FROM IDEA TO ACTION:
ACCELERATING RARE DISEASE DIAGNOSIS

RESOURCE COMPENDIUM

FEBRUARY 2025

GLOBAL COMMISSION | to End the Diagnostic Odyssey for Children with a Rare Disease

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Letter from the Co-chairs

The diagnostic odyssey remains one of the most serious challenges for families confronting the possibility that their child may have a rare disease. About 70% of rare diseases — which affect 300 million people around the world — begin in childhood. It is estimated that it takes an average of five years to receive an accurate diagnosis, even in countries with strong health systems. Delays in diagnosis have devastating consequences, especially for children, including worsening and harder-to-treat symptoms, bleaker prognosis, higher rates of mortality due to lags in receiving life-saving treatment, and an immense social and emotional toll on families.

Earlier this year, the Global Commission to End the Diagnostic Odyssey for Children with a Rare Disease — a group of rare disease advocates, researchers, physicians, and innovators — published a Framework to Guide Action in Accelerating Diagnosis for Children. This framework is intended to support global, regional and national efforts to end the diagnostic odyssey. To bring this framework to life, ***From Idea to Action*** highlights innovations in rare disease diagnosis from across the globe — a resource compendium of case studies with practical guidance on how to adopt similar models to accelerate diagnosis.

We are excited to elevate the creativity, passion and ingenuity of dedicated advocates for children living with a rare disease and share their impactful stories.

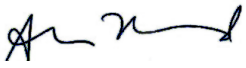
These innovators are driving change within vastly different socio-economic contexts. It is not surprising that their solutions are diverse, reflecting each country's particular barriers to diagnosis. Nonetheless, several cross-cutting themes have emerged that we found to be critical to their success: the transformative power of technology in accelerating diagnosis, the value of global collaboration within the rare disease community, the importance of government engagement for sustainable change and more.

From high-income countries including Australia and Japan to middle-income countries like Brazil and the Philippines and developing countries such as the Democratic Republic of Congo and Ghana, we believe that innovation is happening in every corner of the world. We hope that these success stories can inspire other countries to take action in ending the diagnostic odyssey for children.

We are grateful to the members of the Global Commission and to the rare disease champions whose impressive work is featured in this report. Together, we are optimistic that we can fulfill our vision of a clear path to a timely, accurate diagnosis for all children. Please join us.

Sincerely,

The Global Commission Co-chairs



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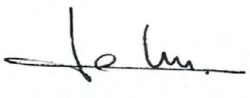

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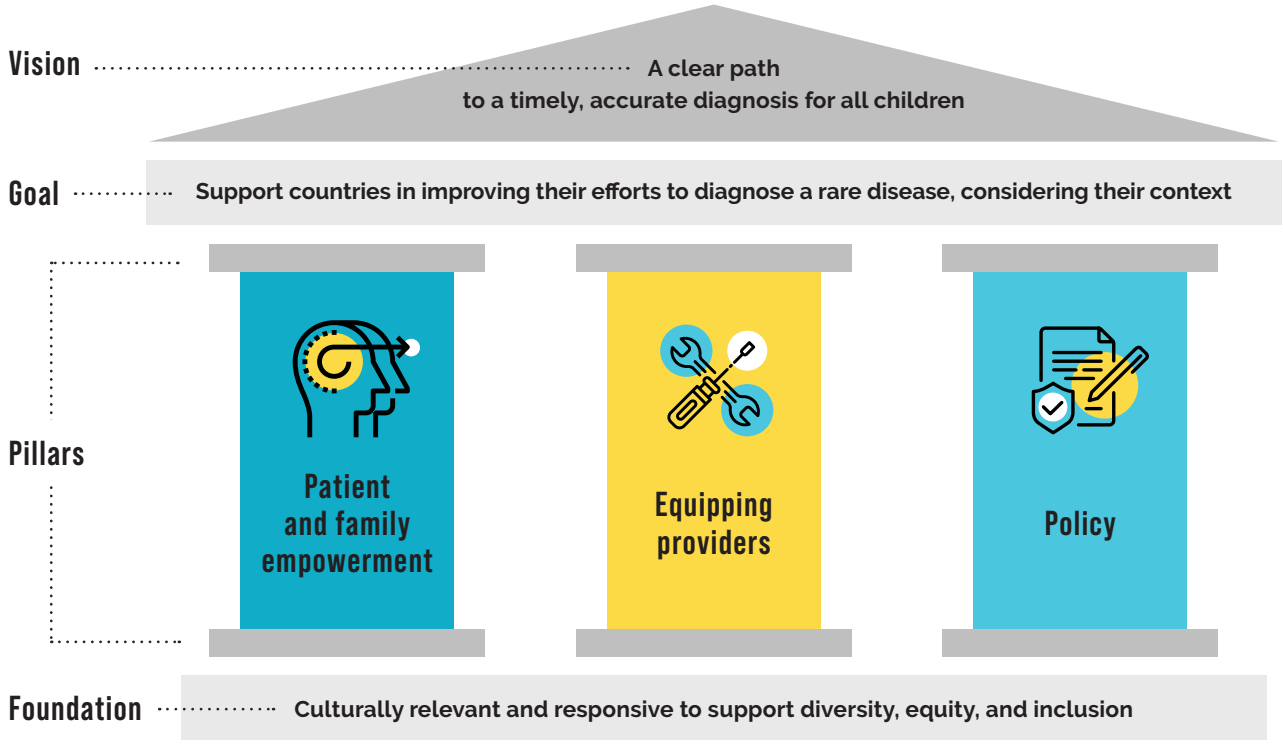
Founder & Past-CEO, EURORDIS – Rare Diseases Europe

