

CURRICULUM VITAE
August 2023

Name: Hans Christoph Andersson, MD, FACMG

Birth: December 23, 1956; New Orleans, LA

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Education:

1978 **Bachelor of Science**; majors: Music and Psychology
Tulane University, New Orleans, Louisiana
1978 - 1980 Post-baccalaureate pre-medical curriculum
Tulane University; Georgetown University, Washington, D.C.
1984 **Doctor of Medicine**
Tulane University School of Medicine, New Orleans, LA
1989 Inborn Errors of Metabolism, Foundation for Advancement of Educ. in Sciences
National Institutes of Health, Bethesda, MD

Postdoctoral Training:

1984 - 1987 **Pediatric Residency**
Tulane University Medical School, Department of Pediatrics
1987 - 1988 **Intramural Research Training Award Fellowship**
Section on Human Biochemical Genetics, Human Genetics Branch
National Institute of Child Health & Human Development, NIH
Lab Chief: William A. Gahl, MD, PhD
1988 - 1990 **Medical Staff Fellow**, Clinical Genetics
Interinstitute Genetics Program, NIH
Director: John J. Mulvihill, M.D.
1990 - 1991 **Medical Staff Fellow**, Clinical and Research Fellow
Section on Human Biochemical Genetics, Human Genetics Branch
NICHD/NIH Lab Chief: William A. Gahl, MD, PhD
1992 - 1993 **Research Fellowship**
Alexander von Humboldt Foundation, German Research Council
Center for Biochemistry and Molecular Cell Biology
University of Göttingen, Germany
Lab Chief: Kurt von Figura, MD

Employment:

1994 - 2000 **Assistant Professor of Pediatrics**, Clinical/Biochemical Geneticist
2000 - 2006 **Associate Professor of Pediatrics** (with tenure)
Hayward Genetics Center, Tulane University School of Medicine
1998 - 2000 **Interim Laboratory Director**, Biochemical and Molecular Genetics Labs
Hayward Genetics Center, Tulane University School of Medicine
2000 - present **Associate Laboratory Director**, Biochemical and Molecular Genetics Labs
Hayward Genetics Center, Tulane University School of Medicine

Employment (cont.):

2002 - 2010 **Director**, Hayward Genetics Training Program, ABMGG-accredited in
 Laboratory Genetics & Genomics
 Hayward Genetics Center, Tulane University School of Medicine

2012 - present
 2006 - present **Director**, Hayward Genetics Center
 Karen Gore Chair in Human Genetics

2006 - present **Professor of Pediatrics** (with tenure)

2006 - present **Director**, Hayward Genetics Center Laboratories
Lab Director, Biochemical Genetics Laboratory

Honors, Awards, Invited Professorships:

1992 - 1993 **Research Fellowship** Alexander von Humboldt Foundation,
 one-year support plus six-month extension

1993 **Guest Researcher Fellowship**,
 German Research Council (Deutsche Forschungsgemeinschaft); five months support

1998 - present **Best Doctors in America: Medical Genetics, Pediatric Medical Genetics,
 Pediatric Metabolic Specialist**

2012 - present **Top Doctor: Clinical Genetics**
 Castle-Connolly Medical Ltd, US News and World Report

2000 **1st Annual Award for Humanism in Medicine**
 Healthcare Foundation of New Jersey

2001 **Invited Professorship**, Kumamoto University School of Medicine
 Kumamoto Japan, Center for Animal Resources and Development (3 months)

2007 **9th Jack Spevak Visiting Professorship**, Blank Children's Hospital, Iowa

2017 **Distinguished Service Award**, Louisiana Academy of Nutrition and Dietetics

2021 **Louisiana Rare Disease Advisory Council**, member, appointed by Governor

Grants:*Previous*

1997 **NIH Travel Grant**: International Inborn Errors of Metabolism meeting
 Vienna, Austria, May 1997. \$1000

1997 - 2000 **Louisiana Board of Regents Grant (LEQSF)**, Principal Investigator
 Research Competitiveness Program, 3 year direct support: \$99,988
 Cloning and Characterization of Lysosomal System h Protein

1999 - 2000 **Teaching Innovations in Medical Education (TIME)**, Principal Investigator
 Tulane School of Medicine, Office of Educational Research and Services
 Development of Dysmorphology Teaching Tools, \$3147

1999 - 2001 **Project MECCA** (Medical Education Community of Collaborative Activities)
 Development of a Web-Based System for Curricular Integration
 Co-Principal Investigator

1999 - 2001 **CDC Cooperative Agreement Award**, Co-Principal Investigator
 Medical Genetics Test Results Reporting, 2 year support, \$95,116/y

2000 - 2005 **Louisiana Board of Regents**, Principal Investigator
 Linkage Analysis of Cobalamin C Disease Gene, \$181,084

2002 - 2003 **Transkaryotic Therapies**, Principal Investigator, \$60,000
 Education of Primary Care Physicians In Mucopolysaccharidoses

2004 - 2007 **HRSA: Enhanced Genetic Services & Newborn Screening Collaborative
 in Region 3**, Telecommunications pilot project director, \$50,000 (0.1 FTE)

2006 - 2007 **HRSA: Enhanced Genetic Services & Newborn Screening Collaborative
 in Region 3**, Co-Principal Investigator, \$125,000 (0.15 FTE)

2007 - 2012 **HRSA: Region 3 Collaborative Regional Genetic and Newborn Screening
 Services**, Co-Principal Investigator, \$996,501 (0.25 FTE)

Grants (cont):

2010 - 2012	Biomarin Investigator Sponsored Trial: Evaluation of Behavior, Executive Function, Neurotransmitter Function and Genomic Expression in PKU "Nonresponders" to Kuvan®, Principal Investigator
2012 - 2020	HRSA: Region 3 Collaborative Regional Genetic and Newborn Screening Services, Co-Principal Investigator (0.2 FTE)
<i>Currently Active</i> 2020-2024	HRSA: Region 3 Collaborative Regional Genetic and Newborn Screening Services, Co-Principal Investigator (0.2 FTE)
2019-2024	NIH/NINDS: Frontiers in Congenital Disorders of Glycosylation (1U54NS115198-01), CO-PI (0.05%)

Teaching Awards:

Tulane University School of Medicine Owl Club Awards (Education Awards)	
1998 - 99	Outstanding Second Year Teacher
1998 - 99	Best Second Year Course: Medical Genetics (Course Director)
2000	Virginia S. Furrow Award for Innovations in Teaching Tulane University School of Medicine
2000	Mortar Board Award for Outstanding Teaching Newcomb College, Tulane University
2000	Honorary Member for Outstanding Service Tulane University Honors Program
2001	Clinical Integration Teaching Award,
2012	Best First Year Professor, Honorable Mention
2015	Best First-Year Professor

Educational Activities:

1994 - present	Tulane University Medical School: Medical Genetics course core: Course Director/lecturer (8 hrs annually) Biochemistry: annual Clinical Correlations (2 hrs annually) Pediatrics: genetics block lecture (6 hrs annually) Medical Residency/Grand Rounds: Pediatric, Pathology, Psychiatry, Internal Medicine, Preventive Medicine residents
1994 - 1999	Morning Report, Department of Pediatrics
1995 - 2010, 2016	Course Director, Medical Genetics , 35-hr 2nd year medical school course * Owl Club Awards: 1998-99, 2000-01
1999 - 2009	Guest Lecturer: Biochemical Genetics, Medical Genetics core curriculum University of South Alabama Medical School
2002 - 2012	ACGME-accredited Clinical Genetics Fellowship Program, Director, Human Genetics Program
2006 - 2023	ABMGG-accredited Fellowship Program in Laboratory Genetics & Genomics Program Director (4 fellows since 2010, all board certified)
2003 - present	<u>Course Director</u> , Intro to Human Genetics , Hayward Graduate Program
2006 - present	<u>Course Director</u> , Biochemical Genetics , Hayward Graduate Program
1996 - present	Dissertation committee member for PhD in Human Genetics Amanda Helip-Wooley, PhD (2002) Margaret Park, PhD (2005) Chunbo Shao, PhD (2007) Congxing Lin (2008) SanSan Ng (2008) Matt Comeaux (2009) Graeme Preston (2020)

Licensure, Board Certifications:

1984 - present	Louisiana Board of Medical Examiners , medical license (017892)
1989 - 1996	American Board of Pediatrics , board certification
1990 - present	American Board of Medical Genetics , board certification * Clinical Genetics * Clinical Biochemical/Molecular Genetics

Reviewership:

1998 - present	Ad Hoc Reviewer: <i>American Journal of Medical Genetics, Neonatology Cellular & Molecular Biology, Journal of Pediatrics, Nutrition, Metabolism & Cardiovascular Diseases, Genetics In Medicine Journal of Inherited Metabolic Diseases, Eur Journal Human Genetics, Biochemie, Molecular Genetics and Metabolism</i>
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Editorial Board Memberships:

2006 - 2021	Journal of Pediatrics: Editorial Board Member; Guest Editor
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Program Committees:

1999 - 2000	Centers for Disease Control (CDC): Expert Panelist, Program on Molecular Genetics Testing
2003 - 2014	Pediatric Academic Societies-Society of Pediatric Research: Program Committee Member (Medical Genetics), CME Liaison, American Society of Human Genetics Annual Meeting
2004, '08, '11, '12	Invited Reviewer (Inborn Error of Metabolism abstracts)
2004, '11, '12	Session Chair, American Society of Human Genetics annual meeting
2006 - 2010	Program Committee Member (inborn Errors of Metabolism)
2007	Organizing Committee Member Tulane University Health Sciences Center 18th Annual Research Days
2008	Invited Member , CDC September 24-26, 2008, Plan I Newborn Screening National Contingency Plan Stakeholder's Workshop
2015	Invited Member , CDC Plan II Newborn Screening National Contingency Plan Stakeholder's Workshop
2017	Newborn Screening and Genetic Technologies Symposium Planning Committee Member, Panel Co-Chair, New Orleans, Sept 10-13, 2017

National & Regional Committees

	Southeast Regional Genetics Group (SERRG)
1995 - 1997	Vice-Chair, Biochemical Genetics Committee
1998 - 2004	Chair, Biochemical Genetics Committee
2004 - 2006	Board of Directors, Biochemical Genetics Chair
2006 - 2008	President-elect
2007	Host and organizer for annual SERRG meeting in New Orleans
2008 - 2010	President
2022 - 2024	President-elect
2024 - 2026	President, incoming
1997 - 2022	International Collaborative Gaucher Group , Board Member and Regional Coordinator
2006 - present	Workgroup On Telegenetics , Co-Chair, National Coordinating Center
2006 - present	Workgroup On Emergency Preparedness , Member, National Coordinating Center
	Louisiana State Genetics Committees
1994 - present	Genetics Advisory Committee, Member
1995 - present	Newborn Screening Subcommittee, Member
2022 - present	Louisiana Rare Disease Advisory Committee, Member

Tulane University Committees

2006 - 2008	University Faculty Tenure, Freedom and Responsibility Committee
2010 - 2012	University Senate
	Tulane University School of Medicine Committees:
1994 - 1998	Curriculum Reform Steering Committee
1994 - 2010	Second Year Curriculum Advisory Committee
1994 - 2000	Research Task Force/Committee, Department of Pediatrics
1998 - 2004	Student Affairs Committee
1999 - 2000	Anatomy Departmental Review Committee
2001 - 2004	Curriculum Committee
2005 - 2008	Center for Continuing Education Advisory Committee
2005 - present	Graduate Medical Education Committee
2007 - 2010	Personnel and Honors Committee
2011 - 2017	ACGME Residency Review Committee: Medical Genetics, Molecular Genetic Pathology

Society Memberships:

1991 - present	American Society of Human Genetics , Member
1993 - present	American College of Medical Genetics , Founding Member, Fellow
1992 - 2017	American Medical Association , Member
1994 - present	Society of Inherited Metabolic Diseases , Member
1994 - 2018	Southern Society of Pediatric Research , Member
1996 - 2000	American Academy of Pediatrics , Fellow Section on Genetics and Birth Defects , Member
1998 - present	Alexander von Humboldt Foundation in America , Member
1999 - present	Association of Professors of Human & Medical Genetics Tulane University School of Medicine Representative
2002 - present	Society of Pediatric Research , Member

Hospital Affiliations:

1994 - present	Tulane University Hospital and Clinic , Full-time Active
1994 - present	Tulane Hospital For Children , Full-time Active
1994 - 2018	Medical Center of Louisiana (Charity Hospital), Full-time
1995 - 2007	Lafayette General Medical Center , Consulting
1997 - present	Women and Childrens Hospital , Lafayette, LA, consulting
1999 - 2005	Ochsner Foundation Hospital and Clinic , consulting
2001 - 2014	H. P. Long Hospital , Pineville LA, consulting
2005 - present	Tulane - Lakeside Hospital
2016 - present	Rapides Regional Medical Center
2019 - present	Childrens Hospital New Orleans , Service Line Chief: Medical Genetics

Languages: German: spoken, reading and written fluency

Bibliography:

Peer-reviewed Articles:

1. Harper GS, Kohn LD, Bernardini I, Bernar J, Tietze F, **Andersson HC**, Gahl WA. Thyrotropin stimulation of lysosomal tyrosine transport in rat FRTL-5 thyroid cells. *J Biol Chem* 263:9320-9325, 1988. (cited by 19)
2. Tietze F, Kohn LD, Kohn AD, Bernardini I, **Andersson HC**, Adamson MD, Harper GS, Gahl WA. Carrier-mediated transport of monoiodotyrosine out of rat thyroid cell lysosomes. *J Biol Chem* 264:4762-4765, 1989. (cited by 38)
3. Adamson MD, **Andersson HC**, Gahl WA. Cystinosis. *Sem Nephrol* 9:147, 1989 (cited by 23)
4. **Andersson HC**, Kohn LD, Bernardini I, Blom HJ, Tietze F, Gahl WA. Characterization of

