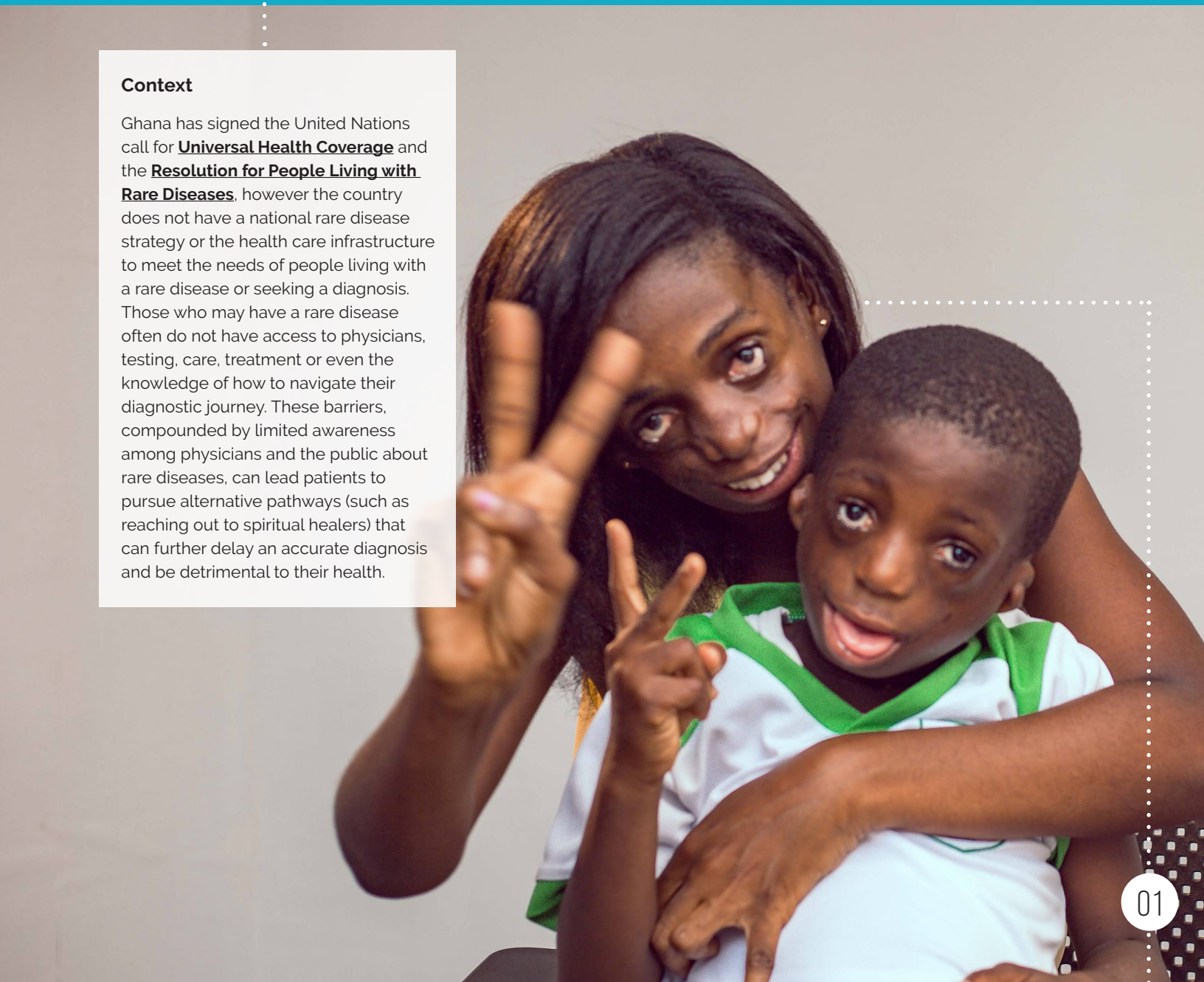
CASE STUDY: Rare Disease Ghana Initiative (Ghana)
PILLAR(S): 1 (Patient and family empowerment)

Increasing access to diagnosis and care in Ghana

Context

Ghana has signed the United Nations call for **Universal Health Coverage** and the **Resolution for People Living with Rare Diseases**, however the country does not have a national rare disease strategy or the health care infrastructure to meet the needs of people living with a rare disease or seeking a diagnosis. Those who may have a rare disease often do not have access to physicians, testing, care, treatment or even the knowledge of how to navigate their diagnostic journey. These barriers, compounded by limited awareness among physicians and the public about rare diseases, can lead patients to pursue alternative pathways (such as reaching out to spiritual healers) that can further delay an accurate diagnosis and be detrimental to their health.



Rare Disease Ghana Initiative

Overview of program

The Diagnostic Access Program — a five-year old effort in Ghana — is demonstrating that people with a rare disease can receive a timely diagnosis even in countries with extremely limited resources. The **Rare Disease Ghana Initiative (RDGI)** is a national nonprofit organization that launched the Diagnostic Access Program in 2019 to help individuals and families get a diagnosis and receive the medical and social care they need throughout their journey.

