

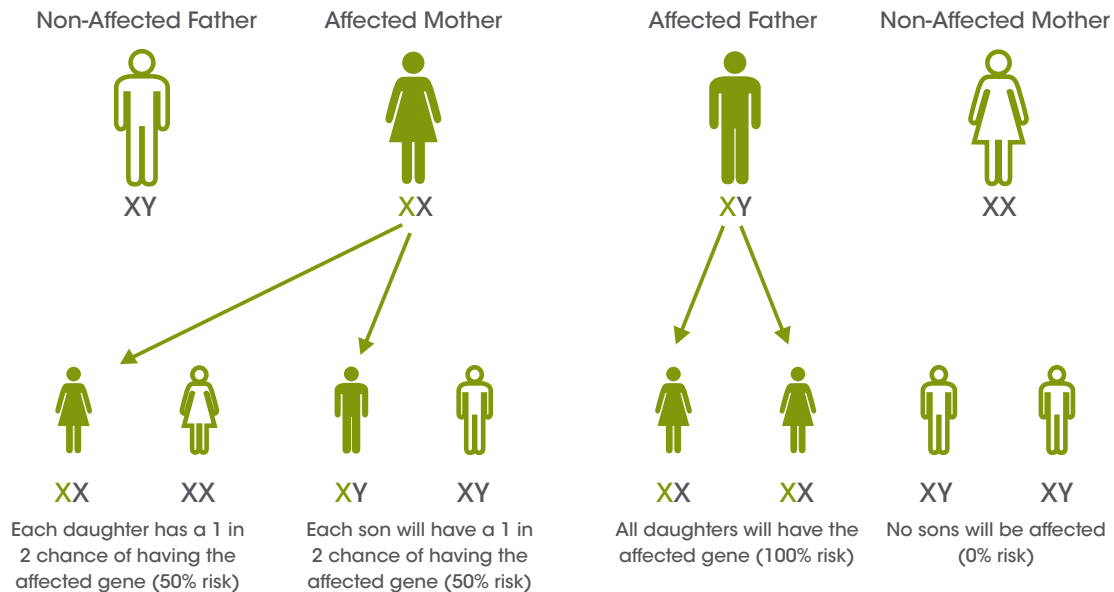
FABRY DISEASE

DIAGNOSTIC TESTING

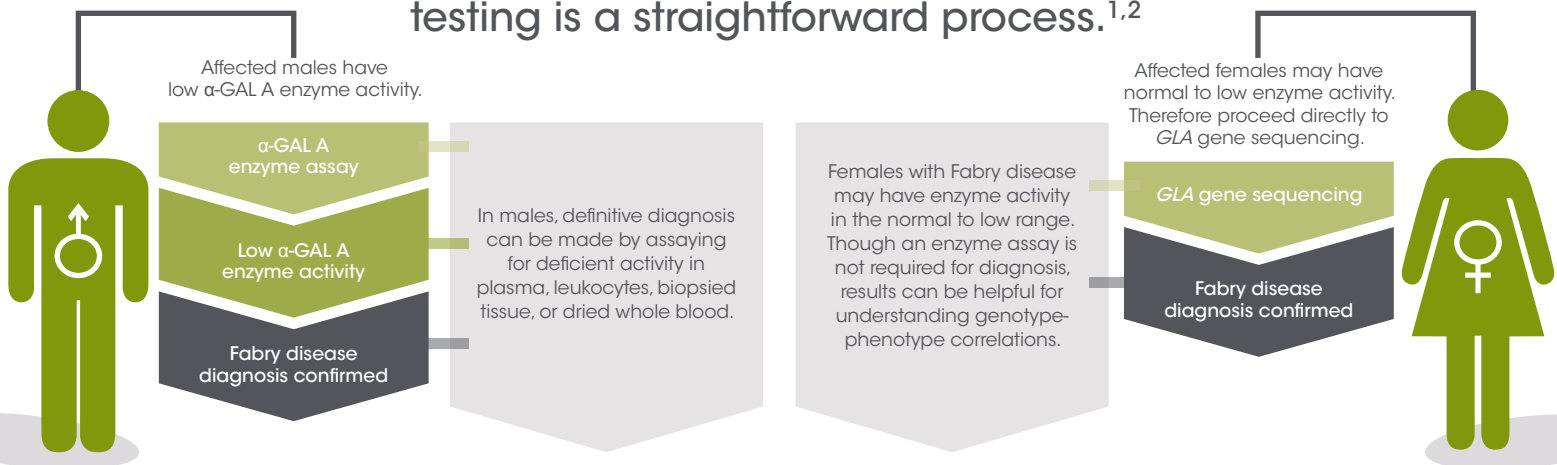
Fabry disease is a progressive and often life-threatening X-linked genetic disorder that affects men, women, and children. Fabry disease is caused by complete or partial deficiency of the lysosomal enzyme α -galactosidase A (α -GAL A), which leads to the accumulation of globotriaosylceramide (GL-3) in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

