

# Second Symposium on ATP1A3 in disease

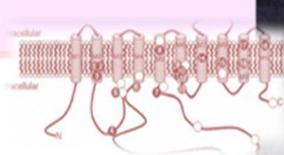
## Genotype/Phenotype Correlations, modelling and identification of potential targets for treatment

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.

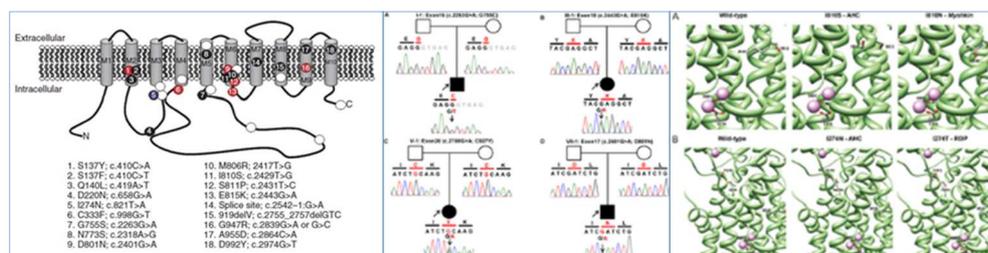
Similarly to what was programmed in the previous ATP1A3 Symposium in Brussels, two sessions will be dedicated to the modelling of AHC and to the results of functional studies on ATP1A3, both normally and in disease.

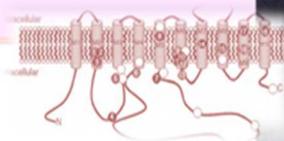
Finally, a new session has been programmed this year, which deals with the pharmacological modulation of ATP1A3 through different compounds that are currently used in the clinical practice for the treatment of other conditions. We hope that the discussion around this topic can pave the way towards the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

## Local Organizing Committee

Fiorella **Gurrieri**, Giovanni **Neri**, Danilo **Tiziano**, Catholic University, Rome Italy

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

## Contacts

Rosaria Vavassori

[info.2ndsymposiumatp1a3@ibahc.org](mailto:info.2ndsymposiumatp1a3@ibahc.org)



UNIVERSITÀ  
CATTOLICA  
del Sacro Cuore



Duke Center for Human Genome Variation  
Duke University School of Medicine





## 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

**Mohamad Mikati**, Duke University, NC, United States

**Giovanni Neri**, Catholic University, Rome, Italy

**Poul Nissen**, Aarhus University, Copenhagen, Denmark

**Laurie Ozelius**, Mount Sinai School of Medicine, NY, United States

**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**

**[scs.2ndsymposiumatp1a3@ibahc.org](mailto:scs.2ndsymposiumatp1a3@ibahc.org)**



## PRELIMINARY PROGRAM

Monday, September 23rd

**11:00 Registration opens**

**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 and Eleni Panagiotakaki

**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**

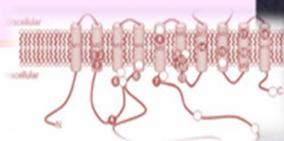
Giuseppe Gobbi

**15:15 A brief genetic update on AHC and RDP**

Hendrik Rosewich

**15:35 Imaging results in RDP provide potential insights**

Allison Brashear



**15:55 Coffee break and poster session**

**16:45 Functional studies of ATPase**

Moderator Boukje de Vries

**16:45 Genetic variation in ATP1A3 in neurological, developmental and neuropsychiatric diseases**

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 The role of Institutions and Patient Associations in the support to the Collaborative Research on AHC**

Moderator Dominique Poncelin

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

Francesca Sofia

**18:00 The Message from the AHC Families Worldwide**

Jeff Wuchich

**18:15 The International Patient Alliance AHCIA**

Lynn Egan

**18:25 The European Federation AHC FE**

Sigurdur Hólmar Johannesson

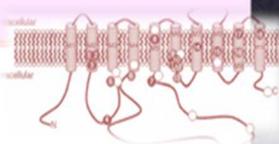
**18:35 The European Network for Research on Alternating Hemiplegia, ENRAH: ten years of facilitating clinical and basic science research on AHC**

Tsveta Schyns

**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 Danilo Tiziano

### 11:30 Endogenous Ouabain and ATPase: possible implications for Rostafuroxin

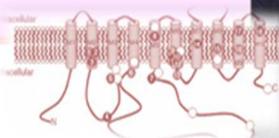
Paolo Manunta

### 12:00 AMPK 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 The Global Alliance for the Cure and the Care of the Patients**

