Massachusetts General Hospital
Department of Neurology
Center for Genomic Medicine
185 Cambridge St, Simches 5-238
Boston, MA 02114-2696

Kathryn J. Swoboda, M.D., FACMG
Director, Neurogenetics Unit
Tel: 617-726-5732
Fax: 617-724-9620

Dear AHC communities,

Dr. Kathryn Swoboda and her team at the Massachusetts General Hospital Neurology Department is conducting a study on "retrospective medical record and database review in children and adults with ATP1A3 related neurologic phenotype". The goals of the project are 1) to increase awareness of the range of outcomes and better document the need for novel therapeutic treatments for individuals with *ATP1A3* mutations, 2) to identify a descriptive cohort of individuals, including their functional status and age range, who are willing to participate in clinical studies and trials, and 3) to review and summarize longitudinal data previously obtained to help determine the best outcome measures across the age span in individuals with AHC. The development of this resource is critically needed to support efforts to develop appropriate clinical outcome measures, identify potential biomarkers for clinical trials, and synergize efforts to develop new therapeutic targets for patients with AHC.

We will prioritize individuals who have previously enrolled in the database to establish a rich natural history dataset across the lifetime. The genetic counselor or study coordinators may be reaching out to the families by phone or email for medical history information and to help fill in any missing critical data. You may be receiving a survey link to complete questionnaires. This study is voluntary, and your participation is greatly appreciated.

If you have any questions or concern, please do not hesitate to contact the study contact person Helen (Jin Yun) Chen at 617-726-4878 or email jin.chen@mgh.harvard.edu

Sincerely,

Kathryn J. Swoboda M.D.

Kathryn J. Swoboda, MD, FACMG
KATHERINE B. SIMS MD CHAIR IN NEUROGENETICS