FIRST SYMPOSIUM ON ATP1A3 IN DISEASE

FROM GENE MUTATIONS TO NEW TREATMENTS

Brussels, Belgium, 10 - 11 December 2012

Organizers

European Network for Research on Alternating Hemiplegia & Duke University

Program Committee

Chairs: David Goldstein, Duke University, United States
Tsveta Schyns, ENRAH, Belgium

Alexis	Arzimanoglou	University Hospitals of Lyon, France
Allison	Brashear	Wake Forest University, NC, United States
Knut	Brockmann	Georg August University, Göttingen, Germany
Steven	Clapcote	University of Leeds, United Kingdom
Kathleen	Freson	Catholic University of Leuven, Belgium
Erin	Heinzen	Duke University, United States
Kamran	Khodakhah	Albert Einstein College of Medicine, United States
Mohamed	Mikati	Duke University, United States
Brian	Neville	University College London, United Kingdom
Poul	Nissen	Aarhus University, Denmark
Laurie	Ozelius	Mount Sinai School of Medicine, NY, United States
Bente	Vilsen	Aarhus University, Denmark
Arn	v/d Maagdenberg	Leiden University Medical Centre, the Netherlands

Registration

To participate in the Symposium, please, complete the Participant registration Form.

Deadline for registration is 1st December 2012.

Venue

Basil & Co Brussels Louise Seminar, Avenue Louise 156, 1050, Brussels , Belgium

Venue Map

PROGRAM

Monday, December 10th, 2012

Opening Session
Chairs: Tsveta Schyns and David Goldstein

10:00	**Registration Opens**	
10:30-10:45	Welcome	*Tsveta Schyns*
10:45-10:55	Family Foundations involvement for AHC international research	*Dominique Poncelin*
11:55-11:05	The parents perspective	*Sigurður Hólmar Jóhannesson*
11:05 -11:35	Genetics of rare disease, application of NGS in clinical care	*David Goldstein*

Introduction to Alternating Hemiplegia of Childhood
Chair: Brian Neville

11:35-11:55	Overview of AHC	*Mohamad Mikati*
15:55-12:10	Is AHC a progressive disease?	*Brian Neville*
12:10-12:20	Discussion	

12:20-13:00	Lunch

AHC Genetics
Chair: Giovanni Neri

13:00-13:15	ATP1A3 and AHC: The Nature Genetics research group	*Erin Heinzen*
13:15-13:30	Identifying the gene associated with AHC: the Lancet Neurology research group	*Hendrik Rosewich*
13:30-13:45	ATP1A3 mutations in sporadic cases from the I.B.AHC Biobank and Clinical Registry	*Fiorella Gurrieri*

13:45-14:00	ATP1A3 mutations in sporadic and familial AHC cases from the Utah registry	*Sandra P. Reyna*
14:00-14:15	Identification of ATP1A3 mutations by exome sequencing as the cause of AHC in Japanese patients	*Atsushi Ishii*
14:15-14:30	Phenotypic analysis of AHC patients with ATP1A3 mutations: Preliminary results.	*Eleni Panagiotakaki*
14:30-14:45	Identifying fields for future clinical research in AHC	*Alexis Arzimanoglou*

Roundtable Discussion
Clinical genetic correlations and search for new genes for AHC

14:45-15:45

Moderators : Arn V.D. Maagdenberg and Alexis Arzimanoglou

- Key challenges
- Directions moving forward
- Collaborative groupings
- Active support and collaboration to researchers in the field of AHC and related diseases - *Filippo Franchini*

15:45- 16:00	Break/Refreshments

Rapid-Onset Dystonia-Parkinsonism
Chair: *Mohamad Mikati*

16:00-16:20	AHC versus Rapid-Onset Dystonia-Parkinsonism: allelic disorders and a phenotypic spectrum	*Knut Brockmann*
16:20-16:40	Expanded RDP phenotype: motor and non-motor features	*Allison Brashear*
16:40-17:00	Dystonia phenotype, circuitry, and, physiology	*Mark Edwards*
17:00-17:20	Cerebellar dysfunction in RDP	*Kamran Khodakhah*

17:20-17:40	Imaging in Dystonia in general	*Kristina Simonyan*
17:40-18:00	The genetics of Dystonia	*Laurie J. Ozelius*
18:00-19:00	Discussion and Refreshments	
19:00-22:00	**Dinner**	
	Dinner talk From genetics to therapy: The story of neonatal diabetes	*Frances Ashcroft*

PROGRAM

Tuesday, December 11th, 2012

Functional Studies of the Na/K ATPase – Structure/Function
Chair: David Goldstein

