DUKE UNIVERSITY MEDICAL CENTER

CURRICULUM VITAE

for
Permanent Record
and the
Appointments and Promotions Committee

July 2, 2024

Name:	Priya S. Kishnani, MD
Primary Academic Appointment:	Professor of Pediatrics
Primary Academic Department (not DUAP):	Department of Pediatrics Division of Medical Genetics
Secondary Appointment:	Professor of Molecular Genetics and Microbiology
Present Academic Rank and Title:	C.L. and Su Chen Professor of Pediatrics Professor of Molecular Genetics and Microbiology
Date and Rank of First Duke Faculty Appointment:	Associate in Pediatrics, July 1995
Specialty Certification(s) and Dates:	American Board of Pediatrics, 1992 American Board of Pediatrics, Recertification 1999, not recertified since American Board of Medical Clinical Genetics, 1996, recertified in January 2020 American Board of Clinical Biochemical Genetics, 1996, recertified in January 2020
Place:	Bombay, India
Citizen of:	US Citizen
Visa Status:	NA

Education	**Institution**	**Date (Year)**	**Degree**
High School	Convent of Jesus and Mary, Bombay, India	1978	Diploma
Jai Hind College	Bombay, India	1980	Diploma

Education	Institution	Date (Year)	Degree
Bachelor of Medicine and Bachelor of Surgery	Bombay University – Bombay, India	10/1985	M.B.B.S.
Diploma in Child Health – Pediatrics	College of Physicians and Surgeons of Bombay – Bombay, India	4/1989	D.C.H.
Pediatrics	Bombay University - Bombay, India	3/1990	M.D.
PL-III Resident	Duke University Medical Center	1991-1992	M.D.
Clinical Fellowship Pediatric Genetics & Metabolism	Duke University Medical Center	1992-1994	
Research Fellowship Pediatric Genetics & Metabolism	Duke University Medical Center	1994-1995	

Professional Training and Academic Career:

Institution	Position/Title	Dates
B.Y.L. Nair Charitable Hospital, Bombay, India	Internship	1986-1987
B.Y.L. Nair Charitable Hospital, Bombay, India	House Officer (Pediatrics)	2/1987-7/1987
B.Y.L. Nair Charitable Hospital, Bombay, India	House Officer (Hematology)	8/1987-1/1988
B.Y.L. Nair Charitable Hospital, Bombay, India	House Officer (ICCU & Cardiology)	2/1988-7/1988
Children's Hospital of Philadelphia, Philadelphia, PA	Research Pediatrician	1990-1991
Duke University Medical Center, Durham, NC	PL-III Resident	1991-1992
Duke University Medical Center, Durham, NC	Clinical Fellowship, Pediatric Genetics & Metabolism	1992-1994
Duke University Medical Center, Durham, NC	Research Fellowship, Pediatrics Genetics & Metabolism	1994-1995

Institution	Position/Title	Dates
Duke University Medical Center, Durham, NC	Associate in Pediatrics	1995-1997
Duke University Medical Center, Durham, NC	Co-Director of Down Syndrome	1996-2021
Duke University Medical Center, Durham, NC	Assistant Professor of Pediatrics	1997-2002
Duke University Medical Center, Durham, NC	Director of Glycogen and Lysosomal Storage Disease Program	1997-Present
Duke University Medical Center, Durham, NC	Director of Biochemical Genetics Training Program	1997-2015
Duke University Medical Center, Durham, NC	Director of Metabolic Clinic	1998-Present
Duke University Medical Center, Durham, NC	Director of Clinical Trials	2002-Present
Duke University Medical Center, Durham, NC	Associate Professor of Pediatrics	2002-Sept. 2007
Duke University Medical Center, Durham, NC	Interim Chief, Division of Medical Genetics	2005-2006
Duke University Medical Center, Durham, NC	Chief, Division of Medical Genetics	Jan. 2007-Present
Duke University Medical Center, Durham, NC	Medical Director, YT and Alice Chen Pediatrics Genetics and Genomics Center	July 2007-Present
Duke University Medical Center, Durham, NC	Professor, Molecular Genetics and Microbiology Department	Feb. 2019-Present
National Organization for Rare Disorder, Danbury, CT	Director, Duke Health NORD Rare Disease Center for Excellence	Oct. 2021-Present

Publications:

Refereed Journals (https://pubmed.ncbi.nlm.nih.gov/?term=Kishnani+P)

1. Silber JH, Radcliffe J, Peckham V, Perilongo G, **Kishnani PS**, Fridman M, Goldwein JW, Meadows AT. Whole-brain irradiation and decline in intelligence: the influence of dose and age on IQ score. J Clin Oncol 1992 Sep; 10(9):1390-6. PMID: 1517781

2. **Kishnani P,** Iafolla AK, McConkie-Rosell A, Van Hove JL, Kanter RJ, Kahler SG. Hemangioma, supraumbilical midline raphe, and coarctation of the aorta with a right aortic arch: single casual entity? Am J Med Genet 1995 Oct 23; 59(1):44-8. PMID: 8849010
3. Van Hove JL, **Kishnani PS**, Muenzer J, Wenstrup RJ, Summar ML, Brummond MR, Lachiewicz AM, Millington DS, Kahler SG. Benzoate therapy and carnitine deficiency in non-ketotic hyperglycinemia. Am J Med Genet 1995 Dec 4; 59(4):444-53. PMID: 8585564
4. **Kishnani PS**, Bengur AR, Chen YT. Pulmonary hypertension in glycogen storage disease type I. J Inherit Metab Dis. 1996; 19(2):213-6. PMID: 8739968
5. McConkie-Rosell A, Wilson C, Piccoli DA, Boyle J, DeClue T, **Kishnani PS**, Shen JJ, Boney A, Brown B, Chen YT. Clinical and laboratory findings in four patients with the non-progressive hepatic form of type IV glycogen storage disease. J Inherit Metab Dis. 1996; 19(1):51-8. PMID: 8830177
6. Bao Y, **Kishnani PS**, Wu JY, Chen YT. Hepatic and neuromuscular forms of glycogen storage disease type IV caused by mutations in the same glycogen-branching enzyme gene. J Clin Invest. 1996 Feb 15; 97(4):941-8. PMID: 8613547
7. Wang M, **Kishnani PS**, Decker-Phillips M, Kahler SG, Chen YT, Godfrey M. Double mutant fibrillin-1 (FBN1) allele in a patient with neonatal Marfan syndrome. J Med Genet. 1996 Sep; 33(9):760-3. PMID: 8880577
8. **Kishnani PS**, Van Hove JL, Shoffner JS, Kaufman A, Bossen EH, Kahler SG. Acute pancreatitis in an infant with lactic acidosis and a mutation at nucleotide 3243 in the mitochondrial DNA tRNALeu(UUR) gene. Eur J Pediatr. 1996 Oct; 155(10):898-903. PMID: 8891562
9. **Kishnani PS**, Bao Y, Wu JY, Brix AE, Lin JL, Chen YT. Isolation and nucleotide sequence of canine glucose-6-phosphatase mRNA: identification of mutation in puppies with glycogen storage disease type Ia. Biochem Mol Med. 1997 Aug; 61(2):168-77. PMID: 9259982
10. **Kishnani PS**, Sullivan JA, Walter BK, Spiridigliozzi GA, Doraiswamy PM, Krishnan KR. Cholinergic therapy for Down's syndrome. Lancet. 1999 Mar 27; 353(9158):1064-5. PMID: 10199357
11. **Kishnani PS**, Boney A, Chen YT. Nutritional deficiencies in a patient with glycogen storage disease type Ib. J Inherit Metab Dis. 1999 Oct; 22(7):795-801. PMID: 10518279
12. Ahmad A, Amalfitano A, Chen YT, **Kishnani PS**, Miller C, Kelley R. Dubowitz syndrome: a defect in the cholesterol biosynthetic pathway? Am J Med Genet. 1999 Oct 29; 86(5):503-4. PMID: 10508998
13. Ahmad A, Kahler SG, **Kishnani PS**, Artigas-Lopez M, Pappu AS, Steiner R, Millington DS, Van Hove JL. Treatment of pyruvate carboxylase deficiency with high doses of citrate and aspartate. Am J Med Genet. 1999 Dec 3; 87(4):331-8. PMID: 10588840
14. Shaiu WL, **Kishnani PS**, Shen J, Liu HM, Chen YT. Genotype-phenotype correlation in two frequent mutations and mutation update in type III glycogen storage disease. Mol Genet Metab. 2000 Jan; 69(1):16-23. PMID: 10655153
15. Van Hove JL, **Kishnani PS**, Demaerel P, Kahler SG, Miller C, Jaeken J, Rutledge SL. Acute hydrocephalus in nonketotic hyperglycemia. Neurology. 2000 Feb 8; 54(3):754-6. PMID: 10680820

16. **Kishnani PS**, Spiridigliozzi GA, Heller JH, Sullivan JA, Doraiswamy PM, Krishnan KR. Donepezil for Down's syndrome. Am J Psychiatry. 2001 Jan; 158(1):143. PMID: 11136652
17. **Kishnani PS**, Faulkner E, VanCamp S, Jackson M, Brown T, Boney A, Koeberl D, Chen YT. Canine model and genomic structural organization of glycogen storage disease type Ia (GSD Ia). Vet Pathol. 2001 Jan; 38(1):83-91. PMID: 11199168
18. Amalfitano A, Bengur AR, Morse RP, Majure JM, Case LE, Veerling DL, Mackey J, **Kishnani PS**, Smith W, McVie-Wylie A, Sullivan JA, Hoganson GE, Phillips JA 3rd, Schaefer GB, Charrow J, Ware RE, Bossen EH, Chen YT. Recombinant human acid alpha-glucosidase enzyme therapy for infantile glycogen storage disease type II: results of a phase I/II clinical trial. Genet Med. 2001 Mar-Apr; 3(2):132-8. PMID: 11286229
19. Mackey J, Treem WR, Worley G, Boney A, Hart P, **Kishnani PS**. Frequency of celiac disease in individuals with Down syndrome in the United States. Clin Pediatr (Phila). 2001 May; 40(5):249-52. PMID: 11388673
20. Beaty RM, Jackson M, Peterson D, Bird A, Brown T, Benjamin DK Jr, Juopperi T, **Kishnani PS**, Boney A, Chen YT, Koeberl DD. Delivery of glucose-6-phosphatase in a canine model for glycogen storage disease, type Ia, with adeno-associated virus (AAV) vectors. Gene Ther. 2002 Aug; 9(15):1015-22. PMID: 12101432
21. Snyder MJ, Bradford WD, **Kishnani PS**, Hale LP. Idiopathic hyperammonemia following an unrelated cord blood transplant for mucopolysaccharidoses I. Pediatr Dev Pathol. 2003 Jan-Feb; 6(1):78-83. Epub 2002 Dec 17. PMID: 12481230
22. Koeberl DD, Young SP, Gregersen NS, Vockley J, Smith WE, Benjamin DK Jr, An Y, Weavil SD, Chaing SH, Bali D, McDonald MT, **Kishnani PS**, Chen YT, Millington DS. Rare disorders of metabolism with elevated butyryl- and isobutyryl-carnitine detected by tandem mass spectrometry newborn screening. Pediatr Res. 2003 Aug; 54(2):219-23. Epub 2003 May 7. PMID: 12736383
23. Heller, JH, Spiridigliozzi GA, Sullivan JA, Doraiswamy PM, Krishnan RR, **Kishnani PS**. Donepezil for the treatment of language deficits in adults with Down syndrome: a preliminary 24-week open trial. Am J Med Genet A. 2003 Jan 15; 116A (2):111-6. PMID: 12494428
24. Koeberl DD, Millington DS, Smith WE, Weavil SD, Muenzer J, McCandless SE, **Kishnani PS**, McDonald MT, Chaing S, Boney A, Moore E, Frazier DM. Evaluation of 3-methylcrotonyl-CoA carboxylase deficiency detected by tandem mass spectrometry newborn screening. J Inherit Metab Dis. 2003; 26(1):25-35. PMID: 12872837
25. Hanna R, McDonald MT, Sullivan JA, Mackey JF, Krishnamurthy V, **Kishnani PS**. Diagnostic and treatment challenges of neuropathic Gaucher disease: two cases with an intermediate phenotype. J Inherit Metab Dis. 2004; 27(5):687-90. PMID: 15669686
26. Hardy O, Worley G, Lee MM, Chaing S, Mackey J, Crissman B, **Kishnani PS**. Hypothyroidism in Down syndrome: screening guidelines and testing methodology. Am J Med Genet A. 2004 Feb 1; 124A (4):436-7. PMID: 14735598

27. **Kishnani PS**, Sullivan JA, Spiridigliozzi GA, Heller JH, Crissman BG. Donepezil use in Down syndrome. Arch Neurol. 2004 Apr; 61(4):605-6. PMID: 15096417
28. **Kishnani PS**, Howell RR. Pompe disease in infants and children. J Pediatr. 2004 May; 144(5 Suppl):S35-43. PMID: 15126982
29. Ing RJ, Cook DR, Bengur RA, Williams EA, Eck J, Dear Gde L, Ross AK, Kern FH, **Kishnani PS**. Anesthetic management of infants with glycogen storage disease type II: a physiological approach. Paediatr Anaesth. 2004 Jun; 14(6):514-9. PMID: 15153218
30. Worley G, Shbarou R, Heffner AN, Belsito KM, Capone GT, **Kishnani PS**. New onset focal weakness in children with Down syndrome. Am J Med Genet A. 2004 Jul 1; 128A (1):15-8. PMID: 15211649
31. Hunley TE, Corzo D, Dudek M, **Kishnani PS**, Amalfitano A, Chen YT, Richards SM, Phillips JA 3rd, Fogo AB, Tiller GE. Nephrotic syndrome complicating alpha-glucosidase replacement therapy for Pompe disease. Pediatrics. 2004 Oct; 114(4):e532-5. PMID: 15466083
32. Heller JH, Spiridigliozzi GA, Doraiswamy PM, Sullivan JA, Crissman BG, **Kishnani PS**. Donepezil effects on language in children with Down syndrome: result of the first 22-week pilot clinical trial. Am J Med Genet A. 2004 Oct 15; 130A (3):325-6. PMID: 15378553
33. Franco LM, Krishnamurthy V, Bali D, Weinstein DA, Arn P, Clary B, Boney A, Sullivan J, Frush DP, Chen YT, **Kishnani PS**. Hepatocellular carcinoma in glycogen storage disease type Ia: a case series. J Inherit Metab Dis. 2005; 28(2):153-62. PMID: 15877204
34. Van Hove JL, Vande Kerckhove K, Hennermann JB, Mahieu V, Declercq P, Mertens S, De Becker M, **Kishnani PS**, Jaeken J. Benzoate treatment and the glycine index in nonketotic hyperglycinemia. J Inherit Metab Dis. 2005; 28(5):651-63. PMID: 16151895
35. Goker-Alpan O, Hruska KS, Orvisky E, **Kishnani PS**, Stubblefield BK, Schiffmann R, Sidransky E. Divergent phenotypes in Gaucher disease implicate the role of modifiers. J Med Genet. 2005 Jun; 42(6):e37. PMID: 15937077
36. An Y, Young SP, **Kishnani PS**, Millington DS, Amalfitano A, Corz D, Chen YT. Glucose tetrasaccharide as a biomarker for monitoring the therapeutic response to enzyme replacement therapy for Pompe disease. Mol Genet Metab. 2005 Aug; 85(4):247-54. PMID: 15886040
37. Quigley DI, McDonald MT, Krishnamuthy V, **Kishnani PS**, Lee MM, Haqq AM, Goodman BK. Triploid mosaicism in a 45,X/69,XXY infant. Am J Med Genet A. 2005 Oct 1; 138A (2):171-4. PMID: 16152633
38. Smith W, **Kishnani PS**, Lee B, Singh RH, Rhead WJ, Sniderman King L, Smith M, Summar M. Urea cycle disorders: clinical presentation outside the newborn period. Crit Care Clin. 2005 Oct; 21(4 Suppl):S9-17. Review. PMID: 16227115
39. **Kishnani PS**, Steiner RD, Bali D, Berger K, Byrne BJ, Case LE, Crowley JF, Downs S, Howell RR, Kravitz RM, Mackey J, Marsden D, Martins AM, Millington DS, Nicolino M, O'Grady G, Patterson MC, Rapoport DM, Slonim A, Spencer CT, Tiff CJ, Watson MS. Pompe disease diagnosis and management guideline. Genet Med. 2006 May; 8(5):267-88. Erratum in: Genet Med. 2006 Jun;

