

iHope Insights:

An iHope Genetic Health Newsletter

iHope Genetic Health
 March 2025



500+ Families and Counting

In its first operational year as a program of Genetic Alliance, iHope successfully reached a major milestone: providing genomic testing to more than 500 families! This achievement was made possible through the collaboration of our incredible iHope network, which includes three top-tier laboratories and 15 clinical sites spanning eight countries. As testing results continue to roll in, our data so far support a diagnostic yield of approximately 45%.

Building on the momentum of 2024, we are excited to continue to expand iHope's reach in 2025. Our goal is to broaden access to genetic health solutions by increasing the number of participating laboratories, clinical sites, and healthcare professionals, and doubling the number of families who receive testing. To that end, we are thrilled to welcome Galatea Bio as a new iHope testing laboratory and Children's Hospital Srebrnjak in Croatia as a new clinical site. Galatea Bio will support both new and existing clinical sites, providing exome and genome sequencing for approximately 250 families in 2025. Children's Hospital Srebrnjak got to work quickly, facilitating testing for nearly 30 families in January 2025.

We are also excited to begin work on our Clinical Genetics Volunteer Corps, which will provide direct support to clinicians and patients in low- to middle-income countries.

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iHope

With gratitude,
The iHope Team



Left: The Genetics Clinic at the University of Zimbabwe facilitating a blood draw for WGS testing. Right: Junior, an 8-year-old boy seen in the Genetics Clinic who received WGS testing and subsequently received a diagnosis of Duchenne Muscular Dystrophy.

Regional Spotlight: iHope in Zimbabwe

At Rare Disorders Zimbabwe (RDZ) (formerly Children and Youth Care Zimbabwe), hope is more than just a concept—it is a tangible pathway to better health and well-being for children with rare and undiagnosed diseases. Through its collaboration with iHope as a clinical site, RDZ is pioneering the integration of genetic testing and counseling into pediatric care in Zimbabwe, ensuring that children and families with suspected genetic disease receive the support they need to navigate complex health conditions.

The First Step Toward a Better Future

For many children with rare diseases, an accurate diagnosis is the first step toward improving their quality of life. In Zimbabwe, access to genetic testing remains a significant challenge. Families often wait years—sometimes a lifetime—without answers, leading to unnecessary suffering and missed opportunities for treatment or management. As an iHope clinical site, RDZ bridges this gap by providing access to whole genome sequencing (WGS), enabling earlier diagnoses and, in some cases, life-changing interventions.

Through iHope, testing is provided at no cost to families by the laboratory at SickKids in Toronto, Canada. In the first year of this partnership, RDZ and SickKids facilitated WGS for 18 children and their families. So far, 16 of these children have received their genetic results, with a diagnosis identified in 63% of cases.

The expertise and dedication of RDZ team members play a crucial role in ensuring quality care. Senior Nursing Sister Phillipa Dingiswayo, who leads patient interactions at the genetic clinic, has specialized training in genetic counseling topics—a rare skill set in Zimbabwe. This expertise enhances patient services and strengthens her additional role as a Research Nurse in the Department of Paediatrics at the University of Zimbabwe.

The Power of Follow-Up Care

RDZ's commitment to children and families extends beyond genetic testing and diagnosis. Recognizing that a diagnosis can bring as many questions as it does answers, RDZ goes the extra mile—literally. Trudy Nyakambangwe, RDZ's founder and a parent to children with rare diseases, initiated home visits to provide emotional support, assess family well-being, offer guidance on diagnosis and management, and connect families with community resources. By stepping into the homes of families, RDZ fosters trust, continuity of care, and a deeper understanding of each family's unique needs. This hands-on approach reflects a holistic vision of care—one that sees the child within the full context of their family, home, and community.

Collaboration Across Borders and Sectors

RDZ advocates that addressing rare diseases requires a united approach, where healthcare providers, researchers, policymakers, community organizations, and families work together. The partnership between RDZ and iHope exemplifies how cross-border cooperation can drive progress in genetic medicine, but collaboration must extend beyond the medical community.

