



# PILOT HIHA RESEARCH GRANT

## 2019

### RESEARCH TOPIC:

#### **Novel Mouse Model to Investigate Pathophysiology of Hyperinsulinism/Hyperammonemia Syndrome (HIHA)**

HIHA is not only a disease of hypoglycemia. Patients with HIHA also have high blood ammonia levels, seizures, and neurodevelopmental differences that currently are not well-understood and do not have any specific treatments. It has been difficult to study each of these features of HIHA in patients because each one can affect the other features.

This project will create a mouse model of HIHA that has the disease-causing mutation only in specific cell types. We will first use this mouse model to study the effects of the HIHA mutation in the brain, but additional future studies will also be able to use this mouse model to study effects of the HIHA mutation in other organs including the liver and kidneys.

Just as diazoxide is used to treat hyperinsulinism, which is an effect of the HIHA mutation in pancreatic beta cells, once we understand what causes the other features of HIHA (high blood ammonia levels, seizures, and neurodevelopmental differences), we will be able to develop treatments for these aspects of the disease.

**AWARD RECIPIENT:**  
Amanda Ackerman,  
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**HOME INSTITUTION:**  
Children's Hospital  
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**AWARD AMOUNT:**  
\$70,000