

CURRICULUM VITAE

PERSONAL STATEMENT

I am a computational biologist with extensive experience in genetics and analyses of omics data, and I am the PI of this application focused on developing methods/tools coupled with high-throughput functional assays for improved variant classification. I am a part of the NHGRI's Baylor College of Medicine Genomics Research to Elucidate the Genetics of Rare Diseases Program (BCM-GREGoR) as a PI of the UTHealth School of Public Health (UTSPH) subaward. My Ph.D. studies in the laboratory of Dr. Barton performed an epigenetic profiling to examine the function of a very well-studied tumor suppressor p53 in normal proliferating liver cells and a potent oncogene Myc in association with Gcn5 during somatic cell programming in mice. During my postdoctoral fellowship with Dr. Lupski, I worked extensively with the data we propose to analyze from the Baylor College of Medicine: Genomics Research to Elucidate the Genetics of Rare diseases (BCM-GREGoR), spearheading a wide variety of projects that improved understanding of the relationship between gene function and human phenotypes and fascinating yet understudied genetic mechanisms of disease in Mendelian disease biology through development of computational algorithms, tools and analytical frameworks. These include development of a nonsense-mediated decay (NMD) escape intolerance score metric to rank genes based on their depletion of protein truncating variants (PTVs) within the gene region predicted to escape NMD in control databases that would render them to cause potential dominant disease traits through potential dominant-negative or gain-of-function (GoF) effects. Collectively, I have been at the forefront of Mendelian genetics and genomics for the last 1- years and carried this role forward as an Assistant Professor of Epidemiology in the UTHealth School of Public Health (UTSPH).

My lab is currently developing novel predictive models and computational tools to examine the prevalence of transcripts present with NMD-escape alleles that are associated with a wide range of human phenotypes due to production of defective proteins. To achieve this, I spearheaded collaborations with investigators at the UTHSC and other institutes all over the world to identify NMD escape alleles associated with diverse human phenotypes including Mendelian and complex disease traits and congenital heart defects. The extension of this proposed approach to the dataset available from the NHLBI's Trans-Omics Precision Medicine (TOPMed) program and Simons Foundation Autism Research Initiative (SFARI) to characterize the impact of predicted NMD-escape alleles on quantitative hematological and hemostatic traits and neurodevelopmental disorders were awarded. We are also working on benchmarking those NMD escape alleles' computational predictions with both molecular validations (Dr. Sujatha Jagannathan). I am highly committed to mentoring the next generation of scientists including mentoring my own graduate students and postdoctoral scholar and serving in the thesis committees of five graduate students from the Baylor College of Medicine, UTHealth McGovern Medical School and University of Pittsburg. Last three summers, I also lectured a course for graduate students introductory for statistical methods and software for analyzing measured genetic variation in human studies. I am also currently teaching an introductory course in epidemiology.

GENERAL BIOGRAPHICAL INFORMATION

A. Personal

1. Zeynep H Coban-Akdemir
2. Citizen (USA, Turkey)

B. Education (include institution/location, degree, and dates of attendance)

Sabancı University, Faculty of Engineering and Natural Sciences, Istanbul, Turkey

B.S. in Biological Sciences and Bioengineering, 2002-2007 Harvard University,

Department of Biostatistics, Boston, MA, USA

A.M. in Biostatistics, 2007-2009

The University of Texas Health Science Center at Houston Graduate School of

Biomedical Sciences, Houston, TX, USA

Ph.D. in Biomedical Sciences, 2010-2014

Thesis: p53 Maintains Hepatic Cell Identity During Liver Regeneration Advisor: Michelle C. Barton

Baylor College of Medicine, Molecular and Human Genetics, Houston, TX, USA Postgraduate

Training, 2014-2019

Advisor: James R. Lupski

C. Academic Appointments

The University of Texas Health Science Center at Houston School of Public Health,

Epidemiology, Human Genetics & Environmental Sciences, Houston, TX, USA Assistant

