Direct-to-Consumer Genetic Testing Expands in Asia

A joint-venture DTC company focuses on the large and growing South Korean market as an entry point to expand into Southeast Asia.

Introduction

It seems that you can't open a magazine or your favorite social media app without seeing an advertisement for direct-to-consumer (DTC) genetic testing. Unlike traditional genetic tests, usually performed through a health care provider, DTC tests enable consumers to send companies a buccal swab for analysis and receive personalized information about ancestry, kinship, common traits, and disease risk. The demand for such tests is growing rapidly. According to Global Market Insights, the global direct-to-consumer (DTC) genetic testing market will exceed $2.5 billion in the next five years.[1]

Min-Seob Lee, PhD. is the Founder and Chairman of Diagnomics, one of the first next-generation sequencing (NGS) consumer genomics companies offering genomic testing services in the United States. Since 2011, Diagnomics has provided research and development services, including consumer genomics solutions and Clinical Laboratory Improvement Amendments (CLIA) testing, to various industry and academic institutions across the globe. In 2013, he cofounded the Eone Diagnomics Genome Center (EDGC) in Incheon, South Korea, a joint US–South Korea venture with the mission of expanding DTC and clinical genomics solutions throughout Asia.

iCommunity spoke to Dr. Lee about the state of consumer genomics in South Korea, the development of the Infinium™ Global Screening Array (GSA), and EDGC's future plans to expand into personalized medicine applications.

Q: What capabilities did Diagnomics and Eone bring to the EDGC joint venture?
Min-Seob Lee (MSL): I came to the US in 1991 as a graduate student in molecular biology and, over the years, worked with several biotech companies. After gaining that experience, I founded Diagnomics as a partner of the Illumina Genome Network (IGN). Diagnomics was selected to be one of the first three bioinformatic and data analysis partners in IGN.

Eone Laboratories is the largest diagnostic laboratory in South Korea. It's been in the diagnostic reference lab business for more than 35 years and was looking to expand into consumer genomics and personalized medicine. It needed to upgrade its existing clinical diagnostic laboratories with NGS systems. We agreed to form a joint venture and founded EDGC in South Korea to prepare for these new opportunities.

Since then, EDGC has worked very closely with Illumina to provide bioinformatics services, including whole-genome sequencing (WGS) and cancer genome sequencing data annotation and interpretation. That expertise and working closely with Illumina have put us in a position to expand our consumer and clinical genetic test offering to South Korea.

Q: What is the status of consumer genomics in South Korea?
MSL: Consumer genomics in South Korea is limited compared to what you would find in the US or other countries. In South Korea, only a few DTC tests have been approved and most genetic testing is performed through clinics, hospitals, or physicians. Even for ancestry analysis, there isn't a DTC test and a person has to go to a clinic for testing.

Q: Do you expect the market to grow and become more competitive in the coming years?
MSL: South Korea has the demand and the infrastructure to support genomics-related businesses. There are several reasons why the market will grow in the coming years. First, South Korea has a single-payer health care system. If the government can be convinced that genetic testing benefits the general public, it will be easier to launch new DTC tests. Second, South Korea is an information technology–focused country and very tech savvy, with high-speed internet connections and vast broadband services. It has the infrastructure to support the growing number of consumer and clinical genetic testing options. Finally, South Korea has the fastest growing life expectancy. It's expected to be the first country in which the average lifespan will grow to 90 years within the next 10 years.

Min-Seob Lee, PhD is the Founder and Chairman of Diagnomics and the Cofounder of EDGC.

As a result, I believe that there is great potential for South Korea to become a leading country in genomic solutions for clinical and consumer applications. Currently, Eone and EDGC have the largest share of the South Korean market for genetic testing.

Q: Has DTC testing demand increased since you founded EDGC?
MSL: We established EDGC in 2013 and the number of samples we've tested has increased nearly 100% each year. We're performing genetic testing in South Korea and actively expanding our services to other Asian countries. In fact, more than half of our samples are from countries such as Thailand, India, and Singapore.

> "I believe that a significant potential market for genomics is in reaching healthy people who want to live longer, happier, and healthier lives."

Q: How did Diagnomics become involved with the development of the GSA?
MSL: Diagnomics is based in San Diego and we have worked closely with Illumina and IGN to align our business needs and goals to expand our personal genomic offering. When I first heard about Illumina forming the largest consortium for a global genotyping array, we immediately joined the project. We were heavily involved in working on the GSA content and providing feedback on how to improve the array and enable development of new applications. While Diagnomics has developed several GSA-based CLIA genetic tests, EDGC has developed most of our GSA-based consumer and clinical genetic tests.

Q: What are some of the DTC tests that EDGC offers?
MSL: MyGenPlan is one of the first DTC tests that we offered and is based on the GSA. It combines a routine blood test with GSA analysis to deliver a simple report. The blood test measures cholesterol and glucose levels, which can help in assessing diabetes and chronic disease risk. Using the GSA, MyGenPlan adds the associated genome information to provide metabolic disease and obesity risk. It's a unique offering. We are also developing new testing options that enable people to understand their family ancestry.

There is a significant potential for personalized medicine based on the GSA. As the technology advances, a GSA chip could be used throughout a person's lifetime to assess their health risks.

Q: When did you begin using the Infinium Asian Screening Array (ASA)?
MSL: The ASA is a very good tool for the discovery of Asian-specific markers in samples. It's also the best choice for EDGC as we develop a new application for Asian populations because it has the markers that cover those populations. Currently, we use it mostly for research studies. It's compatible with the GSA and both perform well in our genetic testing applications.

