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BEACONS Newborn Genome Screening Study Selects Seven Sites, Finalizes Gene List

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NEW YORK – Researchers from the Building Evidence and Collaboration for Genomics in Nationwide Newborn Screening (BEACONS) project said on Wednesday that they have selected newborn screening programs from six states and one US territory to participate in the study and have finalized a gene list for analysis.

The seven participating public health programs are Iowa, Minnesota, New York, Oregon, Puerto Rico, South Carolina, and Texas.

The project, which will assess the integration of whole-genome sequencing into existing public health newborn screening, will focus on 746 genes, associated with 777 conditions that are actionable in the first year of life.

"One thing that was really important to us in this study is to ensure the representation of the United States in performing this feasibility assessment," said Sikha Singh, director of newborn screening and genetics at the Association of Public Health Laboratories (APHL) and a co-investigator of the study. "Right now, the newborn screening system is accessible to nearly all 4 million babies born across the United States, and that is something that we are pressure-testing and stress-testing with this study."

According to Singh, the seven study sites were chosen from roughly 20 state or territorial public health programs that had expressed interest in participating in the project. The BEACONS investigators reviewed a variety of parameters for them — including program readiness, demographics and populations served, number of core and secondary disorders currently screened, and geographic representation — to ensure those selected are representative of the entire country.

BEACONS settled on seven sites for now, given funding limitations, but the project would accommodate more participants moving forward if resources allow, Singh said. "If there is an opportunity in the future to engage additional programs, we are definitely open to it and looking for ways to do that, as well," she noted.

Kicked off in October with $14.4 million in funding from the National Institutes of Health (NIH) Common Fund Venture Program as well as $12.6 million of in-kind support from GeneDx and Illumina, BEACONS, as the nation's first multistate newborn genomic screening initiative, aims to add whole-genome sequencing to existing newborn screening systems with as little disruption as possible.

"This study truly looks at how different states and territories can incorporate whole-genome sequencing as part of their newborn screening program activities, and try not to infringe on anything that we have been doing for the last 60 years as it relates to newborn screening," said Jelili Ojodu, senior director of newborn screening and genetics at APHL and a BEACONS co-investigator.

Unlike existing newborn screening programs, which are mandated by state laws across most of the country, BEACONS will only enroll participants with parental consent, Singh said.

According to Robert Green, professor of medicine at Harvard Medical School and the overall project lead for BEACONS, the study will recruit participants through several avenues.

For one, each participating state or territory will work with the BEACONS team to select two birthing hospitals to serve as primary recruitment sites based on delivery volume, demographics served, and willingness to collaborate with the newborn screening system. The study plans to deploy dedicated coordinators to these hospitals to educate and consent parents with newborns or expecting parents.

In addition, the study plans to enroll through other types of outreach, such as social media and educational pamphlets.

Overall, BEACONS aims to enroll up to 30,000 newborns over three years beginning in March or April, though Green said the team is still deciding whether enrollment will be the same across the sites or driven by their population.

Participation in BEACONS will not interfere with routine newborn screening, which will run in parallel with the study. According to Green, BEACONS will use the same dried blood spot sample collected for routine screening, and the state public health lab will create a subsample to be sent to GeneDx, the study's sequencing partner, for whole-genome sequencing.

Green said the funding from GeneDx and Illumina has "no strings attached," and the companies "will not use any of the data in their own product development."

After sequencing, GeneDx will analyze a list of 746 genes. Nina Gold, a pediatric geneticist at Massachusetts General Hospital who led the gene list development effort for BEACONS, said that a unique aspect of the project's list, which was finalized through a multidisciplinary process, is that it exclusively focuses on 777 conditions that are actionable within the first year of life.

These conditions can be divided into 12 categories, including core conditions from the Recommended Uniform Screening Panel (RUSP), cardiac, endocrine, gastrointestinal, hematologic, and other disorders.

The gene list is also different from some previous studies, Gold pointed out, such as BabySeq, which included a much larger list of over 4,000 genes, or the GUARDIAN study, which has a smaller list and focuses on about 250 conditions.

In addition, Gold said the gene list will be iterative, and the team might update it at the halfway point of the study to incorporate feedback.

Once the analysis is complete, the results from BEACONS will be routed back to the public health laboratory, which will report positive findings to participants using the state's existing framework. "What we are trying to

learn is how clinicians handle these results," Gold noted.

Given BEACONS covers substantially more conditions than the routine newborn screening programs, the study also has a so-called Genome Resource Center to support clinicians as needed in understanding and communicating the results, according to Gold.

In terms of data, BEACONS will follow a "very strict" data governance and usage policy, Singh said, adding that participant families ultimately own their data and will have the ability to opt out of any data storage or sharing beyond the regulatory requirements.

The goal of BEACONS is to provide a window into a potential future national newborn sequencing program, Green noted.

"It's a great deal of responsibility," he said. "If we do it well and the results are positive, then we could accelerate the life-saving benefits of this technology. If we do it poorly, or we find surprising results that dissuade us from thinking it's a good idea, then it may set this back."

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