- lysosomal monoiodotyrosine transport in rat thyroid cells: Evidence for transport by system h. *J Biol Chem* 265:10950-10954, 1990. (cited by 37)
5. Blom HJ, **Andersson HC**, Seppala R, Tietze F, Gahl WA. Defective glucuronic acid transport from lysosomes of infantile sialic acid storage fibroblasts. *Biochem J* 268:621, 1990 (cited by 28)
 6. Sonies B, Ekman EF, **Andersson HC**, Adamson M, Kaler S, Markello T, Gahl WA. Swallowing dysfunction in nephropathic cystinosis. *N Engl J Med* 323:565, 1990. (cited by 75)
 7. Blom HJ, **Andersson HC**, Krasnewich DM, Gahl WA. Pulsed amperometric detection of carbohydrates in lysosomal storage disease fibroblasts: A new screening technique for carbohydrate storage diseases. *J Chromatogr Biomed Appl* 533:11-21, 1990. (cited by 12)
 8. Racusen LC, Fivush BA, **Andersson HC**, Gahl WA. Culture of renal tubular cells from the urine of patients with nephropathic cystinosis. *J Am Soc Nephrol* 1:1028, 1991. (cited by 26)
 9. **Andersson HC**, Markello T, Schneider JA, Gahl WA. The effect of growth hormone treatment on serum creatinine in patients with cystinosis, chronic renal disease. *J Pediatr* 120:716-720, 1992. (cited by 25)
 10. Oerter KE, Friedman TC, **Andersson HC**, Cassorla FG. A familial syndrome of endocrine and neuroectodermal failure. *Am J Med Genet* 44:487-491, 1992. (cited by 7)
 11. **Andersson HC**, Parry DM, Mulvihill JJ. Late-onset hereditary lymphedema complicated by lymphangiosarcoma: case report and nosological implications. *Am J Med Genet* 56:72-75, 1995. (cited by 31)
 12. **Andersson HC**, Kohn A, Gahl WA, Kohn LD. Photoaffinity-labeling of lysosomal membrane proteins with [¹²⁵I]-diiodotyrosine, a system h ligand. *Biochem Molec Med* 55:71-73, 1995. (cited by 1)
 13. Napoleone RM, Varela M, **Andersson HC**. Complex congenital heart malformations in mosaic tetrasomy 8p: a case report and literature report. *Am J Med Genet*, 73:330-3, 1997. (cited by 12)
 14. **Andersson HC**, Shapira, E. Biochemical and clinical response to hydroxocobalamin versus cyanocobalamin treatment in *cb1C* - methylmalonic acidemia/homocystinuria. *J Pediatr*, 132:121-4, 1998. (cited by 88)
 15. **Andersson HC**, Frentz, J, Martinez JE, Tuck-Muller CM, Bellizaire J. Adrenal Insufficiency in Smith-Lemli-Opitz Syndrome. *Am J Med Genet*, 82:382-4, 1999. (cited by 60)
 16. **Andersson HC**, Marble M, Shapira E. Long-term outcome in combined methylmalonic acidemia and homocystinuria (*cb1C*). *Genetics In Medicine*, 1:146-150, 1999. (cited by 68)
 17. Kutcher JS, Kahn MJ, **Andersson HC**, Foundas AL. Neuroacanthocytosis masquerading as Huntingtons Disease: CT/MRI findings. *J Neuroimaging* 9:187-9, 1999. (cited by 41)
 18. Charrow J, **Andersson HC**, Kaplan P, Kolodny E, et al. The Gaucher Registry: Demographics and disease characteristics of 1,698 patients with Gaucher Disease. *Arch Int Med*, 160:2835-43, 2000. (cited by 544)
 19. Burrage L, **Andersson HC**. Phenylketonuria in Louisiana *Louisiana Morbidity Report*, May-June, 2000.
 20. Morava E, Smith C, Pierce M, **Andersson HC**. Management dilemmas in patient with

- hereditary renal adysplasia. *J La Med Jan*;153(1):27-30, 2001. (cited by 2)
21. Waterham HR, Koster J, Romeijn GJ, Vreken P, Hennekam RCM, **Andersson HC**, FitzPatrick D, Kelley RI, Wanders RJA. Mutations in 3beta-hydroxysterol Δ^{24} -reductase cause, desmosterolosis, autosomal recessive disorder of cholesterol biosynthesis. *Am J Hum Genet*, 69:685, 2001. (cited by 326)
 22. Weinreb NJ, Charrow J, **Anderson HC**, Kaplan P, Kolodny EH, et al. Effectiveness of enzyme replacement therapy in 1028 patients with type 1 Gaucher Disease after 2-5 years of treatment: A report from the Gaucher Registry. *Ann Int Med*, 113(2):112-9, 2002. (cited by 620)
 23. **Andersson HC**, Kratz L, Kelley R: Desmosterolosis Presenting with Multiple Congenital Anomalies and Profound Developmental Delay. *Am J Med Genet* 113:315, 2002. (cited by 108)
 24. **Andersson HC**, MA Krousel-Wood, KE Jackson, J Rice, IM Lubin. Medical Genetic Test Reporting in Molecular Cystic Fibrosis and Factor V Leiden Tests Based On Reports Solicited From North American Laboratories. *Gen in Med* 4(5):324-7 2002. (cited by 17)
 25. **Andersson HC**. Disorders of Post-squalene Cholesterol Biosynthesis Leading to Human Dysmorphogenesis. *Cell Molec Biol*, 48:173-178, 2002. (cited by 10)
 26. Krousel-Wood M, **Andersson HC**, Rice J, Jackson KE, Rosner E, Lubin IM. Physicians' Perceived Usefulness of and Satisfaction with Test Reports for Cystic Fibrosis (F508) and Factor V Leiden. *Gen In Med*, vol 5(3):166-171, 2003. (cited by 18)
 27. Charrow J, **Andersson HC**, Kaplan P, Kolodny EH, Mistry P, Pastores G, et al. Enzyme replacement therapy and monitoring for children with type 1 Gaucher disease: consensus monitoring. *J Pediatr*, 144(1):112-20, 2004. (cited by 184)
 28. Morava E, Jackson, KE, **Andersson HC**. Focal skin defect, limb anomalies and microphthalmia. *Clin Dysmorphol* Apr;13(2):113-5, 2004. (cited by 2)
 29. Weinreb NJ, Aggio MC, **Andersson HC**, Andria G, Charrow J, Clarke JT, Erikson A, Giraldo P, Goldblatt J, Hollak C, Ida H, Kaplan P, Kolodny EH, Mistry P, Pastores GM, Pires R, Prakesh-Cheng A, et al. Gaucher disease type 1: Revised recommendations on evaluations and monitoring for adult patients. *Semin Hematol*. 2004 Oct;41(4 Suppl 2):15-22 (cited by 179)
 30. Waterham HR, Koster j, Romeijn GJ, Vreken P, Hennekam RCM, **Andersson HC**, Fitzpatrick D, Kelley RI, RJA Wanders. Moleculaire basis van desmosterolosis, een autosomaal recessief overervend defect in de cholesterol. *Ned Tijdschr Geneesk* 148:253-6, 2004.
 31. Techakittiroj C, Cunningham A, Hooper PF, **Andersson HC**, Thoene J. High Protein Diet Mimics Hypertyrosinemia In Newborn, *J Pediatr*, 146(2):281-282, 2005. (cited by 15)
 32. **Andersson HC**, Charrow J, Kaplan P, Mistry P, et al. Individualization of Long-term Enzyme Replacement Therapy for Gaucher Disease. *Genet in Med* Feb 7:105-110, 2005. (cited by 157)
 33. Techakittiroj C, K.C. Kim, **H. Andersson**, M.M. Li. 9p subtelomere deletion: pathogenic mutation or normal variant? *Beijing Da Xue Xue Bao*. 38(1):92-3, 2006. (cited by 3)

