

Michael H. Guo, M.D., Ph.D.
Curriculum Vitae

Contact Information

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3400 Spruce Street
Philadelphia, PA, 19104

Education and Training

2019-2022 Hospital of the University of Pennsylvania, Philadelphia, PA
Resident, Department of Neurology

2018-2019 University of North Carolina, Chapel Hill, NC
Intern, Department of Internal Medicine

2010-2018 University of Florida College of Medicine, Gainesville, FL
MD, Graduation with Honors in Research
PhD portion of MD/PhD performed at Harvard

2012-2016 Harvard University, Cambridge, MA
PhD, Genetics and Genomics
Dissertation title: Genetic methods and studies of human growth and development across the allelic spectrum

2006-2010 University of Michigan, Ann Arbor, MI
BBA with High Distinction, Business Administration
BS with High Distinction, Cellular and Molecular Biology

Research Positions

2020- Present Residency/Post-doc research
Mentor: Jennifer Philips-Cremens, PhD and Edward Lee, MD, PhD
University of Pennsylvania
Research topic: Short tandem repeats in Alzheimer's disease

2012- 2016 Graduate school (PhD)
Mentor: Dr. Joel Hirschhorn, MD, PhD
Harvard Medical School/Boston Children's Hospital
Research topic: Genetics methods development and studies into genetic basis of human quantitative traits (height, BMI, timing of puberty) and related disorders of human growth and development

2009- 2010 Undergraduate research assistant
Mentors: Dr. Kai Zheng, PhD and Dr. David Hanauer, MD
University of Michigan School of Public Health
Research topic: Impact of health information technology on clinician workflow and patient interactions

2008-2010 Undergraduate research assistant
Mentor: Dr. John Moran, PhD
University of Michigan Medical School/HHMI

Peer-reviewed publications

1. Dratch L, Bardakjian TM, Johnson K, Babaian N, Gonzalez-Alegre P, Elman L, Quinn C, **Guo MH**, Scherer SS, Amado DA. The importance of offering exome or genome sequencing in adult neuromuscular clinics. Manuscript accepted to Biology.
2. **Guo MH***, Francioli LC*, Stenton SL, Goodrich JK, Watts NA, Singer-Berk M, Groopman E, Darnowski PW, Solomonson M, Baxter S, gnomAD Project Consortium, Tiao G, Neale BM, Hirschhorn JN, Rehm HL, Daly MJ, O'Donnell-Luria A, Karczewski KJ, MacArthur DA, and Samocha KE. Inferring compound heterozygosity from large-scale exome sequencing data. Nature Genetics. Online ahead of print. PMID: 38057443.
3. Baldwin A, Copeland J, Azage M, Dratch L, Johnson K, Paul RA, Amado DA, Baer M, Deik AF, Elman LB, **Guo MH**, Hamedani AG, Irwin DJ, Lasker A, Orthmann-Murphy J, Quinn C, Tropea TF, Scherer SS, Ellis CA. Disparities in Genetic Testing for Neurologic Disorders. Manuscript in press at Neurology.
4. Dratch L, Azage M, Baldwin A, Johnson K, Paul RA, Bardakjian TM, Michon S-C, Amado DA, Baer M, Deik AF, Elman LB, **Guo MH**, Hamedani AG, Irwin DJ, Lasker A, Orthmann-Murphy J, Quinn C, Tropea TF, Scherer SS, Ellis CA. Genetic testing in adults with neurological disorders: indications, approach, and clinical impacts. Journal of Neurology. Online ahead of print. PMID: 37891417.
5. Lofrano-Porto A, Pereira SA, Dauber A, Asimow N, de Moraes OL, Araujo PAT, Abreu AP, **Guo MH**, Oliveira SF, Liu H, Lee C, Coelho MS, Jiang R, Carroll RS, Kaiser UB. OSR1 is important for Mullerian duct and endometrial development. Journal of Clinical Investigation. 2023. 133(23):e161701. PMID: 37847567
6. **Guo MH** and Vaishnavi SN. Clinical management in Alzheimer's disease in the era of disease modifying therapies. Current Treatment Options in Neurology. 2023. 25:121–133.
7. Deignan JL, Gregg AR, Grody WW, **Guo MH**, Kearney K, Monaghan K, Raraigh K, Taylor J, Zepeda-Mendoza C, Ziats C. Recommendations for *CFTR* carrier screening: A position statement of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine. 2023. 25(8):100867. PMID: 37310422
8. Mouri K*, **Guo MH***, de Boer CG, Lissner MM, Harten IA, Newby GA, DeBerg HA, Platt WF, Gentili M, Liu DR, Hacohen N, Tewhey R, Ray JP. Prioritization of autoimmune disease-associated genetic variants that perturb regulatory element activity in T cells. Nature Genetics. 2022. 54(5):603-612. PMID: 35513721.
*co-first authors
9. **Guo MH**, Sarma P, LaBarre BA, Lokhande H, Balibalos J, Chu C, Du X, Kheradpour P, Kim CC, Oniskey T, Snyder T, Soghoian DZ, Weiner HL, Chitnis T, and Patsopoulos N. Integrating human genetic and epigenetic data to identify causal mechanisms underlying multiple sclerosis pathogenesis. Genome Biology. 2022. 23(1):127. PMID: 35672799
10. **Guo MH**, Bardakjian TM, Brzozowski MR, Scherer SS, Quinn C, Elman L, Orthmann-Murphy J, Tropea TF, Ellis CA, and Gonzalez-Alegre P. Temporal trends and yield of