Professor, January 2020

D. Awards

Simons Foundation Pilot Award, 2024

TOPMed fellowship, 2023

ASHG/Charles J. Epstein Trainee Award for Excellence in Human Genetics Research (Semifinalist), 2017
Harvard PQG conference - Whole Genome Sequencing Analysis: Comprehensive Capture of Genetic Variants (Stellar Abstract Award), 2016
ASHG/Charles J. Epstein Trainee Award for Excellence in Human Genetics Research (Semifinalist), 2015
The University of Health Science Center at Houston: Genes & Development Research Achievement Award (Post-candidacy category, second place), 2013
The University of Health Science Center at Houston: The Center for Cancer Epigenetics Fellowship, 2013
The University of Health Science Center at Houston: Genes & Development Off-site Course Award (First place), 2013
The University of Health Science Center at Houston: Genes & Development Retreat Poster Award (Pre-candidacy category), 2012
The University of Health Science Center at Houston: Hearst Foundation Student Research & Education Award (Pre-candidacy category), 2012
The University of Health Science Center at Houston: Genes & Development Research Achievement Award (Pre-candidacy category), 2012
Harvard University: Pfizer Fellowship, 2008
Sabanci University: ranked 2nd amongst Biological Sciences and Bioengineering 2007 graduate batch with CGPA 3.72/4.00, 2007
Sabanci University: Certificate of High Honor for 6 semesters, 2002-2007 Sabanci University: Certificate of Honor for 2 semesters, 2002-2007
Sabanci University: Honor Scholarship for ranking 95th out of approximately 1,700,000 test takers in the 2002 annual central nationwide university examination, 2002

II. RESEARCH INFORMATION

A. Research Support

1U01HG011758

Posey (PI), Role: Co-Investigator

07/15/2021 – 07/15/2026

“Baylor College of Medicine - Mendelian Genomics Research Center (BCM-MGRC)”

Development of new algorithms and methods and also integration of different kinds of genomic dataset to facilitate a better understanding of Mendelian disease biology.

NHLBI Trans-Omics for Precision Medicine (TOPMed) fellowship

Zeynep Coban Akdemir, Role: Principal Investigator 03/13/2023 –

03/12/2024

“Systematic profiling and characterization of transcripts that are associated with quantitative blood disease traits through putative gain-of-function variants”

The goal of this project is to establish enhanced predictive models and computational pipelines for comprehensive annotation of nonsense-mediated decay (NMD) outcomes of protein truncating variants (PTVs) (NMD-triggering vs. NMD-escape) using the whole genome sequencing (WGS) coupled with RNA-Sequencing (RNA-Seq) data available from the Genotype-Tissue Expression (GTEx) and TOPMed datasets and assess the impact of NMD-escape PTVs (putative GoF alleles) on quantitative hematological and hemostatic traits in the setting of the TOPMed Hematology and Hemostasis (H&H) Working Group

1X01HL148621-01

Brown (PI), Role: Co-Investigator 05/2020 –

12/2023

“Transcriptome and Whole Genome analysis of HIPS samples: A prospective cohort study of previously untreated patients with hemophilia A during their first 50 exposures to rFVIII”

The goal of this project is to characterize changes in immune cell gene expression during the first 50 exposure days, changes in gene expression in FVIII-specific lymphocytes during the first 50 FVIII exposure days and the changes in gene expression during Immune Tolerance Induction (ITI) in patients with inhibitors.

The Simons Foundation Autism Research Initiative (SFARI) Pilot award

Zeynep Coban Akdemir, Role: Principal Investigator

06/01/2024 – 05/31/2026

“Systematic Identification and Characterization of Transcripts Associated with ASD through Potential Gain-of-Function Alleles”

The objective of this project is to develop advanced predictive models and computational pipelines for thorough annotation of nonsense-mediated decay (NMD) outcomes in protein truncating variants (PTVs) (differentiating between NMD-triggering and NMD-escape) using whole genome sequencing (WGS) combined with RNA-Sequencing (RNA-Seq) data from the Simons Simplex Collection dataset. Additionally, the project aims to evaluate the impact of NMD-escape PTVs (potential gain-of-function alleles) on autism spectrum disorder.

