

# Anokhi Kashiparekh

aak104@pitt.edu

## Education

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### PhD in Human Genetics

2020- 2025

University of Pittsburgh Graduate School of Public Health, Pittsburgh, PA

### Bachelor of Science in Genetics and Genomics *with* Special Honors in Biology

2015- 2018

### Bachelor of Arts in Health and Society

University of Texas at Austin, Austin, TX

### Bachelor of Arts in Hindustani Music: Vocal

2003- 2013

Gandharva Mahavidyalaya, Gujarat, India

Certification: **First Class**

## Technical Experience

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### Rangos Research Center at UPMC Children's Hospital, Pittsburgh, PA

January 2022- Present

*PhD candidate at the Gonzalez Lab*

Supervisor: Dr. Lina Ghaloul Gonzalez

#### **Project: Unveiling novel insights into the pathophysiology of TANGO2 Deficiency Disorder (TDD) using integrated functional and multi-omic studies**

##### **Aim 1: Elucidate the TANGO2 protein network using functional and proteomic studies**

- Developed and optimized a co-immunoprecipitation protocol for isolating and characterizing TANGO2 binding partners from diverse biological sources, including HEK293T cells, mouse tissues, and patient-derived skin fibroblasts.
- Implemented a pipeline to analyze and calculate the statistical Protein-Protein Interaction probabilities.
- Performed subcellular and submitochondrial fractionation to determine the precise localization and expression patterns of TANGO2 within cellular and mitochondrial compartments.

##### **Aim 2: Uncover TANGO2-associated biochemical networks through transcriptomics, metabolomics and lipidomics**

- Generated novel multi-omic datasets (transcriptomic, metabolomic, and lipidomic) from patient fibroblasts, establishing a public resource to support further TANGO2 research.
- Developed and automated data analysis pipelines using RStudio, Unix, and Python, enabling comprehensive pathway analysis across multi-omic datasets.
- Implemented an integrative analysis framework to synergize different omic layers, revealing underlying biochemical dysfunctions and compensatory mechanisms in TDD pathophysiology.

##### **Aim 3: Validate TDD biochemical signatures and downstream effects of targeted therapies on TDD-derived skin fibroblasts**

- Applied multi-omic integration to elucidate the biochemical mechanisms by which Vitamin B5 ameliorates specific TDD symptoms.
- Validated key biochemical dysfunctions identified in omics studies, such as Reactive Oxygen Species (ROS) accumulation, altered Oxygen Consumption Rate (OCR), and post-translational modifications, through targeted functional assays and immunofluorescence imaging.
- Assessed the therapeutic potential of candidate compounds targeting specific biomarkers, evaluating their efficacy in mitigating biochemical disruptions associated with TDD.

**Additional projects** include investigating biochemical changes associated with a novel PNPT1 variant using ROS, OCR, and fatty acid flux assays. Collaborated on a natural history study for GUCY2C, identifying variants through PCR and gel electrophoresis, followed by interpretation of Sanger sequencing results.

### Magee-Womens Research Institute, Pittsburgh, PA

August 2020- December 2021

*Graduate Student Researcher at the Lee/ Osterreich Lab*

Supervisor: Dr. Adrian Lee and Dr. Steffi Osterreich

#### **Project: Invasive Lobular Cancer Cell Line Encyclopedia (ICLE)**

- Contributed to a large-scale characterization of Invasive Lobular Cancer (ILC) and ILC-like cells, establishing a comparative framework with Invasive Ductal Cancer (IDC) cells to develop an open-access resource.
- Conducted phenotypic assays and utilized HTC cluster resources to analyze Whole Exome Sequencing, RNA-sequencing, and RPPA datasets, providing insights into molecular distinctions among ILC, ILC-like, and IDC cell lines.

**Massachusetts General Hospital, Boston, MA**

March 2019- July 2020

*Research Technician II at the Yun Bio-Optics Lab*

Supervisor: Dr. Seok-Hyun (Andy) Yun

**Project 1: Optimizing *in vitro* bioluminescence photodynamic therapy to treat breast cancer in mice models**

- Synthesized luciferase-photosensitizer conjugates to investigate therapy effects and biodistribution *in vitro* and *in vivo*.
- Assessed lymph node retention and filtration capacity by creating mouse window chamber models followed by two-photon imaging

**Project 2: Fabrication of semiconductor microlasers and understanding their impact in biological systems**

- Created tissue models and delivery systems for microlaser tracking for microfluidic cell sorting applications.
- Purified and extracted RNA for subsequent spatial transcriptomic analysis of *in vitro* laser uptake.
- Optimized the ICC staining of endosomes, lysosomes and cytoskeleton to visualize laser location using confocal imaging.

**Managerial duties** included general maintenance of the tissue culture room, acting as biosafety officer and main liaison for the lab, ensuring that the lab and all protocols comply with the IACUC, EHS and Partners policies, maintaining controlled substances, and coordinating lab meetings.

