

US LABS
DIAGNOSIS POMPE DISEASE

Pompe disease often presents with a cluster of non-specific signs and symptoms which makes Pompe disease difficult to distinguish from other muscular dystrophies. Different diseases that all looked the same to physicians were lumped together into one category that they called Limb Girdle Muscular Dystrophy (LGMD). Pompe disease is currently one of 25 different genetically defined forms of LGMD.

Jain Foundation:

<http://www.jain-foundation.org/>

The Jain Foundation has partnered with several other LGMD family foundations to offer free genetic testing (through a saliva sample) for individuals suffering from muscular dystrophy to help them obtain a definitive and specific genetic diagnosis. Currently the free genetic sequencing is only available to individuals living in the US through the following link (found on the Jain Foundation website) <http://www.lgmd-diagnosis.org/>

Consortium of LGMD Disease Foundations www.lgmd-diagnosis.org website:

- **Physician Tools:**
 - The Automated LGMD Diagnostic Assistant (ALDA), is a free online tool to help guide physicians toward the most probable diagnosis. This tool predicts the most likely type(s) of LGMD a patient may have based on clinical presentation and laboratory findings
- **Patient Tools:**
 - Take the quiz on the website to see if you qualify for genetic sequencing sponsorship.

US LABS THAT TEST FOR POMPE DISEASE:

DUKE UNIVERSITY:

- **Biochemical Genetics Lab (Glycogen Storage Disease laboratory):**
<http://pediatrics.duke.edu/divisions/medical-genetics/biochemical-genetics-laboratory/glycogen-storage-disease-laboratory/tes-2>
 - **Enzyme assays:** for acid alpha-glucosidase (GAA) in blood, cultured skin fibroblasts, muscle, and cultured amniocytes and chorionic villi
 - Download the test request form from this link:
<http://pediatrics.duke.edu/divisions/medical-genetics/biochemical-genetics-laboratory/glycogen-storage-disease-laboratory/test->
 - **Urine Hex4 Assay:** The urine Hex4 assay is a biomarker assay for glycogen storage in skeletal and cardiac muscle. It can be used to aid in the diagnosis of Pompe disease and to monitor patients on enzyme replacement therapy.
 - **Urine Hex4 Assay test request form:** The Urine Hex4 Assay test request form can be found at this link:
<http://pediatrics.duke.edu/divisions/medical-genetics/biochemical-genetics-laboratory/glycogen-storage-disease-laboratory/te-10>

US LABS
DIAGNOSIS POMPE DISEASE

- **Duke Molecular Diagnostics laboratory:**
 - **GAA gene sequencing:** Full GAA gene sequencing is performed by the Duke Molecular Diagnostics laboratory, Phone: (919) 684-2698,
 - **Download the test request form from this link:**
<http://clinlabs.duke.edu/DukeMolecular/default.aspx>

GENETIC TESTING REGISTRY:

GAA Gene Sequencing: The Genetic Testing Registry lists other labs that can run the GAA Gene Sequencing:

http://www.ncbi.nlm.nih.gov/gtr/tests/?term=pompe&condition=C0017921&test_purpose=Diagnosis&methods=2:7&locations=840:108,118,133,139,142

Emory Genetics Laboratory (EGL):

Emory University School of Medicine
Department: Department of Human Genetics
2165 North Decatur Road
Decatur, Georgia, United States 30033
Phone: 855-831-7447
Fax: 404-778-8559
Email: eglgc@emory.edu
Website: <http://genetics.emory.edu/egl/>

Greenwood Genetic Center Diagnostic Laboratories, GGC:

Greenwood Genetic Center (GGC)
106 Gregor Mendel Circle
Greenwood, South Carolina, United States 29646
Phone: 800-473-9411
Fax: 864-941-8141
Email: labgc@ggc.org
Website: <http://www.ggc.org/diagnostic.html>

Molecular Genetics Laboratory:

Cincinnati Children's Hospital Medical Center (CCHMC)
3333 Burnet Ave ML 4006
Cincinnati, Ohio, United States 45229-3039
Phone: 513-636-4474
Fax: 513-636-4373
Email: moleculargenetics@cchmc.org
Website: <http://www.cincinnatichildrens.org/molecular-genetics/>

Baylor Miraca Genetics Laboratories (BMGL):

Baylor College of Medicine (BCM)
2450 Holcombe, Grand Blvd. - Sample Receiving Dock

US LABS
DIAGNOSIS POMPE DISEASE

Houston, Texas, United States 77021-2024
Website: <http://www.bmgl.com/>

GeneDx:

207 Perry Parkway
Gaithersburg, Maryland, United States 20877
Phone: 301-519-2100
Email: lisa@genedx.com
Website: <http://www.genedx.com/>

Please contact Marsha Zimmerman (AMDA) if you have questions or need assistance in getting a Pompe disease diagnostic test ordered. Email: Marzim50@gmail.com