B. National Scientific Participation (include dates and titles)

Invited Lectures, Presentations, Seminars

Yale Genetics Walter J Burdette Trainee Symposium, New Haven, Connecticut (2019)

NYU Langone Health Center for Human Genetics and Genomics Talk, New York, NY (2019)

UTHealth Epidemiology, Human Genetics and Environmental Sciences Talk, Houston, TX (2019)

UTHealth SPH HGC Seminar Series Talk, Houston, TX (2021)

UTHealth SPH HGC Seminar Series Talk, Houston, TX (2022)

BCM Human Genome Sequencing Center Virtual Seminar Series Talk, Houston, TX (2022) GREGoR

Science Seminar Series Talk, Houston, TX (2023)

CPH Spring Seminar Series Talk, Houston, TX (2023) TOPMed All

Hands Workshop, Houston, TX (2023)

TOPMed Annual Meeting, Bethesda, TX (2024)

UTHealth SPH HGC Seminar Series Talk, Houston, TX (2024)

GREGoR In-Person Meeting, Boston, MA (2024)

Horizons in Genomics Seminar, Virtual (2024)

Acibadem University Seminar Series, Virtual (2025)

GREGoR Science Seminar Series, Virtual (2025)

C. Publications

- Rai, A., Klonowski, J., Yuan, B., Coveler, K. J., Dardas, Z., Egab, I., ... & **Coban-Akdemir, Z.** (2025). Genomic rare variant mechanisms for congenital cardiac laterality defect: A digenic model approach. *The American Journal of Human Genetics*, 112(7), 1664-1680.
- Kolakada, D., Fu, R., Biziaev, N., Shuvalov, A., Lore, M., Campbell, A. E., ... **Coban-Akdemir, Z.** & Jagannathan, S. (2025). Systematic analysis of nonsense variants uncovers peptide release rate as a novel modifier of nonsense-mediated mRNA decay. *Cell Genomics*.
- Bozkurt-Yozgatli, T., Lun, M. Y., Bengtsson, J. D., Sezerman, U., Chinn, I. K., **Coban-Akdemir, Z.***, & Carvalho, C. M. (2025)*. Investigation of a pathogenic inversion in UNC13D and comprehensive analysis of chromosomal inversions across diverse datasets. *European Journal of Human Genetics*, 1-9. (*Co-corresponding authors).
- Klonowski, J. F., Ganapathiraju, M., Rai, A., Glennon, B., **Coban-Akdemir, Z.**, Kostka, D., & Lo, C. (2025). Analysis of Premature Termination Codons Predicted to Escape Nonsense Mediated Decay Identifies Novel Genes, Pathways, and Networks Contributing to an Oligogenic Etiology of Congenital Heart Disease. *medRxiv*, 2025-08.
- Friedman, R. K., Heath, A. S., Huffman, J. E., Baker, J. T., Hasbani, N. R., Taliun, S. A. G., ... & Hemostasis Working Group. (2025). Genetic study of von Willebrand factor antigen levels ≤ 50 IU/dL identifies variants associated with increased risk of von Willebrand disease and bleeding. *Journal of Thrombosis and Haemostasis*, 23(8), 2410-2421.
- Bozkurt-Yozgatli, T., Pehlivan, D., Gibbs, R.A., Sezerman, U., Posey, J.E., Lupski, J.R., and **Coban-Akdemir, Z.** (2024). Multilocus pathogenic variants contribute to intrafamilial clinical heterogeneity: a retrospective study of sibling pairs with neurodevelopmental disorders. *BMC Med Genomics* 17, 85. 10.1186/s12920-024-01852-4.
- Calame, D.G., Wong, J.H., Panda, P., Nguyen, D.T., Leong, N.C.P., Sangermano, R., Patankar, S.G., Abdel-Hamid, M.S., AlAbdi, L., Safwat, S., Flannery, K.P., Dardas, Z., Fatih, J.M., Murali, C., Kannan, V., Lotze, T.E., Herman, I., Ammouri, F., Rezich, B., Efthymiou, S., Alavi, S., Murphy, D., Firoozfar, Z., Nasab, M.E., Bahreini, A., Ghasemi, M., Haridy, N.A., Goldouzi, H.R., Eghbal, F., Karimiani, E.G., Begtrup, A., Elloumi, H., Srinivasan, V.M., Gowda, V.K., Du, H., Jhangiani, S.N., **Coban-Akdemir, Z.** ... & Lupski, J.R., and Nguyen, L.N. (2024). Biallelic variation in the choline and ethanolamine transporter FLVCR1 underlies a severe developmental disorder spectrum. *Genet Med*, 101273. 10.1016/j.gim.2024.101273.
- Chong, J.X., Berger, S.I., Baxter, S., Smith, E., Xiao, C., Calame, D.G., Hawley, M.H., Rivera-Munoz, E.A., DiTroia, S., Genomics Research to Elucidate the Genetics of Rare Diseases, C., Bamshad, M.J., and

