CURRICULUM VITAE

I. Personal Information

Name: Kristina Patrice Cusmano-Ozog, MD
e-mail address: kcusmano@stanford.edu

II. Educational Background

Undergraduate Education
University of Miami College of Arts and Sciences, Bachelor of Science in Biochemistry & Molecular Biology, General Honors and Departmental Honors in Biochemistry, Miami, FL, August 1995 - May 1998

Graduate/Medical Education
University of South Florida College of Medicine, Doctor of Medicine, Tampa, FL, August 1998 - May 2002

Post-Graduate Training
Resident, Pediatrics, University of South Florida, Tampa, FL, July 2002 - June 2005
Resident, Medical Genetics, Stanford University, Stanford, CA, July 2005 - June 2007
Fellow, Clinical Biochemical Genetics, Stanford University, Stanford, CA, July 2007 - June 2008
Postdoctoral Fellow, Biochemical Genetics, Stanford University, Stanford, CA, July 2008 - January 2011
Fellow, Clinical Molecular Genetics, National Institutes of Health, Bethesda, MD, July 2014 - June 2015

III. Professional Appointments

Clinical Instructor, Department of Pediatrics/Division of Medical Genetics, Stanford University, Stanford, CA, January 2008 – January 2011
Interim Director, Clinical Biochemical Genetics Laboratory, Pathology, Stanford University School of Medicine, January 2011 – April 2011
Director, Biochemical Genetics and Metabolism Laboratory, Laboratory Medicine, Children's National Medical Center, Washington, DC, July 2011 – May 2019
Assistant Professor of Pediatrics & Pathology, George Washington University Medical Center, Washington, DC, July 2013 – May 2019
Director, Molecular Diagnostics Laboratory, Laboratory Medicine, Children's National Medical Center, Washington, DC, July 2015 – May 2019
Program Director, Medical Genetics Residency Program, Children's National Medical Center, Washington, DC, July 2016 – May 2019
Co-Director, Clinical Biochemical Genetics Laboratory, Pathology, Stanford University School of Medicine, July 2019 – Present
Clinical Associate Professor, Pathology, Stanford University School of Medicine, July 2019 – Present

IV. Honors and Awards

Alpha Epsilon Delta Premedical Honor Society, 1996 - 1998
Golden Key National Honor Society, 1998
General Honors and Departmental Honors in Biochemistry, 1998
USF College of Medicine Class of 2002 Vice President, 1999-2000
USF Pediatric Residency Program 'Case of the Week' Winner, 2002-2003, 2003-2004, 2004-2005
Tampa General Hospital's September Resident of the Month, 2003
Travel Award to attend the Western Society of Pediatric Research meeting, 2007
Travel Award to attend the Society for Inherited Metabolic Disorders meeting, 2007 and 2008
American Federation for Medical Research Regional Scholar, 2009
Power of One Award, Children's National, April 2019

V. Scholarly Publications

Complete List of Published Work in MyBibliography:
https://www.ncbi.nlm.nih.gov/myncbi/browse/collection/55000918

A. Peer-reviewed journal articles (original research)

1. **Cusmano**-Ozog K, Manning MA, Hoyme HE. 22q13.3 deletion syndrome: a recognizable malformation syndrome associated with marked speech and language delay. American journal of medical genetics. Part C, Seminars in medical genetics. 2007; 145C(4):393-8. PMID: 17926345

2. Adam MP, Kobori JA, **Cusmano**-Ozog K, Chen KM, Hoyme HE. Progressive and symmetric supraorbital hyperostosis with bony and soft tissue overgrowth in an Ethiopian female: a newly recognized overgrowth syndrome? American journal of medical genetics. Part A. 2008; 146A(5):543-7. PMID: 18241057

3. Cheyette BN, Cheyette SN, **Cusmano**-Ozog K, Enns GM. Dopa-responsive dystonia presenting as delayed and awkward gait. Pediatric neurology. 2008; 38(4):273-5. PMID: 18358407

4. Zhang B, Lo C, Shen L, Sood R, Jones C, **Cusmano**-Ozog K, Park-Snyder S, Wong W, Jeng M, Cowan T, Engleman EG, Zehnder JL. The role of vanin-1 and oxidative stress-related pathways in distinguishing acute and chronic pediatric ITP. Blood. 2011; 117(17):4569-79. PMID: 21325602