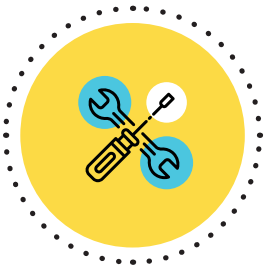
Introduction

In May 2024, the Global Commission to End the Diagnostics Odyssey for Children with a Rare Disease launched its newly developed Framework to Guide Action in Accelerating Diagnosis for Children to inspire innovation in diagnosis and support countries as they seek to improve their efforts to diagnose a rare disease.



The framework has three pillars which are foundational to securing a timely, accurate diagnosis for children around the world:

Patient and family empowerment
Empowering patients and families with the information and resources they need to get closer to a diagnosis



Equipping providers
Ensuring that all health providers — regardless of where they practice — have access to the right tools and knowledge to identify rare disease "red flags" and make quicker referrals

Policy
Mobilizing advocates on national, regional, and global levels to change policies that will help accelerate and improve the quality of diagnosis for more people

To help make this framework actionable, we have developed a Resource Compendium — case studies of exemplars from around the world that are on the path towards ending the diagnostic odyssey. Many are addressing inequities in diagnosis and have strong potential for expansion, including in low-and-middle income countries where diagnostic challenges are greatest.

Thus far, we have been able to publish 10 exemplars. Eight are from countries as diverse as Brazil, Ghana and Japan, one is regional and two are global. We developed these case studies based on in-depth interviews with the rare disease champions who are leading these impressive efforts.

In some cases, this is the first time that these stories have been told in such detail. Although they are quite diverse — from using the power of artificial intelligence to identify potential diagnoses based on symptoms families report to encouraging new parents to participate in a newborn screening pilot project — each one offers learnings that are applicable to many different settings and provides valuable considerations for replication.

Case studies represent eight different countries, two global, and one regional effort:

Through this growing **Resource Compendium**, we are learning about new and innovative approaches to accelerate diagnosis and have identified key themes. We found that while the barriers — or intensity of the barriers — to diagnosis may differ depending on the context, there are common solutions to overcome them that we encourage rare disease advocates to adopt:

- Leverage technology
- Build tailored training programs for health providers
- Understand and respond to community needs
- Generate data to drive policy change

From Idea to Action also highlights elements of success that these case studies share, including respecting the cultural context, engaging government early, collaborating across borders and more. We hope these success factors provide valuable guidance in developing similar initiatives.

Finally, we have included recommendations aligned with the Framework for Action and focused on the activities we encourage patient advocacy groups, health providers and policymakers to undertake to end the diagnostic odyssey.

This resource compendium aims to be a useful tool for rare disease advocates, innovators and policy makers to achieve the vision of a clear path to a timely, accurate diagnosis for all children.

