The 5th Annual Workshop on Krabbe Disease

July 31 - August 1, 2012
Tamarack Lodge – Holiday Valley Resort
Ellicottville, New York

Funded in part by HRSA (Maternal and Child Health Bureau) and Hunter’s Hope Foundation

July 30, 2012: Arrival and Dinner (7:00pm in the Laurel & Snowledge Conference Rooms)

Session 1 (8:30 – 12:30 in Bobsled Conf Rm): July 31, 2012

Neuropathology of treated animals and humans with Krabbe disease

<table>
<thead>
<tr>
<th>Time</th>
<th>Activity</th>
</tr>
</thead>
<tbody>
<tr>
<td>8:00</td>
<td>Breakfast – Crystal Conference Room</td>
</tr>
<tr>
<td>8:30-8:40</td>
<td>Welcome, review of prior workshops, and introductions</td>
</tr>
<tr>
<td>8:40-9:00</td>
<td>Treatment of twitcher using AAVrh10 (Mohammad Rafi)</td>
</tr>
<tr>
<td>9:00-9:10</td>
<td>Discussion</td>
</tr>
<tr>
<td>9:10-9:30</td>
<td>Treatment of twitcher with mesenchymal stem cells (Bruce Bunnell)</td>
</tr>
<tr>
<td>9:30-9:40</td>
<td>Discussion</td>
</tr>
<tr>
<td>9:40-10:00</td>
<td>Treatment of twitcher with HCT and gene therapy (Jacqueline Hawkins-Salsbury)</td>
</tr>
<tr>
<td>10:00-10:10</td>
<td>Discussion</td>
</tr>
<tr>
<td>10:10-10:30</td>
<td>Break</td>
</tr>
<tr>
<td>10:30-10:50</td>
<td>Treatment of Krabbe dogs with HCT (Charles Vite)</td>
</tr>
<tr>
<td>10:50-11:00</td>
<td>Discussion</td>
</tr>
<tr>
<td>11:00-11:20</td>
<td>Neuropathology of untreated humans with Krabbe disease (Reid Heffner)</td>
</tr>
<tr>
<td>11:20-11:40</td>
<td>MRI findings in children transplanted for Krabbe disease (Jim Provenzale)</td>
</tr>
<tr>
<td>11:40-12:00</td>
<td>Neuropathology of children transplanted for Krabbe disease (Evan Snyder)</td>
</tr>
<tr>
<td>12:00-12:30</td>
<td>Discussion</td>
</tr>
<tr>
<td>12:30 – 1:30</td>
<td>Lunch – Laurel &amp; Snowledge Conference Rooms</td>
</tr>
</tbody>
</table>
Session 2 (1:30-5:30 in Bobsled Conf Rm); July 31, 2012
Issues with Newborn Screening for Krabbe disease

1:30-2:50  Statewide NBS for Krabbe disease

1:30-1:50  Newborn Screening Techniques: Comparative
effectiveness study on GALK testing on blood spots
(Dieter Matern)

1:50-2:10  Update on NBS for KD in NY: two tier testing by
MS/MS and genotyping—results and operational costs
(Joe Orsini)

2:10-2:30  Update on proposed statewide NBS for KD (Illinois, Joel
Charrow; Missouri, Bryce Heese; New Jersey, Shari Fallet;
New Mexico, Brenda Romero)

2:30-2:50  Discussion

2:50-3:10  Break

3:10-4:00  How reliable are most genotypes in predicting phenotypes?

3:10-3:30  Genotype/phenotype correlations from symptomatic
patients in the world-wide registry (Denise Kay)

3:30-3:50  Array CGH improves detections of mutations in the
GALK gene in Krabbe disease (Madhuri Hegde)

3:50-4:00  Discussion: What about expanded genotype analysis –
modifiers? (Rod Howell)

4:00-4:20  Confirmatory GALK testing: Can enzyme assays predict
phenotype? Can NYS risk groups be applied to other labs?
(Group Discussion)

4:20-4:50  Is any test reliably predictive of phenotype?

4:20-4:40  How useful are neurodiagnostic tests in predicting
phenotype? (Patti Duffner)

4:40-4:50  Discussion: Psychosine? Where are we? (Joe Orsini)

4:50-5:30  Roundtable discussion: What are community pediatricians to
do when faced with a positive newborn screen for Krabbe
disease from a supplemental newborn screening kit? (Rod
Howell, Mike Watson, Dieter Matern, Bill Slimak, Marc
Patterson)

7:30  Dinner – Laurel & Snowledge Conference Rooms

Session 3 (9:00 – 12:00 in Bobsled Conf Rm); August 1, 2012
Basic and Clinical Research Updates

8:00  Breakfast – Crystal Conference Room

9:00-9:20  Psychosine Toxicity: Strategies for protection and discoveries
of how to make it worse (Mark Noble)

9:20-9:40  Endogenous psychosine induced dysregulation of
oligodendrocyte differentiation and survival (Inderjit Singh)

9:40-10:00  Discussion and Break

10:00-10:20  Global CNS gene delivery platform in non-human primates
utilizing self-complementary AAV9 vectors (Steven Gray)

10:20-10:30  Discussion

10:30-10:50  ERT with and without HCT in LSDs (Jeanine Utz)

10:50-11:10  Alternative approaches to HSCT using reduced intensity
regimens for babies and children with lysosomal storage
disorders (Joanne Kurtzberg)

11:10-11:20  Discussion

11:20-11:40  Remyelination of the CNS: why is it important and how can it
be achieved? (Ian Duncan)

11:40-12:00  Discussion

12:00  Adjournment & Lunch – Laurel Snowledge Conference Rooms