9:00-9:20	Genes and transgenic models in migraine: Lessons for AHC?	*Arn V.D. Maagdenberg*
9:20-9:40	Structure, function, and biological roles of Na, K-ATPase isoforms in excitable tissues	*Kathy Sweadner*
9:40-10:00	Insights to disease mechanisms from structural studies of Na+, K+-ATPase and related ion pumps	*Poul Nissen*
10:00-10:20	Functional consequences of alpha-3 Na, K-ATPase mutations at the molecular and cellular levels	*Bente Vilsen*
10:20-10:40	Cell biological and mutational studies of Na, K-ATPase, insect cell expression system	*Jan Koenderink*
11:40-11:00	Functional and proteomic studies in platelets from AHC patients reveals a lysosomal granule defect	*Michela Di Michele*
11:00-11:20	Coffee and refreshments	

Functional Studies of the Na/K ATPase .Electro Physiology & in vivo work
Chair: Sophie Nicole

11:20-11:40	Electrophysiological studies in oocytes of disease mutations in atp1a2 and 3.	*Thomas Friedrich*
11:40-12:00	Electrophysiological studies in oocytes of Na, K-ATPase mechanisms	*Hanne Poulsen*
12:00-12:20	Electrophysiology of Na, K-ATPase	*David Gadsby*

12:20-13:30	Lunch break	
13:30-13:50	A Mouse Model for ATP1A3-related Alternating Hemiplegia of Childhood	*Steven Clapcote*
13:50-14:10	Increased Susceptibility to Cortical Spreading Depression in the Mouse Model of Familial Hemiplegic Migraine Type 2	*Giorgio Casari*
14:10-14:30	Zebrafish and mouse models of atp1a2 and atp1a3	*Karin Lykke-Hartmann*
14:30-14:50	Mania-like behaviour induced by genetic dysfunction of the neuron-specific Na+,K+-ATPase α3 sodium pump	*Greer S. Kirshenbaum*

Roundtable Discussion
Functional Biology of ATP1A3 and ATP1A3 Mutations

Moderators : Poul Nissen and Bente Vilsen

14:50-15:30
- Key challenges
- Directions moving forward
- Collaborative groupings

Roundtable Discussion
Collaborations and Funding

Moderators: Tsveta Schyns and David Goldstein

15:30-16:20
- Key challenges
- Directions moving forward
- Collaborative groupings
- Outcomes of the Symposium

16:20-16:30 Closing message from the AHC Community - *Jeff Wuchich*

16:30 End of the meeting

Speakers and Chairs

Alexis Arzimanoglou
Associated Professor of Neurology and Child Neurology, University Children's Hospital of Lyon, France

Frances Ashcroft
The Royal Society GlaxoSmithKline Research Professor
University Laboratory of Physiology
Oxford and Fellow of Trinity College, Oxford University, United Kingdom

Allison Brashear
Professor and Chair of Neurology
Wake Forest University School of Medicine, United States

Knut Brockmann
Professor of Paediatrics and Child Neurology ,Georg August University Göttingen, Germany

Giorgio Casari
Neurogenomics Unit
Center for Genomics & Bioinformatics
San Raffaele University School of Medicine, Italy

Steven Clapcote
Lecturer in Pharmacology,
University of Leeds
United Kin15:30-16:20gdom

Mark Edwards
Senior Lecturer, University College London, Institute of Neurology, London
United Kingdom

Filippo Franchini
I.B.AHC Project Manager
Board of Advisors of A.I.S.EA
Italy

Thomas Friedrich
Professor, Institute of Chemistry
Technical University of Berlin
Germany

David Gadsby
Professor Laboratory of Cardiac and Membrane Physiology
Rockefeller University, United States

David B. Goldstein
Professor & Director Center for Human Genome Variation
Duke University
United States

Fiorella Gurrieri
Institute of Medical Genetics
Università Cattolica del Sacro Cuore
Rome, Italy

Erin Heinzen
Assistant Research Professor Center for Human Genome Variation
Duke University
United States

Atsushi Ishii
Department of Pediatrics
School of Medicine, Fukuoka University
Japan

Sigurður Hólmar Jóhannesson
President of AHC Iceland
Board member of ENRAH
Iceland

Kamran Khodakhah
Professor Dominick P. Purpura
Department of Neuroscience
Albert Einstein College of Medicine
United States

Greer S. Kirshenbaum
Postdoctoral Research Scientist
University of Columbia
Canada

Jan Koenderink
Associate Professor
Nijmegen Centre for Molecular Life Sciences
The Netherlands

