

CHERYL LEE GARGANTA

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Birth Date: August 16, 1961
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Education

- 1981, May. B.A. in Chemistry from Wheaton College, Norton, MA.
Thesis: Phase transfer catalysis and ultrasound- an effective combination for the preparation and addition of dichlorocarbene to a Δ^5 steroid.
Advisor: Bojan Jennings
- 1983, May. M.S. in Biochemistry from Medical College of Virginia/Virginia Commonwealth University, Richmond, VA.
Thesis: Characterization of the deficiency of a metalloendopeptidase from murine kidney
Advisor: Judith Bond, Ph.D
- 1991, May. M.D. from Medical College of Virginia/Virginia Commonwealth University, Richmond, VA.
- 1995, August. Ph.D. in Human Genetics from Medical College of Virginia/Virginia Commonwealth University, Richmond, VA.
Thesis: Biochemical studies of mammalian lipoamidase activity
Advisor: Barry Wolf, M.D., Ph.D.

Post-graduate training

- 1991, June- June 1994. Internship and residency in Pediatrics, Medical College of Virginia/Virginia Commonwealth University, Richmond, VA.
- 1998, July- June 2001. Residency in Clinical and Biochemical Genetics, Yale University, New Haven, CT.

Academic Appointments:

- 2001, July-June 2002. Instructor, Yale University, Department of Genetics, New Haven, CT
- 2001, July-Dec. 2004. Director of Biochemical Disease Detection Laboratory (BDDL), Yale University Department of Genetics, New Haven CT
- 2002, July-Dec. 2004. Assistant Professor, Yale University, Dept. of Genetics, New Haven, CT
- 2005, Jan-June 2014. Director, Metabolic Lab, Tufts Medical Center
- 2005, Jan-June 2014. Clinical Geneticist, Tufts Medical Center, Boston, MA. Assistant Professor, Pediatrics, Tufts University School of Medicine.
- 2011, Jan-June 2014. Assistant Professor, Department of Pathology and Laboratory Medicine, Tufts Medical Center, Boston, MA
- 2014, July-present. Associate Professor, Department of Pathology, Immunology and Laboratory Medicine, University of Florida, Gainesville FL

Hospital Appointments:

- 1993, December-May 1995. House Pediatrician, St. Mary's Hospital, Richmond, VA
- 1994, July-May 1995. House Pediatrician, Children's Hospital, Richmond, VA
- 1995, June-June 1998. Pediatrician, Western Mass Pediatrics, Holyoke, MA
- 2001, October-Dec 2004. Attending in Pediatrics, Yale-New Haven Hospital, New Haven, CT
- 2005, January-June 2014. Attending in Pediatrics, Tufts Medical Center, Boston, MA
- 2006, June-June 2015. Attending in Pathology, Tufts Medical Center, Boston, MA
- 2014, July-present. Pathology and Pediatrics, UF, Gainesville, FL

Teaching Responsibilities:

- 2010- 2014, 3 times/month. Scientific writing workshops and discussion of topics related to mass spectrometry, inborn errors of metabolism (IEMs), testing for IEMs and molecular genetics testing with Tufts Pathology residents.

- 2012, Fall. Basics of chromatography, Pathology Rounds, Tufts Medical Center.
- 2012, Summer. Research preceptor for 4 high school students, Tufts School of Medicine Teachers and High School Students.
- 2012, Spring. –Omics series of 3 lectures, Pathology Rounds, Tufts Medical Center.
- 2011, Spring. Mass Spectrometry Basics. Newborn screening. Pathology Rounds, Tufts Medical Center.
- 2010- June 2012, monthly. Discuss coagulation and hemoglobinopathy testing with Tufts Pathology residents.
- 2010, Spring. Chromatography in the clinical lab. Specialized metabolic testing. Pathology Rounds.
- 2009, Spring. Chromatography and mass spectrometry in the clinical lab. Specialized testing for metabolic disease. Pathology Rounds.
- 2008 Summer. Research preceptor for 2 high school students, Tufts School of Medicine Teachers and High School Students
- 2008, January-March. Preceptor, Tufts School of Medicine first year medical student, Metabolic Lab.
- 2007, Fall-Spring 2009. Clinical receptor, Boston University Genetic Counseling Students.
- 2007, Summer-August 2008. Preceptor, Medical Technologist from Saudi Arabia learning techniques used in Biochemical Genetics Lab
- 2007, July. “Metabolic Emergencies”. Lecture for pediatrics residents, Tufts Medical Center.
- 2007 Summer. Research preceptor for 2 high school students, Tufts School of Medicine Teachers and High School Students
- 2007, Spring. Clinical preceptor, First year medical student, Tufts School of Medicine
- 2006, September-January 2007. Preceptor, Boston University genetic counseling students
- 2006, Summer. Research preceptor for 2 high school students, Tufts School of Medicine Teachers and High School Students
- 2005, February and December. Teaching attending in pediatrics. Tufts-New England Medical Center, Boston, MA.
- 1999-2004 Lecturer (Non-Mendelian Genetics) and small group leader for Medical Genetics course for second year medical students at Yale School of Medicine.
- 2003, Summer. Advisor for minority undergraduate students doing research in my lab.
- 2003, Fall. Co-instructor for Human Molecular Genetics. Gave 7 lectures including non-Mendelian genetics, population genetics, epigenetic phenomena, and methods for studying gene function.
- 2002, Spring. Co-instructor for Human Molecular Genetics. Gave 7 lectures including non-Mendelian genetics and genetics of hearing loss, repeat expansion conditions, and skeletal dysplasias.
- 2001-2004. Multiple lectures given to Genetics residents and at off-site clinics to pediatricians, obstetricians, and obstetrics residents on a variety of genetic topics.

