ARSACS Research Progress Update
October 2017

International collaboration on ARSACS continues to advance our knowledge of cellular dysfunctions in the disease. The prestigious Anglo-American journal Human Molecular Genetics published in August a collective work combining work carried out in the laboratories of Paul Chapple in London, Francesca Maltecca in Milan and Bernard Brais and Heather Durham in Montreal. This study demonstrates that the protein, the missing protein in most cases of ARSACS, is essential for normal organization and maintenance of the cytoskeleton of all cells, and more particularly fibroblasts from the skin of patients. This study goes in the direction that a cytoskeletal abnormality is the primary change seen in ARSACS. Moreover, it highlights that the use of fibroblasts of patients can be used for the research and validation of therapeutic molecules. As a matter of fact, the high throughput screening platform of the University of Montreal's Institute for Research in Immunology and Cancer (IRIC), under the direction of Michel Bouvier, works closely with Dr. Brais's team Montreal Neurological Institute to screen molecules using the fibroblasts of patients. The results are very encouraging, to the point where the foundation has chosen to support Dr. Bouvier and his team to accelerate this research. The foundation also supports research on therapeutic molecules by Professor Thomas L. Schwarz of Harvard University Children's Hospital. 2017 is the first year in which we are seeing therapeutic breakthroughs for ARSACS! Ten years of research, because research is healing.

Several researchers from the international collaboration on ARSACS participated in the International Ataxia Research Conference held in Pisa, Italy, from September 27 to 30, 2017. They learned about the therapeutic breakthroughs developing for Friedreich's ataxia, the most common of recessive ataxias, and others more rare ataxias. Progress is very encouraging, especially with new DNA-derived molecules. The Foundation's efforts to increase the international visibility of ARSACS have been successful because ARSACS was specifically mentioned in deliberations as one of the most recessive ataxias on which research was being conducted, and was to be discussed further in this type of international meeting.