

Precision medicine: From dream to reality

Introduction

Mark Bouzyk, PhD, is co-founder and Chief Scientific Officer of AKESOGen, a genomic services company that works with scientists engaged in clinical and basic research around the world. Prior to founding AKESOGen, Bouzyk developed Emory University's genomics facilities and served as director of Emory's Center for Medical Genomics. Dr. Bouzyk was also the director of genetic laboratory sciences at GlaxoSmithKline (GSK) for nearly a decade. He has worked on applying genetic testing technology in the clinical environment and has created state-of-the-art CLIA-compliant and CAP-accredited labs to meet the needs of high-throughput genetic, genomic, and biomarker studies. Thermo Fisher Scientific recently talked with Dr. Bouzyk about his work.

Thermo Fisher: Can you give us an overview of your company AKESOGen?

Bouzyk: The first thing people wonder about is where our name came from. We took it from Akeso, who is the Greek goddess of healing and curing, because ultimately that's what we are working toward. We founded the company in 2010 and have established and expanded it through grants and contracts, with no outside investment funding. This year we were again named to Inc. magazine's list of America's fastest-growing companies, and we are ranked as the 23rd fastest-growing healthcare company. We are CLIA-compliant, CAP-accredited and have a GxP-enabled Quality Management System, enabling us to work with investigators in the clinical and clinical trials areas. We have a 14,000 square foot facility, where we have put an infrastructure in place that covers all the bases for genomics and precision medicine research and development. We support three sectors: academic, government, and pharmaceutical research, with an emphasis on looking at disease risk factors; the diagnostic sector, where we carry out cancer diagnostic testing and are about to launch capabilities for Alzheimer's testing; and we support clinical trials by performing omics testing.

Thermo Fisher: What factors are important to your customers when selecting a service company like yours?

Bouzyk: Quality is the most important. Our core competency is in generating data of the highest quality. It is imperative to make sure that your DNA is at the right concentration or that the RNA is of the right quality so we have rigorous internal QC standards. Many times we ask for the starting material so we can perform the isolations ourselves. Anything less can lead to incorrect conclusions, lost time, and great expense. We apply the strictest process controls, laboratory process precision, and state-of-the-art technology. Other factors are the ability to test exactly what the customer wants, with fast turnaround. These factors are realized through detailed and prompt communication with the customer so we fully understand their needs and priorities.

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Thermo Fisher: One of your customers is the Million Veteran Program. What work are you doing for that project?

Bouzyk: The Million Veteran Program was created by the U.S. Department of Veterans Affairs (VA) with the goal of learning more about how genes affect veterans' health. This project is creating a very large database of genetic, lifestyle, and health information. The VA who created the project chose Affymetrix to build a custom genotyping microarray tailored to specific genetic- and disease-related targets identified by VA scientists and physicians. AKESOgen was chosen to genotype samples in this study, using the Affymetrix™ array platform, and we have completed genotyping for over 200,000 veterans at this point. We are delivering over 750,000 genetic data points to the VA for each of the individuals we genotype. The billions of high-quality genotypes ultimately generated will provide the Million Veteran Program with valuable understanding of how genetic factors affect human health.

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Thermo Fisher: How does a large project like this differ from smaller-scale ones?

Bouzyk: I think that these large-scale projects are groundbreaking in the sense that people at long last are seeing the need to do this kind of work and that this is going to provide a lot of very interesting information. As the projects become large, they become more visible and the public is more likely to hear about them. We are already hearing that the Million Veteran Program is going to be open to active duty personnel in addition to veterans, and that speaks to the growing interest. As more and more people in the community become interested in DNA and about having their own DNA evaluated, it impacts society as a whole. Over time, this will only compound and will result in increased funding for research and more successful clinical applications.

Thermo Fisher: Which Affymetrix products do you provide services for?

Bouzyk: We primarily use the Affymetrix Axiom™ array platform and we are delighted with it. We have three Affymetrix GeneTitan™ instruments, simultaneously running large numbers of samples on both genotyping and expression arrays. We have added some automated liquid handlers to increase throughput, and we have an instrument running almost all of the time. Overall, our experience has been second to none.

I have been working with Affymetrix for many years, since my days at Smith Kline Beecham and GSK, where I used some of the first arrays, so we have a close relationship and hope to continue in that vein with Thermo Fisher Scientific. It really goes beyond genomics and arrays; we are working in proteomics and planning on metabolomics. We are planning on using Affymetrix's ProcartaPlex™ immunoassays for projects such as research on vitamins. We will build on all the work we have already done, even adding technologies like mass spectrometry.

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Thermo Fisher: What motivated you to become one of the first service providers to offer Axiom™ Precision Medicine Research Array and PharmacoScan™ Solution?

Bouzyk: Yes, we are about to start the first project on the Axiom Precision Medicine Research Array and we are very excited about the content. There is a whole multitude of markers on the array that are useful and will have clinical utility. We're getting inquiries from people in all spectrums of disease and therapeutic research areas. I think it will have broad utility and appeal.

We are also eager to get the new PharmacoScan array for pharmacogenomics as it's designed to handle very difficult-to-genotype genes such as CYP 2D6, and we'll be happy to offer this to our clients. A lot of pharmaceutical companies are coming to us for these types of assays. The data will enable them to stratify populations in clinical trials based on certain genes. I think this will be very useful for them because it will cover not only the genes they are interested in, but also others they may not have considered, at "one-stop shop" pricing.

We are increasingly getting calls from non-research sectors such as insurance companies, corporate wellness programs and hospital systems who want to offer this kind of risk-based profile to their employees or patients. For the hospitals, even though the orders must be initiated by the physician, this is a way to position themselves as a factor in the precision medicine world. We will ultimately see more genes as clinically actionable and CPT codes will be created for the array-based tests. I think the industry will be able to show cost/benefit rationale for pharmacogenetic testing as we generate more data, and we will see all of these markets open up over the next few years.

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Thermo Fisher: What sort of increased demand are you seeing for your services in light of President Obama's Precision Medicine Initiative (PMI)?

Bouzyk: Yes, although those of us in the field were already working on precision medicine before this rallying cry, we have definitely seen an uptick in genotyping. Whereas at one time experts thought that sequencing would replace arrays, we now have new products and effective price points that make a strong case for genotyping arrays. The new precision medicine research array that Affymetrix has just announced is allowing investigators to really think differently about the types of experiments they can consider because the cost has come down. I think this will be a game-changer. We have a lot of interest from clients who want to use this array, but don't want to set up their own lab. So they come to us.

I think the precision medicine research array has wide applications. Take biobanking, for example. If you're storing large numbers of samples over time, you can make those samples more valuable and interesting to more people by running the precision medicine research array now that the pricing is so reasonable. I think it's going to revolutionize the way people do research and practice medicine over time.

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