Scientist spotlight

Expanding genetic disease testing in Korea with customizable population genomics microarrays

Introduction

Predictive genomics researchers worldwide are exploring opportunities to leverage population genomics to investigate many different diseases within defined populations. The Applied Biosystems™ Axiom™ Genotyping Solution is designed to enable researchers to optimize and customize microarrays for unique ethnic populations and studies of any size. Investigators can select from over 10 million variants and markers, add their own unique markers, and leverage imputation algorithms to capture additional variants. The Axiom Genotyping Solution is highly scalable, with Thermo Fisher Scientific expertise to support your test development from initiation through testing.

We spoke to Jong Bhak, PhD, chief executive officer (CEO) of Clinomics, about the importance of customized genotyping for Clinomics' work in developing disease-focused microarrays for the Korean population.

Thermo Fisher Scientific: Why did you choose microarrays, and specifically Applied Biosystems™ Axiom™ arrays, for your work?

Jong Bhak (JB): We have been using microarrays in our business for a long time because they are critically important for our human genotyping tests. When we started working with microarrays, we compared several products and determined that Axiom arrays are the most flexible and cost-effective, so we developed our own custom content on that platform. We have used Axiom arrays for over 10 years.

Jong Bhak, PhD, CEO, Clinomics; Professor, Biomedical Engineering, Ulsan National Institute of Science and Technology (UNIST) Genome Lab and Korean Genomics Center (KOGIC)

Clinomics is a leader in genome-based early diagnosis of complex diseases in the Korean population. Clinomics technology and products enable genetic testing from blood, saliva, or epithelial cells for a wide range of individual physical characteristics or diseases, including cancer, complex diseases, rare diseases, mental conditions, nutrition, aging, and ancestry. Multi-omics analyses are conducted using Clinomics' internationally renowned bioinformatics capabilities.

The library of millions of markers for Axiom arrays is fundamental to both our research and our product development. The publicly available markers are the foundation of our microarrays, with additional space dedicated to our Korean-specific or disease-specific markers. The size of the microarray is very important. The size determines the variant coverage, the amount of information we can gather, and the cost. Sometimes, we use large, off-the-shelf microarrays when we want to cover the whole genome for research or if we need a microarray quickly. We use smaller sets of markers to develop tests to target certain diseases. We are trying to expand our marker sets on different types of Thermo Fisher microarrays.

Thermo Fisher: How is customization important for your work?

JB: The ability to customize our microarrays is essential for us to continue to develop tests and expand our business for different diseases. We currently have at least 20 products in our Geno series of DNA tests, which cover up to 11 phenotypes and hundreds of genotypes. Our primary focus is on risk prediction and early diagnosis for cancer, heart disease, and mental conditions such as depression and risk of suicide. We would like to expand the number of markers and diseases that we cover, so we must be able to design custom microarrays for each of them.

We also have tests for nonmedical applications. The Korean government regulates the types of phenotypes that can be reported from direct-to-consumer genetic testing. Only six companies are certified to do genetic tests for nonmedical applications. We are certified to test for 43 phenotypes and hundreds of markers on our arrays. We want to expand to 70 phenotypes.

Thermo Fisher: What challenges have you faced?

JB: Our different types of tests require different numbers of markers and different sizes of microarrays. It can be a challenge to balance the combination of markers needed with microarray size and cost-effectiveness. For example, we can get a lot of information from a million-marker microarray, but it may cost more than a smaller custom microarray, so the large microarray might be best for research purposes. If we only need a minimal number of markers, we will design a custom microarray with our markers to maximize profit. Balancing requirements for size, content, and cost can be difficult.

Thermo Fisher: How is the ability to do genome-based early detection of diseases changing Korean health care?

JB: Korea has among the most advanced health checkup centers in the world. Routine checkups are cheap and subsidized. Everyone over the age of 40 has annual checkups. Clinicians are very busy and in Korea we do not have many genetic counselors, so the owners of the hospitals and health checkup centers make the decisions about which tests to include. Multiple hospitals have adopted our DNA tests for several diseases. Now, Koreans are already beginning to have genetic testing as part of regular checkups.

> "The ability to customize our microarrays is essential for us to continue to develop tests and expand our business for different diseases."
>
> Jong Bhak, PhD, CEO, Clinomics

Thermo Fisher: How will Axiom microarrays be important in the future of genome-based disease diagnosis?

JB: The Korean government is launching a project to sequence the genomes of one million Korean people. The project includes the Korea Biobank Array (KoreanChip), which was developed on the Axiom array platform by the Korea National Institute of Health to provide microarray-based reports to assess the risk of certain diseases. The KoreaChip will be used to genotype 500,000 people.

Microarrays contain the selected targeted marker sets of the whole genome, so we can decide which custom markers to include. The microarray designers like us and the microarray platform companies like Thermo Fisher can design marker sets that are targeted for specific applications, sizes, and costs. I think microarray technology will be used for a long time, even if the cost of next-generation sequencing (NGS) goes down. The critical factor is how we design microarrays for certain markets so that they are very cost-effective for both the microarray designers and the hospitals.

Thermo Fisher: What excites you most about the future of genomics in Korean health care?

JB: Sometime in the next 20 years, genome data will be combined with artificial intelligence computing technologies. Then, from just a blood test, every year when you go to the hospital, you will know exactly what is happening in your body. With genotyping, there will be a multi-omics prediction on your health conditions. That's the future.

Learn more at **thermofisher.com/predictivegenomics**