IAHCRC STUDY VARIA-ATP1A3

PRESENTATION OF THE STUDY VARIA-ATP1A3

LOOKING INTO THE RARE VARIANTS OF THE MOST COMMON GENETIC CAUSE OF ALTERNATING HEMIPLEGIA IN CHILDHOOD

Study Coordinator
Katharina Vezyroglou, University College of London – Great Ormond Street Children Hospital, UK

75-80% of cases of AHC are due to mutations in the ATP1A3 gene. But not all patients with ATP1A3 related AHC carry the same ATP1A3 variant. On the contrary, there are many different variants and previously unknown ones keep being discovered. However, some of these variants, like D801N, E815K and G947R seem to be very common, and researchers have been able to study relatively large cohorts of patients, whilst others are very rare and sometimes only one patient has been described in the literature.

For these most common AHC variants a clear gradient in severity has been shown, with E815K leading to the most severe phenotype and G947R to the mildest. However, we do not know at present whether a similar correlation between phenotype and genotype exists for the rarer variants. The VARIA-ATP1A3 study is a new study spearheaded by the IAHCRC consortium with which we are trying to expand our knowledge of the rarer ATP1A3 variants by using an online survey to collect the phenotypic characteristics of patients carrying them. To do this as effectively as possible we are currently setting up a large network of collaborators from all around the globe and we are happy to hear from all colleagues interested in participation.

With the VARIA-ATP1A3 study we are hoping to:

1. establish the actual width of the ATP1A3-related disease spectrum.
2. establish whether genotype/phenotype correlation exists for the rarer ATP1A3 variants.
3. establish a registry of patients with rarer variants.

If successful, this study will help doctors and researchers give families a more accurate prognosis early on and support patients with appropriate therapies.
The knowledge we gain might help us diagnose patients with atypical AHC earlier. Finally, having a registry of patients carrying rarer variants might be valuable for future research into better understanding ATP1A3 protein function, as well as for clinical application of precision medicine approaches as they become available.