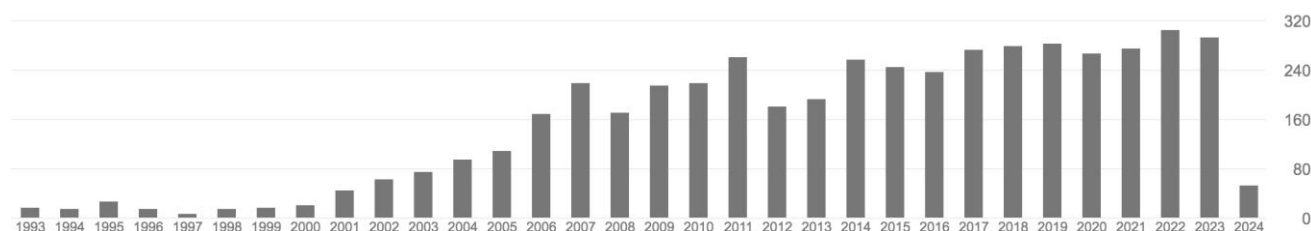
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35. Kaplan P, **Andersson HC**, Kacena KA, Yee J. The Clinical and Demographic Characteristics of Non-Neuronopathic Gaucher Disease in 887 Children at Diagnosis. *Archives Pediatrics & Adolescent Medicine*, 160:604-609, 2006 (cited by 216)
36. Techakittiroj C, **Andersson H**, et al, MM Li. A female infant with hypotonia, developmental delay, transitional hearing loss & 22q13.1 deletion. *World J Pediatr*. 2:245, 2006 (cited by 1).
37. MM Li, MA Nimmakayalu, D Mercer, **H Andersson**, BS Emanuel. Characterization of a Cryptic 3.3 Mb Deletion in a Patient with Developmental Delay and Mild Dysmorphic Features Using High Density Oligo ArrayCGH and Gene Expression Arrays. *Am J Med Genetics*, 146A:368-375, 2008. (cited by 26)
38. **Andersson H**, Kaplan P, et al. Eight-year clinical outcomes of long-term enzyme replacement therapy in 884 children with type 1 Gaucher disease, *Pediatr*,122:1182, 2008. (cited by 179)
39. MM Li, **HC Andersson**. Clinical Application of Microarray-based Molecular Cytogenetics:An Emerging New Era of Genomic Medicine, *J Pediatrics*, Sept 2009: 155(3): 311-7 (cited by 50)
40. B Xiang, Y Shen, R Nasir, M Sobeih, D Miller, K Lu, X Hu, **HC Andersson**, et al. Genome-wide oligonucleotide array CGH for etiological diagnosis of mental retardation: a multi-center experience on 1,499 clinical cases. *J Molec Diagnostics* Mar, 12(2):204-12, 2010 (cited by 71).
41. **HC Andersson**, Perry W, Bowdish B, P Floyd-Browning. Emergency preparedness for genetics centers, laboratories and patients: the SouthEast Region Genetics Collaborative strategic plan. *Genet Med*, ePub: June 2011, October, 2011 (cited by 4).
42. P Floyd-Browning, W Perry, **HC Andersson**. Newborn Screening Results Reporting Survey: A National Snapshot with Implications for Emergency preparedness. *J Pediatr*, 2013 May;162(5):955-7.e1-2. doi: 10.1016/j.jpeds (cited by 1)
43. J Vockley, **HC Andersson**, KM Antshel, NE Braverman, BK Burton, DM Frazier, J Mitchell, WE Smith, BH Thompson, SA Berry. Phenylalanine hydroxylase deficiency: diagnosis and management guideline. *Genet Med* 2014 Feb;16(2):188-200, Epub 2013, Oct 10 (cited by 348).
44. ML Landsverk, VW Zhang, LC Wong, **HC Andersson**. A SUCLG1 Mutation in a patient with mitochondrial DNA Depletion and Congenital Anomalies. *Molec Gen Metab Rep* (2014), pp. 451-454 DOI information: 10.1016/j.ymgmr.2014.09.007 (cited by 13)
45. P Witters, E Debbold, K Crivelly, K Vande Kerckhove, K Corthouts, B Debbold , **H Andersson**, L Vannieuwenborg, S Geuens, M Baumgartner, T Kozicz, L Settles, E Morava. Autism in patients with propionic academia. *Molec Genet Metab* 119(4):317-21, 2016 (cited by 1)
46. G Stuhrman, SJ Perez-Juanazo, K Crivelly, J Smith, **HC Andersson**, E Morava. False-Positive Newborn Screen Using the Beutler Spot Assay for Galactosemia in Glucose-6-Phosphate

