

4th Symposium on ATP1A3 in Disease

Washington, DC USA 27-29 August 2015

DoubleTree Bethesda Hotel & Conference Center



A Collaborative Effort of Advocates, Researchers, and Clinicians to Set the Stage for Treatment Trials



Objectives of the Symposium

- *To gather the researchers involved in ATP1A3 related research, in order to share latest research findings**
- *To recruit new research teams and gain new insights to accelerate the pace of knowledge regarding the mechanisms of the ATP1A3 gene and the discovered mutations**
- *To further involve patient advocates and clinicians to develop approaches aimed at improving quality of life NOW, while we pursue cures/effective treatments for the ATP1A3 mutation-caused diseases**
- * To lay the needed collaborative groundwork for successful clinical trial process, so as to be ready when research surfaces candidate compounds**

Organizing Committee

Chair

* **Jeff Wuchich, Cure AHC** Rolesville, NC USA

Members

* **Sigurdur Johannesson, AHC Assoc. Iceland/AHCFE**, Reykjavik, Iceland

* **Filippo Franchini, A.I.S.EA**, Lecco, Italy

***Tsveta Schyngs, ENRAH**, Brussels, Belgium

***Erin Heinzen, Columbia University** New York, NY USA

***David Goldstein, Columbia University** New York, NY USA

***Mohamad Mikati, Duke University** Durham, NC USA

Scientific Committee

Chair

- * **Erin Heinzen, Deputy Director of Institute for Genomic Medicine
Columbia University New York, NY USA**

Members

- * **Alexis Arzimanoglou, Associated Professor of Neurology and Child Neurology,
University Children's Hospital of Lyon Lyon, France**
- * **Allison Brashear, Professor and Chair of Neurology, Wake Forest
University School of Medicine Winston Salem, NC USA**
- * **Steven Clapcote, Lecturer in Pharmacology, University of Leeds, Leeds, UK**
- * **David Goldstein, Director of Institute for Genomic Medicine, Columbia University,
New York, NY USA**
- * **Mohamad Mikati, Wilburt C. Davison Professor of Pediatrics,
Professor of Neurobiology, and Chief of the Division of Pediatric Neurology
Duke University Durham, NC USA**
- * **Steve Petrou, Associate Director and Head of the Florey Institutes' Division of
Epilepsy, University of Melbourne Melbourne, Victoria Australia**
- * **Hanne Poulson, Junior Group leader at the PUMPKin Centre, Associate Professor
Department of Molecular Biology & Genetics, Aarhus University Aarhus, Denmark**
- * **Hendrik Rosewich, Department of Pediatrics and Adolescent Medicine, Division of
Pediatric Neurology, University Medical Center, Georg August University, Gottingen,
Germany**
- * **Arn van den Maagdenberg, Professor of Molecular and Functional Neurogenetics,
Leiden University Medical Center Leiden, The Netherlands**

Program Agenda

Thursday, August 27, 5:30 -7 PM Symposium Kickoff Reception

***Advocates, families, researchers, clinicians, and potential donors invited for fun and dialog

Friday August 28th, 9 AM -6 PM

Keynote speaker

9:00 am - 10:00 am

Walter Koroshetz - Director of the National Institute of Neurological Disorders and Stroke

The benefits and future of rare disease research

Break

10:00 am - 10:15 am

Session 1: Clinical trials in rare diseases/preparing for clinical trials in AHC/RDP

10:15 am - 1:00 pm

Session Chair(s): Mohamad Mikati and Henrik Rosewich

****First-hand experience of clinical trial in rare disease- Pompe's disease** Priya Kishnani (~45 min)

****Preparing for clinical trials in AHC/RDP**

Introductory remarks: Mohamad Mikati (5 min)

Criteria to define what should be counted as epileptic seizures

Mohamad Mikati (10 min presentation + 10 min discussion)

****Should we lump or split the different manifestations of AHC (hemiplegias, quadreplegias, dystonia, tonic spells, abnormal eye movements) and what scales should we use**

Alexis Arzimanoglou and Kathy Swoboda (10 min presentation + 10 min discussion)

****Should we assess non-paroxysmal symptoms and what scales should we use**

(motor development, ataxia, tone, behavior)

Tiziana Granata and Masayuki Sasaki (10 min presentation + 10 min discussion)

****Should we use biomarkers:**

Allison Brashear and Helen Cross (10 min presentation + 10 min discussion)

