THE GLOBAL COMMISSION TO END THE DIAGNOSTIC ODYSSEY FOR CHILDREN WITH A RARE DISEASE

2024 FRAMEWORK FOR ACTION

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

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Letter from the Co-chairs

Today, more than 300 million people around the world live with a rare disease. About 70% of these diseases appear in childhood.[1] On average, it can take about five years to receive an accurate diagnosis of a rare disease, even in countries with the most sophisticated health systems.[2] A range of obstacles contribute to these delays, including limited awareness of rare disease, a global shortage of genetic and rare disease specialists, minimal rare disease education or training for healthcare professionals, insufficient infrastructure, and lack of appropriate testing tools—among other factors.

The impact of waiting years for a rare disease diagnosis can be devastating, often resulting in a more severe and debilitating evolution of the disease, accelerating potential co-morbidities or even mortality, delaying life-saving treatment, and causing significant socioeconomic and mental health challenges for families in the early years of a child's life.

The Global Commission to End the Diagnostic Odyssey for Children with a Rare Disease is a multidisciplinary group of rare disease advocates, researchers, physicians, and innovators from across the globe working to end this long and arduous diagnostic journey.

Our vision is a clear path to a timely, accurate diagnosis for all children.

The roadmap we developed in our first year included actionable recommendations to address barriers along the diagnostic pathway, with an emphasis on technological solutions. More recently, we have expanded our mission to harness the power of innovation broadly to accelerate the time to diagnose a rare disease for children around the world.

We are excited to share this report which introduces our newly developed framework to guide action in accelerating diagnosis and an initial set of case studies that we hope inspire further innovation in diagnosis.

Ending the diagnostic odyssey in rare disease is an ambitious undertaking—and one that cannot be accomplished by any one discipline or sector. Fortunately, there has been strong momentum in the rare disease field in recent years. Global policy changes—such as the inclusion of rare diseases within the United Nations Political Declarations on Universal Health Coverage in 2019 and 2023 and the first-ever United Nations Resolution on rare diseases released in 2021—have galvanized the rare disease community. The time is ripe for countries and health systems to transform how they help families navigate the complex search for a diagnosis and find answers more quickly.

Our members are committed to global collaboration to bring creative recommendations and learnings to the field to improve the lives of children living with a rare disease. Throughout the year, we will be working closely with the broader rare disease community to build a resource compendium and highlight additional examples of pioneering efforts to end the diagnostic odyssey. We welcome any suggestions you may have and look forward to hearing from you.

Sincerely,

The Global Commission Co-chairs

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Framework to guide action in accelerating diagnosis

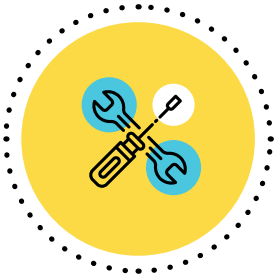
The Global Commission aims to inspire concerted action and mobilize diverse stakeholders to work collaboratively toward a shared ambition of accelerating the time it takes to diagnose a rare disease.

Our view is that the field is filled with creative examples of innovative efforts to diagnose rare diseases more quickly—from integrated care systems and rare disease education networks to advocacy campaigns—and that we have substantial collective knowledge to leverage.

We are excited to share this report which includes the first iteration of a newly developed *Framework to Guide Action in Accelerating Diagnosis for Children*. Our diverse membership has developed this framework to support global, regional, and national efforts to end the diagnostic odyssey.

The framework has **three pillars** which are foundational to 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

