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*
11:05-11:15	Active support and collaboration to researchers in the field of AHC and related diseases	*Filippo Franchini*
11:15 -11:45	Genetics of rare disease, application of NGS in clinical care	*David Goldstein*

Introduction to Alternating Hemiplegia of Childhood

Chair: Brian Neville

11:45-12:05	Overview of AHC	*Mohamad Mikati*
12:05-12:20	Is AHC a progressive disease?	*Brian Neville*
12:20-12:30	Discussion	
12:30-13:00	Sandwich 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 Genetics Consortium	*Fiorella Gurrieri*
13:45-14:00	ATP1A3 mutations in sporadic and familial AHC	*Sandra P. Reyna*

	cases from the Utah registry	
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*

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
15:30-16:30

- Key challenges
- Directions moving forward
- Collaborative groupings and **Outcomes of the Symposium**

16:30 **End of meeting**