Engagement with social services is crucial to bridging gaps in care. Families of children with rare diseases often face overwhelming medical, emotional, and financial burdens, sometimes leading to family breakdown. RDZ has observed a rise in single-parent households following a rare disease diagnosis. Without proper support, families may struggle to access essential resources, underscoring the need to integrate social services, counselors, and advocacy networks into the care system.

iHope in Zimbabwe Continued

Building a Future of Comprehensive Support

To make a lasting impact, RDZ advocates for sustainable, accessible, and comprehensive infrastructure for genetic testing and counseling. Key priorities include:

- Training healthcare providers in genetics and genetic counseling to increase support, awareness, and provider capabilities.
- Integrating social services and support resources into the rare disease care model.
- Strengthening community and government partnerships to advocate for policies that support affected families.
- Building local capacity to enable in-country genetic testing, making results more timely, cost-effective, and widely available.

With continued collaboration and investment, RDZ aims to ensure that children with rare diseases, and their families in Zimbabwe receive the care, answers, and hope they deserve. iHope is incredibly proud to partner with RDZ and to support genetic diagnosis for families in Zimbabwe. Learn more about RDZ at raredisorderszim.org.



Summitting Mt. Kilimanjaro for a Purpose

In August 2024, Trudy Nyakambangwe and seven of her family members embarked on an extraordinary journey to summit Mount Kilimanjaro. This climb had a purpose: to raise funds to establish the first Clinic for Genetic Disorders in Zimbabwe at the University of Zimbabwe. Although the climb is complete, you can still contribute to the clinic here: <https://gofund.me/5a630549>



Launch of First Genetics Clinic in Zimbabwe

The Genetics Clinic at the University of Zimbabwe Medical School officially launched on February 25, 2024. Pictured above are the genetics clinic team which includes a lab scientist, clinical nurse and physician on the first day of clinic. It is currently the only facility offering diagnostic testing nationwide for rare disorders.

Sharon Terry invited Commissioner of Lancet Commission on Rare Diseases

Sharon Terry, CEO of Genetic Alliance, was invited as one of 27 global members of the newly established Rare Diseases International (RDI)-Lancet Commission on Rare Diseases. This initiative is dedicated to improving the lives of Persons Living with a Rare Disease (PLWRD) worldwide, and officially launched with its inaugural convening in Geneva, Switzerland in November 2024. The Commission, co-chaired by Dr. Kym Boycott (CHEO), clinical director of iHope partner lab CHEO, strives to generate evidence-based



recommendations that can be implemented in all countries, and to drive meaningful change in healthcare policy and practice. By generating robust data and concrete recommendations, the Commission aims to ensure that individuals with rare diseases are not only recognized but receive the care, support, and visibility they deserve, regardless of where they live.

This mission deeply aligns with iHope's commitment to expanding access to genetic testing and diagnosis for underserved children with rare diseases. Engaging in broader global initiatives like this not only helps to amplify the voice and impact of iHope but also to establish the network of partners necessary to create lasting change. Read more here: [https://www.thelancet.com/journals/lancet/article/PIIS0140-6736\(25\)00211-9/abstract](https://www.thelancet.com/journals/lancet/article/PIIS0140-6736(25)00211-9/abstract)



Donate to Cover Testing for a Family

In its first operational year at Genetic Alliance, iHope supported nearly 600 families with undiagnosed genetic conditions, but there are so many that we could reach! Your donation is critical —\$100 covers a family, supporting shipping and customs costs to get samples from clinical sites around the world, such as Zimbabwe, Ghana, Peru, Mexico, and North Macedonia, to the iHope lab network for detailed genomic analysis.

DONATE NOW

The success of iHope would not be possible without the generous support of Illumina and our incredible donors. Thank you! We'll see you again in June!