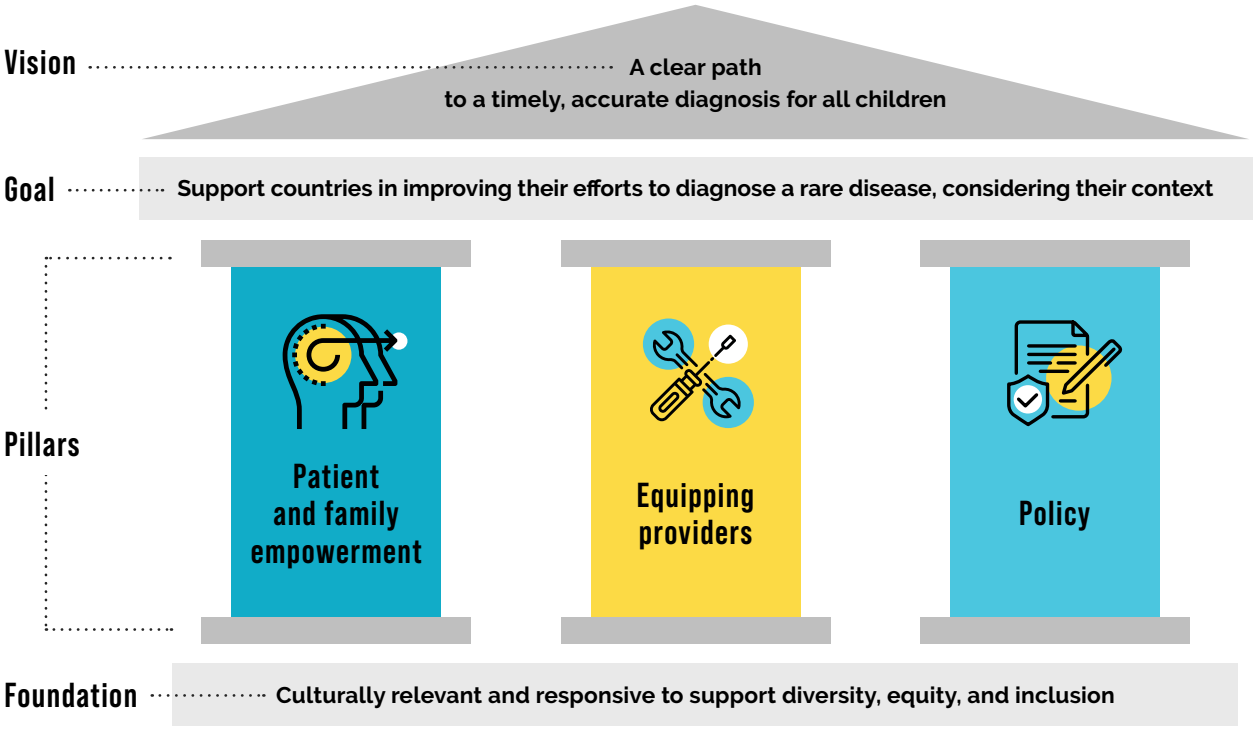
Policy:
Mobilizing advocates on national, regional, and global levels to change policies that will help accelerate and improve the quality of diagnosis for more people

To bring the framework to life, the Global Commission has developed accompanying case studies. Our intent is to showcase innovative efforts to accelerate diagnosis, highlight key learnings that can be applied to different contexts, and inspire the global health community. These case studies are the beginning of a resource compendium that will grow with additional examples of how the rare disease community is making important advances in ending the diagnostic odyssey.

The initial three case examples include: 1) a new integrated diagnostic and treatment center for rare disease in Brazil that has significantly reduced diagnosis time; 2) a community of practice for nurses to learn about rare diseases through knowledge exchange and resource sharing; and 3) a patient-led advocacy effort to influence New Zealand's first rare disorder strategy. We are grateful to the individuals leading these efforts who were generous with their time in providing such detailed information about their work, including key learnings.

Over the next year, with input and guidance from rare disease innovators, we will continue to develop case studies and grow this resource compendium, which we hope will be a valuable tool for the rare disease community. Please contact us at info@globalrarediseasecommission.com if you have an example of a successful or promising initiative to accelerate diagnosis that you recommend we turn into a case study. We welcome your feedback on this framework and how it has assisted your efforts to speed up diagnosis.

FRAMEWORK TO GUIDE ACTION IN ACCELERATING DIAGNOSIS



Our vision is that all patients and their families…

- Feel heard when speaking to health professionals about their child's symptoms
- "Consider rare" when seeking answers about their child's symptoms
- Understand the benefits of genetic counseling and genetic testing
- Are free from stigma in searching for a diagnosis
- Are viewed the same as those with non-rare conditions thanks to greater public awareness and understanding

Our vision is that all providers, regardless of where they practice…

- Are equipped to identify the warning signs of a rare disease, prompting earlier investigation and referrals
- Have access to the most up to date medical expertise, transformative tools, and creative approaches that can facilitate a faster, more accurate referral and/or diagnosis

Our global vision is…

- Rare disease is viewed as directly relevant to and associated with global policies and initiatives to improve health and advance universal health coverage, such as the Sustainable Development Goals, WHA Resolution on Diagnostics, Lancet Commission on Diagnostics, and the WHO Global Patient Safety Action Plan
- Rare disease, including the seriousness of diagnostic error or misdiagnosis, is considered a priority by UNICEF, the World Bank, and WHO

Our regional/national vision is…

- Countries recognize the value of diagnosis, including rapid and equitable access to diagnostics that considers their context
- Diagnosis of rare disease is a regional/national health priority
- Rare disease is embedded within non-communicable disease strategies and resource allocations
- Rare disease policies are culturally relevant, sustainable, and driven by health needs, and include mechanisms for holding governments accountable for implementation to ensure that people living with a rare disease are not left behind

FRAMEWORK PILLAR: Patient & Family Empowerment

CASE STUDY: *Casa dos Raros*, Brazil

A new integrated diagnostic and treatment center in Brazil that has significantly reduced the time to diagnose a rare disease.