Innovative solutions to overcoming barriers to diagnosis

This growing resource compendium of exemplar case studies highlights how passionate members of the rare disease community have developed innovative solutions to the particular challenges to diagnosis in their countries. Many of these barriers are common even though the contexts are vastly different. Lack of awareness and information about rare diseases, limited ability of frontline health providers to identify a potential rare disease, absence of data on the burden of rare disease and its impact on families and the health care system, and inadequate access to genetic testing all contribute to a multi-year diagnostic odyssey.

KEY BARRIERS

Patients and families

- Lack of awareness and information about rare disease
- Challenges in communicating concerns with heath providers
- Limited access to genetic testing

Providers

- Inadequate access to diagnostic/testing tools
- Limited ability to identify rare diseases, given symptoms are often similar to common conditions
- Shortage of rare disease specialists

Policy

- Lack of rare disease strategy
- Absence of data on rare disease and its impact
- Inadequate testing capabilities and/or reimbursement

This section describes four cross-cutting solutions that emerged from the exemplar case studies and are showing promising results.

SOLUTION 1:

Leveraging technology to increase access to diagnosis

Given that an estimated 80% of rare diseases are genetic in origin, genetic testing and genomic sequencing are key diagnostic tools.[1] However, they are expensive and can be difficult to access due to cost and required infrastructure — especially in resource-limited settings.

Advances in technology — including the ability to analyze extensive amounts of data to narrow the search for answers — have the potential to transform the diagnostic journey by making it more affordable and available. From digital platforms that use machine learning to identify symptom patterns and facilitate data sharing among health providers to smartphone applications that enable patients to locate diagnostic centers, technology can accelerate the pace of innovation in diagnosis in a more equitable way.

SPOTLIGHT

DxGPT (Global)

DxGPT, developed by the nonprofit organization Foundation 29, is an easy-to-use, free of charge digital tool that analyzes symptoms that any patient, family member or health provider submits in the quest to find a diagnosis. The emergence of predictive language learning models and generative AI enables users to enter a free-form description of their symptoms and health condition in their own words, eliminating the need for families to decipher medical terms.

The tool then proposes a list of potential rare diseases, making it possible for patients/caregivers to ask targeted questions of their providers and prompting providers to look "beyond the usual suspects" when a case proves difficult to diagnose. More than 62,000 people from countries around the world have used Dx-GPT, which has been integrated into Madrid's electronic medical record system across all primary care settings within the health care system.

SPOTLIGHT

UTOPIA (Australia and Singapore)

UTOPIA is a digital platform that generates a semi-automated summary of a patient's condition based on clinical features (phenotype). The platform deploys a wide range of AI approaches for analysis, including large language models and new concept recognition. The information generated is then filtered against large data sets of diseases.

Medical experts review the output and produce a patient profile, including the likely trajectory of the disease's evolution and how it may affect the child physically and behaviorally — even if a child has not been diagnosed. By capturing what is known about these individual conditions and how they inter-relate and evolve over time, UTOPIA is increasingly able to help create personalized plans for health, education, disability and social services — even without knowing the exact genetic cause.

[1]https://globalgenes.org/rare-disease-facts/

SOLUTION 2:

Understanding and addressing communities' needs

The rare disease diagnostic odyssey is frustrating for individuals and families who are continually bounced around the health care system or, in some cases, completely neglected.

Organizations that understand families' needs, address them holistically and actively engage patients and families have been successful in accelerating testing and diagnosis. They have taken the time to understand cultural, social and economic issues that affect the diagnostic journey. Likewise, they invest in teams with multidisciplinary members who are equipped to meet individualized care needs. These organizations also play an important role in their communities in promoting awareness of rare diseases and reducing stigma.

SPOTLIGHT

Casa dos Raros (Brazil)

Casa dos Raros, also known as the Center for Comprehensive Care and Training in Rare Diseases, provides an interconnected network of care for people living with a rare disease throughout Brazil and Latin America. The Center takes a multidisciplinary approach to meet family and community needs, integrating diagnostic testing, clinical expertise, innovative treatment methods, research, education, training, as well as patient and family support under one roof.

Casa dos Raros' partnership with a patient advocacy group has enhanced its ability to understand the local rare disease community and effectively promote its services. This relationship also facilitates continuous feedback to revise and strengthen Casa dos Raros' programming.

