# JAMA (Journal of the American Medical Association) Recognizes GUARDIAN Newborn Genomic Screening Study Among Its Research of the Year

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*Prestigious recognition underscores the impact of large-scale genomic screening in identifying serious childhood genetic conditions in conjunction with traditional newborn screening*

GAITHERSBURG, Md.--(BUSINESS WIRE)--Dec. 15, 2025-- GeneDx (Nasdaq: WGS), a leader in delivering improved health outcomes through genomic insights, today announced that the GUARDIAN (Genomic Uniform-screening Against Rare Disease In All Newborns) study has been recognized as part of *JAMA*'s annual **Research of the Year Roundup**. This highly selective honor is reserved for the most impactful scientific contributions published across all *JAMA* journals.

The recognition highlights the significance of GUARDIAN's peer-reviewed findings, which demonstrate the power of genomic newborn screening to identify serious, actionable childhood conditions more broadly than traditional newborn screening (NBS).

The published analysis from the first 4,000 newborns enrolled in GUARDIAN showed:

- A **72% parental consent rate**, demonstrating strong family interest in genomic screening
- A **3.7% screen-positive rate**
- **92% of screen-positive newborns had a confirmed diagnosis** for a condition **not included in traditional NBS**

Conditions identified through the study include **long QT syndrome**, a rare cardiac condition associated with sudden infant death syndrome (SIDS) that can be effectively managed with beta-blockers; **Wilson disease**, which can be treated with a liver transplant or managed through early zinc supplementation and a low-copper diet;[1] and **severe combined immunodeficiency (SCID)**, which can be treated with stem cell transplantation or gene therapy.[2] While SCID is included in traditional newborn screening, some variants can be missed. In each case, early diagnosis enables timely intervention and meaningfully improves clinical outcomes.

"Being included in JAMA's Research of the Year Roundup is a tremendous honor and reflects the dedication of the entire GUARDIAN team and the families who participate," said Wendy Chung, MD, PhD, GUARDIAN's principal investigator, clinical and molecular geneticist, and the Chief of Pediatrics at Boston Children's Hospital and Harvard Medical School. "GUARDIAN demonstrates that genomic screening can identify serious childhood conditions earlier than ever before. This recognition strengthens our commitment to bringing these benefits to all newborns." Dr. Chung launched the study at NewYork-Presbyterian and Columbia University Irving Medical Center in 2022.

GUARDIAN is one of the largest genomic newborn screening studies in the world and is open to all babies born at NewYork-Presbyterian hospitals in New York City. To date, the study has screened more than 20,000 newborns, with a long-term goal of enrolling 100,000 newborns. The program is designed to evaluate the feasibility, clinical impact, and equity considerations of implementing genomic screening alongside existing public health newborn screening programs.

"This recognition from *JAMA* reinforces what families and clinicians are seeing firsthand – that earlier, more precise genetic diagnosis can fundamentally change the trajectory of a child's life," said Katherine Stueland, President and CEO of GeneDx. "GUARDIAN is setting the standard for how genomics can responsibly and effectively complement traditional newborn screening, ensuring more children get answers before symptoms begin and opening the door to earlier intervention, prevention of disease progression, and true precision genomic medicine."

"The GUARDIAN program reflects the enormous power of the analysis of an individual's genome in the provision of truly personalized medicine — delivered at the beginning of a lifetime," said Rudolph Leibel, M.D., Chief of Pediatric Molecular Genetics at NewYork-Presbyterian/Columbia University Irving Medical Center and a co-author of the study. "These same genetic data can ultimately also serve to inform diagnostic and therapeutic decisions throughout that lifetime, and will at some point soon likely become a standard of care. Dr. Chung and her associates have pioneered a remarkable advance."