Context

It takes an average of 5.4 years for someone with a rare disease in Brazil to finally be diagnosed after first presenting symptoms.[3] *Casa dos Raros* ("House of Rares") is a new, comprehensive rare disease center in Porto Alegre that has succeeded in diagnosing patients in an average of 58 days from first contact while the average wait time for a first consultation with a rare disease reference center affiliated with the public health system is around two years.

***Casa dos Raros* was established to accelerate diagnosis, facilitate research, develop educational materials for healthcare teams, and improve the accessibility of treatment for people living with a rare disease.** Specifically, it seeks to expand the reach of the rare disease reference centers affiliated with the public health system, which offer clinical care, lab testing, and counselling, but have long wait times and have been unable to fully meet the demand of individuals and families seeking a timely diagnosis.

Casa dos Raros, Brazil

Overview of program

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 from throughout Brazil and Latin America. The Center is a collaboration between Casa Hunter (a well-respected patient advocacy group with a good working relationship with the government) and the Genetics for All Institute (a non-governmental medical and scientific organization that develops courses and trainings on rare disease for healthcare professionals, patients and families, and others).

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. The Center opened its doors in February 2023 and has established agreements with government health services, private insurance companies, private companies, and donors to help finance its operations.

Anyone seeking a diagnosis can contact the Center through its website, email, telephone, WhatsApp, or make an in-person visit. *Casa dos Raros* is active on social media and has become a reference for many different health services and agencies throughout Brazil and Latin America.

Because *Casa dos Raros* is an independent, not-for-profit institution, the staff have the flexibility to triage individuals on the waiting list and expedite consultations for those with more urgent needs. A clinical group analyzes every request and usually follows up with a video consultation for the medical team to learn more about the patient's medical history, family history, and previous tests.

In most cases, patients visit *Casa dos Raros* after the video call for a thorough evaluation by a multidisciplinary team. Any additional procedures, such as MRIs or blood tests, are performed during this time to optimize the visit. After the clinical evaluation is completed and once the test results are received, the team works to determine a diagnosis – a process that takes a few weeks rather than a few years.

The team then designs a disease management plan and, whenever possible, patients are transferred to a care center close to their home. To ensure appropriate care in these situations, *Casa dos Raros* trains the local healthcare team to meet the patient's needs. The staff will either host a training at the Center or travel to consult with the patient's local medical team—from pharmacists to nurses—about the treatment protocol. The patient still has the option to receive treatment at *Casa dos Raros*, however, routine care, such as infusions or physical therapy, is typically delivered by the local healthcare team and the patient visits *Casa dos Raros* for periodic follow-up evaluations.

Casa dos Raros' staffing model is a mix of permanent staff and contracted specialists. There are approximately 50 multidisciplinary team members, including clinical geneticists, speech therapists, physical therapists, dentists, nurses, pharmacists, social workers, as well as laboratory, clinical research, and administrative staff. Certain specialists, such as neurologists and cardiologists, are brought on to assess patients as needed.

Casa dos Raros is open to anyone who is suspected of having a rare disease, genetic or non-genetic, in addition to individuals who already have a diagnosis and are seeking a second opinion or an updated management plan. Among patients who are seeking care, *Casa dos Raros* prioritizes historically medically underserved populations when possible.

Patients do not pay for the services they receive from *Casa dos Raros*. The Center has a partnership with the biggest genomics laboratory in Latin America (Dasa Genomics), which has a facility within *Casa dos Raros* and provides genetic tests for *Casa dos Raros*' patients (at no cost to patients). In addition, the Center has a contract with the state of Rio Grande do Sul to evaluate 25 patients from the state per month.

A combination of payments from neighbouring states, insurance providers, clinical trial sponsors and private laboratories ordering diagnostic tests, and private and public donations covers the costs of all other patients diagnosed and treated at *Casa dos Raros*.

Results

Casa dos Raros has significantly reduced the time to diagnose a patient with a rare disease to an average of 58 days from first contact with the Center. This timeline is considerably shorter than the national average of 5.4 years for patients to receive a diagnosis and much faster than the average two-year wait for a first consultation with rare disease specialists.[3]

In the year since its doors opened in 2023, over 2,500 people have contacted *Casa dos Raros* and the Center has evaluated more than 250 patients. *Casa dos Raros* has a team that evaluates and replies to all requests. Based on the initial assessment of cases presented, the Center selects the most urgent ones (usually children without a diagnosis who have no access to specialized services and may benefit from disease-modifying therapies). About 1 in every 10 patients has been selected for evaluation.[4]

In addition to the health and quality of life benefits of a faster and more accurate diagnosis, *Casa dos Raros* has demonstrated financial benefits as well. **The Center has estimated that the cost of its comprehensive, integrated, and intensive model is at least the same—but probably less—compared to the costs accrued during the 5+-year diagnostic odyssey.[4]**

Learnings

Casa dos Raros' model provides valuable insights regarding the importance of partnerships, innovative funding models, multidisciplinary teams, and passionate staff to accelerate diagnosis.

