

Resourcing research into genomic medicine

A big-data project and one of the world's biggest biobanks will be invaluable resources at the **KOREA NATIONAL INSTITUTE OF HEALTH.**

The future of medicine is personalized, precise and powerful. It's about tailoring diagnostics and treatments to the unique genome of an individual patient and their disease, which can substantially boost the chances of early and accurate diagnosis, and safe and effective therapy.

Precision medicine is built on a foundation of genomic data, which is why the South Korean government launched the pilot National Bio Big Data Project in 2020. Its goal is to create a vast repository of human health, medical, physiological, and genomic data from a South Korean cohort that can be accessed by biomedical researchers everywhere. Korea National Institute of Health is one of the key players behind this project, and it helped to establish the Bio Data Dam, a project for collecting, standardizing and utilizing data generated through public and private networks.

"Precision medicine requires huge amounts of data to identify the main drivers of disease development and treatment effectiveness," says

Hyun-Young Park, director of the Department of Precision Medicine at the Korean National Institute of Health.

THIS ALLOWS RESEARCHERS TO LOOK FOR SPECIFIC GENETIC FACTORS THAT PLAY AN IMPORTANT ROLE IN DISEASE.

ACQUIRING DATA ON RARE DISEASES

Backed by South Korean government agencies and carried out by 16 hospitals nationwide, the pilot project is focused on collecting genomic data and biomedical information from patients with rare diseases. For example, the first patient to be registered with the National Bio Big Data Project in June 2020 was a young man with Duchenne muscular dystrophy, a heritable muscle-wasting disease. By amassing information such as behavioural and environmental factors that could play a role in disease



development, the project will become a valuable source of information for identifying genetic risk factors, which will help inform the development of new treatments.

SPREADING A BROADER NET

However, precision medicine needs genomic data from the broader population, not just those diagnosed with diseases. This information will contribute to a better understanding of the spectrum of genomic diversity present in a population. It will also be valuable for pinpointing genomic risk factors and early warning signs of a disease.

The National Biobank of Korea launched the Korea

Biobank Network in 2008, a network that consists of the National Biobank and 20 biobanks at university hospitals. For more than a decade, this network has been building a repository of standardized biometric data from a million South Koreans from around the country, across all ages and backgrounds and all states of health and disease.

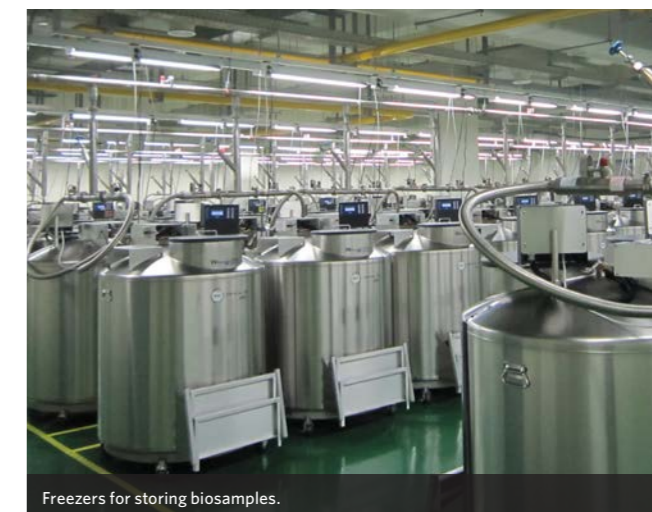
As of the end of 2020, more than 990,000 Koreans have participated in this massive data-collection effort, more than 1,365 papers have been published and 109 patents have been lodged based on research that has used data from the national or regional biobanks.

MORE THAN JUST DATA STORAGE

The National Biobank's role is not simply to collect and store biospecimens and data, says Eugene Kim, the biobank's deputy scientific director. "We provide not only an accelerator to generate large-scale genetic data, but also a bridge to produce biometric data using recently developed technologies," she says. The network has already generated genomic information for more than 200,000 South Koreans using the Korea Biobank Array — a genotyping chip that has been developed specifically for the national population, having been designed using genomic

data from citizens. This allows researchers to look for specific genetic factors that play an important role in disease.

Furthermore, through the National Bio Big Data Project and intramural research, more than 10,000 individuals have had their entire genomes sequenced. In 2021, the National Biobank of Korea simultaneously produced a range of 'omics' data (whole genome-seq, scRNA-seq, TCR/BCR-seq with cytokine profiling) of COVID-19 patients. Park says that these data will be open to the public and used in research aiming to develop vaccines and treatments.



Freezers for storing biosamples.



A tray containing an array of specimens from the biobank.

SETTING HIGH STANDARDS

Another important role of the National Biobank is to develop standard protocols and regulations to govern the collection, storage, and use of human biological samples and biomedical information. Kim says that the biobank is standardizing these protocols, which will both facilitate management of the collection and make it easier for researchers to find the information they need.

South Korean authorities have also ensured that the highest standards of informed consent, anonymization, and research ethics are met in the collection, storage and use of

the biological and biomedical data by the biobanks and the Bio Big Data Project.

Having seen the power of the Bio Big Data Dam, which is based on biobanking infrastructure developed over the past two decades, Park is excited about the research and health possibilities that they offer. "I hope that all our projects can be used for researching and promoting the health of the next generation," she says. ■



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