Michela Di Michele
PostDoc
Katolieke Universiteit Leuven
Belgium

Karin Lykke-Hartmann
Associate Professor
Department of Biomedicine
Aarhus University, Denmark

Mohamad Mikati
Professor of Paediatrics & Neurobiology
Duke University
United States

Giovanni Neri
Professor of Medical Genetics &Director of the Institute of Medical Genetics
Università Cattolica del S Cuore
Rome, Italy

Brian Neville
Emeritus Professor of Childhood Epilepsy
UCL Institute of Child Health
United Kingdom

Sophie Nicole
Institut du cerveau et de la moëlle épinière
INSERM UMR 975, Université Paris 6
Pierre et Marie Curie, France

Poul Nissen
Professor
Department of Molecular Biology and Genetics, Aarhus University
Denmark

Laurie J. Ozelius
Associate Professor Genetics and Genomic Sciences & Neurology
Mount Sinai Hospital, United States

Eleni Panagiotakaki
Hôpital Femme Mere Enfant
Hospices Civils de Lyon
France

Dominique Poncelin
President of Association Française de l'Hémiplégie Alternante, AFHA France

Hanne Poulsen
Department of Molecular Biology and Genetics, Aarhus University, Denmark

Sandra P. Reyna
Assistant Professor
Paediatric Motor Disorders Research Program, University of Utah School of Medicine, United States

Hendrik Rosewich
Department of Paediatrics and Adolescent Medicine, Georg August University
Göttingen, Germany

Tsveta Schyns-Liharska
Consultant Research Management
Founder and Board Member of ENRAH
Brussels, Belgium

Kristina Simonyan
Assistant Professor Neurology & Otolaryngology
Mount Sinai Hospital
United States

Kathy Sweadner
Associate Professor Molecular Neurophysiology, Massachusetts General Hospital, United States

Arn Van den Maagdenberg
Professor of Genetics
Leiden University Medical Centre
The Netherlands

Bente Vilsen
Professor
Department of Biomedicine
Aarhus University, Denmark

Jeff Wuchich
Rolesville, United States

PARTICIPANT REGISTRATION FORM

Symposium ATP1A3 in disease
10 - 11 December 2012, Basil & Co Brussels Louise, Belgium

Please, use CAPITAL LETTERS or TYPE and return this form latest 1 December 2012 to:
ENRAH Brussels Office,
Email : ts@enrah.net Tel. +32 2 325 86 94

Registration Details

☐ Mr. ☐ Ms. ☐ Dr. ☐ Prof.

Family Name: ---------- First Name: ----------

Position: ---------- Department: ----------

Organisation/Company: ----------

Address: ----------

Zip code: ---------- Town: ---------- Country: ----------

Phone: ---------- Email: ----------

- Specific diet requirements (vegetarian, allergies …): ----------

Registration Fee

The registration fee covers the Symposium brochure, coffee breaks and lunches, and dinner on Monday 10 December

- Standard € 500.00
- Reduced Registration Fee * € 300.00

* For Health professionals and non-profit organisations registered before 10 November 2012

Method of Payment:

Bank transfer to:
ENRAH, Bank: Easybank AG, Quellenstraße 51-55, A-1100 Vienna, Austria
IBAN: AT511420020010398003 BIC: EASYATW1.
Billing Information (if different from above):

Cancellation Policy

All cancellations must be in writing. Cancellations received before 1 December 2012 are subject to a charge of 25% of the registration fees for reservation costs. After 1 December 2012 full payment will be requested and refunds will not be provided. Transfer of the registration to other persons is at no extra cost.