5. Arnold GL, Salazar D, Neidich JA, Suwannarat P, Graham BH, Lichter-Konecki U, Bosch AM, **Cusmano**-Ozog K, Enns G, Wright EL, Lanpher BC, Owen NN, Lipson MH, Cerone R, Levy P, Wong LJ, Dezsofi A. Outcome of infants diagnosed with 3-methyl-crotonyl-CoA-carboxylase deficiency by newborn screening. Molecular genetics and metabolism. 2012; 106(4):439-41. PMID: 22658692

6. Moore T, Le A, Niemi AK, Kwan T, **Cusmano**-Ozog K, Enns GM, Cowan TM. A new LC-MS/MS method for the clinical determination of reduced and oxidized glutathione from whole blood. Journal of chromatography. B, Analytical technologies in the biomedical and life sciences. 2013; 929:51-5. PMID: 23660247

7. Manoli I, Sysol JR, Li L, Houillier P, Garone C, Wang C, Zerfas PM, **Cusmano**-Ozog K, Young S, Trivedi NS, Cheng J, Sloan JL, Chandler RJ, Abu-Asab M, Tsokos M, Elkahloun AG, Rosen S, Enns GM, Berry GT, Hoffmann V, DiMauro S, Schnermann J, Venditti CP. Targeting proximal tubule mitochondrial dysfunction attenuates the renal disease of methylmalonic acidemia. Proceedings of the National Academy of Sciences of the United States of America. 2013; 110(33):13552-7. PMID: 23898205, PMCID: PMC3746875

8. Chandler RJ, Tarasenko TN, **Cusmano**-Ozog K, Sun Q, Sutton VR, Venditti CP, McGuire PJ. Liver-directed adeno-associated virus serotype 8 gene transfer rescues a lethal murine model of citrullinemia type 1. Gene therapy. 2013; 20(12):1188-91. NIHMSID: NIHMS520081 PMID: 24131980, PMCID: PMC3855546

9. Enns GM, Moore T, Le A, Atkuri K, Shah MK, **Cusmano**-Ozog K, Niemi AK, Cowan TM. Degree of glutathione deficiency and redox imbalance depend on subtype of mitochondrial disease and clinical status. PloS one. 2014; 9(6):e100001. PMID: 24941115, PMCID: PMC4062483

10. Kruszka PS, Kirmse B, Zand DJ, **Cusmano**-Ozog K, Spector E, Van Hove JL, Chapman KA. Concurrent non-ketotic hyperglycinemia and propionic acidemia in an eight year old boy. Molecular genetics and metabolism reports. 2014; 1:237-240. PMID: 27896094, PMCID: PMC5121296

11. Fraser JL, Vanderver A, Yang S, Chang T, Cramp L, Vezina G, Lichter-Konecki U, **Cusmano**-Ozog KP, Smpokou P, Chapman KA, Zand DJ. Thiamine pyrophosphokinase deficiency causes a Leigh Disease like phenotype in a sibling pair: identification through whole exome sequencing and management strategies. Molecular genetics and metabolism reports. 2014; 1:66-70. PMID: 27896076, PMCID: PMC5121315

12. Le A, Ng A, Kwan T, **Cusmano**-Ozog K, Cowan TM. A rapid, sensitive method for quantitative analysis of underivatized amino acids by liquid chromatography-tandem mass spectrometry (LC-MS/MS). Journal of chromatography. B, Analytical technologies in the biomedical and life sciences. 2014; 944:166-74. PMID: 24316529

13. Johnston JJ, Sapp JC, Curry C, Horton M, Leon E, **Cusmano**-Ozog K, Dobyns WB, Hudgins L, Zackai E, Biesecker LG. Expansion of the TARP syndrome phenotype associated with de novo mutations and mosaicism. American journal of medical genetics. Part A. 2014; 164A(1):120-8. NIHMSID: NIHMS610067 PMID: 24259342, PMCID: PMC4443488

14. Tarasenko TN, Singh LN, Chatterji-Len M, Zerfas PM, **Cusmano**-Ozog K, McGuire PJ. Kupffer cells modulate hepatic fatty acid oxidation during infection with PR8 influenza. Biochimica et biophysica acta. 2015; 1852(11):2391-401. PMID: 26319418

15. Caterino M, Chandler RJ, Sloan JL, Dorko K, **Cusmano**-Ozog K, Ingenito L, Strom SC, Imperlini E, Scolamiero E, Venditti CP, Ruoppolo M. The proteome of methylmalonic acidemia (MMA): the elucidation of altered pathways in patient livers. Molecular bioSystems. 2016; 12(2):566-74. NIHMSID: NIHMS746999 PMID: 26672496, PMCID: PMC4858437

