Application Title: MRI signatures of cervical spine alterations in mucopolysaccharidosis type I (MPS I)

PI: Igor Nestrasil
Institution: University of Minnesota

Mucopolysaccharidosis type I (MPS I) is associated with structural abnormalities affecting all regions of the vertebral column. In the cervical spine region such abnormalities can result in cervical spinal cord (CSC) compression with irreversible and often catastrophic neurological damage. Despite the improvement of the standard-of-care for MPS I patients and a normal or an almost normal life expectancy the risk of C-spine related injury in MPS I patients remains high and further increases with age. The recent cases of unexpected cervical spinal cord injuries in MPS I patients emphasize the importance of meaningful and reliable measures of C-spine related injury risk. The search for a magnetic resonance imaging (MRI) marker that properly predicts surgical outcomes is, however, challenging due to the anatomical localization of CSC. Motivated by the desire to comprehensively evaluate microstructural CSC alteration, our group has recently developed and tested an innovative MRI protocol in MPS I subjects that overcomes technical challenges related to CSC advanced imaging. The main objective of our project is to identify the neuronal and functional signatures of spinal and spinal cord alteration in MPS I, relate them to neurological outcomes, and establish sensitive markers that will be utilized in an evaluation of the C-spine related risk, surgical planning, and in clinical trials.