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KNIH's Initiatives for the Advancement of Genome Medical Services

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Over two decades have passed since the completion of the Human Genome Project, and genome medicine is now transitioning from research to implementation in clinical settings. The Korea National Institute of Health (KNIH) has played a central role in advancing genomic medicine in Korea through infrastructure development, data generation, and translational research. A fundamental resource for genomic research is large-scale bio big data. KNIH has contributed by establishing population-based cohort studies and disease registries, and by systematically collecting human biospecimens through the Korea Biobank Project. These resources have enabled population genomics and disease association studies. To support researchers, KNIH developed the Korean Biobank Array (K-CHIP), a population-optimized tool that has been widely used for genetic studies of complex diseases. Recognizing the need for open science, KNIH has also made its genomic and epidemiological data accessible through curated platforms(CODA), thereby accelerating collaborative research and enabling more scientists to contribute to the field of genomic medicine. In the field of rare diseases, KNIH has been instrumental in developing diagnostic technologies. Through the National Bio Big Data Pilot Project, the Institute has implemented whole genome sequencing (WGS) to improve genetic diagnostics and support the discovery of novel disease-associated genes. This year, KNIH is further expanding its efforts with the launch of two new national initiatives: a WGS-based diagnostic program for critically ill newborns, and a newborn genomic screening project. These programs aim to integrate genomic diagnostics into routine care and improve early detection of rare and genetic disorders. The presentation will highlight KNIH’s ongoing initiatives and strategic vision for accelerating the integration of genome science into healthcare to realize the promise of precision medicine in Korea.

Keywords: Genetics, Precision Medicine, Biobanking, Rare Diseases, Data sharing