GeneDx Rare Disease Data Demonstrates Utility in Drug Candidate Identification for Common Diseases

February 16, 2023

Research published in Nature Genetics underscores the role of genetic insights in the drug discovery and development process

STAMFORD, Conn., Feb. 16, 2023 (GLOBE NEWSWIRE) -- GeneDx Holdings Corp. (GeneDx) (Nasdaq: WGS), a leader in delivering improved health outcomes through genomic and clinical insights, continues to demonstrate how the study of rare disease can improve understanding of the biology of common disorders.

Research published in the October 2022 issue of Nature Genetics, to which GeneDx contributed data and authorship, yields insights into the role of two-pore-domain potassium channels (K2P) in rare disease and explores the role genes might play in the newly described developmental delay with sleep apnea (DDSA). Such connections better enable the exploration of pharmacological solutions to common diseases impacting wide swaths of the general population.

Sleep apnea, characterized by abnormal, interrupted breathing during sleep, is a common disorder that represents a global public health burden thought to affect up to one in five people worldwide and approximately 30 million Americans. Poor sleep quality often results in decreased quality of life and increased risk of comorbidities, such as cardiovascular disease, diabetes and depression.

The study describes DDSA, a newly identified condition caused by de novo gain-of function variants clustering in the X-gate motif of the TASK-1 channel encoded by *KCNK3*. TASK-1 is a K+ channel implicated in the control of breathing, but its link with sleep apnea remains poorly understood. Functional studies indicate that TASK channel inhibitors may be effective at inhibiting abnormal channel activity caused by these variants, illuminating the underlying biology of sleep apnea and pointing to possible therapeutic options for patients with DDSA and common sleep apnea.

“These findings indicate how research into rare disease can uncover findings that are potentially applicable to much broader segments of the general population and also represent important progress for the pharmacological management of sleep apnea,” said Dr. Paul Kruszka, Chief Medical Officer of GeneDx. “Sleep apnea’s high prevalence and the imperfection of existing therapies mean there is sizeable appetite for developing new treatment approaches.”

Through its robust rare disease data set, GeneDx is working to advance drug discovery and development, and accelerate research to help identify therapeutic targets for common diseases. With the deep genomic information available from GeneDx’s exome and genome testing, combined with its clinical insights, the company is able to help connect biopharmaceutical companies with patients to improve clinical trial enrollments and recruitment and improve patient outcomes by connecting them to therapies.

About GeneDx (formerly Sema4)
GeneDx delivers personalized and actionable health insights to inform diagnosis, direct treatment and improve drug discovery. The company is uniquely positioned to accelerate the use of genomic and large-scale clinical information to enable precision medicine as the standard of care. GeneDx is at the forefront of transforming healthcare through its industry-leading exome and genome testing and interpretation, fueled by one of the world’s largest rare disease data sets. For more information, please visit genedx.com and connect with us on LinkedIn, Facebook, and Instagram.

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