Dehydrogenase Deficiency. *JIMD Rep* 2017 doi: 10.1007/8904_2016_34

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48. Rife E, Dunbar A, Nelson SL, **Andersson HC**. Stippled Chondral Calcifications of the Patella in Zellweger Syndrome. *J Pediatr*, January 2018. (cited by 2)
49. Manoli I, Sysol JR, Epping MW, Li L, Wang C, Sloan JL, Pass A, Gagne J, Ktena Y, Li L, Trivedi N, Ouattara B, Zerfas P, Hoffman V, Abu-Asab M, Tsokos M, Kleiner D, Garone C, Cusmano-Ozog K, Enns G, Vernon H, **Andersson HC**, et al, Venditti C. FGF21 underlies a hormetic response to metabolic stress in methylmalonic acidemia. *JCI Insight*, December, 2018, *JCI Insight*. 2018;3(23):e124351. <https://doi.org/10.1172/jci.insight.124351> (cited by 15)
50. Witters P, **H C Andersson**, J Jaeken, C vanKarnebeek, D Lefeber, D Cassiman, E Morava. Dietary supplement with D-galactose improves quality of life in a mild PMM2-CDG patient in a clinical trial pilot. *Orphanet J Rare Dis*, Mar 2021; 16(1):138
51. Jenkins B, CG Fischer, CA Polito, DR Maiese, AS Keehn, M Lyon, MJ Edick, M Taylor, **HC Andersson**, J Bodurtha, M Blitzer, M Muenke, M Watson The 2019 U.S. Medical Genetics Workforce: A Focus on Clinical Genetics *Genet Med*, Aug 2021; 23(8)1458-1464
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54. Adams D, **HC Andersson**, H Bausell, K Crivelly, C Eggerding, M Lah, J Lilienstein, K Lindstrom, M McNutt, JW Ray, H Saavedra, S Sacharow, D Starin, J Tiffany-Amaro, J Thomas, E Vucko, LB Wessenberg, K Whitehall Use of pegvaliase in the management of phenylketonuria: Case series of early experience in US clinics. Aug2021 *Molec Genet Metab Rep*.
55. Upadia J, Y Li, N Walano N, S Deputy, K Gajewski, **H Andersson**. Genotype-phenotype correlation in IARS2-related diseases: A case report and review of literature. Accepted for publication *Clin Case Reports*, Jan, 2022, <http://doi.org/10.1002/ccr3.5401> .
56. Upadia, J, Noh, G, Lefonte, JJ, **HC Andersson**. Biochemical and molecular characteristics among infants with abnormal newborn screen for very-long-chain acyl-CoA dehydrogenase deficiency: A single center experience. *Molecular Genetics and Metabolism Reports*, Volume 37, July 2023,101002, ISSN 2214-4269. <https://doi.org/10.1016/j.ymgmr.2023.101002>

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58. Bier C, Dickey K, Bibb B, Crutcher A, Sponberg R, Chang R, Boyer M, Davis-Keppen L, Matthes C, Tharp M, Vice D, Cooney E, Morand M, Ray J, Lah M, McNutt M, **HC Andersson** Outcomes in 14 live births resulting from Pegvaliase-treated pregnancies in PKU-affected females. *Molec Genet Metabol*, 2024, 141:108152 doi: 10.1016/j.ymgme.2024.108152

Peer-Reviewed Citations: Total citations: 5043; h-index= 29
Citations per year 1993 - 2024 (Google Scholar, February 2024)



Invited Editorials, Reviews, Book Chapters & Letters:

59. **Andersson HC**. Neutral amino acid transport across lysosomal membranes, *Pathophysiology of Lysosomal Transport*, J Thoene, ed., CRC Press., 73-91, 1992.
60. **Andersson HC**, Shapira E. The Mucopolysaccharidoses. in *Cecil's Textbook of Medicine*, 20th Ed.,1996; 21st Ed., 1999.
61. Shapira E, **HC Andersson**. Tay-Sachs Disease. In *Encyclopedia of Neurosciences 3rd Edition*, CD-ROM edition, 2004.
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91. D. F. G. J. Wolthuis, E. V. van Asbeck, **HC Andersson**, E. Morava-Kozicz, MD, PhD
Amyoplasia with congenital eye malformations and wrinkled skin: a new syndrome. American Society of Human Genetics annual mtg, October 2013, Boston (poster).
92. K Kavanagh, **HC Andersson**, E Morava. Clinical Variability of Osteogenesis Imperfecta Within Families. Southern Society for Pediatric Research, New Orleans, LA , Feb 2014, Journal of Investigative Medicine 02/2014; 62(2):422-422. (poster)
93. **HC Andersson**, A Cunningham, K Crivelly, Y Li, D Goldstein, TJ Chen. Evaluation of Behavior, Executive Function, Neurotransmitter Function and Genomic Expression in PKU "Nonresponders" to Sapropterin. SIMD annual mtg, March, 2014 (poster)
94. **HC Andersson**, A Cunningham, K Crivelly, Y Li, D Goldstein, TJ Chen. Evaluation of Behavior, Executive Function, Neurotransmitter Function and Genomic Expression in PKU "Nonresponders" to Sapropterin. (Abstract #2274) Presented at 64th Annual Meeting of the American Society of Human Genetics, October 20, 2014, San Diego, CA
95. Sylvia Mann, Alisha Keehn, **Hans Andersson**. Who Is Using Telegenetics In the United States: A National Survey. American Public Health Association annual meeting, Nov 17, 2014, New Orleans, LA
96. C Dvorak, J Langston, J Hoffman, **H Andersson**. Risk of BRCA Mutation in Patients without Access to Genetics Testing. Amer Col Medical Genetics March 2015, Salt Lake City (poster).
97. S Mann, A Keehn, L Dean, **H Andersson**, B Schaeffer. Increasing Telegenetics Providers in United States. Amer Coll Medical Genetics March 2015, Salt Lake City, UT, (poster).
98. **HC Andersson**, William Perry. Regional Newborn Screening Laboratory Backup Planning: 2015 Snapshot, APHL Newborn Screening and Genetics, March 2016, St. Louis, MO (poster).
99. L. Mao, A. Janssen, Y. Li, C. Dvorak, **H. Andersson**, T. Chen. Partial uniparental disomy results in homozygous 2p21 deletion in a male newborn with hypotonia-cystinuria syndrome. Am Soc Hum Genet annual meeting, October 19, 2016, Vancouver, (poster)
100. PK Mistry, JL Batista, HC. Andersson, M Balwani, TA Burrow, J Charrow, P Kaplan, A Khan, PS Kishnani, EH Kolodny, B Rosenbloom, CR Scott, N Weinreb. Transformation in Pre-treatment Presentations of Gaucher Disease During the First Two Decades of Imiglucerase Enzyme Replacement Therapy: Report from The ICGG Gaucher Registry. *Blood* 2016 128:4877 58th Amer. Soc. Hematol annual meeting (poster)
101. N Weinreb, JL Batista, HC. Andersson, M Balwani, TA Burrow, J Charrow, P Kaplan, A Khan, PS Kishnani, EH Kolodny, B Rosenbloom, CR Scott, PK Mistry. Transformation in Pre-treatment Presentations of Gaucher Disease During the First Two Decades of Imiglucerase Enzyme Replacement Therapy: Report from The ICGG Gaucher Registry. *Molec Genet Metab* 120:S139, 2017, WORLD LSD annual meeting, Feb 2017 (poster)

