GENERAL INFORMATION

Description

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.
The official language of the Symposium is English. No translation service, in any language, will be available.
On the **Official Website www.symposium-atp1a3.tk** all the scientific and logistical information about the Symposium is available, as well as all the forms for the Registration and the Abstract Submission.

Registration

There is a **registration fee** (300 Euros before 31st August 2013, 350 Euros afterwards, all taxes included) that includes all the coffee breaks, the business lunches, the social dinner and the course materials.

Abstracts

Posters and Oral Communications in the Breaking News Session may be presented whose abstracts must be submitted by 31st August 2013.

Venue

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

Local Organizing Secretariat

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Patronage

The Patronage has been granted by:

Italian Society of Human Genetics
www.sigu.net

National Institute of Health
www.iss.it

National Center for Rare Diseases
www.iss.it/cnmr

The Patronage has been requested to:

Italian Ministry of Health
Municipality of Rome Capital City
SIN – Italian Society of Neurology

Directors of the Symposium

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

Speakers and Chairs

Alexis Arzimanoglou, University Children's Hospital of Lyon, France
Allison Brashear, Wake Forest Baptist Medical Center, Winston-Salem, NC, USA
Alexander Chibalin, Karolinska Institute, Stockholm, Sweden
Steven Clapcote, University of Leeds, United Kingdom
Lynn Egan, AHCF Foundation, Southfields, MI, USA
Giuseppe Gobbi, Maggiore Hospital, Bologna, Italy
David B. Goldstein, Duke University, Durham, NC, USA
Tiziana Granata, IRCCS Scientific Institute C. Besta, Milano, Italy
Fiorella Gurrieri, Università Cattolica del Sacro Cuore, Rome, Italy
Erin Heinzen, Duke University, Durham, NC, USA
Sigurdur Hólmar Johannesson, AHCAI Association, Reykjavik, Iceland
Jan Koenderink, Nijmegen Centre for Molecular Life Sciences, The Netherlands
Karin Lykke-Hartmann, Aarhus University, Denmark
Paolo Manunta, San Raffaele University, Milan, Italy
Mohamad Mikati, Duke University, Durham, NC, USA
Giovanni Neri, Università Cattolica del Sacro Cuore, Rome, Italy
Poul Nissen, Aarhus University, Denmark
Eleni Panagiotakaki, Hospices Civils de Lyon, France
Steve Petrou, University of Melbourne, Australia
Dominique Poncelin, Association AFHA, Paris, France
Hanne Poulsen, Aarhus University, Denmark
Hendrik Rosewich, Georg August University, Göttingen, Germany
Masayuki Sasaki, National Center of Neurology and Psychiatry, Tokyo, Japan
Tsveta Schyns-Liharska, ENRAH Association, Brussels, Belgium
Francesca Sofia, Telethon Foundation, Milan, Italy
Kathleen Sweadner, Massachusetts General Hospital, Harvard Medical School, Boston, MA, USA
Kathryn Swoboda, University of Utah School of Medicine, USA
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UNIVERSITÀ CATTOLICA del Sacro Cuore

A.I.S.EA onlus
Associazione Italiana per la Sindrome di Emiplegia Alternante

Second Symposium on ATP1A3 in disease

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

Catholic University School of Medicine
Rome, Italy

Rome, Policlinics "A. Gemelli"
23 - 24 September 2013

Official Website
www.symposium-atp1a3.tk

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

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 Boukje de Vries

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 **The role of Institutions and Patients' Associations in the support of 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 AHCFE**
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

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 **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**