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Concerns Rise Over Potential End of Public Health Genomics in US

Apr 21, 2025 | Turna Ray

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NEW YORK – The elimination of the Public Health Genomics Branch (PHGB) at the US Centers for Disease Control and Prevention is yet another blow to the genomics and precision medicine sector.

As part of the March 27 reduction in force (RIF), the US Department of Health and Human Services has eliminated 2,400 jobs at the CDC, including those within the Division of Blood Disorders and Public Health Genomics, which housed the PHGB. Employees in that branch, who are now on administrative leave and will be let go in June, did not respond to requests for comment or said they didn't have permission to speak to the press.

The branch also employed several contractors, whose contracts will end in the fall. Some who worked in this branch have publicly discussed the shuttering of the division on social media and indicated they are looking for employment. In response to a request for comment, the CDC directed *GenomeWeb* to contact HHS, which did not reply to questions.

The loss of the PHGB comes as cuts across federal healthcare agencies have diminished expertise, resources, and infrastructure that doctors, researchers, the drug and diagnostics industries, and state and local groups committed to advancing precision medicine had come to rely on. "There is a generalized anxiety that the future of basic infrastructure that supports precision medicine may be uncertain," said Scott Topper, chief clinical operations officer at Color, a company that offers cancer care services, including genetic risk screening. Topper has heard of potential disruptions to funding for other areas of the precision medicine sector, and "it is not yet clear who will be left when the dust settles," he said.

The future of many genomics and precision medicine-focused research projects funded by the National Institutes of Health are in limbo. At the NIH's National Human Genome Research Institute, former Director Eric Green has left and former acting Director Vence Bonham Jr. is on administrative leave. Amid cuts at the FDA, reviewers are worried about their ability to ensure the safety and efficacy of *in vitro* diagnostics, key to delivering precision medicines. The Advisory Committee on Heritable Disorders in Newborns and Children, which advised HHS on the heritable conditions that babies should be screened for at birth, has been disbanded. The viral hepatitis lab at the CDC's National Center for HIV, Viral Hepatitis, STD, and TB Prevention has been eliminated. Even a resource that clinicians use regularly to help them diagnose and treat patients with heritable diseases, called GeneReviews, may be in jeopardy.

If the aim of the RIF is to "do more with less," as HHS Secretary Robert F. Kennedy Jr. has said, then the PHGB was particularly adept at that. Muin Khoury, who had been the PHGB's director from its inception in 1998 to "almost until the end," led this group through five name changes, its placement under different organizations within the CDC more than a dozen times, 16 bosses, and six presidential administrations, and kept the program alive despite having its budget decimated by more than 90 percent from $12.3 million in 2010.

When Khoury retired voluntarily last year, he said the PHGB had around a $1 million budget and employed five full-time staff and between five and 15 contractors and students. Reflecting on the PHGB's history through a "politically neutral lens," he acknowledged that "from the beginning, the office was not given a lot of resources, not endowed with hundreds of people," because the expectation was that the PHGB would work with other federal, state, and local groups and leverage their resources to integrate genomics into public health programs, Khoury said.

The PHGB did this work for more than 25 years, often without recognition. Few may know or realize that this small government branch had a hand in their newborn being tested for severe combined immunodeficiency, a genetic condition that can be fatal if not diagnosed and treated early, or that their doctors were offering them screening for a genetic heart condition because the PHGB recommended testing for it. While the genetic testing industry was growing at breakneck speed, the PHGB was pushing for evidence-based, safe, and fiscally responsible implementation of genomics-based interventions. The branch also contributed to infectious disease surveillance and supported healthcare workforce education and training.

"If you truly are a civil servant, often your work is somewhat invisible to others," said Debra Duquette, who directs the graduate program in genetic counseling at Northwestern University.

Now that the PHGB is gone, Duquette fears that patients' access to quality genetic testing is at risk. Duquette, who was the genomics coordinator for the Michigan Department of Community Health from 2004 to 2017, thinks a lot about a phone call she made during that time to a mother whose son had died of sudden cardiac death. The department had learned that this child's heart condition had a genetic cause, and Duquette wanted to make sure, per the CDC's guidance, that this family also got genetic testing and counseling.

At the end of the call, the mother said, "Thank you for telling us this information. I thought we were forgotten and no one cared," Duquette recalled. Absent national leadership on public health genomics, she fears "there's going to be a lot more people feeling like this."

'Shaping the future'

The concept of public health genomics has always been saddled by a philosophical debate as to whether the goals of public health are incongruent with the current capabilities of genomically informed care. In public health initiatives, the focus is on interventions that can broadly benefit a population, such as helping health systems create smoking cessation programs and providing tools that community programs can use to promote healthy eating habits and exercise. But genomics is most readily equated with rare disease and individualized medicine.

