



# Center for NeuroGenetics

UNIVERSITY of FLORIDA

Presents

## Myotonic Dystrophy and the Brain:

### *Brainstorming DM*

**December 15, 2014**

8:00 ***Continental Breakfast—Room LG 110, MBI***

8:50 Welcoming remarks— Laura Ranum, PhD University of Florida  
DeWeese Auditorium

***Session 1*** Clinical features of DM

***Chair: Maurice Swanson, PhD***

9:00-9:20 Richard Moxley, MD University of Rochester  
*"Observations of Brain Dysfunction in DM1 & DM2 Patients: Challenges To Consider in Preparing for Future Trials of Treatment of CNS Manifestations"*

9:20-9:40 Tetsuo Ashizawa, MD University of Florida  
*"Myotonic Dystrophy as a Brain Disorder – therapeutic principle"*

9:40-10:00 S.H. Subramony, MD University of Florida  
*"Loose thoughts on loose connections"*

10:00-10:20 ***Break—Room LG 110 , MBI***

***Session 2*** Neuropathology and Imaging

***Chair: Richard Moxley, MD***

10:20-10:40 Anthony Yachnis, MD University of Florida  
*"Neuropathology of two cases: DM2 and FXTAS"*

10:40-11:00 Brent Clark, MD, PhD University of Minnesota  
*"The CNS neuropathology of myotonic dystrophy".*

11:00-11:20 John Day, MD, PhD Stanford University  
*"Imaging DM1 and DM2"*

11:20-11:40 David Vaillancourt, PhD University of Florida  
*"Neuroimaging in Movement Disorders: A specific example of a progression marker in Parkinson's Disease"*

11:40-12:00 Peg Nopoulos, MD University of Iowa  
*"Brain Imaging in DM1: Lessons From Huntington's Disease"*

12:00-1:20 ***Lunch—Room LG 110, MBI***

*Session 3 Lessons from Other Diseases*

*Chair: Laura Ranum, PhD*

- 1:20-1:40 John Day, MD, PhD Stanford University  
*"Therapeutic Strategies in Spinal Muscular Atrophy"*
- 1:40-2:00 David Borchelt, PhD University of Florida  
*"Molecular mechanisms of ALS"*
- 2:00-2:20 Lee Sweeney, PhD University of Pennsylvania  
*"Therapeutic opportunities and challenges in the muscular dystrophies"*
- 2:20-2:40 Andrew Judge, PhD University of Florida  
*"Skeletal muscle ultrastructural alterations, atrophy and weakness"*
- 2:40-3:00 Damaris Lorenzo Duke University  
*"Anchored No More! Ankyrin-B Promotes Organelle Transport and Axonal Elongation"*
- 3:00-3:20 **Break—Room LG 110, MBI**

*Session 4 Molecular Changes in the CNS*

*Chair: Tetsuo Ashizawa, MD*

- 3:20-3:40 Eric Wang, PhD Massachusetts Institute of Technology  
*"Genomic approaches to understanding the DM brain"*
- 3:40-4:00 Andy Berglund, PhD University of Oregon  
*"Splicing sensitivities in myotonic dystrophy type I differ across events and tissues"*
- 4:00-4:20 Christopher Pearson, PhD The Hospital for Sick Children, Canada  
*"Repeat Instability in DM"*
- 4:20-4:40 Maurice Swanson, PhD University of Florida  
*"Mbnl conditional knockout models for the myotonic dystrophy brain"*
- 4:40-5:00 Laura Ranum, PhD University of Florida  
*"Repeat associated non-ATG (RAN) translation in neurologic disease"*
- 6:30 **Gala Dinner—Invitation only**

## December 16, 2014

8:00 **Continental Breakfast –Room LG 110, MBI**

**Session 5** *Functional Analysis of Mouse Models* **Chair: Lee Sweeney, PhD**  
DeWeese Auditorium, MBI

8:40-9:00 Jada Lewis, PhD University of Florida  
*"The missing link in modeling tauopathy"*

9:00-9:20 Mario Gomes-Pereira, PhD French Institute of Health and Medical Research  
*"Neuroglial miscommunication in DM1 brains through dysregulation of glutamate transport"*

9:20-9:40 Timothy Ebner, MD, PhD University of Minnesota  
*"Abnormalities in Cerebral Cortical Circuits in Mouse Models of Myotonic Dystrophy."*

9:40-10:00 Seiji Nishino, MD, PhD Stanford University  
*"Sleep phenotype characterization in mouse models of myotonic dystrophy"*

10:00-10:20 **Break –Room LG 110, MBI**

**Session 6** *Therapeutic Strategies* **Chair: Andy Berglund, PhD**  
Room LG 110, MBI

10:20-10:40 Matthew Disney, PhD The Scripps Research Institute , Florida  
*"Design of small molecules targeting repeating transcripts"*

10:40-11:00 Steven Zimmerman, PhD University of Illinois  
*"Rational Design of Small Molecules to Target Expanded Trinucleotide Repeat Sequence in Myotonic Dystrophy"*

11:00-11:20 Todd Golde, MD, PhD University of Florida  
*"Using rAAV vectors to manipulate phenotypes in preclinical neurodegenerative disease models"*

11:20-11:40 Charles Thornton, MD University of Rochester  
*"Getting ready for antisense trials"*

11:40-12:40 Advocacy Panel/Q&A