16. Ferreira CR, **Cusmano**-Ozog K. Spurious Elevation of Multiple Urine Amino Acids by Ion-Exchange Chromatography in Patients with Prolidase Deficiency. JIMD reports. 2017; 31:45-49. PMID: 27067078, PMCID: PMC5388643

17. Ferreira CR, Devaney JM, Hofherr SE, Pollard LM, **Cusmano**-Ozog K. Hereditary fructose intolerance mimicking a biochemical phenotype of mucolipidosis: A review of the literature of secondary causes of lysosomal enzyme activity elevation in serum. American journal of medical genetics. Part A. 2017; 173(2):501-509. PMID: 27797444

18. Raval DB, **Cusmano**-Ozog KP, Ayyub O, Jenevein C, Kofman LH, Lanpher B, Hauser N, Regier DS. Diagnosis of LCHAD/TFP deficiency in an at risk newborn using umbilical cord blood acylcarnitine analysis. Molecular genetics and metabolism reports. 2017; 10:8-10. PMID: 27995076, PMCID: PMC5155040

19. Scott AI, **Cusmano**-Ozog K, Enns GM, Cowan TM. Correction of hyperleucinemia in MSUD patients on leucine-free dietary therapy. Molecular genetics and metabolism. 2017; 122(4):156-159. PMID: 29032949

20. Muriello MJ, Viall S, Bottiglieri T, **Cusmano**-Ozog K, Ferreira CR. Confirmation that *MAT1A* p.Ala259Val mutation causes autosomal dominant hypermethioninemia. Molecular genetics and metabolism reports. 2017; 13:9-12. PMID: 28748147, PMCID: PMC5512230

21. Kirmse B, Cabrerra-Luque J, Ayyub O, **Cusmano** K, Chapman K, Summar M. Plasma fibroblast growth factor-21 levels in patients with inborn errors of metabolism. Molecular genetics and metabolism reports. 2017; 13:52-54. PMID: 28920014, PMCID: PMC5586549

22. Wilnai Y, Blumenfeld YJ, **Cusmano** K, Hintz SR, Alcorn D, Benitz WE, Berquist WE,

Bernstein JA, Castillo RO, Concepcion W, Cowan TM, Cox KL, Lyell DJ, Esquivel CO, Homeyer M, Hudgins L, Hurwitz M, Palma JP, Schelley S, Akula VP, Summar ML, Enns GM. Prenatal treatment of ornithine transcarbamylase deficiency. Molecular genetics and metabolism. 2018; 123(3):297-300. PMID: 29396029

23. Beck DB, **Cusmano**-Ozog K, Andescavage N, Leon E. Extending the phenotypic spectrum of Sengers syndrome: Congenital lactic acidosis with synthetic liver dysfunction. Translational science of rare diseases. 2018; 3(1):45-48. PMID: 29682452, PMCID: PMC5904566

24. Tarasenko TN, **Cusmano**-Ozog K, McGuire PJ. Tissue acylcarnitine status in a mouse model of mitochondrial β-oxidation deficiency during metabolic decompensation due to influenza virus infection. Mol Genet Metab. 2018 Jun 23. pii: S1096-7192(18)30218-X. doi: 10.1016/j.ymgme.2018.06.012. [Epub ahead of print] PMID: 30031688

25. Manoli I, Sysol JR, Epping MW, Li L, Wang C, Sloan JL, Pass A, Gagné J, Ktena YP, Li L, Trivedi NS, Ouattara B, Zerfas PM, Hoffmann V, Abu-Asab M, Tsokos MG, Kleiner DE, Garone C, **Cusmano**-Ozog K, Enns GM, Vernon HJ, Andersson HC, Grunewald S, Elkahloun AG, Girard CL, Schnermann J, DiMauro S, Andres-Mateos E, Vandenberghe LH, Chandler RJ, Venditti CP. FGF21 underlies a hormetic response to metabolic stress in methylmalonic acidemia. JCI Insight. 2018;3(23) PMID: 30518688

26. Raible SE, Mehta D, Bettale C, Fiordaliso S, Kaur M, Medne L, Rio M, Haan E, White SM, **Cusmano**-Ozog K, Nishi E, Guo Y, Wu H, Shi X, Zhao Q, Zhang X, Lei Q, Lu A, He X, Okamoto N, Miyake N, Piccione J, Allen J, Matsumoto N, Pipan M, Krantz ID, Izumi K. Clinical and molecular spectrum of CHOPS syndrome. Am J Med Genet A. 2019 Jul;179(7):1126-1138. doi: 10.1002/ajmg.a.61174. Epub 2019 May 6. PubMed PMID: 31058441.

