Roadmap 2 Rare

How Sanofi can support Fabry family tree screening (FTS)

Fabry disease is an X-linked disorder of glycosphingolipid metabolism caused by an alpha-galactosidase A deficiency, which leads to an accumulation of glycosphingolipids and potentially life-threatening complications.[1,2]

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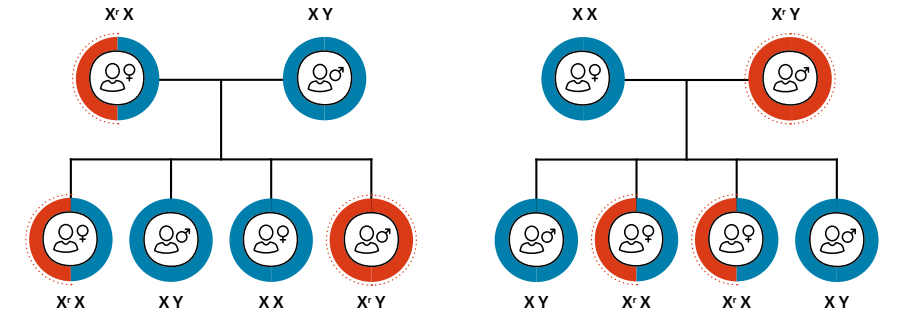
Introduction to Fabry

Due to its X-linked inheritance pattern, an affected mother (XX) will have a 50% chance of passing on a *GLA* mutation to any of her children, and an affected father (XY) a 100% chance of passing on a *GLA* mutation to his daughters (but not to his sons).[1,2]

For this reason, family tree screening (identifying and testing family members for Fabry disease) is important as it may help in diagnosing patients at an earlier stage of their disease.



All children of affected mothers have a 50% chance of inheriting a mutation regardless of gender.

All daughters of affected fathers will inherit a mutation; no sons of affected fathers will inherit the mutation

***GLA* mutation** **No *GLA* mutation**

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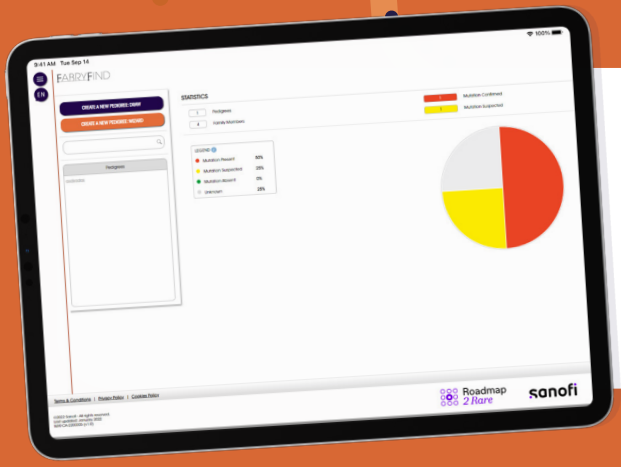
The FABRY FIND Tool

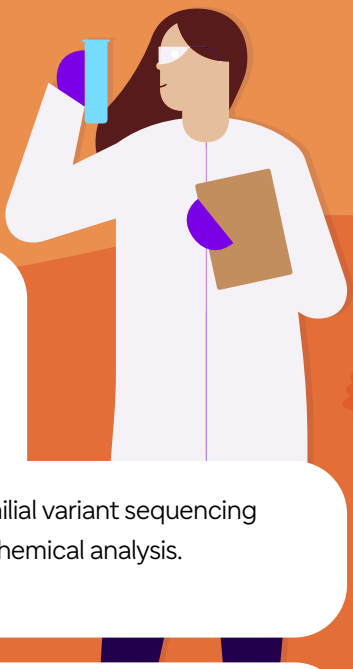
The FABRY FIND tool can help clinicians create family trees and visualize the Fabry risk of family members after the identification of a proband patient.



To use the FABRY FIND tool, visit www.fabryfind.ca to create a new family tree/pedigree.

Reports can be exported as a PDF file.

Fabry Testing



If Fabry disease is suspected, one available testing option is the ***Roadmap2Rare Diagnostic Program (R2RDP)**, a Sanofi Canada-sponsored testing program for rare diseases, including Fabry disease. The R2RDP is offered in collaboration with Revvity Omics.

Testing for Fabry disease within the R2RDP includes familial variant sequencing (if the familial variant is known) or *GLA* sequencing/biochemical analysis. This testing is offered at no charge.

Order kits

Order kits through Revvity's kit order URL for in-clinic specimen collections at
https://apps-omics.revvity.com/roadmap2rare

Collection support

For mobile phlebotomy, upload a completed test requisition and informed consent form to Revvity's test request URL (https://apps-omics.revvity.com/roadmap2rare/order)

Questions about laboratory testing

Contact Revvity at genomics@revvity.com or visit Revvity's Roadmap2Rare landing page at
https://www.revvity.com/ca-en/category/roadmap-2-rare

Clinician Support

If clinicians have ordered testing through the R2RDP, the Revvity Omics team can support clinicians with results interpretation.

Fabry Monitoring

For patients diagnosed with Fabry disease, Lyso-GL3 biomarker monitoring is available at no charge through the Sanofi-sponsored **Rare Disease Specialty Testing Program (RDSTP)**. This biomarker testing can help clinicians monitor disease evolution and is available regardless of a patient's treatment status.

For complimentary support in the procurement of the required RDSTP collection supplies, and/or for in-clinic collection support, please contact your local Medical Lead to be connected with nurses from the Shoppers Drug Mart Specialty Health Network (SHN).

*The Roadmap2Rare Diagnostic Testing Program is not intended to and should not interfere in any way with a clinician's or patient's independent judgment and freedom of choice in the testing and treatment options for these diseases. Clinicians and patients should always consider the full range of testing and treatment options and select those most appropriate for the individual patient. If a patient receives a diagnosis after being tested through the R2RDP, they are in no way obligated to be treated with a medication that Sanofi manufactures.

The identifying information of patients and clinicians is not shared with Sanofi Canada.

References: 1. Germain DP. Fabry disease. *Orphanet J Rare Dis*. 2010;5:30. **2.** Rozenfeld PA, Masllorens FM, Roa N, et al. Fabry pedigree analysis: A successful program for targeted genetic approach. *Mol Genet Genomic Med*. 2019;7(7):e00794.

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