- clinical diagnostic genetic testing in adult neurology. American Journal of Medical Genetics. 2021. 185(10):2922-28. PMID: 34075706
11. Gergics P, Smith C, Bando H, Jorge ALA, Rockstroh-Lippold D, Vishnopolaska S, Castinetti F, Maksutova M, Carvalho LRS, Hoppmann J, Mayer JM, Albarel F, Braslavsky D, Keselman A, Bergadá I, Martí M, Saveanu A, Barlier A, Jamra RA, **Guo M**, Dauber A, Nakaguma M, Mendonça BB, Ozel AB, Fang Q, Ma Q, Li JZ, Brue T, Millán MIP, Kitzman JO, Arnhold IJP, Pfaeffle R, Camper SA. High-throughput splicing assays identify missense and silent splice-disruptive POU1F1 variants underlying pituitary hormone deficiency. American Journal of Human Genetics. 2021. 108(8):1526-39. PMID: 34270938
 12. Vuckovic D, Bao EL, Akbari P, and 100 additional authors including **Guo MH**. The Polygenic and Monogenic Basis of Blood Traits and Diseases. Cell. 2020. 182(5):1214-31. PMID: 32888494
 13. Chen MH, Raffield LM, Mousas A, and 110 additional authors including **Guo MH**. Trans-ethnic and ancestry-specific blood-cell genetics in 746,667 individuals from 5 global populations. Cell. 2020. 182(5):1198-1213. PMID: 32888493
 14. Beagan JA, Pastuzyn ED, Fernandez LR, **Guo MH**, Titus KR, Chandrashekar H, Shepherd JD, Phillips-Cremens JE. 3D genome architecture orchestrates activity-dependent gene expression in neurons. Nature Neuroscience. 2020. 23(6):707-17. PMID: 32451484
 15. Tropea TF, Mak J, **Guo MH**, Xie SX, Suh E, Rick J, Siderowf A, Weintraub D, Grossman M, Irwin D, Wolk DA, Trojanowski JQ, van Deerlin V, Chen-Plotkin AS. *TMEM106B* Exerts Genetic Modifier Effects on Longitudinal Cognitive Change in Parkinson's Disease and Frontotemporal Dementia. Annals of Neurology. 2019. 85(6):801-811. PMID: 30973966
 16. Lareau CA, Ulirsch JC, Bao EL, Ludwig LS, **Guo MH**, Benner C, Satpathy AT, Kartha VK, Salem RM, Hirschhorn JN, Finucane HK, Aryee MJ, Buenrostro JD, and Sankaran VG. Interrogation of human hematopoiesis at single-cell and single-variant resolution. Nature Genetics. 2019. 51(4):683-93. PMID: 30858613
 17. **Guo MH** and Gregg AR. Estimating yields of carrier screening and implications for design of expanded carrier screening panels. Genetics in Medicine. 2019. 21(9):1940-47. PMID: 30846881.
[feature article for September 2019 issue]
 18. Ulirsch JC, Verboon JM, Kazerounian S, **Guo MH**, Yuan D, Ludwig LS, Handsaker RE, Abdulhay NJ, Fiorini C, Genovese G, Lim ET, Cheng A, Cummings BB, Chao KR, Beggs AH, Genetti CA, Sieff CA, Newburger PE, Niewiadomska E, Matysiak M, Vlachos A, Lipton JM, Atsidaftos E, Glader B, Narla A, Gleizes PE, O'Donohue MF, Montel-Lehry N, Amor DJ, McCarroll SA, O'Donnell-Luria AH, Gupta N, Gabriel SB, MacArthur DG, Lander ES, Lek M, Da Costa L, Nathan DG, Korostelev AA, Do R, Sankaran VG, and Gazda HT. The Genetic Landscape of Diamond-Blackfan Anemia. American Journal of Human Genetics. 2018. 103(6):930-47. PMID 30503522.
 19. **Guo MH**, Plummer LC, Chan Y-M, Hirschhorn JN, and Lippincott MF. Burden testing of rare variants identified through exome sequencing via publicly available control data. American Journal of Human Genetics. 2018. 103(4):522-534. PMID: 30269813