B. Peer-reviewed publications other (i.e. editorials, commentaries, etc.) – None

C. Non peer reviewed journal articles, reviews, editorials, etc. – None

D. Book Chapters
Kristina **Cusmano**-Ozog and Kimberly Chapman. Metabolic Acidosis in the Newborn. In: Stevenson, David K, Cohen, Ronald S, Sunshine, Philip. Neonatology Clinical Practice and Procedures. New York, NY: McGraw-Hill; 2015: 907-924.

E. Books – None

F. Special materials (editorials, letters, epitomes)
1. D'Aco K, Mooney R, **Cusmano**-Ozog K, Hofherr S, Lichter-Konecki U. What is in the can? The dilemma with dietary supplements. Molecular genetics and metabolism. 2014; 113(4):239-40. PMID: 25453401

2. Ayyub OB, Hofherr S, **Cusmano**-Ozog K, Ah Mew N. Time-dependent negative bias in plasma ammonia samples in a clinical setting. Clinica chimica acta; international journal of clinical chemistry. 2017; 471:126-127. PMID: 28595139

G. Digital publications (software, online writings, and other scholarly digital creations) – None

H. Abstracts not published in other forms
1. **Cusmano** KP, Kousseff BG. Congenital diaphragmatic hernia in genetics. Abstract #787. Presented at the 49th Annual Meeting of The American Society of Human Genetics (October 19-23, 1999; San Francisco, California).

2. **Cusmano**-Ozog K, Chen K, Kwan A, Hudgins L. Brachydactyly A1: Is it a single condition? J Invest Med. 2006;54:S137.

3. **Cusmano**-Ozog K, Rutledge SL, Boneh A, Gottesman G, Tuchman M, Harding C, Pickler L, Van Hove JL, Enns GM. Severe liver disease and urea cycle disorders. Abstract #1335. Presented at the 56th Annual Meeting of The American Society of Human Genetics (October 9-13, 2006; New Orleans, Louisiana).

4. **Cusmano**-Ozog K, Hoyme E, Hudgins L. Asymmetric Crying Facies: A common facial malformation of heterogeneous etiology. Proceedings of the Greenwood Genetic Center. Vol 26, p. 82-83, 2007.

5. **Cusmano**-Ozog K, Lorey F, Levine S, Martin M, Nicholas E, Packman S, Rosenblatt DS, Cowan TM, Enns GM. Cobalamin C disease and expanded newborn screening: the California experience. J Invest Med. 2007;55:S90.

6. **Cusmano**-Ozog K, Martin M, Nicholas E, Packman S, Rosenblatt DS, Cowan TM, Enns GM. Early identification and treatment of cobalamin C disease. Mol Genet Metab. 2007;90:249.

7. **Cusmano**-Ozog K, Sweet VT, Enns GM. D409H homozygosity and the cardiovascular form of Gaucher disease: A case report. Abstract #1445. Presented at the 57th Annual Meeting of The American Society of Human Genetics (October 23-27, 2007; San Diego, California).

8. **Cusmano**-Ozog K, Lorey F, Levine S, Martin M, Nicholas E, Packman S, Rosenblatt DS, Cederbaum SD, Cowan TM, Enns GM. Cobalamin C disease and expanded newborn screening: the California experience. Mol Genet Metab. 2007;90:240.

9. Manning M, **Cusmano** K, Enns G. 4q34>ter: Possible critical region for tracheal development. Proceedings of the Greenwood Genetics Center. Vol. 27, p. 68-69, 2008.

10. **Cusmano**-Ozog K, Waterson JR, Enns GM, Cowan TM. Neonatal Presentation of Glutaric Aciduria Type I. J Invest Med. 2008;56:196.

11. **Cusmano**-Ozog K, Lorey F, Feuchtbaum L, Sweet VT, Cowan TM, Enns GM. Outcomes among newborns with abnormal C5-OH on expanded newborn screening in California. Mol Genet Metab. 2008;93:233.

12. **Cusmano**-Ozog K, Lorey F, Kazerouni N, Roberson M, Cowan TM. Confirmatory Testing Outcomes Following Newborn Screening for Biotinidase Deficiency. Abstract #605. Presented at the 58th Annual Meeting of The American Society of Human Genetics (November 11-15, 2008; Philadelphia, Pennsylvania).

13. **Cusmano**-Ozog K, Kwan A, Enns GM, Cowan TM. Ethylmalonic Encephalopathy: Expanding the Phenotype. J Invest Med. 2009;57:115.

