Sema4|GeneDx collaborates on new research demonstrating genome and exome sequencing deliver more diagnostic certainty than multi-gene panels

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Exome and genome tests report 30% fewer inconclusive results and a higher diagnostic yield compared to multi-gene panel tests; research to be presented at American Society of Human Genetics Annual Meeting

STAMFORD, Conn., Oct. 26, 2022 (GLOBE NEWSWIRE) -- Sema4 (Nasdaq: SMFR), a health insights company, has collaborated on research debunking a commonly held belief that exome and genome sequencing deliver more inconclusive results than multi-gene panels. The study demonstrates that using exome or genome sequencing results in a significantly lower rate of variants of uncertain significance (VUS) compared to panel-based testing, underscoring the importance of genomic sequencing for improving diagnoses and health outcomes. It is one of more than a dozen studies Sema4|GeneDX is either presenting, or has participated in, being showcased at the American Society of Human Genetics (ASHG) Annual Meeting.

The study of more than 1.5 million tests shows genomic sequencing generates more diagnostic certainty than panel-based testing. Led by Dr. Heidi Rehm, Chief Genomics Officer in the Department of Medicine at Massachusetts General Hospital, the research compared VUS rates across multi-gene panels of varying sizes and both genome and exome tests. It demonstrated that both genome and exome testing have lower VUS rates compared to traditional gene panels, with trio-based genomic testing having the fewest reported VUS rates.

Specifically, the research revealed that genomic tests reported 30% fewer inconclusive results compared to multi-gene panel tests (23% vs 33%) and a higher diagnostic yield (18% vs 10%). The study also supported testing parents in addition to affected individuals as doing so increases the probability of a genetic answer from 15% to 20%.

“We are proud to work with Dr. Rehm and the other study partners on this research illustrating the power of genomic sequencing for improving diagnostic certainty,” said Dr. Paul Kruszka, Chief Medical Officer of GeneDx at Sema4. “Genome and exome sequencing have improved diagnostic yields and contribute greatly to ending the diagnostic odyssey, which is a goal of affected families, healthcare providers and payers.”

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