SPOTLIGHT

African Rare Diseases Initiative (Democratic Republic of Congo)

The African Rare Diseases Initiative (ARDI) is a new locally led effort to expand genetic testing, build a network for genomic research and advance provider training to better manage rare diseases in Africa. Launched in the Democratic Republic of Congo, a country with low awareness of rare disease and extremely limited resources, ARDI hosts informational meetings at churches, hospitals and schools to explain the project's objectives, the benefits of genetic testing and the value of receiving a diagnosis.

A deep appreciation of local cultural practices has enabled the team to be more effective when looking for genetic patterns among families and within communities in addition to offering genetic counseling. The team has also been sensitive to language issues, careful to avoid stigma and speak with families in their own language.

SOLUTION 3:

Building tailored and strengthened health provider training programs

Many health providers have limited exposure to a person living with a rare disease and most medical schools spend minimal time on rare disease education. With more than 7,000 rare diseases, it is unrealistic to expect every general practitioner to be able to make a fast and accurate diagnosis.[2]

Equipping frontline health care providers with the knowledge to recognize a possible rare disease and the information to make an appropriate referral and care plan is a huge step toward ending the diagnostic odyssey.

SPOTLIGHT

Global Nursing Network for Rare Diseases (Global)

The Global Nursing Network for Rare Diseases is an effort to consolidate resources on rare diseases and tailor them for nurses — often the first health provider a family will encounter — better equipping them to diagnose and care for people living with a rare disease. The Network is creating a global community to facilitate cross-learning about rare diseases and educate nurses on rare disease symptomology.

This virtual network connects nurses through a website, enabling them, as well as nursing students, to access educational resources (e.g., webinars, learning modules), opportunities to collaborate on projects, a forum to discuss questions about cases, and information about travel scholarships to attend conferences or workshops related to rare disease. As of November 2024, the Network's members include more than 500 nurses from 51 countries, more than half of which are low- or middle-income.

SPOTLIGHT

Rare Disease Ghana Initiative (Ghana)

Rare Disease Ghana Initiative (RDGI), a nonprofit organization, is helping individuals and families receive genetic testing and the medical and social care they need throughout their diagnostic journey. RDGI has trained more than 50 primary care clinicians across 12 facilities in Ghana to identify rare disease signs and symptoms.

These providers make referrals to RDGI when they suspect a patient may have a rare disease but do not have the diagnostic capabilities to conduct genetic testing. Through collaborations with labs in the country and in other countries, RDGI has been able to provide genetic testing at no cost to more than 200 people — helping determine a definitive diagnosis for 70% of them.

[2]https://www.nih.gov/about-nih/what-we-do/nih-turning-discovery-into-health/promise-precision-medicine/rare-diseases

SOLUTION 4:

Generating data to drive policy

Data generation is essential to drive change — especially policy change that leads to action. Government and health system leaders want to see hard facts to understand why they should adopt a new policy — particularly compelling statistics on the economic and social impact.

Generating data on the societal benefit of diagnosing rare diseases has been an effective strategy to advance policies that support testing and quality care and, subsequently, guide allocation of resources to maximize impact. Additionally, data on the prevalence of rare diseases helps raise awareness and guides advocacy efforts.

SPOTLIGHT

Newborn Screening Act (Philippines)

A group of physicians from 24 hospitals across Manila implemented a pilot program to test newborns for select rare diseases, generating valuable data on parents' willingness to have their newborn tested and the economic value of newborn screening. The data was foundational in advocating for successful passage of The Philippines Newborn Screening Act which led to national implementation of newborn screening in the public health system and screening access for all newborns.

As a result of the Act and the power of early detection, close to 300,000 babies have been screened for a rare disease. The number of facilities offering newborn screening in the Philippines increased from 24 in 1996 to more than 7,000 in 2023 and the number of disorders being screened increased from 5 to 30.

SPOTLIGHT

Aotearoa Rare Disease Strategy (New Zealand)

Rare Disorders NZ (New Zealand) — an umbrella organization — led the charge in mobilizing and uniting a diverse set of patient groups, including from the marginalized Indigenous (Māori) community, to provide input on the country's first rare disease strategy. As part of its advocacy to reflect the patient perspective in the strategy, the organization facilitated the government's access to key information about rare disorders.

