

**7<sup>th</sup> Ataxia Investigators Meeting (AIM) 2018**  
**“From Bench to Bedside: Entering a New Era of Ataxia Therapies”**  
**Philadelphia Downtown Marriott, Philadelphia, PA**  
(Schedule as of 3-6-18-2:45 p.m.)

**MONDAY, April 2, 2018**

- 3:30-6:30 p.m. AIM Check-in and Poster Boards available to hang posters
- 5:30-6:30 p.m. Welcome Reception & Opening Remarks  
Gülin Öz, PhD, University of Minnesota
- 6:30 p.m. *Buffet Dinner at Marriott Hotel Salon G*

**TUESDAY, April 3, 2018**

- 7:30-8:30 a.m. *Continental Breakfast in Salons I-L*

**Theme 1 – Basic cerebellar function and dysfunction**

**Session Chair:** Marija Cvetanovic, PhD, University of Minnesota

- 8:30-9:05 a.m. **Key Note:** Chris de Zeeuw, MD, PhD, Erasmus Medical Center  
*Common Default Pathways in Ataxia*
- 9:05-9:30 a.m. Wade Regehr, PhD, Harvard University  
*Purkinje cell synapses*
- 9:30-9:55 a.m. Baljit Khakh, PhD, University of California, Los Angeles  
*Cells that tile your brain: astrocyte roles in neural circuits*
- 9:55-10:15 a.m. *Morning Break*
- 10:15-10:40 a.m. Esther Becker, PhD, University of Oxford  
*Stem cell-derived models to elucidate the disease mechanisms in spinocerebellar ataxia*
- 10:40-11:05 a.m. \*Adam Avery, PhD, University of Minnesota  
*Molecular and cellular consequences of SCA5 mutations in  $\beta$ -III-spectrin*
- 11:05-11:30 a.m. Ray Truant, PhD, McMaster University  
*Reactive Oxygen Stress and DNA Damage Define ATM as a Node in Age-Onset Neurodegenerative disease*
- 11:30-11:50 p.m. Theme 1 Open Discussion led by Theme Chair
- 11:50-1:00 p.m. *Lunch at Marriott in Salons I-L*



## WEDNESDAY, April 4, 2018

7:30-8:30 a.m. Continental Breakfast in Salons I-L

### **Theme 3 – Translational research and disease models**

**Session Chair:** Patricia Maciel, PhD, University of Minho

8:30-9:05 a.m. **Key Note:** Huda Zoghbi, MD, Baylor College of Medicine  
*Lessons we continue to learn from studying SCA1*

9:05-9:30 a.m. \* Maria do Carmo Costa, PhD, University of Michigan  
*Drug and target discovery for Spinocerebellar ataxia type 3*

9:30-9:55 a.m. Lisa Ellerby, PhD, Buck Institute for Age Research  
*Disease Modeling and Therapeutic Targets in Huntington's Disease*

9:55-10:15 a.m. Theme 3 Open Discussion led by Theme Chair

10:15-10:35 a.m. “Hot Chair” Junior Poster Presenters Group 2  
(3 minutes each, 2 min talk + 1 min questions)

Chandrakanth Edamakanti, PhD	SCA1	Northwestern University
Luis Pereira de Almeida, PhD	SCA3	University of Coimbra
Vincenzo Gennarino, PhD	PUM1	Columbia University
Amy Salovin, MS	FRDA	Children's Hosp of Philadelphia
Jorge da Silva, PhD Candidate	SCA3	University of Minho
Francesca Tiano, PhD Candidate	FRDA	University of Rome “Tor Vergata”

10:40 a.m. Official Photo of all AIM Attendees will be taken at this time

10:45 a.m.-12 p.m. Scientific Poster Session (Coffee, Tea, Snack)

12:00-1:30 p.m. Networking Lunch - Offsite

### **Theme 4 – Enhancing Clinical Trial Readiness**

**Session Chair:** George Wilmot, MD, PhD, Emory University, Atlanta, GA

1:30-2:05 p.m. **Key Note:** Frank Bennett, CSO, Ionis Pharmaceuticals  
*Antisense based drugs for the treatment of triplet repeat diseases*

