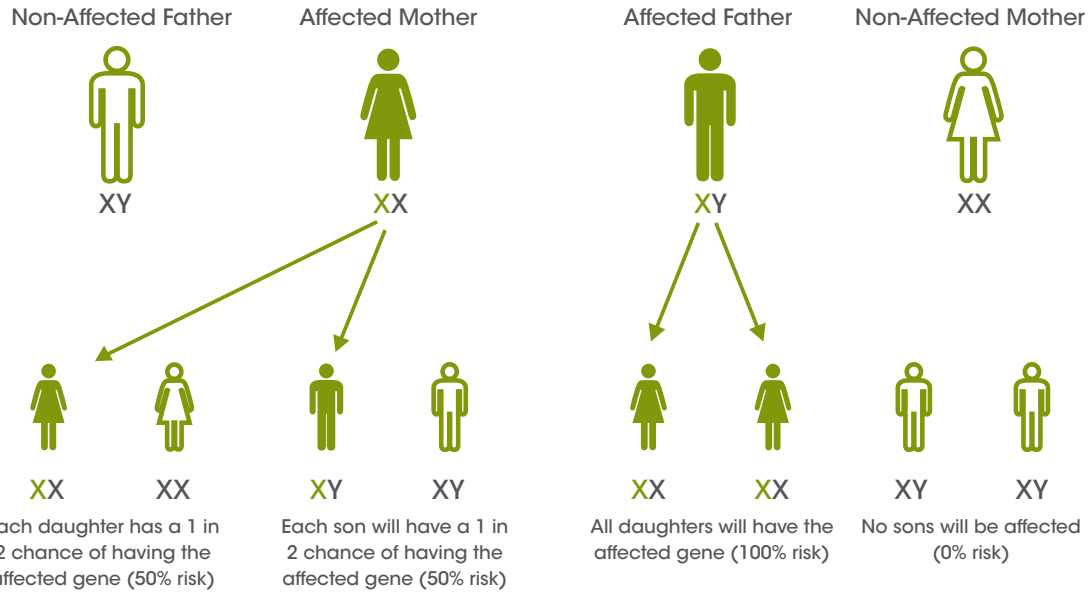
FABRY DISEASE

DIAGNOSTIC TESTING

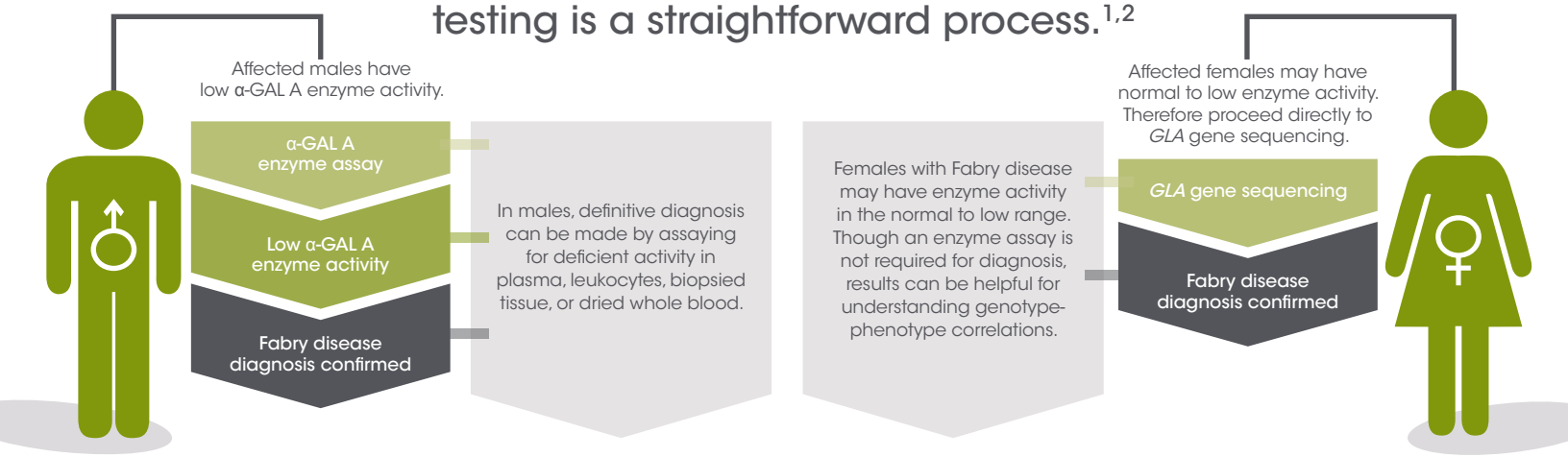
Fabry disease is a progressive and often life-threatening X-linked genetic disorder that affects men, women, and children. Fabry disease is caused by complete or partial deficiency of the lysosomal enzyme α-galactosidase A (α-GAL A), which leads to the accumulation of globotriaosylceramide (GL-3) in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

Identification of one family member can enable earlier screening for others[3]

- 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/lysosom/ -al-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.com **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:** asklGclientservices@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 Nephrology 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
CGC Genetics	Idiopathic Renal Failure on Young (5015)	171	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
GeneDx	Nephrotic Syndrome/FSGS (TG99)	55	WB: 2-5 mL EDTA (lavender) tube (preferred); Buccal swab	Blood, Buccal	4 w	No	Yes	Self-Pay, Ins	**P:** 301-519-2100 **E:** zebras@genedx.com **W:** https://www.genedx.com
Invitae	Progressive Renal Disease (75000)	195	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
Iowa Institute of Human Genetics	KidneySeq Comprehensive Panel	330	WB: 6mL EDTA (lavender) tube	No	30 d	No	No	Inst	**P:** 319-335-3688 **E:** clinicaldivision@healthcare.uiowa.edu **W:** https://medicine.uiowa.edu/humangenetics
	KidneySeq Glomerulopathies Panel	68							
Johns Hopkins DNA Diagnostic Laboratory	RenalZoom Glomerular Diseases and Complement Testing	118	WB: 3-6 mL EDTA (lavender) tube; Saliva	No	6-8 w	No	No	Inst, Self-Pay, Ins	**P:** 410-955-0483 **E:** ddl@jhmi.edu **W:** https://www.hopkinsmedicine.org/dnadiagnostic
Natera	Renasight	385	WB: 6 mL EDTA (lavender) tube; Buccal swab	Blood, Buccal	3 w	Yes	Yes	Inst, Self-Pay, Ins	**P:** 415-619-5054 **W:** https://www.natera.com
Prevention Genetics	Comprehensive Inherited Kidney Diseases (13990)	326	WB: 3-5 mL EDTA (lavender) or ACD (yellow) tube; DBS: 5 spots; Saliva	Blood, Saliva,	18 d	No	No	Inst, Self-Pay, Ins	**P:** 715-387-0484 **E:** support@preventiongenetics.com **W:** https://www.preventiongenetics.com

Avg TAT = average turnaround time; d = days; DBS = dried blood spot; FSGS = focal segmental glomerular sclerosis; ins = insurance; inst = institution; WB = whole blood; w = weeks

References: 1. Smid BE, et al. *J Med Genet*. 2015;52:262–268. **2.** Ortiz A, et al. *Mol Genet Metab*. 2018;123:416–427. **3.** Laney DA, Fernhoff PM. *J Genet Counsel*. 2008;17:79-83.