Specifically, Rare Disorders NZ highlighted data captured by its bi-annual Voice of Rare Disorders Survey which showed that access to diagnosis was a major concern: over half of respondents reported that they waited more than one year to get a diagnosis and, for almost one in five respondents, the journey took over 10 years. Aotearoa New Zealand's first Rare Disorders Strategy was released in July 2024, providing guidance to health entities on how to better support people living with a rare disorder, including the importance of collecting information on rare disorders in national data sets.

Innovations in Diagnosis: Ingredients for Success

The innovations featured in the following 10 case studies — although diverse in geography, health system context, population and solution — share common elements that have contributed to their success and, in many cases, their sustainability. We encourage rare disease advocates and innovators to integrate these features in efforts to accelerate diagnosis equitably.

Harness the passion of advocates
These examples are powered by courageous advocates who are not afraid to break new ground. They have deep personal and professional experience and an unwavering commitment to accelerating diagnosis for people living with rare disease.

Co-design with patients and families
Whether technology tools or advocacy efforts, involving patients and families throughout the development process made these solutions more effective.

Elevate the patient voice
Solutions focused on hearts and minds and that elevate the lived experience of people with a rare disease and their families continue to be integral in efforts to strengthen rare disease policies.

Reflect the cultural context
A deep understanding of the cultural practices, beliefs and norms in each community underpinned many of these solutions, which led patients and families to adopt them more readily and resulted in greater equity.

Provide holistic, cross-sector support
Efforts to support people living with a rare disease and their families didn't stop at diagnosis — they considered their broader needs, regardless of whether they received a diagnosis or are still searching for one.

Engage government early
Working with government at the outset helped ensure that solutions addressed policy makers' concerns — such as impact, feasibility, cost, equity — which set the foundation for sustainability.

Generate data to enable decision-making
The initiatives that were intentional about building evidence during implementation were well-equipped to advocate for policy change that led to sustainability.

Collaborate across borders
Drawing on expertise from countries with more mature rare disease policies and programs inspired lower and middle-income countries to pursue innovations in their context. The technical assistance they received strengthened their capacity to implement solutions more quickly and demonstrate results.

Benefit multiple stakeholders
Addressing the varied needs and priorities of patients/caregivers, health providers and policy makers garnered more champions and visibility, resulting in more expansive reach.

Don't reinvent the wheel
Leveraging emerging technology, existing educational platforms and testing resources from other countries accelerated innovation and progress in implementation.

Recommendations

We have developed actionable recommendations to support countries in their efforts to diagnose a rare disease. Although ending the diagnostic odyssey requires a collective effort, these recommendations address distinct challenges each stakeholder group is equipped to solve.

Generate greater awareness

Build a stronger knowledge base on the challenges of the diagnostic odyssey and solutions to accelerate rare disease diagnosis.

Patient advocacy groups
Elevate success stories — including the case studies included in this report — and build networks through various channels (e.g., conferences, webinars, newsletters, social media) to strengthen advocacy for more innovation and investment in accessible technology and diagnosis.

Providers
Become more knowledgeable about rare disease in the communities they serve, "think rare," and take advantage of the resources available to promptly identify signs and symptoms for early investigation and referral. "Think care" for children and families with and without a diagnosis, and "think again." Conditions evolve, as do technology and knowledge — persistence in searching for a diagnosis is important.

Policymakers
Consult with families, patient advocacy groups and medical societies to better understand the rare disease landscape and how to strengthen policies and support education that would lead to faster diagnosis.

Promote innovation

Foster a collaborative and "connected" environment — within and across countries — to stimulate innovations in diagnosis that integrate families' lived experience, consider cultural context, and leverage advances in science and technology.

Patient advocacy groups
Encourage innovators to design new solutions to the diagnostic odyssey and collaborate with them to provide the patient/family insights needed to ensure that families will use what they create.

Providers
Take advantage of emerging and available technologies (e.g., AI tools, WhatsApp, telehealth) to promote equitable access to diagnosis. Support knowledge sharing through diagnostic networks.