- Rehm, H.L. (2024). Considerations for reporting variants in novel candidate genes identified during clinical genomic testing. *Genet Med* 26, 101199. 10.1016/j.gim.2024.101199.
- Coban-Akdemir, Z.**, Song, X., Ceballos, F. C., Pehlivan, D., Karaca, E., Bayram, Y., ... & Lupski, J. R. (2024). The impact of the Turkish population variome on the genomic architecture of rare disease traits. *Genetics in Medicine Open*, 101830.
- Copeland, I., Wonkam-Tingang, E., Gupta-Malhotra, M., Hashmi, S.S., Han, Y., Jajoo, A., Hall, N.J., Hernandez, P.P., Lie, N., Liu, D., Xu, J., Rosenfeld, J., Haldipur, A., Desire, Z., **Coban-Akdemir, Z.H.**, Scott, D.A., Li, Q., Chao, H.T., Zaske, A.M., Lupski, J.R., Milewicz, D.M., Shete, S., Posey, J.E., and Hanchard, N.A. (2024). Exome sequencing implicates ancestry-related Mendelian variation at SYNE1 in childhood-onset essential hypertension. *JCI Insight* 9. 10.1172/jci.insight.172152.
- Dardas, Z., Fatih, J.M., Jolly, A., Dawood, M., Du, H., Grochowski, C.M., Jones, E.G., Jhangiani, S.N., Wehrens, X.H.T., Liu, P., Bi, W., Boerwinkle, E., Posey, J.E., Muzny, D.M., Gibbs, R.A., Lupski, J.R., **Coban-Akdemir, Z.***, and Morris, S.A.* (2024). NODAL variants are associated with a continuum of laterality defects from simple D-transposition of the great arteries to heterotaxy. *Genome Med* 16, 53. 10.1186/s13073-024-01312-9. (* Co-corresponding authors)
- Du, H., Dardas, Z., Jolly, A., Grochowski, C.M., Jhangiani, S.N., Li, H., Muzny, D., Fatih, J.M., Yesil, G., Elcioglu, N.H., Gezdirici, A., Marafi, D., Pehlivan, D., Calame, D.G., Carvalho, C.M.B., Posey, J.E., Gambin, T., **Coban-Akdemir, Z.**, and Lupski, J.R. (2024). HMZDupFinder: a robust computational approach for detecting intragenic homozygous duplications from exome sequencing data. *Nucleic Acids Res* 52, e18. 10.1093/nar/gkad1223.
- Duan, R., Marafi, D., Xia, Z.J., Ng, B.G., Maroofian, R., Sumya, F.T., Saad, A.K., Du, H., Fatih, J.M., Hunter, J.V., Elbendary, H.M., Baig, S.M., Abdullah, U., Ali, Z., Efthymiou, S., Murphy, D., Mitani, T., Withers, M.A., Jhangiani, S.N., **Coban-Akdemir, Z.**, Calame, D.G., Pehlivan, D., Gibbs, R.A., Posey, J.E., Houlden, H., Lupashin, V.V., Zaki, M.S., Freeze, H.H., and Lupski, J.R. (2023). Biallelic missense variants in COG3 cause a congenital disorder of glycosylation with impairment of retrograde vesicular trafficking. *J Inherit Metab Dis* 46, 1195-1205. 10.1002/jimd.12679.
- Petit, F., Longoni, M., Wells, J., Maser, R.S., Bogenschutz, E.L., Dysart, M.J., Contreras, H.T.M., Frenois, F., Pober, B.R., Clark, R.D., Giampietro, P.F., Ropers, H.H., Hu, H., Loscertales, M., Wagner, R., Ai, X., Brand, H., Jourdain, A.S., Delrue, M.A., Gilbert-Dussardier, B., Devisme, L., Keren, B., McCulley, D.J., Qiao, L., Hernan, R., Wynn, J., Scott, T.M., Calame, D.G., **Coban-Akdemir, Z.**, Hernandez, P., Hernandez-Garcia, A., Yonath, H., Lupski, J.R., Shen, Y., Chung, W.K., Scott, D.A., Bult, C.J., Donahoe, P.K., and High, F.A. (2023). PLS3 missense variants affecting the actin-binding domains cause X-linked congenital diaphragmatic hernia and body-wall defects. *Am J Hum Genet* 110, 1787-1803. 10.1016/j.ajhg.2023.09.002.
