

## Roadmap2Rare Diagnostic Program Overview



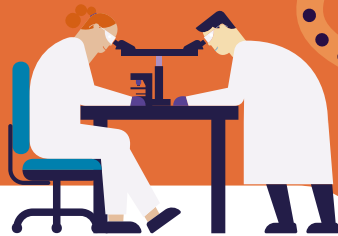
The Roadmap2Rare Diagnostic Program is a Sanofi Canada-sponsored testing program for Pompe, Fabry, Gaucher, ASMD, and MPS-I diseases, offered in collaboration with Revvity Omics.



## Roadmap *2 Rare*



The program includes single-disorder diagnostic testing for Pompe, Fabry, Gaucher, MPS-I, and ASMD. Healthcare providers can order specimen collection kits for in-office collections or can elect to have Revvity Omics manage the collection by coordinating mobile phlebotomy. Diagnostic test results are provided within 5 weeks from specimen receipt at the laboratory.



Revvity Omics manages the specimen shipping logistics, diagnostic testing, and offers complimentary results interpretation/consultation for healthcare providers. All Roadmap2Rare Diagnostic Program services offered by Revvity Omics are provided at no charge.

Patient and physician identifying information provided to Revvity Omics remains confidential and is not shared with Sanofi.

	Tests	Eligibility criteria	Sample types	Sample Collection Options
<b>Pompe</b>	Acid alpha-glucosidase enzyme assay with reflex to <i>GAA</i> sequencing	Clinical suspicion of Pompe disease (for suspected IOPD cases, STAT testing is available)	DBS	Clinician collection or mobile phlebotomy
<b>Fabry (male)</b>	Alpha-galactosidase A enzyme assay with reflex to <i>GLA</i> sequencing and Lyso-GL3	Clinical suspicion of Fabry disease	DBS	Clinician collection or mobile phlebotomy
<b>Fabry (female)</b>	<i>GLA</i> sequencing with reflex to Lyso-GL3	Clinical suspicion of Fabry disease	DBS	Clinician collection or mobile phlebotomy
<b>ASMD</b>	Acid sphingomyelinase (ASM) enzyme assay with reflex to <i>SMPD1</i> sequencing	Clinical suspicion of Acid Sphingomyelinase Deficiency (ASMD)	DBS	Clinician collection or mobile phlebotomy
<b>Gaucher</b>	Glucocerebrosidase enzyme assay (includes ASM enzyme assay in parallel) with reflex to <i>GBA</i> and Lyso-GL1 or <i>SMPD1</i> sequencing as appropriate	Clinical suspicion of Gaucher disease (ASMD testing is performed in parallel)	DBS	Clinician collection or mobile phlebotomy
<b>MPS-1</b>	Alpha-iduronidase enzyme assay with reflex to <i>IDUA</i> sequencing	Clinical suspicion of Mucopolysaccharidosis Type I (MPS-I)	DBS	Clinician collection or mobile phlebotomy



In addition to the above, familial variant testing is available if there is family history and the genetic variant is known. Standalone enzyme assays are available for confirmatory analysis if genetic testing was completed elsewhere.



For more detailed information about the Roadmap2Rare Diagnostic Program services, please visit the Roadmap2Rare landing page on the Revvity Omics website. You can be directed to this landing page by visiting [www.roadmap2rare.ca/laboratory](http://www.roadmap2rare.ca/laboratory)

*Or scan the QR code on the right.*



If you are ready to test, please navigate to the Roadmap2Rare landing page on the Revvity Omics website by visiting [www.roadmap2rare.ca/laboratory](http://www.roadmap2rare.ca/laboratory).

Here you can request a kit to collect your patient's sample or indicate your preference for Revvity Omics to coordinate the collection. If you would like to create an account with Revvity Omics, please contact [genomics@revvity.com](mailto:genomics@revvity.com).

Patient consent is required prior to testing.