KCNT1 EPILEPSY FOUNDATION LAUNCHES DIGITAL NATURAL HISTORY STUDY IN PARTNERSHIP WITH INVITAE CIITIZEN

Novel approach aims to accelerate the analysis of patient data for rare disease drug development

CONTOOCOOK, NH (Feb. 25, 2022) – The KCNT1 Epilepsy Foundation today announced in partnership with Invitae, the launch of its Digital Natural History Study (NHS) using Invitae's Ciitizen technology, a patient-mediated platform that allows patients to collect and store their medical information. It is a novel approach that supports the nonprofit patient advocacy group's work to accelerate the development of treatments for those with a KCNT1 gene mutation.

The new digital model with the Ciitizen platform provides the ability to rapidly acquire and analyze all data points across a larger cohort of KCNT1 patients, which is critical to accurately understanding the disease. It aims to augment traditional studies by amassing retrospective data that ultimately reduces the time needed for research and drug development.

"We are proud to be at the forefront of rare disease research and collaborate with Invitae to redefine the drug development process for KCNT1," said Justin West, MD, co-founder and Director of Clinical Medicine for the KCNT1 Epilepsy Foundation. "These analytics are expected to provide insights to improve patient care in the short term, and to identify endpoints for drug development and trial design for the long term."

Mutations in the KCNT1 gene can result in neurodevelopmental issues and intractable epilepsy, presenting differently in children: some infants have clinical seizures within days or months of birth, while others have seizures that go undetected for years, leading to a lengthy diagnostic odyssey. Observations of how these symptoms of individuals with a KCNT1 mutation progress over time, or the natural history, are essential to improve care and inform drug development in the effort to move rapidly toward disease-modifying treatments.

With the digital NHS on Invitae's Ciitizen platform, medical records can be collected on the patient's behalf - always patient directed and consented - from providers, hospitals and specialty clinics designated by the patient during a short onboarding process. Parents and caregivers of KCNT1 patients will have direct access to their consolidated, digitized data through one online portal. It uniquely summarizes their experience, which can facilitate second opinion consultations, insurance claims and eligibility determinations, and be used to inform providers across disciplines to improve patient care. With patient consent, anonymized data can be extracted and made available to researchers and pharmaceutical companies investing time in understanding KCNT1, thereby providing them with access to a common database of natural history data for cross-patient analysis.

"We are honored to support the KCNT1 Epilepsy Foundation to find treatments for patients suffering from this disease," said Robert Nussbaum, M.D., chief medical officer at Invitae.

"Invitae's Ciitizen technology is intended to generate patient-consented, real-world data that can be utilized by advocacy groups to help patients understand their disease and can support the longitudinal natural history studies needed by biopharma in planning clinical trials for treatments that more effectively target the disease."

The model also reduces barriers to patient participation inherent in the traditional NHS by eliminating the cost of site visits and travel requirements, dramatically reducing the cost per patient and providing access to study data. It allows for the participation of substantially more, and potentially all, of a patient population, a critical factor for rare diseases like KCNT1 that have fewer than 1,000 patients worldwide. By leveraging collaborative data practices and innovative technology with this Ciitizen Digital Natural History Study among its network of patients, the KCNT1 Epilepsy Foundation is opening a new chapter for those living with the rare disease.

About the KCNT1 Epilepsy Foundation
Founded in 2019, the KCNT1 Epilepsy Foundation is a 501(c)(3) parent-led organization dedicated to facilitating research and drug development through material and financial support to reduce the time needed to find novel therapies, and ultimately a cure, for patients living with KCNT1-related epilepsy. For more information, visit kcnt1epilepsy.org.

About Invitae
Invitae Corporation (NYSE: NVTA) is a leading medical genetics company, whose mission is to bring comprehensive genetic information into mainstream medicine to improve healthcare for billions of people. Invitae's goal is to aggregate the world's genetic tests into a single service with higher quality, faster turnaround time and lower prices. For more information, visit the company's website at invitae.com.

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