- Poli, M.C., Rebolledo-Jaramillo, B., Lagos, C., Orellana, J., Moreno, G., Martin, L.M., Encina, G., Bohme, D., Faundes, V., Zavala, M.J., Hasbun, T., Fischer, S., Brito, F., Araya, D., Lira, M., de la Cruz, J., Astudillo, C., Lay-Son, G., Cares, C., Aracena, M., Martin, E.S., **Coban-Akdemir, Z.**, Posey, J.E., Lupski, J.R., and Repetto, G.M. (2024). Decoding complex inherited phenotypes in rare disorders: the DECIPHERD initiative for rare undiagnosed diseases in Chile. *Eur J Hum Genet*. 10.1038/s41431-023-01523-0
- Holmstrom, L., Chaudhary, N.S., Nakamura, K., Chugh, H., Uy-Evanado, A., Norby, F., Metcalf, G.A., Menon, V.K., Yu, B., Boerwinkle, E., Chugh, S.S., **Akdemir, Z.**, and Kransdorf, E.P. (2023). Rare Genetic Variants Associated With Sudden Cardiac Arrest in the Young: A Prospective, Population-Based Study. *Circ Genom Precis Med*, e004105. 10.1161/CIRCGEN.123.004105.
- Pan, X., Yun, J., **Coban Akdemir, Z.H.***, Jiang, X., Wu*, E., Huang, J.H.*, Sahni, N.*, and Yi, S.S.* (2023). AI-DrugNet: A network-based deep learning model for drug repurposing and combination therapy in neurological disorders. *Comput Struct Biotechnol J* 21, 1533-1542. 10.1016/j.csbj.2023.02.004. (*Co-corresponding authors)
- Klonowski, J., Liang, Q., **Akdemir, Z.C.**, Lo, C., and Kostka, D. (2023). aenmd: Annotating escape from nonsense-mediated decay for transcripts with protein-truncating variants. *bioRxiv*. 10.1101/2023.03.17.533185.
- Li, M.M., Awasthi, S., Ghosh, S., Bisht, D., **Coban Akdemir, Z.H.**, Sheynkman, G.M., Sahni, N., and Yi, S.S. (2023). Gain-of-Function Variomics and Multi-omics Network Biology for Precision Medicine. *Methods Mol Biol* 2660, 357-372. 10.1007/978-1-0716-3163-8_24.
- Pan, X., **Coban Akdemir, Z.H.**, Gao, R., Jiang, X., Sheynkman, G.M., Wu, E., Huang, J.H., Sahni, N., and Yi, S.S. (2023). AD-Syn-Net: systematic identification of Alzheimer's disease-associated mutation and co-mutation vulnerabilities via deep learning. *Brief Bioinform* 24. 10.1093/bib/bbad030.
- Calame, D.G., Guo, T., Wang, C., Garrett, L., Jolly, A., Dawood, M., Kurolap, A., Henig, N.Z., Fatih, J.M., Herman, I., Du, H., Mitani, T., Becker, L., Rathkolb, B., Gerlini, R., Seisenberger, C., Marschall, S., Hunter, J.V., Gerard, A., Heidlebaugh, A., Challman, T., Spillmann, R.C., Jhangiani, S.N., **Coban- Akdemir, Z.**, Lalani, S., Liu, L., Revah-Politi, A., Iglesias, A., Guzman, E., Baugh, E., Boddaert, N., Rondeau, S., Ormieres, C., Barcia,