Rosaria Vavassori

**15:30 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 Gurreri

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

### Sigurður Hólmur Johannesson

President of the Icelandic Association for Alternating Hemiplegia of Childhood AHCAI  
Reykjavik, Iceland



## Speakers and Chairs

### Jan Koenderink

Associate Professor Nijmegen Centre for Molecular Life Sciences  
Nijmegen, The Netherlands

### Karin Lykke-Hartmann

Associate Professor Department of Biomedicine  
Aarhus University, Denmark

### 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 Petrou

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

### **Danilo Tiziano**

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

### **Rosaria Vavassori**

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

### **Edvige Veneselli**

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

### **Federico Vigevano**

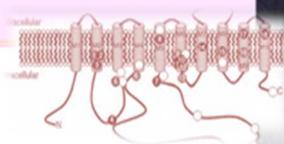
Director Neurology Division, IRCCS Scientific Institute Bambino Gesù Children's Hospital,  
Rome, Italy

### **Boukje de Vries**

Postdoctoral Researcher, Human Genetics, Leiden University Medical Centre  
The Netherlands

### **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 both the Posters and the Oral Presentations, must be received before **31 August 2013**.

All the accepted abstracts will be admitted as poster presentations.

Only five abstracts, among those submitted for the Oral Presentation, will be selected by the Scientific Committee to be presented during the Breaking News Session.

The selection of the Scientific Committee is unquestionable.

To submit an abstract please fill the **Abstract Submission Form** and send it by email to **[abstract.2ndsymposiumatp1a3@ibahc.org](mailto:abstract.2ndsymposiumatp1a3@ibahc.org)**

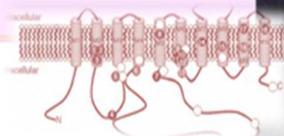
The **Abstract Submission Form** can be downloaded from **[www.ibahc.org/2ndsymposiumatp1a3/AbstractSubmissionForm.doc](http://www.ibahc.org/2ndsymposiumatp1a3/AbstractSubmissionForm.doc)**

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

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

**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.



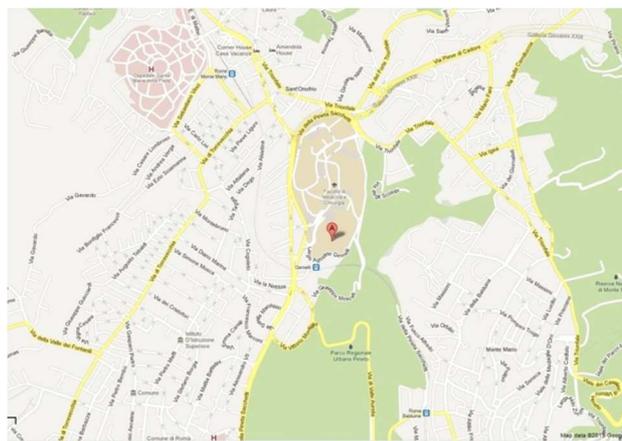
## Venue

Meeting Room N. 617, VI° Floor

Policlinics “Agostino Gemelli”

Largo A. Gemelli, 8

Rome, Italy



## Hotels

### COURTYARD ROME CENTRAL PARK (walk)

Via Giuseppe Moscati, 7

00168 Rome

[www.marriott.com/hotels/travel/romcp-courtyard-rome-central-park](http://www.marriott.com/hotels/travel/romcp-courtyard-rome-central-park)

### HOTEL ZONE

Via Alfredo Fusco, 118

00168 Rome

[www.zonehotel.com](http://www.zonehotel.com)

### Hotel SISTO V

Via Lardaria, 10

00168 Rome

[www.hotelsistov.com](http://www.hotelsistov.com)

### Hotel DESIDERIO

Via della Pineta Sacchetti, 225

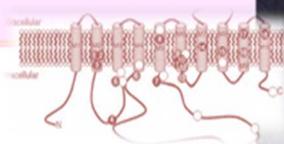
00168 Rome

[www.hoteldesiderioroma.com](http://www.hoteldesiderioroma.com)



A.I.S.EA onlus

UNIVERSITÀ  
CATTOLICA  
del Sacro Cuore



## How to reach the Venue

### Shuttle Service

Thanks to an agreement between A.I.S.EA Onlus and a SHUTTLE SERVICE Company in Rome, a reduced rate will be granted to the participants to the “Second Symposium on ATP1A3 in Disease” (Rome, 23 – 24 September 2013), for the transfer between any airport or train station in Rome and the venue or any hotel of the Symposium (and vice versa) .

