Cambridge-led natural history study identifies MTRFR/C12orf65 deficiency to improve diagnosis and therapy development

This research will potentially show a method that may be quicker and better at identifying baseline disease phenotypes to support research and clinical trials.

NEW YORK, NEW YORK, UNITED STATES, April 14, 2023 /EINPresswire.com/ -- Cambridge-led retrospective natural history study funded by the Hereditary Neuropathy Foundation identifies MTRFR/C12orf65 deficiency with a method to improve diagnosis, patient care, and therapy development.

At the Department of Clinical Neurosciences, University of Cambridge Rita Horvath, MD, PhD (Director of Research in Rare Neurogenetic Diseases) has opened a study collecting retrospective data on rare disease patients to assist the Hereditary Neuropathy Foundation (HNF), and its Therapeutic Research In Accelerated Discovery (TRIAD) research partners to advance a gene therapy to treat these patients with MTRFR/C12orf65 deficiency, a rare Charcot-Marie-Tooth disease.

MTRFR/C12orf65 deficiency is a rare complex axonal hereditary motor neuropathy that presents with a series of diseases that cause blindness and cognitive impairment. MTRFR/C12orf65-related disease may result in CMT6, Leigh Syndrome, Spastic Paraplegia-55, Behr Syndrome and COXPD.

Dr. Horvath and her research team developed a retrospective natural history study to collect and review the symptoms of patients who have a genetic defect in the MTRFR/C12orf65 gene.

“We felt it was vital to identify as many patients as possible to learn as much as we can about

this variant of CMT and to enable a better understanding of the disease. So far, Dr. Horvath's method has identified 33 patients who were previously reported with MTRFR/C12orf65 deficiency internationally", says Allison Moore, CEO, HNF

"By using Dr. Horvath's method, perhaps we can find additional patients in the US, UK and worldwide, which has been extremely difficult. Patients are most likely not getting diagnosed due to the vast genetic testing panels, which may or may not include these genes. Furthermore, neurologists and other medical practitioners would not typically know how to order the appropriate test.

"It is our hope that our approach will enable a more robust characterization of the disease caused by MTRFR/C12orf65 mutations. We are looking forward to seeing as many patients as possible with this condition worldwide. The final aim of our study is to use the data to develop new treatments for this progressive neurodegenerative condition" says Rita Horvath, MD, PhD

Patients will also be invited to enroll in the HNF patient registry, Global Registry for Inherited Neuropathies (GRIN). The registry's goal is to acquire, record, and analyze patient-reported data and associated genetic reports, Electronic Health Records (EHRs) and clinical notes to identify the burden, diagnostic journey, and prevalence of disease that will aid scientists in their work toward finding a cure.

The data collected has helped HNF and its partners in industry, academia, and government identify previously unknown genotype/phenotype correlations, uncover important comorbidities such as pain or respiratory issues, and target research spending based on actual patient need and likelihood of success. By including patients with MTRFR/C12orf65-related disease, additional data will be captured to gain more insight on direct patient and caregiver reported outcomes for research and clinical trials.

Since whole genome sequencing (WGS) is more commonly becoming 'standard of care' in the first-line diagnosis of neurogenetic disease including mitochondrial disease and CMT, we predict that WGS will identify more patients with MTRFR/C12orf65 deficiency, even with less characteristic phenotype.

To support patient diagnosis, HNF's CMT Genie was developed to assist patients and healthcare providers access to affordable options for genetic testing.
If you think you or a family member might have MTRFR/C12orf65 deficiency you can contact HNF for genetic testing. If you have MTRFR/C12or65 deficiency and would like to participate in Dr. Horvath's research, please contact the study team directly.

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About the Research Team at the University of Cambridge

The Cambridge Clinical Mitochondrial Research Group studies the genetic basis and mechanisms of mitochondrial diseases, including mitochondrial eye diseases, and other rare inherited neurological disorders, such as Charcot-Marie-Tooth (CMT) disease and spastic ataxias. Our aim is to provide a precise diagnosis and to discover new treatments. Our research group is embedded within the Department of Clinical Neurosciences and the MRC Mitochondrial Biology Unit, both based on the Cambridge Biomedical Campus.

Mitochondrial diseases are genetic disorders that impair the energy production in our cells, affecting about 1 in 5,000 people in the UK. They cause progressive disease that often leads to significant disability and sometimes a reduced life expectancy.

Our research programme aims to better understand the mechanisms of rare inherited neurological diseases, and to develop new and effective treatments for patients affected with mitochondrial disease and CMT through investigator-led experimental medicine studies, novel gene therapy approaches, and clinical trials in partnership with the pharmaceutical industry.

For more information, visit us on Twitter or on our website: https://www-neurosciences.medschl.cam.ac.uk/mitocamb/.

About The Hereditary Neuropathy Foundation

HNF is the Patient-Centered CMT Advocacy group driven by patients, for patients. HNF developed the Therapeutic Research in Accelerated Discovery (TRIAD) as a collaborative effort with academia, government, and industry to develop treatments for CMT. The Global Registry for Inherited Neuropathies (GRIN) has provided critical patient-reported data to TRIAD partners since 2013. This data was also critical in supporting the HNF-hosted Patient-Focused Drug Development (PFDD) meeting to inform the FDA about the burden of CMT and the critical, unmet need for treatment in 2018. In 2022, HNF launched the CMT Genie Program to accelerate genetic diagnosis and offer virtual genetic counseling to patients to explain the sometimes complicated genetic reports, as well as to increase the number of participants in GRIN.

For more information, visit us at www.hnf-cure.org, or on Facebook, Twitter, Instagram, Vimeo and YouTube.

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