Discovery of a gene that causes Joubert Syndrome

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C5ORF42 was identified as the gene that causes Joubert Syndrome in a number of families in the Lower St. Lawrence region of Quebec where the causal gene had remained unknown since the initial description of the syndrome in 1969. This is what a study in the April issue of The *American Journal of Human Genetics* reveals. The study was conducted by researchers from the Sainte-Justine University Hospital Research Center and the Centre of Excellence in Neuromics of Université de Montréal (CENUM).

Joubert Syndrome is a condition that affects brain development and manifests itself through delayed psychomotor development, abnormal coordination of eye movements and respiratory abnormalities. Since Dr. Marie Joubert and her colleagues described it for the first time in 1969, a number of related genes have been identified in various populations, but the causal gene of the Quebec form of the syndrome has remained until now unknown.

"No studies had been done to identify the genetic origin of the disease in Quebec, more specifically in the exact area of the Lower St. Lawrence where most cases in Quebec are concentrated," Dr. Jacques Michaud, the study's principal investigator, explained. "This is the first study to present a picture of Joubert syndrome in the Quebec population. It will allow family members affected by the syndrome to assess their children's genetic risks with a simple DNA test."

The finding is interesting, both genetically and historically, since, while

Joubert Syndrome is present around the world, genetic strains can vary regionally. Distribution is related to the history of various population groups.

Quebec is no exception. In fact, 6,000 French Canadian settlers from Quebec City and its surroundings settled in the Lower St. Lawrence region about the end of the 17th and beginning of the 18th centuries. The fact that most present day inhabitants of the Lower St. Lawrence region descend from this small group of settlers suggests a genetic founder effects. Indeed, certain founder mutations are transmitted to a large number of descendants, which increases the risk of genetic diseases in offspring.

As a matter of fact, Dr. Michaud's team identified three C5ORF42 mutations that are common to most of the families in the study. In all, seven families that are carriers of the gene were identified in a 400-km area along Route 132.

In the space of about two centuries, the first colonists settled in the Lower St. Lawrence region and from there other families headed out to settle along the river heading east as far as Mont-Joli and then along the Matapedia River. According to Myriam Srour, a doctoral student and co-author of the study, the mutations likely spread in the population along these migration routes.

So far 15 genes that play a role in the expression of the syndrome have been revealed elsewhere in the world. Dr. Michaud and his team will continue their research to better understand the exact function of the gene and the specific effect of each mutation.

Provided by University of Montreal

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