Identification of one family member can enable earlier screening for others³

- A female with Fabry disease may have inherited it from either her mother or her father
- A male 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}



For every index patient diagnosed, an average of 5 affected family members may be identified. It is important to conduct family testing.³

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 NEXT PAGES

Testing Options for Fabry Disease

Some of the laboratories offering diagnostic testing for Fabry disease are listed below. There may be other diagnostic testing appropriate for your patient, and this is not an endorsement of any specific lab. Other testing options can be found at www.concertgenetics.com or www.ncbi.nlm.nih.gov/gtr. Consult each laboratory for a full range of options. Content is current at time of publication, and tests may not be available in all states; please call laboratory to confirm test availability, sample shipping information, and all other logistics. Sanofi does not review or control the content of non-Sanofi websites. This listing does not constitute an endorsement by Sanofi of information provided by any other organizations.

Lab	Available Testing	Sample Requirements	Kits	Avg TAT	Mobile Blood Draw	Billing	Contact
Centogene	Enzyme	WB: 1ml EDTA (lavender) tube; DBS card: 10 circles	Blood, DBS, Saliva	7 d	Yes	Inst, Self-pay, Ins	P: 617-580-2102 E: customer.support-US@centogene.com W: https://www.centogene.com
	Sequencing	WB: 1ml EDTA (lavender) tube; DBS card: 10 circles; Saliva; Buccal swab		15-25 d			
	Del/Dup	WB: 1ml EDTA (lavender) tube; DBS card: 10 circles; Saliva; Buccal swab		15 d			
Duke University	Enzyme	WB: 3-5 ml EDTA (lavender) tube; DBS card: 5 circles	No	15 d	No	Inst	P: 919-613-8400 E: clientservices@dm.duke.edu W: https://testcatalog.duke.edu
	Sequencing	WB: 2 ml EDTA (lavender) tube; DBS card: 5 circles		28 d			
	Lyso-GL3	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 or 404-778-8518 E: fabry.testing@emory.edu W: www.genetics.emory.edu/patient-care/lysosomal-storage-disease-center
	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: 5-6 ml EDTA (lavender) tube; DBS card: 3 circles; Saliva		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	7-10 d	Yes	Inst, Ins, Self-pay	LabCorp Customers: P: 800-345-4363 W: www.labcorp.com Integrated Customers: P: 800-848-4436 E: askIGclientservices@integratedgenetics.com W: www.integratedgenetics.com
	Sequencing	WB: 7 ml EDTA (lavender) tube or ACD (yellow) tube		18-21 d			
The Lantern Project (performed at PerkinElmer Genomics)	Enzyme	WB: 2-10 ml EDTA (lavender) tube (volume varies with age); DBS card: 3 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: 2-10 ml EDTA (lavender) tube (volume varies with age); DBS card: 3 circles; Saliva; (Oragene)		3 wks			
	Lyso-GL3	DBS: 2 circles		3 d			
Mayo Clinic 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: mcl@mayo.edu W: www.mayocliniclabs.com
	Sequencing	WB: 3 ml EDTA (lavender) or ACD (yellow) tube; DBS card: 2-5 spots		14-20 d			
	Lyso-GL3 (LGB3S, LGBWB, LGBBS)	WB: 1 ml EDTA (lavender); Serum: 1 ml (red top tube); DBS: 2 circles		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)	Blood, Saliva	7-10 d	Yes	Inst, Ins, Self-pay	P: 800-298-6470 E: clientservices@sema4.com W: www.sema4.com
	Sequencing	WB: 5-10 ml ACD (yellow) AND 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; Ins = insurance; Inst = institution; lyso-GL3 = globotriaosylsphingosine; WB = whole blood; wks = weeks.