Diagnosing rare genetic conditions is often slow and complex, with many children waiting five years or more for an accurate diagnosis – if they receive one at all.[3,4] During this diagnostic odyssey, pediatric patients typically undergo multiple uninformative tests and incur thousands of dollars in healthcare costs before a diagnosis is reached.[5] Earlier genetic diagnosis can fundamentally change this paradigm by enabling timely treatment, guiding clinical management, reducing unnecessary interventions, and connecting families to critical resources and support – ultimately improving outcomes for children and easing the burden on families and health systems.[6,7]

"Next-generation sequencing technologies, including whole genome sequencing, have radically improved the diagnostic yield for pediatric genetic disease and will make an equally deep impact in the context of genomic newborn screening, which we believe will save many children's lives," said Swaroop Aradhya, PhD, Global Head of Medical Affairs at Illumina. "JAMA's recognition of

the GUARDIAN study underscores this profound importance."

The GUARDIAN study is an ongoing collaboration between Columbia University Irving Medical Center and NewYork-Presbyterian, and the New York State Department of Health, with GeneDx providing genomic testing and interpretation and Illumina supporting sequencing technology and scientific collaboration. Together, they are generating critical evidence to inform the future of newborn screening and precision medicine.

**About GeneDx**
GeneDx (Nasdaq: WGS) is the global leader in rare disease diagnosis, transforming the way medicine is practiced by making genomics the starting point for health, not the last resort. We bring together unmatched clinical expertise, advanced technology, and the power of GeneDx Infinity™ – the largest rare disease dataset – built over 25 years from millions of genomic tests and deep clinical insights. This unparalleled foundation powers our ExomeDx™ and GenomeDx™ tests, giving clinicians the highes likelihood of delivering a timely, accurate diagnosis. GeneDx is shaping the future of healthcare by moving the standard of care from sick care to proactive healthcare. While our roots are in rare disease diagnosis, our commitment extends beyond – growing with the families we serve – as a trusted partner at every stage of life. For more information, visit genedx.com and connect with us on LinkedIn, Facebook, and Instagram.

**About GUARDIAN**
GUARDIAN (Genomic Uniform-screening Against Rare Diseases In All Newborns) is a research study conducted at Columbia University in collaboration with NewYork-Presbyterian, the New York State Department of Health, and Illumina, using whole genome sequencing (WGS) provided by GeneDx to screen 100,000 newborns for more than 250 genetic conditions not currently included in standard newborn screening. The goals of the study are to drive earlier diagnosis and treatment to improve the health of the babies who participate, generate evidence to support the expansion of newborn screening through genomic sequencing, and characterize the prevalence and natural history of rare genetic conditions. More information about GUARDIAN can be found at https://guardian-study.org/.

References:


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**Forward Looking Statements**

This press release may contain "forward-looking statements" within the meaning of Section 21E of the Securities Exchange Act of 1934, as amended, and the U.S. Private Securities Litigation Reform Act of 1995. These forward-looking statements generally are identified by the words "believe," "project," "expect," "anticipate," "estimate," "intend," "strategy," "future," "opportunity," "plan," "may," "should," "will," "would," "will be," "will continue," "will likely result," and similar expressions. Forward-looking statements are predictions, projections and other statements about future events that are based on current expectations and assumptions and, as a result, are subject to risks and uncertainties. Many factors could cause actual future events to differ materially from the forward-looking statements in this press release, including but not limited to: (i) our ability to implement plans to accelerate scientific studies and unlock the full potential of precision medicine and other value in the rare disease space, (ii) the risk of downturns and a changing regulatory landscape in the highly competitive healthcare industry, (iii) the size and growth of the market in which we operate, (iv) our ability to pursue our new strategic direction. The foregoing list of factors is not exhaustive. A further list and description of risks, uncertainties and other matters can be found in the "Risk Factors" section of our Annual Report on Form 10-K for the fiscal year ended December 31, 2024 and our Quarterly Reports on Form 10-Q for the fiscal quarters ended March 31, 2025, June 30, 2025, and September 30, 2025 and other documents filed by us from time to time with the SEC. These filings identify and address other important risks and uncertainties that could cause actual events and results to differ materially from those contained in the forward-looking statements. Forward-looking statements speak only as of the date they are made. Readers are cautioned not to put undue reliance on forward-looking statements, and we assume no obligation and do not intend to update or revise these forward-looking statements, whether as a result of new information, future events, or otherwise. We do not give any assurance that we will achieve our expectations.

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