Title: Longitudinal Studies of Brain Structure and Function in MPS Disorders

IRB# 2010-046
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Synopsis:

This is a five year prospective longitudinal study carried out at multiple centers in North America. The purpose of this study is to examine changes to the central nervous system (CNS) in both structure and function over time in up to 75 patients who have been diagnosed with mucopolysaccharides (MPS) type I, II or VI.

Patients will have an annual 3-Tesla MRI of the brain, age specific neuropsychological testing of cognitive ability, memory attention, spatial ability, visual motor ability, adaptive functions, behavior, and quality-of-life. A blood sample for biomarkers and urine sample for glycosaminoglycan (GAG) testing will also be collected.

The primary objective of the study is to identify abnormalities of CNS structure and function over time in both treated and untreated patients diagnosed with MPS I, II and VI by collecting brain MRI sequences, and to track disease progression over time. The secondary objective is to identify those quantitative measures that are most sensitive to change and to examine the degree to which independent variables, such as age at first treatment, severity of disease, types of medical abnormalities, mutation, medical events, and sensory abnormalities, have an impact on both functional and structural outcomes as well as on quality-of-life.