G., Tan, Q.K.G., Thiffault, I., Pastinen, T., Sheikh, K., Biliciler, S., Mei, D., Melani, F., Shashi, V., Yaron, Y., Steele, M., Wakeling, E., Ostergaard, E., Nazaryan-Petersen, L., Undiagnosed Diseases, N., Millan, F., Santiago-Sim, T., Thevenon, J., Bruel, A.L., Thauvin-Robinet, C., Popp, D., Platzer, K., Gawlinski, P., Wiszniewski, W., Marafi, D., Pehlivan, D., Posey, J.E., Gibbs, R.A., Gailus-Durner, V., Guerrini, R., Fuchs, H., Hrabe de Angelis, M., Holter, S.M., Cheung, H.H., Gu, S., and Lupski, J.R. (2023). Monoallelic variation in DHX9, the gene encoding the DEXH-box helicase DHX9, underlies neurodevelopment disorders and Charcot-Marie-Tooth disease. *Am J Hum Genet* *110*, 1394-1413. 10.1016/j.ajhg.2023.06.013.

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Petit, F., Longoni, M., Wells, J., Maser, R.S., Bogenschutz, E.L., Dysart, M.J., Contreras, H.T.M., Frenois, F., Pober, B.R., Clark, R.D., Giampietro, P.F., Ropers, H.H., Hu, H., Loscertales, M., Wagner, R., Ai, X., Brand, H., Jourdain, A.S., Delrue, M.A., Gilbert-Dussardier, B., Devisme, L., Keren, B., McCulley, D.J., Qiao, L., Hernan, R., Wynn, J., Scott, T.M., Calame, D.G., **Coban-Akdemir, Z.**, Hernandez, P., Hernandez-Garcia, A., Yonath, H., Lupski, J.R., Shen, Y., Chung, W.K., Scott, D.A., Bult, C.J., Donahoe, P.K., and High, F.A. (2023). PLS3 missense variants affecting the actin-binding domains cause X-linked congenital diaphragmatic hernia and body-wall defects. *Am J Hum Genet* *110*, 1787- 1803. 10.1016/j.ajhg.2023.09.002.

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Dawood, M., Akay, G., Mitani, T., Marafi, D., Fatih, J.M., Gezdirici, A., Najmabadi, H., Kahrizi, K., Punetha, J., Grochowski, C.M., Du, H., Jolly, A., Li, H., **Coban-Akdemir, Z.**, Sedlazeck, F.J., Hunter, J.V., Jhangiani, S.N., Muzny, D., Pehlivan, D., Posey, J.E., Carvalho, C.M.B., Gibbs, R.A., and Lupski, J.R. (2023). A biallelic frameshift indel in PPP1R35 as a cause of primary microcephaly. *Am J Med Genet A* *191*, 794-804. 10.1002/ajmg.a.63080.

