Sema4|GeneDx Commends New Evidence-Based Guidelines from the National Society of Genetic Counselors Recommending Exome Sequencing as a First-Tier Genetic Test for Unexplained Epilepsies

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Guideline adoption can help end the diagnostic odyssey experienced by many families

STAMFORD, Conn., Oct. 31, 2022 (GLOBE NEWSWIRE) -- Sema4 (Nasdaq: SMFR), a health insights company, supports the new National Society of Genetic Counselors guidelines recommending exome sequencing as a first-tier test, to be prioritized over multi-gene panels for all individuals with unexplained epilepsy, and applauds the American Epilepsy Society for endorsing these guidelines. This is the first evidence-based guideline for genetic testing for individuals with unexplained epilepsy, and the implications are vast, as a significant amount of unexplained epilepsy has a genetic cause[1]. In many cases, identifying a molecular genetic cause of epilepsy can have implications for treatment and management, including avoiding, stopping, or initiating specific medication or diet recommendations and clinical trial eligibility.

These recommendations mirror guidelines for other neurodevelopmental disorders such as intellectual disabilities and developmental delays in that they recommend genome and exome sequencing as first-tier tests. A child with neurodevelopmental disorders is likely to wait[2] more than six years on average for a genetic diagnosis; accrue more than $10,000 in additional health costs; and undergo more than five uninformative tests. These new guidelines demonstrate the importance and advantage of ordering exome sequencing first rather than chromosomal microarray analysis panel testing; in which diagnostic rates are highest[3] for whole genome sequencing (48%), followed by whole exome sequencing (24%). Chromosomal microarray analysis has the lowest diagnostic rate (9%).

Leveraging one of the world's largest data sets of more than 400,000 clinical exomes, we have seen first-hand the power of genomic sequencing for improving diagnostic certainty and believe these new guidelines will unlock insights and improve patient care.

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

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[1] Source: Smith, L., Malinowski, J., *et al*. (2022). Genetic testing and counseling for the unexplained epilepsies: An evidence-based practice guideline of the National Society of Genetic Counselors. *Journal of genetic counseling*, 10.1002/jgc4.1646. Advance online publication. https://doi.org/10.1002/jgc4.1646

[2] Source: Soden, S. E., Saunders, C. J., *et al.* (2014). Effectiveness of exome and genome sequencing guided by acuity of illness for diagnosis of neurodevelopmental disorders. *Science translational medicine*, *6*(265), 265ra168. https://doi.org/10.1126/scitranslmed.3010076

[3] Source: Sheidley, B. R., Malinowski, J., *et al.* (2022). Genetic testing for the epilepsies: A systematic review. *Epilepsia*, *63*(2), 375–387. https://doi.org/10.1111/epi.17141