**University of Regensburg, Regensburg, Germany**

June 2018- August 2018

*Undergraduate Research Assistant at the Brembs Lab*

Supervisor: Dr. Björn Brembs

**Project: Understand the effect of calcium signalling on spontaneous behaviour in fruit flies**

- Created *SERCA KD* *Drosophila* flies using RNAi and monitored them in a specialized flight simulator.
- Analyzed fly wing patterns to determine spontaneous directional preference using RStudio.

**University of Texas at Austin, Austin, TX**

August 2017 - December 2018

*Undergraduate Research Assistant at the Hofmann Lab*

Supervisors: Dr. Hans Hofmann and Dr. Rebecca Young-Brim

**Honours thesis: Characterise the transcriptomic and developmental mechanisms underlying a maternally-induced growth acceleration in House finches**

- Analyzed RNA-seq data obtained from House finch embryos using a Linux/ Unix environment (TACC) for transcriptomic analysis including quality control, pseudoalignment and differential gene expression.
- Carried out methods for data clustering, statistical analysis and visualization to compare gene expression profiles.

**Indian Institute of Science, Bangalore, India**

June 2017- August 2017

*Undergraduate Research Assistant at Prof. Visweswariah Lab*

Supervisor: Dr. Sandhya Visweswariah and Dr. Sveta Chakrabarti

**Project 1: Understand the effect of biotic stressors on the *CG16717* knockout *Drosophila melanogaster***

- Used the Gal4/UAS system to create *CG16717* overexpressing and rescue *Drosophila* fly lines.
- Inoculated multiple bacterial strains to infect *Drosophila* flies orally and systemically, and conducted a survival analysis.

**Project 2: Create *D. melanogaster* transgenic lines expressing cAMP and cGMP FRET sensors**

- Used an ApE reference sequence assembly to generate recombinant plasmids expressing a FRET pair and GAFa or Epac domain, to sense cAMP or cGMP in infected tissues.

**Insitome, Inc., Austin, TX**

December 2017- January 2018

*Bioinformatics Intern*

Supervisors: Dr. Spencer Wells and Dr. Gareth Highnam

- Optimized and reimplemented the HIRISplex method in Python to predict the hair and eye colour from 23 DNA markers to be incorporated into the Insitome suite of genetic testing apps.

**University of Texas at Austin, Austin, TX**

September 2016- May 2017

*Undergraduate Research Assistant at the Preston Lab*

Supervisor: Dr. Alison Preston

**Project: Examine the extent to which awake rest promotes the consolidation of integrated memories by the functional coupling between the hippocampus and the medial prefrontal cortex in adults versus children**

- Operated the fMRI to collect functional data and analysed it using MATLAB.
- Collaborated in creating tasks and stimuli for testing spatial navigation and memory integration, and interviewing test subjects

## University of Texas at Austin, Austin, TX

January 2017- December 2018

### Statistical Analyst for the College of Natural Sciences

- Analysed student success and predicted enrollment statistics for each college course taught at CNS to ultimately recommend changes in certificates and introductory STEM courses taught at UT
- Looked at student satisfaction through surveys and maintained student records using R and Python

## University of Texas at Austin, Austin, TX

January 2017- December 2018

### STEM Study Center Head Tutor for the College of Natural Sciences

- Drop-in tutor for Introductory Biology and Chemistry, Genetics, Biochemistry and Biostatistics courses
- Proctor for UT STEM examinations for incoming freshmen to assess student placement in specific STEM classes
- Compiled CNS testing and tutor schedules (Excel and R) to ensure tutor availability on testing dates
- Interviewed and supervised new tutors, and coordinated with the program manager regarding student feedback

## University of Texas at Austin, Austin, TX

September 2016-December 2016

### Undergraduate Tutor for the Sanger Learning Center

- Drop-in and one-on-one tutor for Introductory Biology, Chemistry and Calculus courses to about 25 students per week
- Led review sessions for different classes of 10-15 people for upper-division Biology and Chemistry courses

## Publications

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- Shah, O., Chen, F., Webn, A., [Kashiparekh, A.](#), Savariau, L., Clifford, B., ... & Lee, A. V. (2023). The invasive lobular cancer cell line encyclopedia (ICLE) for studying potential biomarkers and exploring therapeutic opportunities. *bioRxiv*, 2023-09.
- Yan, H., Forward, S., Kim, K. H., Wu, Y., Hui, J., [Kashiparekh, A.](#), & Yun, S. H. (2023). All-natural-molecule, bioluminescent photodynamic therapy results in complete tumor regression and prevents metastasis. *Biomaterials*, 296, 122079.
- Dannenber, P. H., Kang, J., Martino, N., [Kashiparekh, A.](#), Forward, S., Wu, J., Liapis, A., Wang, J., & Yun, S. H. (2022). Laser particle activated cell sorting in microfluidics. *Lab on a Chip*, 22(12), 2343-2351.
- Dannenber, P. H., Liapis, A. C., Martino, N., Kang, J., Wu, Y., [Kashiparekh, A.](#), & Yun, S. H. (2021). Multilayer Fabrication of a Rainbow of Microdisk Laser Particles Across a 500 nm Bandwidth. *ACS Photonics*, 8(5), 1301-1306.

