authorities** can provide leadership and resourcing, embed rare disease data strategies in public health planning and ensure that insights feed back into equitable policy and system design.
- **Investors and industry** can apply data-driven insights to de-risk R&D, identify new market opportunities and scale innovation responsibly through cross-sector partnerships.

TABLE 3 | **Summary of rare disease data strategies and stakeholder roles**

Why	How	Who
Define and track a minimum dataset across countries		
Lack of alignment in data collection creates blind spots around one of the largest cost areas in healthcare systems, weakening the collective case for investment	Standardize data collection to build a global knowledge base, starting with a core set of metrics aligned with international data standards and able to collect across all health systems	Health authorities lead, with governments providing resources, patient groups ensuring relevance and payers and employers contributing data and insights
Strengthen patient engagement in data collection		
Disease registries and other patient-centred datasets yield insights and evidence that improve care and help clinicians, drug developers, regulators and payers make better decisions	Design and fund data systems that are participatory, equitable and interoperable across borders, engaging patients as co-creators in governance and implementation	Patient groups lead, supported by public and private funders, with efforts aligned and integrated into registries initiated by industry, government and healthcare systems
Improve newborn screening and diagnostic capacity		
Early diagnosis leads to better health outcomes while generating more accurate and unbiased prevalence estimates, strengthening the overall evidence base for rare diseases	Strengthen newborn screening and diagnostic testing, using next-generation sequencing and AI where appropriate, and ensuring data is preserved, shared and used to improve care	Healthcare systems lead, working with private screening and diagnostic firms, laboratories and payers to align incentives
Enable trusted data sharing across health systems		
Fragmented and siloed data systems limit the visibility, usability and trustworthiness of rare disease data, causing duplication of effort and slowing innovation	Build secure, federated data systems that connect datasets across health systems, aligning technical standards, governance models and regulatory approaches to enable ethical, transparent and interoperable data sharing	Regulatory authorities lead, partnering with health ministries, technology firms, industry and patient organizations to ensure effective implementation
Use AI and digital tools to address evidence gaps		
Rare disease data is often unavailable, fragmented or hard to analyse, limiting its value for patients, researchers, healthcare systems, payers and policy-makers	Apply AI and digital tools to generate, integrate and analyse data while maintaining privacy, interoperability and utility across settings	Tech providers lead by developing solutions, with implementation driven by patient groups, healthcare systems, payers, employers, governments, industry or other users

Source: World Economic Forum, Rare Disease Community analysis

With better data and smarter investment, countries can align around a shared agenda for rare diseases and unlock a multitrillion-dollar opportunity for economic and social value. Rare disease research and innovation have already reshaped what medicine can achieve; with global coordination, they can help build a fairer, more resilient future for all.

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Endnotes

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