Privacy

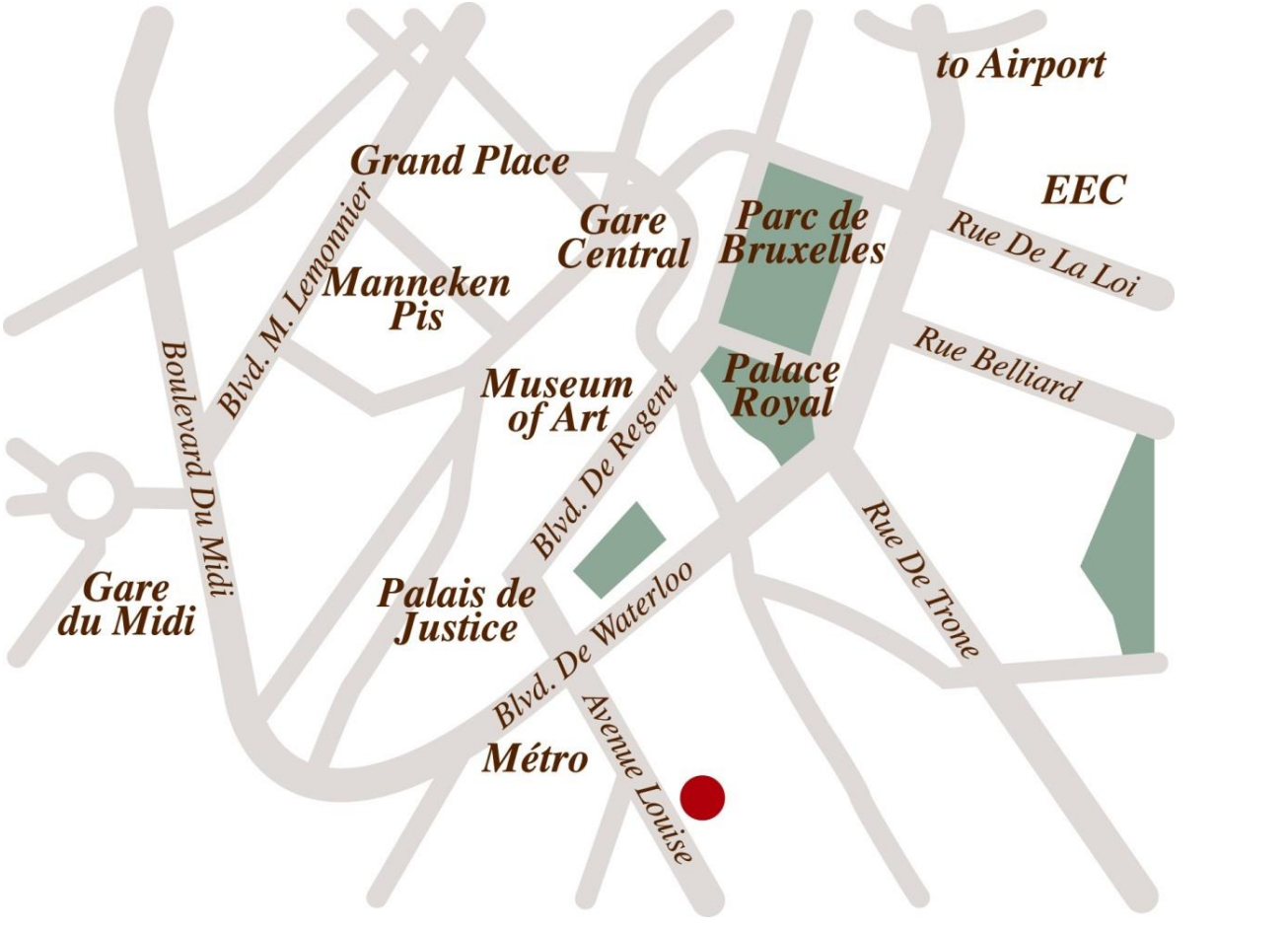
By filling out the registration form, the participant gives consent that ENRAH can process the data provided within the framework of the Symposium This includes, all handling needed for the applicant's participation at the event and for the drafting of a list of participants which will be distributed at the Symposium.

Important

Written confirmation will be sent after the payment has been received. If you have not received your confirmation within five working days, please contact the ENRAH office.

VENUE MAP

Basil & Co Brussels Louise, Avenue Louise 156, 1050 Brussels



SPONSORS

enrah EUROPEAN NETWORK FOR RESEARCH ON ALTERNATING HEMIPLEGIA	**ENRAH** **European Network for Research on Alternating Hemiplegia** www.enrah.net
RTD Services Bringing science and business together	**RTD Services** **Research and Technological Development and innovation projects** www.rtd-services.com
afha	**AFHA** **French Association for Alternating Hemiplegia** www.afha.org
AHC association of Iceland	**AHCAI** **Icelandic Association for Alternating Hemiplegia** www.ahc.is
A.I.S.EA onlus Associazione Italiana per la Sindrome di Emiplegia Alternante	**A.I.S.EA Onlus** **Italian Association for Alternating Hemiplegia** www.aisea.org
AESHA	**AESHA** **Spanish Association for Alternating Hemiplegia** www.aesha.org
AHC Vereniging Nederland	**AHC Vereniging Nederland** **Dutch Association for Alternating Hemiplegia** www.ahckids.nl
AHC AHC-Deutschland e.V. Alternating Hemiplegia of Childhood www.ahckids.de	**AHC-Deutschland e.V.** **German Association for Alternating Hemiplegia** www.ahckids.de
ahcf	**AHCF** **US Foundation for Alternating Hemiplegia** www.ahckids.org