8(6):382. ACMG Work Group of Management of Pompe Disease (removed); Case, Laura (corrected to Case, Laura E). PMID: 16702877

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41. Ansong AK, Li JS, Nozik-Grayck E, Ing R, Kravitz RM, Idriss SF, Kanter RJ, Rice H, Chen YT, **Kishnani PS**. Electrocardiographic response to enzyme replacement therapy for Pompe disease. Genet Med. 2006 May; 8(5):297-301. PMID: 16702879
42. Zhang H, Kallwass H, Young SP, Carr C, Dai J, **Kishnani PS**, Millington DS, Keutzer J, Chen YT, Bali D. Comparison of maltose and acarbose as inhibitors of maltase-glucoamylase activity in assaying acid alpha-glucosidase activity in dried blood spots for the diagnosis of infantile Pompe disease. Genet Med. 2006 May; 8(5):302-6. PMID: 16702880
43. Jack RM, Gordon C, Scott CR, **Kishnani PS**, Bali D. The use of acarbose inhibition in the measurement of acid alpha-glucosidase activity in blood lymphocytes for the diagnosis of Pompe disease. Genet Med. 2006 May; 8(5):307-12. PMID: 16702881
44. Cook AL, **Kishnani PS**, Carbonic MP, Kanter RJ, Chen YT, Ansong AK, Kravitz RM, Rice H, Li JS. Ambulatory electrocardiogram analysis in infants treated with recombinant human acid alpha-glucosidase enzyme replacement therapy for Pompe disease. Genet Med. 2006 May; 8(5):313-7. PMID: 16702882
45. Case LE, **Kishnani PS**. Physical therapy management of Pompe disease. Genet Med. 2006 May; 8(5):318-27. Review. PMID: 16702883
46. **Kishnani PS**, Hwu WL, Mandel H, Nicolino M, Yong F, Corzo D; Infantile-Onset Pompe Disease Natural History Study Group. A retrospective, multinational, multicenter study on the natural history of infantile-onset Pompe disease. J Pediatr. 2006 May; 148(5):671-676. PMID: 16737883
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49. Crissman BG, Worley G, Roizen N, **Kishnani PS**. Current perspective on Down syndrome: selected medical and social issues. Am J Med Genet C Semin Med Genet. 2006 Aug 15; 142C (3):127-30. Review. PMID: 17048353
50. Dixon N, **Kishnani PS**, Zimmerman S. Clinical manifestations of hematologic and oncologic disorders in patients with Down syndrome. Am J Med Genet C Semin Med Genet. 2006 Aug 15; 142C (3):149-57. Review. PMID: 17048354

51. Thurberg BL, Lynch Maloney C, Vaccaro C, Afonso K, Tsai AC, Bossen E, **Kishnani PS**, O'Callaghan M. Characterization of pre- and post-treatment pathology after enzyme replacement therapy for Pompe disease. Lab Invest. 2006 Dec; 86(12):1208-20. Epub 2006 Oct 30. PMID: 17075580
52. Heller JH, Spiridigliozzi GA, Crissman BG, Sullivan JA, Eells RL, Li JS, Doraiswamy PM, Krishnan KR, **Kishnani PS**. Safety and efficacy of rivastigmine in adolescents with Down syndrome: a preliminary 20-week, open-label study. J Child Adolesc Psychopharmacol. 2006 Dec; 16(6):755-65. PMID: 17201619
53. **Kishnani PS**, Corzo D, Nicolino M, Byrne B, Mandel H, Hwu WL, Leslie N, Levine J, Spencer C, McDonald M, Li J, Dumontier J, Halberthal M, Chien YH, Hopkin R, Vijayaraghavan S, Gruskin D, Bartholomew D, van der Ploeg A, Clancy JP, Parini R, Morin G, Beck M, De la Gastine GS, Jokic M, Thurberg B, Richards S, Bali D, Davison M, Worden MA, Chen YT, Wraith JE. Recombinant human acid [alpha]-glucosidase: major clinical benefits in infantile-onset Pompe disease. Neurology. 2007 Jan 9; 68(2):99-109. Epub 2006 Dec 6. Erratum in: Neurology. 2008 Nov 18; 71(21):1748. PMID: 17151339
54. Demo E, Frush D, Gottfried M, Koepke J, Boney A, Bali D, Chen YT, **Kishnani PS**. Glycogen storage disease type III-hepatocellular carcinoma a long-term complication? J Hepatol. 2007 Mar; 46(3):492-8. Epub 2006 Nov 9. Review. PMID: 17196294
55. Kallwass H, Carr C, Gerrein J, Titlow M, Pomponio R, Bali D, Dai J, **Kishnani PS**, Skrinar A, Corzo D, Keutzer J. Rapid diagnosis of late-onset Pompe disease by fluorometric assay of alpha-glucosidase activities in dried blood spots. Mol Genet Metab. 2007 Apr; 90(4):449-52. Epub 2007 Jan 31. Erratum in: Mol Genet Metab. 2007 Nov; 92(3):285. PMID: 17270480
56. Koeberl DD, **Kishnani PS**, Chen YT. Glycogen storage disease types I and II: treatment updates. J Inherit Metab Dis. 2007 Apr; 30(2):159-64. Epub 2007 Feb 16. Review. PMID: 17308886
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58. Spiridigliozzi GA, Heller JH, Crissman BG, Sullivan-Saarela JA, Eells, R, Dawson D, Li J, **Kishnani PS**. Preliminary study of the safety and efficacy of donepezil hydrochloride in children with Down syndrome: a clinical report series. Am J Med Genet A. 2007 Jul 1; 143A (13):1408-13. PMID: 17542008
59. Wang LY, Ross AK, Li JS, Dearmey SM, Mackey, JF, Worden M, Corzo D, Morgan C, **Kishnani PS**. Cardiac arrhythmias following anesthesia induction in infantile-onset Pompe disease: a case series. Paediatr Anaesth. 2007 Aug; 17(8):738-48. PMID: 17596219
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61. Raben N, Takikita S, Pittis Mg, Bembi B, Marie SK, Roberts A, Page L, **Kishnani PS**, Schoser BG,

Chien YH, Ralston E, Nagaraju K, Plotz PH. Deconstructing Pompe disease by analyzing single muscle fibers: to see a world in a grain of sand… Autophagy. 2007 Nov-Dec; 3(6):546-52. Epub 2007 Jun 15. PMID: 17592248

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63. Smith WE, Sullivan-Saarela JA, Li JS, Cox GF, Corzo D, Chen YT, **Kishnani PS**. Sibling phenotype concordance in classical infantile Pompe disease. Am J Med Genet A. 2007 Nov 1; 143A (21):2493-501. Review. PMID: 17705024
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65. Kemper AR, Hwu WL, Lloyd-Puryear M, **Kishnani PS**. Newborn screening for Pompe disease: synthesis of the evidence and development of screening recommendations. Pediatrics. 2007 Nov; 120(5):e1327-34. Review. PMID: 17974725
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67. Koeberl DD, Pinto C, Sun B, Li S, Kozink DM, Benjamin DK Jr, Demaster AK, Kruse MA, Vaughn V, Hillman S, Bird A, Jackson M, Brown T, **Kishnani PS**, Chen YT. AAV vector-mediated reversal of hypoglycemia in canine and murine glycogen storage disease type Ia. Mol Ther. 2008 Apr; 16(4):665-72. Doi: 10.1038/mt.2008.15. Epub 2008 Mar 11. PMID: 18362924
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33. Hijazi G, Murala S, **Kishnani PS**. Pompe disease, PG Textbook of Pediatrics, 3rd Edition. February 2022.
34. Hijazi G, **Kishnani PS**. Defects in metabolism of carbohydrates, Nelson Textbook of Pediatrics, 22nd Edition. Submitted February 2022.
35. Korlimarla A, Gibson R, **Kishnani PS**. Glycogen Storage Diseases, Springer, Cham. In: Bernstein, L.E. Rohr, F, van Calcar S, editor Nutrition Management of Inherited Metabolic Diseases. Published June 2022. Dio.org/10.1007/978-3-030-94510-7_25. Print ISBN: 978-3-030-94509-1; Online ISBN: 978-3-030-94510-7.
36. Reuser A, van der Ploeg A, **Kishnani PS**, Pim Pijnappel WWM. Pompe Disease from PART 2: The Individual Diseases, Wiley & Sons, New York, 2nd Edition, Chapter 17. Atul B. Mehta and Bryan Winchester, Book Editors, Published July 2022. Doi.org/10.1002/9781119697312.ch17. Print ISBN: 9781119697282 and Online ISBN: 9781119697312.

Other

Published Scientific Reviews for Mass Distribution:

1. Abstracts and scientific presentations in newsletters for the National Association for Glycogen Storage Disease.
2. Abstracts and scientific presentations in newsletters for Pompe Disease for International Pompe Association (IPA), Acid Maltase Disease Association (AMDA), Genzyme Corporation.
3. Abstracts & commentaries for the Triangle Down Syndrome Network.
4. Abstracts & commentaries for the Canadian Down Syndrome Network.

Editorial Experience:

Editorial Boards:

Molecular Genetics and Metabolism, 2015- current

Ad Hoc scientific review journals:

- American Journal of Medical Genetics
- Clinical Pediatrics
- European Journal of Pediatrics
- Genetics in Medicine
- JAMA
- JCI
- Journal of Genetic Counseling
- Journal of Inherited Metabolic Disorders
- Journal of Pediatrics
- Lancet
- Molecular Genetics and Metabolism
- Molecular Therapy
- Muscle and Nerve
- NEJM
- Neurology
- Pediatrics

Invited Editor for American Journal of Medical Genetics for the Down syndrome, supplement, 2006
Invited Editor for American Journal of Medical Genetics for Pompe disease, supplement, 2012
Invited Editor for Pediatrics for Newborn Screening in Pompe Disease, supplement, 2017

Patents/Inventions:

29 Duke Invention Disclosures
45 Issued Patents (US and foreign)
13 Pending Patent Applications (US and foreign)
Representative Patents
US Patent #10,647,969, "Method of Treating Glycogen Storage Disease," issued 5/12/2020
US Patent #8,809,282, "Methods of Treatment in Diseases Treated with Therapeutic Proteins," issued 8/19/2014
US Patent #8,679,478 "Methods of Lysosomal Storage Disease Therapy," issued 8/19/2014

Professional Awards and Special Recognitions:

I.C.S.E.	Delhi Board	November 1978	Distinction
H.S.C.E.	Maharashtra	March 1981	Distinction
I M.B.B.S.	Bombay	October 1982	Ranked **5th/583** overall at University of Bombay Distinction in Anatomy, I M.B.B.S.
II M.B.B.S.	Bombay	April 1984	Distinction in Microbiology, II M.B.B.S. **First in Pathology** - Awarded Dr. S.B. Kekre's and Dr. Manek Bilimoria's Prizes in II M.B.B.S. Distinction in Pharmacology, II M.B.B.S.
III M.B.B.S.	Bombay	October 1985	**Ranked 3rd/603** overall at University of Bombay
D.C.H.	Bombay	April 1989	Distinction
M.D.	Bombay	March 1990	**Ranked first** overall at institution (Pediatrics)

- Fellow Travel Award ***for Outstanding Research to the Society of Inherited Metabolic Disorders Conference in Perdido Beach, AL,*** March 1995.
- Fellow Travel Award from the ***Society for the Study of Inborn Errors of Metabolism in Toledo, Spain,*** September 1995.
- Special Recognition by University President Nannerl O. Keohane at Annual Meeting of Duke University Board of Trustees, ***for original research in Down syndrome,*** 1999.
- Special Recognition Honors from Triangle Down Syndrome Network Board of Directors, ***for contributions professionally and personally in the lives of individuals with Down syndrome,*** 2001.
- Interviewed by Wall Street journal for work in Down syndrome research, published ***in article "New Hope for Treating Down Syndrome",*** April 18, 2005.
- Exceptional Parent Maxwell J. Schleiffer Distinguished Service Award, ***for passion, dedication, professionalism and inspiration to people with disabilities, particularly those with Pompe disease,*** 2005.
- Interviewed by Wall Street Journal for work in Pompe disease, published ***in article "Genzyme Drug for Rare Pompe Disease Approval,"*** April 29, 2006.
- Ruth and A. Morris Williams, Jr Faculty Research Prize, ***for intellectual vigor, dedication, and scientific ingenuity needed to make a critical impact on the future of medical research,*** 2008.
- Recognized in list of North Carolina Best Doctors, 2009, 2011-current.
- National Down Syndrome Congress, Christian Pueschel Memorial Research Award, ***for outstanding clinical research that improves lives through greater understanding of Down syndrome,*** 2010.
- Anna's Angels Award, recognition by Anna's Angels Foundation ***for passion and devotion to Down syndrome research,*** 2012.
- Association of Glycogen Storage Disease, ***Lifetime Membership,*** September 2014.
- Dr SS Agarwal Oration Award for **"Novel Therapies for Genetic Disorders",** given by the Society for Indian Academy of Medical Genetics, during the IAMGCON, 2015.
- ***Rare Disease Hero Award in Lysosomal Disease,*** given by the Rare Disease Report, October 2016.
- ***Sandor Oration Award,*** given by the Indian Society of Inborn Errors of Metabolism for work in rare diseases in India, February 2017.
- Recognized as one of the 2021 ***Pioneering Women in Science for Duke Health,*** February 2021.
- Recipient of the Duke Medical Alumni Association's ***Distinguished Faculty Award***, February 2021.
- Featured in the 9th Annual Women's Issue of *Durham Magazine* as ***Women of Achievement***, May 2021.
- ***2021 Emmanuel Shapira Award*****,** given to a Society for Inherited Metabolic Disorders Member for best paper in the field of biochemical genetics and metabolism published in *Molecular Genetics and Metabolism,* "Investigation of ALPL variant states and clinical outcomes: An analysis of adults and adolescents with hypophosphatasia treated with asfotase alfa", 2021.
- **2022 Recipient of the North Carolina Award for Science**, State Highest Honor, administrated by the N.C. Department of Natural and Cultural Resources, November 2022.
- Received the 2022 HSNC (Hindu Society of North Carolina) ***award for Pioneering research and Treatment for Glycogen Storage Diseases (GSD)***
- Received the **2024 Faculty Mentoring Award** for the Duke University School of Medicine, Department of Pediatrics, June 2024.