102. Walano, N.; Preston, G.; Dvorak, C.; Nelson, S.; **Andersson, H.**; Morava, E.
Homicidal tendencies at early pediatric age in males carrying variants in IQSEC2.
Poster presented at the 35th annual Southeast Regional Genetics Group Conference,
July 2017
103. Crivelly, KS, Cunningham AC, Stuhrman, GA, Morava E, TJ Chen, **Andersson HC**, “The Super Responders: Kuvan Provides Significant Dietary Liberalization in 17 of 72 PKU Patients.”
Poster presented at Genetic Metabolic Dietitians International Conference and Southeastern Regional Genetics Group Annual Meeting, 2018
103. **Andersson HC**, Debbold E, Nelson S, Settles L. “Establishment of a Developmental Neurogenetics Clinic: Characterization of a Cohort of 56 Consecutive Patients with Autism, Developmental Delay +/- Epilepsy”, Southeastern Regional Genetics Group Annual Meeting, July 2018 (platform)
104. Adviento B, Sarkasian A, Walano N, **Andersson HC**, and JJ Karlitz. Feasibility of utilizing PREMM score4 for Lynch Syndrome identification in an urban, minority patient population. *Gastroenterology* 2019 (April):156(6):S-823-S-824 Digestive Disease Week, San Diego, May 18, 2019 (poster)
105. Thakore P, Raja Jawad Kiani, Y Li, **H Andersson**, MC Steinhardt, Alston Dunbar. Infant with 16p11.2 microdeletion syndrome with multiple congenital malformations. Southern Regional Meeting, New Orleans, Feb 21 2019 (poster)
106. M. Shinawi, J. Fleischer, **H. Andersson**, L. White, S. Hughes, T. Hsu. Familial cases of DeSanto Shinawi syndrome with previously unreported nonsense and missense variants in WAC
107. Upadia, J, Walano, N, Crivelly, K, **H Andersson**. An adult male with combined malonic and methylmalonic aciduria: Case report. Annual SERGG mtg, Asheville, July, 2019 (poster)
108. Walano, N, **Andersson HC**. An Unusual Case of Cornelia De Lange Syndrome with Triple X. Annual SERGG mtg, Asheville, July, 2019 (poster)
109. **Andersson H**, Bodurtha J, Blitzler M, Watson M, Lyon M, et al. What does the Medical Genetics workforce look like today? Data from the 2019 Medical Geneticist Workforce Survey, ACMG Annual Meeting March 2020 (platform)
110. Upadia, J, **Andersson HC**. Carnitine deficiency: A case report of an adult female presenting with hyperammonemic encephalopathy. SERGG Annual mtg , Asheville, July 14-16, 2022 (poster)
111. Crivelly K, Cerminaro C, Cunningham A, Noh GS, Upadia J, **Andersson HC**. Palynziq: Is It Worth the Weight? SERGG Annual mtg , Asheville, July 14-16,, 2022 (poster)
112. Li Y, Upadia J, Chenevert M, McKoin M, Liu J, Cen TJ, **H Andersson**. A Family Review of Phenotypic Spectrum Associated with the Interstitial duplications at 4q31.1 and / or 16p13.3 SERGG Annual mtg , Asheville, July 14-16, 2022 (poster)

113. Gurung SR, Kenneson A, **Andersson HC**, and Singh RH. Patient and provider virtual summit: The current state of medium chain acyl-CoA dehydrogenase deficiency management. SERGG Annual Mtg, Charleston, SC, July 13-15, 2023 (platform)
114. Upadia, J, Noh, G, **Andersson HC**. Biochemical and molecular characteristics among infants with abnormal newborn screen for very-long-chain acyl-CoA dehydrogenase deficiency: A single center experience. SERGG Annual Mtg, Charleston, SC, July 13-15, 2023 (platform)
115. Bier C, Dickey K, Bibb B, Crutcher A, Sponberg R, Chang R, Boyer M, Davis-Keppen L, Matthes C, Tharp M, Vice D, Cooney E, Morand M, Ray J, Lah M, McNutt M, **HC Andersson** Outcomes in 14 live births resulting from Pegvaliase-treated pregnancies in PKU-affected females. SERGG Annual Mtg, Charleston, SC, July 13-15, 2023 (poster)
116. Bier C, Dickey K, Bibb B, Crutcher A, Sponberg R, Chang R, Boyer M, Davis-Keppen L, Matthes C, Tharp M, Vice D, Cooney E, Morand M, Ray J, Lah M, McNutt M, **HC Andersson** Outcomes in 14 live births resulting from Pegvaliase-treated pregnancies in PKU-affected females. Amer Coll Medical Genetics annual meeting, April 2024 (poster)
117. Cooney E, Ammous Z, **Andersson H**, Bender T, Clague G, Clifford M, Crutcher A, Davis-Keppen L, Havens K, Lah M, Sacharow S, Sanchez-Valle A, Vucko E, Wardley B, L Wessenberg. LESSONS LEARNED FROM 5-YEARS OF EXPERIENCE WITH PEGVALIASE IN US CLINICS: A CASE SERIES. Society Inherited Metab Diseases annual meeting, April 2024 (poster)

INVITED PRESENTATIONS

Intramural Lectures:

I. Hayward Genetics Center Seminars

- The Natural History of Treated Methylmalonic Acidemia/Homocystinuria (Cbl C) 1997
- Cholesterol Biosynthesis Defects: Lessons in (Dys)Morphogenesis, June 2002
- Identifying the Cobalamin C Disease Gene, March 2005
- Disaster Preparedness for Medical Specialty Groups, April, 2006
- Patient Registries in Rare Disease Research: Establishing Nat'l History/Clinic Outcomes, Sept 2008
- Personalized Medicine and Direct-To-Consumer Genetic Testing, 2011
- Newborn Screening for LSDs, 2018
- Diagnostic Outcomes in Developmental Neurogenetics, 2018

II. Tulane Pediatric Grand Rounds

- Contiguous Gene Syndromes, May 1994
- The Sick Infant – Beyond Ruling-out Sepsis, April 1996
- Enzyme Replacement Therapy in the Lysosomal Storage Diseases, December 1998
- Molecular and Metabolic Advances in the Diagnosis of Mental Retardation, August 1999
- Phenylketonuria and Maternal PKU, August 2002
- Lysosomal Storage Diseases Update., December 2005
- A New Epoch in Screening and Treatment for Lysosomal Storage Disorders, April 2007
- Disaster Preparedness for Medical Specialty Groups, October 2008
- Newborn Screening in Lysosomal Storage Disorders, April 2018

III. Tulane Pathology Department Grand Rounds,

- Lysosomal Storage Disorders, May 1994
- A New Epoch in Lysosomal Storage Disease Understanding: Breakthroughs in Screening, Treatment and Pathophysiology, March 2010

IV. Tulane Internal Medicine Department Grand Rounds

- Novel Clinical Aspects of Diagnosis and Treatment In Managing Adult Disease, October 2007

V. CHNOLA Grand Rounds

Establishing a Developmental Neurogenetics Clinic: Characterization of 56 Patients With Autism, Developmental Delay +/- Epilepsy, September 11, 2019

Extramural/Invited Lectures:

Care of the Newborn/Regional Pediatric Update Conference:

The Approach to the Dysmorphic Child, February 1998.
 The Genetic Approach To Limb Anomalies, February 1999
 Common Genetic Syndromes, February 2005
 Newborn Screening: It's Not Just PKU Anymore, April 2007
 Newborn Screening: 50-Year Anniversary!! April 2015

University of Brescia, Italy, Department of Biochemistry, Lysosomal Membrane Transport in Eukaryotes April 1994

Louisiana State University School of Medicine, Molecular and Human Genetics Center of Excellence, Clinical and Biochemical Aspects of Lysosomal Membrane Transport, April 1996.

