The May 2024 kickoff event in Changsha, Hunan Province

Expanding hope to more families with rare diseases in China

The Illumina iHope China program will provide 1800 families with genetic testing in 2024, broadening its reach and impact nationwide

THIS YEAR MAY BE the most impactful yet in the short history of iHope China. In Changsha, Hunan Province, the program's leaders recently announced plans to deepen its impact by increasing reagent donations and expanding its coverage area.

Since its inception in 2022, iHope China has embodied its namesake for families dealing with rare genetic diseases. The program was established by the Special Fund for Diagnosis and Treatment of Genetic Diseases under the March of Dimes Birth Defects Foundation of China, and it receives support from the Illumina Corporate Foundation and Give2Asia.

In collaboration with key hospitals across the country, iHope China has made significant strides. It provides free genetic testing to families with rare disease patients and offers systematic training in genetic disease diagnosis to front-line clinicians nationwide. By mid-May 2024, the program had helped 513 families in collaboration with seven hospitals, ensuring valuable treatment opportunities for many children.

Addressing the issue of limited access to timely health care for grassroots patients in remote areas, the program has also implemented volunteer clinics at participating hospitals.

iHope China's reach promises to be extensive in 2024, covering 13 provinces and cities in China. By the end of the year, the program aims to support 1800 families with rare diseases through genetic testing and during the kick-off event, more than 200 clinicians joined the offline genetic counseling workshop. This training includes guidance on the entire diagnostic process, from technology selection to report interpretation.

Early screening and diagnosis of rare disease are critical for improving patients' prognoses. Professor Wu Lingqian, director of the Department of Genetics in the School of Life Sciences at Central South University in Changsha, says, "The level of awareness of rare diseases among front-line clinicians is crucial to the ultimate benefit of tens of millions of rare disease families in China. The program's expansion and increased medical education efforts mean more patients will benefit from an enhanced rare disease prevention and treatment system."

Xue Jingjie, secretary general of the March of Dimes

Birth Defects Foundation of China, notes the program's role in enhancing early diagnosis, providing timely treatment, and supporting families both psychologically and financially. "With the support of strong national policies, medical advancements, and broad public opinion, the early diagnosis rate of rare diseases in China is improving. The iHope China program has been dedicated to educating patients and their families about rare diseases and providing comprehensive support," Xue says.

According to a study published in the European Journal of Human Genetics, 71.9% of known rare diseases are caused by genetic variations, and nearly 70% manifest in childhood.[1] Over the past decade, whole genome sequencing has become increasingly vital for enabling timely and accurate diagnosis of genetic disease, and increasing treatment time for children with rare diseases.

"At Illumina, we are dedicated to accelerating access to genomics to achieve health equity for billions worldwide," remarks Sharon Vidal, global lead of Corporate Social Responsibility at Illumina. "Expanding the reach of genetic testing enables families and communities to benefit from next-generation sequencing. To ensure treatments are effective across diverse populations, we strive for equitable representation in genomic data. This effort helps reduce biases and improves medical outcomes for a broader range of patients."

Jenny Zheng, Illumina senior vice president and general manager of the Greater China region, adds, "The iHope China program reflects our commitment to expanding genomics accessibility and reducing the burden of disease on patients. In China, we uphold the spirit of 'genomics for good,' driving public welfare through technological innovation and collaboration with partners. Our goal is to expand hope for more families with rare diseases through science and care."

Guided by the mission of contributing to China's goals for preventing disease, the program will continue to bolster clinical acumen in diagnosing and treating rare diseases. It will also harness the collaborative strengths of philanthropic organizations, medical institutions, and enterprises, thereby amplifying public awareness and fostering societal advocacy for rare diseases. ◆

For more information about iHope China or to inquire about applying, please contact ihope@csqx.org.cn.

1. Nguengang Wakap S, Lambert DM, Olry A, et al. Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database. Eur J Hum Genet. 2020;28(2):165-173. doi:10.1038/s41431-019-0508-0