Mentoring Activities:

A) FACULTY

NAME	MENTORSHIP ROLE	CURRENT POSITION
Laura Case, PT, DPT, MS, PCS, C/NDT	Involvement and design of PT endpoints for clinical trials of neuromuscular disorders such as Pompe disease, SMA, Duchenne muscular dystrophy	Associate Professor of Orthopedic Surgery Duke Medical Center Pediatrics Physical Therapy Duke Children's Hospital Durham, NC
Jennifer L. Cohen, MD	Gene therapy for Pompe disease IUERT to treat lysosomal disorders	Assistant Professor of Pediatrics, Department of Pediatrics, Division of Medical Genetics, Duke Medical Center, Durham, NC
Mai ElMallah, MD, PhD	Pulmonary aspects of Pompe disease. Role of AAV gene therapy in correcting systemic and neurologic aspects of Pompe disease	Chief , Division of Pulmonary and Sleep Medicine' Department of Pediatrics, Duke University Medical Center, Durham, NC
Areeg El-Gharbawy, MD	Metabolomics – role in GSD I and other liver GSDs	Associate Professor of Pediatrics, Department of Pediatrics, Division of Medical Genetics, Duke University Medical Center, Durham, NC
Harrison Jones, PhD	Development of respiratory muscle strength training (RMST) as an adjunctive therapy for Pompe disease. Speech and articulation challenges in Down syndrome	Associate Professor of Surgery, Department of Surgery, Head and Neck Surgery and Communication Sciences, Duke University Medical Center, Durham, NC
Sujay Kansagra, MD	Clinical research approaches for rare diseases	Associate Professor of Pediatrics Pediatric Neurologist School of Medicine Duke Medical Center Durham, NC
Gwen O'Grady, PhD	Development of an algorithm for systematic evaluation of hearing in Pompe disease	Audiologist Division of Speech Pathology and Audiology Duke Medical Center and Duke Children's Hospital Durham, NC
Grace Prakalapakorn, MD	Ophthalmologic endpoints as a way to measure treatment response in clinical trials for infantile Pompe disease.	Associate Professor of Ophthalmology; Assistant Professor in Pediatrics, Duke Medical Center, Durham, NC

Baodong Sun, PhD	Linking data between bench and bedside in the design of preclinical experiments	Associate Professor of Pediatrics, Department of Pediatrics, Division of Medical Genetics, Duke University Medical Center, Durham, NC

B) FELLOWS, RESIDENTS, POST DOCS, DOCTORAL

NAME (Fellows and Residents)	**TITLE OF RESEARCH PROJECT**	**CURRENT POSITION**
Ayesha Ahmad, MD	Role of low cholesterol in Dubowitz syndrome. Role of high dose citrate and aspartate in pyruvate carboxylase deficiency.	Associate Professor, Biochemical Genetics C.S. Mott Children's Hosp. Ann Arbor, MI
Martha Decker-Phillips, MD	Double mutant fibrillin-1 (FBN1) allele in a patient with neonatal Marfan syndrome.	Associate Professor, Maternal-Fetal Medicine Wake Forest Baptist Health
Areeg El-Gharbawy, MD	Expanding the clinical spectrum of late-onset Pompe disease: dilated arteriopathy involving the thoracic aorta. An approach to desensitize infants and young children with Pompe disease with significant reactions to alglucosidase alfa infusions.	Associated Professor, Department of Pediatrics, Division of Medical Genetics Duke Children's Duke University Medical Center
Paula Goldenberg, MD	Mechanisms for immune responses In Pompe Disease. Negative impact of CRIM negative status.	Assistant Professor, Mass General Hospital, Director of NE Regional Center for Chromosome22, Stickler/ Marshall Syndrome
William Hannah, MD	Wolman disease and HLH: Lessons Learned. Very early on-set IBD-is it a part of GSD1a clinical spectrum?	Research Associate Department of Genetics and Genome Science Case Western Reserve University Cleveland, OH
David Jackson, MD	Role of muscle ultrasound in Pompe disease.	Assistant Professor Medical Genetics and Genomics East Carolina University Health Brody School of Medicine Greensville, NC
Vellore Kasturi, MD	Recognition of Menorrhagia as a feature in patients with type 1 glycogen storage disease. Pregnancy outcomes in Pompe disease.	Obstetrician/Gynecologist Community Health Centers Santa Maria, CA

John Koepke, MD	Hepatocellular carcinoma as a complication of GSDIII.	Clinical Geneticist, UNC Hospital
Vidya Krishnamurthy, MD	Diagnostic and treatment challenges of neuropathic Gaucher disease.	Medical Genetics Physician Northside Hospital, GA
Michele Spencer-Manzon, MD	Liver transplantation outcomes for glycogen storage disease type Ia.	Assistant Professor of Genetics and of Pediatrics Pediatric Genomics Discovery Program (PGDP) Yale Medicine
Dietrich Matern, MD, PhD	Liver complications in glycogen storage diseases type I, III and IV.	Chair, Division of Laboratory Genetics and Genomics, Department of Laboratory Medicine and Pathology Mayo Clinic
Margarita Nieto Moreno, MD	Complications in GSD I	Resident Physician, University of Miami Jackson Health System Miami, FL
Mari Mori, MD	Odontohypophosphatasia as a presenting feature of childhood/adult-onset hypophosphatasia. Limitations of whole exome sequencing in detecting infantile and late-onset Pompe disease. Immune phenotyping provides insights into the disease pathology of Pompe disease.	Assistant Professor, Department of Pediatrics, Ohio State University College of Medicine and Clinical and Clinical and Biochemical Geneticist, Genetic and Genomic Medicine, Nationwide Children's Hospital
Ricardo Ong, MD	Hypersensitivity reactions and management in hypophosphatasia	Emergency Medicine Physician Womack Army Medical Center Fort Liberty Hospital Fort Bragg, NC
Pankaj Prasun, MD	Neuropsychiatric manifestations in propionic academia.	Assistant Professor, Genetics And Genomic Sciences, Mt. Sinai Hospital
Wendy Smith, MD	Clinical trial design and implementation for infantile Pompe disease. Sibling concordance in infantile Pompe disease.	Faculty Member, Clinical & Biochemical Genetics Maine Medical Center
Khoon Tan, MD	Identification of premature pubarche as a complication in children with Pompe disease.	Pediatrics Specialist Cleveland Clinic Center For Personalized Genetic Healthcare

		Cleveland, OH
Jariya Upadia. MD	Natural history of liver and muscle outcomes in glycogen storage disease type III. Long-term complications in propionic academia.	Clinical Geneticist, Children's Hospital, New Orleans, LA
Kim Choong Wen Ng, MD	Phenotyping for GSDIV and Use of EMPA in GSD1b	Genetics Specialist Children's Hospital of Philadelphia Philadelphia, PA
Sarah Young, PhD	Urine hex 4 as a biomarker for Pompe disease	Professor of Pediatrics, Division of Medical Genetics Duke University Medical Center
NAME (MD/MBBS Post Docs)	**TITLE OF RESEARCH PROJECT**	**CURRENT POSITION**
Suhrad Banugaria, MBBS	High-sustained antibody titers is a poor prognostic factor to clinical outcome in patients treated with a therapeutic protein. Development of a successful immune modulation protocol to prevent immune response in high-risk patients with Pompe disease.	Pediatrician, The Brooklyn Hospital Center, Brooklyn, New York
Ankit Desai, MBBS	Role of transient low dose methotrexate as an immune tolerance induction agent. Emerging Phenotype of late-onset Pompe disease. Safe rate escalation protocol to administer enzyme replacement therapy.	Senior Research Associate, Department of Pediatrics-Division of Medical Genetics Duke University Medical Center
Luis Franco, MD	Identification of hepatocellular carcinoma as a long-term complication of Glycogen storage disease type Ia.	Assistant Clinical Investigator, Clinical Research Laboratory of Immune System Biology National Institute of Allery and Infectious Disease
Carine Halaby, MD	Natural history of liver and muscle involvement in glycogen storage type III.	Resident Physician, Pediatrics UT Southwestern Medical Center, Dallas, TX
Rabi Hanna, MD	Diagnostic and treatment challenges of neuropathic Gaucher disease. Fracture risk in children with Pompe disease.	Chairman, Department of Pediatric Hematology-Oncology and Bone-Marrow Transplant Cleveland Children's Clinic Cleveland, OH

Mrudu Herbert, MBBS, MPH	Cardio protective role of c.-32-13T>G variant in late on-set Pompe disease. Identification of early markers of disease in late on-set Pompe disease picked by newborn screening. Continuous glucose monitoring as a way to monitor individuals with Glycogen storage disorders.	Pediatrician, General Pediatrics University of Kentucky Albert B. Chandler Hospital Lexington, KY
Ghada Hijazi, MD, MRCPCH, FCCMG	Natural history of glycogen storage disease type III and type IX γ2	Postdoctoral Associate Pediatrics-Medical Genetics Duke University Medical Center
Zoheb Kazi, MBBS	Role of bortezomib to abrogate an entrenched immune response to a therapeutic protein. Role of transient low dose methotrexate as an immune tolerance induction agent.	Physician/Clinical Research Immunovant, Inc. (IMVT) New York, NY
Aleena Khan, MBBS	Role of increased dose in Pompe disease and use of WBMRI as a biomarker in Neuromuscular disease	General Pediatrician in Emergency Medicine UT Southwestern Medical Center Dallas, TX
Aditi Korlimarla, MBBS	CNS manifestations in Pompe disease.	Associate Medical Director Ono Pharma UK Ltd. London, UK
Ju-Li Lin, MD	Isolation and nucleotide sequence of canine glucose-6-phosphatase mRNA: identification of mutation in puppies with glycogen storage disease type Ia.	Professor of Genetics, Dept. of Pediatrics, Division of Medical Genetics, Chang-Gung Memorial Hospital, Taiwan
Neelam Makhijani, MD	Newborn Screening in Pompe disease-clinical phenotyping and treatment outcomes	2nd Year Medical Genetics Genomics Fellow Duke Medical Center School of Medicine Department of Pediatrics
Sireesha Murala, MBBS	Role of DTI in monitoring white matter hyperintensities in Pompe disease.	Associate Scientific Director HMP Global Malvern, PA
Mugdha Rairikar, MBBS, DCH	Role of high dose IVIG as an immune modulating agent for patients with high antibody titers to enzyme replacement therapy. Role of substrate reduction therapy in thrombocytopenia in	Pediatrics Nephrology, Fellowship Baylor College of Medicine Houston, TX

	patients with Gaucher disease type I.	
Maheen Sheikh, MBBS	Natural history of glycogen storage disease type VI Comparison of histopathology outcomes in glycogen storage disease	Postdoctoral Associate, Pediatrics-Medical Genetics Duke University Medical Center
NAME (DVM Post Docs)	**TITLE OF RESEARCH PROJECT**	**CURRENT POSITION**
Elizabeth Brooks, MS, DVM	Characterization of canine glycogen storage disease Type I, and III and treatment interventions.	Staff Veterinarian, Pediatrics-Medical Genetics Duke University Medical Center
NAME (PhD Post Docs)	**TITLE OF RESEARCH PROJECT**	**CURRENT POSITION**
Su Jin Choi, PhD	Immune modulation in the setting of gene therapy	Research Associate, Senior Duke Medical Center Department of Pediatrics, Division of Medical Genetics Durham, NC
Shelly Goomber, PhD	Functional studies to understand VUS in Pompe and other rare diseases	Postdoctoral Associate, Department of Pediatrics, Division of Medical Genetics, Duke University Medical Center
Rebecca L. Koch, PhD, RDN	Clinical and lab correlations in GSD IV. Role of gene therapy. Focused on Natural History and Biomarker development for GSD IV, VI and IX.	Postdoctoral Associate, Duke Medical Center Department of Pediatrics, Division of Medical Genetics, Durham, NC
Jeong-A Lim, PhD	AAV9 gene therapy for GSD IIIa	Senior Research Associate, Pediatrics-Medical Genetics Duke Medical Center
Han-Hyuk Lim, MD, PhD	Using rapamycin-carrying synthetic vaccine particles (SVP) in conjunction with enzyme replacement therapy to induce immune tolerance in Pompe disease.	Assistant Professor, Department of Pediatrics Chungnam National University Hospital
Jianjun Shen, PhD	Identification of a protective mutation in non-progressive hepatic form of type IV Glycogen storage disease. Genotype-phenotype correlations and mutations update in type III glycogen storage disease.	Professor, Dept. of Epigenetics And Molecular Carcinogenesis Texas Anderson Cancer Center, Houston, TX

Haiqing Yi, PhD	A modified approach to measuring glycogen content in GSD IV alglucosidase alfa as a treatment approach for GSD III and IV. AAV gene therapy for GSD IV	Research Associate, Senior Department of Pediatrics Division of Medical Genetics Duke Medical Center
NAME (Medical Students – Research Year)	**TITLE OF RESEARCH PROJECT**	**CURRENT POSITION**
Chris Carter	Natural history of GSD VI	3rd Year Medical Student Research year with Division of Medical Genetics, Department of Pediatrics, Duke Medical Center, Durham, NC
Sammi Fernandes	Natural history of GSD IX. Role of WBMRI in setting of infantile Pompe disease.	Med/Peds resident at Vanderbilt University Medical Center Nashville, TN
Matthew Gayed	Natural history of APBD	3rd Year Medical Student Research year with Division of Medical Genetics, Department of Pediatrics, Duke Medical Center, Durham, NC
Rebecca Gibson	Characterization of mouse model for GSD IX (Gamma Variant), AAV gene therapy approach to treating GSD IX.	MD/PhD Candidate, Duke University, Durham, NC Research year with Division of Medical Genetics, Department of Pediatrics
Bridget Kiely	Natural history of GSD IV and APBD	Medical Student Duke University School of Medicine
Anna Paschall	Natural history of GSD III and liver manifestation in patients with GSD IX	Medical Student Duke University School of Medicine
Trusha Patel, MD	The negative impact of antibodies in the late-onset Pompe disease. Skeletal muscle pathology of infantile Pompe disease during long-term enzyme replacement therapy	Attending Physician, Division of Gastroenterology, Hepatology and Nutrition, Children's Hospital of Philadelphia, PA
Sean Prater, MD	Recognition of the long-term phenotype of Infantile Pompe disease survivors treated with enzyme replacement therapy	Psychiatrist, Department of Psychiatry and Behavioral Medicine, Wake Forest Baptist Medical Center, Winston Salem, NC

C) MEDICAL STUDENTS

NAME	**TITLE OF RESEARCH PROJECT**	**CURRENT POSITION**

Alexandria Beckemeyer	Reviewing literature of existing therapies and unmet needs in Pompe disease and helped with research and etc. for Clinical Trials	Owner, Holistic Nutritionist Allure Wellness Los Angeles, CA
Justin Chan	Systematic literature review of emerging phenotype of late-onset Pompe disease	Medical Student Duke University School of Medicine Durham, NC
Stuti Das	Role of Stbd1 in cytoplasmatic transport of glycogen to lysosomes	Physician, Ophthalmology University of Utah Salt Lake City, UT
Rebecca Eells	Role of rivastigmine and donepezil to enhance cognition in Down syndrome disease	Associate Director of Biophysics Reaction Biology Corporation Malvern, PA
Christine Ha	Muscle pathology in Pompe disease; recognition of glycogen accumulation in interstitium	Duke University School of Medicine Durham, NC
Sneha Jain	Recognition of small-fiber neuropathy in Pompe disease	Resident Physician California Pacific Medical Center San Francisco, CA
Cindy Li	Cognitive and academic outcomes in long-term survivors of infantile-onset Pompe disease	Clinical Research Coordinator, Division of Medical Genetics, Department of Pediatrics, Duke University Medical Center Durham, NC
Paul McIntosh	Characterization of gait abnormalities in Pompe disease. Neuroimaging findings in infantile Pompe long-term survivors. Strategy for evaluation of family members with Pompe disease	PGY-3 Senior Assistant Resident Department of Neurology Duke University Medical Center Durham, NC
Kunal Potnis	Bone complications in neuropathic Gaucher disease despite ERT	MD Candidate, Yale School of Medicine New Haven, CT

