**5TH SYMPOSIUM ON ATP1A3 IN DISEASE**

**UCL Institute of Neurology, Queen Square, London, United Kingdom**

**24th-26th August 2016**

**PROGRAM**

## Wednesday, 24th August, 2016

19.00 P.M. – 22.00 P.M. Welcome to London

### The Holiday Inn Bloomsbury, 1st Floor Reception Area

| Time | Agenda |
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| **19:00-21:30** | **Registration & Buffet** |
| **19:45** | Welcome Speech – AHCUK |
| **19:50** | Opening Message – Sanjay Sisodiya, Chair 5th Symposium |
| **20:00** | Victoria A. Platt – Secretary AHC Foundation, USA<br>A presentation of the topics and discussions raised at the July 2016 AHCF Family Meeting in Indianapolis, Indiana, USA |
| **20:15** | Sigurdur Holmar Jóhannesson – President, AHC Federation of Europe. AHC today and in the future. |
| **20:30-22:00** | Interaction and discussions with AHC Family Associations |

## Thursday, 25 August 2016

### Morning session: Clinical developments in ATP1A3-related disease

Chairs: Helen Cross, GOSH, London &Mohamad Mikati, Duke, USA

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| **09:00** | Allison Brashear, Wake Forest School of Medicine, USA<br>&<br>Kathleen Sweadner, Massachusetts General Hospital and Harvard Medical School, USA | Plenary Introduction: Where we are in ATP1A3-related disease today? |

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| **09:45** | Diane Doummar, Hospital Armand Trousseau, Paris, France | Encephalopathies associated with ATP1A3 mutation |
| **10:00** | Hendrik Rosewich, Medical University Göttingen, Germany | CAPOS |
| **10:15** | Madeleine Scharf, Inst. of Experimental Immunology, Lübeck, Germany | Neuronal Na+/K+ ATPase as an autoantibody target in paraneoplastic neurologic syndrome |
| **10.30** | Coffee Break | |

**11:00** **What are the clinical events in ATP1A3-related disease?**
Chairs: Hendrik Rosewich, Medical University Göttingen, Germany and Sanjay Sisodiya, UCL, London

A video and clinical panel session to discuss events that are seizures confirmed on EEG recording, those that are hemiplegic episodes and that are undiagnosed.

- Facilitators:

murine experts (Steve Clapcote, Leeds, UK; Karin Lykke-Hartmann, Aarhus University, Denmark, Mohamad Mikati, Duke, USA), human epilepsy (Mohamad Mikati, Duke, USA; Helen Cross, GOSH, London; Beate Diehl, UCLH, UK; Alexis Arzimanoglou, Lyon, France), human movement disorders (Kailash Bhatia, UCL and Lucinda Carr, GOSH, London).

- Intended outcome:

consensus definitions of events in AHC, key for therapy trials.

**12:30** **Lunch in Foyer of the UCL Institute of Neurology**

**and**

**Poster session**

**Afternoon session Research developments in ATP1A3-related disease**

Chairs: Erin Heinzen, Columbia University, USA and Steve Clapcote, Leeds University, UK

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| **14:30** | Arn van den Maagdenberg, LUMC The Netherlands | AHC Gene 2 – an update |
| **14:50** | Jennifer Kearney, Northwestern University, USA | Genetic modifiers for ATP1A3 |
| **15:10** | Karin Lykke-Hartmann , Aarhus University, Denmark | Further analysis of the Atp1a3 D801Y knock-in mouse model |
| **15:30** | Bente Vilsen<br>Aarhus University,<br>Denmark | ATP1A3 neurological disease mutations affecting Na+-site III: Structural and functional perspectives and rescue of compromised function |
| **15:50** | Discussion | |
| **16.00** | Coffee Break | |
| **16.30** | Hugh Piggins, The University of Manchester, UK | Circadian Disruption in the Myshkin Mouse Model of Mania Independent of Deficits in Molecular Clock Function |
| **16:50** | Minako Hoshi, Kyoto University, Japan | ATP1A3 as target of beta-amyloid assembly |
| **17.10** | Ronald Melki, CNRS Paris, France | α-synuclein interaction with α3-Na+/K+-ATPase and relation to decline? |
| **17.30** | Jan Koenderink, Radboud UMC,<br>The Netherlands | Biochemical and electrophysiological analysis of ATP1A3 mutations |
| **17:50** | Discussion | |
| **18:00** | End of Day 1 Program | |
| **19.00** | **Conference Dinner, Holiday Inn London Bloomsbury, Turner Suite** | |

**Friday, 26 August 2016**

**Morning session: Translation to Treatments in ATP1A3-related disease**

Chairs Sanjay Sisodiya, UCL, UK and Tsveta Schyns, ENRAH, Belgium

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| **09:00** | Francesco Muntoni, GOSH, London, UK | Plenary Introduction: Gene therapy in a neurological disease, the journey: Duchenne Muscular Dystrophy |
| **09:45** | Natalya Fedosova, Aarhus University, Denmark | On the way to isoform – specific drugs |
| **10:00** | Steve Clapcote, Leeds University, UK | Update on the Myshkin mouse model of AHC |
| **10:15** | Emmanuel Roze, Paris, France | Sharing the experience of a clinical trial in alternating hemiplegia |
| **10:30** | Coffee break | |
| **11:00** | Helen Cross, GOSH London, UK | The role of dietary therapy in AHC |
| **11:15** | Elisa de Grandis, University of Genoa, Italy | Flunarazine and AEDs – where are we now? |
| **11:45** | Juan Kaski, GOSH, London, UK | Cardiac involvement in AHC: treatment beyond the brain? |
| **12:15** | **Poster Bursary Award Winner** Christine Simmons, Northwestern University, USA | AHC Patient-specific iPSC-derived Neurons Exhibit Depolarized Resting Membrane Potential and Altered Excitability |

**CONCLUSIONS, LUNCH AND END OF MEETING**