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

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

Patronage

The Patronage has been granted by:



Italian Society of Human Genetics www.sigu.net



National Institute

of Health

www.iss.it



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

Rosaria Vavassori, Association A.I.S.EA, Lecco, Italy

Edvige Veneselli, IRCCS Scientific Institute G. Gaslini, Genova, Italy

Federico Vigevano, IRCCS Scientific Institute Bambino Gesù Children's

Hospital, Rome, Italy

Boukje de Vries, Leiden University Medical Centre, The Netherlands

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

Scientific Secretariat

Fiorella Gurrieri, Danilo Tiziano

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

Tel. +39 06 301 54 927

Email scs.2ndsymposiumatp1a3@ibahc.org





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

Email info.2ndsymposiumatp1a3@ibahc.org
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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,KATPase
 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