Second Symposium on ATP1A3 in disease

Genotype/Phenotype Correlations, modelling and identification of potential targets for treatment

Catholic University School of Medicine
Rome, Italy

Rome, 23 - 24 September 2013

The aim of the Symposium is to present the further progress of the research on Alternating Hemiplegia of Childhood (AHC), after the finding of the ATP1A3 gene as the primary cause of this rare neurological disease, to promote the international collaboration and to recruit new teams of researchers.
Presentation of the Symposium

The aim of the Symposium is to present the further progress of the research on Alternating Hemiplegia of Childhood (AHC), after the finding of the ATP1A3 gene as the primary cause of this rare neurological disease, to promote the international collaboration and to recruit new teams of researchers.

With respect to the clinical aspects, one session of the meeting will focus on genotype/phenotype correlations based on reports from different research groups who have studied patients from all over the world.

Similarly to what was programmed in the previous ATP1A3 Symposium in Brussels, two sessions will be dedicated to the modelling of AHC and to the results of functional studies on ATP1A3, both normally and in disease.

Finally, a new session has been programmed this year, which deals with the pharmacological modulation of ATP1A3 through different compounds that are currently used in the clinical practice for the treatment of other conditions. We hope that the discussion around this topic can pave the way towards the establishment of clinical trials for AHC.

Last, but not least, we want to express our gratitude for the crucial and generous effort of the family support groups in the organization of this symposium.

We look forward to seeing you in Rome,

Giovanni Neri and David Goldstein, Chairmen
Organizing Committee

Sharon Ciccodicola, AHCF, USA
Filippo Franchini, A.I.S.EA, Lecco, Italy
David Goldstein, Duke University, North Carolina
Fiorella Gurrieri, Catholic University, Rome, Italy
Erin Heinzen, Duke University, North Carolina
Giovanni Neri, Catholic University, Rome, Italy
Dominique Poncelin, AFHA, Paris, France
Tsveta Schyns, ENRAH, Brussels, Belgium
F. Danilo Tiziano, Catholic University, Rome, Italy
Maria Rosaria Vavassori, A.I.S.EA, Lecco, Italy
Jeff Wuchich, CureAHC, USA

Contacts

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Chairs

Giovanni Neri
Institute of Medical Genetics, Università Cattolica del Sacro Cuore, Rome, Italy

David Goldstein
Center for Human Genome Variation, Duke University, Durham NC, United States

Scientific Committee

Alexis Arzimanoglou, University Hospitals of Lyon, France
Steve Clapcote, University of Leeds, United Kingdom
David Goldstein, Duke University, NC, United States
Fiorella Gurrieri, Catholic University, Rome, Italy
Erin Heinzen, Duke University, NC, United States
Arn van den Maagdenberg, Leiden University Medical Centre, the Netherlands
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Steve Petrou, University of Melbourne, Australia
Hendrik Rosewich, Georg August University Göttingen, Germany
Kathryn Swoboda, University of Utah School of Medicine, United States
Danilo Tiziano, Catholic University, Rome, Italy
Edvige Veneselli, Gaslini Scientific Institute, Genoa, Italy

Contacts

Fiorella Gurrieri, Danilo Tiziano
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## PRELIMINARY PROGRAM
### Monday, September 23rd

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<th>Time</th>
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<tr>
<td>11:00</td>
<td>Registration opens</td>
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| 12:00  | **Welcoming address**  
Giovanni Neri and David Goldstein                                       |
| 12:15  | **Clinical and genetic aspects of AHC**  
Moderator Edvige Veneselli                                                 |
| 12:15  | **Clinical outcome measures and biomarkers for AHC patients**  
Mohamed Mikati                                                               |
| 12:45  | **International collaborative group on genotype/phenotype correlations**  
Alexis Arzimanoglou                                                         |
| 13:15  | Lunch                                                                    |
| 14:15  | **Clinical aspects of AHC in different patients’ groups**  
Moderator Fiorella Gurrieri                                                 |
| 14:15  | **Catastrophic outcomes in AHC: an overview of clinical features and neuropathologic findings from the U.S/International AHCF database**  
Kathy Swoboda                                                                |
| 14:35  | **Genotype/phenotype correlation in Japanese patients with AHC**  
Masayuki Sasaki                                                             |
| 14:55  | **Natural history of the disease**  
Giuseppe Gobbi                                                              |
| 15:15  | **A brief genetic update on AHC and RDP**  
Hendrik Rosewich                                                           |
| 15:35  | **Imaging and pathology results provide potential insights**  
Allison Brashear                                                           |
15:55 Coffee break and poster session

16:45 Functional studies of ATPase
   Moderator Arn van den Maagedenberg

16:45 Searching for somatic mutation in ATP1A3 “negative” patients
   Erin Heinzen

17:05 Structural and biochemical studies addressing the AHC mutations
   Poul Nissen

17:25 Electrophysiological studies of sodium pump mutants
   Hanne Poulsen

17:45 Telethon Italy and the alliance with the patients in the support to the research on rare genetic diseases
   Francesca Sofia