Medical Licensure and Boards

- Florida, unrestricted license ME120970. Expires 01/31/2016.
- Massachusetts, unrestricted license 80976, expires 8/2015.
- American Board of Medical Genetics (2002035), Clinical Geneticist and Clinical Biochemical Geneticist, September 2002, recertified 2012, expires December 2022. Participant in MOC.
- American Board of Pediatrics (053335), October 1994. Recertified January 2002, expired 2008.
- Diplomate of the National Board of Medical Examiners, July 1992.

Membership in Professional Organizations

- American Society of Human Genetics
Society for Inherited Metabolic Disorders
Society for the Study of Inherited Metabolic Disease
American Association of Clinical Chemists

Other Professional Activities

Observer, CLSI Newborn Screening by Tandem Mass Spectrometry; (I/LA32-A), July 2010. Wrote Chapter 17 (Additional Assays). Active participant in document editing.
Invited speaker, CPSA Metabolomics, February 2015, Gainesville FL. Metabolic Disease Testing: More than Organic and Amino Acids, Acylcarnitines and Inborn Errors.

PUBLICATIONS

Articles

- Jennings BJ, Boudreau SM, Braasch B, and **Garganta CL**. 5 α , 6 α - and 5 β , 6 β -dichloromethylene adducts of 3 β -acetoxy-androsten-17-one. *Steroids* 39(1982) 371-380.
- Jones LA, Kirby SL, **Garganta CL**, Gerig TM, and Mulik JD. Optimization of separation number in gas chromatography with fused silica capillary columns under temperature programmed conditions. *Analytical Chemistry* 55(1983) 1354-1360.
- Garganta CL**, Hazelett J, and Shaw JM. Synthesis of a photoactivable phospholipid containing an aromatic sulfonylazide and its interaction with proteins. *Biochimica et Biophysica Acta* 812(1985) 261-267.
- McKay MJ, **Garganta CL**, Beynon RJ, and Bond JS. Deficiency of a mouse kidney metalloendopeptidase activity: immunological demonstration of an altered gene product. *Biochemical and Biophysical Research Communications* 132(1985) 171-177.
- Garganta CL** and Bond JS. Assay and kinetics of arginase. *Analytical Biochemistry* 154(1986) 388-394.
- Garganta CL** and Wolf B. Lipoamidase activity in human serum is due to biotinidase. *Clinica Chimica Acta* 189(1990) 313-326.
- Garganta CL** and Bodurtha JN. Report of another family with Simpson-Golabi-Behmel Syndrome and a review of the literature. *American Journal of Medical Genetics* 44(1990) 129-135.
- Garganta CL** and Wolf B. A colorimetric assay of lipoyl-lysine hydrolase activity using 2,6-dibromoquinone-4-chlorimide. *Analytical Biochemistry*, 240(1996)177-184.
- Garganta C** and Seashore MR. Universal screening for congenital hearing loss. *Pediatric Annals*. 29(2000)302-308.
- Mudd SH, Braverman N, Pomper M, Tezcan K, Kronick J, Jayakar P, **Garganta C**, Ampola MG, Levy HL, McCandless SE, Wiltse H, Stabler SP, Allen RH, Wagner C, Borschel MW. Infantile hypermethioninemia and hyperhomocysteinemia due to high methionine intake: a diagnostic trap. *Molecular Genetics and Metabolism* 79 (2003)6-16.
- Murakami T, Fukai K, Oiso N, Hosomi N, Kato A, **Garganta C**, Barnicoat A, Poppelaars F, Aquaron R, Paller AS, Ishii M. New KIT mutations in patients with piebaldism. *Journal of Dermatological Science* 35 (2004)29-33.
- Bolteus AJ, **Garganta C**, Bordey A. Assays for measuring extracellular GABA levels and cell migration rate in acute slices. *Brain Research Protocols* 14(2005)126-134.
- Garganta CL** and Smith WE. Metabolic Evaluation of the Sick Neonate. *Seminars in Perinatology* 29(2005)164-172.
- Taylor P, Raman T, **Garganta CL**, Njalsson R, Carlsson K, Ristoff E, and Carey HB. Recurrent High Anion Gap Metabolic Acidosis Secondary to 5-Oxoproline (Pyroglutamic Acid). *American Journal of Kidney Diseases* 46(2005)E4-10.
- Grindle S, **Garganta C**, Sheehan S, Gile J, Lapierre A, Whitmore H, Paigen B, DiPetrillo K. Validation of high-throughput methods for measuring blood urea nitrogen and urinary albumin concentrations in mice. *Comp Med*. 2006 Dec;56(6):482-6.
- Arnold GL, Koeberl DD, Matern D, Barshop B, Braverman N, Burton B, Cederbaum S, Fiegenbaum A, **Garganta C**, Gibson J, Goodman SI, Harding C, Kahler S, Kronn D, Longo N. A Delphi-based consensus clinical practice protocol for the diagnosis and management of 3-methylcrotonyl CoA carboxylase deficiency. *Molecular Genetics & Metabolism* 93(2008)363-370.