Du, H., Jolly, A., Grochowski, C.M., Yuan, B., Dawood, M., Jhangiani, S.N., Li, H., Muzny, D., Fatih, J.M., **Coban-Akdemir, Z.**, Carlin, M.E., Scheuerle, A.E., Witzl, K., Posey, J.E., Pendleton, M., Harrington, E., Juul, S., Hastings, P.J., Bi, W., Gibbs, R.A., Sedlazeck, F.J., Lupski, J.R., Carvalho, C.M.B., and Liu, P. (2022). The multiple de novo copy number variant (MdnCNV) phenomenon presents with peri-zygotic DNA mutational signatures and multilocus pathogenic variation. *Genome Med* *14*, 122. 10.1186/s13073-022-01123-w.

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D. Abstracts Presented Based on Posters or Oral Presentations

American Society of Human Genetics Conference 2022

Unraveling the genomic basis of congenital heart disease from Gabriella Miller Kids First Pediatric Research Program: A Preliminary data.

American Society of Human Genetics Conference 2022

Escape from Nonsense-Mediated Decay: Annotation of Transcripts with Protein Truncating Variants.

American Society of Human Genetics Conference 2022

Comparison of chromosomal inversions in three different datasets.

European Society of Human Genetics Conference 2021

Elucidating the non-canonical rules of NMD in human using genomic approaches.

American Society of Human Genetics Virtual Conference 2021

Organizer of the invited session entitled "Nonsense-mediated decay: A double-edged sword in cancer and genetic diseases"

International Society of Computational Biology 2021

Organizer of the invited session entitled "Emerging gain-of-function mutations and their characterization by multi-omics network biology"

European Human Genetics Virtual Conference 2021

De novo mutation in ancestral generations evolves haplotypes contributing to disease

American Society of Human Genetics Houston, TX (2019)

Long-sized AOH regions reveal IBD haplotypes that drive Mendelian disease through mutational burden at a locus

Genomics of Rare Disease, Wellcome Genome Campus, Hinxton, UK (2018)

A Systematic Analysis of De Novo Mutations in 837 trios with clinical suspicion of Mendelian phenotypes

American Society of Human Genetics San Diego, CA (2018)

A Systematic Analysis of De Novo Mutations in 837 Trios with Clinical Suspicion of Mendelian Phenotypes

The NHGRI Genome Sequencing Program (GSP) In-Person Meeting, Bethesda, MD (2017)

When are (LOF) mutations not LOF mutations?

American Society of Human Genetics Orlando, FL (2017)

When are predicted loss-of-function (LOF) mutations not LOF mutations?

III. TEACHING INFORMATION

A. Educational Leadership Roles (title, dates, responsibilities)

N/A

B. Didactic Coursework

The University of Texas Health Science Center at Houston, School of Public Health, Houston, Texas
USA

Instructor

Duties have included leading lectures and preparing homework and exams

- Epidemiology I, Spring 2024

The University of Texas Health Science Center at Houston, School of Public Health, Houston, Texas
USA

Instructor

Duties have included leading lectures and preparing homework and exams

- Epidemiology I, Spring 2023

The University of Texas Health Science Center at Houston, School of Public Health, Houston, Texas
USA

Instructor

Duties have included leading lectures, weekly labs and grading homework and exams

- Genetic Epidemiology, Summer 2024.

The University of Texas Health Science Center at Houston, School of Public Health, Houston, Texas
USA

Instructor

Duties have included leading lectures, weekly labs and grading homework and exams

- Genetic Epidemiology, Summer 2023.

