

## \$3M TO ACCELERATE THE INTEGRATION OF GENOMICS INTO CLINICAL PRACTICE

**Montréal, December 14, 2020 –** Génome Québec, in partnership with Genome Canada, is pleased to announce that two CARTaGENE scientific directors, Guillaume Lettre, PhD and Simon Gravel, PhD, have been awarded funding of more than \$3 million for the *GenoRef-Q Initiative*. This initiative will sequence the genome of more than 2000 participants from previously genotyped individuals recruited for the CARTaGENE population-based cohort.

The funding comes from Genome Canada's *Regional Priorities Partnership Program* (RP3), which aims to develop initiatives that advance genomics research and its practical translation capacity in areas of strategic priority specific to a given region. Génome Québec and Genome Canada contributed \$2 million to the initiative, while CARTaGENE and the CHU Sainte-Justine each invested \$100,000. This unprecedented achievement will also be made possible with significant support from the tech company Illumina.

According to Guillaume Lettre, CARTaGENE is a unique initiative that will produce valuable information not only in Québec but around the world as well. "Our end goal is to create a catalogue of genetic variants found in the population of Québec and ultimately provide clinical geneticists and other clinicians with a powerful tool to interpret the results of genetic tests and to pinpoint the cause of various genetic diseases."

In addition to supporting multiple initiatives in precision medicine, the *GenoRef-Q Initiative* will generate sequencing information useful for deciphering existing genetic data and prioritizing further development of more specific drug targets. "Given that the project will lead to a better understanding of genetic risks in Quebec and Canadian populations, it aligns perfectly with the priorities of many universities and research centers, as well as those of the provincial and federal governments," said Simon Gravel.

The *GenoRef-Q Initiative* is part of an overall strategic vision focused on making available to researchers and clinicians safe, robust infrastructure for receiving data from clinical activities and genomics research. "This is a unifying project that will leverage existing infrastructure and bring together researchers from the clinical genomics research ecosystem in Québec. This cutting-edge approach will mark a major step forward in helping clinicians with decision-making and support the shift toward precision medicine and its alignment with big data, two priority areas in Québec," said Génome Québec President and CEO Daniel Coderre, PhD.

Asked to comment on the relevance of such a tool, Anne-Marie Laberge, M.D., a researcher at the CHU Sainte-Justine Research Centre and co-researcher for GenoRef-Q, explained that "This tool will be very useful in interpreting sequencing data both for researchers and for molecular diagnostic labs. With this tool, it will be possible to interpret the benign or pathogenic character of variants, understand how common they are in the Québec population and, in turn, better categorize them."



Another feature of this unique initiative is that it will make use of the *BALSAC* population database, a structured tool that tracks the genealogy of individuals it includes. Access to *BALSAC* paves the way for research on the characteristics of the Québec population in terms of the founder effect, growth, diversity and regional structures. According to Hélène Vézina, Director of the *BALSAC* project and a professor at Université du Québec à Chicoutimi, "the synergy between the *GenoRef-Q Initiative* and *BALSAC* will not only enhance the initiative by introducing a sociohistorical perspective but it will also reaffirm the relevance of *BALSAC* as the project nears its 50<sup>th</sup> anniversary."

## **About Génome Québec**

Génome Québec's mission is to catalyze the development and excellence of genomics research and promote its integration and democratization. It is a pillar of the Québec bioeconomy and contributes to Québec's influence and its social and sustainable development. The funds invested by Génome Québec are provided by the ministère de l'Économie et de l'Innovation du Québec (MEI), the Government of Canada through Genome Canada and private partners. To learn more, visit <a href="https://www.genomequebec.com">www.genomequebec.com</a>

## **About CARTAGENE**

CARTaGENE is a public research platform of the CHU Sainte-Justine created to accelerate health research. It consists of both biological samples and health and lifestyle data from 43,000 Québec residents aged between 40 and 69 years at recruitment. Its mission is to accelerate research and innovation and support evidence-based decision-making, while reducing the cost of health research. To learn more about the CARTaGENE population cohort, visit <a href="https://www.cartagene.qc.ca">https://www.cartagene.qc.ca</a>

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