



HUNTER JAMES KELLY
RESEARCH INSTITUTE



2014 Medical Symposium **Krabbe Disease**

Newborn Screening | Clinical Research | Basic Science

July 27 – 29, 2014

Holiday Valley Resort – Ellicottville, NY

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2014 Medical Symposium Agenda

Sunday July 27, 2014

**Arrive at Holiday Valley Resort, Ellicottville NY
Dinner: 6pm – Sunrise Room, Holiday Valley Lodge**

Monday July 28, 2014

Breakfast: 7-8am – Sunrise Room, Holiday Valley Lodge

**Session I: NBS: Diagnosis & Predicting Long-Term Outcome
8am – 12pm – Foxfire Conference Room, Holiday Valley Lodge**

8:00 – 8:05

Welcome and Introduction (5 min)	Tom Langan
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8:05 – 8:35

The New York State Experience (2006-2014) (30 min) Overall Update, Protocol, GALC, Mutation, STFU, LFTU, MRI, Psychosine <i>Moderator: Tom Langan</i>	Joe Orsini
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8:35 – 9:05

The Missouri Experience (2012-2014) (30 min) NBS GALC, 2 nd Tier Mutation Results, Confirmatory GALC Results & Patient Outcomes <i>Moderator: Tom Langan</i>	Sharmini Rogers Patrick Hopkins
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9:05 – 9:20

Illinois NBS Program (15 min) Protocol and Plan for Follow Up - Current Status <i>Moderator: Tom Langan</i>	George Dizikes
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9:20 – 9:35

New Jersey NBS Program (15 min) Protocol and Plan for Follow Up - Current Status <i>Moderator: Tom Langan</i>	Scott Shone
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9:35 – 9:55

Discussion (20 min)

9:55 – 10:15

Family Perspective (20 min) How Krabbe Newborn Screening Impacts Families	Krabbe Families
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Break 10:15-10:30am

10:30 – 11:10

The Multi-State Krabbe Consortium (30 min) World Wide Registry, The NY-Model, Expansion, Protocol, Follow Up Discussion (10 min)	Tom Langan
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11:10 – 11:50

Overview of Krabbe Newborn Screening (30 min) Current Status and Action Items Discussion (10 min)	Tom Langan
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Lunch 12-1pm – Sunrise Room, Holiday Valley Lodge

2014 Medical Symposium Agenda

Monday July 28, 2014

Session II: Clinical Research: Diagnosis & Long Term Follow Up

1pm – 5pm – Foxfire Conference Room, Holiday Valley Lodge

1:00 – 1:30

Psychosine Measured in Blood - Is this a useful marker to monitor patients with abnormal newborn screening results for Krabbe Disease? (20 min)	Dieter Matern
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Discussion (10 min)

1:30 – 1:55

Spectrum of <i>GALC</i> Variants Identified by Newborn Screening for Krabbe Disease in New York State (15 min)	Denise Kay
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Discussion (10 min)

1:55 – 2:20

MRI Scans in Newborns with Myelin Disorders – Update (15 min)	Jim Provenzale
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Discussion (10 min)

2:20 – 2:45

Long Term Outcomes of Transplantation in Krabbe Patients (15 min)	Maria Escolar
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Discussion (10 min)

2:45 – 3:10

New York State – Short Term and Long Term Follow Up for Krabbe (15 min) Electronic Data Capture System	Joe Orsini
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Discussion (10 min)

Break 3:10 – 3:25pm

3:25 – 3:55

Newborn Screening Translation Research Network (20 min) Longitudinal Pediatric Data Resource	Amy Brower
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Discussion (10 min)

3:55 – 4:25

Rare Disease Clinical Research Network (20 min) Lysosomal Disease Network	Chet Whitley
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Discussion (10 min)

4:25 – 4:55

Unsolved Leukodystrophies: Needs, Toolkit and Models for Centers of Excellence (20 min)	Adeline Vanderver
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Discussion (10 min)

4:55 – 5:25

Clinical Care Center of Excellence for Leukodystrophies at HJKRI (20 min)	Tom Langan
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Discussion (10 min)

Dinner 6pm – Sunrise Room, Holiday Valley Lodge

2014 Medical Symposium Agenda

July 29, 2014

Breakfast: 7am – 8am – Sunrise Room, Holiday Valley Lodge

Session III: Understanding Krabbe Disease

8:00 – 10:00 am – Foxfire Conference Room, Holiday Valley Lodge

8:00 – 8:05

Welcome and Introduction (5 min)	Larry Wrabetz
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8:05 – 8:30

MyRF Promotes Myelination (15 min)	Yungki Park
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Discussion (10 min)

8:30 – 9:00

Modeling the Natural Evolution of Pelizaeus Merzbacher Disease (20 min)	Ian Duncan
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Discussion (10 min)

9:00 – 9:35

From Molecular Pathology to Regenerative Medicine: Combining Neuroprotection with Gene and Cell Therapies in Krabbe Disease (25 min)	Ernesto Bongarzone
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Discussion (10 min)

9:35 – 10:10

Trafficking and Processing of GalC mutants (25 min)	Daesung Shin
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Discussion (10 min)

Break 10:10 – 10:30am

Session IV: Improving Cord Blood Transplant and Discovering Additional Therapies

10:30 – 12:30 pm

10:30 – 11:05

Translational Approaches for Krabbe Disease Gene Therapy (25 min)	Steve Gray
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Discussion (10 min)

11:05 – 11:40

Updates on Gene Therapy with AAVrh10 in the Mouse and Dog models of Krabbe Disease (25 min)	David Wenger
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Discussion (10 min)

11:40 – 12:15

Novel Cell Therapy Approaches to Treat Krabbe Disease (25 min)	Joanne Kurtzberg
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Discussion (10 min)

12:15 – 12:30

Discussion and Closing Remarks (15 min)	Larry Wrabetz Tom Langan
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Lunch: 12:30pm