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Today HCU Network America announces the recipient of their first Cobalamin G research grant, awarding The Research Institute of the McGill University Health Centere, to identify potential treatment for Cobalamin G (CblG) deficiency. The research, led by Dr. Brian Gilfix, aims to identify the use of a specific antibiotic to allow full expression of the protein in patients with stop codon mutations. Dr. Gilfix is an Associate Professor in the Department of Medicine and Division of Experimental Medicine. He is also the program director for residency training in Medical Biochemistry at the McGill University Health Centre.

Cobalamin G Deficiency (cblG) is an inborn error of metabolism which disrupts the utilization of vitamin B12 to convert homocysteine to methionine in the remethylation pathway. Typically, patients present during their first year of life with metabolic crisis, anemia, failure to thrive, seizures or developmental delay. Patients have elevated plasma homocysteine and low methionine levels. Unlike combined cobalamin disorders such as cobalamin C, patients do not have elevated levels of methylmalonic acid; making early detection on newborn screening tests nearly impossible for this group of rare disorders. Unless there is a family history of this disorder, patients are not diagnosed until they become symptomatic. The disease prevalence is unknown for this disorder.

The current treatment consists of hydroxocobalamin injections (a very specific form of B12) and betaine anhydrous. If a patient is symptomatic at the time of diagnosis, treatment will not reverse the side effects. Many patients despite current treatment standards still become symptomatic.

According to the principal investigator, Dr. Brian Gilfix, the project aims to pilot a new strategy for treating cblG disorder and other inborn errors of cobalamin metabolism due to stop mutations for which treatments are now currently limited. It is known that aminoglycoside antibiotics allow the cell to overcome the stop codons by allowing the insertion of a random amino acid, and thereby expressing a full-length protein. It is our intention to demonstrate that in the culture cells we are able to restore at least partially cblG function and demonstrate an alternative means of treating cblG disorders.

Board and Cobalamin Steering Committee member Dana Hunt said:" Words cannot begin to describe how thrilled we are to finally be able to fund this potentially lifesaving research grant. Carson and others continue to defy the odds but are in desperate need of new medical treatments to manage this rare disorder. We are beyond grateful for the support of our families, friends and the entire HCU community."

Cobalamin Steering Committee Lead and Board member for HCU Network America, Brittany Parke said "It is an exciting day for our community to be able to help fund an incredible project

for such a rare disorder. The implications to improve the quality of life for cblG patients is incredible. I am honored to have been apart of the process to extend this grant to Dr. Gilfix. Finally, all our hard work is beginning to come together to improve the future for those we love with this disorder."

HCU Network America thanks their community of supporters who contributions made this grant possible.

About HCU Network America:
HCU Network America is a 501c(3) non-profit organization founded in 2016 dedicated to helping patients and their families affected by Homocystinuria (HCU) and related disorders. The mission of the organization is to inform and provide resources for patients and families, create connections, influence state and federal policy, and support advancement of diagnosis and treatment for HCU and related disorders.