UGDH - Grant Amount: $40,000

Jamuar syndrome is caused by recessive loss-of-function mutations in the UGDH gene and is a neurodevelopmental disorder characterized by developmental and epileptic encephalopathy. Affected individuals present with global developmental delay, speech impairment, intellectual disability and epilepsy. Other features include axial hypotonia, movement disorder and feeding difficulties. Brain imaging may show non-specific findings including delayed myelination, enlarged ventricles, cerebral atrophy, cerebellar atrophy, and thin corpus callosum. Facial features may include protruding ear lobes, ptosis, deep set eyes, epicanthal folds, short flat philtrum, full lower lip and pointed chin. The UGDH Foundation is offering one $40,000 grant for research that will facilitate the development of an effective treatment for Jamuar syndrome. This would include, but is not limited to, biomarkers, model development, characterization of the natural history or therapeutic approaches.