18:00 What has changed after the discovery of ATP1A3? The families speak
   Tsveta Schyns, Rosaria Vavassori, Dominique Poncelin, Jeff Wuchich, Lynn Egan

18:45 Breaking News if any

19:30 End

20:30 Social Dinner
PRELIMINARY PROGRAM
Tuesday, September 24th

8:30  ATP1A3opathies modeling
      Moderator Steven Clapcote

8:30  The myshkin mouse
      Steven Clapcote

9:00  A knock-in mouse model for RDP/AHC
      Karin Lykke Hartmann

9:30  Oocyte modeling of ATP1A3 mutations
      Steve Petrou

10:00 The knock-in mouse model
      Mohammed Mikati

10:30 Structure-function studies and symptoms in a mutant mouse
      Kathy Sweadner

11:00 Coffee break

11:30 Pharmacologic modulation of ATPases activity
      Moderator to be assigned

11:30  Endogenous Ouabain and ATPase: possible implications for Rostafuroxin
      Paolo Manunta

12:00 AMPA activators as potential candidates to the treatment of AHC
      Alexander Chibalin

12:30 Binding of digitalis-like compounds to Na,K-ATPase
      Jan Koenderink
13:00 Lunch

14:00 Round table: Clinical trials
Moderator Federico Vigevano

14:00 Are we ready for clinical trials?
Tiziana Granata

14:15 General discussion
Mohammed Mikati, Eleni Panagiotakaki, Masayuki Sasaki, Paolo Manunta, Jan Koenderink, Alexander Chibalin, others ....

15:00 Conclusions and future priorities
David Goldstein

15:15 End
Speakers and Chairs

Alexis Arzimanoglou
Associated Professor of Neurology and Child Neurology, University Children’s Hospital of Lyon, France

Allison Brashear
Professor & Chair, Department of Neurology, Wake Forest Baptist Medical Center, Winston-Salem, NC, United States

Alexander Chibalin
Associate Professor of Cell Physiology, Dept. of Molecular Medicine and Surgery, Karolinska Institute, Stockholm, Sweden

Steven Clapcote
Lecturer in Pharmacology, University of Leeds, United Kingdom

Lynn Egan
President of the AHCF Foundation for Alternating Hemiplegia of Childhood, Southfields, MI, United States

Giuseppe Gobbi
Director Child Neurology Unit, Maggiore Hospital, Bologna, Italy

David B. Goldstein
Professor & Director Center for Human Genome Variation, Duke University, Durham, NC United States

Tiziana Granata
Division of Child Neurology, IRCCS Scientific Institute C. Besta, Milano, Italy

Fiorella Gurrieri
Professor of Medical Genetics at the Institute of Medical Genetics, Università Cattolica del Sacro Cuore, Rome, Italy

Erin Heinzen
Assistant Research Professor Center for Human Genome Variation, Duke University, Durham, NC United States

Jan Koenderink
Associate Professor Nijmegen Centre for Molecular Life Sciences, Nijmegen, The Netherlands
Speakers and Chairs

Karin Lykke-Hartmann  
Associate Professor Department of Biomedicine  
Aarhus University, Denmark

Arn van den Maagdenberg  
Professor of Genetics, Leiden University Medical Centre  
The Netherlands

Paolo Manunta  
Professor and Chair of the School of Nephrology, San Raffaele University  
Milan, Italy

Mohamad Mikati  
Professor of Paediatrics & Neurobiology, Duke University  
Durham, United States

Giovanni Neri  
Professor of Medical Genetics & Director of the Institute of Medical Genetics,  
Università Cattolica del Sacro Cuore, Rome, Italy

Poul Nissen  
Professor Department of Molecular Biology and Genetics, Aarhus University  
Denmark

Eleni Panagiotakaki  
Hôpital Femme Mere Enfant, Hospices Civils de Lyon  
France

Steve Petrrou  
Associate Professor Department of Center of Neuroscience  
University of Melbourne, Australia

Dominique Poncelin  
President of the French Association for Alternating Hemiplegia of Childhood AFHA  
France

Hanne Poulsen  
Department of Molecular Biology and Genetics, Aarhus University,  
Denmark

Hendrik Rosewich  
Department of Paediatrics and Adolescent Medicine, Georg August University  
Göttingen, Germany
Speakers and Chairs

Masayuki Sasaki
Department of Child Neurology, National Center of Neurology and Psychiatry, Tokyo, Japan

Tsveta Schyns-Liharska
Consultant Research Management, Founder and Board Member of ENRAH
Brussels, Belgium

Francesca Sofia
Head of the Telethon Career Program – DTI, Research Program Manager, Telethon Foundation
Milan, Italy

Kathleen Sweadner
Associate Neurobiologist, Associate Professor of Cellular & Molecular Physiology Department of Neurosurgery, Massachusetts General Hospital, Harvard Medical School, Boston, MA, United States

Kathryn Swoboda
Associate Professor, Neurology and Pediatrics and Director Pediatric Motor Disorders Research Program, University of Utah School of Medicine, United States

Rosaria Vavassori
I.B.AHC Project Coordinator and President of the Italian Association for Alternating Hemiplegia of Childhood A.I.S.EA, Italy

