Newborn Genomic Screening Takes Center Stage at ACMG as Stakeholders Discuss Progress, Concerns

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Premium

NEW YORK – As newborn genomic screening initiatives continue to unfold around the world, the ethical and responsible implementation of the approach into public health sparked vibrant discussions at the American College of Medical Genetics and Genomics (ACMG) annual meeting last week.

In addition to updates from notable newborn genomic screening studies — such as the UK's Generation Study and the Building Evidence and Collaboration for Genomics in Nationwide Newborn Screening (BEACONS-NBS, formerly known as BEACONS) program — during the meeting, clinicians, genetic counselors, and researchers also voiced their opinions and concerns about the approach, which many perceive as becoming inevitable.

"This is probably the next disruption that is coming to genetics," ACMG President Mira Irons, a medical geneticist and pediatrician at Boston Children's Hospital, told a packed audience during an open forum last Thursday, where conference attendees were encouraged to express their comments on the future of newborn screening. Many scientific talks, posters, and a plenary session that formally opened this year's meeting also covered genomic newborn screening.

Recognizing the shifting landscape, and in response to the termination of the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) by the US Department of Health and Human Services (HHS) last year, ACMG has also recently established a newborn screening working group, though details of its action plans remain to be determined.

"What about the poor baby?"

Of the many questions raised by the audience, health equity and follow-up care were among the top concerns. "If the purpose of the newborn screen is to diagnose and treat, [and] we are diagnosing everybody but only treating the rich babies, what about the poor baby?" Aaliya Ahmad, a pediatric genetic counselor at University of Florida, asked during the forum, and was met by applause.

In her practice, 90 percent of patients rely on Florida Medicaid, Ahmad explained, and many of them do not own a car and live hours away from her treatment center. She has witnessed first-hand that some patients were not able to receive appropriate treatment following positive spinal muscular atrophy newborn screening results due to their resource constraints.

When introduced routinely, newborn genomic screening will possibly expand the list of conditions covered by current methods by at least an order of magnitude — from dozens to hundreds, exacerbating people's health equity concerns.

The Generation Study, a partnership between the UK's National Health Service (NHS) and Genomics England, for instance, currently tests for 208 conditions that are linked to 462 genes, with 97 genes to be added in the upcoming weeks, according to David Bick, principal clinician of the newborn genomes program at Genomics England, who presented updates on the project on Wednesday.

"The key point here is that our list is not set in stone," Bick said. "One of the aspects of our program was not only to come up with a list and implement it, but also to figure out how to improve the list over time."

While enrollment for the BEACONS-NBS project has not commenced yet, it published its gene list earlier this year and will focus on 746 genes associated with 777 conditions that are actionable in the first year of life.

"One of the things that we need to be talking about is: How do we develop programs that can grow our newborn screening panels but at the same time assure equitable access to treatment?" said Aaron Goldenberg, a bioethics professor at Case Western Reserve University, who presented on the ethical and social considerations of newborn screening on Wednesday. Goldenberg helps oversee outcomes research and guide the community advisory board for the BEACONS-NBS study.

According to Ingrid Holm, a professor of pediatrics at Boston Children's Hospital and a lead investigator of BEACONS-NBS who also presented on Wednesday, the scope of the project, which is a feasibility pilot, ends when the genomic screening results are routed back to the public health laboratory, which will report positive findings to participants using the state's existing framework.

The study does not encompass follow-up care for participants, though it will establish a so-called Genome Resource Center to support clinicians as needed in understanding and communicating the genomic results. Still, by selecting seven public health programs with various levels of resources to participate — Iowa, Minnesota, New York, Oregon, Puerto Rico, South Carolina, and Texas — BEACONS-NBS aims to investigate the barriers patients might be facing in seeking appropriate care after genomic screening.

The NHS, meanwhile, will pay for treatments associated with the conditions screened for by the Generation Study, Bick said. "We could not put a condition into the Generation Study unless its treatment were available to anybody in the NHS," he noted. "If there was even the possibility of someone living in Bristol not being able to access the care that was available in London, we could not include that condition."

Data storage and sharing

Another discussion point is how genomic data and information should be stored and handled for newborn genomic screening programs. "Over the last five to 10 years, there has been a huge increase in public concern around privacy of all sorts of things, including genetic information," Goldenberg said. "We need to think about data governance and control after screening."

For BEACONS-NBS, there is "no plan" for sharing the data, Holm said. GeneDx, an industry partner for BEACONS-NBS, will hold the study data for five years, she said, and the company will not use the data for anything other than quality assurance purposes. After that, BEACONS-NBS will discard the data unless parents opt in to keep it. "We are hoping that this puts the decisions on data sharing into parents' hands, which will help with their trust in our program," Holm said.