D) Research Mentor for MD Fellows, Other Specialties

NAME	**TITLE OF RESEARCH PROJECT**	**CURRENT POSITION**
Annette Ansong, MD (Cardiology Fellow)	Electrocardiographic response to enzyme replacement therapy for Pompe disease and role of EKG in long term follow-up.	Pediatric Cardiologist, Inova Affiliated Hospitals Fairfax, VA
Srinivas Reddy, MD (Surgical Fellow)	Role of liver transplantation for glycogen storage disease type Ia.	Associate Professor of Oncology, Department of Surgical Oncology

	Outcome of patients with glycogen storage disease type Ia who have had adenoma liver resection.	Co-Director, Liver and Pancreas Tumor Center Roswell Park Comprehensive Cancer Center, Buffalo, New York
Amanda Cook, MD (Cardiology Fellow)	Role of Holter monitoring to capture arrhythmia risk in infants treated with recombinant human acid alpha-glucosidase enzyme replacement therapy for Pompe disease.	Pediatric Cardiologist, Sanger Heart & Valve Institute Iredell Health System Salisbury, NC
Rodney McDowell, MD (Cardiology Fellow)	Arrhythmias in patients receiving enzyme replacement therapy for infantile Pompe disease, a new long-term complication.	Pediatric Cardiologist, Baptist Health Louisville Clark Memorial Hospital New Albany, IN
Shawyntee (Vertilus) Mayo, MD (Cardiology Fellow)	Echocardiographic manifestations of Glycogen storage disease III-cardiac hypertrophy is a complication.	Assistant Professor, Department of Pediatrics, University of Florida College of Medicine, Jacksonville, FL
Luke Wang, MD (Anesthesia Fellow)	Anesthesia precautions in infantile-onset Pompe disease.	Assistant in Pain Medicine Instructor in Anesthesia Anesthesiology Perioperative and Pain Medicine Boston Children's Hospital Boston, MA
Natalia Dixon, MD (Hematology Fellow)	Clinical manifestations of hematologic and oncologic disorders in patients with Down syndrome. Identification of iron deficiency as an under recognized feature in children with Down syndrome.	Assistant Professor, Pediatrics Pediatric Hematology-Oncology Wake Forest Baptist Health Wake Forest, NC
Jeffrey Horvath (Radiology Fellow)	Correlation between quantitative whole-body muscle MRI and clinical muscle weakness in Pompe disease. Identification of lingual weakness: MRI approach in patients with glycogen storage disorder Type IIIa.	Radiologist, Vascular and Interventional Radiology/Diagnostic Radiology Triad Radiology Associates and Durham Veterans Affairs Medical Center Durham, NC
Divya Ajay, MD (Urology Resident)	Lower urinary tract symptoms and incontinence in Children with Pompe Disease.	Urologist, University of Rochester Medical Center Rochester, NY
Ramin Javan, MD (Radiology Fellow)	Generating color-coded anatomic muscle maps for correlation of quantitative magnetic resonance	Associate Professor of Neuroradiology Division of Neuroradiology

	imaging analysis with clinical examination in neuromuscular disorders.	George Washington University Medical Center Washington, DC
Erin McNamara, MD, MPH (Urology Fellow)	Lower urinary tract symptoms and incontinence in adults with Pompe disease and Gaucher disease.	Associate Director, Colorectal and Pelvic Malformation Center; Associate, Department of Urology Instructor in Surgery Harvard Medical School Boston, MA
Rachel Torok, MD (Cardiology Fellow)	Delineation of phenotype in patients with PRKAG2 mutations. Understanding cardiac manifestations in GSD II.	Assistant Professor of Pediatrics, Division of Cardiology, School of Medicine, Duke University Medical Center Durham, NC
Jordan E. Ezekian, MD, MPH (Cardiology Fellow)	Cardiac Manifestations of Pompe Disease	House Staff – Pediatrics Department of Pediatrics Duke University Medical Ctr. Durham, NC

E) Research Mentor for Allied Health Professionals

NAME	**RESEARCH INTEREST**	**CURRENT POSITION**
Joanne F. Mackey, RN, PNP Certified Nurse Practitioner July 1995-2013	Celiac disease in Down syndrome.	Retired
Jennifer A. Sullivan, MS, CGC Certified Genetic Counselor June 1997-Present	Cholinergic therapy as a treatment approach for Down syndrome.	Senior Genetic Counselor Pediatrics-Medical Genetics Duke Medical Ctr. Durham, NC
Stephanie DeArmey, PA-C Physician Assistant July 2001-Present	Practical approaches to desensitization in patients with hypersensitivity reactions to a therapeutic protein Hypophosphatasia: Clinical spectrum.	Physician Assistant Pediatrics-Medical Genetics Duke Medical Ctr. Durham, NC
Jennifer Goldstein, PhD, MS Certified Genetic Counselor May 2005-August 2014	Mutation analysis to determine CRIM status in Pompe disease. Natural history of GSD IX.	Assistant Research Professor University of North Carolina at Chapel Hill Chapel Hill, NC
Stephanie Austin, MS, MA, CGC	Cardiac complications in GSD III. Role of WBMRI as a tool to evaluate treatment response to ERT. Developmental outcome in	Medical Science Liaison, Associate Director, US Field Medical Division, Amicus Therapeutics Cranbury, NJ

Certified Genetic Counselor and Senior Research Program Leader January 2006-2021	long-term survivors of infantile Pompe disease.	
Katie Berrier, MD, CSGC Certified Genetic Counselor August 2010-May 2017	Spectrum of mutations in CRIM negative Pompe disease.	Patient Education Liaison (patient and family education and support) Sanofi Genzyme, U.S. Rare Disease Cambridge, MA
Crista Walters, NP Certified Nurse Practitioner February 2013-2019	Practical approaches to rate escalation of ERT. Careful approaches in Phase I study design.	Nurse Practitioner Pediatrics-Rheumatology Duke Medical Ctr. Durham, NC
Lauren Flueckinger, MS, CGC Certified Genetic Counselor June 2013-2019	Hypophosphatasia and complex counseling issues. Phenotypic spectrum of hypophosphatasia.	Medical Science Liaison Sanofi Genzyme, U.S. Rare Disease Cambridge, MA
Sarah Hart, MS, MA, PhD Genetic Counselor July 2014-June 2021	Down syndrome disintegrative disorder: dissecting the phenotype.	Genetics Counselor Pediatrics-Medical Genetics Duke Medical Ctr. Durham, NC
Erin Huggins, MS, CGC Clinical Research Genetic Counselor 2019-Present	Emerging phenotype in late onset Pompe disease identified by NBS	Clinical Research Genetic Counselor, Department of Pediatrics, Division of Medical Genetics Duke Medical Ctr. Durham, NC
Erica Nading, MS, CGC Certified Genetic Counselor 2014-Present	Variant reclassification in hypophosphatasia. Identification of non-coding pathogenic variants inborn errors of metabolism.	Genetics Counselor Pediatrics-Medical Genetics Duke Medical Ctr. Durham, NC

Mentorship Committee for Faculty Members

Faculty Members	Type	Approximate Start	Approximate End
Dwight Koeberl, MD, PhD	Committee Chair	November 2005	2010
Edward Smith, MD	Committee Chair	2008	2023
Yong-Hui Jiang, MD, PhD	Committee Chair	2010	2019
Loren Pena, MD, PhD	Committee Chair	July 2012	2018
Mai Elmallah. MD, PhD	Committee Chair	July 2017	Present
Areeg El-Gharbawy, MD	Committee Member	October 2019	Present
Jennifer L. Cohen, MD	Committee Member	October 2019	Present

Mentorship Committee for Graduate Students

Student	Type	Approximate Start	Approximate End
Rebecca Gibson	Committee Chair (primary faculty advisor)	2018 – Kishnani Lab	2023

Veronica Gough	Member, Pre-Prelim Committee	2018 – Gersbach Lab	2022
Apoorva Iyengar	Member, Pre-Prelim Committee	2021 – Reddy Lab	2023
Alan Rosales	Member, Pre-Prelim Committee	2021 – Asokan Lab	2023
Tong Chen	Member, Pre-Prelim Committee	2021 – Bissig Lab	2024
Makenzie Beaman	Member, UPGG Thesis Committee	2021 – Crawford Lab	2024

Mentorship Committee for Undergraduate Students

Student	Type	Approximate Start	Approximate End
Emily Duerr	Committee Chair (primary faculty advisor)	2022 – Kishnani Lab	2023

Mentor to High School Students (select list of more than 50 students that I have mentored)

Student	Time Period	Current
Vivek Ramaswamy	2001-2002	Chairman and President, Roivant Science
Shankar Ramaswamy	2005-2006	Co-Founder and Chief Executive Officer, Kriya Therapeutics
Rahul Jawaney	2010-2011	Cardiology Fellowship at Cleveland Clinic
Karishma Desai	2011-2012	Dermatology residency, Leonard M. Miller School of Medicine, University of Miami
Shaylee Boger	Aug. 3-Aug 12, 2017	Sophomore at Trinity College, who was honored with 2017 Sanofi Genzyme Torch award (for contributions to the LSD community)
Rasika Rajagoplan		Senior Manager, People and Culture Business Partner at Curology
Neha Jumani	2022 – Present	

Education / Teaching Activities:

- Resident teaching, medical student and fellows training at Duke University Medical Center.
- Genetic counselor teaching of biochemical genetics at Duke University Medical Center.
- Medical student genetics teaching course at Duke University Medical Center.
- Participation and presentations at Pediatric Grand Rounds.
- Participation and presentations in Pediatric Faculty Research Conferences.
- Participation and presentations in weekly Genetics and Metabolism research and teaching seminars.
- Presentations at Grand Rounds in Outreach centers for Duke University and by invitation to outside institutions.
- Participation and presentations in the Duke Clinical Research Unit Rounds.
- Participation in Liaison Committee on Medical Education (LCME) of Department of Pediatrics, Duke University Medical Center.
- Invited lecturer at Clinical Correlation Molecules, Cells & Tissues Course, "Glycogen Storage Disease", Duke University Medical Center.

Invited Lectures and Presentations:

1. Pulmonary Hypertension in Glycogen Storage Disease Type I (GSDI). Annual American Glycogen Storage Disease Association, Atlanta, GA, 1995.

2. Down Syndrome – Overview and management, Pediatric Grand Rounds, Rex Hospital, Durham, NC, 1996.
3. Down syndrome – Overview and management, Pediatric Grand Rounds, Rocky Mount, NC, 1997.
4. Nutritional deficiencies in Glycogen Storage Disease Type I (GSDI). Annual American Glycogen Storage Disease Association, Raleigh, NC, 1997.
5. Mutation in canine Glucose-6-Phosphatase Deficiency (glycogen storage disease type Ia
6. , GSD-Ia), American Maltese Association, 1997.
7. Clinical & Molecular Evaluation in Glycogen Storage Disease type III. Association for Glycogen Storage Disease Annual meeting, Davenport, IA, 1998.
8. Overview of Management Issues and Current Trends in Down syndrome. Coastal Down syndrome Quarterly Meeting, Shallotte, SC. March 1998.
9. Cholinergic Therapy and overview of current trends in Down syndrome. Down syndrome Association of Charlotte, NC, October 1998.
10. Cholinergic Therapy and Celiac disease in Down syndrome. Down syndrome Family Alliance of Greenville, SC, November 1998.
11. Overview of Management Issues and Current Trends in Down syndrome. Triangle Down Syndrome Society, Durham, NC, July 1999.
12. Canine model of Glycogen Storage Disease Type Ia. Association for Glycogen Storage Disease Annual meeting, Seattle, WA, September 1999.
13. Down syndrome: Treatment with Donepezil. Pfizer and Eisai Pharmaceuticals, October 1999.
14. Down syndrome: A Health care update Conference at Children's Hospital of Pittsburgh, Research Panel and speaker for treatment strategies in Down syndrome, November 1999.
15. "Life during CAP" session at the NIH. Invited speaker to serve as a role model to discuss what CAP's should be trying to accomplish during their tenure to make them remain competitive in academic medicine, Washington, DC, March 2000.
16. Participation in Australian Radio National network presentation regarding Donepezil hydrochloride on four individuals with Down syndrome, March 2000.
17. Participation in the BBC Radio Network presentation regarding Donepezil hydrochloride in the treatment of individuals with Down syndrome, March 2000.
18. Invited speaker at launch meeting for multicenter trial for role of Donepezil in cognition in individuals with Down syndrome, Charleston, SC, December 2000.
19. Invited speaker at 2001 Down Syndrome Family Conference, Columbia, SC, May 2001.
20. Invited speaker at Association for Glycogen Storage Disease, "Recent advances in treatment trials for glycogen storage disease, type III", Tampa, FL, September 2001.
21. NPR Radio presentation on clinical and treatment aspects in Down syndrome, October 2001.
22. Invited panel member of medical experts and speaker at "Fired Up" Down syndrome meeting, "Cholinergic therapy in Down syndrome - updates and advances", Nashville, TN, November 2001.

23. Invited speaker at National Down Syndrome Society (NDSS) meeting, "Cholinergic therapy in Down syndrome - an update", Nashville, TN, July 2002.
24. Invited speaker, It's in Your Genes Program by Haddassah Group, Screening for Jewish Genetic Diseases Charlotte, NC, 2002.
25. Invited speaker to Triangle Down Syndrome Network meeting "Down Syndrome, an update on management and new therapies", Cary, NC, 2003.
26. Invited speaker to II International Conference on Prospects in the treatment of rare diseases, Trieste, 2002.
27. Invited speaker to a teaching course for medical students, Yale "Lysosomal storage diseases, from bench to bedside," Yale, Connecticut, March 2003.
28. Invited Grand Round Speaker, Johns Hopkins "Pompe Disease: An update", Baltimore, MD, September 2003.
29. Invited speaker at Association for Glycogen Storage Disease, "Hepatocellular carcinoma in glycogen storage disease, type I, Portland, OR, September 2003.
30. Invited speaker at International Pompe Association meeting "Enzyme replacement Therapy with recombinant human acid alpha glycosidase (rhGAA) in Infantile Pompe Disease: Duke Experience", Heidelberg, Germany, October 2003.
31. Invited speaker at The Challenge of Lysosomal Storage Disorders: From Clinical Manifestations to Therapy. "Pompe Disease an update of ERT in infantile Pompe disease," Los Angeles, CA, November 2003.
32. Invited speaker at Neurogenetics teaching Seminar Advances in the Diagnosis and Management of Lysosomal Storage Disorders "Pompe disease, an overview and update of ERT," New York University, NY, November 2003.
33. Invited speaker at International Down Syndrome meeting "Therapeutic interventions in Down Syndrome," Barcelona, Spain, March 2004.
34. Invited Grand Round Speaker, LeBonhers Children Hospital "Pompe Disease: The Past, present and Future," Baltimore, MD, April 2004.
35. Invited speaker at The Annual symposium on Lysosomal Storage Disorders, "Pompe Disease, current and future therapeutic options," Madrid, Spain, April 2004.
36. Invited speaker at National Down Syndrome Society (NDSS) meeting "Treatment with cholinesterase inhibitors," Washington, DC, April 2004.
37. Invited speaker at Annual Symposium of WORLD Lysosomal Disease Clinical Research Network "Longitudinal data and follow up of patients with infantile Pompe disease," Minneapolis, Minnesota, May 2004.
38. Invited speaker at Latin American LSD meeting "Pompe Disease, therapeutic options and results of treatment trials," Santiago, Chile, August 2004.
39. Invited speaker at Clinical Advances in Pediatrics meeting "Overview of Glycogen Storage Disease" and

"Update on Pompe Disease," Kansas City, Missouri, November 2004.

