Using PEDI-CAT to Assess Functional Capabilities in CLN3 (Batten) Disease
Heather R. Adams PhD, Lauren Winschel, Erika F. Augustine, MD, MS on behalf of the University of Rochester Batten Center
Batten Disease Center of Excellence at the University of Rochester School of Medicine and Dentistry, Rochester, NY USA

Background and Study Aims:
- CLN3 disease is an autosomal recessive lysosomal storage disease (LSD) with childhood-onset. Symptoms include vision loss, motor decline, dementia, and seizures.
- The Pediatric Evaluation of Disability Inventory – Computer Adaptive Test (PEDI-CAT) evaluates functional capacity in young persons (approx. age 1-21 years).
- The aims of the present study were to (1) Evaluate parent-reported PEDI-CAT ratings of functional abilities in CLN3 disease, and (2) Examine associations between PEDI-CAT parent ratings and a clinician-based capability assessment from the Unified Batten Disease Rating Scale (UBDRS).

Methods:
- Parents of 6 children and young adults with genetically confirmed CLN3 disease completed the PEDI-CAT. Assessments were completed in-person at the 2017 annual meeting of the Batten Disease Support and Research Association (BDSRA) or administered via a phone interview within 2-3 months after the meeting.
- All parents gave permission for their child’s participation in this study; a waiver of assent was granted for children <18 and a consent waiver was granted for adults with CLN3 disease because of limited capacity due to dementia.
- All research activities were conducted through a research protocol approved by the University of Rochester Research Subjects Review Board (RSRB00020390).

Fit Scores
Higher fit scores (>2.00) suggests ‘misfitting’ response pattern (unexpected pattern of scores)

<table>
<thead>
<tr>
<th>Daily Activities</th>
<th>Mobility</th>
<th>Soc. / Cog.</th>
<th>Resp.</th>
</tr>
</thead>
<tbody>
<tr>
<td>mean</td>
<td>-0.53</td>
<td>-1.66</td>
<td>-0.73</td>
</tr>
<tr>
<td>sd</td>
<td>0.91</td>
<td>0.86</td>
<td>2.82</td>
</tr>
<tr>
<td>min</td>
<td>-1.59</td>
<td>-2.67</td>
<td>-6.18</td>
</tr>
<tr>
<td>max</td>
<td>0.91</td>
<td>-0.46</td>
<td>1.54</td>
</tr>
</tbody>
</table>

All 4 PEDI-CAT Domains Correlate with UBDRS Capability Score

Conclusions
- Floor effects are seen on the PEDI-CAT T scores at a moderate level of disease severity, among children and young adults with CLN3 disease. This suggests that age-adjusted scores have limited utility for evaluating meaningful changes in adaptive function over time, in this disease population.
- Item-level scores and Standard Scores (which are not age-adjusted) show a pattern of decreasing functional capacity with increasing age. It is unclear whether item content in some PEDI-CAT domains is fully relevant to characterizing disease phenotype and progression in CLN3 disease. Interestingly, an earlier non-CAT version of PEDI was adapted for another lysosomal storage disease (Pompe-PEDI; see Henly et al., Pediatr Rehabil 2003 Apr-Jun;6(2):77-84) to best capture all aspects of disease impact on functional capacity. A similar item-level analysis approach may be needed for CLN3 disease.
- Despite these limitations, the PEDI-CAT provided good initial external validation of the UBDRS Total Capability Score.

Acknowledgements: This research was sponsored by Abeona Therapeutics and by the University of Rochester Child Neurology Endowment Fund. We thank the Batten Disease Support and Research Association (BDSRA) for permitting the research to be conducted at the BDSRA 2017 Annual Conference in Pittsburgh, PA (July 7-9, 2017). We especially thank the children and families who have participated in this research.