

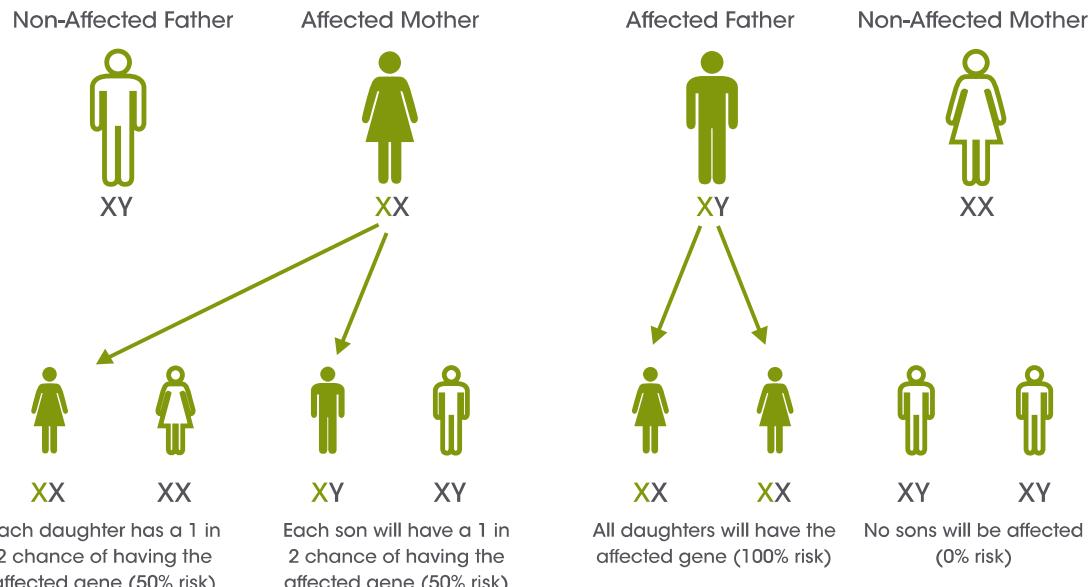
# FABRY DISEASE

## DIAGNOSTIC TESTING

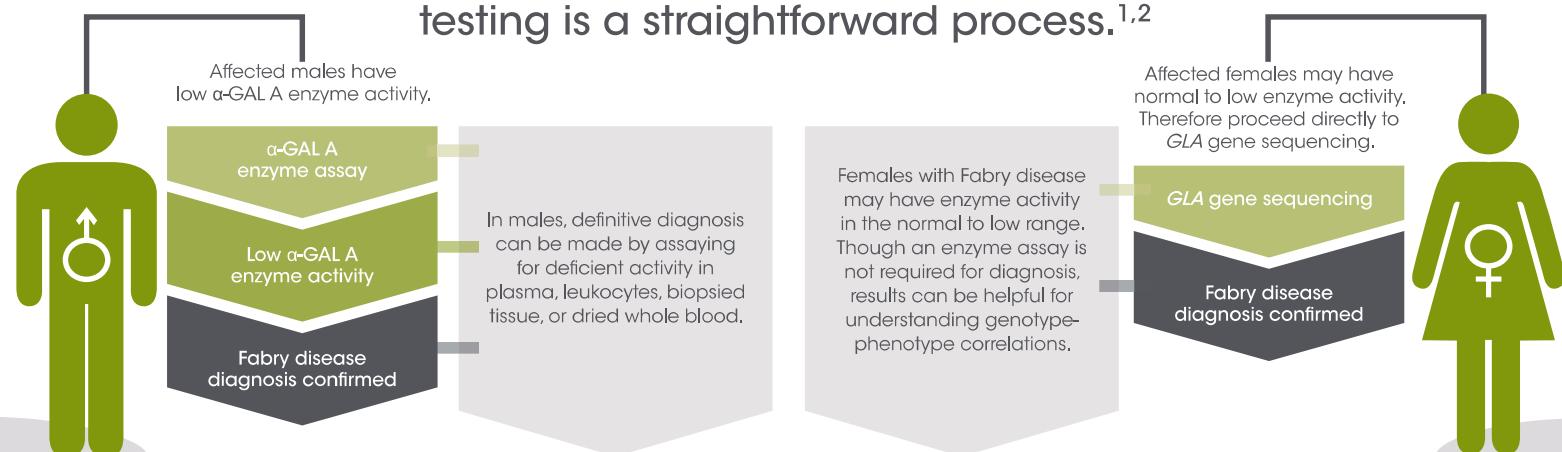
Fabry disease is a progressive and often life-threatening X-linked genetic disorder that affects men, women, and children. Fabry disease is caused by complete or partial deficiency of the lysosomal enzyme  $\alpha$ -galactosidase A ( $\alpha$ -GAL A), which leads to the accumulation of globotriaosylceramide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

### Identification of one family member can enable earlier screening for others<sup>3</sup>

- 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.<sup>1,2</sup>



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

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

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.ncbi.nlm.nih.gov/dtr](http://www.ncbi.nlm.nih.gov/dtr). 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 Genzyme does not review or control the content of non-Sanofi Genzyme websites. This listing does not constitute an endorsement by Sanofi Genzyme of information provided by any other organizations.

