Roadmap 2 Rare

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 (formerly PerkinElmer Genomics).

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The program includes access to two multi-gene panels, the **Muscle Disorders Panel** (122 genes) and the **Hypertrophic Cardiomyopathy Panel** (60 genes), that cover several differential diagnoses for Pompe and Fabry disease, respectively. The program also 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 patient self-collection or 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, as well as genetic counselling services for patients. 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.

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	Tests	Eligibility criteria	Sample types	Sample Collection Options
Pompe	Acid alpha-glucosidase enzyme assay with reflex to *GAA* sequencing	Clinical suspicion of Pompe disease (for suspected IOPD cases, STAT testing is available)	DBS	Clinician collection *or* mobile phlebotomy
Pompe	122-gene Muscle Disorders panel (focused whole exome sequencing)	Laboratory evidence suggestive of a muscle pathology IN ADDITION to muscle weakness OR unexplained respiratory insufficiency OR other symptom(s) supporting muscle involvement	DBS or Saliva swab	Clinician collection *or* patient self-collection
Fabry & Pompe	60-gene Hypertrophic Cardiomyopathy panel (focused whole exome sequencing)	Clinical diagnosis of cardiomyopathy or cardiac arrythmia of unknown etiology OR confirmed family history of hypertrophic cardiomyopathy of unknown etiology	DBS or Saliva swab	Clinician collection *or* patient self-collection
Fabry (male)	Alpha-galactosidase A enzyme assay with reflex to *GLA* sequencing and Lyso-GL3	Clinical suspicion of Fabry disease	DBS	Clinician collection *or* mobile phlebotomy
Fabry (female)	*GLA* sequencing with reflex to Lyso-GL3	Clinical suspicion of Fabry disease	DBS	Clinician collection *or* mobile phlebotomy
ASMD	Acid sphingomyelinase (ASM) enzyme assay with reflex to *SMPD1* sequencing	Clinical suspicion of Acid Sphingomyelinase Deficiency (ASMD)	DBS	Clinician collection *or* mobile phlebotomy
Gaucher	Glucocerebrosidase enzyme assay (includes ASM enzyme assay in parallel) with reflex to *GBA* and Lyso-GL1 or *SMPD1* sequencing as appropriate	Clinical suspicion of Gaucher disease (ASMD testing is performed in parallel)	DBS	Clinician collection *or* mobile phlebotomy
MPS-1	Alpha-iduronidase enzyme assay with reflex to *IDUA* 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

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.

Here you can request a kit to collect your patient's sample or indicate your preference for Revvity Omics to coordinate the collection, either through mobile phlebotomy or patient self-collection. If you would like to create an account with Revvity Omics, please contact genomics@revvity.com.

Patient consent is required prior to testing.

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