



# 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.

I am riding to increase awareness and research funding for patients with rare and complex diseases like hyperinsulinism, who need new treatment options.

## RIDING FOR RESEARCH!

AWARD RECIPIENT:  
Amanda Ackermann,  
MD, PhD

HOME INSTITUTION:  
Children's Hospital  
of Philadelphia

AWARD AMOUNT:  
\$84,080



Penn Medicine  
Orphan Disease Center