







Testing for Pompe disease

There are several programs and laboratories that offer lysosomal acid alpha-glucosidase (GAA) enzyme activity testing and/or *GAA* gene sequencing for Pompe disease (also known as acid maltase deficiency).

PROGRAMS OFFERING FREE TESTING

Program Name	Testing Available (At No Cost)	Kits Available	Information/Contact
MDA Pompe Testing Program	<ul style="list-style-type: none"> <i>GAA</i> gene sequencing (1 of 35 genes included in the sequencing panel for a range of neuromuscular disorders) 		Contact your local MDA office for more information

LABORATORIES OFFERING FEE-BASED TESTING

Laboratory Name	Testing Available (Fee-Based)	Kits Available	Information/Contact
Duke Biochemical Genetics Laboratory	<ul style="list-style-type: none"> <i>GAA</i> enzyme testing <i>GAA</i> gene sequencing 		URL: https://pediatrics.duke.edu/divisions/medical-genetics/biochemical-genetics-laboratory Phone: 919-684-0338 or 919-684-2722
Emory Genetics Lab-Eurofins	<ul style="list-style-type: none"> <i>GAA</i> enzyme testing <i>GAA</i> gene sequencing 	 Account setup is required	URL: https://www.egl-eurofins.com/tests/DW For account setup inquires contact client services: 855-831-7447 or https://www.egl-eurofins.com/client-portal/create-account/
GeneDx	<ul style="list-style-type: none"> <i>GAA</i> gene sequencing 	 (max 10 per order)	URL: https://www.genedx.com/wp-content/uploads/crm_docs/Neuro_requisition.pdf For kit order: http://www.genedx.com/order-a-test/test-kit-ordering/ Phone: 301-519-2100 or genedx@genedx.com
Icahn School of Medicine at Mount Sinai (sema4)	<ul style="list-style-type: none"> <i>GAA</i> enzyme testing <i>GAA</i> gene sequencing 	 Account setup is required	URL: http://icahn.mssm.edu/research/genomics/genetic-testing General Questions: 800-298-6470 For account setup inquiries contact client services: 212-241-3104 or 212-241-4499
Integrated Genetics	<ul style="list-style-type: none"> <i>GAA</i> enzyme testing <i>GAA</i> gene sequencing 	 Account setup is required	URL: www.integratedgenetics.com For account setup and inquiries, contact Client Services: 800-848-4436 Specimen services/pickup: 877-246-1226
Invitae	<ul style="list-style-type: none"> <i>GAA</i> gene sequencing 	 Account setup is required	URL: https://www.invitae.com/en/physician/genes/20772/?-cat=CAT000009 For Requisition Forms: https://www.invitae.com/en/order-forms/ Phone: 800-436-3037 or clientservices@invitae.com
Mayo Medical Laboratories	<ul style="list-style-type: none"> <i>GAA</i> enzyme testing <i>GAA</i> gene sequencing 	 Account setup is required	URL: www.mayomedicallaboratories.com Phone: 855-516-8404
Seattle Children's Hospital (Biochemical Genetics Department)	<ul style="list-style-type: none"> <i>GAA</i> enzyme testing <i>GAA</i> gene sequencing 		Test catalog listing: http://seattlechildrenslab.testcatalog.org/show/A-Glu-Pom-1 Phone: 206-987-2216

Contact individual testing programs and/or laboratories for information on obtaining test requisitions, sample requirements, and turnaround time.

www.pompe.com

This is not an all-inclusive list. For additional testing centers that perform diagnostic testing, please visit www.genetests.org. This list of testing programs and/or laboratories is provided for informational use only. It should not be considered a recommendation or endorsement of any company, test or service. 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, appropriate coding and obtaining reimbursement is the responsibility of the provider submitting a claim for the item or service.