- Dericioglu N, **Garganta CL**, Petroff OA, Mendelsohn D, Williamson A. Blockade of GABA synthesis only affects neural excitability under activated conditions in rat hippocampal slices. *Neurochem Int*. 53(2008):22-32. doi: 10.1016/j.neuint.2008.04.006
- Arnold GL, Van Hove J, Freedenberg D, Strauss A, Longo N, Burton B, **Garganta C**, Ficicioglu C, Cederbaum S, Harding C, Boles RG, Matern D, Chakraborty P, Feigenbaum A. A Delphi clinical practice protocol for the management of very long chain acyl-CoA dehydrogenase deficiency. *Mol Genet Metab* 96(2009)85-90. doi: 10.1016/j.ymgme.2008.09.008
- Poduri A, Chitsazzadeh V, D'Arrigo S, Fedrizzi E, Pantaleoni C, Riva D, Busse C, Küster H, Duplessis A, Gaitanis J, Sahin M, **Garganta C**, Topcu M, Dies KA, Barry BJ, Partlow J, Barkovich AJ, Walsh CA, Chang BS. The syndrome of perisylvian polymicrogyria with congenital arthrogryposis. *Brain Dev*. 2010 Aug;32(7):550-5. Epub 2009 Sep 13.
- Shen Y, Dies KA, Holm IA, Bridgemohan C, Sobeih MM, Caronna EB, Miller KJ, Frazier JA, Silverstein I, Picker J, Weissman L, Raffalli P, Jeste S, Demmer LA, Peters HK, Brewster SJ, Kowalczyk SJ, Rosen-Sheidley B, McGowan C, Duda AW 3rd, Lincoln SA, Lowe KR, Schonwald A, Robbins M, Hisama F, Wolff R, Becker R, Nasir R, Urion DK, Milunsky JM, Rappaport L, Gusella JF, Walsh CA, Wu BL, Miller DT; Autism Consortium Clinical Genetics/DNA Diagnostics Collaboration. Clinical genetic testing for patients with autism spectrum disorders. *Pediatrics*. 2010 Apr;125(4):e727-35. Epub 2010 Mar 15. doi: 10.1542/peds.2009-1684
- Kronn D, Mofidi S, Braverman N, Harris K, Diagnostics Guidelines Work Group. Diagnostic guidelines for newborns who screen positive in newborn screening. *Genet Med* 2010 Dec;12(12 Suppl):S251-5. doi: 10.1097/GIM.0b013e3181fe5d8b
- Pittas AG, Nelson J, Mitri J, Hillmann W, **Garganta C**, Nathan DM, Hu FB, Dawson-Hughes B, Dpp Research Group. Plasma 25-Hydroxyvitamin D and Progression to Diabetes in Patients at Risk for Diabetes. *Diabetes Care* 2012 35:565-573. doi: 10.2337/dc11-1795
- Mitri J, Nelson J, Ruthazer R, **Garganta C**, Nathan DM, Hu FB, Dawson-Hughes B, Pittas AG, Diabetes Prevention Program Research Group. Plasma 25-hydroxyvitamin D and risk of metabolic syndrome: an ancillary analysis in the Diabetes Prevention Program. *Eur J Clin Nutr* 2014 Mar;68(3):376-83. doi: 10.1038/ejcn.2013.293
- Sahai I, **Garganta CL**, Bailey J, James P, Levy HL, Martin M, Neilan E, Phornphutkul C, Sweetser DA, Zytovicz TH, Eaton RB. Newborn screening for Glutaric Aciduria-II: The New England experience. *JIMD Rep* 2014;13:1-14. doi:10.1007/8904_2013_262
- Alroy J, **Garganta C**, Wiederschain G. Secondary biochemical and morphological consequences in lysosomal storage diseases. *Biochemistry (Mosc)*. 2014 Jul;79(7):619-36. doi: 10.1134/S0006297914070049
- Zhang FF, Driban JB, Lo GH, Price LL, Booth S, Eaton CB, Lu B, Nevitt M, Jackson B, **Garganta C**, Hochberg MC, Kwok K, McAlindon TE. Vitamin D deficiency is associated with progression of knee osteoarthritis. *J Nutr*. 2014 Dec;144(12):2002-8. doi: 10.3945/jn.114.193227. Epub 2014 Oct 1

