New Research from Sema4|GeneDx Highlights the Importance of Rapid Exome Sequencing for Diagnosing Mitochondrial Diseases in the NICU

December 5, 2022

Data underscores the value of mitochondrial DNA testing and the opportunity to improve health outcomes for critically ill newborns with early medical intervention

STAMFORD, Conn., Dec. 05, 2022 (GLOBE NEWSWIRE) -- Sema4 (Nasdaq: SMFR), a health insights company, today announced the results of a new study on mitochondrial diseases in newborns. The findings support the addition of mitochondrial DNA (mtDNA) testing to rapid exome sequencing, showing it can lead to earlier diagnoses and more immediate potential changes in clinical management. The research evaluates the incidence of mitochondrial disease caused by both nuclear DNA (nDNA) and mtDNA in 966 infants in the NICU who received both rapid exome sequencing and mtDNA sequencing and deletion testing concurrently.

Mitochondrial diseases are chronic, genetic disorders that occur when mitochondria fail to produce enough energy for the body to function properly. These can affect multiple organ systems at different ages. Mitochondrial diseases are individually rare but, collectively, as this study shows, this group of disorders is not uncommon in clinical settings.

In the study conducted by Sema4|GeneDx, nearly 10% of infants referred for exome sequencing plus mtDNA testing were diagnosed with a mitochondrial disorder, accounting for 29% of the overall diagnostic cases. 1.3% of diagnoses were made via mtDNA testing. The findings are especially important for infants presenting with the most common symptoms, including lactic acidosis, seizures, and hypotonia.

“Our study shows that including both rapid exome sequencing and mt-DNA sequencing in the NICU will increase the diagnostic yield for many infants,” said Dr. Paul Kruszka**,** Chief Medical Officer of GeneDx at Sema4. “Mitochondrial disease is difficult to diagnose as it can affect many different parts of the body and clinicians may not be looking for it in the NICU. Although there is no cure for mitochondrial disease, earlier diagnoses can lead to a more immediate change in medical management and significantly reduce the diagnostic odyssey for critically ill pediatric patients.”

With the largest curated database of 400,000 clinical exomes and more than 100,000 mtDNA genomes sequenced – as compared to approximately 57,000 in the public database – Sema4|GeneDx is a leader in understanding the genetic etiologies that cause mitochondrial disease. This is due in part to analysis of the entire mitochondria genome, as opposed to the more common approach to analyze pathogenic variants alone.

Sema4|GeneDx is presenting the findings from this research at the Hot Topics in Neonatology conference in National Harbor, MD, which runs from December 4-7, 2022.

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.

Media contact
Radley Moss
radley.moss@sema4.com