Q: What software pipeline are you using for data analysis?
MSL: We have created our own data analysis platform. Illumina software processes the data initially. When we have the raw genotype or NGS data, we use our own software pipeline for analysis in the cloud. Our consortium partners can also use our pipeline to analyze their data.

The beauty of the GSA is that it enables researchers to share information. In the old days, many institutes developed their own unique arrays and it was difficult to share the data to support new research studies. The GSA provides a standardized format. While there is some custom content, most of the content can be shared across the consortium. This enables us to work closely with other consortium members to develop new genetic tests. Many of the consortium partners are also using the informatics pipelines, like the EDGC Annotator, that we have developed.

> "There is a significant potential for personalized medicine based on the GSA. As the technology advances, a GSA chip could be used throughout a person's lifetime to assess their health risks."

Q: What is the EDGC Annotator?
MSL: The EDGC Annotator is a bioanalysis software pipeline that we developed at Diagnomics and EDGC. It is available as a BaseSpace™ App. It enables researchers to analyze genome information quickly to identify clinically relevant information or variations in the genome. A researcher can input their data into the pipeline, select the VCF file format, and the software annotates all functional genetic variants. It provides allele frequency data and clinical insights using ClinVar, OMIM (Online Mendelian Inheritance in Man), COSMIC (Catalogue of Somatic Mutations in Cancer), 1000 Genomes Project, dbSNP, and VEP (Variant Effect Predictor) databases.

We made the analysis process very simple. There are only a few steps and it provides a complete list of all the annotations that we've gathered from public sources and some of our own research efforts so that others can easily perform genome annotation and minimize their informatics development time.

The output of the EDGC Annotator is an Excel file. It enables a researcher to easily perform their own sorting and searches. It's a very easy and convenient tool for graduate students and postdocs to use.

Q: How do you reach potential customers?
MSL: One of the good things about being part of a joint venture is the reach and access that it provides to large markets. Eone is the largest diagnostic laboratory in South Korea and has been there for 35 years. It has many existing relationships with clinics

and hospitals, enabling EDGC to reach South Korean customers and those customers to get to know EDGC.

While we rely on the clinical side as an entry point, we have active outreach programs to engage consumers and increase their understanding of the value of genomics. I believe that a significant potential market for genomics is in reaching healthy people who want to live longer, happier, and healthier lives.

> "I believe that GSA-based tests and the GSA consortium activities will ultimately accelerate the rate at which WGS testing is performed."

Q: What do you see as EDGC's role in the future of genomics?
MSL: Illumina and I have a similar vision about the future of genomics. NGS is as powerful a tool as PCR was in the early 1990s and has revolutionized the field. I remember when Jay Flatley, the former CEO of Illumina, said that every newborn baby will have their whole genome sequenced by 2019. That hasn't happened yet and the question is why. Why aren't we at a point where every person is having their genome sequenced and analyzed? The cost of sequencing has dropped dramatically, so I don't think it's just because of the cost. I believe that the delay is due to the amount of information we get from NGS.

When people perform whole-genome sequencing (WGS), they get > 100 gigabytes of data. Most people don't even know how all that data can help them and what the benefits are to their health and lifestyles. It's important that people have access to the data and that there is an ecosystem that enables them to understand it. That's why we developed MyGenomeBox, an app where people can deposit their genomic information and access hundreds of different informatics tools from ancestry analysis to diet and exercise recommendations.

The GSA consortium provides a great opportunity to understand genomic information in a new way. I believe that GSA-based tests and the GSA consortium activities will ultimately accelerate the rate at which WGS testing is performed. The data the GSA provides to consumers will fill in the gaps in their understanding so that people will appreciate the value of genomic information.

Q: What are EDGC's next steps for expanding its personal genomics business?
MSL: Our immediate goal is to expand our business into the precision medicine area. We are trying to introduce more genetic testing products into existing clinics. We are working on various research projects to apply genetic information to prevent, diagnose, and manage chronic diseases such as Alzheimer's disease and cancer. We believe that genomics will enable us to tailor and personalize treatment and disease management for patients in the future. That's our long-term goal.

We also want to expand our offerings into the precision lifestyle area with tests that enable customized skincare, diet, and exercise, all based on an individual's genome. I see genomics as the basis of providing individuals with personalized information that will improve their health and their well-being.

Q: How will Illumina products, systems, and services support you in reaching those goals?
MSL: We are very happy with the NGS revolution that Illumina has spearheaded over the last two decades. We appreciate its efforts to lower the costs of service offerings so that we can build our business. We will continue to work closely with Illumina to develop new genetic tests that maximize the use of its platforms. It's been a very good relationship and we plan to continue developing many new DTC and precision health tests based on Illumina technologies.

Learn more about the products mentioned in this article:

Infinium Global Screening Array, www.illumina.com/products/by-type/microarray-kits/infinium-global-screening.html

Infinium Asia Screening Array, www.illumina.com/products/by-type/microarray-kits/infinium-asian-screening.html

BaseSpace Apps, www.illumina.com/products/by-type/informatics-products/basespace-sequence-hub/apps.html

References

1. Ugalmugale S and Mupid S. DTC Genetic Testing Market size to exceed $2.5 billion by 2025. Global Market Insights. www.gminsights.com/pressrelease/direct-to-consumer-dtc-genetic-testing-market. Accessed August 16, 2019.