14. **Cusmano**-Ozog K, Enns GM, Cowan TM. Riboflavin-responsive multiple acyl-CoA dehydrogenase deficiency with atypical acylcarnitines and normal in vitro enzyme activity. Abstract #92. Presented at the 2009 American College of Medical Genetics Annual Clinical Genetics Meeting (March 25-29, 2009; Tampa, Florida).

15. **Cusmano**-Ozog K, Abdenur JA, Barshop BA, Cederbaum S, Lorey F, Packman S, Powell B, Waterson J, Cowan TM, Enns GM. Outcomes in cobalamin C disease identified by expanded newborn screening in California. Mol Genet Metab. 2009;98:121.

16. **Cusmano**-Ozog K, Le A, Enns GM, Cowan TM. A new tandem mass spectrometry (MS/MS) method for detecting Pompe disease in dried blood spots. Abstract #3033. Presented at the 59th Annual Meeting of The American Society of Human Genetics (October 20-24, 2009; Honolulu, Hawaii).

17. **Cusmano**-Ozog K, Moore T, Cowan TM, Enns GM. Glutathione Deficiency In Methylmalonic Acidemia Is Responsive To Treatment With N-Acetylcysteine. J Invest Med. 2010;58:124.

18. **Cusmano**-Ozog K, Moore T, Niemi A-K, Zadeh N, Cowan TM, Enns GM. Evidence Of Redox Imbalance In A Patient With Methylmalonic Acidemia (mut0). Mol Genet Metab. 2010;99:210-211.

19. C Nenon, K **Cusmano**-Ozog, N AhMew. Clinical Utility of Alloisoleucine in Maple Syrup Urine Disease. Abstract #218. Presented at the 2013 American College of Medical Genetics Annual Clinical Genetics Meeting (March 19- 23, 2013; Phoenix, Arizona).

20. EL MacLeod and KP **Cusmano**-Ozog. Dramatic Effect Of High Performance Activity On Phenylalanine Levels In An Adolescent Female With Phenylketonuria. 12th International Congress of Inborn Errors of Metabolism. Journal of Inherited Metabolic Disease September 2013, Volume 36, Issue 2 Supplement, pp 343-360

21. SE Hofherr and KP **Cusmano**-Ozog. Development Of A GC-MS Sim/Scan Method For Organic Acid Analysis. 12th International Congress of Inborn Errors of Metabolism. Journal of Inherited Metabolic Disease September 2013, Volume 36, Issue 2 Supplement, pp 343-360

22. DS Regier, NA Banks, EL Leon, U Lichter-Konecki, KP **Cusmano**-Ozog. Mevalonate Kinase Deficiency Presenting As Neonatal Liver Failure. 12th International Congress of Inborn Errors of Metabolism. Journal of Inherited Metabolic Disease September 2013, Volume 36, Issue 2 Supplement, pp 343-360

23. K. **Cusmano**-Ozog, P. Tanpaiboon, L. Harris, J. Turner, L. Kehoe, T. Biagi, B. Lanpher. Atypical Rett Syndrome: Is it really more common in females? Presented at the 63rd Annual Meeting of The American Society of Human Genetics (October, 2013; Boston, MA).

24. Kristina **Cusmano**-Ozog, Sean Hofherr, Tina Cowan, Debra Regier. Portable Learning Solutions to Support Biochemical Genetics Trainees: a Model for Training in Small Graduate Subspecialty Fields. Abstract 458 Presented at the 2015 American College of Medical Genetics Annual Clinical Genetics Meeting (March 24-28, 2015; Salt Lake City, Utah).

25. Kristina **Cusmano**-Ozog, Michael Sheppard, Sean Hofherr. Detection Rate of Chromosomal Microarray: Our first year of experience at Children's National Health Systems Abstract 205 Presented at the 2015 American College of Medical Genetics Annual Clinical Genetics Meeting (March 24-28, 2015; Salt Lake City, Utah).

26. Sean Hofherr, Mary Beth Seprish, Julien Weinstein, Veronique Weinstein, Kristina **Cusmano**-Ozog. Utility of Chromosomal Microarray Analysis for Autism: A One Year Retrospective Analysis of Testing in Patients with Autism at Children's National Health System. Abstract 197 Presented at the 2015 American College of Medical Genetics Annual Clinical Genetics Meeting (March 24-28, 2015; Salt Lake City, Utah).

