

National Ataxia Foundation
AIM 2012 – 4th Ataxia Investigators Meeting
San Antonio, Texas at Grand Hyatt San Antonio
March 13-16, 2012

TUESDAY, MARCH 13, 2012

- 3-7 PM Registration
- 3-4 PM Junior Lecturer Welcome Reception (By Invitation)
- 4-6 PM Welcome Reception and Registration
- 6-7 PM **Dinner**
- 7 PM Opening Remarks – Henry Paulson, M.D., Ph.D., University of Michigan
- 7 PM **Keynote Address: Huda Zoghbi, M.D.**, Baylor College of Medicine
Toward Therapy in SCA1

WEDNESDAY, MARCH 14, 2012

- 7:00 AM **Breakfast**
- Theme 1 – Cerebellar function and dysfunction in ataxia**
(Chairs: Drs. Christopher Gomez & Khalaf Bushara)
- 8:00 AM **Keynote Address: Nancy Bonini, Ph.D.**, University of Pennsylvania
Multiple triggers to neuronal death
- 8:40 AM Jeremy Schmahmann, M.D., Massachusetts General Hospital
Cerebellum and Neuropsychiatry: Radical Implications of a New Field
- 9:10 AM Vikram Shakkottai, M.D., Ph.D., University of Michigan
Targeting potassium channels for the treatment of cerebellar ataxia
- 9:40 AM **Break 30 Minutes**
- 10:10 AM Joanna Jen, M.D., Ph.D., University of California-Los Angeles
RNA exosome in cerebellar and spinal motor neuron maldevelopment and degeneration
- 10:40 AM Mandy Jackson, Ph.D., University of Edinburgh
Progressive cerebellar dysfunction in a mouse model of SCA5: the beta-III spectrin connection
- 11:10 AM Junior Lecturer: Xiaofei Du, M.D., University of Chicago
P/Q calcium channel $\alpha 1A$ C termini regulation of Purkinje cell gene expression, dendritic morphology and synaptic input are abolished with SCA6.
- 11:30 AM Junior Lecturer: Yuanzheng Gao, Johns Hopkins University
(now at University of Florida, Gainesville)
Beta III sepectrin is critical for development of Purkinje neuron dendrites and spines
- 11:50 AM Discussion – All participants

12:20 PM **Lunch 40 Minutes**

Theme 2 – Pathogenesis of dominant ataxias (Chairs: Drs. Harry Orr & Joseph Savitt)

1:00 PM **Keynote Address: Christian De Zeeuw, M.D., Ph.D.**, Erasmus MC, Netherlands
New insights into the role of the cerebellum in health and disease

1:40 PM Hirokazu Hirai MD, Ph.D., Gunma University Graduate School of Medicine, Japan
Mutant PKC γ in spinocerebellar ataxia type 14 impairs synapse elimination and synaptic plasticity in Purkinje cells in vivo.

2:10 PM Hidehiro Mizusawa, M.D., Tokyo Medical and Dental University, Tokyo
Progress in the research on SCA31

2:40 PM Laura Ranum, Ph.D., University of Florida,
Repeat Associated Non-AUG Translation: Lessons from SCA8

3:10 PM **Break 15 minutes**

3:25 PM Patricia Maciel, Ph.D., University of Braga, Portugal
Modeling MJD/SCA3 in the mouse and in C. elegans: probing pathogenesis towards therapies

3:55 PM Junior Lecturer: Keyi Zhu, M.D., Ph.D., Baylor College of Medicine
Identification of Common Pathogenic Mechanisms of Spinocerebellar Ataxias (SCA) Using Drosophila Models

4:15 PM Junior Lecturer: Tyisha Hathorn, Ph.D., University of Florida
mGluR1 mislocalization and LTP deficits in a mouse model of spinocerebellar ataxia type 5

4:35 PM Discussion – All Participants

5:00-7:00 PM **Poster Session**

Evening **Networking Dinner on your own in San Antonio**

THURSDAY, MARCH 15, 2012

7:00 AM **Breakfast**

Theme 3 – Pathogenesis of recessive and other ataxias (Chairs: Drs. Rob Wilson and Joanna Jen)

8:00 AM **Keynote Address: Dr. Robert (Berch) Griggs, M.D.**, University of Rochester
Designing Clinical Trials for Rare Diseases

8:40 AM Marek Napierala, Ph.D., University of Texas MD Anderson Cancer Center
Epigenetic silencing in Friedreich's ataxia as target for therapy

9:10 AM Yosef Shiloh, Ph.D., Tel Aviv University, Tel Aviv, Israel
Ataxia-telangiectasia: do the known functions of ATM explain the phenotype?

