TUESDAY, MARCH 29, 2016

4:00-6:15 p.m.  AIM Check-in and Poster Boards available to hang posters
5:15-6:15 p.m.  Welcome Reception and Check-in continues
6:30 p.m.  Dinner
7:15 p.m.  Opening Remarks – David Lynch, M.D., Ph.D. CHOP
7:20 p.m.  Keynote Address
Jennifer Raymond, M.D., Stanford University School of Medicine
Neural learning rules in the cerebellum

WEDNESDAY, MARCH 30, 2016

Theme 1 – Cerebellar Circuitry and its role in Ataxia

7:30-8:30 a.m.  Continental Breakfast

8:30-8:55 a.m.  Key Note: Timothy J. Ebner, M.D., Ph.D., University of Minnesota
Cerebellum Acts as a Forward Internal Model and Computes Sensory Prediction Errors

Theme 1 Session Chair: David Lynch, M.D., Ph.D. Children’s Hospital of Philadelphia

9:00-9:20 a.m.  *Lin Hong, Ph.D. Children’s Hospital of Philadelphia
Cerebellar circuit deficits in a frataxin-deficient mouse model of Friedreich’s ataxia

9:25-9:45 a.m.  *Abigail Person, Ph.D., University of Colorado School of Medicine
A feedback pathway from the cerebellar nuclei to cerebellar cortex regulates sensory gain

9:50-10:10 a.m.  *Jonathan Schisler, Ph.D. University of North Carolina at Chapel Hill
The characterization of a mouse model of SCAR16 generated with CRISPR gene editing

10:15-10:45 a.m.  Morning Break

10:50-11:10 a.m.  *Andreia Castro, University of Minho, Portugal
Serotonergic signaling suppresses SCA3 pathogenesis: tackling the when and how

11:15-11:35 a.m.  Liliana S. Mendonça, PharmD, PhD, University of Coimbra
Transplantation of Neural Stem Cells as a therapeutic strategy in Machado-Joseph disease

11:40-12:00 p.m.  Tycho Hoogland, Ph.D., The Netherlands
Complex Spike Asynchrony in Cerebellar Ataxia: Cause and/or Consequence?

12:00-1:00 p.m.  Lunch

Theme 2 – Biological Basis of Cerebellar Degeneration
1:00-1:25 p.m.  **Key Note:** Dineke Verbeek, Ph.D. University Medical Center Groningen  
*The role of opioid signalling in cerebellar neurodegeneration*

**Theme 2 Session Chair:** Harry T. Orr, Ph.D. University of Minnesota, Minneapolis

1:30-1:50 p.m.  Andrew P. Lieberman, M.D., Ph.D. University of Michigan  
*Non-cell autonomous mechanisms in SBMA*

1:55-2:15 p.m.  Olaf Riess, M.D., University of Tuebingen, Germany  
*Mechanisms in SCA3*

2:20-2:40 p.m.  *Jana Schmidt, University of Tuebingen, Germany  
*Assessing in vivo the role of ubiquitination in the pathogenesis of SCA3*

2:45-3:15 p.m.  *Afternoon Break*

3:20-3:40 p.m.  *Jordi Magrane, Ph.D., Weill Cornell Medical College  
*Cellular mechanisms in FRDA,*

3:45-4:05 p.m.  *Sara Duarte-Silva, PhD, University of Minho, Portugal  
*TUDCA reduces neuroinflammation and improves motor symptoms in a transgenic mouse model of Machado-Joseph disease*

4:10-4:30 p.m.  Laura Ranum, Ph.D. University of Florida, Gainesville  
*Identification of novel spectrin, beta, non-erythrocytic 2 (SPTBN2) mutations in a large patient cohort*

4:30-4:45 p.m.  *“Hot Chair” Junior Poster Presenters Group 1 – 2 minutes each*

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5:00-6:30 p.m.  Scientific Poster Session A (Wine and Cheese)

6:30 p.m.  **Networking Dinner on your own**

**THURSDAY, MARCH 31, 2016**

**Theme 3 – Genomic and Genetic Approaches to cerebellar function and ataxia and Epigenetics**

7:30-8:30 a.m.  **Continental Breakfast**

8:30-8:55  **Key Note:** John Hardy, Ph.D., University College London, Institute of Neurology  
*Genomic analysis of neurodegeneration*