Policymakers
Engage diverse stakeholders (e.g., patients/families, advocacy groups, health providers, innovators, academia) to provide input on rare disease policies, including guidance on setting and meeting targets for faster diagnosis. Ensure the right balance between safeguarding the privacy of individual and organizational data and making data available to enhance the value of technology in diagnosis.

Drive greater investment in diagnosis

Build a compelling investment case for rare disease diagnosis by quantifying the impact of early diagnosis.

Patient advocacy groups
Highlight available data on the positive economic and societal impact of shortening the time to diagnosis when communicating with funders and policymakers, and strive to generate new data through surveys on the negative patient/family experience of the protracted wait for a diagnosis.

Providers
Collaborate with peers and health institutions to collect data from patient records on health care utilization to strengthen the evidence base for greater investment in faster diagnosis.

Policymakers
Engage economists to quantify the value of diagnosis — and accept relevant data from similar countries — to inform investment decisions in rare disease diagnosis.

CONCLUSION

The Global Commission to End the Diagnostic Odyssey for Children with a Rare Disease is committed to elevating the right of all children to a timely, accurate diagnosis. ***From Idea to Action*** reintroduces our ***Framework for Action in Accelerating Diagnosis***, developed in 2024, and its three pillars: 1) Patient and Family Empowerment; 2) Equipping Providers; and 3) Policy. We hope that the 10 exemplars featured in this report bring the framework to life and inspire rare disease advocates to consider similar innovations as they strive to speed up diagnosis of rare diseases in their country.

The report highlights the determination and achievements of rare disease champions from around the world because we believe that learning from peers — regardless of country context — is one of the most effective ways to end the diagnostic odyssey.

As a resource compendium, ***From Idea to Action*** provides practical guidance on how to design and implement solutions that can lead to significant progress on the path to diagnosis. The ingredients for success that we have identified are cross-cutting. Elevating the patient voice, reflecting the cultural environment, engaging the government early, collaborating across borders — to name a few — are not bound by geography or resources.

Likewise, the recommendations we have outlined for patient advocacy groups, health providers and policymakers will hopefully resonate in high-income countries as well as low- and middle-income ones where greater attention to rare disease is needed most.

Answering families' questions about their child's health should be an urgent priority for all countries. Through grit, deep understanding of communities, multisector collaboration and adoption of technology and scientific advances, the potential for faster and more equitable diagnosis is enormous.

We are excited to promote these stories of impact and are optimistic about the power of advocates and innovators to end the diagnostic odyssey for children with a rare disease.

Please contact us with other examples that could be valuable for the rare disease community or if you would like more information about our work: info@globalrarediseasecommission.com. We look forward to hearing from you.

ACKNOWLEDGMENTS

The Global Commission is grateful to the many people who contributed their time and expertise to develop this report. We would like to thank the individuals leading the initiatives featured in the 10 case studies for being so generous with their time: Sue Baker — Program Director, Rare Care Centre, Perth Children's Hospital, Co-founder of Global Nursing Network Rare Diseases; Dr. Gareth Baynam — Medical Director of the Rare Care Centre and co-founder of UTOPIA; Helene Cederroth — Co-Founder, Wilhelm Foundation; Dr. Roberto Giugliani — Full Professor of Medical Genetics, Federal University of Rio Grande do Sul, Founder and Director of Casa dos Raros; Chris Higgins — Chief Executive, Rare Disorders NZ; Julián Isla — Founder of Foundation 29, Data and Artificial Intelligence Resource Manager at Microsoft; Dr. Aime Lumaka — Principal Investigator of African Rare Diseases International, Directeur du Réseau National Maladies Rares et Non-Diagnostiquées, Professor of Genetics at University of Kinshasa; Yukiko Nishimura — President NPO ASrid; Dr. Carmencita Padilla — Emeritus Professor, College of Medicine, University of the Philippines, Manila; Dr. Samuel Agyei Wiafe — Executive Director, Rare Disease Ghana Initiative & Clinical Psychologist, Ghana Health Service.

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DISCLAIMER

The Global Commission to End the Diagnostic Odyssey for Children with a Rare Disease is an association composed of a diverse group of organizations and individuals committed to shortening the time for an accurate diagnosis for all children with rare diseases. The views expressed herein are solely those of the Commission and do not represent any individual, co-chair, or member organization's views or opinions.

