

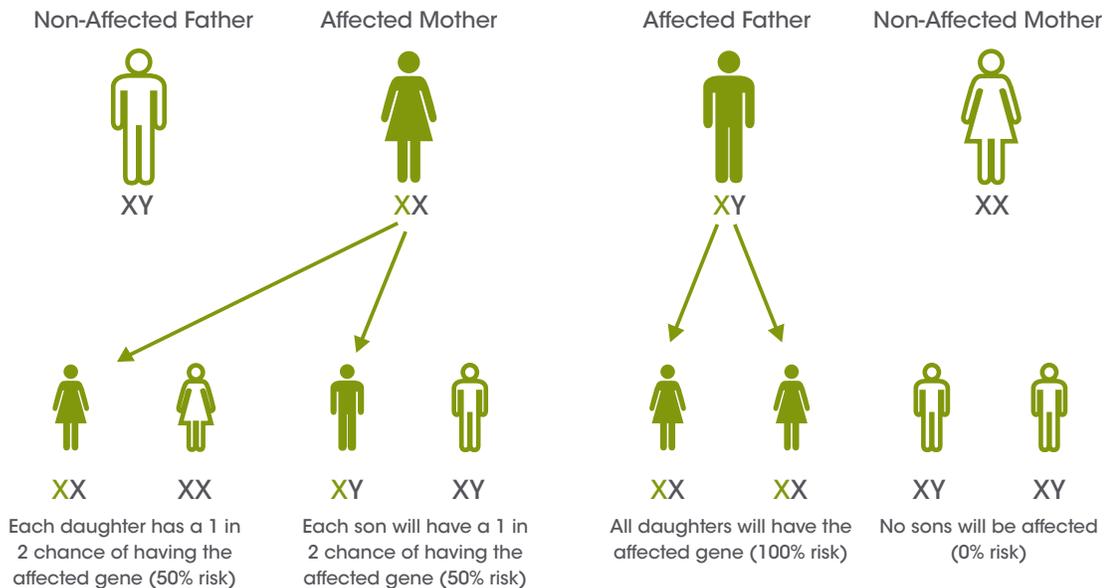
FABRY DISEASE

DIAGNOSTIC TESTING

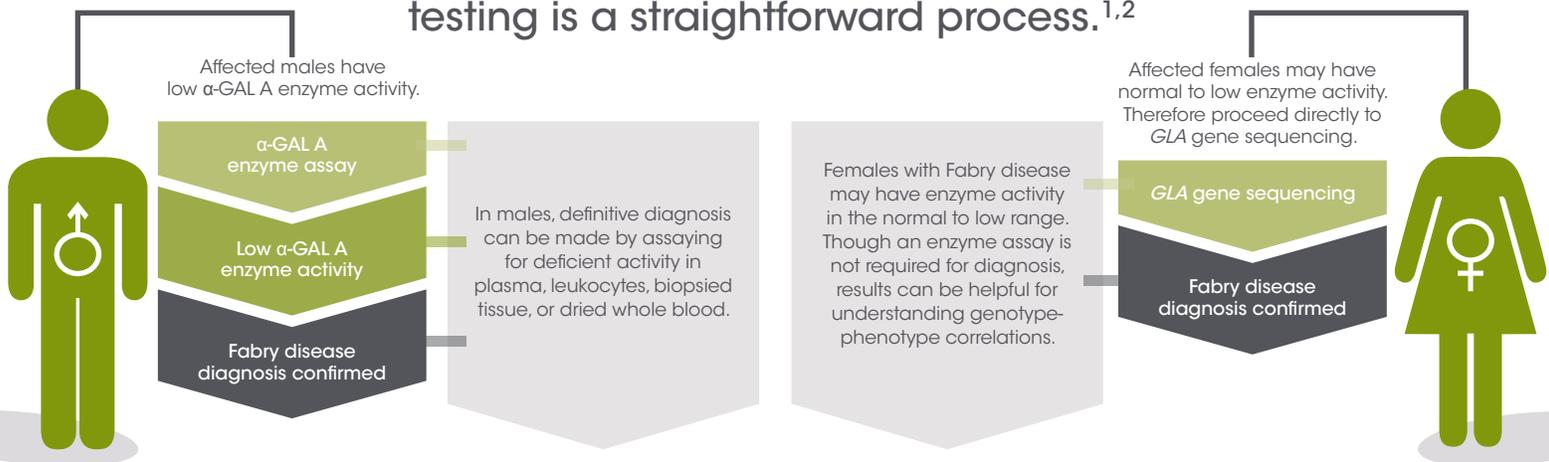
Fabry disease is a progressive and often life-threatening X-linked genetic disorder that affects men, women, and children. Fabry disease is caused by complete or partial deficiency of the lysosomal enzyme α -galactosidase A (α -GAL A), which leads to the accumulation of globotriaosylceramide (GL-3) in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

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 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.

Testing Options for Fabry Disease

| 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.