20. **Guo MH**, Monir RL, Wright A, Holland NP. Case of serotonin syndrome initially presenting as diffuse body pain. American Journal of Case Reports. 2018. 19:1227-31. PMID: 30318504
21. **Guo MH**, Hirschhorn JN, Dauber A. Insights and implications of genome wide association studies of height. Journal of Clinical Endocrinology and Metabolism. 2018. 103(9):3155-68. PMID: 29982553. [invited review]
22. Cesana M*, **Guo MH***, Cacchiarelli D*, Wahlster L, Doulatov S, Salvatori B, Trapnell C, Ziller MJ, Clement K, Cahan P, Tsanov K, Sousa P, Barragan J, Tazon-Vega B, Giorgi FM, Bolondi A, Califano A, Rinn JL, Meissner A, Hirschhorn JN, and Daley GQ. Alternative splicing of HMGA2 by CLK3 governs human hematopoietic stem cell ontogeny. Cell Stem Cell. 2018. 22(4):575-588. PMID: 29625070. *co-first authors [cover article]
23. **Guo M***, Liu Z*, Willen J*, Shaw CP, Richard D, Dingwall H, Jagoda E, Doxey A, Hirschhorn JN and Capellini T. Epigenetic profiling of growth plate chondrocytes sheds insight into genetic variation influencing height. eLife. 2017. e29329. PMID: 29205154. *co-first authors
24. **Guo MH***, Nandakumar SK*, Ulirsch JC, Zekavat SM, Buenrostro, JD, Natarajan P, Salem RM, Chiarle R, Mitt M, Kals M, Pärn K, Fischer K, Milani L, Mägi R, Palta P, Gabriel SB, Metspalu A, Lander ES, Kathiresan S, Hirschhorn JN, Esko T, and Sankaran VG. Comprehensive Genome Sequencing in a Population-based Biobank Provides Insight into Hematopoietic Regulatory Mechanisms. Proceedings of the National Academy of Sciences. 2017. 114(3):E327-36. PMID: 28031487. *co-first authors
25. Ocaranza P*, Golekoh MC*, Andrew SF*, **Guo MH**, Kaplowitz P, Saal S, Rosenfeld RG, Dauber A, Cassorla F, Backeljauw PF, and Hwa V. Expanding genetic and functional diagnoses of IGF1R haploinsufficiencies. Hormone Research. 2017. 87(6):412-22. PMID: 28395282.
26. Swartz JM, Ciarlo R, **Guo MH**, Abrha A, Diamond DA, Chan YM, and Hirschhorn JN. A 46,XX ovotesticular disorder of sex development likely caused by a steroidogenic factor-1 (NR5A1) variant. Hormone Research. 2017. 87(3):191-95. PMID: 27855412
27. **Guo MH**, Dauber A, Lippincott MF, Chan YM, Salem RM, Hirschhorn JN. Determinants of Power in Gene-based Burden Testing for Monogenic Disorders. American Journal of Human Genetics. 2016. 99(3):527-39. PMID: 27545677
28. Swartz JM, Ciarlo R, **Guo MH**, Abrha A, Diamond DA, Chan YM, and Hirschhorn JN. Two unrelated undervirilized 46,XY males with inherited NR5A1 variants identified by whole exome sequencing. Hormone Research. 2016. 87(4):264-70. PMID: 27553487
29. Nilsson O, Isoherranen N, **Guo MH**, Lui, JC, Jee YH, Guttmann-Bauman I, Acerini C, Lee W, Allikmets R, Yanovski JA, Dauber A, and Baron, J. Accelerated skeletal maturation in disorders of retinoic acid metabolism: A case report and focused review of the literature. Hormone and Metabolic Research. 2016. 48(11):737-744. PMID: 27589347
30. Giani FC, Ludwig LS, Wakabayashi A, Salem RM, Jobaliya CD, Regan SN, Ulirsch JC, Liang G, Steinberg-Shemer O, **Guo MH**, Esko T, Tong W, Brugnara C, Hirschhorn JN, Weiss MJ, Zon LI, Chou ST, French DL, Musunuru K, Sankaran VG. Targeted

