



Advocacy for Neuroacanthocytosis Patients



**Brain, Blood and Iron:  
Joint International Symposium on Neuroacanthocytosis  
and Neurodegeneration with Brain Iron Accumulation**

October 1-2, 2010  
Doubletree Hotel, Bethesda, MD

Organizers: Ruth Walker, MB, ChB, PhD & Susan Hayflick, MD

Sponsored by  
National Institute of Neurological Disorders and Stroke  
Office of Rare Diseases Research  
National Institute of Child Health & Human Development

Additional Sponsors



*The Movement Disorder Society*  
Advocacy for Neuroacanthocytosis Patients  
AISNAF  
Hoffnungsbaum e.V.  
NBIA Disorders Association

**Program**

**Thursday, September 30<sup>th</sup>**

7.00 Buffet dinner and welcome session

**Friday, October 1<sup>st</sup>**

**9.00 Welcome and General Introduction** Adrian Danek, MD, Neurologische Klinik, Ludwig Maximilians University, Munich, Germany

**1: Observations from the clinic**

**9.10 The neuroacanthocytosis syndromes** Ruth Walker, MB, ChB, PhD, Department of Neurology, James J. Peters VAMC, Bronx and Mount Sinai Medical School, New York, NY, USA

**9.30 The neurodegeneration with brain iron accumulation syndromes** Susan J. Hayflick, MD, Molecular and Medical Genetics, Oregon Health and Science University, Portland, OR, USA

**9.50 Questions for and from basic science** Rainer Prohaska, PhD, Max F. Perutz Laboratories, Medical University of Vienna, Vienna, Austria

10.10 Discussion

10.30 Break

**2: The fate of red blood cells**

**10.50 Early erythropoiesis** Mohandas Narla, DSc, Kimbal Research Institute, New York Blood Center, New York, NY, USA

**11.10 Maturation of reticulocytes by autophagy** Mondira Kundu, MD, PhD, St. Jude Children's Research Hospital, Memphis, TN, USA

**11.30 Red cell aging** Lucia de Franceschi, MD, PhD, Department of Clinical and Experimental Medicine, University of Verona, Verona, Italy

**11.50 Determinants of red cell shape** Lesley J. Bruce, PhD, Bristol Institute of Transfusion Services, Bristol, UK

12.10 Discussion

12.30 Lunch

### **3: Mitochondria, iron, and neurodegeneration**

1.30 **Mitochondria in cellular iron homeostasis** Alex Sheftel, PhD, University of Marburg, Marburg, Germany

1.50 **Mitochondrial defects in fumble fly and pantethine rescue** Ody Sibon, PhD, Department of Radiation and Stress Cell Biology, University of Groningen, Groningen, The Netherlands

2.10 **Mitochondrial ferritin in neurodegeneration** Sonia Levi, PhD, Vita-Salute San Raffaele University, Milan, Italy

2.30 **MIN and NBIA** Tobias Haack, PhD, Ludwig Maximilians University, Munich, Germany

2.50 Discussion

3.15 **Iron and neurodegeneration** Tracey Rouault, MD, Section on Human Iron Metabolism Head, Molecular Medicine Program, NIH, Bethesda, MD, USA

4.15 Poster session

7:00 Dinner

### **Saturday, October 2<sup>nd</sup>**

9.00 **Cellular CoA defects**, Grant A. Mitchell, MD, Service of Medical Genetics, Hôpital Sainte-Justine, Montreal, QC, Canada

9.45 Discussion

10.00 Break

### **4: Membrane trafficking and turnover (1) lipids and phospholipase**

10.20 **Mechanisms of membrane trafficking** Kai Simons, MD, Max Planck Institute for Molecular Cell Biology and Genetics, Dresden, Germany

10.40 **CoA metabolism and consequences of reduced CoA** Suzanne Jackowski, PhD, Infectious Diseases, St. Jude Children's Research Hospital, Memphis, TN, USA

11.00 **Role of PLA2s in neuronal mitochondrial lipid metabolism and membrane structure** Paul T. Kotzbauer, MD, PhD, Molecular Biology and Pharmacology, George Washington School of Medicine, St. Louis, MO, USA

11.20 **iPLA2 in synaptic vesicle trafficking** Kevin L. Seburn, PhD, The Jackson Laboratory, Bar Harbor, ME, USA

11.40 **Alteration of autophagy in the brain of the INAD animal model** Alex Ma, PhD, Division of Experimental Diabetes and Aging, Mount Sinai School of Medicine, New York, NY, USA

12.00 **Fatty acid metabolism in NBIA** Michael Kruer, MD, Department of Pediatrics, Oregon Health and Science University, Portland, OR, USA

12.15 Discussion

12.30 Lunch

### **5: Membrane trafficking and turnover (2) VPS functions**

1.20 **Functions of the VPS family** Antonio Velayos Baeza, PhD, Wellcome Centre for Human Genetics, University of Oxford, Oxford, UK

1.40 **A direct requirement for Vps13p in TGN to late endosome trafficking in yeast** Robert S. Fuller, PhD, Biological Chemistry, University of Michigan, Ann Arbor, USA

2.00 **Zebrafish vacuolar protein sorting (Vps39) and endosome trafficking** Ralf Dahm, PhD, Spanish National Cancer Research Centre, Madrid, Spain

2.20 Discussion

### **6: Autophagy in neurodegenerative disease**

2.40 **Autophagy and neurodegeneration** Zhenyu Yue, PhD, Molecular & Cellular Neurobiology Laboratory, Department of Neurology and Neuroscience, Mount Sinai School of Medicine, New York, NY, USA

3.00 **Multivesicular bodies in neurodegeneration** Scott D. Emr, PhD, Weill Institute for Cell and Molecular Biology & Department of Molecular Biology and Genetics, Cornell University, Ithaca, NY

3.20 **Formation and regulation of autophagosomes in health and disease** Fulvio Reggiori, PhD, Cell Microscopy Center, Utrecht, The Netherlands

3.40 Discussion

4.00 Break

**4.15 Proteomics of neurodegeneration** Jing Zhang, MD, PhD, Department of Pathology, University of Washington School of Medicine, Seattle, WA, USA

**7: Future directions**

**5.00 Closing summary** Rainer Prohaska, PhD

**5.15 Discussion to develop a Research Strategic Plan for NBIA** Susan J. Hayflick, MD

**5.45 Discussion to develop a Research Strategic Plan for NA** Adrian Danek, MD

7:00 Dinner

**The price of the two-day symposium including breakfast, lunch, breaks and dinner is \$350.**

**Accommodation at the Doubletree Hotel can also be arranged.**

**Scholarships will be available for early career investigators on a competitive basis.**

**NINDS, ORDR and NICHD researchers are welcome to stop by at any time during the symposium to hear topics of interest.**

**Other interested participants may attend morning, afternoon or all day sessions on one or both days for a fee.**

**Please contact [glenn@naadvocacy.org](mailto:glenn@naadvocacy.org)**