40. Invited expert speaker at American College of Medical Genetics at Pompe Disease Management Guideline Working Group meeting "Overview of Pompe" and "Surgery and Anesthesiology in Pompe disease," Miami, FL, October 2004.
41. Invited speaker at Exceptional Parents on line seminar on "Lysosomal Storage disorders," January14, and January 28, 2005.
42. Invited speaker to the Society of Inherited Metabolic Disorders "Long term issues and complications of Glycogen Storage Disease," Asilomar, CA.
43. Invited speaker at 6th Annual Taiwan Human genetics Society meeting "Pompe disease, natural history and change after ERT," March 2005.
44. Invited speaker to the American College of Medical Genetics "Glycogen Storage Diseases, long term outcome and complications," Dallas, TX, March 2005.
45. Invited expert speaker at ENMC meeting "Clinical spectrum and Natural course of children and adults with GSDII," "Tetrasaccharides as markers for GSDII," and "How should muscle strength and function be assessed in infants/children/adults?" Naarden, Netherlands, April 2005.
46. Invited panel member and speaker at NDSS research conference on "Cognitive research in Down syndrome," Cincinnati, OH, May 2005.
47. Invited to co-chair "Annual LSD Registries Meeting," Phoenix, AZ, May 2005.
48. Invited Speaker at Annual Symposium of WORLD Lysosomal Disease Clinical Research Network "Update and long term follow up data of patients with infantile Pompe disease," Phoenix, AZ, May 2005.
49. Invited expert speaker at MDA meeting on "Challenges in Drug Development for Muscle Disease: A Stakeholders' Meeting," Bethesda, MD, August 2005.
50. Invited speaker at "Pompe Disease Management Forum," Cambridge, MA, August 2005.
51. Invited speaker at "1st Disease Severity Index Meeting," Paris, France, September 2005.
52. Invited speaker and Chairperson of Association for Glycogen Storage Disease on "GSD's overview and long-term complications," Calgary, Alberta, Canada, September 2005.
53. Invited speaker at "International Conference on Inborn Errors of Metabolism," New Delhi, India, September 2005.
54. Invited speaker at International Collaborative Gaucher Group (ICGG) US Regional Coordinators Meeting, Palm Beach, FL, October 2005.
55. Invited speaker at ASHG Annual Meeting, Salt Lake City, UT, October 2005.
56. Invited speaker at 10th Annual LSD Latin American Symposium "Evidenced Based Medicine in LSD: From Experimentation to Individualization," San Jose, Costa Rica, November 2005.
57. Invited speaker at Issues in Presymptomatic Diagnoses of Lysosomal Storage Diseases on "Clinical Overview and Update of Clinical Trials of Myozyme for Pompe Disease: Role of Inborn Screening," Bethesda, MD, December 2005.
58. Invited speaker at the Zavesca® Consultant Meeting, San Francisco, CA, December 2005.

59. Invited speaker at FDA meeting, College Park, MD, January 2006.
60. Invited speaker at Grand Rounds Meeting, New York, NY, February 2006.
61. Invited speaker at FDA Expert Panel Meeting for Pompe Disease, Bethesda, MD, February 2006.
62. Invited speaker at Steps Forward in Pompe Disease Symposium and chairperson for "Cardiology & Anesthesiology Perspectives" workshop, Berlin, Germany, April 2006.
63. Invited speaker at Conference of Kennedy Krieger, sponsored by Taishoff Family Foundation. Improving speech, language and cognitive outcomes: novel approaches combining pharmacotherapy and educational intervention in children with DS "Rivastigmine for language and memory enhancement in DS," Baltimore, MD, April 2006.
64. Invited speaker at Genzyme Pompe Disease Consultant Meeting, Dallas, TX, April 2006.
65. Invited keynote speaker at Dr. M.P. Bhagat oration at GSMC & KEMH "Therapy for Lysosomal Storage disorders – No longer elusive!" Mumbai, India, June 2006.
66. Invited speaker at the Department of Pediatrics, University of Malaya "LSDs: the burden of disease" and "ERT: Medical milestones," Malaysia, June 2006.
67. Invited speaker at Hospital Kuala Lumpur "Clinical Spectrum of LSDs" and "Role of ERT in treating LSDs," Malaysia, June 2006.
68. Invited speaker at The Children's Medical Institute of the National University Hospital "LSDs: the burden of disease" and "ERT: Medical milestones," Singapore, June 2006.
69. Invited speaker at KK Women's and Children's Hospital "Down Syndrome, management issues and trends" and "Updates in LSD's," Singapore, June 2006.
70. Invited speaker at Pediatric Grand Rounds at Morgan Stanley Children's Hospital of New York, Columbia University Medical Center "Cardiac Manifestations of LSDs," New York, July 2006.
71. Invited speaker at National Down Syndrome Congress "Cholinergic Therapy in Down syndrome: An update of clinical trials at Duke," Atlanta, GA, July 2006.
72. Invited speaker and chairperson at the International Congress of Inborn Errors of Metabolism, Chiba, Japan, September 2006.
73. Invited speaker to the American Association of Neuromuscular & Electrodiagnostic Medicine (AANEM) Annual Meeting "Pompe Disease: Diagnostic Advances and New Treatment Options." To discuss "Epidemiology and Pathophysiology of Pompe disease," Washington DC, October 2006.
74. Invited keynote speaker at the AMDA/IPA Pompe Disease Conference, San Antonio, TX, November 2006.
75. Invited speaker to the Health Resources and Services Administration (HRSA) Evidence-base Evaluation and Decision Process for the Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children (ACHDGDNC). To discuss "Evaluation of Pompe Disease, for newborn screening," Washington, DC, October 2006.
76. Invited speaker to the Immune Tolerance Network NSC Meeting – Session on Enzyme Deficiency Disease and Network Steering Committee Meeting. To discuss "Clinical Consequences of Immune

Responses to Enzyme Replacement Therapy in Pompe's Disease," Washington, DC, November 2006.

77. Invited keynote speaker at the AMDA/IPA Pompe Disease Conference, "Pompe disease, clinical update, long term effects of ERT and prognostic factors," San Antonio, TX, November 2006.
78. Invited Speaker at Annual Symposium of WORLD Lysosomal Disease Clinical Research Network "Myozyme as a treatment for Pompe disease, updates in research", Orlando, Florida, Dec 2006.
79. Invited speaker at Pediatric Grand Rounds at New York Presbyterian Hospital-Cornell "LSD's - Cardiac Manifestations as clues in diagnosis and management updates," New York, Jan 2007.
80. Invited speaker at Idiopathic and Primary cardiomyopathy in children, research, directions and strategies conference "Role of ERT in metabolic cardiomyopathies," Bethesda, MD, Jan 2007.
81. Invited speaker at Grand Rounds presentation to faculty and graduate students in the Department of Human Genetics at Emory University "Down Syndrome Clinical and research Updates," Atlanta, GA, Feb 2007.
82. Invited speaker and Chairperson for ACMG "Innovations in the management of Pompe disease," Nashville, TN, March 2007.
83. Invited speaker to the AAN meeting "First FDA approved treatment for muscle disease," Boston, MA, April 2007.
84. Invited speaker to the Centers for Disease Control and Prevention (CDC), the Georgia Division of Public Health, and the Georgia Chapter of the March of Dimes for the 5th Annual Birth Defects Monitoring and Prevention "Clinical Trials, Research Updates in Down Syndrome," Atlanta, GA, August 2007.
85. Invited speaker to the United Pompe Foundation Patient and Family Meeting "Pompe Disease: Diagnostic Advances and New Treatment Options," Los Angeles, CA, August 2007.
86. Invited speaker to the United Pompe Foundation Patient and Family Meeting "New Advances for Pompe Disease," San Francisco, CA, August 2007.
87. Invited speaker to the United Pompe Foundation Patient and Family Meeting "New Advances for Pompe Disease," Los Angeles, CA, August 2007.
88. Chairperson for the Latin American Training Meeting and Symposium, "Early Diagnosis and Treatment," Uruguay, Montevideo, September 2007.
89. Invited speaker to the Latin American Training Meeting and Symposium, "Monitoring Patient Care in a Multidisciplinary Approach," Uruguay, Montevideo, September 2007.
90. Invited as Distinguished speaker to the American Society of Human Genetics 57th Annual Meeting "The Bridge between Research and Practice," San Diego, CA, October 2007.
91. Invited speaker to the American Society of Human Genetics 57th Annual Meeting "Early Intervention for LSDs: What is Appropriate Care and Management of the Pre-Symptomatic Individual?" and "A Collaborative Discussion," San Diego, CA, October 2007.
92. Invited speaker to the CDC Workshop: Setting a Public Health Research Agenda for Down syndrome, Atlanta, GA, November 2007.
93. Invited speaker to the MDA Directors Meeting, "Pompe disease a treatable neuromuscular disorder,"

Providence, RI, November 2007.

94. Invited speaker to the American College of Medical Genetics Diagnostic Confirmation of LSD Work Group Meeting, "Variability in clinical presentation of LSDs," Washington DC, November 2007.
95. Invited speaker and International Board Representative, India Charitable Access Program, Mumbai, India, February 2008.
96. Invited speaker and Chairperson to the "Clinical Experts Forum and Pompe Meeting," Prague, Czech Republic, February 2008.
97. Invited speaker to the "Diagnostic Guidelines on Acid Maltase Deficiency (Pompe Disease) Development Meeting," Dallas, TX, February 2008.
98. Invited Highlights Plenary Speaker to the American College of Medical Genetics Annual Clinical Genetics Meeting, "Alglucosidase Alfa: One Small Step for Patients with Pompe, One Giant Leap for Neuromuscular Diseases," Phoenix, AZ, March 2008.
99. Invited Guest Speaker at All India Institute of Medical Sciences Annual Genetics Meeting, "New advances in diagnosis and management of Lysosomal Storage Disorders," and "Advances in Down Syndrome," New Delhi, India, August 2008.
100. Invited Guest Speaker at KEM Hospital Pediatrics Meeting, "Updates in Glycogen Storage Disorders," Mumbai, India, August 2008.
101. Invited speaker at Association of Glycogen Storage Disease Annual Conference, "The Era of Therapeutics", Moline, IL, September 2008.
102. Invited speaker at Association of Glycogen Storage Disease UK Pompe Workshop and IPA Meeting, "Management Guidelines for the Pompe Infant," and "Clinical and Scientific Updates in Pompe disease," Reading, UK, September 2008.
103. Invited speaker at Association of Glycogen Storage Disease, UK Meeting, "Updates in GSD I and GSD III," Reading, UK, September 2008.
104. Invited speaker at FDA Panel Meeting, at the closed and open session, "Pompe Disease -Clinical Spectrum, Pathophysiology and Treatment response to Myozyme," Washington, DC, October 2008.
105. Plenary Speaker at American Society of Human Genetics Meeting, "Pompe Disease, course of the disease and treatment options," Philadelphia, PA, November 2008.
106. Plenary Speaker at National MDA Clinic Director's Conference, "Updates in Pompe Disease, from bench to bedside," Las Vegas, NV, January 2009.
107. Invited speaker at Neurology Grand Rounds at University of Rochester, "Pompe Disease, lessons learned and application to other neuromuscular disorders," New York, NY, March 2009.
108. Plenary lecture at American College of Medical Genetics Meeting, "Stuffed too Full Lysosomal Storage Disorders and the Heart," Tampa, FL, March 2009.
109. Invited speaker at Royal Children's Hospital, "Early diagnosis and efficacy of treatment with Myozyme for Pompe Disease," Melbourne, Australia, April 2009.
110. Invited speaker at Westmead Metabolic Meeting, "Importance of diagnosis of Glycogen Storage Disease

and efficacy of treatment of Pompe Disease," Sydney, Australia, April 2009.

111. Invited speaker at Australian Gaucher Treaters Meeting, "Gaucher Disease – Updates in Management," Sydney, Australia, April 2009.
112. Invited speaker at Pompe Treaters Meeting, "Diagnosis of Pompe Disease," "Enzyme Replacement Therapy with Myozyme," "Multidisciplinary Disease Management," "Differential Diagnosis & High-Risk Testing for Pompe Disease," Sydney, Australia, April 2009.
113. Invited speaker at Association for Glycogen Storage Disease Meeting, Research Updates in GSD I, II, III, Denver, CO, September 2009.
114. Invited chair for "Clinical presentation and multidisciplinary management of Pompe Disease at the 3rd European Symposium "Steps Forward in Pompe Disease," Munich, Germany, November 2009.
115. Invited speaker for 3rd European Symposium "Steps Forward in Pompe Disease "and "Immunological Aspects of Treatment of Pompe Disease," Munich, Germany, November 2009.
116. Invited speaker for University of Pittsburgh Neurology Grand Rounds, "Pompe Disease – A Treatable Neuromuscular Disease, Lessons, Learned and Challenges Ahead," Pittsburgh, PA, December 2009.
117. Invited speaker for Lysosomal Disease Network (LDN) Annual WORLD Symposium, "Immunological Aspects of Treatment of Pompe Disease," Miami, FL, February 2010.
118. Invited speaker for 13th North American Lysosomal Storage Disease (LSD) Registries Meeting. Panel session: "Increase knowledge, Improving Outcomes: The Evidence for Early Intervention," Dallas, TX, May 2010.
119. Invited guest speaker at KEM Hospital "Updates in Pompe disease," Mumbai, India August 2010.
120. Invited speaker at ENMC Workshop on Pompe "Long-term Outcome in Infantile Survivors of Pompe Disease," Naarden, - Netherlands, September 2010.
121. Invited speaker at AGSD Annual Conference, "Development of guidelines for GSD's and recent advances in the field," Orlando, FL, October 2010.
122. Co-chair and invited speaker at FDA/NIH Rare Disease Investigators Training Course, "Role of Urinary Hex4 as a Biomarker for Pompe Disease," Rockville, MD, October 2010.
123. Invited speaker for 4th European Symposium at Steps Forward in Pompe Disease "Updates on Immunological Aspects of Treatment of Pompe Disease," London, UK, November 2010.
124. Co-Chaired American College of Medical Genetics "Newborn Screening Translational Research Network Meeting," Las Vegas, NV, February 2011.
125. Invited speaker for Lysosomal Disease Network (LDN) 7th Annual WORLD Symposium, "The Pompe Registry: Baseline Data from the First Five Years," Las Vegas, NV, February 2011.
126. Invited member for the NICHD Workshop on Cognition, Bethesda, MD, March 2011.
127. Invited guest speaker for Jaikei University "Immune Modulation in Pompe disease, Issues at Hand," Tokyo, Japan, June 2011.
128. Invited speaker for Jaikei University "Pompe Disease – Updates and Emerging Issues," Tokyo, Japan, June 2011.