RDGI was founded by Dr. Samuel Wiafe, a clinical psychologist, who saw first-hand the frustration and trauma families experience when searching for a diagnosis in Ghana. RDGI's aim is to advocate for greater government investment in health care services, research, education 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 health care professionals to identify rare disease signs and symptoms. RDGI has reached more than 500 health care professionals and trained over 50 clinicians through webinars, workshops, symposiums and other collaborative educational programs, including training programs conducted by **FYMCA Medicals**, a non-profit organization that provides education about rare disease to health providers globally.

RDGI works with clinicians across 15 health care facilities who make 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 (20% of the results were inconclusive and the remaining 10% were negative). Through this initiative, more than 100 **different rare diseases** have been reported.

RDGI is also involved with the **iHope Genetic Health program** — a philanthropic partnership between Illumina and Genetic Alliance to make whole-genome sequencing accessible to children across the globe with rare and undiagnosed genetic diseases. The test findings have highlighted that genetic testing is critical to supplement clinicians' knowledge and experience.

Beyond aiming to provide a diagnosis, the Diagnostic Access Program focuses on meeting patients' broader needs. The team conducts a **needs assessment** for everyone referred to identify their highest priority concerns — which may be financial or psychosocial — and develops an individualized roadmap. A care coordination specialist is paired with every patient to help overcome the challenges in navigating care and may accompany patients to appointments. Nurses, psychologists, social workers and genetic counselors are all part of the care coordination team.

As part of the assessment process, the team also asks a series of questions about health-seeking behaviors: How long has it taken you to get a diagnosis? How did you try to get a diagnosis? How many clinicians did you consult? Were any spiritual healers involved in your journey? The answers inform each patient's care and also generate evidence that will support RDGI's advocacy for greater government investment in people living with a rare disease. RDGI is actively engaging policymakers to develop a comprehensive plan for rare diseases in Ghana.

RDGI is an entirely volunteer run and led organization. It currently has more than 15 active volunteers, and donations and sponsorships cover operational costs, such as stipends for care coordinators' transportation expenses and office space. The goal is to evolve the Diagnostic Access Program — which is a proof of concept — into a sustainable model by convincing the government of its value in providing genetic testing to diagnose people living with a rare disease.

Results

Since 2019, RDGI has provided genetic testing for more than 200 patients, with over 70% receiving a definitive diagnosis. In Q1 of 2024 alone, the organization received over 50 requests for genetic testing. The organization has demonstrated that even in a country with very limited health resources and minimal genetic testing capacity, it is possible to meet the needs of individuals and families searching for a diagnosis.

Learnings

- **Training primary care physicians is an effective path to referral:** RDGI's training efforts have succeeded in increasing awareness and understanding of rare diseases. Primary care physicians are making appropriate referrals — more than 250 as of early 2024 — and see the value of genetic testing, especially because a high proportion of test findings have showed different results than what clinicians suspected.
- **Keep the patient at the core:** RDGI understands that patients must be treated as partners in their care, particularly important in patient/family education efforts during a time of uncertainty. The needs assessment process has been essential in identifying and addressing patients' diverse barriers to diagnosis and care.
- **Be sensitive to health seeking behaviors:** Many families in Ghana turn to alternative pathways for their healthcare needs, such as consulting spiritual healers. As a Ghanaian organization, RDGI knew that it was important to understand where people go to receive their care and developed respectful questions that reflect the country's cultural context as part of the needs assessment.
- **Collaborate with the right partners to build credibility:** To ensure that the organization is recognized as a credible organization — especially given its advocacy goals — the team has built partnerships with prominent global rare disease organizations. RDGI is currently a member of several alliances, such as Rare Disease International, International Rare Disease Research Consortium and Undiagnosed Disease Network International. Membership has enabled the sharing of knowledge and expertise to support RDGI's efforts to improve the wellbeing and quality of life of people living with rare diseases in Ghana and Africa.

Goals & next steps

Looking ahead, the team aims to:

- **Continue its advocacy:** RDGI is on a mission to encourage the government to develop a national rare disease strategy; establish a center of excellence for rare disease; support education of health care providers on rare disease; step up surveillance and monitoring of rare disease; and provide adequate genetic testing in the country.
- **Publish its findings:** A near-term goal is writing and publishing a paper on the impact of the Diagnostic Access Program in improving the quality of life for people living with a rare disease. The paper will focus on RDGI's genetic test findings and highlight the value of patient needs assessments and care coordination.
- **Create a more sustainable funding model:** An urgent goal is to secure sustainable funding – RDGI acknowledges that dependence on free lab services from outside the country and volunteer staff is not sustainable. RDGI is collecting data to make the case for investing in rare disease resources beyond treatment.

Success factors & considerations