9:40 AM Peter Todd, M.D., Ph.D., University of Michigan
Toward understanding Fragile X Tremor Ataxia Syndrome (FXTAS)

10:10 AM **Break 30 Minutes**

- 10:40 AM Tetsuo Ashizawa, M.D., University of Florida
Sequence complexity of expanded ATTCT repeats and its phenotypic correlation in SCA10
- 11:10 AM Junior Lecturer: Floriana Licitra, Ph.D Student, IGBMC, Illkirch, France
Pathophysiological Mechanisms Underlying Recessive Ataxia Associated With Coenzyme Q10 Deficit
- 11:30 AM Junior Lecturer: Natalia Gromak, Ph.D., Oxford
Molecular function of human senataxin protein, mutated in AOA2/ALS4 disorders, in the regulation of gene expression
- 11:50 AM Discussion – All Participants
- 12:20 PM **Lunch and Posters 40 Minutes**

Theme 4 – Toward therapies through disease mechanisms

(Chairs: Drs. Sarah Ying & Al LaSpada)

- 1:00 PM Luis Pereira de Almeida, Ph.D., University of Coimbra, Portugal
Disease-modifying strategies for Machado-Joseph disease
- 1:30 PM Robert Wilson, M.D., Ph.D., University of Pennsylvania
Experimental Therapeutics for Friedreich Ataxia
- 2:00 PM Edgardo Rodriguez-Lebron, Ph.D., University of Iowa
Selective targeting of the polyQ-encoding Cav2.1 splice isoform
- 2:30 PM Puneet Opal, M.D., Ph.D., Northwestern University, Chicago, IL
Vascular Endothelial Growth Factor in Spinocerebellar Ataxia
- 3:00 PM **Break 30 Minutes**
- 3:30 PM Gulin Oz, Ph.D., University of Minnesota
Surrogate Marker Potential of in vivo Magnetic Resonance Spectroscopy in Ataxia Trials
- 4:00 PM Junior Lecturer: Peter Breuer, Ph.D., University of Bonn Medical Center, Germany
Calpain-mediated Ataxin 3 Aggregation in Mice and Men
- 4:20 PM Junior Lecturer: Sara Lagalwar, Ph.D., University of Minnesota
Inhibition of Ataxin-1 S776 phosphorylation: A strong candidate for SCA1 therapeutic
- 4:40 PM Discussion – All Participants
- 5:15-6:15 PM **Poster Session for Patients**
- 6:30 PM **Dinner**
- 7:00 PM **Keynote Address: Helene Puccio, Ph.D.**, Research Director INSERM, IGBMC
New insights into mechanisms of recessive ataxias

FRIDAY, MARCH 16, 2012

7:00-8:00 **Breakfast**

Theme 5 – Working together to advance ataxia research and therapy

(Chairs: Drs. S.H. Subramony & Wendy Galpern)

8:00 AM Thomas Klockgether, M.D., University Hospital Bonn, Germany
Spinocerebellar ataxias: The prodromal phase

8:30 AM John Ferguson, M.D., National Institutes of Health
Office of Rare Disease Research

9:00 AM Ludger Schöls, M.D., Hertie-Institute for Clinical Brain Research Tuebingen, Germany
Move and improve - how coordinative training helps ataxia

9:30 AM Shoji Tsuji, M.D., Ph.D., University of Tokyo
Whole genome resequencing to elucidate the molecular basis of multiple system atrophy

10:00 AM **Break 30 Minutes**

10:30 AM David Lynch, M.D., University of Pennsylvania
Collaborative research efforts in Friedreich ataxia

11:00 AM Theresa Zesiewicz, M.D., FAAN, University of South Florida
Review of Clinical Trials for Ataxia

11:30 AM Junior Lecturer: Andreia Castro, Ph.D., University of Minho, Portugal
*Searching for therapeutic compounds for Machado-Joseph disease:
a C. elegans-based screening*

11:50 AM Junior Lecturer: Anna Duarri, Ph.D., University of Groningen, The Netherlands
Exome sequencing reveals disease gene underlying spinocerebellar ataxia 19

12:10 PM Discussion – All Participants

12:40 PM **Closing Lunch**

2-5 PM Opportunity to participate in small patient groups (Birds of a Feather sessions)

Schedule as of 2/21/12