Moreover, government-based public health programs have typically focused on producing health gains over the short term, but the value of incorporating genomics in public health will take much longer to realize. "When people think of genomics in public health, there are people who'll say, 'Well, this doesn't look like public health.' That's right, it doesn't look like public health because it's not a traditional public health program," said Scott Grosse, an economist who researched the impact of newborn screening and genetic testing at the CDC from 1996 to 2024 and often collaborated with the PHGB. "The

genomics and public health [program] was all about how to get people, companies, and healthcare systems thinking about genomics as it affects everything they do. That's a long game."

The public health genomics group was created in 1998 in parallel with the completion of the Human Genome Project to translate the data and discoveries emerging from the first sequenced human genome. Around the time the public health genomics group's budget was cut by 90 percent in 2011, the HGP was approaching its 10-year anniversary, and many were taking stock of the medical advances that had resulted from this $3 billion project and wondering if the cost was worth it.

In a *Science* article published at the time, entitled "Deflating the Genomics Bubble," several leading healthcare ethicists and researchers wrote that it will take decades to realize the full potential of genomics in healthcare, and in the near term, there may be more public health gains from figuring out how to help people stop smoking and exercise more. "US National Institutes of Health and Department of Energy spending on genomics vastly exceeds the budget for behavioral and social science research," University of Alberta's Timothy Caufield and colleagues wrote. "Given that even a small improvement in our ability to alter behaviors could yield major benefits, we suggest a reappraisal of the apportioning of funds to promote the promise of improved human health."

Amid this debate, when it came to public health genomics, "people didn't know what to do with us because we were kind of shaping the future," Khoury said. During one of the many lean budgetary years, Khoury recounted how one of the CDC directors he'd served under was asking programs to estimate how many lives they could save in the next 100 days. The director didn't even bother asking Khoury this question, he said, probably because the answer was going to be, "Not that many," but Khoury was still able to make a case for the future value of public health genomics.

The PHGB's work was setting the groundwork for a future where genomics would play a much bigger role in population health than it does today. "Everyone wants to sell the genome," Khoury said. "But all the millions and billions [of dollars] in investment and all that discovery work, it needs that final step of population health benefits."

Cutting through the hype

Although patients have been hearing about the "promise of precision medicine" for a long time, continued government investment in public health infrastructure and policy is critical to ultimately realizing that promise, said Alicia Zhou, CEO of the Cancer Research Institute, a non-profit that funds research programs. "Government-led public health programs, like the CDC's public health genomics program, play a key role in driving evidence-based evolution of policy," Zhou said. "To ensure equitable access for all patients, it will be important for these types of non-commercial programs to exist."

The supporters of the PHGB pointed out that the group's charge was to cut through the hype and advance evidence-based implementation of genomic tests. Khoury often describes the PHGB's mission as the "three 'I's": "Identify" evidence-based genomics interventions that could improve public health if appropriately adopted, and "inform" and help state and local programs "integrate" them.

Toward this end, the public health genomics group in 2004 set up the Evaluation of Genomic Applications in Practice and Prevention (EGAPP) Working Group, an independent body that reviewed the evidence on genomic tests and made recommendations about whether they were ready for broad clinical deployment.

For example, in 2009, the working group did not find enough evidence to definitively recommend for or against using gene-expression profiling tests like Agendia's MammaPrint or Exact Sciences' (then Genomic Health's) Oncotype DX. In subsequent years, the field gathered more data on the ability of

these tests to determine a breast cancer patient's risk of recurrence, and oncologists regularly perform these tests now in clinical practice.

On the other hand, in 2010, this working group found "insufficient evidence" on eight tests, which together gauged 58 gene variants associated with cardiovascular disease and which labs were marketing. Unconvinced that testing for most of the gene variants in these tests would benefit the public health, the group urged doctors to wait for more evidence before using them routinely in practice. At least one of companies marketing these tests is no longer in business and its unclear the extent to which the other tests are still being sold as they were back in 2010.

The EGAPP Working Group had its last meeting in 2014, but as more genomically informed treatments have entered the market, the evidence evaluation framework it developed for genetic tests have been adopted by other bodies like the American College of Medical Genetics and Genomics and cancer groups that put out treatment and testing guidelines, Khoury said.

"Precision medicine has become an essential part of caring for patients in the most effective and efficient way possible," said Kathryn Phillips, a health economics and health services research professor at the University of California, San Francisco, who was an EGAPP Working Group member. "The future use of genomics has even more opportunities coming, for example, gene therapies to cure diseases such as sickle cell anemia."

