



MILLION DOLLAR BIKE RIDE GRANT WINNERS

2019

RESEARCH TOPIC:

Towards new therapeutic treatments for the hyperinsulinism/hyperammonemia syndrome (HI/HA)

Dr. Thomas Smith and his research team are using a wide array of techniques to develop drugs to treat the hyperinsulinism/hyperammonemia (HI/HA) syndrome. HI/HA is one of the more common genetic hyperinsulinism subtypes. Patients suffering from HI/HA have a higher basal level of insulin in their blood and over secrete insulin upon consumption of protein. This disease not only affects the pancreas but is also linked to seizures and developmental problems. These pathologies are due to mutations in glutamate dehydrogenase (GDH) that affect how the enzyme is regulated. While diazoxide is effective at controlling insulin secretion, it comes with side effects and does not target GDH and therefore does not treat the other affected organs.

Our goal is to target GDH directly to treat all symptoms associated with HI/HA throughout the body. To this end, we have been creating the various HI/HA mutants to better understand their subtle differences. We are collaborating with a pharmaceutical company and have performed more than a half million screens looking for active compounds. The company is helping us to analyze and optimize activity. As soon as possible, we will test the best compounds in whole cell and animal assays.

AWARD RECIPIENT:

Thomas Smith,
PhD

HOME INSTITUTION:

University of Texas
Medical Branch

AWARD AMOUNT:

\$72,014



RIDING FOR RESEARCH!



Penn Medicine
Orphan Disease Center