Southeast Genetics Group Biochemical Genetics Workshop, Clinical Presentation and Diagnostic Second Regional Conference on Down Syndrome, Atlanta, Ga, Validity of Amino Acid Analysis and Therapy in Down Syndrome

Louisiana State University School of Veterinary Medicine, Humboldt Society in America Lecture, Clinical and Biochemical Aspects of Lysosomal Membrane Transport, May 1998.

Southern Genetics Group annual meeting, Ft. Walton, FLA, Controversial Metabolic Therapies in Trisomy 21, July, 1998.

Louisiana Maternal & Child Health Conference: Learning From The Past/ Designing the Future, Genetics In The MCH Population, March 1999

American Association of Mental Retardation, New Molecular Insights In Developmental Disabilities, May 1999.

Genetics Of the Acadian People: Inherited Metabolic Diseases Breakout Session, McNeese State University, August 1999.

First International Workshop on Niemann-Pick C Disease, National Institutes of Health, Bethesda, MD; Clinical Outcome In N-P C Following Liver Transplant In Infancy. October 1999 (poster)

Ochsner Foundation Hospital Grand Rounds, Molecular and Metabolic Advances in the Diagnosis of Mental Retardation, February, 2000

Genetics of the Acadian People, Nichols State University, Incidence of hyperphenylalaninemia in Louisiana from 1985 – 1999: a 15 year retrospective. March 2001

Family Practice 2001 - A Case-based Clinical Update, Tulane University Department of Family Practice, Evaluation of Dysmorphic Children, May 2001

Secretary's Advisory Committee on Genetic Testing, Issues in Medical Genetic Test Reporting, Bethesda, MD, November 16, 2001.

University of Kumamoto International Symposium, Disorders of Cholesterol Biosynthesis: Lessons for Dysmorphology, keynote address, Kumamoto, Japan, January 2002.

University of Kumamoto Medical School, Department of Pediatrics, Advances in Molecular and Metabolic Diagnosis of Mental Retardation, Kumamoto, Japan, January, 2002.

Association of Molecular Pathologists Annual Meeting, Invited Workshop Speaker, Issues in Medical Genetic Test Reporting, Dallas, November 2002

9th Ross Metabolic Conference, Biochemistry and Outcome of Patients With Combined MMA and Homocystinuria. Houston, April 2003.

International Investigators Meeting, International Collaborative Gaucher Group, Meeting Co-Chair; Pediatric Gaucher Disease: Can the Phenotype Be Reversed; Miami, May, 2003

Louisiana Chapter of American Academy of Pediatrics Annual Meeting, Newborn Screening in the New Millennium. New Orleans, October 2003

Louisiana Perinatal/Maternal Child Health meeting, Genetic Care and the Human Genome Project: The Sky's The Limit and Who'll Pay For It, March, 2004

National Tay-Sachs and Allied Diseases ann conf., Tay-Sachs Disease in Louisiana, April 14, 2005.

- Pediatric Academic Societies Annual Meeting**, Approaches to Diagnosis of Genetic Disorders: Which is the Right Test? Wash. DC, May, 2005
- Clinical Advances in Pediatrics 2005**, , Kansas City, MO, November 2005
Lysosomal Storage Diseases Update. (keynote address); The Future of Genetics
- American College of Medical Genetics Annual Meeting**, Plenary session of ACMG Public Health Genetics Special Interest Group Forum: Genetic and Metabolic Health Care Delivery During and After Hurricanes Katrina and Rita, San Diego, March, 2006
- Pediatric Academic Societies Annual Meeting**, Co-Chair, plenary session: Inherited Disorders Caused by Inappropriate Apoptosis; Intro and Overview of Apoptosis, San Francisco, May, 2006
- 10th Annual Lysosomal Storage Registries Meeting**, Current Progress in the Gaucher Disease Registry (platform presentation), Orlando, May, 2006
- New York - Mid-Atlantic Consortium for Genetic and Newborn Screening Services, Work Group #3 Meeting**, Louisiana Disaster Response Experience, Baltimore, June, 2006
- Disaster Preparedness Workshop**, Southeast Regional Genetics Group, Workshop organizer and speaker, New Orleans, August 27-28, 2006.
- Acadian Pediatrics Grand Rounds**, Expanded Newborn Screening In Louisiana: It's Not Just PKU Anymore, September 2006.
- 3rd Meeting of the National Coordinating Center for the Regional Genetics and Newborn Screening Collaborative Groups**; Report from the Telegenetics Workgroup; Report of Disaster Preparedness from SERGG, Virginia, October, 2006.
- Louisiana State University Health Sciences Center**, Department of Genetics Seminar Series, A New Epoch in Lysosomal Storage Disease Understanding: Breakthroughs in Screening, Treatment & Pathophysiology, November, 2006.
- Tulane Pediatric CME Series**, Expanded Newborn Screening In Louisiana: It's Not Just PKU Anymore, November 2006
- National Coordinating Center for Genetics and Newborn Screening Collaborative Groups**: Emergency Preparedness for Newborn Screening and Genetic Services National Workgroup Meeting, Southeastern Regional Genetics Group. McLean, VA, February, 2007
- Spevak Visiting Professorship** to Blank Children's Hospital, Des Moines, Iowa, October 3-5, 2007.
- * Treatment of Genetic Diseases Without Gene Therapy
 - * Expanded Newborn Screening: It Isn't Just PKU Anymore.
 - * Disaster Preparedness for Medical Specialty Groups: Lessons Learned from Katrina and Rita
 - * Diagnostic Approaches to Genetic Cases
- National Gaucher Foundation**, Pediatrics Type 1 Gaucher Disease, October 15, 2007
(audiostreaming @ <http://www.gaucherdisease.org/audio9.php>)
- Biology Department Seminar**, SUNY-Potsdam College: Cholesterol Biosynthesis Defects: Lessons for (Dys)Morphogenesis, December 5, 2007. Given as web-based distance learning lecture.
- American College Medical Genetics** annual meeting, Phoenix. Controlling Phe Levels: Current and Emerging PKU Treatment Options, part of an educational symposium: Emerging Insight and Therapeutic Guidance to Optimize PKU Outcomes; March 15, 2008
- Lysosomal Disease Network WORLD Symposium**: Clinical outcome following 8-year enzyme replacement therapy in 884 symptomatic children with type 1 Gaucher Disease. Feb 2008.
- 12th Annual North American Lysosomal Disease Registries Meeting**, Montreal, CANADA
What does the Registry have to share regarding clinical outcomes of long-term Cerezyme therapy in pediatric type 1 Gaucher disease? Friday May 9, 2008
- Southeast Region Genetics Group** annual meeting, Charleston, SC, Safety and Efficacy of BH4 in Treatment of PKU July 31, 2008 (Biomarin-sponsored satellite meeting).
- Greenwood Genetics Center**, Greenville, SC, Emergency Preparedness For Genetics Centers: Steps Every Clinician Can Take To Mitigate An Emergency, October 22, 2008.