Edvige Veneselli
Director Child Neuropsychiatry Unit, Department of Neuroscience, IRCCS Scientific Institute G. Gaslini, Genova, Italy

Federico Vigevano
Director Neurology Division, IRCCS Scientific Institute Bambino Gesù Children’s Hospital, Rome, Italy

Jeff Wuchich
Co-Founder & President, Cure AHC-Alternating Hemiplegia of Childhood, Rolesville, NC, United States
Posters and Oral Presentations

Posters and Oral Presentations in the Breaking News Session will be accepted in the following areas, with reference to Alternating Hemiplegia of Childhood:

1. Clinics
2. Genetics and modelling
3. Quality of life for the underage and adult patients
4. IT and Organizational Models for multidisciplinary research networks

The abstracts for the Posters must be received before **31 July 2013**.
The abstracts for the Oral Presentations must be received before **31 August 2013**.

To submit an abstract please fill the **Abstract Submission Form** and send it by email to **abstract.2ndsymposiumatp1a3@ibahc.org**

The **Abstract Submission Form** can be downloaded from **www.ibahc.org/2ndsymposiumatp1a3/AbstractSubmissionForm.doc**

**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.

The acceptance of an Abstract will be notified to the Presenting Author within 15 days from the submission.

Not more than 5 abstracts will be chosen for oral presentation.

The accepted Abstracts will be published on the Official Website of the Symposium.
Venue

Meeting Room N. 617, VI° Floor
Policlinics “Agostino Gemelli”
Largo A. Gemelli, 8
Rome, Italy

How to reach the Venue

Railway
From the Railway Station “TERMINI” take the subway Line A to the stop “Valle Aurelia - Cornelia” (8 stops). Take the train Line FM3 “Roma-Viterbo” towards “Cesano” to the stop “Gemelli”.

Subway
Line A to the stop “Valle Aurelia - Cornelia” or “Battistini”.
Take one of the following City Bus Lines, all of them having a stop at the entrance of the Policlinics Gemelli:

ATAC City Bus Lines:
- line 994 (direction: Valle Aurelia - Pineta Sacchetti - Ospedale San Filippo Neri);
- line 446 (direction: Circonvallazione Cornelia - Pineta Sacchetti - Cortina d’Ampezzo - Ponte Milvio - Piazza Mancini);
- line 146 (direction: Battistini - Pineta Sacchetti - Policlinico Gemelli)

Car
Arriving from South (Naples), on the A1 Highway, enter the “Grande Raccordo Anulare – GRA” (GRA – City Ring Road) and take the EXIT N. 1 – Via Aurelia (direction: Piazza Irnerio - Via della Pineta Sacchetti) or the EXIT N. 2 – Via Bocca (direction: Via di Torrevecchia - Via della Pineta Sacchetti).
Arriving from North (Florence), on the A1 Highway, enter the “Grande Raccordo Anulare – GRA” and take the EXIT N. 3 (direction Via Trionfale - Via della Pineta Sacchetti).

Plane
At the “FIUMICINO” Airport, take the train Line FR1 “Settebagni” at the stop “Fiumicino”, to the stop “Trastevere” (five stops). Take the train line FR3B “Cesano” to the stop “Gemelli” (5 stops).
Hotels

COURTYARD ROME CENTRAL PARK (walk)
Via Giuseppe Moscati, 7
00168 Rome

Hotel SISTO V
Via Lardaria, 10
00168 Rome
www.hotelsistov.com

Hotel DESIDERIO
Via della Pineta Sacchetti, 225
00168 Rome
www.hoteldesiderioroma.com

Additional Information

The official language of the Symposium is English. No translation service, in any language, will be available.
CME Credits are being requested.

Local Organizing Secretary

Rosaria Vavassori, A.I.S.E.A Onlus
Luciana Amato, Catholic University School of Medicine
Tel. and Fax +39 039 95 180 46
Email info.2ndsymposiumatp1a3@ibahc.org
Registration

In order to register to the Symposium, please fill the Registration Form and send it to registration.2ndsymposiumatp1a3@ibahc.org by 10 September 2013.

The Registration Form can be downloaded from here www.ibahc.org/2ndSymposiumATP1A3/RegistrationForm.doc

Registration Fee

The registration fee covers the Symposium brochure, coffee breaks and lunches.

<table>
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<tr>
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<tr>
<td>Standard Fee</td>
<td>€350.00</td>
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<tr>
<td>Early Registration Fee *</td>
<td>€300.00</td>
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* before 31 August 2013

Method of Payment:

Bank transfer to:

A.I.S.EA Onlus

Bank       CREDITO EMILIANO – CREDEM, MERATE (LC) - ITALY
IBAN*     IT08 0030 3251 5300 1000 0010 200
BIC       BACRIT21918

* Warning! The first character after “IT08” is the letter “O” while all the others are the number 0 (zero)

Cancellation Policy

All cancellations must be in writing. Cancellations received before 31 August 2013 are subject to a charge of 25% of the registration fees for reservation costs. After 31 August 2013 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 info.2ndsymposiumatp1a3@ibahc.org
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