

Minutes of the Ataxia of Charlevoix-Saguenay research team meeting held at the Montreal Neurological Institute on January 14, 2014

Participating at the meeting held at the Montreal Neurological Institute on January 14, 2014 were twelve researchers working on the ataxia of Charlevoix-Saguenay, four members from the Dr. Paul Chapple's team remotely from London UK and Dr. Kemal Payza, project director at the Institute of Montreal NeoMed.

Drs Shoubridge , Heather Durham , Alana Watt and Jason Young presented the selected projects in the last competition of the Foundation. This provided an opportunity to share with the whole team the new research directions chosen by the Foundation.

The production of new antibodies by Dr. Peter McPherson against the sacsins was discussed. The exercise once again appears complex and other commercial approaches will be pursued.

The results generated by the team of Dr. Paul Chapple were reviewed for more than an hour. The work of his team are complementary to those of the Canadian team. They continue to demonstrate the importance of the mitochondria in the pathophysiology of ataxia of Charlevoix -Saguenay.

Some of the results generated during a first short-term study of an experimental drug in the KO (knockout) mouse, which does not express the sacsins, were presented by Roxanne Larivière from the laboratory of Dr. Brais. These results suggest that this drug could improve motor performance of patients on a daily basis. The lasting effect of this experimental drug will be evaluated on the mice over the upcoming months.

The first results of a screening study of more than 110,000 molecules, some of which are potentially therapeutic, were presented. The team of Dr. Kalle Gehring in collaboration with Dr. Jean Duchaine, director of the high screening platform of the Institute for Research in Immunology and Cancer (IRIC) of the Université de Montreal, achieved this first major research of new molecules that can influence the function of the protein. They identified more than 400 substances that could be powerful tools to better understand the functioning of the sacsins.