- application of human genetic variation can improve red blood cell production from stem cells. Cell Stem Cell. 2015. 18(1):73-8. PMID: 26607381
31. Chan Y, Salem RM, Hsu YH, McMahon G, Pers TH, Vedantam S, Esko, T, **Guo MH**, Lim ET, GIANT Consortium, Davey Smith G, and Hirschhorn JN. Genome-wide association analysis of body proportion classifies height-associated variants by mechanism of action and implicates genes important for skeletal development. American Journal of Human Genetics. 2015. 96(5):695-708. PMID: 25865494.
 32. Quintos JB, **Guo MH**, Dauber A. Idiopathic short stature due to novel heterozygous mutation of the aggrecan gene. Journal of Pediatric Endocrinology and Metabolism. 2015. 28(7-8):927-32. PMID: 25741789.
 33. Zhu J, Choa REY, **Guo MH**, Plummer L, Buck C, Palmert MR, Hirschhorn JN, Seminara SB, Chan YM. A shared genetic basis for self-limited delayed puberty and idiopathic hypogonadotropic hypogonadism. Journal of Clinical Endocrinology and Metabolism. 2015. 100(4):E646-54. PMID: 25636053.
 34. Wegman MP*, **Guo MH***, Bennion DM, Shankar MN, Chrzanowski SM, Goldberg LA, Xu J, Williams TA, Hsu SI, Anton SD, Leeuwenburgh C, Brantly ML. Practicality of Intermittent Fasting in Humans and its Effect on Oxidative Stress and Genes Related to Aging and Metabolism. Rejuvenation Research. 2015. 18(2):162-72. PMID: 25546413.
*co-first authors
 35. Majithia AR, Flannick J, Shahinian P, **Guo M**, Bray MA, GoT2D Project, NHGRI JHS/FHS Allelic Spectrum Project, SIGMA T2D Consortium, T2D-GENES Consortium, Rosen ED, and Altshuler D. Rare variants in PPARG with decreased activity in adipocyte differentiation are associated with increased risk of type 2 diabetes. Proceedings of the National Academy of Sciences. 2014. 111(36):13127-32. PMID: 25157153.
 36. Kerns SL, Guevara-Aguirre J, Andrew S, Geng J, Guevara C, Guevara-Aguirre M, **Guo M**, Oddoux, C., Geng J, Shen Y, Rosenfeld RG, Ostrer H, Hwa V, and Dauber A. A novel variant in CDKN1C is associated with intrauterine growth restriction, short stature, and early-adulthood-onset diabetes. Journal of Clinical Endocrinology and Metabolism. 2014. 99(10):E2117-22. PMID: 25057881.
 37. Nilsson O*, **Guo MH***, Dunbar N, Popovic J, Flynn D, Jacobsen C, Lui JC, Hirschhorn JN, Baron J, and Dauber A. Short stature, accelerated bone maturation, and early growth cessation due to heterozygous aggrecan mutations. Journal of Clinical Endocrinology and Metabolism. 2014. 99(8):E1510-8. PMID: 24762113.
*co-first authors
 38. **Guo MH**, Shen Y, Miller TC, Walvoord, E.C., Moon JE, Hirschhorn JN, and Dauber A. Whole exome sequencing to identify genetic causes of short stature. Hormone Research in Pediatrics. 2014. 82(1):44-52. PMID: 24970356
[Selected as Editor's Choice article and featured on cover]
 39. Okada Y, Wu D, Trynka G, and 92 other authors including **Guo MH**. Genetics of rheumatoid arthritis contributes to biology and drug discovery. Nature. 2014. 506(7488):376–381. PMID: 24390342
 40. **Guo MH**, Stoler J, Lui J, Nilsson O, Bianchi DW, Hirschhorn JN, and Dauber, A. Redefining the progeroid form of Ehlers-Danlos syndrome: Report of the fourth case of