Lab	Available Testing	Sample Requirements	Kits	Avg TAT	Mobile Blood Draw	Billing	Contact
Centogene	Enzyme Sequencing	WB: 1ml EDTA (lavender) tube; DBS card: 10 circles WB: 1ml EDTA (lavender) tube; DBS card: 10 circles; Saliva; Buccal swab	Blood, DBS, Saliva	7 d 15-25 d	Yes	Inst, Self-pay, Ins	P: 617-580-2102 E: customersupport-US@centogene.com W: <a href="https://www.centogene.com">https://www.centogene.com</a>
	De/Dup	WB: 1ml EDTA (lavender) tube; DBS card: 10 circles; Saliva; Buccal swab		15 d			
Duke University	Enzyme Sequencing	WB: 3-5 ml EDTA (lavender) tube; DBS card: 5 circles WB: 2 ml EDTA (lavender) tube; DBS card: 5 circles	No	15 d 28 d	No	Inst	P: 919-613-8400 E: clientservices@dm.duke.edu W: <a href="https://testcatalog.duke.edu">https://testcatalog.duke.edu</a>
	Lyso-GL3	WB: 4 ml EDTA (lavender) tube		28 d			
The Fabry Diagnostic Project (performed at EGL)	Enzyme Sequencing	WB: 5-10 ml heparin (green) tube WB: 5-10 ml EDTA (lavender) or ACD (yellow) tube	Blood, Saliva	7-10 d 4 wks	No	No charge*	P: 800-200-1524 or 404-778-8518 E: <a href="mailto:fabry.testing@emory.edu">fabry.testing@emory.edu</a> W: <a href="https://www.genetics.emory.edu/patient-care/fabryosomal-storage-disease-center">www.genetics.emory.edu/patient-care/fabryosomal-storage-disease-center</a>
	De/Dup	WB: 5-10 ml EDTA (lavender) or ACD (yellow) tube					
Greenwood Genetic Center	Enzyme Sequencing	WB: 5-10 ml heparin (green) tube; DBS card: 3 circles WB: 5-6 ml EDTA (lavender) tube; DBS card: 3 circles; Saliva	Blood, DBS, Saliva	2 wks 2 wks 26 d	No	Inst, Self-pay, Ins (SC residents only)	P: 800-473-9411 E: <a href="mailto:labgc@b6c.org">labgc@b6c.org</a> W: <a href="https://www.b6c.org">www.b6c.org</a>
	De/Dup	WB: 5-7 ml EDTA (lavender) tube					
LabCorp/ Integrated Genetics	Enzyme	WB: 5-10 ml ACD (yellow) tube		7-10 d	Yes	Inst, Ins, Self-pay	<b>LabCorp Customers:</b> P: 800-345-4463 E: <a href="mailto:askLGI@labcop.com">askLGI@labcop.com</a> W: <a href="https://www.integratedgenetics.com">www.integratedgenetics.com</a>
	Sequencing	WB: 7 ml EDTA (lavender) tube or ACD (yellow) tube	Blood	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		3 d			P: 866-354-2910 E: <a href="mailto:genomics@perkinelmer.com">genomics@perkinelmer.com</a> W: <a href="https://www.lanternprojectdx.com">www.lanternprojectdx.com</a>
	Sequencing (including De/Dup)	WB: 2-10 ml EDTA (lavender) tube (volume varies with age); DBS card: 3 circles, Saliva: (Oragene) Lyso-GL3 DBS: 2 circles	Blood, DBS, Saliva	3 wks	Yes	No charge*	
Mayo Clinic Laboratories	Enzyme	WB: 6 ml ACD (yellow) tube; DBS card: 2 spots; Serum: 2 ml (red top tube)		8-15 d			P: 800-533-1710 E: <a href="mailto:mci@mayo.edu">mci@mayo.edu</a> W: <a href="https://www.mayocliniclabs.com">www.mayocliniclabs.com</a>
	Sequencing	WB: 3 ml EDTA (lavender) or ACD (yellow) tube; DBS card: 2-5 spots	DBS (in some cases), Saliva	14-20 d	Yes	Inst (ins can be billed in some cases, inst acct required)	
Mount Sinai International Center for Fabry Disease	Enzyme	WB: 1 ml EDTA (lavender); Serum: 1 ml (red top tube); DBS: 2 circles		8-15 d			
	Sequencing	WB: 20 ml sodium heparin (green) tube	Blood, Buccal	7-10 d 10-14 d	No	No charge*	P: 866-322-7963 E: <a href="mailto:fabry.disease@mssm.edu">fabry.disease@mssm.edu</a> W: <a href="https://icahn.mssm.edu/research/fabry">https://icahn.mssm.edu/research/fabry</a>
Sema4	Enzyme	WB: 20 ml sodium heparin (green) tube					
	Sequencing	WB: 2 x 5-10 ml sodium heparin (green)	Blood, Saliva	7-10 d 2-3 wks	Yes	Inst, Ins, Self-pay	P: 800-298-6470 E: <a href="mailto:clientservices@sema4.com">clientservices@sema4.com</a> W: <a href="https://www.sema4.com">www.sema4.com</a>

\*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; WB = whole blood; wks = weeks.