129. Invited speaker for Patient Symposium for Pompe Disease, "Lessons Learned from Pompe Disease," Tokyo, Japan, June 2011.
130. MUSC Grand Rounds, "Clinical and Research Updates on Pompe Disease," Charleston, SC, July 2011.
131. Invited speaker for 33rd Annual Association for Glycogen Storage Disease "GSD 101;" "Care Guidelines Update (all types);" Breakout Session #1: Research Reviews and Updates Type 0, III, VI, and IX;" Breakout Session #2 Teens/Young Adult GSD," Indianapolis, IN, September 2011.
132. Invited speaker for Drug Information Association (DIA) 1st Annual Rare Diseases & Orphan Products Summit, Washington, DC, October 2011.
133. Invited guest speaker at the Carolina Institute for Developmental Disabilities Investigator Forum Series 2011-2012, "Clinical Trials to Enhance Cognition in Down Syndrome," Chapel Hill, NC, December 2011.
134. Invited speaker for Steps Forward in Pompe Disease, "Registry data on current practices of diagnosing LOPD and infantile Pompe disease: Emphasis on the need for early recognition;" "Immunological Challenges in Pompe Disease," Budapest, Hungary, December 2011.
135. Invited speaker for Indian Association for Study of the Liver (INASL) Mid-Term Single Theme Conference – Metabolic Liver Disease, "Metabolic liver disease – getting the definitions right;" "Glycogen storage disorders and the liver;" "How do I interpret tests for Metabolic liver disease?" Mumbai, India, January 2012.
136. Invited presenter for Lysosomal Disease Network (LDN) 8th Annual World Symposium, "Long Term Outcome and Clinical Experience on Immune Tolerance Induction Therapies in Infantile Pompe Disease" "Characteristics Associated with Delays in Diagnosis of Pompe Disease Among Patients Enrolled in the Pompe Registry," San Diego, CA, February 2012.
137. Invited speaker for 2012 Annual Clinical Genetics Meeting (ACMG), "Cardinal Signs and Symptoms of Common and Rare Important Inborn Errors of Metabolism," Charlotte, NC, March 2012.
138. Invited speaker for 2012 Society for Inherited Metabolic Disorders (SIMD), "The use of non-depleting anti-CD4 monoclonal antibody for immune tolerance induction in Pompe disease," Charlotte, NC, March 2012.
139. Lectures and Case Presentations at Instituto Fernandes Figueira (IFF), Brazil, "Duke University Hospital experience on Pompe disease – Diagnosis, Treatment and Management and Immunomodulation Discussion," Rio de Janeiro, Brazil, April 2012.
140. Lectures and Case Presentations in Brazil at IGEIM – Genetic and Inborn Errors of Metabolism Institute, "Patient Evaluation: Duke Experience and multidisciplinary discussion," San Paulo, Brazil, April 2012.
141. Lectures and Case Presentations at AACD Symposium, "Duke University Hospital experience on diagnosis, treatment and overall management of Pompe disease," Sao Paulo, Brazil, April 2012.
142. Speaker for Challenging the Paradigms: Liver Transplantation for Metabolic Disease at Children's Hospital of Pittsburgh of UPMC, "Glycogen Storage Disease," Pittsburgh, PA, May 2012.
143. Expert Speaker for FDA PMC Meeting, "Success with Immune Modulation Protocols in naïve setting

and in entrenched immune response setting in infantile Pompe disease," Washington, DC, May 2012.

144. Speaker and Chairperson for the Workshop on Natural History Studies for Rare Diseases: Meeting the Needs of Drug Development and Research at NIH, "The Role of Retrospective Chart Reviews In Natural History Studies: Lessons Learned From Pompe Disease" Bethesda, MD, May 2012.
145. Invited Presenter for Myozyme™ Launch South Africa, "Clinical Updates in Pompe disease," Johannesburg, SA, June 2012.
146. Invited Presenter for Myozyme™ Launch South Africa, "Lysosomal Storage Disease updates and therapies on the horizon," Durban, SA, June 2012.
147. Invited Presenter for Myozyme™ Launch South Africa, "A treatable cardiomyopathy: Pompe disease clinical findings and updates," Cape Town, SA, June 2012.
148. Invited speaker at Portland Pompe Conference, "Clinical and Research Updated in Pompe Disease," Portland, OR, July 2012.
149. Invited speaker for annual Association of Glycogen Storage Disease (AGSD) Annual Conference, "On the Horizon – Type 2 & 3," Las Vegas, NV, August 2012.
150. Invited speaker for The 2nd Middle East Lysosomal Storage Disease Meeting, "Updates in Pompe Disease", Dubai, October 2012.
151. Speaker at Indian Association of Neurology Annual Conference (IANCON 2012), "Treatable Neuromuscular Disorders in Pompe Disease", Ahmedabad, INDIA, October 2012.
152. Invited speaker for DIA/NORD's US Conference on Rare Diseases & Orphan Products, "Policy and Special Challenges in Rare Diseases: Role of Academic Centers in Orphan Product Development, Washington, DC, October 2012.
153. Invited Speaker for DIA/NORD's US Conference on Rare Diseases & Orphan Products, "Research and Regulation: Immune Tolerance", Washington, DC, October 2012.
154. Invited Speaker for Genzyme Dinner Symposium at the American Society of Human Genetics (ASHG) Symposium, "Addressing the Needs of Newly-Diagnosed Patients with Lysosomal Storage Diseases: San Francisco, November 2012.
155. Neuromuscular Grand Rounds Speaker at University Hospitals Case Medical Center, "Pompe disease: A treatable Neuromuscular Disease", Cleveland, OH, December 2012.
156. Genetics Grand Rounds Speaker at University Hospitals Case Medical Center, "Clinical Updates in Pompe disease, Cleveland, OH, December 2012.
157. Co-Chair on Translational Research Session and Invited Presenter at Lysosomal Disease Network WORLD Symposium, "Approach to Management of Cross-Reactive Immunologic Material (CRIM) – Negative Infantile Pompe Patients Treated with ERT: Role of Immune Modulation in Changing the Natural History", Orlando, FL, February 2013.
158. Panelist at Medscape Neurology CME Activity, Town Hall Live Symposium, "Advances in Treatment of Lysosomal Diseases in Today's World: Translating Science in to Practice", at Lysosomal Disease Network WORLD Symposium, Orlando, FL, February 2013.

159. Invited speaker at Medigencon Program at Azad Medical College, "Therapeutics in LSDs – strides", New Delhi, INDIA, February 2013.
160. Invited speaker at Medigencon Program at Azad Medical College, "Pompe Disease – unfolded: New Delhi, INDIA, February 2013.
161. Invited speaker and Chair at ACMG Annual Clinical Genetics Meeting, "Hypophosphatasia: Clinical Expression and Differential Diagnosis of Genetic Metabolic Bone Disorder", Phoenix, AZ, March 2013.
162. Invited speaker at ACMG Annual Clinical Genetics Meeting, "HCA Happenings –Lysosomal Storage Disorders: Beyond the Classic Phenotype", Phoenix, AZ, March 2013.
163. Invited speaker at 2nd Working Group on Aldurazyme use post-transplant Genzyme Meeting, Amsterdam, Netherlands, May 2013.
164. Invited speaker at Soft Bones, an HPP Patient Group First Educational Patient Meeting, "Hypophosphatasia: Current Knowledge and Clinical Update", Chicago, IL, June 2013.
165. Invited speaker at UC Irvine Neurology Grand Rounds, "Bench to Bedside Research in Pompe Disease", Orange, CA, June 2013.
166. Invited speaker at Miller Children's Hospital UCI/CHOC Pediatrics Residents, "Clinical Update in Liver Glycogen Storage Disease (GSDs)", Long Beach, CA, June 2013.
167. Invited speaker at Pompe Patient and Family Meeting at UC Irvine & United Pompe Foundation, "Glycogen Storage Diseases – An Overview", Marian Del Rey, CA, June 2013.
168. Invited speaker for Genzyme Pompe Patient Meeting, "LOPD – "Emerging Issues", San Jose, CA, June 2013.
169. Invited speaker at Kaiser Santa Clara Facility for Pompe Disease HCP Program, "Pompe disease update to Healthcare Providers at Kaiser", Santa Clara, CA, July 2013.
170. Invited speaker for Children's Hospital of Oakland, CHORI Seminar Services – "Pompe Disease", Oakland, CA, July 2013.
171. Invited speaker for Arkansas Children's Hospital CHAR Lectureship, "Pompe Disease: a treatable Neuromuscular Disorder, Little Rock, AR, July 2013.
172. Invited speaker for the NYMAC Pompe Disease Newborn Screening Symposium; "Infantile Pompe Disease: Diagnosis & Management, Westchester Medical Center, Tarrytown, NY, November 2013.
173. Invited presenter for Grand Rounds at the 3rd Annual UCLA Intercampus Lysosomal Disease Symposium, "Pompe Disease: Clinical Research and Updates, Los Angeles, CA, December 2013.
174. Invited panelist for the Children's Liver Foundation and Indian Academy of Pediatrics, "Liver failure in a neonate", "Hepatomegaly and seizures – what is the link?"; "Recurrent Vomiting", Splenohepatomegaly – How do we approach?"; Parel, Mumbai, INDIA, December 2013.
175. Invited speaker for IAP, Pune in collaboration with Genzyme India, CME on Lysosomal Storage Disorders, "Diagnosis & Management of Lysosomal Storage Disorders, Pune, INDIA, December 2013.
176. Invited speaker for IAP, Nagpur in collaboration with Genzyme India, CME on Lysosomal Storage Disorders, "Diagnosis & Management of Lysosomal Storage Disorders, Nagpur, INDIA, December

2013.

177. Invited speaker for Genetics Chapter, IAP in collaboration with Genzyme India, CME on Lysosomal Storage Disorders, "Diagnosis & Management of Lysosomal Storage Disorders: Janpath, New Delhi, INDIA, December 2013.
178. Invited speaker for Lysosomal Storage Disease Network World Symposium, "Emerging Natural History of Cross-reactive Immunologic Material (CRIM) Negative Infantile Pompe Disease patients treated with Recombinant Human GAA", San Diego, CA, February 2014.
179. Invited speaker for India Charitable Access Program (INCAP) at the IMAB Meeting, "Presentation of data on infantile Pompe in India", Bengaluru, INDIA, February 2014.
180. Invited speaker for India Charitable Access Program (INCAP) at the IMAB Meeting, "Updates in Management of Pompe Disease", Bengaluru, INDIA, February 2014.
181. Invited speaker for 2nd National Symposium Approach to Rare, Treatable Genetic Diseases for Medigencon 2014, "Pompe disease: Past, present and future", Bengaluru, INDIA, March 2014.
182. Invited speaker for American College of Medical Genetics (ACMG), "Diagnosis of Inherited Metabolic Bone Disorders; The Critical Role of the Medical Geneticist", Nashville, TN, March 2014.
183. Invited presenter for Alexion Hypophosphatasia Advisory Board Meeting, "HPP Disease Overview/Advisor Feedback, Denver, CO, May 2014.
184. Invited presenter for Alexion Hypophosphatasia Advisory Board Meeting, "Adult HPP Cases", Denver, CO, May 2014.
185. Invited speaker for Food and Drug Administration (FDA)-Center for Drug Evaluation and Research (CDER) Workshop on Immune Responses to Enzyme Replacement Therapies: Role of Immune Tolerance Induction, "Immune responses to enzyme replacement therapy (ERT), Silver Spring, MD, June 2014.
186. Invited facilitator and speaker for Newborn Screening Advisory Board Meeting, "Roles, Responsibilities, and Timing for IOPD Patient Follow-up, San Diego, CA, June 2014.
187. Invited presenter for Texas Department of State Health Services Newborn Screening Grand Rounds, "Pompe Disease: Bench to Bedside Journey and Newborn Screening Initiatives, Austin, TX, July 2014.
188. Invited speaker and panelist for Action Plan Workgroup 2 member for Muscular Dystrophy Coordinating Committee Muscular Dystrophies Workshop, "Diagnosis, Screening, and Biomarkers for Muscular Dystrophy", Bethesda, MD, July 2014.
189. Invited speaker for Department of Pediatrics at Armed Forces Medical College, "Newborn Screening", Pune India, August 2014.
190. Co-chair of the Newborn Screening Translational Research Network (NBSTRN) LSD Workgroup Meeting, Bethesda, MD, September 2014.
191. Facilitator and Presenter for Alexion Hypophosphatasia Registry Scientific Advisory Board Meeting, "Adult case presentation: how sure are we of nomenclature", London, UK, September 2014.
192. Chair and Presenter for 7th European Symposium Steps Forward in Pompe Disease, "Improving the

management of Pompe disease: Immunological factors in Pompe Disease Management: Clinical experience and implications for newborn screening, Turin, Italy, November 2014.

193. Invited speaker for 11th Annual Lysosomal Disease Network World Symposium, "Prophylactic immune modulation in infantile Pompe disease; collective experience treating CRIM-positive and negative patients in the naïve setting", Orlando, FL, February 2015.
194. Invited speaker for Medigencon, "Glycogen Storage Diseases – Advances in management and Pompe Disease – Ask the Expert", New Delhi, India, February 2015.
195. Invited speaker for IMAB, "Newborn Screening for Pompe Disease", New Delhi, India, February 2015.
196. Invited speaker for Brazil Genzyme Pompe Disease National Meeting, "Challenges and controversies in the diagnosis of Pompe Disease – Experience at Duke University, Rio de Janeiro, Brazil, March 2015.
197. Invited speaker for Brazil Genzyme Pompe Disease National Meeting, "Immunomodulation and multidisciplinary management in Pompe disease – Clinical Experience at Duke University, Rio de Janeiro, Brazil, March 2015.
198. Invited Presenter at Pompe Round Table, "Infantile Pompe Disease – CRIM and Immune Modulation, the emergent phenotype", Buenos Aires, Argentina, April 2015
199. Invited Presenter at Pompe Round Table, "Pompe Disease, What have you learned? New Challenges", Buenos Aires, Argentina, April 2015
200. Invited participant in Roundtable Discussion at Japan Pompe Meeting, "Significance of Early Treatment in IOPD and Opportunities for Newborn Screening, Tokyo, Japan, June 2015.
201. Invited presenter at the Japan Pompe Meeting, "Immunotolerance Induction for ERT in IOPD, Tokyo, Japan, 2015.
202. Keynote Presenter at the Symposium on ATP1A3 in Disease, "First-hand experience of a clinical trial in a rare disease, Washington, DC, August 2015.
203. Invited presenter at the Meet-the-Expert Session at SSIEM Symposium, "Improving outcomes in Pediatric – onset Hypophosphatasia, Lyon, France, September 2015.
204. Invited presenter at the Pompe Newborn Screening Meeting, "National Pompe Newborn Screening Update, New York, NY, October 2015.
205. Invited speaker for the AMDA/IPA Pompe Patient and Scientific Conference, "ERT: What Have We Learned So Far Roundtable, San Antonio, TX, October 2015.
206. Invited speaker for IAMGCON 2015 2nd Annual Meeting of Society for Indian Academy of Medical Genetics, Dr S Agarwal Oration "Novel Therapies for Genetic Disorders, Jodhpur India, December 2015.
207. Invited speaker for IAMGCON 2015 2nd Annual Meeting of Society for Indian Academy of Medical Genetics, Diagnostic Revolutions Session "Pompe Disease – Importance of early diagnosis", Jodhpur India, December 2015.
208. Invited presenter at "Stay the Course to Success" – 1st Off Site Meeting for the Liver & Metabolic Therapeutic Area Team at uniQure, Introduction to the top 3 liver disease indications and brief introduction to the tier 2 liver disease indications, Amsterdam, Netherland, January 2016.

