Study Demonstrates the Utility of Exome Sequencing for Diagnosing Autism Spectrum Disorders

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Research from 19,000 patients with Autism Spectrum Disorders found approximately 90% had prior negative tests, likely contributing to a longer time to diagnosis

Data will be presented at the Child Neurology Society (CNS) Annual Meeting on October 13, 2022

STAMFORD, Conn., Oct. 13, 2022 (GLOBE NEWSWIRE) -- Sema4 (Nasdaq: SMFR), a health insights company, is presenting research today on autism spectrum disorders (ASD) at the Child Neurology Society (CNS) Annual Meeting. The research underscores the positive outcomes of exome analysis for individuals with ASD, supporting the use of a broader genetic testing approach to reach a faster diagnosis. This study of almost 19,000 individuals is one of the few to examine the genetic basis of autism and current guidelines around screening and testing.

The study evaluated exome analysis for individuals with ASD against current testing methods and provided proof points that exome sequencing should be a first-tier test for ASD. The American Academy of Pediatrics recommends screening for ASD between the ages of 18-24 months. The median ages at testing in this analysis were 6.4 and 8.4 years for both isolated and syndromic ASD; exome sequencing provided higher diagnostic rates of 7.9-21%. Step-wise testing likely contributed to a delayed molecular diagnosis, as nearly 90% of the 18,911 individuals in the study with ASD had at least one prior negative test.

The study also suggests completing exome sequencing early on in a child's life can help identify other disorders that a child with ASD may be at risk for, including epilepsy, as that affects up to a third of people with autism, but only 1-2% of the general population. Overall, this genetic testing approach helps break down an individual's genetic etiology, leading to better, more informed health interventions and outcomes.

Additionally, this study revealed a high number of emerging genes not previously connected to autism. Initial analysis identified 1,337 emerging genes, 113 of which were upgraded to disease-causing starting in 2015. Moreover, 60% of genes identified in this study as associated with ASD were not found in the Simon Foundation Autism Research Initiative (SFARI) Gene online database – an evolving resource for the autism research community centered on genes implicated in autism susceptibility.

"This dataset illustrates that relying on exome testing as a proactive testing approach will help more patients and clinicians reach faster diagnoses," said Dr. Paul Kruszka, Chief Medical Officer of GeneDx at Sema4. "As we see in our research, updating the standard of care to include exome sequencing as the front-line test has the promise to help support children's development."

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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