**Theme 3 Session Chair:** Alexandra Durr, M.D., Ph.D., Hopital de la Salpetriere, Paris, France

9:00-9:20 a.m.  *Yogesh K. Chutake, Ph.D. University of Oklahoma HSC  
*Epigenetic Silencing of the FXN gene in Friedreich ataxia*
9:25-9:45 a.m. *Vikram Khurana, MBBS, Ph.D., Brigham and Women’s Hospital and Harvard Medical School, Boston, MA
Neurodegenerative Proteinopathies: Genome Scale investigations

9:50-10:10 a.m. *Bing Yao, Ph.D., Emory University, Atlanta, GA
5-hydroxymethylcytosine-mediated Epigenetic Dysregulation in Cerebellar Degeneration

10:15-10:30 “Hot Chair” Poster Presenters Group 2 – 2 minutes each
Matthew Scaglione Theme 2-Poly SCAs Medical College of Wisconsin
Vincenzo Gennarino Theme 2-SCA1 Baylor College of Medicine
Wei-Ling Tsou Theme 3-SCA6 Wayne State University School of Medicine
Percy Tumbale Theme 3-AOA1 Natl Inst Environmental Health Sciences
Edgardo Rodriguez Theme 4-SCA1 University of Florida
Charles Isaacs Theme 5-FRDA Children’s Hospital of Philadelphia

10:35 a.m.-12 p.m. Scientific Poster Session B (Coffee, Tea, Snack)

12:00 noon Official Photo of all AIM Attendees will be taken at this time

12:00-1:00 p.m. Lunch

**Theme 4 – Therapeutic Strategies for ataxia: Translational Research**

1:00-1:25 p.m. **Key Note:** Beverly Davidson, Ph.D., Children’s Hospital of Philadelphia
Progress in advancing RNAi for the spinocerebellar ataxias to human application

**Theme 4 Session Chair:** Albert LaSpada, M.D., Ph.D., University of California, San Diego

1:30-1:50 p.m. *Elisabetta Soragni, Ph.D. The Scripps Research Institute, La Jolla, CA
Identification of biomarkers for clinical trials in Friedreich’s ataxia

1:55-2:15 p.m. *Eric Wang, Ph.D., University of Florida, Gainesville, FL
Post-transcriptional regulation of gene expression in neuromuscular disease

2:15-2:45 p.m. Afternoon Break

2:50-3:10 p.m. *Hayley McLoughlin, Ph.D., University of Michigan, Ann Arbor, MI
Antisense Oligonucleotide Therapy for SCA3

3:15-3:35 p.m. *Marek Napierala, Ph.D., University of Alabama at Birmingham
A next generation perspective on the molecular mechanism of Friedreich’s ataxia

3:40-4:00 p.m. Richard A. Gatti, M.D., UCLA School of Medicine
ATM mutations (splicing, nonsense and large genomic deletions) and therapeutic opportunities

5:15-6:15 p.m. Poster Session for Patients and Families

6:30 p.m. Dinner

7:15 p.m. The Patient/Family Perspective
FRIDAY, APRIL 1, 2016

Theme 5 – Therapeutic strategies for ataxia: Clinical Trials, Outcome Measures and Drug Development

7:30-8:30 a.m.    Continental Breakfast

8:30-8:55    Key Note: Merit Cudkowicz, M.D., M.Sc., Massachusetts General Hospital
             Clinical Trials in ALS and Ataxia – Lessons From Other Fields

Theme 5 Session Chair: Gulin Oz, Ph.D., University of Minnesota

9:00-9:20 a.m.    Kurt Fischbeck, M.D., NINDS, NIH
             Clinical trials in neurodegenerative disease

9:25-9:45 a.m.    *Fanny Mochel, M.D., Ph.D., University Hospital La Salpêtrière, Paris
             MRS studies in ataxias

9:45-10:15 a.m.    Morning Break

10:20-10:40 a.m.   *Kathrin Reetz, M.D., Aachen University, Germany
             Multimodal Imaging in Ataxias

10:45-11:05 a.m.    Robert Wilson, M.D., Ph.D. University of Pennsylvania Medical Center
             High throughput screening in drug development

11:10-11:30 a.m.    *Christophe Lenglet, University of Minnesota
             Cross-sectional and longitudinal diffusion MRI and MRS of the spinal cord in Friedreich’s Ataxia

11:30-11:45 p.m.    Closing Remarks – David Lynch, University of Pennsylvania
             AIM 2016 Adjourns

2-5 p.m.    “Birds of a Feather” Sessions at the Annual Ataxia Conference
             Junior Investigators are welcome to attend these small groups

As of 2/16/16 - sah

*indicates a Junior Investigator