IAHCRC-CLOUD PLATFORM
A SERVICE FOR THE RESEARCH ON ALTERNATING HEMIPLEGIA OF CHILDHOOD
by a partnership between the Euro-Mediterranean Institute of Science and Technology and the IAHCRC International Consortium

Palermo, 17 September 2018

The President of the Euro-Mediterranean Institute of Science and Technology (I.E.ME.S.T.) Bartolomeo Sammartino communicates that on Tuesday October 30 2018, at 3.30pm, the press conference will take place at the headquarters of the Institute (via Michele Miraglia, 20 – IT Palermo), for the presentation of the Project “IAHCRC-CLOUD Platform”, aimed at establishing an international platform for the data collection and sharing serving the collaborative studies of the IAHCRC Consortium.

The technical and ethical aspects will be presented, as well as the innovative features of the IAHCRC-CLOUD Platform, making it a Second Generation Rare Disease Registry, implementing the principles and methods of patient engagement and platform cooperativism.

The current studies of the IAHCRC Consortium using the Platform will also be presented; among them, the longitudinal Study OBSERV-AHC, coordinated by the Division of Pediatric Neurology - AHC and Related Disorders Program of the Duke University in Durham, NC USA. In addition, some anticipations will be given on future developments already foreseen for the Platform, to make it an open, scalable and adaptable tool for any other rare disease.

The IAHCRC-CLOUD Project is coordinated by Rosaria Vavassori, Founder and Past President of the Italian Patient Association for AHC, former Coordinator of the Project I.B.AHC – Italian Biobank and Clinical Registry for AHC and former Registry Manager for the European Network ENRAH. The project team is composed of some researchers of the Institute expert in computer science, including Fabio Cernigliaro, Maurizio La Rocca and Andrey Megvinov. Giosuè Lo Bosco and Francesco Cappello, professors at the University of Palermo, are also participating in the project, the latter is also Scientific Director of I.E.ME.S.T.

Alternating Hemiplegia of Childhood (AHC) is a severe neurological disease affecting one in a million people. It is a very rare disease, complex and heterogeneous, still little known and studied; the research for the development of an effective treatment is almost entirely funded by patient organizations. Precisely because of these characteristics, the international community of patients and research and care centers has always been aware of the necessity to collaborate, creating networks, defining multicentre studies and sharing their resources through tools such as Clinical Registries and Biobanks.

Thanks to one of these multicentre studies, the genetic cause of the disease was identified in 2012, consisting in a group of specific mutations in the ATP1A3 gene that encodes a protein with a sodium-potassium ion-pump function at the neuronal level. Following this finding, the IAHCRC International Consortium was formally established, a network of clinical and basic research centers, working with the financial, technical and logistical support of the international patient alliance.

The IEMEST Institute is a member of the IAHCRC Consortium.

Please confirm your participation via email to segreteria@iemest.eu by Friday October 26 2018.

For further information about the IAHCRC-CLOUD Project: www.iemest.eu/en/e-u-projects-2#iahcrcCloud
For further information about the IAHCRC Consortium: www.iahcrc.net