Chapters

- Bond JS, Unger DF, and **Garganta CL**. "Properties and regulation of mouse liver arginase" in Manganese in Metabolism and Enzyme Function. Ed. Schramm VL and Wedler FC. 1986, 236-257. ISBN 978-0-12-629050-9.
- Garganta CL**. "Inborn Errors of Metabolism" in Contemporary Practice in Clinical Chemistry. Ed. William Clarke and D. Robert Dufour. 2006 and 2010 editions.

Abstracts

- Bond JS, **Garganta CL**, Bellingham DL, Beynon RJ. Deficiency of a kidney metalloendoproteinase in mice. Fed Proc 42(1983)1915.
- Garganta CL** and Carpenter TO. Cellular localization of PHEX using green fluorescent protein. Journal of Bone and Mineral Research 15(2000)S211.
- Garganta CL**, Seashore MR, Rosengren SS, Tezcan K. Branched chain ketoaciduria in respiratory chain disorders (RCDs). Presented at SIMD meeting, March 2001.
- Gillis LA, Rand EB, Russo P, Hirsch B, **Garganta CL**, Tanguay RM, Berry GT. Hepatocellular carcinoma with vascular extension at presentation in a child with atypical tyrosinemia. Presented by LA Gillis at Society of Inherited Metabolic Diseases (SIMD) meeting, March 2002.
- Williamson A, **Garganta CL**, Petroff OA. Disruptions in glutamate-glutamine cycling alter GABA function. Epilepsia 45(2004) supp 7:360.
- Garganta CL** and Czyzyk DJ. Separation and quantification of branched chain amino acids by LC/MS/MS. American Journal of Human Genetics 75(2004)supp.
- Petroff OA, Cavus I, **Garganta CL**, Vives KP, Spencer DD. Glutamine synthesis is impaired in regions with above normal extracellular glutamate levels. American Epilepsy Society meeting, 2007.
- Pittas AG, Nelson J, Mitri J, Hillman W, **Garganta C**, Nathan DM, Hu FG, Dawson-Hughes B and the Diabetes Prevention Program Research Group. Vitamin D status and progression to diabetes in patients at risk for diabetes: an ancillary analysis in the Diabetes Prevention Program randomized controlled trial. *Diabetes* 2011:(Suppl 1):117-OR.
- O'Brien JJ, Cassalman CL, Johnson S, Hunter R, Pilichowska M, **Garganta C**. Further Evaluation of False Positive Hexagonal Phase Phospholipid Assays Due to Elevated C-Reactive Protein. Presented by CL Cassalman at ASCP meeting 11-2012.
- Abbott MA, Berry G, **Garganta C**, Newton S, Spence S, Sullivan J. Phenotypical variability in brothers with HSD10 disease. Poster presented by MA Abbott at American College of Medical Genetics meeting, March 2013.