

Labs Offering Diagnostic Testing for Pompe Disease

also known as Acid Maltase Deficiency and Glycogen Storage Disease Type II (GSD-II)

Diagnostic Lab List

Updated March 2011

GAA Enzyme Activity Assay – Blood Tests

Laboratory	Ship-To Address	Contact Person
The Buffalo General Hospital www.rgbmgl.org	Robert Guthrie Biochemical & Molecular Genetics Laboratory Room A-762, 100 High Street Buffalo, NY 14203	Georgirene D. Vladutiu PhD, Director 716-859-7741 Email: gdv@buffalo.edu
Duke University Medical Center http://medgenetics.pediatrics.duke.edu	Duke University Hospital Glycogen Storage Disease Laboratory Biochemical Genetics Laboratory 801 Capitola Drive, Suite 6 Durham, NC 27713	Deeksha Bali, PhD, FACMG 919-684-0025 Email: bali0001@mc.duke.edu or Gwen Harmon Dickerson 919-684-0338 Email: gharmon@duke.edu
Emory University, Department of Human Genetics www.geneticslab.emory.edu	Emory Genetics Laboratory 2165 N. Decatur Road Decatur, GA 30033	Ymkje Cuperus, MS, GC 404-778-8550 Email: icuperu@emory.edu
Genzyme Genetics www.genzymegenetics.com	Genzyme Genetics 2000 Vivigen Way Santa Fe, NM 87505	Client Services 800-848-4436
Seattle Children's Hospital www.seattlechildrens.org/geneticslab	Seattle Children's Hospital Laboratory A6901 Attn: Rhona Jack, PhD 4800 Sand Point Way NE Seattle, WA 98105	Rhona Jack, PhD 206-987-2216 Email: rhona.jack@seattlechildrens.org

GAA Enzyme Activity Assay – Skin Biopsy

Laboratory	Ship-To Address	Contact Person
Duke University Medical Center http://medgenetics.pediatrics.duke.edu	Duke University Hospital Glycogen Storage Disease Laboratory Biochemical Genetics Laboratory 801 Capitola Drive, Suite 6 Durham, NC 27713	Deeksha Bali, PhD, FACMG 919-684-0025 Email: bali0001@mc.duke.edu or Denise Peterson 919-684-2722 Email: pet01@duke.edu
Mayo Clinic www.mayomedicallaboratories.com/test-info/biochemical/index.html	Mayo Medical Laboratories 3050 Superior Drive NW Rochester, MN 55901-1995	Biochemical Genetics Laboratory Genetic Counselor on Call 800-533-1710 Email: biochemicalgenetics@mayo.edu
Seattle Children's Hospital www.seattlechildrens.org/geneticslab	Seattle Children's Hospital Laboratory A6901 Attn: Rhona Jack, PhD 4800 Sand Point Way NE Seattle, WA 98105	Rhona Jack, PhD 206-987-2216 Email: rhona.jack@seattlechildrens.org
University of Alabama at Birmingham	Metabolic Disease Laboratory University of Alabama at Birmingham 648 Kaul Building 720 20th St. South Birmingham, AL 35233	Paula Huffman, BS 205-934-6370 Email: phuffman@uab.edu

For additional information about laboratories that offer diagnostic testing for Pompe Disease, visit www.genetests.org

This listing includes only laboratories that have agreed to be listed by Genzyme and is not intended to be exhaustive. It is for informational purposes only, and no endorsement of or representations regarding the services offered are either intended or implied. Please note that, while Genzyme has endeavored to obtain information that is current as of the time of publication, it makes no representations as to accuracy, and physicians are directed to the individual laboratories for the specific details of the services provided.

Contact the laboratories listed above for information on obtaining test requisitions, sample requirements, and turnaround time.

GAA Enzyme Activity Assay – Muscle Biopsy

Laboratory	Ship-To Address	Contact Person
Athena Diagnostics www.athenadiagnostics.com	Athena Diagnostics, Inc. 377 Plantation Street Worcester, MA 01605	Sat Dev Batish, PhD, FACMG Phone 508-756-2886 ext 3222 Fax 508-753-5601 Email: dev.batish@athenadiagnostics.com
The Buffalo General Hospital www.rgbmgf.org	Robert Guthrie Biochemical Genetics Laboratory Room A-762, 100 High Street Buffalo, NY 14203	Georgirene D. Vladutiu PhD, Director 716-859-7741 Email: gdv@buffalo.edu
Duke University Medical Center http://medgenetics.pediatrics.duke.edu	Duke University Hospital Glycogen Storage Disease Laboratory Biochemical Genetics Laboratory 801 Capitola Drive, Suite 6 Durham, NC 27713	Deeksha Bali, PhD, FACMG 919-684-0025 Email: bali0001@mc.duke.edu or Denise Peterson 919-684-2722 Email: pet01@duke.edu

GAA Enzyme Activity Assay – Prenatal Screening

Laboratory	Ship-To Address	Contact Person
Duke University Medical Center http://medgenetics.pediatrics.duke.edu	Duke University Hospital Glycogen Storage Disease Laboratory Biochemical Genetics Laboratory 801 Capitola Drive, Suite 6 Durham, NC 27713	Deeksha Bali, PhD, FACMG 919-684-0025 Email: bali0001@mc.duke.edu or Denise Peterson 919-684-2722 Email: pet01@duke.edu
Emory University, Department of Human Genetics <i>Known familial mutations only</i> www.geneticslab.emory.edu	Emory Genetics Laboratory 2165 N. Decatur Road Decatur, GA 30033	Ymkje Cuperus, MS, GC 404-778-8550 Email: icuperu@emory.edu

DNA Analysis

Laboratory	Ship-To Address	Contact Person
Cincinnati Children's Hospital www.cincinnatichildrens.org	Molecular Genetics Laboratory 3333 Burnet Avenue R-1042 Cincinnati, OH 45229	Laurie Bailey, CGC 513-636-4474 Email: laurie.bailey@cchmc.org
Duke University Medical Center http://medgenetics.pediatrics.duke.edu	Duke University Hospital Glycogen Storage Disease Laboratory Biochemical Genetics Laboratory 801 Capitola Drive, Suite 6 Durham, NC 27713	Deeksha Bali, PhD, FACMG 919-684-0025 Email: bali0001@mc.duke.edu or Gwen Harmon Dickerson 919-684-0338 Email: gharmon@duke.edu or Catherine Rehder, PhD, FACMG 919-613-8434 email: catherine.rehder@duke.edu
Emory University, Department of Human Genetics www.geneticslab.emory.edu	Emory Genetics Laboratory 2165 N. Decatur Road Decatur, GA 30033	Ymkje Cuperus, MS, GC 404-778-8550 Email: icuperu@emory.edu
Genzyme Genetics www.genzyme genetics.com	Genzyme Genetics 2000 Vivigen Way Santa Fe, NM 87505	Client Services 800-848-4436
Prevention Genetics www.preventiongenetics.com	Prevention Genetics LLC Attn: Diagnostics Lab 3700 Downwind Drive Marshfield, WI 54449	James Weber, PhD 715-387-0484 Email: jim.weber@preventiongenetics.com
Seattle Children's Hospital www.seattlechildrens.org/geneticslab	Seattle Children's Hospital Laboratory A6901 Attn: Molecular Genetics Laboratory 4800 Sand Point Way NE Seattle, WA 98105	Monica Wellner (206) 987-5306 Email: monica.wellner@seattlechildrens.org

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