

NIH funds multi-state pilot for genome sequencing in newborn screening

Mass General Brigham

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Mass General Brigham and Ariadne Labs, alongside Boston Children's Hospital, Albert Einstein College of Medicine, the Association of Public Health Laboratories (APHL), Case Western Reserve University, Baylor College of Medicine, and sequencing partners GeneDx and Illumina, today announced the launch of BEACONS (Building Evidence and Collaboration for GenOmics in Nationwide Newborn Screening), the nation's first multi-state genomic newborn screening initiative.

Funded by a \$14.4 million award from the National Institutes of Health (NIH) Common Fund Venture Program, BEACONS will pilot the integration of whole genome sequencing into existing state newborn screening systems. The study will recruit, consent, and enroll up to 30,000 newborns in as many as 10 states over the next three years. If successful, BEACONS will provide the first national evidence that genome sequencing can be implemented responsibly, fairly, and sustainably through public health newborn screening programs. The project is intended as the beginning of a broader national initiative, with the potential to scale beyond the initial 30,000 newborns.

APHL's Newborn Screening and Genetics program directors Jelili Ojodu, MPH and Sikha Singh, MHS, PMP, co-investigators, will provide critical leadership to align the study with state public health laboratories and programs. By working collaboratively with existing newborn screening programs, the initiative aims to build a sustainable roadmap for incorporating genome sequencing into U.S. public health practice.

“For more than 50 years, newborn screening has been a beacon of trust, saving children's lives by analyzing a few drops of blood after birth. BEACONS brings the next generation of hope, giving families the option of genomic screening for hundreds of additional conditions that can be treated from birth, in an era when gene and cell therapies are rapidly expanding treatment possibilities. Together, families who volunteer in participating state programs will be collaborating in a national effort to give every child the brightest start to life.”

Robert Green, MD, MPH, contact and co-lead investigator at Mass General Brigham and Ariadne Labs

BEACONS is led by a multidisciplinary leadership team, combining scientific, clinical and ethical expertise to guide every aspect of the project. "Knowledge is power. Identifying early that a child has a treatable or preventable condition gives the child the best chance for a healthy life. It also means parents won't have to go through the long diagnostic odyssey that is so common in children with rare diseases, sometimes delaying a diagnosis until it is too late to provide treatment that could change the course of their medical outcome," said Ingrid Holm, MD, MPH, co-lead investigator at Boston Children's Hospital.

Early in the study, the team will select which genetic conditions to include in screening with input from rare disease advocates, state public health laboratories, and evidence from prior research studies. "We will be developing a list of conditions which, when identified early, can meaningfully improve the health of a child," said Nina Gold, MD, MS, co-lead investigator at Mass General Brigham.

Parental and community perspectives are integral to BEACONS and will be incorporated throughout the project. Newborns will be enrolled only after parental education and consent, and parents will be invited to participate in a series of surveys and interviews about their experiences to ensure that their voices are heard. A Community Advisory Board will contribute critical insight to help address the complex ethical, legal, privacy, and social considerations of newborn sequencing.

"We will rely on the input of parents and community leaders to make sure that newborn screening expansion is done in a thoughtful and transparent way," said Melissa Wasserstein, MD, co-lead investigator from Albert Einstein College of Medicine and Children's Hospital at Montefiore Einstein. "This is about building trust as much as building science."

Source:

Mass General Brigham