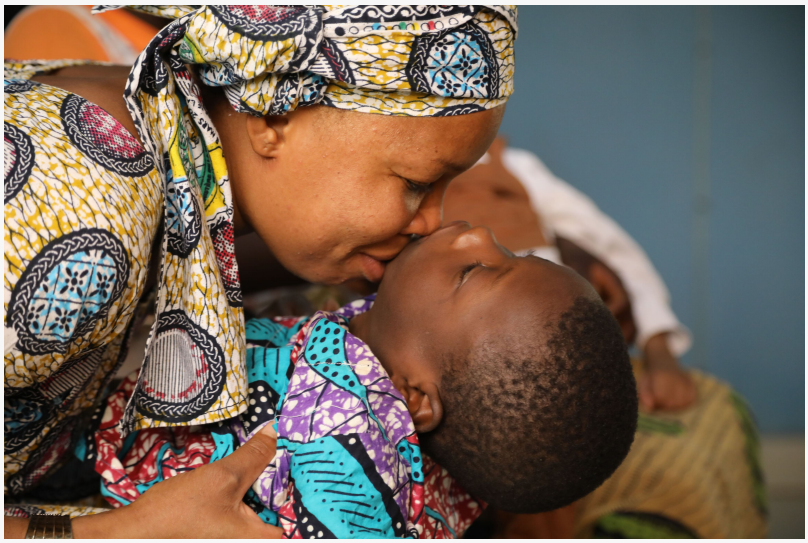
iHope shows benefits of genome sequencing across global population of genetic disease patients

International philanthropic effort provided genomic testing to 1,004 underserved individuals and shows it may reduce health disparities.


Mother and child with a rare genetic disease in Africa

DAMASCUS, MARYLAND, UNITED STATES OF AMERICA, June 6, 2024 /EINPresswire.com/ -- The iHope program, an international consortium dedicated to providing genetic testing to those in need, has published findings demonstrating the widespread benefit of clinical genome testing in the American Journal of Human Genetics.

The silent epidemic of rare genetic diseases affects at least 250 million people worldwide, but few receive the genetic testing they need. This diagnostic odyssey leads to missed opportunities for care, unnecessary medical interventions and prolonged psychological suffering. iHope was initiated at Illumina Inc. more than a decade ago to close this gap, especially for patients in low- and middle-income countries (LMICs).

The iHope program's publication focused on more than 1,000 patients who were tested over a five year period. Their analysis showed that at least 41% of patients received a definitive molecular diagnosis and a majority had a change in management, including new medications and avoided surgeries.

For patients in LMICs the impacts were even more pronounced, with at least 57% receiving a diagnosis and up to 78% receiving changes in the care because of clinical genome testing. Patients in LMIC that benefited from clinical genomic testing included a young woman in Peru whose diagnosis of a movement disorder initiated treatment that led to a pronounced improvement of her condition, and a child who received tailored molecular intervention for spinal muscular atrophy, a potentially devastating and life threatening disorder. Based on these findings the iHope authors argue that increased access to genomic testing can decrease health

disparities globally.

“This study shows it is entirely feasible to provide genome sequencing to resource limited populations, and that doing so leads to tangible changes in care. It is imperative that we make genomic testing accessible worldwide,” said Ryan J Taft PhD, Chief Scientific Officer of Genetic Alliance.

These outcomes have catalyzed the expansion of the iHope program, now managed and operated by Genetic Alliance, to include multiple testing laboratories across two continents and additional clinical sites in LMICs. Over the next five years iHope is slated to benefit thousands of patients per year and enable in-country capacity building and education.

“This publication is proof that one of the best things we can do for genetic disease patients, no matter where they are in the world, is to get them a precision diagnosis. We want patients to be empowered with information about themselves, and to get the care they both need and deserve,” said Sharon Terry, CEO of Genetic Alliance.

To learn more about iHope, Genetic Alliance, and how you can contribute please visit iHope Genetic Health.

Publication Reference
Thorpe et al. (2024) The impact of clinical genome sequencing in a global population with suspected rare genetic disease. The American Journal of Human Genetics. 10.1016/j.ajhg.2024.05.006.

About iHope
iHope program, managed and operated by Genetic Alliance, provides comprehensive genetic testing to undiagnosed individuals from underserved communities. An in-kind donation from Illumina has enabled Genetic Alliance to partner with clinical genomic testing laboratories to offer free testing to children with suspected genetic conditions. Contribute to our fund to increase access to this testing.

About Genetic Alliance
Genetic Alliance is a nonprofit organization dedicated to transforming health through genetics. By fostering collaboration among diverse stakeholders, Genetic Alliance accelerates the translation of genetic research into practical health solutions, advocating for patients and families affected by genetic conditions.

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