

“Neurodegeneration with Brain Iron Accumulation: Research Cohort Development”

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Abstract

This project focuses on cohort development in a group of rare disorders which share the feature of abnormal brain iron. "Neurodegeneration with Brain Iron Accumulation" (NBIA) comprises many different disorders, six of which have been genetically defined. While the OHSU research registry and biorepository has been in existence for many years, its scope and productivity have grown enormously in the last 3-4 years as international collaborations have been forged, the pace of gene discovery has picked up, basic science interest in the disorders has increased, and patient advocacy groups have found strength in numbers. Along with this success and growth have come challenges and opportunities. The OCTRI catalyst funding will allow us to 1) develop and begin testing a clinical outcome test battery, and 2) investigate a tantalizing connection between a newly-discovered form of NBIA and Rett syndrome.