The University of Texas Health Science Center at Houston, School of Public Health, Houston, Texas
USA

Instructor

Duties have included leading lectures, weekly labs and grading homework and exams

- Genetic Epidemiology, Summer 2022.

The University of Texas Health Science Center at Houston, School of Public Health, Houston, Texas
USA

Instructor

Duties have included leading lectures and preparing homework and exams

- Epidemiology I, Spring 2022

The University of Texas Health Science Center at Houston, School of Public Health, Houston, Texas USA

Co-instructor

Duties have included leading lectures, weekly labs, grading homework and exams

- Applied Genetic Methods in Public Health, Summer 2021.

The University of Texas Health Science Center at Houston, Graduate School of Biomedical Sciences,
Houston, Texas USA

Teaching Assistant

Duties have included leading discussion classes and grading exams

- Molecular Biology of Eukaryotic Cells, Spring 2013.

Harvard University, Boston, Massachusetts USA

Teaching Assistant

- Analysis of Rates&Proportions, Fall 2009

Duties have included office hours, leading weekly labs and grading homework and exams Sabanci

University, Istanbul, Turkey

Teaching Assistant

Duties have included leading weekly labs

- Statistical Modeling, Spring 2007

Teaching Assistant

Duties have included leading weekly labs

- Introduction to Probability & Statistics, Fall 2006.

Peer-Teaching Assistant, Academic Support Program (ASP)

Duties have included helping students in their studies by organizing workshops, seminars, and social events and providing guidance to student and providing assistance for the Science of Nature course

C. Curriculum Development Work (include institution where work was done and whether work was disseminated beyond home institution)

1. N/A
2. N/A
3. N/A

D. Non-didactic Teaching (include institution where work was done)

1. N/A
2. N/A
3. N/A
4. N/A
5. N/A

E. Faculty Development or Continuing Medical Education (list contributions as a course developer, presenter, etc.)

N/A

IV. SERVICE CONTRIBUTIONS

A. Administrative Assignments and Committees

Faculty advisor to the students:

Renee Acquaye
(MPH, Graduated) Shoaib
Aziz (MPH, Graduated)
Barbara Bell (MPH)
Sasha Borisewitz (MPH)
Christopher Burris (MPH)
Amelia Collins (MPH)
Harsh Chauhan (MPH)
Sagar Mukhida (MPH)
Iman Egab (PhD)
Jiaoyang Xu (PhD)
Sagar Sayroz Mukhida (Practicum)
Raj Patel (Practicum)
Janica Gala (Practicum)
Barbara Bell (Practicum)

Thesis committee member to the students:

Jonathan Klonowski (UPittsburg, Ph.D. Thesis Committee, Graduated)
Andy Rivera Munoz (BCM, Ph.D. Thesis Committee)
Zain Dardas (BCM, Ph.D. Thesis Committee, Graduated)
Angad Jolly (BCM, Ph.D. Thesis Committee, Graduated)
Moez Dawood (BCM, Ph.D. Thesis Committee)
Nikhita Gogate (BCM, Ph.D. Thesis Committee)
Rachel Friedman (UTHealth SPH, Master's Thesis Committee)
Tugce Bozkurt (Acibadem University, Ph.D. Thesis Committee)
Alanna Cecchi (GSBS, Ph.D. Thesis Committee)
Katherine Ludorf (UTHealth SPH, Ph.D. Thesis Committee)
Shaghayegh Beheshti (BCM, Ph.D. Thesis Committee)

B. National, Regional or Local Participation in Professional or Voluntary Organizations

Member, American Society of Human Genetics Faculty Member,
Faculty, GSBS, MD Anderson
Adjunct Faculty, BCM, HGC
Early Career Reviewer, NIH GCAT study section Member,
Epidemiology Curriculum Committee
Member, The Genomics Research to Elucidate the Genetics of Rare Diseases (GREGoR) program Data Standards and Analysis Committee Member

C. Other Pertinent Information (not given above, including community service)

N/A