



# Fourth International Symposium on Neuroacanthocytosis

July 1-2, 2008

London and Oxford

## CHAIRS:

**Prof. Kailash Bhatia**, MD, FRCP, Institute of Neurology, University College London

**Prof. Anthony P. Monaco**, MD, PhD, Wellcome Trust Centre for Human Genetics, University of Oxford

**How do thorny red blood cells, severe movement disorders due to degeneration of the basal ganglia, and mutations of single genes relate?** This and the latest advances in care of patients with neuro-acanthocytosis syndromes will be the questions discussed in meetings at the Institute of Neurology, Queen Square, London and in Oxford.

## The program includes:

**Introduction: movement disorder phenomenology** - chaired by Prof. Kailash Bhatia, DM, FRCP, Sobell Institute, Institute of Neurology, University College London

- **Follow-up of cases reported by Dr. R. Hardie in 1991** - Sonia Gandhi, MD, Institute of Neurology, University College London
- **International efforts at case collection (“Virtual Institute”)** - Prof. Adrian Danek, Neurologische Klinik, Ludwig-Maximilians-Universität, Munich
- **Epilepsy in neuroacanthocytosis** - Christian Vollmar MD, The National Society for Epilepsy, Chalfont St. Peter, Bucks
- **McLeod syndrome: brain and neuromuscular pathology** - Hans Jung, MD, Department of Neurology, University Hospital Zurich, Zurich
- **Chorein (VPS13A) expression/neuropathology** - Benedikt Bader, MD, Zentrum für Neuropathologie und Prionforschung, Universität München, Munich
- **Caudate nucleus pathology and obsessive-compulsive disorder in Chorea-Acanthocytosis** - Mark Walterfang, MD, Neuropsychiatry Unit, Royal Melbourne Hospital, Melbourne

## Treatments

- **Drug therapies including botulinum toxin** - Susanne Schneider MD, Sobell Institute, Institute of Neurology, University College London
- **Rehabilitation - Qualitative Interviewing - A patient’s perspective** - Jacqueline McIntosh, Principal Speech and Language Therapist, The Wolfson Neuro-rehabilitation Centre, St. George’s Healthcare, Wimbledon
- **Biofeedback in dystonic syndromes** - MIWM Horstink, MD, Department of Neurology, Radboud University Medical Centre, Nijmegen
- **Deep brain stimulation for NA patients** - forum led by Patricia Limousin, MD, Institute of Neurology UCL with Prof. Philippe Coubes, MD, PhD, Gui de Chauliac Hospital, Montpellier (invited), Pierre Burbaud, MD, PhD, Centre Hospitalier Pellegrin, Bordeaux, Jorge Guridi, MD, PhD, Clínica Universitaria, Universidad de Navarra, Pamplona (invited), and Lars Timmermann, MD, University Hospital, Cologne

## Related syndromes

- **Mediators and modulators in Huntington’s Disease** - Sarah Tabrizi, MD, MRC Prion Unit, Institute of Neurology, London
- **Pantothenate kinase-associated neurodegeneration** - Susan J. Hayflick, MD, Professor and Interim Chair, Molecular and Medical Genetics, Oregon Health & Science University, Portland, Oregon
- **Familial Acanthocytosis with Paroxysmal Exertion-induced Dyskinesias and epilepsy (FAPED)** - Prof. Alexander Storch, MD, Department of Neurology, Technical University of Dresden, Dresden
- **Huntington’s disease like-2** - Ruth H. Walker, MB, ChB, PhD, Department of Neurology, James J. Peters Veterans Affairs Medical Center, Bronx and Mount Sinai School of Medicine, New York, NY

## Organizers:

Antonio Velayos-Baeza, PhD; Susanne Schneider, MD  
Glenn Irvine; Tel (+44) 20 7937 2938 email: glenn@naadvocacy.org

## Exploring the clues to neuropathogenesis in neuroacanthocytosis

Chair: Prof. Anthony P. Monaco, Pro-Vice-Chancellor (Planning & Resources) and Head of Neurogenetics Laboratory, Oxford University

### Genes and proteins

- **Vps13 in yeast** - Prof. Robert S. Fuller, PhD, Associate Chair of Biological Chemistry, University of Michigan Medical Center, Ann Arbor, Michigan
- **The *Tetrahymena thermophila* Vacuolar Protein Sorting 13A protein (VPS13A) localizes to the membrane of phagosomes** - Prof. Larry Klobutcher, Associate Dean of the Graduate School, Department of Molecular Microbial and Structural Biology, University of Connecticut Health Center, Farmington, Connecticut
- **Chorein and other human VPS13 proteins** - Clotilde Lévecque, PhD, Monaco Group, Wellcome Trust Centre for Human Genetics, Oxford
- **Chorein (VPS13A) state and Proteome analysis in chorea-acanthocytosis red blood cells** - Masayuki Nakamura, MD PhD, Kagoshima University, Kagoshima
- **Identification of genes regulated by genetic variation in VPS13A** - Eric K. Moses, PhD, Associate Scientist, Southwest Foundation for Biomedical Research, San Antonio, Texas
- **Recent advances in Kell and XK research** - Soohee Lee, PhD, New York Blood Center, New York, NY
- **Pathogenesis of Huntington's disease-like 2** - Dobrila Rudnicki, PhD, Department of Psychiatry, Johns Hopkins Medical Institutions, Baltimore, Maryland

### Reports on current research sponsored by the Advocacy

- **Production of antibodies against human VPS13 proteins** - Antonio Velayos Baeza, PhD, Monaco Group, Wellcome Trust Centre for Human Genetics, Oxford
- **Erythrocyte membrane research** - Lucia de Franceschi, PhD, Department of Clinical and Experimental Medicine, Section of Internal Medicine, University of Verona, Verona
- **Proteomic inventory of erythrocytes** - Giel Bosman, PhD, Nijmegen Centre for Molecular Life Sciences, Nijmegen

### Mechanisms of red cell membrane shape changes

- **The molecular basis of red cell membrane disorders** - Jean Delaunay, MD, Hôpital de Bicêtre, Service d'Hématologie, Faculté de Médecine Paris-Sud, Le Kremlin-Bicêtre
- **Protein interactions in the erythrocyte membrane** - Mohandas Narla, DSc, Director, Kimball Research Institute, New York Blood Center, New York, NY

### Animal models

- **Molecular understanding of ChAc using ChAc mice** - Prof. Akira Sano, Chairman, Department of Psychiatry, Kagoshima University, Kagoshima
- **Huntington's disease mice** - Anthony J. Hannan, PhD, Neural Plasticity Group, Howard Florey Institute, University of Melbourne, Melbourne

### Reviews and critique

- **Genes and proteins** - Prof. John Hardy, PhD, Institute of Neurology, UCL, London
- **Structural biology** - Rainer Prohaska, PhD, Max F. Perutz Laboratories (MFPL) Medical University of Vienna

**The price of the symposium including three dinners, lunches and coffees is £250. Accommodation for three nights at Worcester College Oxford with coach transport to and from the opening day at Queen Square London can be provided for an additional £220. Please send cheques payable to the Brain Research Trust to GM Irvine, 32 Launceston Place, London W8 5RN.**