ISSUE #1

iHope Newsletter 2024 Year in Review

iHope Genetic Health

December 2024

Same Mission, New Model

Welcome to the inaugural issue of the iHope Newsletter! We're thrilled to have you join us as we reflect on a truly transformative year for iHope and share our vision for the future.

In January 2024, iHope became part of Genetic Alliance, a 501(c)(3) organization, marking an exciting new chapter. This transition allowed us to establish a distributed network of laboratories, enabling free clinical genomic testing for underserved children referred by iHope clinical sites. While the program structure has evolved, our mission remains steadfast: to reduce barriers to access complex genetic testing for underserved children around the world suspected to have a genetic condition.

In 2024, we launched two grant cycles selecting three best-in-class labs: The Hospital for Sick Children (SickKids) and CHEO (Children's Hospital of Eastern Ontario) in Canada, the New York Genome Center in the U.S., and Mendelics in Brazil. Alongside this expansion, we re-engaged 15 clinical sites—nine of which are in low- and middle-income countries—each chosen for their exceptional patient care and commitment to serving communities with limited access to advanced genetic diagnostics.

With a reimagined program foundation established in the first half of the year, testing operations launched last summer and we're proud to have already supported nearly 400 families. Looking ahead, we're filled with optimism and excitement for what 2025 and beyond will bring as we build on the momentum of 2024. Thank you for being here and supporting our mission!

With gratitude,

The iHope Team

In this issue:

From left to right: collaborating iHope clinicians at the Hospital Edgardo Rebagliati Martins-ESSALUD, Instituto Nacional de Salud del Niño - San Borja, and Instituto Nacional de Ciencias Neurológicas, all in Lima, Peru.

Regional Spotlight - iHope in Latin America

Established in 2012, Mendelics is Brazil's first and largest genetic diagnostic laboratory specializing in next-generation sequencing. As iHope's first partner lab in Latin America, Mendelics supported all five iHope clinics in the region during 2024: Hospital Edgardo Rebagliati Martins-ESSALUD, Instituto Nacional de Ciencias Neurológicas, and Instituto Nacional de Salud del Niño - San Borja, in Lima, Peru; the Padrino Children's Foundation in Todos Santos, Mexico; and Hospital Infantil de as Californias in Tijuana, Mexico. Additionally, iHope collaborated with Mendelics to expand its efforts to support indigenous children in the Amazon, providing no-cost exome sequencing to 25 families. "The purpose of iHope fits perfectly with our mission, which is to make genetic diagnosis quick, accurate, and accessible to all who need it. Patients deserve access to the best service, quality, and care, no matter where they are," said David Schlesinger, CEO and founder of Mendelics.

"Exome Day" at Hospital Infantil de las Californias: A Legacy of Impact

iHope's longstanding partnership with Hospital Infantil de las Californias is one of the programs most enduring clinical collaborations. Under the guidance of Dr. Marilyn Jones and the leadership of Diane Masser-Frye, in-person "Exome Days" resumed this year and provided exome sequencing to 47 families. These marked the 14th and 15th such clinics that have supported more than 200 families through iHope since 2016.

"Exome Week" at Padrino Children's Foundation: Coordinated Care in Action

In October, a dedicated clinical team at the Padrino Children's Foundation in Todos Santos, Mexico, led by Dra. Alejandra Peña and Dra. Mitzy Cervantes, provided 52 children and their families with exome sequencing through iHope. To ensure seamless multidisciplinary care, additional specialist appointments were coordinated during the same week for the families, many of whom traveled long distances to participate.

Donate to Cover Testing for a Family

In 2024, iHope supported nearly 400 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

iHope in the Scientific Literature

In 2024, two key iHope manuscripts were published in peer-reviewed scientific journals, showcasing the transformative impact of equitable access to genomic testing.

The first study, published in *The American Journal of Human Genetics*, analyzed data from 1,004 individuals who received genome sequencing through iHope. These patients were referred from 24 clinical sites across eight countries, with 41% receiving a genetic diagnosis. Importantly, at least 40% of patients experienced changes in their medical care as a direct result of their test results. Notably, patients referred from low- and middle-income countries had similar rates of management changes as those from high-income countries, underscoring that a precision diagnosis is a critical driver of improved care, regardless of geography. Read the full article **here.**

The second study, published in *npj Genomic Medicine*, focused on the impact of clinical genome sequencing (cGS) for 247 patients from three clinics in Peru. Over 50% of patients received a diagnosis, with candidate variants identified in an additional 22%. Genome sequencing significantly improved care, influencing diagnostic evaluations (85%), genetic counseling (72.1%), and management plans (71.3%). Changes included new referrals, treatments, testing strategies, and imaging, highlighting the critical need to expand genomic testing access in resource-limited settings like Peru. Read the full article **here.**

In The News:

- Munduruku indigenous children are monitored by neuropediatricians from the Ministry of Health
- Free genomic sequencing offers hope for children living with a rare disease
- Sequencing Benefits Even Low-Income Rare Disease Patients, iHope Study Shows

The success of 2024 would not have been possible without the generous support of Illumina and our incredible donors. Thank you! We'll see you again in March with 2025 updates, partner spotlights, stories of the impact of iHope and more.