- **Partnerships are critical to amplify impact:** *Casa dos Raros* has forged partnerships with laboratories, companies, academic centers, and patient advocacy groups that each contribute to the success of the initiative. The collaborations with laboratories have provided state-of-the-art diagnostic capabilities at a fraction of the cost as well as access to advanced treatments at no cost to the patient. The partnership with Casa Hunter, a patient advocacy group, has enhanced the Center's ability to promote its services within the community and receive feedback to help with its programming. The partnership with the Genetics for All Institute has helped expand the reach of *Casa dos Raros*' training and research programs.
- **Innovative funding models are required to support sustainability:** The fact that *Casa dos Raros*' consultation, testing, and treatment services are free has helped attract patients from throughout Brazil and neighboring countries. *Casa dos Raros* supports its operations by soliciting donations from both public and private sources, corporate partnerships, fundraising campaigns, and funds generated through courses for healthcare professionals, sponsored clinical research, provision of laboratory services, and other convenings. The diversity of revenue streams from various local sources facilitates the sustainability of the Center as a valuable institution in the community.
- **A multidisciplinary approach promotes comprehensive care:** *Casa dos Raros*' multidisciplinary model ensures that patients have access to medical, psychological, and logistical support throughout the diagnosis process and beyond—critical to support patients and families who have been searching for a diagnosis.
- **Passionate and motivated staff drive excellence in care:** In large part, the success of *Casa dos Raros* is attributed to its dedicated and specialized staff. In addition to the staff's expertise and specialization in rare diseases, they are passionate about the work they do. Patients receive expert, compassionate care throughout their diagnostic journey and beyond.
- **Scalability is a challenge:** The largest challenge *Casa dos Raros* now faces is its ability to scale across Brazil. Expanding services to reach more patients, setting up new facilities and replicating sustainable funding models to maintain them, and hiring additional staff require significant financial resources, including upfront capital investments. While the state government of Rio Grande do Sul as well as the local community have shown strong support, funding agreements in other regions and with the federal government are currently under negotiation and are not yet secured.

