New Illumina SNP Array Fuels European Consortium Founded to Foment 'Third Generation GWAS Era'

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NEW YORK (GenomeWeb) – Three major European genomics service providers have founded a consortium to offer cost-effective, whole-genome genotyping on a new Illumina SNP array.

Illumina created the chip, dubbed the Global Screening Array, to meet demand from biorepositories and large cohort studies that are keen to genotype their holdings at a high scale and a low price. "We see this as a very significant offering and are seeing a strong resurgence in interest in our array offering," said Jason Johnson, senior director of Illumina's array business. He noted that Illumina customers have already committed to run at least three million samples on the new chip.

One user will be André Uitterlinden, head of Erasmus University Medical Center's Human Genomics Facility and Genetic Laboratory in Rotterdam, the Netherlands, who told GenomeWeb this week that his lab has partnered with the French National Genotyping Center in Paris and Life&Brain, a commercial service provider in Bonn, Germany, to offer genotyping on the array as a new service.

Uitterlinden said that the response to the offering from the consortium's network of biorepositories and scientific partners has been "enormously positive" and that the consortium hopes to have a million samples committed for genotyping by July 31.

He would not divulge the names of the consortium's partners citing confidentiality, and declined to comment on pricing. Illumina's Johnson said that the company is looking to offer the array for less than $70 per sample.

It's a price point that the consortium believes should entice researchers involved in array-based genome-wide association studies, as well next-generation sequencing-based projects, to take advantage of the offer, and could even fuel another round of large association studies.

"We think that with the new Global Screening Array, we will enter the [third] generation GWAS era," said Per Hoffmann, who runs the genomics platform at Life&Brain, housed at Bonn University Hospital. "This new array will allow [us] to increase sample numbers from 10,000 to millions at a [never-before] seen price," he told GenomeWeb.

According to Uitterlinden, the data generated on the new chip will remain with the biobanks and cohort studies that supply the samples for genotyping. He noted that the service is open to all researchers, not just those based in Europe. His hope is that investigators will be able to use the massive amount of data generated as part of new association studies. Uitterlinden underscored that the new consortium is not being funded through any EU mechanism. Rather, institutions will pay to have their samples genotyped using their own sources of funding.

"If you contact many people and explain the big opportunity, they are able to mobilize money for this, even from regular funding schemes," he said.

There certainly is money to be had. Both Illumina and Santa Clara, California-based Affymetrix, its main competitor in the SNP chip market, have long courted biobanks, looking to balance high throughput with low cost to enable them to genotype their vast repositories. Affymetrix, which was acquired by Thermo Fisher Scientific earlier this year, in 2013 agreed to genotype the UK Biobank's 500,000 samples on a custom microarray and has announced similar high-volume deals. Illumina advertises a menu of arrays for biobanks on its website, such as its Infinium Multi-Ethnic Global BeadChip, each of which enables researchers to genotype eight samples across more than 1.7 million markers.

Moreover, Johnson said that Illumina has seen interest in the new chip from healthcare systems, clinical researchers, and consumer genomics firms, in addition to biorepositories.

The members of the new European consortium see the San Diego vendor's new Global Screening Array as something distinct from what has been offered so far.

"There has been a small history of SNP arrays over the past 10 years, but this array is a game changer in that the price is so low and the content is so rich," said Uitterlinden. Using the GSA, customers can screen 24 samples per chip, he said. Each array contains 660,000 markers covering 26 populations, including about 50,000 markers specific to clinical research. Customers can also add up to 50,000 other markers relevant to their studies.

"It's very economical versus the content we are providing," said Johnson, noting that the chip also contains 10,000 markers for quality control and sample tracking, as well as population stratification, making it, in his words, "ideal for biobanks."

Because of this, Life&Brain's Hoffman called the new chip "truly multi-ethnic" and noted that it has been optimized for imputation, making it easier for researchers to compare new data with data from samples genotyped with older chips. "The clinical content will also allow physicians to implement the array into clinical diagnostics for specific diseases," he predicted.

Uitterlinden said that the GWAS community needed such an array to make more discoveries.

"We know as researchers that there are many samples in biobanks, in big cohort studies, in clinical case series, stored in freezers, that can use such a cheap array badly in order to give us much more data, to enlarge our sample size, and to enrich it with more phenotypes, in order to empower the very successful research line we started with GWAS in 2005," he said.

While initial rounds of array-based GWAS were characterized by some as not delivering the desired amount of scientific discoveries, an outcome that pushed many investigators towards using NGS to identify rare variants linked to common diseases, Uitterlinden maintained that increased collaboration among once-protective researchers, as well as ever-enlarging datasets, have borne out the effectiveness of the array-based scientific approach.

He cited work in schizophrenia as an example of this success, specifically the Schizophrenia Psychiatric GWAS Consortium that his lab took part in, along with Life&Brain and many other collaborators. "Before we reached 100,000 samples, we weren't able to see anything," he said of variants related to schizophrenia. "Once we passed that number, we got hits in the GWAS. So we passed a threshold."

It is this "bigger is better" lesson that Uitterlinden and his partners in Paris and Bonn are trying to apply via the new consortium. "By increasing sample size and by involving many more researchers, you come up with many more hundreds of discoveries," he said. "This successful research line needed a cheaper array to unleash the power of all of the biobanks. Finally, the vendors listened to us."

According to Uitterlinden, the chip might also be attractive to researchers who have been using NGS, the results of which he said have been "a little bit disappointing."

"The original idea was that by sequencing, we would discover the rare alleles that would account for a big portion of missing heritability," said Uitterlinden. "It seems to be case that you have rare alleles of

modest to moderate effect size, so sequencing has not been the answer to find the missing heritability," he said. He acknowledged that investigators that rely on NGS often argue back that they have not yet reached the sample size achieved by array-based studies. "That indeed is a valid remark, but still I think the big answer is not coming from additional sequencing data. It's a more complicated story."

While Uitterlinden is focused on lining up the million samples for genotyping by the July 31 deadline, he believes the data generated on the new chip could also contribute to his own research interests. As an investigator, he coordinates the genetic and genomic analyses in the Rotterdam Study on 12,000 elderly individuals and the Generation R birth cohort, which includes 7,000 children and 12,000 patients. He is also on the steering committee of the Cohorts for Heart and Aging Research in Genomic Epidemiology (CHARGE) consortium.

"All consortia will see the data coming in from all of these individual biobanks" on the new array, said Uitterlinden. "That has to do with a successful mechanism of collaboration, providing more data leading to more discoveries," he said.

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