

NAF SUPPORTED RESEARCH FUNDING UPDATE

Harry T. Orr, PhD, University of Minnesota



DREAM IT.
HOPE IT.

National Ataxia Foundation
Annual Ataxia Conference
April 1-3, 2016 ★ Orlando, Florida
Hosted by the Southeast Region

DISCLAIMER

- The information provided by speakers in any presentation made as part of the 2016 NAF Annual Ataxia Conference is for informational use only.
- NAF encourages all attendees to consult with their primary care provider, neurologist, or other health care provider about any advice, exercise, therapies, medication, treatment, nutritional supplement, or regimen that may have been mentioned as part of any presentation.
- Products or services mentioned during these presentations does not imply endorsement by NAF.



PRESENTER DISCLOSURES

- **No relationships to disclose or list**



NAF Funded Research – Mission

- ◆ **Bring new investigators into ataxia research
(Seed grants, Young Investigator. Post-doc grants)**
- ◆ **Foster development of treatments for ataxias
(All grants – SCA Translational Grants, anonymous donor)**



NAF Funded Research – The Big Picture

2015 - \$1,067,595

2016 - \$876,496

NAF Funded Research – 2015, Seed Grants (\$10,000-\$30,000)

SCA3, SCA1, SCA36, AOA2, Sleep-Ataxia, Ataxia Database

Rossoll, Wilfrid, Ph.D., Emory University School of Medicine, Atlanta

Wojcikiewicz, Richard Ph.D., SUNY-Upstate Medical University, Syracuse

Teixeira-Castro, Andreia, Ph.D., University of Minho, Braga, Portugal

Manley, James L., Ph.D., Columbia University, New York


Kim, Do-Hyung, Ph.D., U of MN, Minneapolis (Funded by BAARC)

Hampe, Christiane S., Ph.D., University of Washington, Seattle

Lim, Janghoo, Ph.D. Yale University, New Haven

Perlman, Susan, M.D. University of California, Los Angeles

Schmahmann, Jeremy D., M.D. Massachusetts General Hospital



**NAF Funded Research – 2015, SCA Pioneer Grants
(\$70,000-\$100,000)**

Potential Therapies – SCAs, SCA1, SCA3/MJD, SCA8

Ismael Al-Ramahi, Ph.D. Baylor College of Medicine, Houston

**Beverly Davidson Beverly, Ph.D. Children's Hospital of Philadelphia
(Partnered with BAARC)**

Laura Ranum, Ph.D. University of Florida, Gainesville

Thorsten Schmidt, Ph.D. University of Tübingen, Germany



NAF Funded Research – 2015, Young Investigators (\$32,000-\$50,000)

Friedreich's, SCA3/MJD, SCA1, SCA7

YOUNG INVESTIGATOR AWARD

Donatello, Simona, Ph.D. Université Libre de Bruxelles, Brussels,

POST-DOC FELLOWSHIP AWARDS

Marife Arancillo, Ph.D. Baylor College of Medicine, Houston

Angela Bhalla, Ph.D. University of Alabama at Birmingham

Gautam Rajpal, Ph.D. University of Michigan, Ann Arbor,

Carlos R CastilloHernandez, Ph.D. Universidad Nacional Autonoma de Mexico

Tommaso Vannocci, Ph.D. King's College, London

Yu Miyazaki, M.D., Ph.D. University of Chicago,

YOUNG INVESTIGATOR - SCA AWARDS

Maria do Carmo Pereira Costa, Ph.D. University of Michigan

Miao Zhang, Ph.D. Virginia Commonwealth University, Richmond

Marija Cvetanovic, Ph.D. University of Minnesota, Minneapolis



RESEARCH GRANTS (SEED) - 2016

Michael Downey, Ph.D. University of Ottawa, Canada, "Ataxia 7 as a regulator of substrate selection by KAT2a acetyltransferase."

Brent L. Fogel, M.D., Ph.D. & Esther B. E. Becker, Ph.D. UCLA & University of Oxford, UK Development of Cellular Model for Characterization of SCA41 Mutations

Susan Perlman, M.D. UCLA, "Ataxia Data Base."



Paul Rosenberg, M.D., Duke University Med Center, "Contribution of store-operated calcium entry to calcium dysregulation in SCAs."

RESEARCH GRANTS (SEED) - 2016

**Manolis Fanto, Ph.D. King's College London,
RBfox proteins as critical determinants for cell
toxicity in DRPLA and other Spinocerebellar
Ataxias**



**Liliana Simões Mendonça, PharmD, Ph.D.
University of Coimbra, Portugal “Transplantation
of induced pluripotent stem cells-derived neural
stem cells in Machado-Joseph disease.”**

**Katia Aquilano, Ph.D. University of Rome,
“Lipid dysmetabolism in the pathogenesis of
Friedreich's ataxia”**

YOUNG INVESTIGATOR RESEARCH AWARDS - 2016

Ricardo Mouro Pinto, Ph.D. Mass General, Harvard Medical School, "Identification of genetic modifiers of somatic GAA instability in Friedreich Ataxia by *in vivo* CRISPR-Cas9 genome editing."



Patricia Richard, Ph.D., Columbia University, New York "Role of the SETX/CHD3 interaction in the DNA damage response and its connection to AOA2"

Percy Tumbale, Ph.D. National Institute Environmental Health Sciences, NIH, Research Triangle Park, NC "Expanded Roles for Aprataxin Mutations in Ataxia Oculomotor Apraxia 1 (AOA1)"

POST-DOC FELLOWSHIP AWARDS - 2016



Rachael L. Cohen, DVM Johns Hopkins University School of Medicine, Baltimore, “Molecular Pathogenesis of SCA 12.”

Su Yang, Ph.D. Emory University, Atlanta, “Developing the MANF-based therapeutic approach for SCA 17.”

POST-DOC FELLOWSHIP AWARDS - 2016

Pankaj Kumar Singh, Ph.D.
Translational Medicine and Neurogenetics,
IGBMC, Ellkirch-Strasbourg, France,
“Unravelling pathomechanism of muscle
dysfunction in an autosomal recessive
cerebellar ataxia 2 (ARCA2) mice model.”

Magda Matos Santana, PharmD, Ph.D.
Center Neuroscience & Cell Biology,
Coimbra, Portugal, “Advanced Induced
Pluripotent Stem Cell-based Models of
Machado-Joseph disease



PIONEER SCA TRANSLATIONAL AWARDS - 2016

Wendy Raskind, M.D., Ph.D.
University of Washington,
Seattle, "Oligonucleotide-based
Therapy in BAC-Mouse Models
of SCA14.."



Laura Ranum, Ph.D.
Univ of Florida, "ASO targeting
of bidirectional transcripts
& RAN translation in SCA8."

YOUNG INVESTIGATOR SCA RESEARCH AWARDS - 2016

Giorgio Grasselli, Ph.D., University of Chicago, "Role of SK channels in cerebellar Purkinje cells in Pathophysiology of SCA."



Kenneth Scaglione, Ph.D.
Medical College of Wisconsin, Milwaukee,
"Investigation into polyglutamine in Dictyostelium."

Wei-Ling Tsou, Ph.D. Wayne State
University School of Medicine, Detroit,
"Mechanisms of Neuroprotection by DnaJ-1 in SCA6."

Sarah C. Goetz, Ph.D.
Duke University, Durham, NC
"Exploring the Role of Primary Cilia in SCA11 Pathogenesis."



**THANK YOU FOR
YOUR SUPPORT OF
THE RESEARCH**