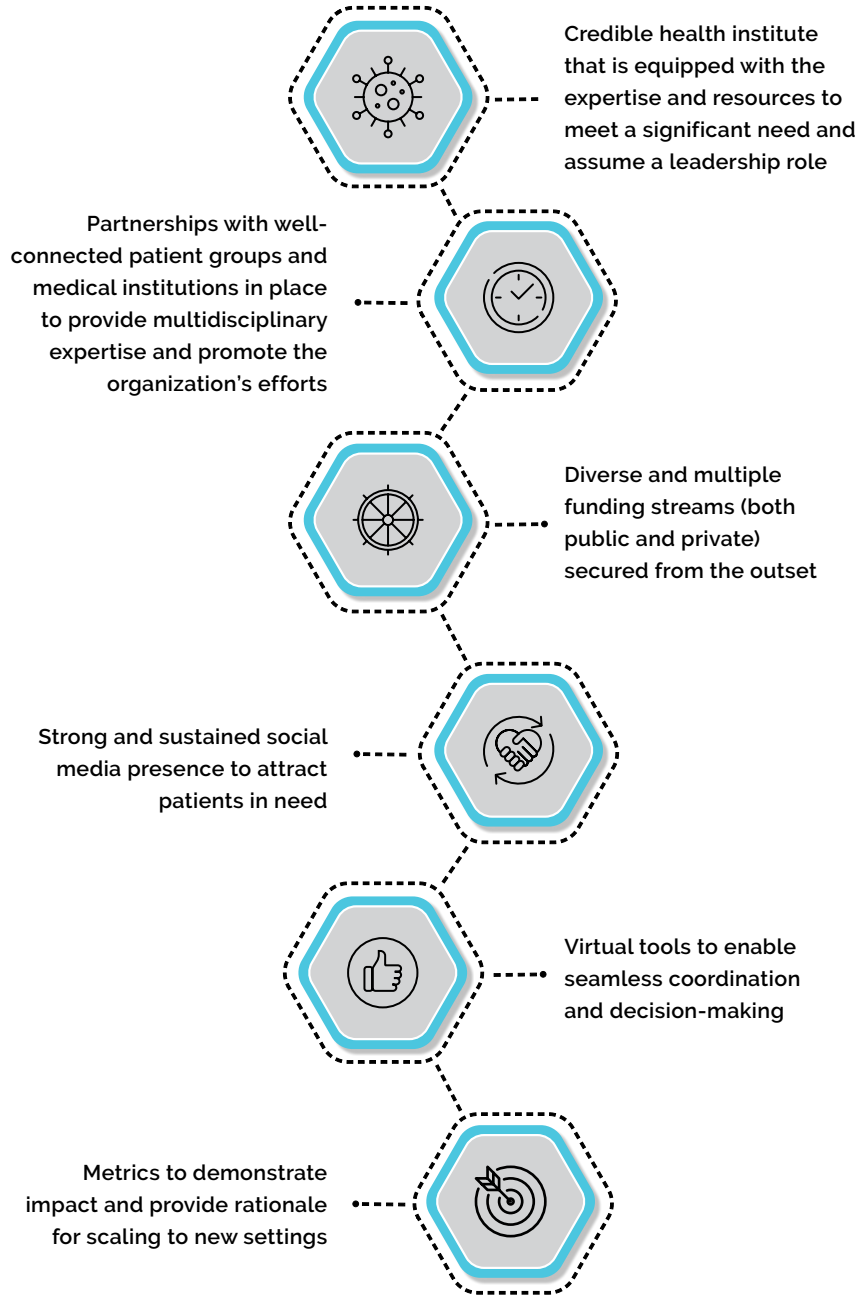
Goals & next steps

Casa dos Raros plans to replicate its integrated care model by establishing a network of similar centers throughout Brazil, with the ultimate goal of having one center in each of the country's five regions. Immediate plans include replicating the current model in São Paulo, which will be a hospital and the first one in the region dedicated entirely to rare diseases.

Casa dos Raros also aims to strengthen its research and training capabilities by developing a patient registry, a biobank of samples from patients with rare diseases, a specific program for undiagnosed diseases that will provide expert opinions and free diagnostic testing for patients who were not able to receive a diagnosis from other reference centers, and an education program with courses on rare diseases. To help execute these plans, *Casa dos Raros* hopes to secure partnerships with academic and research institutions from Brazil and abroad.

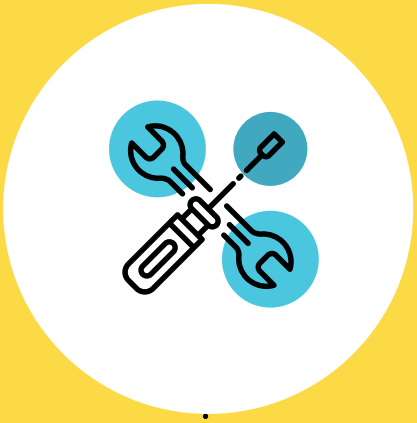
Based on its success in just one year, *Casa dos Raros* has quickly become a model for accelerating the time to diagnosis and providing comprehensive care for those with a rare disease.

Success factors & considerations when developing a similar model



FRAMEWORK PILLAR: Equipping Providers

CASE STUDY: The Global Nursing Network for Rare Diseases



A community of practice for nurses from around the world to learn about rare diseases through knowledge exchange and resource sharing.

Context

Nurses make up approximately half the global healthcare workforce. They are often the first, if not the only, healthcare professional whom patients encounter—making them an essential resource when searching for a diagnosis.

Nevertheless, nurses are not always actively involved in efforts to accelerate diagnosis for people living with a rare disease. For example, educational resources for nurses tend to focus narrowly on disease presentation, diagnostic pathways for specific rare diseases, or specialized areas (such as genomics), rather than support nurses to develop broader expertise in the complex rare disease field.

The Global Nursing Network for Rare Diseases

Overview of program

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, better equipping them to diagnose and care for people living with a rare disease.

The brainchild of the Clinical Centre of Expertise for Rare and Undiagnosed Diseases at Perth Children's Hospital (Rare Care Centre) in Australia, the Network is creating a global community to facilitate cross-learning about rare diseases and educate nurses on rare disease symptomology. The Network aims to shorten the time to diagnosis and provide better care for children and adults throughout the patient journey.

In developing the Network, the team explored models of care for people living with a rare disease and the role of nurses, and benchmarked essential elements across the patient journey—including screening, care coordination, psychosocial support, ongoing management, and access to clinical trials and treatment.

These elements underpinned a framework introduced at the "Nursing Roundtable," held in Singapore in March 2023. The event, co-hosted by the Rare Care Centre, SingHealth Duke-NUS Genomic Medicine Centre (an academic medical center), and Curtin Singapore (a public research university), included people living with a rare disease, their families, and 33 nurses from 25 countries across various regions. The input the Centre received laid the foundation for the GNNRD, establishing a community of practice that strives to be equitable and scalable.

Global Nursing Network's objectives include:

Connect and increase global collaboration among nurses working with people affected by rare or undiagnosed diseases.

Provide nurses with resources to promote and implement comprehensive care for people living with rare or undiagnosed diseases.

