INTRODUCTION
The University of Rochester Batten Center has engaged in research on the Natural History of all forms of Batten Disease. “Natural History” refers to the usual course of development of a disease. High quality natural history data are important for evaluating treatments in rare diseases. This poster summarizes our work in CLN3 disease (JNCL, Juvenile Batten Disease).

RATING SCALE: THE UBDRS
Beginning in 2002, we developed and have used the Unified Batten Disease Rating Scale (UBDRS) to measure key features of Batten Disease. The UBDRS was designed to be used in all forms of Batten Disease.

The UBDRS consists of 4 domains:
1) Physical Impairment
2) Seizures
3) Mood and Behavior
4) Functional Capability

It also captures information about age-at-onset of different symptoms.

Cognition is assessed with separate tools.

The UBDRS has allowed us to measure key features of Batten Disease in order to determine how affected individuals change with age, and which aspects contribute most to disability.

We have used the UBDRS to evaluate over 150 individuals with some form of NCL, including 110 individuals with CLN3 disease. We have performed a total of 400 evaluations, with as many as 15 annual evaluations in an individual.

In addition, we have used data from our natural history work to develop a staging system for potential use in CLN3 disease.

PHYSICAL IMPAIRMENT
Physical impairment progresses continuously over the course of the disease.

SEIZURES
In most individuals with CLN3 disease, most seizures are “grand mal” (generalized tonic-clonic), are relatively infrequent, and respond well to medications. Other types of seizures do occur. Fewer than 10% of individuals are on >2 medications for seizures.

PROPOSED STAGING SYSTEM
Staging can facilitate division into groups for possible clinical trials and provides a broad sense of disease progression.

COGNITION
Cognitive decline progresses over the course of the disease and may precede physical impairment.

SYMPTOM ONSET AGES
Typical CLN3 disease starts with vision loss, followed by cognitive and behavior problems, then seizures and finally motor decline.

FUNCTIONAL CAPABILITY
Functional capability declines with age and correlates with Physical Impairment. Functional capability assessed included school, chores, game playing, and activities of daily living.

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