209. Invited Presenter at LDN World Symposium, "New perspective for ERT in Pompe disease: extending the action of the enzyme to cytosolic targets, San Diego, CA, March 2016.
210. Invited Presenter at LDN World Symposium, Amicus Satellite Dinner Symposium, "The Many Faces of Lysosomal disease: A Global Perspective", San Diego, CA, March 2016.
211. Invited speaker at the 98th Annual Meeting of the Endocrine Society (ENDO 2016), "Biochemical and Physical Function Outcomes in Adolescents and Adults with Hypophosphatasia Treated with Asfotase Alfa for up to 4 years: Interim Results from a Phase II Study, Boston, MA, April 2016.
212. Invited speaker at the Society of Inherited Metabolic Disease (SIMD) Annual Meeting, "Ask the Experts Table" – Lysosomal Disorders, Ponte Vedras Beach, FL, April 2016.
213. Invited speaker at the Society of Inherited Metabolic Disease (SIMD) Annual Meeting, "Diet Therapy in GSD", Ponte Vedras Beach, FL, April 2016.
214. Invited speaker at the NDSC DSMIG USA Meeting, Pharmacological Interventions to Improve Cognition and Adaptive Functioning in Down Syndrome: Strides to Date", Orlando, FL, July 2016.
215. Invited Speaker by Baebies to present at the FDA Meeting, "Lysosomal storage disorder", Gaithersburg, MD, August 2016.
216. Invited speaker for the SSIEM Annual Conference, Alexion Satellite Symposium, "Improving functional outcomes in adolescents and adults with hypophosphatasia", Rome, Italy, September 2016.
217. Invited Presenter at the Association of Glycogen Storage Disease Annual Meeting, "GSD II Newborn screening & GSD III and IV Mouse Model – what we have learned", Toronto, Ontario, Canada, September 2016.
218. Lead Discusser for 2nd South Asia LSD Symposium, "Symposium 1: LSDs in South Asia. Regional Perspectives, and "Symposium 4: Pompe Disease", New Delhi, India, September 2016.
219. Chairperson for 2nd South Asia LSD Symposium, "Symposium 4: Pompe Disease", New Delphi, India, September 2016.
220. Plenary Speaker for 2nd South Asia LSD Symposium – "Present status & new frontiers in Pompe Disease", New Delhi, India, September 2016.
221. Invited Speaker and leader (introductions, closing remarks and wrap up) for Alexion's HPP Registry Scientific Advisory Board and Alexion Supporter Hypophosphatasia and Physician Education (SHaPE), "Adults Clinical Data"; "HPP Patients, Case Study Session"; "Case Studies: Asfotase alfa in clinical practice and the patient perspective"; Q & A for The HPP Registry, Prague, Czech Republic, October 2016.
222. Invited Presenter at 8th Steps Forward in Pompe Disease, "Immunomodulation in Infantile Pompe Disease", Schiphol, Amsterdam, Netherlands, November 2016.
223. Invited lecture at St. Xavier's College, "Today's Science is Tomorrow's Future – bench to bedside experience with rare diseases", Mumbai, India, February 2017.
224. Invited speaker at ISIEM 2017 (IV National Conference of the India Society of Inborn Errors of Metabolism, "Sandor Oration Award: Bench to bedside experience in developing a treatment for Pompe

Disease: The Journey Continues", Chennai, India, February 2017.

225. Invited presenter for the 13th Annual WORLD Symposium, Satellite Symposium, "Newborn Screening for Lysosomal Diseases: Recent Progress and Unanswered Questions", San Diego, CA, February 2017.
226. Invited presenter for Satellite Symposium at the Annual ACMG 2017 Meeting, "Pompe Disease: Early Diagnosis and Optimal Management, Phoenix, AZ, March 2017.
227. Invited Faculty Member for Round Table Workshops at the Gaucher Leadership Academy: Substrate Reduction Therapy Eliglustat Clinical Efficacy, and invited speaker on Eliglustat in Switch Patients – real world experience", Mainz, Germany, June 2017.
228. Invited presenter for three sessions at the IGSD 2017 Annual Meeting, "Causes & Development of Myopathy in GSD IIIa", "Immunomodulatory enzyme replacement therapy using tolerogenic nanoparticles containing rapamycin for Pompe Disease", Population Newborn Screening for Pompe Disease", Groningen, The Netherlands, June 2017.
229. Invited speaker for the Association of Public Health Laboratories (APHL) National Newborn Screening Meeting on New Disorders (Pompe, MPS I, X-ALD), "Clinical Considerations: Pompe Disease: Bethesda, MD, June 2017.
230. Invited speaker for Shire's R & D Day Research Series: Shaping What's Next in LSDs – Hot Topics, "Immune tolerance induction for patients who have developed ADAs to ERT: London, UK, July 2017.
231. Invited faculty speaker for IANCON 2017 Meeting Symposium XVI: Neurology and Humanities Session talk on "Pompe disease, Chennai INDIA, September 2017
232. Invited speaker for the 2nd MMM Genetics Meeting – a symposium on "Genetics and Genomics in Cardiovascular Diseases – 'Cardiac Manifestations in Pompe Disease-Infantile and Late Onset, Chennai INDIA, September 2017.
233. Invited speaker for the annual AGSD Conference, "GSD VI &IX Diagnosis and Management Guidelines", Chicago, IL, September 2017.
234. Invited speaker for the annual ASHG Conference, "Clinical and Translational Insights in Pompe Disease: Disease Pathology, Patient Identification, and Treatment, Orlando, FL, October 2017.
235. Invited leader and presenter for Alexion's HPP Registry SAB Meeting, "A Review of the best practices to report CRIM data in relation to the genotype," Madrid SPAIN, November 2017.
236. Invited presenter for the 14th Annual WORLD Symposium, Satellite Symposium – Excel Continuing Education Talk, "Overcoming the challenges of Pompe Disease", San Diego, CA, February 2018.
237. Invited presenter for the 14th Annual WORLD Symposium, Satellite Symposium, Early Treatment of Lysosomal Disorders: A closer look at Fabry and Pompe disease, "What have we learned about early treatment of Pompe Disease", San Diego, CA, February 2018.
238. Invited opening speaker for The Council of Entrepreneurial Development (CED), the North Carolina Biotechnology Center, and North Carolina Biosciences Organization (NCBIO), CED Life Science 2018 Conference, "Innovation Delivers A Better Tomorrow: The Pompe Journey", Raleigh, NC, February 2018.

239. Chair of CME Accredited Roundtable Discussion for EOCME/Exerpta Medica, "Focus on Lysosomal Diseases: New Frontiers for Accelerating Diagnosis and Optimizing Treatment", Durham, NC, June 2018.
240. Invited speaker for the HPP Soft Bones Meeting, "Role of Functional Testing in Adults patients with Hypophosphatasia (HPP), Chicago, IL, June 2018.
241. Invited presenter at the 40th Annual Association for Glycogen Storage Disease Conference, "GSD1a Research Updates", Davenport, IA, September 2018.
242. Invited speaker at the 5th Annual Conference of Indian Society of Pediatric Gastroenterology, Hepatology and Nutrition and the 28th Annual Conference of the Pediatric Gastroenterology Chapter of IAP, "Diet in GI/Liver disease: Culprit? Saviour? Or both?" Mumbai INDIA, October 2018.
243. Panel Participant at the Indian Medical Advisory Board (IMAB), "Review and Status updates of INCAP cases under respective members and centers", New Delhi INDIA, October 2018.
244. Invited presenter at the Steps Forward in Pompe Disease 2018, "Immune-modulation in infants treated with ERT with alglucosidase alfa: Global experience over a decade of learning", Copenhagen, Denmark, November 2018.
245. Invited speaker for Pompe Registry North American Board Meeting, "Infantile early treatment – WG analysis: Review of IOPD early treatment analysis", Toronto, CANADA, November 2018.
246. Invited faculty presenter for the 15th Annual LDN World Symposium Genzyme/Med-IQ Satellite Symposia. "Bridging Gap between Research and Clinical Advances in Gaucher Disease Type 3", Orlando, FL, February 2019.
247. Invited presenter for the 15th Annual LDN World Symposium Satellite Symposium, "Biomakers: Ready for Prime Time in the Clinical Management of Lysosomal Disorders?" Orlando, FL, February 2019.
248. Co-Chair for the 15th Annual LDN World Symposium for the Translational Research IB Session, "Implementation and Impact of Newborn Screening, Orlando, FL, February 2019.
249. Invited presenter for the 15th Annual LDN World Symposium Pompe Satellite Symposium, "Hand in Hand --- Patient and Physicians Journey in Pompe Disease", Orlando, FL, February 2019.
250. Invited speaker for the LuMind and Alzheimer's Association and the Global Down Syndrome Foundation co-sponsored Alzheimer's & Down Syndrome Workshop, Session 5: Practical Considerations for Clinical Trials, "Inclusion/Exclusion Criteria Considerations, Bethesda, MD, March 2019.
251. Invited speaker for the LuMind and Alzheimer's Association and the Global Down Syndrome Foundation co-sponsored Alzheimer's & Down Syndrome Workshop, Session 5: Practical Considerations for Clinical Trials, "Inclusion/Exclusion Criteria Considerations, Bethesda, MD, March 2019.Presenter for ACMG Annual Clinical Genetics Meeting Platform Presentations – Clinical Genetics and Therapeutics, "Factors Affecting Outcomes in Classic Infantile Pompe Disease Patients Treated with Enzyme Replacement Therapy: Lessons Learned from the Pompe Registry", Seattle, WA, April 2019.
252. Presenter, Welcome and Introduction, Case Studies Panel for ACMG Annual Clinical Genetics Meeting Satellite Symposia – Improving Care for Patients with Pompe Disease: A Look at Emerging Therapies

and Multidisciplinary Management Strategies, Seattle, WA, April 2019.

253. Presenter for ACMG/SIMD Joint Plenary Session Part 1 – Thinking Metabolic for the Genetics Clinician: You Took the Call from the Department of Health: What Now? "You got the NBS call for presumptive positive Pompe. What Now? Seattle, WA, April 2019.
254. Invited Presenter for 18th Annual Asian Oceanian Myology Center Meeting (AOMC) for Session VIII "Pompe", Mumbai INDIA, June 2019.
255. Invited Speaker for Gene Connect Masterclass a series of webinars, "Pompe Disease: Recognize Early – Treat Early, Lessons learned from New Born Screening", Mumbai INDIA, June 2019.
256. Invited Speaker for the 19th Annual Meeting of the Federation of Clinical Immunology Societies (FOCIS) Thematic Session, "Immune Barriers to Protein/Gene Replacement Therapies – "Response Antibodies Challenges in Pompe Disease", Boston, MA, June 2019.
257. Invited presenter at the 5th International Forum of Lysosomal Disorders – Pompe Workshop, "Treatment of Pompe Disease", Tokyo JAPAN, July 2019.
258. Invited Panelist for Amicus Newborn Screening Summit, "Ethical Considerations for Newborn Screening and "Newborn Screening Initiatives: Going Beyond State NBS, Webex, July 2019.
259. Invited presenter at the 41st Annual Association for Glycogen Storage Disease Conference, "GSD III", Houston, TX, September 2019
260. Invited presenter at the 2019 AMDA/IPA International Pompe Patient and Scientific Conference, Session: "Natural History of Pompe vs. New History of Treatment in infantile onset Pompe disease", San Antonio, TX, October 2019
261. Invited Chair at the 2019 AMDA/IPA International Pompe Patient and Scientific Conference, Session: The Basics of Clinical Trials; Member of Expert Roundtable: "20 Years of Treatment and Care of Pompe Patients"; and Session: "Next Generation Therapy: Enzyme Replacement Therapy (ERT) – Update on Valerion's approach", San Antonio, TX, October 2019
262. Invited Plenary Speaker at the 5th International GSD Conference (IGSD2019), Plenary Title: "Ketotic Hepatic GSD", Porto Alegre, Brazil, November 2019
263. Invited Plenary Speaker at the 5th International GSD Conference (IGSD2019), Plenary Title: "Gene Therapy for Pompe disease: What's new?" Porto Alegre, Brazil, November 2019.
264. Invited Session Speaker at the 5th International GSD Conference (IGSD2019), Session Title: "Immunological and treatment aspects in high sustained rhGAA antibodies in Pompe patients", Porto Alegre, Brazil, November 2019
265. Invited Session Speaker at the 5th International GSD Conference (IGSD2019), Session Title: "First-in-human study of ATB200/AT2221 in patients with Pompe disease: 24 months functional assessment results from ATB200-02 trial", Porto Alegre, Brazil, November 2019
266. Invited Speaker, Session Chairperson and Panelists at the Indo-US Symposium on Genetic Neuromuscular Disorders & Sixth Annual National Conference of the Society for Indian Academy of Medical Genetics (SIAMGCON 2019), "Understanding the natural history of muscle involvement in

Glycogen storage disorders" and "Metabolic manipulation with Pompe disease prototype"; Chairperson for Session – "Emerging therapies for genetic neuromuscular disorders; Panelist on Panel discussion – "Creation of an Indian and Indo-US Consortium on Genetic Neuromuscular Disorder, Hyderabad, India, November 2019

267. Invited speaker at Taipei Veterans Hospital – "Management of pre-symptomatic patients with late onset Pompe Disease", Taipei Taiwan, January 2020
268. Invited Speaker at satellite symposium at The LDN World Symposium, "Gaucher disease: Can we build a better roadmap for patient care"?, Orlando, FL, February 2020
269. Invited Speaker at satellite symposium at the LDN World Symposium, "Respiratory Involvement in Pompe disease", Orlando, FL, February 2020
270. Co-Chair of Clinical Trials II Session for the LDN World Symposium, "Clinical Outcomes", Orlando, FL, February 2020
271. Invited Speaker at the Virtual ACMG Digital Edition– "Lessons Learned: The Role of NBS for Pompe Disease and Mucopolysaccharidosis Type 1: Early Intervention Influences outcome", April 2020
272. Co-Chair for Session "Pediatric/Development Conditions" for the National Institute of Health (NIH) Clinical Trials in Down syndrome for Co-Occurring conditions across the lifespan virtual workshop, May 2020
273. Invited Speaker for the National Institute of Health (NIH) Clinical Trials in Down syndrome for co-occurring conditions across the lifespan virtual workshop, "Overview of Clinical Trials in the Down syndrome population", May 2020
274. Moderator for Panel "Consideration for Participation in Clinical Trials in Pediatric and Adult Populations" for the National Institute of Health (NIH) Clinical Trials in Down syndrome for co-Occurring conditions across the lifespan virtual workshop, May 2020
275. Invited presenter for Rare Disease Day, JPAC (Japan, Australia, and South Korea) Pompe Virtual Learning Summit, "The Story Behind Development of a Treatment for Pompe Disease and the importance of early diagnosis" May 2020
276. Invited speaker for the Virtual Conference on Lysosomal Diseases (Med-Lysosomal 2020), Jerusalem, Israel, "Immune modulation for patients receiving therapeutic proteins", June 2020
277. Presenter at the Virtual MDA Pompe Disease Patient-Focused Drug Development Meeting, "Scientific Overview of Pompe disease across the disease spectrum", July 2020
278. Invited Presenter at the Virtual Online Symposium for International Congress on Neuromuscular Diseases (ICNMD), "Insights into the Phenotypes of Pompe Disease", September 2020
279. Association for Glycogen storage disease Virtual Annual Conference, "Updates in Glycogen Storage Disease Type III, September 2020
280. Invited Lecturer for hypophosphatasia Lecture Series Virtual "Understanding Immunogenicity to therapeutic proteins", October 2020
281. Keynote Speaker for the New England Consortium of Metabolic Programs Virtual Annual Meeting,