27. C. Ferreira, J. Devaney, S. Hoffher, Laura Pollard, K. **Cusmano**-Ozog. Hereditary Fructose Intolerance Mimicking A Biochemical Phenotype Of Mucolipidosis. Molecular Genetics and Metabolism 114 (2015) 337

28. K. **Cusmano**-Ozog; M. Seprish; S. Hofherr. Our experience at Children's National Health Systems using CytoScan® Dx Assay. Presented at the Annual Meeting of The American Society of Human Genetics (October 6-10, 2015; Baltimore, MD).

29. K. **Cusmano**-Ozog; M. Seprish; S. Hofherr. Using Areas of Homozygosity identified by Chromosomal Microarray to leverage Next Generation Sequencing: The Children's National Health System Experience. Abstract 519 Presented at the 2016 American College of Medical Genetics Annual Clinical Genetics Meeting (March 8-12, 2016; Tampa, FL).

30. B. Comstock, M. Seprish; S. Hofherr, K. **Cusmano**-Ozog. Chromosomal Microarray Results From a Large Pediatric Clinical Setting. Abstract 266 Presented at the 2016 American College of Medical Genetics Annual Clinical Genetics Meeting (March 8-12, 2016; Tampa, FL).

31. Kristina **Cusmano**-Ozog, S. Berger, J. Fraser, K. Chapman, D. Regier, B. Meltzer, J. Devaney. MTHFR common variants are NOT associated with autism in our patient population. Abstract 390

Presented at the 2017 American College of Medical Genetics Annual Clinical Genetics Meeting (March 21-25, 2017; Phoenix, AZ).

32. Kristina **Cusmano**-Ozog, C. Ferreira. Abnormal amino acid pattern in a patient with COQ9 mutations. Abstract 112 Presented at the 2017 American College of Medical Genetics Annual Clinical Genetics Meeting (March 21-25, 2017; Phoenix, AZ).

33. Charles Billington; C. Ferreira; E. MacLeod; N. Ah Mew; K. **Cusmano**-Ozog. A Tale of Two Variants: Salvadoran founder mutations in DBT cause classical MSUD. Abstract 79 Presented at the 2018 American College of Medical Genetics Annual Clinical Genetics Meeting (April 10-14, 2018; Charlotte, NC).

34. Debra Regier, Jessica Albert, Nancy Cheng, Kristina **Cusmano**-Ozog. Revising the Biochemical Genetics Laboratory Rotation. Abstract Presented at the 2019 Association of Prefessors of Human and Medical Genetics (APHMG) Annual Workshop and Special Interest Groups Meeting (April 30-May 03, 2019; Clearwater Beach, FL).

VI. Editorial Service

A. Editorial board memberships – None

B. Other peer review activities
Ad Hoc Reviewer, Prenatal Diagnosis, 2008
Ad Hoc Reviewer, Journal of Pediatric Genetics, 2010
Ad Hoc Reviewer, Molecular Genetics and Genomic Medicine, 2014 - 2015

VII. Grants (Include role (eg PI), granting agency, type (eg K-08, R-01), and term)

A. Current Funding – None

B. Pending Funding – None

C. Prior Funding - None

VIII. Service as Grant Review

Grant reviewer, US Food and Drug Administration, 2016 - 2018

IX. **Patents** - None

X. University Administrative Service

A. Committee service
Fellowship Executive Committee, National Human Genome Research Institute Training Program, Bethesda, MD, 2012 - 2014
Clinical Competency Committee, National Human Genome Research Institute Training Program, Bethesda, MD, 2015 - Present
Pediatric Residency Program Intern Selection Committee, Children's National Medical Center, Washington, DC, 2013 - 2018
Laboratory Medicine Quality Assurance Committee, Children's National Medical Center, Washington, DC, 2016 - Present
Graduate Medical Education (GME) Committee, Children's National Medical Center, Washington, DC, 2016 - Present
Program Director, Medical Genetics Residency Program, Children's National Medical Center, Washington, DC, July 2016 – Present

B. Leadership roles – None

XI. Service to Professional Organizations

A. Membership
Hillsborough County Medical Association 1998 - 2005
Florida Medical Association 1998 - 2002
American Academy of Pediatrics 2001 - 2008
American Society of Human Genetics 2006 - Present
Society for Inherited Metabolic Disorders 2007 - Present
American Medical Association 1998-2002, 2012 - Present
American College of Medical Genetics 2012 – Present