Inform and influence the development of nursing education resources to upskill and raise awareness of rare and undiagnosed diseases across the nursing workforce.

Strategically influence local, national, global policy and advocacy platforms by bringing nurses to the table of high-level organizations to advocate for people living with rare and undiagnosed diseases.

Guide nurses in assisting families to navigate the clinical trial space.

Connect and share expertise with other regional and disease-specific networks.

The Network is virtual, and nurses are connected through a website, enabling them as well as nursing students to access educational resources (e.g., webinars, learning modules, rare disease country recommendations), 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.

Results

Now at its one-year mark, the Network's members include 280 nurses from almost 40 countries, more than half of which are low or middle-income.[9] 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.

While the Network is in its infancy, it has already had a notable impact in various countries around the world:

- The Dean of the Faculty of Nursing at the University of Colombo, Sri Lanka, who is a Network member, included a genetics module in the Nursing Bachelor of Science program offered by the University. With 100 nursing students starting the degree each year, adding this content to the curriculum will significantly augment future nurses' knowledge of rare and undiagnosed diseases.
- The Network connected the Universitas Gadjah Mada Academic Hospital in Indonesia with the National Taiwan University Hospital to undertake a benchmarking exercise to support Indonesia's newly developed clinical genetics service. A member of the GNNRD also contributed technical expertise to help establish the Indonesian hospital's genetics program and biobank, which will support the government's Biomedical & Genome Science Initiative to enhance precision medicine in the country.
- Thanks to a Network member, the GNNRD has been included in the refreshed Wales Rare Disease Action Plan (2022-2026) as a partner to develop education and training for nurses, building their knowledge and capacity related to rare and undiagnosed diseases.

In addition, the Network has partnered with Medics4Rare (a charitable organization that creates resources to educate doctors and medical students about rare disease) to develop e-learning modules specifically for nurses and nursing students, helping them identify the signs of rare diseases in children.

The Network is also planning to create resources for teachers, such as animated videos, to help guide conversations with children who have a rare disease and their families. The goal is to make these materials culturally relevant and translate them into multiple languages so they can be used across a variety of regions.

Learnings

The success of the Global Nursing Network for Rare Disease, although in its early days, underscores the power of global collaboration and the value of partnerships, co-creating solutions, and creativity in testing new ways to engage health professionals to deepen knowledge of rare disease.

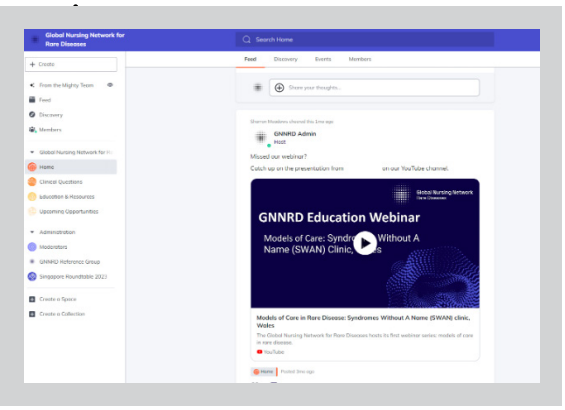
- **Partnerships are critical to establishing credibility:** A key part of forming the Network was leveraging relationships with global rare disease leaders who contributed their expertise—providing credibility, which was essential to secure financial support—and promoted the Network through their channels.
- **Co-design with target audience:** The Rare Care Centre was intentional and methodical about collaborating closely with nurses to design the GNNRD. For example, the Centre organized a series of meetings and workshops to gather nurses' input and gain buy-in for this project—well before launch. The nurses involved were asked to validate findings from the workshops to ensure that the Centre accurately captured their needs, highlighted the most significant accomplishments to funders, and outlined agreed-upon milestones for the upcoming year.

  In addition, the Network partnered with academic institutes to build connections with nursing education specialists and advise on the best platforms and content to use when developing educational resources for nurses. Likewise, teachers will be brought in for a workshop to help craft the animated video to ensure it reflects their needs across the globe, spanning cultural and linguistic diversity.
- **Creative formats of engagement are key to sustaining momentum:** The Network created a robust plan to ensure that nurses would stay engaged once the website went live. For example, it organized webinars and workshops to attract nurses from different geographies, provided travel scholarships for in-person meetings, and added functionality to make the website interactive, including hosting a discussion forum and spotlighting different Network members each month.
- **It's important to recognize when to bring in help:** Understanding the limitations within the Network's own team helped set reasonable expectations for what it could achieve by itself. Ultimately, the Centre determined which services should be subcontracted and hired the technical support required for website development and management.

