Duke Center for Human Genome Variation
Duke University School of Medicine

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EUROPEAN NETWORK FOR RESEARCH ON ALTERNATING HEMIPLEGIA

SYMPOSIUM ATP1A3 IN DISEASE:
From gene mutations to new treatments

Basil & Co Brussels Louise Seminar, Avenue Louise 156, 1050, Brussels, Belgium 10 - 11 December 2012

Organizers

European Network for Research on Alternating Hemiplegia & Duke Center for Human Genome Variation

Program Committee

Chairs: David Goldstein, Duke University, United States
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Arn van den Maagdenberg, Leiden University Medical Centre, the Netherlands

PROGRAM

Monday, December 10th, 2012

	Opening Session *Chairs: Tsveta Schyns and David Goldstein*	
10:00	**Registration Opens**	
10:30-10:45	Welcome	*Tsveta Schyns* *Irene Norstedt*
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	The genetics of Dystonia	*Laurie J. Ozelius*
17:40-19:00	Discussion and Refreshments	
19:00-22:00	**Dinner**	
	Dinner talk From genetics to therapy: The story of neonatal diabetes	*Frances Ashcroft*

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*
12:20-12:30	Discussion	
12:30-13:30	Lunch break	
13:30-13:50	A Mouse Model for ATP1A3-related Alternating Hemiplegia of Childhood	*Steven Clapcote*
13:50-14:10	Zebrafish and mouse models of atp1a2 and atp1a3	*Karin Lykke-Hartmann*
14:10-14:30	Mania-like behaviour induced by genetic dysfunction of the neuron-specific Na+,K+-ATPase α3 sodium pump	*Greer S. Kirshenbaum*
14:30-14:50	Discussion	

	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

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