B. Committee service
Member, Lab Quality Assurance Committee, Biochemical Genetics Subcommittee, American College of Medical Genetics and Genomics, 2013 - 2017
Member, Program Committee, American College of Medical Genetics and Genomics, 2018 - Present
Member, Medical Genetics and Genomics Milestone Working Group, Accreditation Council for Graduate Medical Education, 2018
Member, Clinical Biochemical Genetics Milestone Working Group, Accreditation Council for Graduate Medical Education, 2019
Biochemical and Molecular Genetics Committee, College of American Pathologists (CAP)/American College of Medical Genetics and Genomics (ACMG), 2019

C. Leadership roles – None

D. Other
ABMGG
Item Writer, Biochemical Genetics, American Board of Medical Genetics, 2014 - 2017
Item Writer, Maintenance of Certification Part 2 Module, Biochemical Genetics, American Board of Medical Genetics, 2014
Clinical/Medical Biochemical Genetics Certification Examination Content Based Standard Setting Exercise, American Board of Medical Genetics and National Board of Medical Examiners, 2017
General Certification Item Writer, American Board of Medical Genetics and Genomics, 2018
ASHG
Judge, DNA Day Essay Contest, American Society of Human Genetics, 2011 - Present
Lecturer & Group Facilitator, High School Workshop, American Society of Human Genetics, 2009 - 2015
Platform Session Moderator (Mechanisms and Treatment of Metabolic Disease), 60th Annual Meeting of The American Society of Human Genetics, Washington, DC, 2010
Platform Session Moderator (Inborn Errors of Metabolism: Novel Disorders, Models, and Observations), 65th Annual Meeting of The American Society of Human Genetics, Baltimore, MD, 2015

XII. Presentations

A. National and Regional Meetings

National
1. Asymmetric Crying Facies: A Common Facial Malformation of Heterogeneous Etiology. 27th Annual David W. Smith Workshop on Morphogenesis and Malformations, Lake Arrowhead, CA, September 2006

2. Cobalamin C Disease and Expanded Newborn Screening: The California Experience. Society for Inherited Metabolic Disorders Meeting, Nashville, TN, March 2007
3. Outcomes Among Newborns with Abnormal C5-OH on Expanded Newborn Screening in California. Society for Inherited Metabolic Disorders Meeting, Asilomar, CA, March 2008
4. Atypical Rett Syndrome: Is it really more common in females? 63rd American Society of Human Genetics Annual Meeting, Boston, MA, October 2013
5. Impact on the clinic: CytoScan Dx the first FDA-cleared microarray test. American College of Medical Genetics Annual Meeting Exhibit Theater, Salt Lake City, Utah, March 2015
6. Kynureninase deficiency – an under recognized malformation syndrome. American College of Medical Genetics Annual Meeting, Charlotte, NC, April 2018
7. QI Project: Enhancing Situational Awareness for Inborn Errors of Metabolism. Association of Professors of Human and Medical Genetics, Santa Fe, NM, May 2018
8. Exon-level array: Bridging the gap between chromosomal microarray and next-gen sequencing. American Cytogenomics Conference, Snow Bird, UT, June 2018
9. Using Quality Improvement Tools to Enhance Patient Care and Training in Genetics. Putting the PD into PDSA: Implementing a Meaningful and Publishable QI Project in a Training Program. American College of Medical Genetics Annual Meeting, Seattle, WA, April 2019
10. Serving an Entire Patient Population with Genetic Testing: How National Children's Hospital Supports Precision Medicine Across all Clinical Service Lines, Precision Medicine Institute Symposium, New Orleans, LA, May 2019