For the Generation Study, anonymized data are stored in a secure research database called the National Genomic Research Library. Besides a newborn's DNA, antenatal and health data are also collected, but only select individuals at Genomics England can access data about participants' identity and contact details. Approved healthcare researchers can study the data within the library, where "researchers bring their tools into the environment, bring out the conclusion, but none of the primary data," Bick said.

As newborn genomic screening sheds lights on a baby's genetic conditions, some researchers also wondered how the information might be handled in accordance with the Genetic Information Nondiscrimination Act (GINA), which prevents discrimination in both health insurance and employment based on genetic information. "I do think that these programs are prompting us to think long and hard about what protections are in place and what gaps we have had to live with since the inception of GINA," Goldenberg said.

Taking Over Resources

Alongside ethical and practical concerns on long-term implementation, evidence of newborn genomic screening's clinical utility for certain conditions continues to grow.

The Generation Study, which began enrollment in 2024, has analyzed around 33,000 cases so far, according to Bick. Of these, about 125 were referred to specialists for follow-up care including metabolic, immunology, neurology, hematology, nephrology, endocrine, and bone conditions that are currently not included in the UK newborn screening panel.

In a talk on Friday, Rebecca Reimers, a clinical geneticist at Rady Children's Institute for Genomic Medicine (RCIGM), gave an update on BeginNGS, a newborn genomic screening program led by RCIGM. Currently in its prospective phase, the study has enrolled over 1,300 participants across different ancestry, ethnicity, and race backgrounds from four states: California, Colorado, Tennessee, and New Jersey.

The study currently screens for 507 severe, childhood-onset conditions. Of the more than 1,000 cases analyzed so far, about 4.5 percent had a positive result, compared to less than 1 percent with the current standard-of-care screening method, Reimers noted.

Meanwhile, of the over 20,000 babies enrolled and analyzed in the GUARDIAN study, which stands for Genomic Uniform-screening Against Rare Diseases in All Newborns, has seen a 3.1 percent positive rate.

According to Brenna Boyd, a genetic counselor and program manager at Columbia University who presented in a separate talk on Friday, the GUARDIAN study currently screens for 321 genes correlating with metabolic, cardiac, hematological, and immunodeficiency conditions for all its participants. The study also screens for 135 optional genes for epilepsy and neurodevelopmental disorders.

Of the positive genomic screening results, more than 420 cases had glucose-6-phosphate dehydrogenase (G6PD) deficiency, an X-linked disorder associated with an increased risk of neonatal hyperbilirubinemia and hemolytic anemia, making it the most frequent finding in the GUARDIAN study. Even so, most babies screened positive for G6PD deficiency were not previously ascertained despite standard-of-care screening standards and known family history, Boyd pointed out.

Despite its clinical utility, newborn genomic screening does demand lots of resources and comes with a steep cost.

Having recruited 76 hospitals among 48 NHS Trusts, with three more sites that are currently onboarding, the Generation Study is sequencing at a speed of roughly 1,500 genomes per week, said Bick, and roughly costs £962 ($1,285) per newborn. "It is a lot of money for a nationwide newborn screening program," Bick said. However, he noted that more than 70 percent of the cost comes from sequencing, and with rapidly declining sequencing cost, Bick believes the cost of newborn genomic screening will also "drop dramatically."

Still, some people fear that genomic newborn programs, and their demand for resources, can compete with the current newborn screening approach. "There are concerns among many newborn screening programs that the idea of adding whole-genome sequencing might divert resources from the current, proven newborn screening programs and actually reduce the benefits of those programs over time," Goldenberg said.

Monica Wojcik, a neonatologist and clinical geneticist at Boston Children's, said she feels that the excitement over genomic newborn screening "has somewhat confused" the dialogue around diagnostic genomic sequencing for sick babies in the NICU.

"I will say that I feel a sense of frustration as a neonatologist and geneticist that we are talking about sequencing availability to healthy babies in the country when there are very sick babies in neonatal intensive care units who still cannot access rapid diagnostic genomic sequencing that could transform their lives," Wojcik told *GenomeWeb*. "Genomic newborn screening is an important issue to address, but remember that these are presumably healthy babies — and the findings on genomic newborn screening are not returning within a timeframe to help babies that are already symptomatic and in the NICU."

Despite the chatter and debate, one thing seems to be increasingly clear to stakeholders: newborn genomic screening is coming, and the shift is not a matter of if but when. "There is an understanding that this is the direction that we are heading in," Carla Cuthbert, branch chief of the newborn screening and molecular biology unit at the US Centers for Disease Control and Prevention, told the audience during the open forum. "We know that it is where we are probably going to wind up being."

Cuthbert said her team has been working with state partners, who are also seeing this as an inevitable trend. According to her, the CDC has recently introduced an NGS training course, which could not fit all of the people who applied to come in. "I hope that we can find a path, and I hope we can figure out how we will take our next steps," she said.