

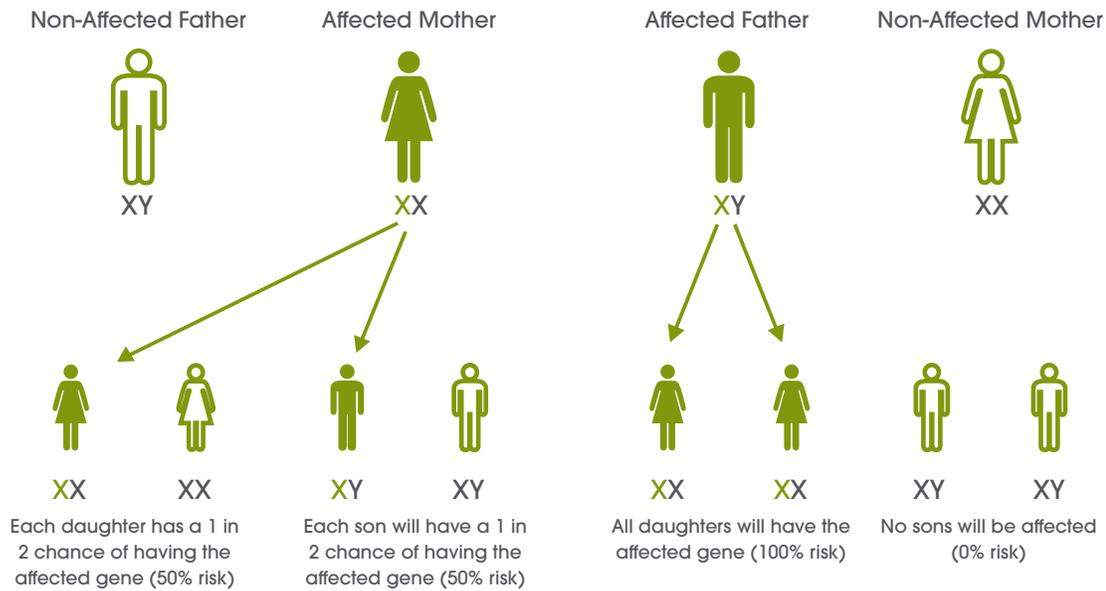
FABRY DISEASE

DIAGNOSTIC TESTING

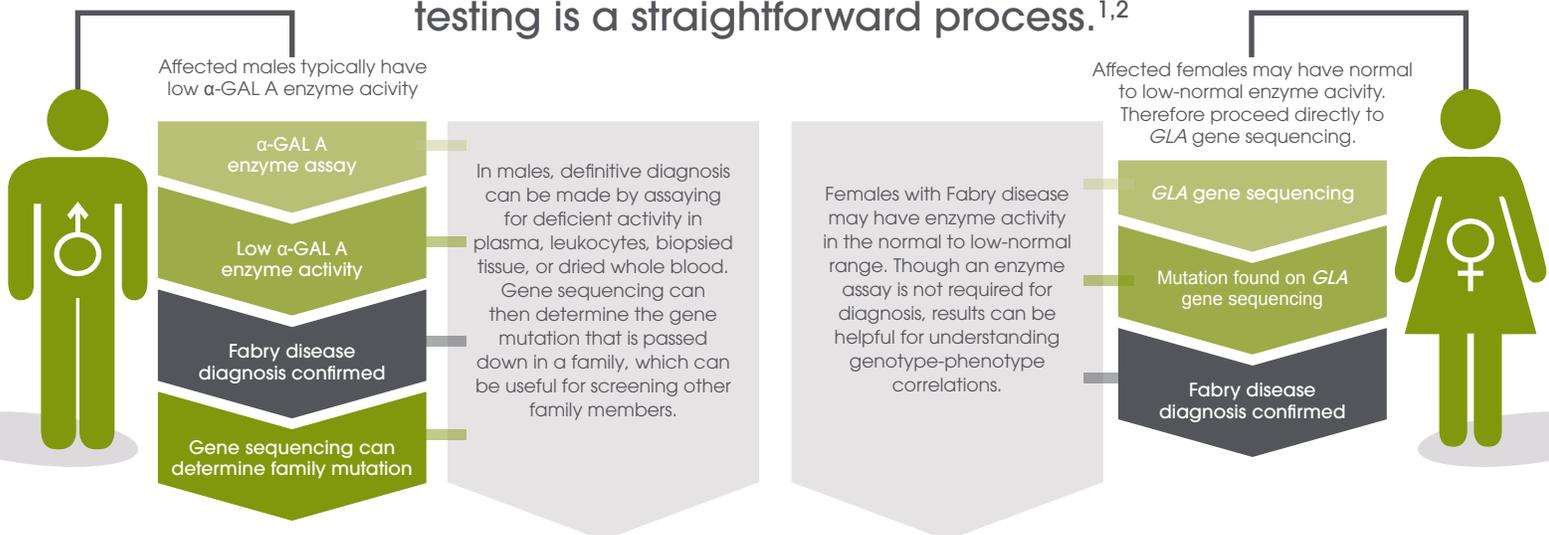
Fabry disease is a progressive and often life-threatening X-linked genetic disorder that affects men, women, and children. Fabry disease is caused by complete or partial deficiency of the lysosomal enzyme α -galactosidase A (α -GAL A), which leads to the accumulation of globotriaosylceramide (GL-3) in cells in the kidney, heart, and skin.^{1,2}

Identification of one family member can enable earlier screening for others³

- A woman with Fabry disease may have inherited it from either her mother or her father
- A man with Fabry disease could only have inherited it from his mother. Because the disease is X-linked, a father will not pass it on to his sons but will pass it on to all of his daughters



If you suspect **Fabry disease** in a patient, testing is a straightforward process.^{1,2}



Physicians are directed to contact the individual testing programs and/or laboratories for information on obtaining test requisitions, sample requirements, turnaround time, and the specific details of the services provided.