Goals & next steps

Immediate goals for the GNNRD are to build chapters in new geographies to establish national/regional networks and links to specialist disease groups to create a repository of resources specific to each disease. Nurses interested in leadership roles to represent their country or region, or a specific disease, can contact hello@gnnrd.org

The near-term goals are to expand the Network to one thousand members by the end of 2024; establish an advisory group of patients with lived experience; refine a strategic framework to inform the Network's future direction, including clear goals and deliverables; and add more translation options for greater linguistic diversity.



Success factors & considerations when developing a similar model

- Clearly identified need, ideally as part of a rare disease needs assessment
- Respected organization willing to invest time and resources to meet that need and assume governance
- Existing resources—national, regional, global—that can be leveraged to avoid reinventing the wheel
- Partnerships already in place to provide expertise and credibility
- Mechanisms in place for involving the beneficiaries in co-creating the solution
- Channels to promote the initiative and ensure it reaches the target audience
- Evaluation metrics and plan for course corrections in place

FRAMEWORK PILLAR: Policy

CASE STUDY: Informing New Zealand's first rare disorder strategy

An initiative to mobilize patient advocates—including from indigenous (Māori) communities—to provide input on the country's first rare disorder strategy.

Context

New Zealand has a robust community of patient advocacy groups supporting people living with a rare disorder, however, the country does not have a formal rare disorder strategy. The New Zealand government has not prioritized the needs of those with rare disorders—including timely diagnosis—or ensured a coordinated approach to supporting people with a rare disorder, raising concerns about adverse health outcomes and restricted access to lifesaving treatments that have been approved for similar patients in neighboring Australia.

As part of a major review of the country's drug purchasing agency, a fortuitous recommendation emerged in 2022 to develop a Rare Disorder Strategy—a longstanding goal of the rare disorder community. The National government tapped the Ministry of Health (MoH) to develop the new strategy.

Seizing an unprecedented opportunity for advocacy, Rare Disorders NZ (New Zealand)—an umbrella organization—led the charge in mobilizing a diverse group of patient groups to provide input on the strategy from the patient perspective, including from the marginalized Indigenous (Māori) community.

Informing New Zealand's first rare disorder strategy

Background

Rare Disorders NZ is a well-established organization that publishes a bi-annual Voice of Rare Disorders Survey, highlighting challenges the rare disease community faces.[5] The 2023 survey results 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 more than 10 years.

This situation did not improve between 2019 and 2021. There were multiple contributors to these delays, including lack of rare disease data in electronic health record systems—which can lead to challenges accessing rare disease services—insufficient communication between healthcare professionals and specialists, and a limited emphasis on rare disorder research.

Over the past few years, Rare Disorders NZ has been able to significantly increase the number of people participating in this survey, from 300 to 1100—including 140 Māori—excellent representation from this population.

With the announcement of a forthcoming Rare Disorder Strategy, Rare Disorders NZ set out to work closely with the MoH and built a strong relationship to ensure that the strategy reflected the community's concerns. The organization facilitated the government team's access to key information about rare disorders to inform the strategy, highlighting, for example, the extensive wait times for diagnosis.

Rare Disorders NZ also collaborated with the government to gather input from patients and clinical experts to inform the strategy. They organized online focus groups with questions that prompted responses on devices that were shared in real-time to spark conversation. The government also interviewed 20 families of Māori descent, which was valuable in designing a strategy that must address the needs of the most underserved population in the country.

When the draft strategy was shared in late 2023, patient advocates were surprised that there were several sections yet to be completed even though they had provided specific recommendations. In response, Rare Disorders NZ made a formal submission outlining the community's demands for what the strategy should include, describing how to complete the missing sections, and raising additional areas that the strategy should address.

Rare Disorders NZ helped provide a unified voice on priority issues for the government to consider and include in the strategy. Together with other patient advocacy groups, they stated their demands more openly than before and made their expectations clear—especially for an accurate and rapid diagnosis. The group's advocacy was noticed with admiration within and beyond the rare disorder community.

Rare Disorders NZ published a white paper on Rare Disease Day 2024 that included the results from the 2023 Voice of Rare Disorders Survey as well as the organization's four advocacy priorities for the forthcoming national strategy:[5]

1. **Develop a Rare and Undiagnosed Disorders Centre of Expertise** to provide early and accurate diagnosis, evidence-based best practices for both diagnosed and undiagnosed patients, and family support through coordination experts who help navigate the health system.
2. **Ensure patients can access essential medications** for rare diseases.
3. **Ensure data on rare disorders is incorporated in the electronic health system** to expedite diagnosis.
4. **Implement the Rare Disorders Strategy** in partnership with Rare Disorders NZ to ensure it is accepted by the rare disorder community and implemented.

The same clinical experts whom the government had consulted when conducting initial interviews publicly supported these demands, enhancing their credibility and, hopefully, encouraging the government to meet them. The white paper also highlighted how the country's current systems suffered from institutional racism and must do better when engaging with Māori communities to avoid a tokenistic way of addressing health concerns.