Open panel discussion (30 min)

Conclusions: Rosewich (5 min)

Lunch

1:00 pm - 2:00 pm

Session 2. Expanding phenotyping spectrum of patients with ATP1A3 mutations and gene discovery in ATP1A3 negative patients

2:00 pm - 3:30 pm

Session Chair(s): Allison Brashear

Program Agenda (Continued)

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Kathy Sweadner - New manifestations, cell loss and prevention (~10 min)
Lyndsey Prange - Compilation of case reports from Duke, Baylor, UC (25 min)
Tamar Michaeli - Novel *ATP1A3* mutation identified in a patient with dystonia (~10 min)
Arn van den Maagdenberg - TBD (~10 min)
Abha Gupta - AHC2 search (~15 min)
Erin Heinzen - Plans for reporting new disease-causing *ATP1A3* mutations (~20 min)

Planning session for the 2016 meeting in London

3:30-4:00

Discussion moderator: Tsveta Schyns

Poster Session

4:00 pm - 6:00 pm

Saturday August 29th

Session 3. Molecular characterization of *ATP1A3* mutations

9:00 am - 11:30 am

Session Chair(s): *Steve Petrou*

In vitro

Melodi McSweeney - In vitro assessments of atpase activity of AHC and RDP mutations (~10 min)

Danilo Tiziano - TBD (~10 min)

In vivo

Aaron Talsma - Identifying modifiers of Na/K ATPase mutants and how such modifiers might be useful, development of a panel of FHM and RDP mutants in flies (~15 min)

Hanne Poulsen - CAPOS mutation in drosophila (~15 min)

Mohamad Mikati - D815K and D801N mouse models of AHC (~20 min)

Karin Lykke-Hartmann - D801Y mouse model (~15 min)

David Goldstein - MEA analyses of neuronal activity from mouse models of AHC (~15 min)

Steve Petrou - Triheptanoin treatment in mouse model of epilepsy - prospects for use in AHC (~10 min)

Where do we go from here: effects of mutations in human neurons

Kevin Eggan - Past, present and future of molecular modeling in neuropsychiatric disease (~30-45 min)

Brainstorming for future directions, future collaborations, and funding opportunities (~1 hr)

Session Moderators: *Erin Heinzen* and *Jeff Wuchich*

11:30 am - 1:00 pm

Jeff Wuchich - Collective efforts of the family foundations in driving the science.

Group discussion and brainstorming

Presentation of AHC documentary

Sigurður H. Jóhannesson

Abstract Submission

Posters and Oral Presentations in the for the Symposium will be accepted in the following areas, with reference to ATP1A3 in Disease (including, but not limited to Alternating Hemiplegia of Childhood, Rapid Onset Dystonia-Parkinsonism, and CAPOS Syndrome):

- **Clinics & Treatment of symptoms for the underage and adult patients**
- **Genetics and modelling**
- **Molecular functional studies**

The abstracts, both for Posters and Oral Presentations must be received before 31 July, 2015.

To submit an abstract please leverage our Easy Chair submission site

<http://bit.ly/AbstractATP1A315>

You will be prompted to create an account (if you do not have one already) Once logged in as “author” (the account type you should have), can click “New Submission” in upper left to fill out the Form.

Abstracts may be accepted for oral presentation or as posters.

Posters must be in portrait orientation and measure not more than 0.8m wide and 1.0m high.

Oral Presentations must not exceed 10 minutes and 10 slides.

Registration

In order to register to the Symposium, please fill the Registration Form and send it to jeff@cureahc.org by 15 August 2015. To download the Registration Form, visit

<https://atp1a32015.wordpress.com/registration/>

Registration Fee

The Registration Fee covers the Symposium brochure, coffee breaks, lunches and the social dinner.

Standard Fee \$420.00

Early Registration Fee (before 30 June 2015) \$385.00

Method of Payment

Bank transfer to:

Bank Name: Wells Fargo Bank, N.A.

Bank Address: 420 Montgomery, San Francisco, CA 94104

SWIFT/BIC code (for USD payments): WFBIUS6S

Wire Routing Transit Number (RTN/ABA): 121000248

Account Number: 2299415766

Beneficiary account name: CURE AHC Inc.

