

**OFFICIAL CONSTITUTION OF THE**

**INTERNATIONAL RESEARCH CONSORTIUM IAHCRC**

**FOR THE RESEARCH ON ALTERNATING HEMIPLEGIA OF CHILDHOOD AND OTHER ATP1A3-RELATED DISEASES**

**Press Release – 28 November 2014**

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Alternating Hemiplegia of Childhood (AHC) is a very rare neurological disease (one affected individual in a million), caused by specific groups of mutations in the ATP1A3 gene.

Rapid Onset Dystonia Parkinsonism (RDP) and, recently, a new syndrome (CAPOS) have been recognized involving other mutations of the same gene.

All these conditions are very rare and partly overlapping in their neurological manifestations. Common pathogenetic mechanisms could be involved.

The International Consortium IAHCRC, for the research on AHC and other ATP1A3 related rare diseases, was formed in 2012 to carry out a collaborative research that led to the identification of the ATP1A3 gene as the main cause of AHC (Heinzen et al., Nature Genetics, July 2012).

The Consortium involves clinicians, geneticists and researchers working at University centers in Europe, USA and Australia; it operates in close collaboration with health professionals and patient organizations, most of whom were already involved in the EU-funded projects “ENRAH for SMEs” (FP6, 2005-2007) and nEUroped (PH, 2008-2011). Its European branch was represented at the recent meetings organized by the European Commission for the creation of European Reference Networks.

In 2013 the Consortium launched a new collaborative study GPC-AHC, directed by Prof. Alexis Arzimanoglou and Doctor Eleni Panagiotakaki (FR), aimed to identify possible correlations between the clinical phenotype associated with AHC and mutations in the ATP1A3 gene, with the goal to investigate whether different mutations can, in part, be responsible for the clinical heterogeneity observed in the disease. The data were collected from the largest international cohort of AHC patients to date (155 patients), and the results of their analysis are now submitted for publication.

Another study ECG-AHC, coordinated by Prof. Sanjay Sisodiya (UK), on some dysautonomic features of AHC, including 55 patients, is also submitted for publication.

Further collaborative studies, clinical, genetic and molecular, are currently in progress by the centers of the Consortium.

The objectives of the IAHCRC Consortium are the active contribution to the collaborative study of the pathogenetic mechanisms of the ATP1A3 diseases and the development of effective treatments. In close collaboration with patient associations, but also European and international networks, the IAHCRC will contribute to the promotion of a better care for all the affected patients, by developing specific standards for the diagnosis and the management of the diseases and by disseminating the information.

Last October 22<sup>nd</sup>, 15 founding member centers officially constituted the Consortium, through the approval of the IAHCRC Charter; the Charter contains a set of rules for the functioning of the Consortium and for the sharing of the information and patient data for its collaborative studies.

All the studies of the Consortium are carried out according to standard methods and protocols, defined by the Standardization Workgroups; currently there are three Workgroups: *genotyping*, led by Prof. Arn van den Maagdenberg (NL); *phenotyping*, led by Dr. Eleni Panagiotakaki (FR); *modeling*, led by Prof. Mohamad Mikati (USA).

The Scientific Coordinator of the Consortium is Prof. Alexis Arzimanoglou (FR) and the Data Manager is Dr. Rosaria Vavassori (IT).

All the research laboratories, the clinical centers and the scientific organizations in the world with active involvement in the research and in the care for AHC or for any other ATP1A3 diseases, and with an interest to work in collaboration with the other members, are invited to join the IAHCRC Consortium.

All the patient organizations worldwide and any other mixed or non-scientific organizations, public and private, can support the activities and the studies carried out by the Consortium, as funders, logistical supporters or appointed staff.

For more information, visit the website [www.iahcrc.net](http://www.iahcrc.net) or send an email to [info@iahcrc.net](mailto:info@iahcrc.net)