MILLION DOLLAR BIKE RIDE GRANT WINNERS

2018

RESEARCH TOPIC:

Vitamin E Supplementation in Hyperinsulinism/Hyperammonemia Syndrome

Hyperinsulinism/hyperammonemia (HI/HA) syndrome is the 2nd most common type of congenital HI. It is caused by activating mutations in glutamate dehydrogenase (GDH). Patients with HI/HA syndrome have fasting and protein-induced hyperinsulinemic hypoglycemia due to GDH over-activity in the pancreatic beta cells, which can be treated with diazoxide. However, these patients also have other medical problems due to GDH over-activity in other cell types, for which there is no treatment: hyperammonemia (high blood ammonia levels), seizures, and developmental delays. Vitamin E has been tested in human cell lines and in mice with activating GDH mutations and shows potential promise as a treatment for HI/HA syndrome. This is a two-part pilot clinical study involving children and adults with HI/HA syndrome to 1) determine if oral Vitamin E supplementation is well-tolerated and 2) determine if oral Vitamin E supplementation is effective in reducing hypoglycemia, hyperammonemia, and seizures. This pilot study will potentially lead to a larger and longer-term clinical trial evaluating Vitamin E treatment of HI/HA syndrome.

AWARD RECIPIENT:
Amanda Ackermann, MD, PhD

HOME INSTITUTION:
Children's Hospital of Philadelphia

AWARD AMOUNT:
$84,080

Riding for research!

Penn Medicine
Orphan Disease Center