

**Time is stated in local Swedish time – time zone UTC/GMT +2 hours / CEST**

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## Thursday 23 September

8:45 Welcome and opening remarks: Organizing Committee (Anita Aperia, Eli Gunnarson)

### *Interactions between ion transporters and the plasma membrane*

9:00-9:30 **Gunnar von Heijne**, Stockholm University, Sweden:

*Biogenesis of integral membrane proteins*

9:30-10:00 **Sara Liin**, Linköping University, Sweden:

*Natural and synthetic lipids as activators of voltage-gated potassium channels*

10:00-10:15 Oral presentation: **Elisa De Grandis**

*Alternating Hemiplegia of Childhood: genotype-phenotype correlations in a cohort of 39 Italian patients*

10:15-10:45 **Steve Karlisch**, Weizman institute, Israel:

*Specific Na,K-ATPase-lipid interactions. A role in neurological disease?*

**Break 10:45-11:00**

11:00-11:30 **Erdinc Sezgin**, Karolinska Institutet, Sweden:

*Structure and function relationship in plasma membrane*

11:30-12:00 **Maria Dahlin**, Karolinska Institutet, Sweden:

*The ketogenic diet – clinical aspects and mechanisms of action*

12:00-12:30 General discussion

### **Lunch break 12:30-13:15**

#### *Pain*

Moderators: **Eli Gunnarson** and **Hendrik Rosewich**

13:15-14:00 **Håkan Olausson**, Linköping university, Sweden:

*A newly discovered system for ultrafast pain signalling*

14:00-14:30 **Philipp Mittermaier**, Karolinska University Hospital, Sweden:

*Pediatric aspects of pain and pain treatment*

14:30-15:15 **Cecilia Lidbeck** and **Kickie Löwing**, Karolinska Institutet/Karolinska University Hospital, Sweden:

*Exploring motor function and everyday activity in a child with ATP1A3*

### **Break 15:15-15:30**

Moderator: **Hjalmar Brismar**

15:30-15:45 Oral presentation: **Michael Habeck**:

*Cryo-EM structures of human Na,K-ATPase  $\alpha 1$  and  $\alpha 3$  isoforms*

15:45-16:00 Oral presentation: **Sofia Ygberg:**

*Molecular pathology of ATP1A mutations*

16:00-16:30 **Kathleen Sweadner**, Harvard university, USA:

*The impact of cellular responses on the spectrum of ATP1A3 disease*

## Friday 24 September

Moderators: **Eli Gunnarson** and **Anita Aperia**

9:00-10:00 **Keynote lecture: Majken Nedergaard**, Copenhagen University, Denmark:

*The Glymphatic System*

10:00-10:15 Oral presentation: **Maria Papadopoulou**

*Sleep disorders in children with Alternating Hemiplegia of Childhood: preliminary results of the HEPNOS study*

*ATP1A3 in motor neurons, role for motor function*

Moderators: **Anita Aperia** and **Eli Gunnarson**

10:15-10:45 **Ihtsham Ul Haq**, University of Miami, USA:

*Rapid Onset Dystonia Parkinsonism in ATP1A3 disease: one point in an expanding spectrum*

10:45-11:15 **Gareth Miles**, St Andrews University, UK:

*ATP1A3 expression within the spinal cord regulates movements and may contribute to motor disorders*

**Break 11:15-11:30**

11:30-11:45 Oral presentation: **Evgeny Akkuratov**

*Mechanisms by which the T613M mutation causes mobility and gait disturbances in Rapid-Onset Dystonia-Parkinsonism*

11:45-12:00 Oral presentation: **Frankie Sorell**

*Effects of the Rapid-Onset Dystonia-Parkinsonism gene mutation T613M on spinal motor networks in mice*

12:00-12:30 **Eva Weidenhielm Broström**, Karolinska Institutet/Karolinska University Hospital, Sweden:

*Gait and motion analysis in pediatric disorders causing motor dysfunction, – how, what and why?*

**Lunch break 12:30-13:15**

*Molecular manifestation and genetics*

Moderators: **Anita Aperia** and **Eli Gunnarson**

13:15-13:45 **Hendrik Rosewich**, Göttingen University, Germany:

*Core Phenotype of the ATP1A3 Related Disorders*

13:45-14:15 **Anna Lindstrand**, Karolinska Institutet, Sweden:

*Reverse phenotyping after whole genome sequencing allows for high diagnostic rates across a broad spectrum of rare diseases*

14:15-14:30 Oral presentation: **Monica Dahlstrup Sietam**:

*Spontaneous development of motor deficits in a mouse model carrying the CAPOS mutation*

**Break 14:30-14:45**

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14:45-15:30 **Christopher A. Walsh**, Harvard Medical School, USA:

*Early role for a ATP1A3 in brain development*

15:30-16:00 **Mohamad Mikati**, Duke University, USA:

*Gene Therapy of Alternating Hemiplegia of Childhood*

16:00-16:30 General discussion and closing remarks (Anita Aperia and Eli Gunnarson)

The symposium is conducted with the support of

