



Sema4|GeneDx To Provide Whole Genome Sequencing and Interpretation Services for Landmark Genomic Newborn Screening Study

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The largest study of its kind in North America, GUARDIAN is expected to improve population health by identifying more than 250 preventable and treatable conditions, many of which are not covered today by standard newborn screening

STAMFORD, Conn., Oct. 05, 2022 (GLOBE NEWSWIRE) -- [Sema4](#) (Nasdaq: SMFR), a health insights company, today announced it is a partner in the recently launched GUARDIAN study (Genomic Uniform-screening Against Rare Diseases In All Newborns), a collaboration among premier non-profit, academic, government, and industry organizations. GUARDIAN is using whole genome sequencing (WGS) to screen 100,000 newborns for more conditions than those 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.

Newborn screening identifies conditions that can affect a child's long-term health or survival. Early detection, diagnosis and intervention can prevent death or disability and enable children to reach their full potential.¹ The conditions GUARDIAN will screen for were identified based on select criteria, including: median age of onset before 5 years of age, greater than 90% probability of the condition developing based on the genetic result, effective treatments, and/or a well-established natural history of the condition.

Sema4|GeneDX was selected to provide WGS and interpretation services given its industry-leading curated dataset. Sema4|GeneDX has performed approximately 400,000 clinical exomes to date, which increases the ability to provide more answers to more patients and serves as the foundation of its genome informatics and reporting platform.

"We are relentlessly focused on accelerating the adoption and use of genomic information to impact the lives of as many people as possible, particularly newborns and children," said [Katherine Stueland](#), President and CEO, Sema4. "As the first commercial laboratory to launch a rapid whole genome sequencing offering, to address broad unmet needs for early diagnosis, participation in this study is an important step forward for healthcare and in delivering on our goal to sequence once, analyze forever."

More than 180 hospitals across the country currently use the Sema4|GeneDX rapid exome and genome offerings.

"We'd like to thank the families in advance for volunteering to participate in GUARDIAN," said Dr. [Paul Kruszka](#), Chief Medical Officer of GeneDx at Sema4. "The results from this study will help us understand the true impact sequencing at birth can have on newborns and their families in comparison to the current standard of care, particularly as we'll evaluate clinical outcomes in addition to the psychosocial effect on families."

The study is open to all babies born in the GUARDIAN partner health system in New York City regardless of race, income, or health insurance coverage. Dr. Kruszka will further discuss the study during a roundtable with other industry and academic colleagues at the inaugural International Conference on Newborn Sequencing (IcoNS), which is being held from October 5-6, 2022, in Boston, Massachusetts.

About Sema4|GeneDx

Sema4|GeneDx is a patient-centered health intelligence company dedicated to advancing healthcare through data-driven insights. Sema4 is transforming healthcare by applying AI and machine learning to multidimensional, longitudinal clinical and genomic data to build dynamic models of human health and defining optimal, individualized health trajectories. Centrellis™, our innovative health intelligence platform, is enabling us to generate a more complete understanding of disease and wellness and to provide science-driven solutions to the most pressing medical needs. Sema4 believes that patients should be treated as partners, and that data should be shared for the benefit of all. For more information, please visit [sema4.com](#) and connect with us on [LinkedIn](#), [Twitter](#), [Facebook](#), and [Instagram](#).

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¹ Source: Centers for Disease Control and Prevention