"But without groups like this one [on public health genomics] to figure out which tests are useful and which are useless, patients and their providers face a bewildering marketplace, and many patients who would benefit from valuable genomic testing to target their care won't get it," Phillips said.

After funding for public health genomics was slashed, the group had to get creative and advance its mission by collaborating with others in the federal government. The group provided weekly updates on COVID-19 genomics and conducted epidemiology research with the CDC's Center for Surveillance, Epidemiology, and Laboratory Service to identify groups at risk of hospitalization or death if infected. With the CDC's Office of Advanced Molecular Detection, the public health genomics group launch pilot projects that explored how genomics can be used in the surveillance and treatment of infectious diseases like Ebola and tuberculosis. It researched the usefulness of family history data in screening patients for the risk of obesity, diabetes, and heart and blood conditions using data from NIH's All of Us Research project.

"I knew from the beginning that this is not just about genomics," Khoury said. "It's about cancer and heart disease and infectious diseases and birth defects and reproductive health."

One of public health genomics' most impactful partnerships was with the National Cancer Institute Epidemiology and Genomics Research Program, which led to the creation of a framework for scanning the evidence on genomic tests and placing them into a three-tier system. Tier 1 tests were those that the CDC had determined could improve population health if appropriately implemented. These include genomic tests for identifying patients at risk for hereditary breast and ovarian cancer syndrome, Lynch syndrome, and genetic cardiac conditions like familial hypercholesterolemia.

This tiering system has influenced molecular diagnostic companies' commercialization strategies, payors' coverage policies, and guidelines bodies' recommendations on which tests to use. Although these recommendations are very "narrow, conservative, and often misunderstood," Color's Topper said, they provide "broad legitimacy" to screening for hereditary disorders. "The fact that they existed was profound," he said. "It shifted the conversation from 'Is screening okay?' to implementation questions: Which people, when, which genes, [and] what protocols [will] maximize benefit and minimize harm? What support do patients and doctors need?"

From the start, Color aligned its genetic test offerings with the Tier 1 conditions hoping to have an impact on population health. One of it strategies is to provide genetic testing within employee benefits programs for the Tier 1 conditions, an approach that employers looking to cost-effectively improve the health of their workforce seem to appreciate at a time when the market is crowded with labs offering large panels that gauge dozens of genes with varying evidence. "Offering a three-condition panel based on the PHGB recommendations communicated our eagerness to focus on maximizing beneficial outcomes in a way that respects the important differences in each employee population versus just taking the approach of 'more information is always better,'" Topper said.

Laurence Sperling, chief medical officer of the non-profit Family Heart Foundation, worked with CDC's public health genomics group to get genetic cardiovascular conditions on the Tier 1 list, including FH and hypertrophic cardiomyopathy, and collaborated on a tool to help families discuss their history of heart disease. (He noted that his comments about the public health genomics group do not reflect the views of his employer.)

FH, which occurs in 1 out of 250 people worldwide, "is not considered rare enough to be a rare disease, but it also was not considered common enough to be a public health concern," Sperling said, suggesting that by adding it to the Tier 1 list, the public health genomics group was helping change that perception.

Some of the contributions of the PHGB might go overlooked, because while the group often brought key stakeholders together in the same room to discuss public health initiatives, others often get the credit for implementing them, reflected Grosse, the CDC economist and newborn screening expert who retired last year. He recalled how in 2001, the then Office of Genetics and Disease Prevention organized a workshop on primary immunodeficiency, where public health experts and clinicians discussed the importance of screening newborns for severe combined immunodeficiency (SCID). At the time, researchers had been thinking about developing an absolute lymphocyte count test for this genetic disorder that causes defects in the immune system and makes infants more prone to infections. The condition can be fatal if not diagnosed quickly and treated early with bone marrow transplant or gene therapy.

However, the test healthcare providers were initially considering would have been "unwieldy," Grosse said, since as some pointed out during the meeting, lymphocyte counts may be normal in some SCID patients, and the test would have to be performed in a hospital on blood samples, and couldn't be done on dried blood spots taken from babies as a standard practice within state newborn screening programs.

Eventually, NIH researchers came up with a SCID test that measured the circular DNA fragments that are created as T cells mature. Patients who have low levels of these so-called T-cell receptor excision circles (TRECs) may have SCID. The NIH-developed test used quantitative PCR to measure TRECs from standard dried blood spots.

The first molecular newborn screening test was for SCID, Grosse recalled, and it took the CDC's public health genomics program bringing together key stakeholders at that 2001 meeting for the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) to eventually recommend SCID be added to the list of conditions that all babies, at a minimum, should be screened for. By 2020, all 50 US states were screening for it.