- B4GALT7* deficiency and review of the literature. *American Journal of Medical Genetics*. 2013. 161(10):2519-27. PMID: 23956117.
41. Zheng K, **Guo MH**, and Hanauer DA. Using the time and motion method to study clinical work processes and workflow: Methodological inconsistencies and a call for standardized research. *Journal of the American Medical Informatics Association*. 2011. 18(5):704–10. PMID: 2152740.

Manuscripts in Submission or Preparation

1. **Guo MH**, Lee WP, Vardarajan B, Schellenberg GD, Phillips-Cremens JE. Polygenic burden of short tandem repeat expansions promote risk for Alzheimer’s disease. Manuscript in submission to *Nature Communications*. Preprint: <https://www.medrxiv.org/content/10.1101/2023.11.16.23298623v1>.

Editorials

1. **Guo MH** and Beslow LA. Basal ganglia calcifications–etiological relationship to strokes from mild head trauma? 2021. *European Journal of Pediatric Neurology*. 33:A3-A4. PMID: 34247930
2. Mittwede PN, Noch EK, and **Guo MH**. A closer look at attrition in MD–PhD programs. *Academic Medicine*. 2014. 89(7):958-9. PMID: 24979156.

Book Chapters

1. **Guo MH** and Gregg AR (2018). Prenatal Genetic Carrier Screening. In Norton M, Kuller J, Dugoff L, and Saade G (Eds.) *Perinatal Genetics*. (pp 65-70). Elsevier.
2. **Guo MH** and Dauber A (2016). Application of Genomics to the Study of Human Growth and Stature. In Hutton JJ (Editor), *Pediatric Biomedical Informatics*. New York, NY: Springer.

Selected Abstracts and Conference Presentations

1. **Guo MH**, Lee WP, Vardarajan B, Schellenberg G, Phillips-Cremens J. Short tandem repeats as a novel genetic driver of Alzheimer’s disease. *American Neurological Association Annual Meeting*. Philadelphia, PA. September, 2023. [Selected for Travel Award]
2. **Guo MH**, Mouri K, de Boer CG, Newby GA, Gentili M, Liu DR, Hacoheh N, Tewhey R, Ray JP. Prioritization of autoimmune disease-associated genetic variants that perturb regulatory element activity in T cells. *American Neurological Association Annual Meeting* (Virtual). October, 2021. [Featured Poster Presentation].
3. **Guo MH**, Cesana M, Cacchiarelli D, Wahlster L, Doulatov S, Salvatori B, Trapnell C, Ziller MJ, Clement K, Cahan P, Tsanov K, Sousa P, Barragan J, Tazon-Vega B, Giorgi FM, Bolondi A, Califano A, Rinn JL, Meissner A, Hirschhorn JN, and Daley GQ. Global transcriptomic analysis of human hematopoietic stem cells identifies alternative splicing of *HMG2* in mediating stem cell properties. *American Society of Human Genetics*. Orlando, FL. October, 2017. [Selected for Platform Presentation].
4. **Guo MH**, Nandakumar SK, Ulirsch JC, Zekavat SM, Buenrostro JD, Natarajan P, Salem RM, Chiarle R, Gabriel SB, Metspalu A, Lander ES, Kathiresan S, Hirschhorn JN, Esko T, and Sankaran VG. Comprehensive population-based genome sequencing provides insight into hematopoietic regulatory mechanisms. *Joint Meeting of the American Society for Clinical Investigation and the Association of American Physicians*. Chicago, IL, April 2017. [Selected for travel award].

