

## **Genomic Sequencing, a Powerful Ally Against Rare Diseases: The Care4Rare-EXPAND Initiative**

**MONTREAL, March 5, 2025** – Ending diagnostic uncertainty for thousands of families affected by a rare disease through genome sequencing: this is the ambition of the Care4Rare-EXPAND, with the establishment of a federated database including more than 7,000 Canadian families. Today, Genome Canada and its provincial partners announced a \$21 million funding for this large-scale initiative as part of the Canadian Precision Health Initiative, of which \$7.2 million are dedicated to the Quebec component led by Dr. Jacques L. Michaud, geneticist, researcher and director of the Centre de recherche Azrieli du CHU Sainte-Justine and Director of the Centre québécois de génomique clinique. These funds are jointly awarded by Genome Canada, Genome Quebec, Santé Québec and Québec's ministère de la Santé et des Services sociaux.

Leveraging emerging genomic technologies and powerful data analysis tools, Care4Rare-EXPAND promises to increase the diagnostic yield of genomic approaches for investigating rare diseases, in addition to being a strong driver of innovation. It thus represents an important milestone in the ambition of the Centre de recherche Azrieli du CHU Sainte-Justine, which is to develop true precision health by unlocking the full potential of genomics and data.

This initiative plans to generate data on more than 7,000 Canadian families living with an undiagnosed rare disease, including the sequencing of 15,000 genomes. The data generated as part of the project will reflect the diversity of genetic ancestries by reaching populations from all backgrounds (urban, rural, remote areas), French Canadians and English Canadians, members of Indigenous communities, as well as new Canadians. The data will be integrated into the Genomics4RD databases (Alberta and Ontario) and the Centre québécois de données génomiques (Quebec), which will be interfaced with the Pan-Canadian Genomic Library to facilitate their sharing with researchers in compliance with the most rigorous ethical and security standards.

Care4Rare-EXPAND is a research project led jointly by hospitals in Alberta (Alberta's Children), Ontario (Children's Hospital of Eastern Ontario, Hospital for Sick Children), and Quebec (CHU Sainte-Justine, CHU de Sherbrooke, MUHC, CHU de Québec) with the support of three provincial genomic organizations (Ontario Genomics, Genome Alberta, and Génome Québec) and industry (Illumina, PacBio, ONT). In Quebec, Care4Rare-EXPAND relies on the Réseau québécois de diagnostic moléculaire (RQDM), which federates molecular diagnostic laboratories and Santé Québec's Centre québécois de génomique clinique hosted at CHU Sainte-Justine. The participation of the RQDM in the project will immediately allow the return of sequencing results to patients,

as well as the rapid evaluation and integration of new technologies into its service offering.

Ultimately, the project will lead to the discovery of new rare disease genes, while also increasing the impact of genomic sequencing for the diagnosis of rare diseases in Quebec. In coherence with the Plan d'action québécois sur les maladies rares 2023-2027, it will have positive impact in Canada and around the world.

## Quotes

"Rare diseases still represent a huge burden for some 3 million Canadian families, including 900,000 children. For many of them, science has not yet been able to make a diagnosis, although this is the first crucial step to medical care. CHU Sainte-Justine is proud to contribute its expertise to this large-scale project that promises to transform the care trajectory of families through faster and more accurate diagnoses, in order to ensure optimal care for children living with a rare genetic disease and their families."  
– **Mrs. Isabelle Demers, President and CEO, CHU Sainte-Justine**

"Powered by precision health, the Care4Rare-EXPAND project leverages the latest sequencing technologies, the vast potential of genomic data, and the cutting-edge expertise of Canadian genomic researchers. Relying on a vast network of stakeholders and scientific platforms, in Quebec and across the country, Care4Rare-EXPAND promises not only to advance knowledge on rare diseases but also and above all to translate this new knowledge into personalized and innovative treatments for the benefit of the entire population." – **Dr. Jacques L. Michaud, Principal Investigator and Director, Centre québécois de génomique Clinique**

"Genome Québec is proud to support the Care4Rare-EXPAND project, an initiative that demonstrates the tangible impact of genomics on the diagnosis of rare diseases. By collaborating with leading partners such as the Centre de recherche Azrieli du CHU Sainte-Justine, we are helping to advance research and generate critical evidence for the integration of genomics into clinical practice. These advancements pave the way for more precise and accessible healthcare for Quebec families." – **Mrs. Stéphanie Lord-Fontaine, Vice President, Scientific Affairs, Genome Québec**

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### **About de Réseau québécois de diagnostic moléculaire**

The Réseau québécois de diagnostic moléculaire (RQDM) is mandated to coordinate the deployment of diagnostic analyses involving next-generation sequencing within the OPTILAB network of Santé Québec. The analyses supported by the RQDM aim to assist in the investigation of cancers and genetic diseases in patients of all ages. The RQDM brings together the nine molecular diagnostic laboratories of Santé Québec and the Centre québécois de génomique clinique.

### **About the Centre québécois de génomique clinique**

The Centre québécois de génomique clinique (CQGC) is mandated to provide the entire OPTILAB network of Santé Québec with a high-throughput next-generation sequencing test offering, including exome, genome, and transcriptome sequencing, to more effectively identify and treat rare genetic diseases and certain cancers in patients of all ages. The CQGC, hosted at CHU Sainte-Justine, is an integral part of the OPTILAB network and the RQDM.

### **About the Quebec Genomic Data Center**

The Quebec Genomic Data Center is a data harmonization and sharing platform for clinical and genomic datasets produced by Québec's clinical and research studies. In accordance with its rigorous governance framework and the strictest standards for data security and protection, it offers the research community access to available research data and a powerful cloud computing facility for data analysis. Learn more: <https://cqdg.ca/en>

**About the Centre de recherche Azrieli du CHU Sainte-Justine**

The Centre de recherche Azrieli du CHU Sainte-Justine is a leading mother-child research institution affiliated with the Université de Montréal. It brings together 295 research investigators, including over 160 clinician-scientists, as well as more than 580 graduate and postgraduate students focused on finding innovative prevention means, faster and less invasive treatments, as well as personalized approaches to medicine. The centre is an integral part of [CHU Sainte-Justine](#), which is the largest mother-child centre in Canada. Learn more: [recherche.chusi.org](https://recherche.chusi.org)

**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, de l'Innovation et de l'Énergie du Québec (MEIE), the government of Canada, through Genome Canada, and private partners. To learn more, visit [www.genomequebec.com](http://www.genomequebec.com).

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