The IAHCR Consortium

The International Consortium IAHCR for the Research on Alternating Hemiplegia of Childhood (AHC) 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. The Consortium involves clinicians, geneticists and researchers working at University centers in Europe, USA and Australia; it works in close collaboration with health professionals and patient organizations, most of whom were already implicated in the EU-funded projects "ENRAH for SMEs" (2005-2007) and eEuroped (2008-2011).

The Studies

In 2013 the Consortium launched a new collaborative study GPC-AHC, 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 under publication.

Further collaborative studies, clinical, genetic and molecular, are currently in execution by the centers of the Consortium. Each center has the right to initiate and/or pilot a study with all or part of the centers members of the Consortium and to develop external collaborations.

Objectives

The IAHCR Consortium aims to accelerate clinical and basic science research in the field of AHC and of all the ATP1A3 related diseases, and to improve the quality of life of the affected patients and of their families.

Its specific objectives are:
1. Contribute actively to the collaborative study of the pathogenetic mechanisms of the ATP1A3 diseases and to the development of an effective treatment for all of them;
2. Promote 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;
3. Define standard formats, protocols and procedures for the production, the assessment, the collection and the sharing of the information and data for the collaborative studies carried out by the members and for the dissemination of the information inside and outside the Consortium;
4. Collaborate with the patient associations and any other non-scientific organization in the pursuance of the first three objectives;

Members

Any Clinical Center, Research lab and Scientific organization in the world that can provide the evidence of its active involvement in the research and in the care for AHC or for any other ATP1A3 diseases, or an interest to be involved and work in collaboration with the other members, can be a member of the Consortium.

A Clinical Center in the Consortium is usually the Reference Center for the care for AHC and/or other ATP1A3 diseases, at the national level; there can be more than one Reference Center member of the Consortium in each country.

A Node is a cluster of Clinical Centers that has in charge a single cohort of patients; it collects the patient data in its Database that may have a linked biobank.

Key Features

- network of homogenous clinical databases and linked biobanks managed by the Nodes, based on an organizational and IT model developed in compliance to the ENRAH and eEuroped models
- no central database, light coordination and management of the network activities and of the data
- clear rules, included in a charter, for the sharing of the patient data and of the working information regarding the studies carried out by the centers
- common formats and procedures for the data assessment, collection and keeping at the Nodes, to be usable for all the collaborative studies of the Consortium, current and future
- easy inclusion of any new centers in the network
- fast and ethic access to a large number of patients
- fast and efficient sharing of the patient and research data for both retrospective and prospective studies, in the future, this could be strategic also for therapeutic trials
- easy and fast translation of any new research results and information into the clinical practice, thanks to all the clinical Reference Centers working together with the research labs according to this model
- can be considered as an international reference network both for the progress of the research of the associations and mixed networks (such as ENRAH) as a guarantee for the development of a higher quality of life for all the affected patients and their families.

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