5. **Guo MH**, Plummer LC, Hirschhorn JN, Lippincott MF. Rare Variant Gene-Based Case-Control Burden Testing in Hypogonadotropic Hypogonadism Using Publically Available Control Data. *Endocrine Society Annual Meeting*. Orlando, FL. April 2017 [Selected for oral and poster presentation].
6. **Guo MH**, Nandakumar SK, Ulirsch JC, Zekavat SM, Buenrostro JD, Natarajan P, Salem RM, Chiarle R, Gabriel SB, Metspalu A, Lander ES, Kathiresan S, Hirschhorn JN, Esko T, and Sankaran VG. Comprehensive population-based genome sequencing provides insight into hematopoietic regulatory mechanisms. Vancouver, British Columbia, Canada, October, 2016. *American Society of Human Genetics Annual Meeting*. [Selected for Oral Presentation].
7. **Guo MH**, Willen J, Dingwall H, Hirschhorn JN and Capellini TC. Epigenetic Profiling of Murine Growth Plates Demonstrate Enrichment for Height-associated GWAS SNPs. *Endocrine Society Annual Meeting*. Boston, MA, April, 2016. [Featured Poster Award; Oral and Poster Presentation].
8. **Guo MH** and Hirschhorn JN. Evaluating Power and Sample Size Considerations for Gene Discovery in Rare Diseases. *American Society of Human Genetics Annual Meeting*. Baltimore, MD, October, 2015. [Poster Presentation]
9. **Guo MH**, Nilsson O, Quintos JB, Dunbar N, Popovic J, Flynn D, Jacobsen C, Lui JC, Baron J, Hirschhorn JN, and Dauber A. Heterozygous Mutations in Aggreca cause Short Stature, Accelerated Bone Maturation, and Early Growth Cessation. *Joint Meeting of the American Society for Clinical Investigation and the Association of American Physicians*. Chicago, IL, April 2015. [Selected for travel award].

Research Funding

Pending	NIH/NIA K08 Award “Short tandem repeats as a novel genetic driver of Alzheimer’s disease” Score: 24 (eligible for AD/ADRD payline; awaiting Council meeting in January 2024)
2022-ongoing	Penn ADRC Clark Scholars Award Award to support clinical and research training in dementia field.
2021-ongoing	NIH/NINDS R25 grant (NIH R25 NS065745; J. Detre PI) “Unraveling the mechanisms and consequences of <i>RBFOX1</i> dysregulation in Alzheimer’s disease” Research education grant to support research during and after neurology residency
2013-2016	NIH T32 grant (NIH 5T32GM096911; F. Winston PI) Graduate school training grant

Awards and Honors

2023	Best Poster Award, Penn Institute of Aging Annual Symposium
2023	American Neurologic Association Annual Meeting Travel Award
2022	University of Pennsylvania Neurology Citizenship Award
2019	UNC Internal Medicine Research Day Prize
2018	American Academy of Neurology Medical Student Prize for Excellence in Neurology

2017	American Society of Human Genetics Charles J. Epstein Trainee Award-Semifinalist
2017	Travel award, Joint Meeting of the American Society for Clinical Investigation and Association of American Physicians
2016	Featured Poster award, Endocrine Society Annual Meeting
2015	Travel award, Joint Meeting of the American Society for Clinical Investigation and Association of American Physicians
2009-2010	James B. Angell Scholar (given to students who achieve an "A" in all courses in consecutive terms), University of Michigan
2007-2010	University Honors, University of Michigan (Semesters: Fall '07, Spring '08, F '08, S '09, F '09, S '10)
2006-2007	Honors Program, College of Literature, Science, and the Arts, University of Michigan
2006-2010	Regents Merit Scholarship Award (\$20,000 per year; awarded to 20 incoming students per year based on merit), University of Michigan
2006	Interfraternal Council Outstanding Freshman Award, University of Michigan
2006	Valedictorian, Class of 2006, Gulf Coast High School
2006	AP National Scholar, Advanced Placement Program
2006	National Merit Finalist