As of April 2024, a revised strategy has not been released publicly, but the rare disease advocacy community is hopeful that the government will incorporate its demands into the final strategy.

Learnings

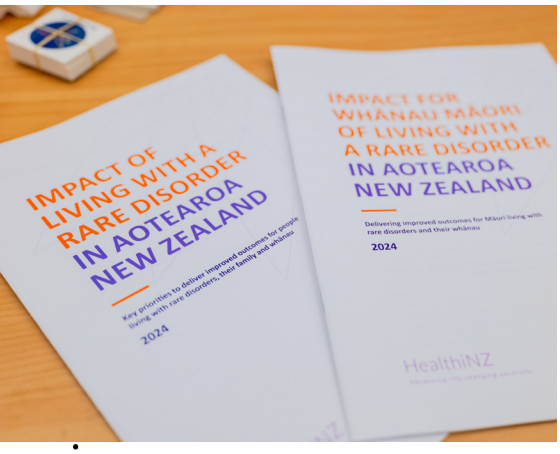
Rare Disorders NZ's perseverance and successful mobilization of patient advocates—including those from underserved communities—highlight the value of uniting behind a shared vision to advocate effectively with government on behalf of all people living with a rare disorder.

- **Be bold and push to get your voice heard:** Rare Disorders NZ realized that it was not being invited to co-create the country's rare disorder strategy (as had happened in Australia) and, therefore, should not assume that the government would reflect the patient perspective. The organization was effective in coordinating multiple opportunities to mobilize rare disease advocates who provided their input on the forthcoming strategy.
- **Elevate community voices:** Rare Disorders NZ was contacted by the government to ensure that the rare disorder strategy it was developing truly served the community. First-hand data from people living with a rare disorder was the best source to elevate their experiences and needs. For example, patients reported that they were being denied the basic right to access disability services because they had not received a definitive diagnosis—a compelling argument to accelerate diagnosis.
- **Start with a focus on the most underserved:** Rare Disorders NZ felt strongly that it should underscore the needs of the indigenous population, which are often neglected, and place these front and center to advance equity. Ensuring that the government understood the challenges of rare disorders among the Māori was an important initial step toward making progress for all New Zealanders with a rare disorder.

- **Get the clinical community on board:** Given that physicians are highly influential with the government, it was important to join forces with the clinical community. Rare Disorders NZ briefed healthcare professionals on patients' demands for what should be included in the strategy. The fact that the clinical community ultimately endorsed Rare Disorders NZ's submissions to the government was a win.
- **Recognize the value of advocacy – even if it may not lead to success:** According to Rare Disorders NZ, even if the Rare Disorder strategy does not reflect everything patients are seeking, mobilizing and uniting the advocacy community —patients and clinicians— has been critical in raising awareness of rare disorders.

Goals & next steps

New Zealand's Rare Disorder Strategy is expected to be released in 2024. The general sentiment within the rare disorder community is that while the strategy may not be perfect, it is an important milestone to leverage and build upon to improve the lives of people living with a rare disorder.



Success factors & considerations when developing a similar model

- Working relationship between rare disease patient advocates and the national government
- History of patient advocates influencing health policies
- Feasibility to unite disparate rare disorder patient groups behind a joint cause
- Identified patient advocacy group that has the credibility, support, and trust to lead an umbrella effort
- Channels in place to encourage diverse voices to contribute their perspectives
- Commitment and ability to actively include underrepresented groups to ensure equitable solutions
- Strong relationships with the clinical community and alignment on relevant policy issues

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.

We recognize that tackling a challenge as complex and multi-faceted as rare disease diagnosis can be daunting. Our Framework for Action in Accelerating Diagnosis and its Three Pillars (Patient and Family Empowerment, Equipping Providers, and Policy) is intended to support national, regional, and global efforts to help families find resolution more quickly as they search for answers about their child's health. The stakes are high because we know that in many cases a faster diagnosis can lead to life-saving treatment as well as peace of mind.

To advance the field, we believe it is critical to learn from peers about how they are making strides in diagnosis for rare disease. The three case studies included in this report illustrate the importance of perseverance and ingenuity to make progress. This report is our first attempt to highlight important work and valuable learnings that we hope will inspire others to consider similar initiatives. We plan to capture more of these compelling stories over the coming months.

Moving forward, we will be developing additional case studies as part of a resource compendium. We encourage you to contact us with your recommendations of examples to potentially turn into case studies as well as other tools for diagnosis that could be useful for the rare disease community. And we welcome your feedback on the framework to make sure it is as valuable as possible. Please contact us at: info@globalrarediseasecommission.com

We look forward to collaborating with you to end the diagnostic odyssey for children with a rare disease.

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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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