

The Ataxia of Charlevoix-Saguenay at the SPATAX Congress in Paris in June 2013- Summary

The SPATAX is mostly an European network of researchers and clinicians interested in spastic paraparesis and cerebellar ataxia. From 11 to 13 June 2013, this group, in association with The Ataxia Study Group (ASG) and The European Friedreich's Ataxia Consortium for Translational Studies (EFACTS) jointly held an international conference. Many of the wellknown names in the ataxia research field not only in Europe but also in America participated in this conference.

More than 200 researchers interested in this subject met together. Ataxia of Charlevoix-Saguenay (ARSACS) was a subject of discussion. It is clear from this conference that ARSACS is really a relatively common recessive ataxia in Europe and in other countries. Six presentations focused on the theme of ARSACS. In the wider context of the presentations on the frequency of mutations in the SACS gene from the different countries, it appears that ARSACS is often the second most common after Friedreich's ataxia, whether in France, Holland and Austria. Phylogeny work on the origin and structure of the SACS gene was presented by an Italian group. A French group of researchers from Bordeaux and the team of Paul Chapple, which team has been funded for several years by the Ataxia of Charlevoix-Saguenay Foundation, presented the results of their study on the fibroblast cells of ARSACS patients.

Finally, the work of the ARSACS team on the KO mice was selected for a special presentation at the end of the congress. This meeting helped to build more links between researchers interested in ARSACS. The Foundation hopes that these contacts will develop a truly international research network on ataxia of Charlevoix-Saguenay to accelerate the discovery of treatments.