Associated Maternal Child Health Programs (AMCHP) annual meeting February, 2009, Washington DC; Newborn Screening: Developing a National Contingency Plan.

American College Medical Genetics Annual Meeting, March 25-29, 2009, Examining the Neurocognitive and Behavioral Aspects in PKU: A Focus on Special Populations

12th Abbott Nutrition Metabolic Conference "Advances in Management of Inherited Metabolic Disorders", May 14-16, 2009: 2 platform presentations:
 *Medical Management of Cobalamin C-Methylmalonic Acidemia
 *Interactive Management Workshop: PKU and Kuvan.

New York - Mid-Atlantic Consortium (NYMAC) For Genetic and Newborn Screening Services Advisory Council Meeting, May 18-19, 2009, Special Presentation on Contingency Planning/Emergency Preparedness for Clinical Genetic Services

National Coordinating Center for Regional Genetic Collaboratives: Telehealth Policy Meeting: Reimbursement and Financing for Telegenetics, November 5-6, 2009

XIII Latin America Symposium on Lysosomal Storage Diseases (Mexico): Gaucher Disease in Pediatric Patients: Outcomes of Imiglucerase, December 11-12, 2009

PKU and BH4 Scientific Development Summit: Phenylketonuria, Tetrahydrobiopterin and Neuropsychiatric Phenotype. San Diego, CA, June 3, 2010; Washington, DC, November 1, 2010.

American Council of Life Insurers Medical Section Scientific Program: Genetics and Genomics in Health Outcomes Prediction. New Orleans, LA, February, 2011.

New England Regional Genetics Collaborative, Special Presentation on Contingency Planning/Emergency Preparedness for Clinical Genetic Services. Boston, April 1, 2011.

American Telemedicine Association Annual Meeting, Telegenetics in Region 3 (Southeast Regional Emory University Genetics Department, Telemedicine In Genetics: National and Regional, June 23, 2011([http://southeastgenetics.org/presentation.php/18/Telegenetics_in_Region_3_\(SERC\)_Past_Successes_and_Plans_for_the_Future](http://southeastgenetics.org/presentation.php/18/Telegenetics_in_Region_3_(SERC)_Past_Successes_and_Plans_for_the_Future))

New Horizons In Pediatrics - Childrens Healthcare of Atlanta (Emory University): Paul Fernhoff Memorial Lecture - Translating Genetic Advances to the Child's Bedside, Atlanta, April, 2012

American Academy of Pediatrics – National Conference 2012 Genetic and Metabolic Unknowns, October, 2012. New Orleans, LA

5th Gaucher Leadership Conference - Approaches to monitoring for Gaucher disease: Variability and Special Cases, Madrid, November, 2013

American College of Medical Genetics 2014 Annual Mtg - Community Conversation- Transitioning Genetics and Rare Conditions into Adulthood. March, 2014 Nashville, TN

Genetic and Metabolic Dieticians International 2014 - ACMG Guidelines on Treatment of Phenylalanine Hydroxylase Deficiency. April, 2014, Dallas/Ft. Worth, TX

Fostering Value-Based Medicine - Regional LSD Registries Meeting. November 2014, Atlanta, GA
 * Value-Based Medicine in LSDs
 * Newborn Screening in Lysosomal Storage Disorders

Using Telemedicine To Increase Access To Clinical Genetic Care, University of Mississippi Division of Medical Genetics Seminar, June 4, 2015

Emergency Preparedness for PKU Families – Are You Prepared, Nat'l PKU Alliance annual meeting, St. Louis, MO, July 30, 2016 (breakout session)

Telegenetics Workshop – Southeast Region Genetics Collaborative, Chair, annual SERC meeting, Jacksonville, July 29, 2016

International Collaborative Gaucher Group: 25 Years of Achievement, Western Regional Rare Disease Registries Meeting. Salt Lake City, October 13, 2016, keynote platform

16th Abbott Nutrition Metabolic Conference, TeleMedicine For Metabolics, March 23-25, 2017, Charleston.

- Mountain States Regional Genetics Collaborative**, Annual Meeting, Katrina, Emergency Preparedness and What We Learned. Apr 19-20, 2017, Salt Lake City.
- Parent/Patient Panel Discussion**, Co-Chair, APHL Newborn Screening and Genetic Technology Symposium, New Orleans, September 12, 2017.
- Newborn Screening Dilemmas in Lysosomal Storage Disorders**, Cedars-Sinai Annual Symposium on Lysosomal Storage Disorders, Keynote Speaker, Dec 8, 2017
- Newborn Screening and Emergency Preparedness: Lessons Learned in Louisiana**, APHL Newborn Screening Short-term Follow-up Workgroup, live webinar, Oct 22, 2018.
- How Expanded Newborn Screening and Next Gen Sequencing Has Revolutionized Genetic Disease Outcomes** Invited speaker at Gulf Coast Regional Genetics Meeting of Foundation of Genetic Technologists, May 2019.
- Northwest (La) Pediatric Society Annual Meeting**, December 11, 2019, Establishing a Developmental Neurogenetics Clinic (invited keynote).
- American Society of Medical Genetics 2020 Annual Meeting** What does the Medical Genetics workforce look like today? Data from the 2019 Medical Geneticist Workforce Survey May, 2020, annual mtg (platform presentation, virtual)
- Univ of Mississippi Pediatric Genetics ECHO** What's Happening in the SouthEast Region Genetics Network (SERN). Invited speaker at. May 10, 2021
- Univ of Mississippi Pediatric Grand Rounds**, Establishing a Developmental Neurogenetics Clinic: Genetics of Autism, Epilepsy and Developmental Delay. April 27, 2022 (platform)
- SouthEast Regional Genetics Group** 2022 annual meeting, Asheville: The Medical Genetics Workforce in 2022, July 16, 2022 (platform)
- Emory School of Medicine**. Medium Chain Acyl CoA Dehydrogenase Deficiency – Medical Overview. Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCADD), Patient & Family Education Summit with Providers. Febr 25, 2023. (virtual lecture).