NHGRI-Funded Population Genomic Screening Network to Study 'Full Cycle of Implementation'

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NEW YORK – The Population Genomic Screening Network, a new research consortium funded by the National Human Genome Research Institute, plans to study the clinical implementation of genomic screening in primary care, including outcomes of patients who test positive for any of the genetic conditions to be included.

Under the five-year initiative, six clinical groups will implement a shared protocol for population genomic screening in primary care settings. The Human Genome Sequencing Center at Baylor College of Medicine will sequence all patient samples, while the University of Washington's Genetic Analysis Center will oversee the network in its role as coordinating center. The initiative formally kicked off last month and has thus far received $8 million in funding, which will support the clinical groups and sequencing center for fiscal year 2026 and the coordinating center for the next two fiscal years

NIH did not respond to a request for comment on how much funding it intends to award to the PGSN in total. Two years ago, NHGRI published funding opportunity notices promising $38 million in funding over five years.

Each of the clinical groups will enroll at least 5,000 patients and screen them for the same six or seven genetic conditions that are "sufficiently prevalent and actionable enough that the entire adult population should be screened for them," said Jason Vassy, an associate professor of medicine at Harvard Medical School who is leading one of the clinical groups and co-chairing the network's steering committee. Three of these will be the Center for Disease Control and Prevention's Tier 1 conditions: Hereditary breast and ovarian cancer syndrome, Lynch syndrome, and familial hypercholesterolemia. The network will select the others in the coming months —chronic kidney disease and inherited cardiomyopathies have been proposed — and there will likely be very few exclusion criteria for screening, Vassy said.

According to Vassy, a major impetus for the PGSN was a 2023 study that found population genome screening for CDC Tier 1 conditions would likely be cost-effective for all US adults younger than 40 if the cost of testing was low and probands had access to preventive interventions. Other genetic conditions are, by now, similarly penetrant and actionable, and primary care is a natural setting for screening for them, he added. "All of our other screening and preventative health — that really kind of belongs in primary care. That's where you get

your mammograms, your colonoscopy, your flu shots," said Vassy, who is also a primary care physician.

Caitlin Allen, an associate professor of implementation science at Wake Forest University School of Medicine, who, like Vassy, is leading a clinical group and co-chairing the network's steering committee, noted that "there have been a lot of population-based screening projects happening around the country, but they are kind of siloed and may or may not be happening in primary care settings." In contrast, the PGSN will have the clinical groups share data, provide regular updates, and jointly issue recommendations by the end of the initiative.

Early days

The network formally launched on June 25, when all participating groups met for the first time. The first year of the initiative will be dedicated to deciding which research questions it should prioritize, Vassy said, and to beginning to recruit patients. In addition to the list of conditions that should be included in screening, outstanding questions include how to integrate the screening orders and results into electronic health records and how to involve non-physician caregivers.

A guiding principle of the network is to avoid overwhelming an already-overburdened workforce of primary care physicians. That's one of the reasons for limiting the screening to a small set of high-confidence genes, Vassy said. The clinical groups will additionally explore allowing participants to enroll and get their blood drawn outside of standard primary care visits, and Allen added that engaging genetic counselors to follow up with patients who receive positive results will also be a priority.

Each clinical site will have some flexibility in how it deploys the screening, even though all will implement the same basic protocol. "For scientific rigor, there will need to be some amount of harmonization of those methods across the sites, but because … we're implementing this in clinical care in different healthcare systems that have different cultures, processes, government structures, etc., there will be some local customization," Vassy said.

That could prove important given the different types of primary care settings represented across the clinical sites. For instance, Katherine Nathanson, a professor at the Abramson Cancer Center of the University of Pennsylvania and a clinical site lead, will be recruiting individuals seen in primary care and family medicine clinics in West Philadelphia. Pamela Ganschow, director of the Cancer Prevention and Survivorship Clinical Programs at the University of Illinois Cancer Center and another PI in the network, will target primary care sites within an academic medical center and a federally qualified health center network.

Sequencing strategy and potential outcomes

Donna Muzny, director of operations of Baylor's Human Genome Sequencing Center, said she expects to start receiving patient samples — most likely blood but possibly also saliva — around June 2027. Before then, her center will advise on which genes to prioritize beyond the Tier 1 list and what sequencing approach to take, though she anticipates it will be whole-exome sequencing. "Right now, the default is really to do whole-exome sequencing, but that's still part of the discussion going forward," she said, though "no one has talked about doing specific panels."

Ginger Metcalf, executive director of strategy and partnerships at the HGSC, explained that "it's not a huge cost savings to do a panel over an exome, because most of the costs that you incur for an exome is in the capture and in the library, [and] you have those same costs if you're doing a gene panel." Furthermore, "the value-add for an exome or genome would be the research utility," she said. "Doing a program like this, it's such a big endeavor to collect the samples, consent, and get things moving, that it makes sense to invest in something that has broader utility."

The Baylor team will use Illumina's NovaSeq X for sequencing and the DRAGEN workflow for analysis. Genomic data will be deposited in the NHGRI's Analytics Visualization and Informatics Lab-space (AnVIL) database along with participant phenotype information and outcomes tracked by the clinical sites, like the healthcare interventions participants receive in the years after getting screened. According to Metcalf, this sets the network apart from earlier initiatives.

"We did clinical genomes for All of Us, we returned a lot of really actionable, important information, but there was no closing of that loop in terms of being able to understand what the outcomes were, the actual clinical utility of that intervention," she said. Additionally, the NHGRI's Electronic Medical Records and Genomics (EMERGE) network "set the foundation for these types of studies … but to my knowledge, [the PGSN] is one of the first programs in primary care to think about the full cycle of implementation for clinical genomics."

With genomic and outcomes data from at least 30,000 individuals, Metcalf is hopeful that the network will catalyze the introduction of population genomic screening into primary care and provide guideposts for doing so. But beyond healthcare providers and the public, there's another group she'd like to realize the value of genomic screening: insurance companies.

The data, "could be used to potentially shape policy decisions around reimbursement," she said, noting that payors typically only cover single-gene tests in limited circumstances, such as a BRCAtest for someone with a family history of breast cancer. And care that comes downstream of a preventative screen, such as an echocardiogram due to the presence of a

cardiomyopathy variant, might be denied coverage due to a perceived lack of a clinical indication.

"The program," Metcalf said, "is hoping to show the data that would maybe move the needle on that."