

Local researchers link rare disease to Parkinson's, ALS

Deadly neurological ailment afflicts children, almost exclusively in Quebec

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An international team of researchers led by scientists from the Montreal Neurological Institute at McGill University have discovered the origins of a rare neurological disease known as ARSACS that afflicts children, almost exclusively in Quebec.

The discovery, published in this week's issue of the Proceedings of the National

Academy of Sciences of the United States of America and singled out for special mention by the American Society for Cell Biology, revealed the disease is linked to a defect in the function of mitochondria, the energy-producing power plants of cells, which gives it a link to more common neurological diseases like Parkinson's and ALS.

This brings hope the extensive research done on those diseases will advance treat-

ment of the rare disorder and further research in the more common diseases.

"We think that by studying this disease we will not only bring treatment to those patients, but may also help to better understand how other neuro-degenerative diseases like Parkinson's – and to a lesser degree, Alzheimer's – function," said Dr. Bernard Brais of the Neuro.

Autosomal Recessive Spastic Ataxi of Charlevoix-Saguenay is so named because it was diagnosed among descendants of roughly 10,000 French settlers who emigrated to that region of New France. It was first

recognized as a separate neurological disease in 1979. Victims show symptoms between the ages of 2 and 6 years old that include trouble walking and clumsiness. By their early 40s, sufferers must use wheelchairs and have trouble speaking. Most die in their 50s. The disease affects about 300 people in Quebec, and another 100 worldwide.

In 2000, the gene that was mutated in patients was identified. All genes make proteins that carry out a certain function, but scientists didn't know what the mutated proteins did. This year, a team of 30 scientists working at nine laboratories worldwide final-

ly discovered that the mitochondria in neurons (cells that carry nerve impulses), particularly those found in the brain, were mutated and would shut down, leading to neurological degeneration.

"You can't even imagine what to do in terms of therapy until you know at the cellular level what's going on," said Dr. Peter McPherson of the Neuro. Scientists can now try different medications and chemical treatments to slow or cure the disease. Most importantly, because links were found to diseases like Parkinson's and ALS that are also affected by mitochondrial defects, the multitude of re-

search on those diseases, and the treatments used, can be applied to ARSACS.

"For us, it definitely gives hope," said Sonia Gobeil and Jean Groleau, whose two children were diagnosed in 2006. The Montreal couple have raised more than \$2 million for research. Their children, now 6 and 8, are still fine but "we know where its going," Gobeil said.

Anything that can help to slow or cure the disease is key, the couple said. This week's announcement was a huge step.

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