Last year, the CDC shifted the PHGB under the National Center on Birth Defects and Developmental Disabilities so that it could start a genomics program focused on rare diseases and newborn screening. "Of course, that never got off the ground because of what is happening now," Khoury said. HHS has also disbanded the ACHDNC.

Countering the view that genomics is relegated to rare diseases and incongruent with the goals of public health, Sperling highlighted that the PHGB had been publishing blogs and articles on Lipoprotein(a), or Lp(a), a little-known biomarker that when elevated increases one's risk of heart disease. One out of five people globally have elevated Lp(a), but less than 1 percent of Americans know they have it.

Recognizing that Lp(a) testing is not standard practice, drugmakers developing treatments for heart conditions that reduce Lp(a) have launched campaigns to encourage people to get tested. The FH Foundation also has an education campaign and a program to identify high-risk patients who should get Lp(a) testing. "Lp(a) is a public health concern, but our public health entities are likely unaware that this should be a public health concern," Sperling said.

The PHGB's efforts with FH Foundation to improve diagnoses of genetic cardiovascular conditions is just one example of how it was "working across and breaking down silos and emphasizing the CDC's mission of bringing science to public health," Sperling said.

Unfinished business

A leaked draft of President Donald Trump's fiscal year 2026 budget proposal provides some insight into how his administration is hoping to reorganize CDC. For example, the administration proposes in this document to cutback CDC's focus on chronic diseases and shift some of the work that the national birth defects center did under a maternal and child health program within the newly created Administration for a Healthy America (AHA) — a plan that HHS recently articulated.

While the administration seems committed to advancing genomic surveillance of pathogens, HHS doesn't appear to be considering any program resembling what the PHGB did. In fact, a lot of PHGB's contributions are now archived on CDC's website or have disappeared entirely.

One of the virtues of his group, Khoury said, was that it could slide into lots of divisions at the CDC and integrate genomics within their programs, for example, in the Office of Science during the COVID-19 pandemic and then at the national birth defects center. "If AHA is to succeed in maternal and child health and in chronic diseases such as cancer, heart disease, and diabetes, then some focus on public health genomics is essential," Khoury said, adding that without careful planning public health genomics may "continue to be in a coma or severely underfunded for the foreseeable future."

More than 100 public health leaders, including past HHS secretaries and CDC directors, have written to Congress that the HHS RIF "demonstrates a stunning disregard for science, public health, and what America needs to be healthy," especially when CDC's expertise and technical support are needed to curtail an influenza outbreak among livestock and rein in a measles outbreak. These public health leaders have asked Congress to hold oversight hearings and strengthen the nation's public health infrastructure.

"The RIF is being done in a completely immoral, haphazard, and inefficient manner," said James Tabery, a medical ethicist at the University of Utah. "I don't think there's any reason to think good is going to come of it."

In his book, *Tyranny of the Gene: Personalized Medicine and its Threat to Public Health*, Tabery argues that the focus on developing pricey precision medicines for small populations has shifted resources away from researching social determinants health that could benefit many more people. "What's unfolding is bigger than my concerns about precision medicine," he said. "The hyping of precision medicine is far below the devastation being done to the federal government by the Trump Administration."

While Tabery been critical of the research community's overpromising of the potential of precision medicine, he recognized the PHGB's work in Tier 1 conditions, encouraging screening of rare versions of common diseases like cancer and heart disease, has "real value." And in its work on newborn screening and pathogen genomics, the PHGB was trying to "maximize the potential" for genomics for everybody, he noted.

"That's a loss if it's not there, and we should be frustrated by the fact that it's been cut without any good reason given," said Tabery. Initially, Tabery was hopeful that maybe the upside of having a new head of HHS with fresh priorities would mean a greater focus on some of the public health research that hasn't received much attention in recent decades, but he's seen "no evidence yet of that being the case."

The recent cuts to the CDC, Sperling fears, will diminish US leadership in promoting public health. "Many other countries have developed their own CDCs based on the model we have in this country," he said. "There's a concern that the US CDC will no longer be a leader, … which would be a big loss globally, but it's also an opportunity for the CDCs of other countries to step in."

Duquette said the work of the CDC and the PHGB influences her work daily. At Northwestern University, she is encouraged that her genetic counseling students espouse the principles that the CDC and the PHGB were working to advance, like equitable access to healthcare services. While she hopes that one day the public health genomics effort will be resurrected in some form, in its absence, she'll be urging industry, non-profits, and patient advocacy groups to carry the torch of evidence-based genomic screening.

In the present upheaval, Khoury said his former colleagues and collaborators often say that he left government service just at the right time, but he wonders if that's true. "Maybe it was the wrong time to retire because there's so much unfinished business."

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