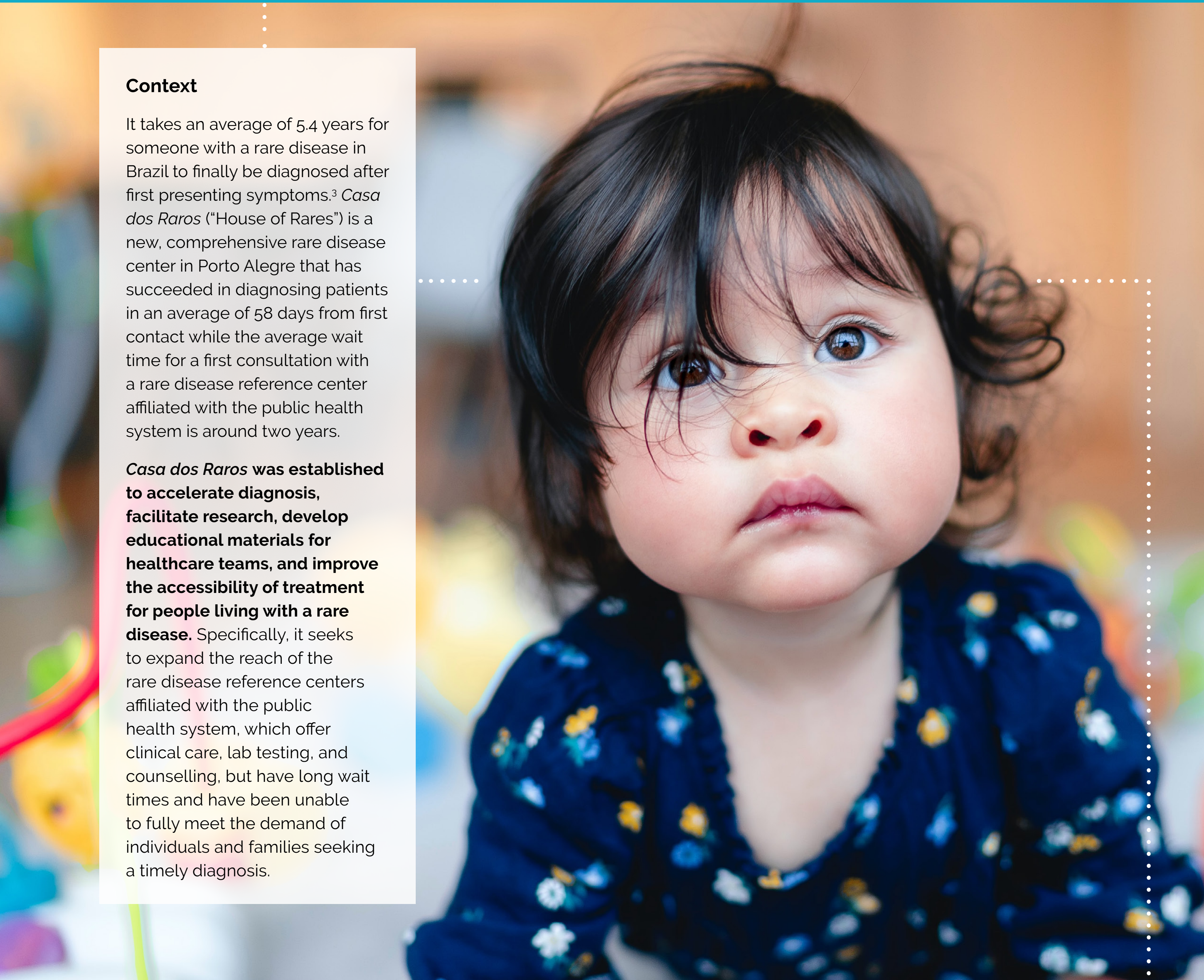
FRAMEWORK PILLAR: Patient & Family Empowerment
CASE STUDY: *Casa dos Raros*, Brazil

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

Credible health institute that is equipped with the expertise and resources to meet a significant need and assume a leadership role

Partnerships with well-connected patient groups and medical institutions in place to provide multidisciplinary expertise and promote the organization's efforts

Diverse and multiple funding streams (both public and private) secured from the outset

Strong and sustained social media presence to attract patients in need

Virtual tools to enable seamless coordination and decision-making

Metrics to demonstrate impact and provide rationale for scaling to new settings