2:05-2:30 p.m. \* Hayley McLoughlin, PhD, University of Michigan (Theme 3)  
*Antisense Oligonucleotides provide therapeutic benefit in Spinocerebellar ataxia type 3 mice*

2:30-2:55 p.m. \* Ian Harding, PhD, Monash University  
*The ENIGMA-Ataxia Global Neuroimaging Research Consortium: Big Data meets Rare Disorders*

2:55-3:20 p.m. Amy Bastian, PhD, PT, Johns Hopkins University  
*Motor Learning and Rehabilitation for Ataxia*

3:20-3:30 p.m. Afternoon Break

## WEDNESDAY, April 4, 2018 (continued)

- 3:30-3:55 p.m. \*Pierre-Gilles Henry, PhD, University of Minnesota  
*Longitudinal MRS, MRI and DTI in the Spinal Cord in Friedreich's Ataxia: 24-month follow-up*
- 3:55-4:20 p.m. Dobrila D. Rudnicki, Ph.D., NCATS  
*Catalyzing the Translation of Basic Science Discoveries into the Clinic*
- 4:20-4:45 p.m. Debate: "*Should treatments be shown to be effective in animal disease models prior to human trials?*"  
Harry Orr, PhD, University of Minnesota  
vs.  
Stefan Pulst, MD, University of Utah
- 4:45-5:05 p.m. Theme 4 Open Discussion led by Theme Chair
- 5:05-5:15 p.m. Albert La Spada, MD, PhD, FACMG, Duke University  
*Vera Cruz, Mexico – SCA 7 Research Trip*
- 5:15-6:45 p.m. Poster Session for Patients and Families
- 6:45 p.m. *Posters must be taken down*
- 6:45 p.m. *Dinner at the Marriott in Salons I-L*
- 7:30 p.m. **The Patient/Family Perspective**  
Bill Sweeney, NAF Board President  
Michael, Karen and Jennifer Leader  
Linda Snider, MD and Kyle Bryant

## THURSDAY, April 5, 2018

7:30-8:30 a.m. *Continental Breakfast in Salons I-L*

### **Theme 5 – Ataxia Trials: What can we learn from trials in other rare diseases?**

- Session Chair:** Jeremy Schmahmann, MD, Harvard, Boston, MA
- 8:30-9:05 a.m. **Key Note:** John Day, MD, PhD, Stanford University  
*Treating SMA with Gene Modification and Gene Replacement*
- 9:05-9:30 a.m. James D. Berry, MD, MPH, Harvard University  
*Coordinating Ataxia Research: Lessons from the Northeast ALS Consortium*
- 9:30-9:55 a.m. Ludy Shih, MD, MMSc, Biogen, Inc.  
*Focus on biomarkers: preparing for clinical trials in the spinocerebellar ataxias*
- 9:55-10:15 a.m. *Morning Break*

**THURSDAY, April 5, 2018** (continued)

- 10:15-10:40 a.m.     \*Manuela Corti, PT, PhD, University of Florida  
*Gene Therapy for Friedreich’s Ataxia*
- 10:40-11:05 a.m.     Robert Berman, MD, Biohaven Pharmaceuticals  
*Trigiluzole: A Phase 2/3 Randomized Controlled Trial in Patients with Spinocerebellar Ataxia*
- 11:05-11:30 a.m.     Debate: “*Catching the horse in the barn – Should treatment trials begin before ataxia symptoms develop?*”  
                              Thomas Klockgether, MD, University of Bonn  
  vs.  
                              George Wilmot, MD, PhD, Emory University
- 11:30-11:50 a.m.     Wrap-up of entire AIM – summary by chairs Gülin Öz and Al La Spada, followed by open discussion and brainstorming
- 11:50-12:00 p.m.     Closing Remarks – Albert La Spada, MD, PhD, FACMG, Duke University

*\*indicates a Junior Investigator*