For commercial lab testing, coding accurately and obtaining reimbursement are the responsibility of the provider submitting a claim for the item or service.

SEE LISTING OF LABS ON REVERSE SIDE

Testing Options for Fabry Disease

Sanofi Genzyme does not review or control the content of non-Sanofi Genzyme websites. These listings do not constitute an endorsement by Sanofi Genzyme of information provided by any other organizations. Below is a selection of laboratories offering both α -GAL A enzyme assay and *GLA* sequencing. This is not an exhaustive list of labs that offer one or the other or an endorsement of any one lab. Other testing options can be found at www.concertgenetics.com or www.ncbi.nlm.nih.gov/gtr. To test individuals with a family history of Fabry for a known familial mutation, please contact your lab of choice to discuss. Content is current at time of printing, and tests may not be available in all states, so please call the laboratory to confirm.

Lab	Available Testing	Sample Requirements	Kits	Avg TAT	Mobile Blood Draw	Billing	Contact
Duke University	Enzyme	WB: 3-5 ml EDTA (lavender) tube; DBS card: 5 spots	No	15 d	No	Inst	P: 919-613-8400 W: https://testcatalog.duke.edu
	Sequencing	WB: 2 ml EDTA (lavender) tube; DBS card: 5 spots		28 d			
	Lyso-GL3 Biomarker	WB: 4 ml EDTA (lavender) tube		28 d			
The Fabry Diagnostic Project (performed at EGL)	Enzyme	WB: 5-10 ml heparin (green) tube	Blood, Saliva	7-10 d	No	No charge*	P: 800-200-1524 E: fabry.testing@emory.edu W: www.genetics.emory.edu
	Sequencing	WB: 5-10 ml EDTA (lavender) or ACD (yellow) tube		4 wks			
	Del/Dup	WB: 5-10 ml EDTA (lavender) or ACD (yellow) tube		4 wks			
Greenwood Genetic Center	Enzyme	WB: 5-10 ml heparin (green) tube; DBS card: 3 circles	Blood, DBS, Saliva	2 wks	No	Inst, Self-pay, Ins (SC residents only)	P: 800-473-9411 E: labgc@ggc.org W: www.ggc.org
	Sequencing	WB: 5-7 ml EDTA (lavender) tube; DBS card: 3 circles; Saliva (EDTA blood only for del/dup)		2 wks			
	Del/Dup	WB: 5-7 ml EDTA (lavender) tube		26 d			
LabCorp/Integrated Genetics	Enzyme	WB: 5-10 ml ACD (yellow) tube	Blood	7-14 d	Yes	Inst, Ins, Self-pay	LabCorp Customers: P: 800-345-4363 W: www.labcorp.com Integrated Customers: P: 800-848-4436 E: ask!Gclientservices@integratedgenetics.com W: www.integratedgenetics.com
	Sequencing	WB: 7 ml EDTA (lavender) tube or ACD (yellow) tube		21 d			
The Lantern Project (performed at PerkinElmer Genomics)	Enzyme	WB: 3-5 ml heparin (green) tube; DBS card: 5 circles	Blood, DBS, Saliva	3 d	Yes	No charge*	P: 866-354-2910 E: genomics@perkinelmer.com W: www.LanternProjectDx.com
	Sequencing (including Del/Dup)	WB: 3-5 ml EDTA (lavender) tube; DBS card: 5 circles, Saliva: (Oragene)		3 wks			
	Lyso-GL3 Biomarker	DBS: 2 circles		3 d			
Mayo Medical Laboratories	Enzyme	WB: 6 ml ACD (yellow) tube; DBS card: 2 spots; serum: 2 ml (red top tube)	DBS (in some cases), Saliva	8-15 d	Yes	Inst (Ins can be billed in some cases, Inst acct required)	P: 800-533-1710 E: mml@mayo.edu W: www.mayomedicallaboratories.com
	Sequencing	WB: 3 ml EDTA (lavender) or ACD (yellow) tube; DBS card: 2-5 spots		14-20 d			
	Lyso-GL3 Biomarker	Serum: 0.5-1 ml red top tube		8-15 d			
Mount Sinai International Center for Fabry Disease	Enzyme	WB: 20 ml sodium heparin (green) tube	Blood, Buccal	7-10 d	No	No charge*	P: 866-322-7963 E: fabry.disease@mssm.edu W: https://icahn.mssm.edu/research/Fabry
	Sequencing	WB: 20 ml sodium heparin (green) tube; 2 buccal brushes		10-14 d			
Sema4	Enzyme	WB: 2 x 5-10 ml sodium heparin (green) or EDTA (lavender) tubes. Minimum of 3 ml	Blood, Saliva	7-10 d	Yes	Inst, Ins, Self-pay	P: 800-298-6470 E: clientservices@sema4.com W: www.sema4.com
	Sequencing	WB: 2 x 5-10 ml EDTA (lavender); Saliva		2-3 wks			

***Testing is performed at no charge; local charges may apply for sample collection, processing, or shipping.**

acct = account; avg TAT = average turnaround time; d = days; DBS = dried blood spot; del = deletion; dup = duplication; E = email, Ins = insurance; Inst = institution; lyso-GL3 = globotriaosylsphingosine; P = phone, W = website, WB = whole blood; wks = weeks.