If you are interested in this service, please fill the first page of the **SHUTTLE RESERVATION FORM** and send it to the e-mail addresses specified in the Form. Any further information and instructions to make the reservation are provided in the second page of the Form.

The **SHUTTLE RESERVATION FORM** can be downloaded from here  
[www.ibahc.org/2ndSymposiumATP1A3/ShuttleReservationForm.doc](http://www.ibahc.org/2ndSymposiumATP1A3/ShuttleReservationForm.doc)

---

### Public Transportation

#### 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 Boccea (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).



A.I.S.EA onlus

UNIVERSITÀ  
CATTOLICA  
del Sacro Cuore



## Additional Information

The official language of the Symposium is English. No translation service, in any language, will be available.

## Local Organizing Secretary

Rosaria Vavassori, A.I.S.EA Onlus

Luciana Amato, Catholic University School of Medicine

Tel. and Fax +39 039 95 180 46

Email [info.2ndsymposiumatp1a3@ibahc.org](mailto:info.2ndsymposiumatp1a3@ibahc.org)

## Patronages



Ministero della Salute

Italian Ministry of Health

[www.salute.gov.it](http://www.salute.gov.it)



SIGU

Italian Society of Human Genetics

[www.sigu.net](http://www.sigu.net)



ISS

Italian National Institute of Health

[www.iss.it](http://www.iss.it)

Centro Nazionale Malattie Rare



CNMR

CNMR

Italian National Center for Rare Diseases

[www.iss.it/cnmr](http://www.iss.it/cnmr)



UNIVERSITÀ  
CATTOLICA  
del Sacro Cuore



## Registration

In order to register to the Symposium, please fill the **Registration Form** and send it to [registration.2ndsymposiumatp1a3@ibahc.org](mailto:registration.2ndsymposiumatp1a3@ibahc.org) by 10 September 2013.

The **Registration Form** can be downloaded from here  
[www.ibahc.org/2ndSymposiumATP1A3/RegistrationForm.doc](http://www.ibahc.org/2ndSymposiumATP1A3/RegistrationForm.doc)

## Registration Fee

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

| Registration             |          |
|--------------------------|----------|
| Standard Fee             | € 350.00 |
| Early Registration Fee * | € 300.00 |

\* *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

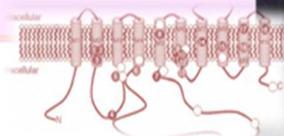
\* Warning! The first character after "IT08" is the letter "O" while all the others are the number 0 (zero)

## 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](mailto:info.2ndsymposiumatp1a3@ibahc.org)



## Sponsors



**RECORDATI**

**RECORDATI INDUSTRIA CHIMICA E FARMACEUTICA S.p.A**  
 RECORDATI Chemical and Pharmaceutical Industry, Milan, Italy  
[www.recordati.com](http://www.recordati.com)



**A.I.S.EA Onlus**  
 Italian Association for Alternating Hemiplegia  
[www.aisea.org](http://www.aisea.org)



**AFHA**  
 French Association for Alternating Hemiplegia  
[www.afha.org](http://www.afha.org)



**CureAHC**  
 Cure Alternating Hemiplegia of Childhood  
[www.cureahc.org](http://www.cureahc.org)



**AHCF**  
 US Foundation for Alternating Hemiplegia of Childhood  
[www.ahckids.org](http://www.ahckids.org)



**AESHA**  
 Spanish Association for Alternating Hemiplegia of Childhood  
[www.aesha.org](http://www.aesha.org)



AHC Association of Iceland

**AHCAI**  
 AHC Association of ICELAND  
[www.ahc.is](http://www.ahc.is)



Association Canadienne de l'Hémiplégie Alternante  
 Canadian Association for Alternating Hemiplegia

**AHC CANADA**  
 Canadian Association for Alternating Hemiplegia of Childhood  
[www.ahc-canada.ca](http://www.ahc-canada.ca)



AHCUK  
 Alternating Hemiplegia group

**AHCUK**  
 Alternating Hemiplegia of Childhood UK Support Group  
<http://ahcuk.co.uk/>



**AHC-Deutschland e.V.**  
 German Association for Alternating Hemiplegia of Childhood  
[www.ahckids.org](http://www.ahckids.org)