Alternating Hemiplegia of Childhood (AHC) is a rare neurological disease 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 IAHCRC Consortium (www.iahcrc.net) was formed in 2012 to carry out its first international collaborative research that led to the identification of the ATP1A3 gene as the main cause of AHC (Heinzen et al; Nature Genetics, July 2012).

In 2013, the Consortium launched further collaborative studies, including the largest international cohort of AHC patients to date, whose results have already been published: the study of the genotype-phenotype correlations (Panagiotakaki et al, European Journal of Rare Diseases, August 2015) and the study on some dysautonomic features of AHC (Jaffer at al, Brain, September 2015).

The project for the identification of the secondary gene(s) for AHC is in progress, as well as a second study on some common dysautonomic features of all the ATP1A3 related conditions. The launch of further studies, clinical, genetic and molecular has also been planned within the next year.

The IAHCRC Consortium is a network of Clinical Centers, Research Laboratories and Scientific Organizations in Europe, North America, Asia and Australia, with active involvement in the research and in the care for the ATP1A3 rare diseases.

All the IAHCRC Centers have an interest to work in collaboration according to common rules defined in the Charter of the Consortium for the sharing of data, information and resources, and according to formats and methods defined by the IAHCRC Standardization Workgroups in compliance to the international standards. They also work in close collaboration with the patient associations and with external scientific and mixed organizations.

The Euro-Mediterranean Institute of Science and Technology (IEMEST, www.iemest.eu), based in Palermo, is a no-profit research organization, recognized by the Italian Ministry of Education, University and Research (MIUR) and registered at the National and European Registries for research institutions. In 2012, it received the recognition of legal personality and in 2014 the Award “HR - Excellence in Research” from the European Commission.

The Institute pursues its statutory objectives to generate, transfer, share and disseminate knowledge and technological innovation. It supports scientific and technological research of high quality, in all the most relevant areas such as computer science and biomedical research. It has as main purpose to carry out basic and industrial research, and to disseminate results through publications, technology transfer and / or teaching.

In pursuing its objectives, the IEMEST has recently decided to support the development of the research on the ATP1A3 rare diseases and of a better healthcare for the affected patients, by becoming active member of the IAHCRC Consortium.
As such, last July 2017 it launched the IAHCRC-CLOUD Project, for the establishment of an international Platform serving the collaborative studies and initiatives carried out by the IAHCRC Consortium in collaboration with its supporting partners, both at the national and international levels.

The IAHCRC-CLOUD Platform will be operational by next January 2018, hosted by IEMEST, based on the RedCap© Electronic Data Capture Tool. The Platform, implementing the principles and methodologies of the platform cooperativism and patient engagement, can be considered as a second generation Rare Disease Registry, allowing the secure and informed data collection and sharing by all the stakeholders involved in the research and health-care process: patients, physicians, researchers, scientific and academic organizations, public healthcare institutions and services.

Dr. Rosaria Vavassori, founder and past president of the Italian Patient Association for AHC and current Data Manager of the IAHCRC Consortium, coordinates the IAHCRC-CLOUD Project for the development and management of the Platform. The Project is carried out by the IEMEST Informatics Team directed by Professor Giosuè Lo Bosco, with the supervision of Professor Francesco Cappello, Scientific Director of the Institute.

In the short term, the Platform will serve the OBSERV-AHC Study, a Prospective Observational Natural History and Therapy Study on AHC that will be launched in the first quarter 2018, with the Scientific Coordination of Professor Mohamad Mikati, at the Duke University, School of Medicine, Durham, NC, USA. Prof. Mikati is the Deputy Coordinator of the IAHCRC Consortium; Prof. Alexis Arzimanoglou, at the University Hospital of Lyon, France, is its Scientific Coordinator.

Both the IAHCRC-CLOUD Project and the OBSERV-AHC Study are sponsored by the main patient associations for AHC, in USA (Cure AHC, www.cureahc.org), France (AFHA, www.afha.org), Great Britain (AHCUK, www.ahcuk.uk), Holland (AHC Vereniging Nederland, www.ahckids.nl) and Iceland (AHC-Iceland, www.ahc.is). All these associations, together with others that are members of the International Patient Alliance AHCIA (www.ahcia.org) are also actively involved in the design, management and logistical support for both projects.

The IEMEST Institute, in the person of its President Professor Bartolo Sammartino, joins the IAHCRC Scientific Coordinators to thank all these associations heartily, for their precious support in pursuing the common goal of an effective treatment for all the patients affected by the ATP1A3 rare diseases, and for a better quality of life for all of them.

Other sponsors, with an interest to support these international projects on the rare diseases related to the ATP1A3 gene, would be more than welcome and we invite them since now to contact Dr. Rosaria Vavassori at info@iahcrc.net

For more information about the IAHCRC-CLOUD Project and about the IAHCRC Consortium and its activities, contact Dr. Vavassori by email or visit the Official Website at www.iahcrc.net