Regional
1. The Mucopolysaccharidoses, a review. University of South Florida Pediatrics Case Conference, Tampa, FL, January 2004
2. The Genetic Causes of Hypotonia. University of South Florida Pediatrics Case Conference, Tampa, FL, August 2004
3. Velocardiofacial syndrome; case report and review of the literature. University of South Florida Pediatrics Case Conference, Tampa, FL, January 2005
4. Noonan syndrome; case report and review of the literature. University of South Florida Pediatrics Case Conference, Tampa, FL, March 2005
5. Genetics and Metabolism Board Review. University of South Florida, Pediatrics Board Review Course, Tampa, FL, May 2005
6. Double Trisomy: Is it double trouble? Stanford University Medical Genetics Grand Rounds, Stanford, CA, November 2005
7. Brachydactyly A1: Is it a single condition? Western Society of Pediatric Research Meeting, Carmel-by-the-Sea, CA, February 2006
8. MMACHC and Cobalamin C disease. Stanford University Human Genetics Journal Club, March 2006
9. Ethnicity and MMA MUT mutations - Is there a correlation? Stanford University Human Genetics Journal Club, Stanford, CA, December 2006
10. Cobalamin C Disease and Expanded Newborn Screening: The Northern California Experience. Western Society of Pediatric Research Meeting, Carmel-by-the-Sea, CA, February 2007
11. Retinitis Pigmentosa and Genetics, a review. Stanford University Medical Genetics Grand Rounds, Stanford, CA, March 2007
12. Could a Spoonful of Sugar Treat Gaucher Disease? A Closer Look at the Iminosugar, Isofagomine. Stanford University Human Genetics Journal Club, Stanford, CA, December, 2007
13. Neonatal Presentation of Glutaric Aciduria Type I. Western Society of Pediatric Research Meeting, Carmel-by-the-Sea, CA, February 2008
14. MMADHC and Cobalamin D disease. Stanford University Human Genetics Journal Club, Stanford, CA, November 2008
15. Lessons Learned from Newborn Screening. Stanford University Medical Genetics Grand Rounds, Stanford, CA, June 2008
16. Ethylmalonic Encephalopathy: Expanding the Phenotype. Western Society of Pediatric Research Meeting, Carmel-by-the-Sea, CA, February 2009

17. The Genetic Causes of Fetal Hydrops. Stanford University Medical Genetics Grand Rounds, Stanford, CA, November 2009
18. Lysosomal Storage Diseases: A review. Stanford University Medical Genetics Grand Rounds, Stanford, CA, June 2009
19. Glutathione Deficiency in MMA is Responsive to Treatment with NAC. Western Society of Pediatric Research Meeting, Carmel-by-the-Sea, CA, February 2010
20. Prenatal Diagnosis and Management of Ornithine Transcarbamylase (OTC) Deficiency: Survival Beyond Liver Transplantation. 11th Annual Northern California Clinical Genetics Exchange, Stanford, CA, May 2010
21. Of Mice and MMA - Tell me about the pathophysiology. Stanford University Human Genetics Journal Club, Stanford, CA, June 2010
22. What is Molecular Genetics & Molecular Diagnostics: Diagnosing Diseases in the 21st Century - Diagnosis, Prognosis & Therapeutics. Children's National Medical Center, Summer Student Lecture Series, Washington, DC, July 2014
23. Inborn Errors of Metabolism and the NICU, NICU Nursing staff, Children's National Medical Center, Washington, DC, July 2014
24. TARP syndrome; Expanding the Phenotype. National Human Genome Research Institute Case Conference, Bethesda, MD, September 2014
25. Molecular Assay - Fragile X. National Human Genome Research Institute Sign Out Conference, Bethesda, MD, October 2014
26. PHGDH: A gene with more than one phenotype. National Human Genome Research Institute Journal Club, Bethesda, MD, February 2015
27. Molecular Medicine in the 21st Century: Applications of Genetic Testing. Children's National Medical Center, Student Lecture Series, Washington, DC, June 2015
28. Inborn Errors of Metabolism Amenable to Liver Transplant. Transplant Academic Conference MedStar Georgetown University Hospital, Washington, DC, February 2015
29. Linear Sebaceous Nevus Syndrome and Post Zygotic KRAS Mutations. National Human Genome Research Institute Case Conference, Bethesda, MD, June 2015
30. Newborn Metabolic Screening...So Much More Than "The PKU Test." Georgetown Pediatrics Grand Rounds, Washington, DC, June 2015
31. Molecular Diagnostics for Genetic Diseases, Children's National Future of Pediatrics, Bethesda, MD, June 2016
32. A 5mo with poor weight gain. Children's National Professorial Rounds, Washington, DC, July 2016
33. Let's get personal: a phenotype-driven approach to sequencing. The Foundation for Genetic Technology Southeastern Regional Genetics Meeting, Chantilly, VA, September 2017

B. International Meetings – None

C. Visiting Professorships – None

XI. **Community Service**

Faith Children's Home, Volunteer, Tampa, FL, 1991 - 1995
Physical Therapy Department, Volunteer, St. Joseph's Hospital, Tampa, FL, 1996
Camillus House Soup Kitchen, Volunteer, Miami, FL, 1995 - 1998
Krewe of Agustina de Aragon, Member, Tampa, FL, 1999 - 2005
Florida Diabetes Camp, Counselor, Winona, FL, 2001
Camp Huber, Maryland Alliance of PKU Families, Chestertown and Westminster, MD, 2013 - 2016
Special Olympics DC MedFest, Volunteer, Washington, DC, 2016 - 2018