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Pillar 1:
Empowering Patients and Families

1

Casa dos Raros (Brazil):
Casa dos Raros, also known as the Center for Comprehensive Care and Training in Rare Diseases, opened in 2023 to provide an interconnected network of care for people living with a rare disease from throughout Brazil and Latin America. Casa dos Raros takes a multidisciplinary approach, integrating diagnostic testing, clinical expertise, innovative treatment methods, research, education, training, and patient and family support under one roof.

As of early 2024, more than 2,500 people have contacted the Center and about 1 in 10 was selected for evaluation based on a combination of symptomology, medical history, family history and previous test results. The Center reached a conclusion about these individuals' conditions in an average of 58 days. This timeline is considerably shorter than the national average of 5.4 years to receive a diagnosis and much faster than the average two-year wait for a consultation with rare disease specialists.

Read the full case study

2

DxGPT (Global):
Dx29 is an easy-to-use, free of charge digital tool that supports patients/caregivers as well as providers in reporting and analyzing symptoms and creating and sharing medical histories in the quest to reach a diagnosis. Predictive language learning models and generative AI enables users to enter a free-form description of their symptoms and health condition in their own words. The tool then proposes a list of potential rare diseases, making it possible for patients/caregivers to ask targeted questions of their providers and prompting providers to look "beyond the usual suspects" when a case proves difficult to diagnose.

Through streamlining the diagnostic process, the tool helps reduce the time and resources required to reach a diagnosis, potentially improving patient outcomes and reducing health care system costs. DxGPT is experiencing growing popularity, with more than 62,000 new users in the past year from across the world. Madrid's health care system recently integrated a branded version of DxGPT into the electronic medical record system across all primary care settings.

Read the full case study

3

Diagnostic Access Program (Ghana):
Started by the Rare Disease Ghana Initiative (RDGI) in 2019, the Diagnostic Access Program helps individuals and families receive genetic testing and the medical and social care they need throughout their diagnostic journey. RDGI's aim is to advocate for greater government investment in research, education, health care services and social support to help individuals and families affected by a rare disease — including those who are undiagnosed.

A key component of the program is training primary care physicians to identify rare disease signs and symptoms. RDGI has trained more than 50 clinicians across 12 facilities who are making referrals to the organization when they suspect a patient may have a rare disease but do not have the diagnostic capabilities to conduct genetic testing. Through collaborations with labs in Ghana and other countries, RDGI has been able to provide genetic testing at no cost to more than 200 people — helping determine a definitive diagnosis for 70% of them.

Read the full case study

Pillar 2:
Equipping Providers

4 **The Global Nursing Network for Rare Diseases (Global):**
The Global Nursing Network for Rare Diseases (GNNRD), established in 2023, is an effort to consolidate resources on rare diseases and tailor them for nurses — often the frontline provider — better equipping them to diagnose and care for people living with a rare disease. The Network aims to shorten the time to diagnosis and provide better care for children and adults throughout the patient journey.

The Network is creating a global community to facilitate cross-learning about rare diseases and educate nurses on rare disease symptomology. As of early 2024, the Network's members include more than 500 nurses from 51 countries, more than half of which are low or middle-income. Momentum is growing rapidly, with interest in establishing both regional and national versions of the Network, such as an Asia-Pacific chapter and a Brazil chapter.

Read the full case study

5 **UTOPIA (Australia and Singapore):**
In 2022, The Rare Care Centre at Perth Children's Hospital in Western Australia and KK Hospital in Singapore co-designed a digital platform to generate a semi-automated summary of a patient's condition called UTOPIA (Unlocking Treatment Options Personalized In-Time Access). Based on an individual's clinical features (phenotype), the platform deploys a wide range of artificial intelligence approaches for analysis. UTOPIA's power is the ability to help determine the care a child needs even if a diagnosis has not been confirmed — streamlining the care pathway during the lengthy diagnostic journey.

UTOPIA has cut in half the time to develop a personalized care plan by reducing the number of hours that clinicians otherwise would have to spend finding the relevant information, tailoring it to the individual child and presenting it in an easy-to-understand format. UTOPIA is being implemented for daily use in the clinical care of patients in the Rare Care Centre in Australia and approximately 180 patients, as of July 2024, have received reports that are guiding their individualized care plan.

