



Roadmap
2Rare

Genetic counselling services offered through Roadmap2Rare



Sanofi Canada is working in collaboration with Revvity Omics (formerly PerkinElmer Genomics) to offer the Roadmap2Rare Diagnostic Program (R2RDP), a complimentary (no charge) genetic and biochemical testing program available to Canadian clinicians with eligible patients.

The program provides diagnostic testing for Pompe, Fabry, MPS-I, ASMD, and Gaucher diseases and provides access to a **122-gene Muscle Disorders panel** and a **60-gene Hypertrophic Cardiomyopathy panel**. As part of the Roadmap2Rare Diagnostic Program, complimentary pre- and post-test genetic counselling is available and provided by Metis Genetics.

What is genetic counselling?

Genetic counselling is a process in which trained professionals, known as genetic counsellors, help individuals and families understand and adapt to the medical, psychological, and familial implications of genetic conditions or the risk of such conditions. The goal of genetic counselling is to provide support and guidance to individuals and families as they make informed decisions about genetic testing and the management of genetic conditions.

As part of the Sanofi Canada-sponsored Roadmap2Rare Diagnostic Program (R2RDP), genetic counselling is available at no charge through Metis Genetics.

Is genetic counselling mandatory?

No, genetic counselling is not required for a patient to have testing through Roadmap2Rare nor for a patient to receive their test result.

What role does Sanofi Canada play?

The R2RDP is a Sanofi Canada-sponsored initiative that includes the Metis Genetics counselling service, which means it is available at no charge. As the program sponsor, Sanofi Canada may receive de-identified and aggregated data for program evaluation, education planning purposes, and/or scientific study.

De-identified data are data from which personal identifiers have been removed. The identifying information of both patients and clinicians is not shared with Sanofi Canada. If a patient receives pre-test genetic counselling from Metis Genetics, they are not obligated to proceed forward to testing through the program. If a patient receives a diagnosis after being tested through the program, they are in no way obligated to be treated with a medication that Sanofi manufactures.



What topics can Metis Genetics cover with my patient?

Metis Genetics can provide both pre-test counselling and post-test counselling for patients.

Pre-test genetic counselling

Pre-test appointments are educational and can help a patient determine if testing is appropriate based on their personal and family medical history. If testing is clinically appropriate, Metis's genetic counsellors can support patients in deciding if they want to pursue testing. This may include:

- a general overview of genetics and DNA analysis
- the benefits and limitations of testing
- potential results and their clinical implications
- how a test result can be useful to a patient's and family's healthcare

Post-test genetic counselling

Post-test appointments are for patients who have received a result through the R2RDP, including on either of the R2RDP's two multi-gene panels, regardless of the result. Metis's genetic counsellors can help with:

- test result interpretation
- explanation of test result implications
- recommended next steps for the patient or their family members

Note: The Roadmap2Rare Diagnostic Program is not intended to and should not interfere in any way with a healthcare professional's or patient's independent judgement and freedom of choice in the testing and treatment options for these diseases. Healthcare professionals and patients should always consider the full range of testing and treatment options and select those most appropriate for the individual patient.

Who should I contact if I have questions, and how do I access the genetic counselling service for my patient?

To access this service, a referral form must be completed and submitted to Metis Genetics. To request a referral form for your patient, or if you have any questions about this service, please contact Revvity Omics (formerly PerkinElmer Genomics) at genomics@revvity.com. Referral forms can also be obtained by contacting Sanofi Canada's Roadmap2Rare team using the Contact Us form on www.roadmap2rare.ca.



Preparing for a Metis Genetics appointment

All Metis Genetics appointments are held over the phone or through a video call and can typically be arranged within two days of request. For post-test counselling, patients must consent on the referral form to their results being shared with Metis.