Leadership Experience

American Physician Scientist Association

2014-2015	President (national organization)
2013-2014	President-elect (national organization)
2012-2013	Chair of Finance Committee (national organization)
2011-2012	President (University of Florida chapter)

Asian Pacific American Medical Student Association

2011-2012	President (University of Florida chapter)
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University of Florida College of Medicine

2010-2012	Medical school class treasurer
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Presentations and Seminars

2023	University of Pennsylvania Alzheimer's Disease Research Center Grand Rounds
2021	American Neurologic Association Annual Meeting oral presentation
2017	American Society of Human Genetics Annual Meeting oral presentation
2017	Florida Chapter of the American Academy of Pediatrics research talk
2017	University of Florida Department of Obstetrics and Gynecology Grand Rounds
2016	American Society of Human Genetics Annual Meeting oral presentation
2016	Endocrine Society Annual Meeting oral presentation
2016	Boston Children's Hospital Endocrinology Division seminar
2016	Broad Institute Functional Genomics seminar
2016	Harvard Program in Genetics and Genomics research talk
2015	Harvard Biomedical and Biological Sciences Recruitment research talk
2015	Boston Children's Hospital Endocrinology Division seminar
2014	Harvard Program in Genetics and Genomics research talk
2014	Harvard Biomedical and Biological Sciences Annual Retreat invited talk
2014	Boston Children's Hospital Endocrinology Division seminar

Service and Outreach

2020-	Committee member, American College of Medical Genetics CFTR Screening Diversity Equity and Inclusion Workgroup
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2016 Public outreach speaker, Science in the News, Boston, MA
 Gave lecture series on human genetic to public in various venues
 2013-2014 Massachusetts General Hospital Center for Community Health Improvement, Boston, MA
 Mentored underprivileged high school students preparing for college and careers. Assisted with local middle school science fair.
 2011-2012 Equal Access Clinic, Gainesville, FL
 Provided free medical care and basic social work needs for an underserved population in Alachua County

Teaching Experience

2018 Invited Guest Lecture
 Biology 105: Modern Biological Concepts; Meredith College (Raleigh, NC)
 2016 Teaching Assistant
 Genetics 202: Human Genetics; Harvard Medical School
 2016 Teaching Assistant
 First Year Genetics Course; Harvard Medical School
 2014 Teaching Assistant
 Genetics 202: Human Genetics; Harvard Medical School
 2013 Teaching Assistant
 Genetics 201: Principles of Genetics; Harvard Medical School

Mentorship

2023 Skenlanda Marseille, American Physician Scientist Association
 undergraduate summer program for under-represented minorities
 2020 Danying Guan, American Physician Scientist Association undergraduate
 summer program for under-represented minorities
 2015-2016 Kristin Tsoi, undergraduate student in Hirschhorn lab
 2014 Beryl Cummings, rotation student in Hirschhorn lab

Ad hoc Journal Reviews

American Journal of Medical Genetics, BMC Medical Genomics, Clinical Endocrinology, eLife, European Journal of Human Genetics, European Journal of Medical Genetics, Frontiers in Genetics, Genome Research, Human Molecular Genetics, Nature Communications, Orphanet Journal of Rare Diseases, PLoS Computational Biology, PLoS Genetics, Stem Cell Research

Also participated in reviews for *Journal of Clinical Endocrinology and Metabolism* and *New England Journal of Medicine* (with Andrew Dauber); *Nature Genetics* and *Cell* (with Jennifer Phillips-Cremens)

Other Employment

2014-2015 MedIQ/First Aid USMLERx
 Editor, USMLE Step 1 Video Series
 2013-2014 MedIQ/First Aid USMLERx
 Question writer for USMLE Step 1 Test Bank
 2009 Deutsche Bank AG, New York, NY
 Global Markets Sales and Trading Summer Analyst
 2008 Lehman Brothers, Washington, DC
 Equity Research Summer Intern

Professional Membership

2020-Present Member, American Neurological Association
 2018-Present Member, American College of Medical Genetics
 2018-Present Member, American Academy of Neurology
 2013-2018 Member, American Society of Human Genetics

2010-Present

Member, American Physician Scientists Association