Beneficiary account address: 545 Irina Dr., Rolesville, NC 27571

Beneficiary Telephone: (919) 488-4217

Cancellation Policy

All cancellations must be in writing. Cancellations received before 31 July 2015 are subject to a charge of 25% of the registration fees for reservation costs. After 31 July 2015 full payment will be requested and refunds will not be provided.

Transfer of the registration to other persons is at no extra cost.

Important

Written confirmation will be sent after the payment has been received.

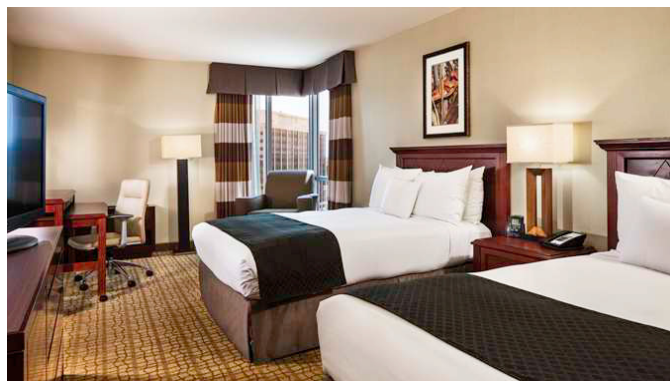
If you have not received your confirmation within five working days, please contact the Organizing Secretary at jeff@cureahc.org.

4th Symposium on ATP1A3 in Disease

Washington, DC USA 17-19 August 2015

Hotel/Venue Information

DoubleTree Bethesda Hotel & Conference Center



The DoubleTree by Hilton Hotel Bethesda - Washington DC is a top choice among Bethesda hotels to make the best of your time in and around the nation's capital. Check in at this stylish hotel and receive a warm DoubleTree welcome – our famous chocolate chip cookie provided by our friendly, professional team. Relax in your modern guest room on an ultra-comfortable bed while watching a movie on the 42-inch flat-screen HDTV. Finish work at our oversized desk while staying connected with our free WiFi. The Metro train station is just a 10-minute walk away.

Keep active during your stay with a workout in the fully equipped fitness center or swim some laps under open skies in the seasonal rooftop pool. Use our business center to prepare for the next day or to print travel documents.

Maximize the return on your meeting investment at our PURE allergy-friendly, IACC-certified conference center.

To Reserve your Room, use this link ONLY <http://bit.ly/ATP1A3HTL>

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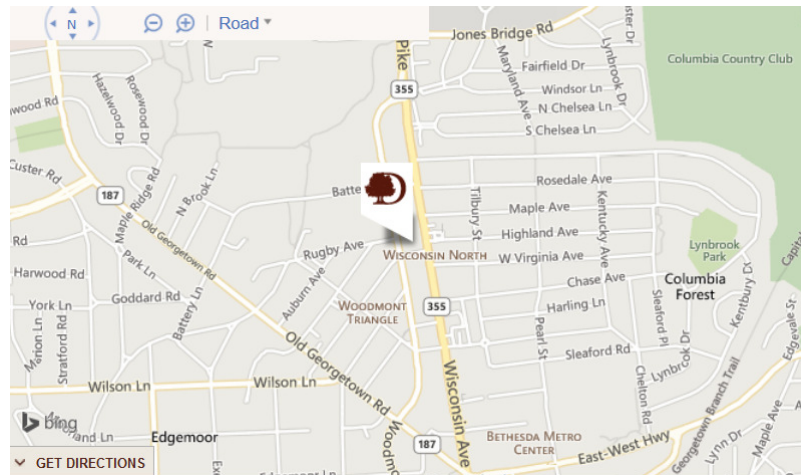
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Directions to Venue

DoubleTree Bethesda Hotel & Conference Center



**DoubleTree by Hilton Hotel
Bethesda - Washington DC
8120 Wisconsin Ave.,
Bethesda, Maryland,
20814-3624 USA
TEL: +1-301-652-2000
FAX: +1-301-652-4525**



Detailed Directions from major airports and roads is at <http://bit.ly/ATP1A315LOCALE>

Shuttle service is available coordinate through Erin Heinzen
eh2682@cumc.columbia.edu

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Contacts

Symposium Chairs

Jeff Wuchich, Cure AHC 919.412.1870 jeff@cureahc.org

Erin Heinzen, Columbia University eh2682@cumc.columbia.edu

Venue Contact

DoubleTree Bethesda Hotel & Conference Center 301.652.2000



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