Mass General Brigham

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# First U.S. National Genomic Newborn Screening Initiative Launched with $14.4 Million NIH Award

## ‘BEACONS,’ a multi-state collaboration led by Mass General Brigham and Ariadne Labs, will enroll up to 30,000 newborns to explore adding genomic sequencing in U.S. newborn screening

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

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."

For more information about BEACONS, see www.BEACONSnbs.org.

Read the NIH's announcement: https://commonfund.nih.gov/venture/nbsxwgs/news/nih-venture-program-announces-first-award-beacons-initiative

BEACONS is supported by the following award from the National Institutes of Health: 1OT2OD040029-01

To connect with BEACONS investigators, please email info@BEACONSnbs.org

**QUOTE SHEET**

Leading experts in the ethical, legal, and social dimensions of genomics, including Aaron Goldenberg PhD, MPH, co-investigator at Case Western Reserve University School of Medicine and Stacey Pereira, PhD, co-investigator at Baylor College of Medicine, will oversee outcomes research and guide the Community Advisory Board composed of parents, advocates, community leaders, clinicians, genetic counselors, bioethicists, and data privacy experts.

> "Evaluating the feasibility of genomic newborn screening requires careful attention to ethical, legal, and social concerns," said Aaron Goldenberg, PhD, MPH, co-investigator at Case Western Reserve University School. "Our study will explore ways programs can reflect parental and public values, protect a newborn's privacy, preserve universal access, and enhance the benefits of newborn screening for all families."

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> "APHL is proud to join this partnership in bringing whole genome sequencing to newborn screening in a way that will provide more information to families and save even more babies' lives. We welcome the opportunity to bring innovation to the public health laboratory community and the newborn screening system," said Jelili Ojodu, MPH, senior director, Newborn Screening and Genetics, APHL.

GeneDx will perform genomic sequencing and interpretation for the study, with additional material support from Illumina. These partners bring expertise to the technical aspects of testing, while the overall governance, data protections, and decision-making will remain firmly anchored in the public health and academic sectors which will be guided by parental consent preferences. With advanced genomic technology, the world's largest rare disease dataset, and unmatched clinical expertise, GeneDx has diagnosed more children with rare diseases – and screened more newborns with genomic sequencing – in the U.S.

> "By bringing the power of genomics to the very start of life, we have the potential to shorten, and one day eliminate, the diagnostic odyssey that so many families endure," said Britt Johnson, PhD, Senior Vice President of Medical Affairs at GeneDx. "This collaboration marks an exciting and transformative step toward a future where every child has the opportunity to live a healthier, fuller life."

> "Integrating genomics into healthcare starting at birth to reduce the burden of genetic diseases has been an important concept since the Human Genome Project," said Swaroop Aradhya, PhD, Global Head of Medical & Scientific Affairs at Illumina. "Research conducted by this group will produce critical insights that inform proper use of whole genome sequencing for newborn screening in the US public healthcare setting."

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**About Mass General Brigham**

Mass General Brigham is an integrated academic health care system, uniting great minds to solve the hardest problems in medicine for our communities and the world. Mass General Brigham connects a full continuum of care across a system of academic medical centers, community and specialty hospitals, a health insurance plan, physician networks, community health centers, home care, and long-term care services. Mass General Brigham is a nonprofit organization committed to patient care, research, teaching, and service to the community. In addition, Mass General Brigham is one of the nation's leading biomedical research organizations with several Harvard Medical School teaching hospitals. For more information, please visit massgeneralbrigham.org.