
Research Focus Areas:

Glucose Transporter Type 1 Deficiency Syndrome (Glut1DS): One \$61,901 grant is available to support a project that will lead to a deeper understanding of this disease so it can be diagnosed earlier and treated more easily and more effectively.

Glut1 Deficiency Syndrome is a rare genetic condition that impairs brain metabolism. It is caused by variants in the *slc2a1* gene, which encodes the glucose transporter protein type 1 (GLUT1). GLUT1 is the principal transporter of glucose and also moves other important sugars across the blood-brain barrier. Impaired glucose transport associated with Glut1 Deficiency creates a metabolic crisis in the brain and often results in a range of neurological symptoms such as seizures, speech and movement disorders, and developmental delays. Not all patients experience all symptoms, and there is a wide spectrum of severity. Symptoms may change and evolve over time.

Potential areas of emphasis for this RFA may include but are not limited to: open source resource development (cell lines, assays, functional studies, etc.); GLUT1 at the blood brain barrier; brain glucose metabolism; ketogenic diets as metabolic therapies; basic science to better understand underlying disease mechanism; identification of new biomarkers and outcome measures to be used in future clinical studies; and understanding how the involvement of GLUT1 in different diseases can lead to the development of better treatments for Glut1 Deficiency. Projects with novel concepts and collaborative/team approaches are especially encouraged.

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