Testing with Cardiology Genetic Panels

Some of the laboratories offering diagnostic testing for Fabry disease are listed below. There may be other diagnostic testing appropriate for your patient, and this is not an endorsement of any specific lab. Content is current at time of publication, and tests may not be available in all states; please call laboratory to confirm test availability, sample shipping information, and all other logistics. Sanofi does not review or control the content of non-Sanofi websites. This listing does not constitute an endorsement by Sanofi of information provided by any other organizations.

Lab	Panel (Test Code)	# of Genes	Sample Requirements	Kits	Avg TAT	Mobile Blood Draw	Genetic Counselor Available to Patients	Billing	Contact
Amry Genetics	Comp. Cardiology (8911)	92	WB: 6-10 mL EDTA (lavender) tube; Saliva	Blood; Saliva	14-21 d	Yes	No	Inst, Self-Pay, Ins	P: 919-900-5500 E: info@amrygen.com W: https://www.amrygen.com
	Cardiomyopathy (8887)	56							
	HCM (8936)	30							
ARUP Laboratories	Comp. Cardiology (2010183)	78	WB: 3 mL EDTA (lavender) or ACD (yellow) tube	Shipping containers	3-6 w	No	Yes	Self-Pay, Ins	P: 1-800-522-2787 E: clientservices@aruplab.com W: https://www.aruplab.com
	HCM (3001579)	34							
Blueprint Genetics	Comp. Cardiology (CA1301)	260	WB: 1 mL EDTA (lavender) tube; Saliva	Blood	4 w	No	No	Inst, Self-Pay, Ins	P: 650-452-9340 Ext. 0 E: support.us@blueprintgenetics.com W: https://blueprintgenetics.com
	Cardiomyopathy (CA1201)	217							
	HCM (CA1901)	92							
CGC Genetics	Comp. Cardiology (5568)	140	WB: 3mL EDTA (lavender) tube	No	60 d	No	Yes	Inst, Self-Pay, Ins	P: 973-623-1264 E: info@cgcgenetics.com W: https://www.cgcgenetics.com
	HCM (5569)	68							
Fulgent	Comp. Cardiology	252	WB: two 4 mL EDTA (lavender) tubes; Saliva; Buccal swab	Blood; Saliva; Buccal	3-5 w	No	No	Inst, Self-Pay, Ins	P: 626-350-0537 E: info@fulgentgenetics.com W: https://www.fulgentgenetics.com
	Cardiomyopathy	129							
	HCM	86							
GeneDX	Comp. Cardiology (935)	138	WB: 2-5 mL EDTA (lavender) tube (preferred); Buccal swab	Blood; Buccal	4 w	No	Yes	Inst, Self-Pay, Ins	P: 301-519-2100 E: zebras@genedx.com W: https://www.genedx.com
	Cardiomyopathy (694)	101							
	HCM (J553)	42							
Invitae	Cardiomyopathy (02251)	121	WB: 3 mL EDTA (lavender) tube (preferred); Saliva; Buccal swab	Blood; Saliva; Buccal	10-21 d	Yes	Yes	Inst, Self-Pay, Ins	P: 800-436-3037 E: clinconsult@invitae.com W: https://www.invitae.com
	HCM (02261)	44							
	Detect	100						*no charge	
PerkinElmer Genomics	Cardiomyopathy (D4101)	122	WB: 5-10 mL EDTA (lavender) tube (preferred); DBS: 1 card; Saliva	Blood, DBS, Saliva	3-5 w	No	No	Inst, Self-Pay, Ins	P: 617-580-2102 W: https://www.perkinelmergenomics.com
	HCM (D4103)	60							
Prevention Genetics	Comp. Cardiology (2663)	164	WB: 3 mL EDTA (lavender) tube (preferred); Saliva; Buccal swab	Blood, Saliva	18 d	No	No	Inst, Self-Pay, Ins	P: 866-354-2910 E: support@preventiongenetics.com W: https://www.preventiongenetics.com
	Cardiomyopathy (5263)	99							
	HCM (1313)	50							

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

Avg TAT = average turnaround time; cardiomyopathy = comprehensive cardiomyopathy panel; comp. cardiology = comprehensive cardiology panel; d = days; DBS = dried blood spot; Detect = Detect Cardiomyopathy and Arrhythmia Panel; HCM = hypertrophic cardiomyopathy panel ins = insurance; inst = institution; WB = whole blood; w = weeks