Read the full case study

6 **African Rare Diseases Initiative (Democratic Republic of Congo):**
The African Rare Diseases Initiative (ARDI) is a new African-led effort that seeks to expand genetic testing, build a network for genomic research and advance provider training to better diagnose and manage rare diseases in Africa. Under the auspices of the National Institutes of Health in the U.S., ARDI aims to fill a major global gap in rare disease research by including previously overlooked populations in genetic studies.

While still in its early stages, ARDI started in 2023 in the Democratic Republic of Congo (DRC), a country with only three medical geneticists. The initiative is an example of how to build local capacity to advance genetic research and innovation — with potential to scale to other African countries. In its first year of operation, ARDI expanded to 11 of the 26 provinces across the DRC. More than 160 families have contacted ARDI, and about 20% qualified to receive genetic testing based on their phenotype trajectory.

Read the full case study

7 **Undiagnosed Hackathon (Global):**
The Undiagnosed Hackathon is an annual event that fosters global collaboration to diagnose genetic conditions that have eluded medical experts. Hosted by the Wilhelm Foundation in collaboration with the Chan Zuckerberg Initiative, the Hackathon brings together multidisciplinary teams — which include expert clinicians, bioinformaticians, molecular biologists, scientists, technology developers and AI specialists — to tackle some of the most intractable cases.

The patients who participate have been nominated by their doctors around the world because their conditions remain undiagnosed despite extensive efforts by their care teams to determine a diagnosis. The most recent Hackathon in 2024 included more than 120 "hackers" representing 28 countries. The individuals with undiagnosed diseases hailed from China, Democratic Republic of Congo, Ghana, India, Pakistan, Sweden, Turkey and the U.S. Forty percent of participants were diagnosed within 48 hours.

Read the full case study

Pillar 3: Policy

8 **Aotearoa's Rare Disease Strategy (New Zealand):**
Rare Disorders NZ (New Zealand) — an umbrella organization — led the charge in mobilizing a diverse group of patient advocates to provide input on the country's first rare disease strategy. The organization worked closely with the Ministry of Health and built a strong relationship with government officials to ensure that the strategy reflected the community's concerns from the patient perspective, with a focus on the marginalized Indigenous (Māori) community.

Rare Disorders NZ collaborated with the government to gather input from patients and clinical experts. They organized online focus groups with questions that prompted responses on devices that were shared in real-time to spark conversation. Rare Disorders NZ helped provide a unified voice on priority issues for the government to consider and include in the strategy. In 2023, the organization made a formal submission to the government outlining the community's demands for what the strategy should include, describing how to complete the missing sections of the proposed strategy, and recommending additional areas that the strategy should address. Aotearoa New Zealand's Rare Disorders Strategy was released in July 2024.

Read the full case study

9 **Newborn Screening Program and Rare Disease Act (Philippines):**
A group of physicians from 24 hospitals across Manila implemented a pilot program to test newborns for select rare diseases, generating valuable data on parents' willingness to have their newborn tested and the economic value of newborn screening. The data were foundational in advocating for successful passage of The Philippines Newborn Screening Act which led to national implementation of newborn screening in the public health system and screening access for all newborns.

As a result of the Act and the power of early detection, close to 300,000 babies have been spared the potential harm of a rare disease. The number of facilities offering newborn screening in the Philippines increased from 24 in 1996 to more than 7,000 in 2023 and the number of disorders being screened increased from 5 to 30.

Read the full case study

10 **NANBYO Act (Japan):**
As a result of advocacy efforts championed by patients, physicians, researchers and policymakers to recognize people living with a rare disease, Japan passed the Act on Medical Care for Patients with NANBYO (which translates to "difficult disease") in 2014. The NANBYO Act was pivotal in bringing attention to the often-hidden population of children and adults living with rare conditions. It formalized government reimbursement for their medical services and provided support for research on rare disorders, including diagnostics.

Since the Act's implementation, the number of diseases that qualify for reimbursement of associated medical expenses has grown from only five conditions in 1972 to 341 diseases in 2024 — a significant increase in the number and proportion of people living with a rare condition who now benefit financially and from more research on their disease. With greater investment in research, Japan has also stepped up its use of advanced technology in medical diagnostics — including next-generation sequencing.

Read the full case study