"Updates and Insights into Hepatic Glycogen Storage Disorders", November 2020

282. Invited Speaker for the Virtual AMDA (Acid Maltase Deficiency Association) – "Efficacy of the Vaccines for COVID-19 Vaccines for Pompe Patients", January 2021
283. Invited Speaker for North American Rare Disease Registries Virtual Meeting, "Newborn Screening for Pompe Disease in the US: Early Findings from the 2020 Pompe Registry Data Analysis, January 2021
284. Serve as a Panelist for the Translation Session at the NIMH Virtual Workshop Gene-based Therapeutics for Rare Genetic Neurodevelopmental therapeutics, "What aspects of success stories can be translated to new neurodevelopmental therapeutics?", January 2021
285. Invited Speaker at The LDN World Virtual Symposium, Late Breaking Science Session, "Efficacy and safety results of the avalglucosidase alfa phase 3 COMET trial in late-onset Pompe disease patients", February 2021
286. Invited Speaker for the Virtual Immunogenicity Advisory Board, "Discussion on immune strategies to be considered in Hypophosphatasia treated with asfotase alfa", March 2021
287. Distinguished Speaker Panelist for the WuXi AppTec Rare Disease Webinar, "Closing the Gap in Pompe Disease: Where Are We Heading", Webinar, April 2021
288. Speaker for the Indian Society for Inborn Errors of Metabolism (ISIEM) and Indian Academy of Medical Genetics (IAMG) sponsored 4th South Asia LSD Symposium, Pompe Disease Session "Rx outcomes in IOPD with treatment initiation in 1st month of life", Virtual, April 2021
289. Presenter at the Lysosomal Disease Network Annual Meeting, Clinical Research Studies Session, "Pompe Disease Study", Virtual, April 2021
290. Speaker at the 16th International Congress on Neuromuscular Diseases (ICNMD), " A journey of long-term commitment in the Pompe Community", Virtual, May 2021
291. Planning Committee and Speaker for NIH Workshop (NCATS) on Early Diagnostic Strategies to Identify Patients for Gene-Targeted Therapies Meeting, June 2021
292. Grand Round speaker for the UPMC Children's Hospital of Pittsburg Pediatric Grand Rounds, "Lessons Learned from Rare Diseases: Bench to Bedside and Back", June 2021
293. Invited Speaker at Sanofi Genzyme Pompe Academy 2021 Meeting, "Early Treatment for Pompe Disease: Lessons learned from NBS", Virtual, June 2021, Argentina
294. Speaker during the Pompe Session of the APHL (Association of Public Health Laboratories) hosted New Disorders and Short-term Follow-up Virtual National Meeting, "Treatments and Therapies for Newborn Screening Conditions", August 2021
295. Invited participant in the FDA CBER 2-day Advisory Committee on Gene Therapy toxicity risks of AAV Therapy, September 2021
296. Talk for Indian Society of Inherited Errors of Metabolism (ISIEM) and Indian Academy of Medical Genetics (IAMG) South Asia Gaucher Summit Celebrating 30 Years of Gaucher Care, "Journey so far….!!!", October 2021
297. Presenter for the Hybrid Patient Forum sponsored by the Australian Pompe Association – Australia and

New Zealand Pompe Down-Under, “Infantile Pompe Disease and Newborn Screening”, virtual, October 2021

298. Participant in the Virtual FDA-CERSI Workshop: "Prenatal Somatic Cell Gene Therapies -- Charting a Path Forward for Clinical Applications”, October 2021
299. Panelist for NORD Rare Disease Center for Excellence Corporate Council Meeting – Access to Care discussions on “Collaborative Science to Address unmet needs in Rare Diseases and Disease Management Guidelines”, virtual, November 2021
300. Cedar-Sinai Keynote Talk for the 11th Annual Symposium of Lysosomal Storage Disease, “Updates in Pompe Disease”, virtual, December 2021
301. Keynote Speaker at Sanofi Genzyme 14th International Lysosomal Disease Web Conference, “Updates in Pompe Disease”, virtual, January 2022
302. Platform Speaker for 18th Annual WORLD Symposium, Late-Breaking Science Session, “Avalglucosidase alfa improves health-related quality of life (HRQoL) in patients with late-onset Pompe disease (LOPD) vs. alglucosidase alfa: Patient-reported outcome measures (PROMs) from the phase 3 COMET trial, San Diego, CA, February 2022
303. Participated as a panelist for the Accredited Satellite Symposium, “Improving Diagnosis and Enhancing the Standard of Care for Pompe Disease”, ACMG Annual Clinical Genetics Meeting, Nashville, TN, March 2022
304. Presented as the 2021 Emmanuel Shapira Award Winner for the SIMD Annual Meeting, “Investigation of ALPL variants states and clinical outcomes: An analysis of adults and adolescents with Hypophosphatasia treated with asfotase alfa”, SIMD Annual Meeting, Orlando, FL, April 2022
305. Speaker for Satellite Symposium, “Exploring Next Generation ERT for Pompe Disease: Strategies for Individualizing Treatment Decisions”, SIMD Annual Meeting, Orlando, FL, April 2022
306. Invited Speaker for at the Organization for Rare Disease India (ORD) Pompe Foundation International Pompe Day Virtual Webinar, “Management of Pompe – Yesterday, Today And Tomorrow”, April 2022
307. Invited speaker at the BIO and NC BIO and in partnership with Rare Disease Innovations Institute, Inc. legislative breakfast educating legislators on Gene & Cell Therapy & Access, “Transforming the clinical outcomes of individuals with Rare diseases: The Gene Therapy Journey, at the Legislative Building, Raleigh, NC, June 2022
308. Invited didactic speaker for Soft Bones Project ECHO-TeleECHO, “Functional Assessments of Hypophosphatasia in Adults”, virtual seminar, July 2022
309. Invited Keynote speaker for the annual SERN-SERGG meeting, “New Insights into Hepatic Glycogen Storage Disease”, Asheville, NC, July 2022
310. Invited session speaker for the annual SERN-SERGG meeting, “Long-term Follow-up of Cipaglucosidase Alfa/Miglustat in Ambulatory Patients with Pompe Disease: An Open-label Phase I/II Study (ATB200-02), Asheville, NC, July 2022
311. Invited presenter for the Sanofi sponsored Satellite Symposium for the Society for the Study of Inborn

Errors of Metabolism (SSIEM), "Pompe disease, a new era: the latest avalglucosidase alfa clinical data and early real-world experiences", Freiburg, Germany, August 2022

312. Invited presenter for NORD Center of Excellence for Rare Disorders at Boston Children's Hospital sponsored Combined Pediatrics and Metabolism Grand Rounds, "New Insights into hepatic GSDs: clinical implications and a need for definitive therapies, Virtual, September 7, 2022
313. Virtual Presenter at the National Italian GSD Patient Association GSD/Pompe Conference, "Pompe Disease Overview and Recent Updates", Bari, Italy, October 2022
314. Invited Speaker at the Annual APS-Jahrestagung 2022 Hepatic Glycogen Storage Diseases Focuses Meeting, "The different types of gene therapy in hepatic GSDs", Kassel, Germany, October 2022
315. Invited Speaker at the 2022 Sanofi Steps Forward in Pompe Disease Meeting, "IOPD Management US Experience – Lessons Learned After 15 Years of Experience", and "Clinical Trials in Patients with LOPD – New Treatment Options Available: Avalglucosidase Alfa" for Session Titled Pompe Disease: Patient's Outcome and Management Insights with Enzyme Replacement Therapy (ERT), Berlin, Germany, November 2022
316. Invited Speaker for a Satellite Symposium at the WORLD Symposium, "Exploring Next-generation Therapies to Mitigate Disease Progression in Pompe Disease, Orlando, FL, February 2023
317. Presented at the Sanofi Medical Booth at WORLD Symposium, "30-Year Registry", Orlando, FL, February 2023.
318. Invited Speaker for Sanofi Satellite Symposium at the WORLD Symposium, "The Next Step Forward: Real World Experience in Patients with Late-Onset Pompe Disease, Orlando, FL, February 2023.
319. Invited Presenter for the Clinical Application Session at the WORLD Symposium, "Efficacy and safety of avalglucosidase alfa in participants with late-onset Pompe disease after 145 weeks of treatment during the COMET trial, Orlando, FL, February 2023.
320. Invited Speaker for a Satellite Symposium at the Society of Inherited Metabolic Disease Annual Meeting, "Managing Late-onset Pompe Disease in the Era of Newborn Screening", Salt Lake City, UT, March 2023.
321. Invited Speaker for the South Asia LSD Symposium, "Newer Therapies in Pompe disease and other Glycogen Storage Diseases", Bangalore, India, April 2023.
322. Invited Speaker for India Charitable Access Program (INCAP) at the Indian Medical Advisory Board (IMAB) Meeting, "Managing Pompe Disease with Nexviazyme (Avalglucosidase alfa-ngpt)", Bengaluru, India, April 2023.
323. Invited to do pre-recorded presentation for the Abbott Nutrition Health Institute (ANHI) Metabolic Conference, "Pompe Disease: Clinical Management and Updates", San Antonio, TX, May 2023
324. Invited speaker for the Annual Association for Glycogen Storage Disease Conference, "Newborn Screening in Pompe, Orlando, FL, June 2023
325. Platform Session Speaker for Southeast Regional Genetics Network (SERN)/Southeastern Regional Genetics Group (SERGG), "Long-term Efficacy and Safety of Cipaglucosidase Alfa/Miglustat in

Ambulatory Patients with Pompe Disease: A Phase III Open-label Extension Study (ATB200-07)", Charleston, SC, July 2023

326. Invited Speaker for the Amicus Consulting Internal Speaking Event, "A Journey through time-lessons learned in development of a therapy for a rare disease", Washington, DC, July 2023
327. Invited Speaker for the Society for Inherited Metabolic Disorders (SSIEM) Annual Conference for Amicus, "Switching treatment from alglucosidase alfa to cipaglucosidase alfa plus miglustat positively affects motor function and quality of life in patients with late-onset Pompe disease", Jerusalem, Israel, August 2023
328. Invited Speaker for the Society for Inherited Metabolic Disorders (SSIEM) Annual Conference, Sanofi Pompe Medical Satellite Symposium, "Efficacy and safety of avalglucosidase alfa in participants with late-onset Pompe disease after 145 weeks' treatment during the COMET Trial", Jerusalem, Israel, August 2023
329. Invited Speaker for Sanofi sponsored Satellite Symposium for the World Muscle Society Annual Meeting, "Optimizing Care: Real World Experience in Patients with Late-Onset Pompe Disease", Charleston, SC, October 2023
330. Invited Poster Presenter for the World Muscle Society Annual Meeting, " COMET: Effects of avalglucosidase alfa and treatment switch from alglucosidase alfa on week 145 QMFT individual item responses", Charleston, SC, October 2023
331. Invited Virtual Presenter for the Australian Pompe Association Meeting, "Newborn screening for late-onset Pompe disease and infant on-set Pompe disease, October 2023
332. Serve as a panelist at the North America Rare Disease Summit, "Addressing Management of Rare Diseases. Hospital Networks & Reference Centers, New York, NY, November 2023
333. Served on the special panelist at the Duke Medical Alumni Association All-Alumni Luncheon, host by the Golden Blue Devils, "Panel Discussion: - Team Medicine: From Breakthrough Research to Lifesaving Care", Durham, NC, November 2023
334. Plenary Session Speaker for The Indian Society of Human Genetics (ISHG) 2024 Conference, "Pompe disease: An exemplar for diagnosis, treatment, and prevention, Satellite, Ahmedabad INDIA, January 2024.
335. Serve as a speaker for the Indian Medical Advisory Board (IMAB) Meeting, "Pompe disease and it's management", Ahmedabad, Gujarat INDIA, January 2024.
336. Serve as chair and speaker for a Satellite Symposium at WORLD Symposium, "Optimizing Outcomes in Late-Onset Pompe Disease: Integrating New Therapies, Whole-person Makers of Disease Monitoring, and Shared Decision-making into Practice", San Diego, CA, February 2024.
337. Served as a speaker at a Satellite Symposium at the WORLD Symposium, "From Bench to Bedside: the Example of Pompe Disease", San Diego, CA, February 2024.
338. Serve as speaker at a Satellite Symposium at the WORLD Symposium, Gaucher Disease: The Evolving Big Picture (Part II - Gaucher Disease Across the Lifespan, "Implications for Pregnancy, Gaps in

Knowledge, Future Directions", San Diego, CA, February 2024

339. Speaker at a Satellite Symposium at the WORLD Symposium, "A Focus on Bone Manifestations in Gaucher Disease Type 1 Patients", San Diego, CA, February 2024.
340. Serve as a speaker at a Satellite Symposium at the American College of Medical Genetics Annual Clinical Genetics Meeting, "A New Treat Approach for Late-Onset Pompe Disease", Toronto, Canada, March 2024.
341. Speaker at the Newborn Screening virtual Webinar for the Neuromuscular Disease Network for Canada (NMD4C), "A new understanding of Pompe disease in the era of Newborn Screening", April 2024
342. Speaker/ Chair at the AMDA/IPA International Pompe Patient and Scientific Conference, "Natural History of Pompe vs. New History with ERT", San Antonio, TX, May 2024.
343. Expert Panelist at the AMDA/IPA International Pompe Patient and Scientific Conference, "Expert Roundtable: 25 Years of Treatment and Care of Pompe Patients", San Antonio, TX, May 2024.
344. Speaker for Adult Polyglucosan Body Disease (APBD) GSD IV Focus Group Meeting, "Where GSD IV and APBD Align", Virtual, May 2024
345. Speaker at the AGSD (Association for Glycogen Storage Disease) Annual Conference, "GSD VI and IX Updates and "Pompe Disease Updates, Minneapolis, MN, June 2024

Clinical Activity:

- Director, Metabolic Clinic which serves as a tertiary referral center for inborn errors of metabolism and lab interpretations with clinical correlations. Serves as international center for Glycogen Storage disorders.
- Director, Lysosomal Storage Disease Program, Durham, NC.
- Clinical Medical Genetics staff physician, full hospital privileges, consultant for Inpatient Metabolic and Genetics Services.
- Clinical Research – Principal investigator for several trials (over 100 to date) –investigator initiated, foundation and pharmaceutical supported: Enzyme replacement therapy (ERT) for Pompe disease, small molecule and ERT for Gaucher disease, hypophosphatasia, SMA, gene therapy approaches for Duchenne Muscular dystrophy, Pompe disease, SMA, clinical trials of small molecules for enhancement of cognition in Down syndrome, SMA, natural history studies and registries for Pompe disease, MPSI, MPS IV, Gaucher disease, Glycogen Storage diseases I and III, Down syndrome and hypophosphatasia.

Participation in academic and administrative activities of the University and Medical Center:

- Chief, Division of Medical Genetics, Duke University Medical Center, 2007 – present
- Medical Director, Alice and YT Chen Pediatrics Genetics and Genomics Research Center, 2007 – present
- Committee Member for Chancellor's Duke Medicine Innovation Council, 2011 – present
- Committee Member for Duke University Medical Alumni Awards, 2009 – present
- Faculty advisor and mentor – Duke undergraduate and 3rd year medical students, 2008 – present
- Mentorship committee for junior faculty members in Pediatrics and Medicine, 2008 – present

- Distinguished Professor Selection Committee, Duke University Medical Center, School of Medicine, 2013 – present
- Focused Group Committee Member on Rare Diseases, Duke University Medical Center, 2013 – present
- Committee Member, Regenerative Medicine, Duke University Medical Center, 2013
- Member of the Human Genetics Genomics Summit– Working Group, 2014
- Member of Duke Translational Research Advisory Committee, Duke University Medical Center, 2014-present
- Duke O'Brien Center for Kidney (DOCK) Research Executive Committee, 2015 – present
- Evaluator for "Pitch Your Innovative Clinical Products and Business Idea for Investment", Duke Health Innovation Jam, Duke Institute for Health Innovation, September 2015-2020
- Committee member, Internal Advisory Group for Children's Discovery Institute (CDI), Duke Pediatrics, February 2017- present
- Affiliate Member of the CHDI (Children's Health and Discovery Institute), March 2018-present
- Committee member, Duke University School of Medicine, Precision Medicine and Genetics/Genomics Work Group, January 2018-present
- Faculty Collaborator for Duke Innovation and Entrepreneurship Initiative, July 2018-present
- Faculty Expert Member for Rare Diseases, Duke School of Medicine Gene Therapy Initiative, October 2019-present
- Participate in Strategic Education Sessions on Research, Translation and Commercialization with the Duke Board of Trustees for Academic year 2019-2020
- Member of the Duke Precision Genomics Collaborators Executive Leadership Committee, January 2020-present
- Member of the Duke Initiative on Research Translation and Commercialization (RTC) Working Group, October 2020-present
- Steering Committee Member for Duke University School of Medicine Marcus Center for Cellular Cures (MC3), July 2022-present
- Member of the Duke Office of Translation & Commercialization (OTC) Oversight Board, May 2023