## Conference & Research Presentations

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- [Kashiparekh, A.](#), Powers, A., Mohsen, W., Vockley, J., Ghaloul-Gonzalez, L. Unveiling novel insights into the pathophysiology of TANGO2 deficiency disorder (TDD) using integrated functional and multiomic studies. PittGene 2024, Pittsburgh, PA, 2024. (Oral Presentation)
- [Kashiparekh, A.](#), Mohsen, W., Vockley, J., Ghaloul-Gonzalez, L. Multi-omics approach to characterize the TANGO2 function: dance partners and potential therapies., Society for the Study of Inherited Errors of Metabolism, Porto, Portugal, 2024. (Guided Poster Talk)
- [Kashiparekh, A.](#), Powers, A., Mohsen, W., Vockley, J., Ghaloul-Gonzalez, L. Unveiling novel insights into the pathophysiology of TANGO2 deficiency disorder (TDD) using integrated functional and multiomic studies. International Network for Fatty Acid Oxidation Research and Management, Porto, Portugal, 2024. (Poster)
- [Kashiparekh, A.](#), Mohsen, W., Vockley, J., Ghaloul-Gonzalez, L. Multi-omics approach to characterize the TANGO2 function: dance partners and potential therapies., Society of Inherited Metabolic Disorders, Charlotte, 2024. (Poster)
- [Kashiparekh, A.](#), Powers, A., Mohsen, W., Vockley, J., Ghaloul-Gonzalez, L. Multi-omics approach to characterize the TANGO2 function: dance partners and potential therapies. American Society of Human Genetics Annual Meeting, Washington DC, 2023. (Oral Presentation)
- [Kashiparekh, A.](#), Powers, A., Mohsen, W., Vockley, J., Ghaloul-Gonzalez, L. Multi-omics approach to characterize the TANGO2 function: dance partners and potential therapies. PittGene, Pittsburgh, PA, 2023 (Poster)

- Kashiparekh, A., Powers, A., Mohsen, W., Vockley, J., Ghaloul-Gonzalez, L. Multi-omics approach to characterize the TANGO2 function: dance partners and potential therapies, Children's Hospital Research Symposium, Pittsburgh, PA, 2023 (Poster)
- Kashiparekh, A., Vockley, CW., Badway, G., Powers, A., Karunanidhi, A., Ghaloul-Gonzalez, L. Investigating a novel PNPT1 variant in the Amish population. Children's Hospital Research Symposium, Pittsburgh, PA, 2023 (Poster)
- Kashiparekh, A., Vockley, CW., Badway, G., Powers, A., Karunanidhi, A., Ghaloul-Gonzalez, L. Investigating a novel PNPT1 variant in the Amish population. Dean's Day, Pittsburgh, PA, 2023 (Poster)
- Kashiparekh, A., Vockley, CW., Badway, G., Powers, A., Karunanidhi, A., Ghaloul-Gonzalez, L. Investigating a novel PNPT1 variant in the Amish population. PittGene, Pittsburgh, PA, 2022 (Poster)
- Forward, S., Yan, H., Kim, K.H., Kashiparekh, A., Kowk S.J., Yun S.H. Luciferase-Chlorin e6 Conjugates for Bioluminescent Photodynamic Therapy. MRS Fall Meeting & Exhibit, Boston, MA, 2019. (Invited Oral Presentation)
- Kashiparekh, A., Chakrabarti, S., Visweswariah, S. Understand the effect of biotic stressors on the *CG16717* knockout *Drosophila melanogaster*. Harvard National Collegiate Research Conference, Cambridge, MA, 2018. (Poster)
- Kashiparekh, A., Hayward, M. Prisoner Rights and the Human right to healthcare. The Undergraduate Awards, Dublin, Ireland, 2018. (Invited Oral Presentation)
- Kashiparekh, A., Chakrabarti, S., Visweswariah, S. Understand the effect of biotic stressors on the *CG16717* knockout *Drosophila melanogaster*. Gulf Coast Undergraduate Research Symposium, Houston, TX, 2018. (Speaker)
- Kashiparekh, A., Hayward, M. Prisoner Rights and the Human right to healthcare. Liberal Arts Dean's Research Reception, Austin, TX, 2018. (Poster)
- Kashiparekh, A., Young R.L., Hofmann H. Characterize the Transcriptomic and Developmental Mechanisms Underlying a Maternally-Induced Growth Acceleration in House Finches. UTSA College of Sciences Research Conference, San Antonio, TX, 2018. (Invited Poster)
- Kashiparekh, A., Chakrabarti, S., Visweswariah, S. Understand the effect of biotic stressors on the *CG16717* knockout *Drosophila melanogaster*. Fall Undergraduate Research Symposium, Austin, TX, 2017. (Oral Presentation)
- Kashiparekh, A., Coughlin C., Preston A., Rest Effects on Memory Integration: A Developmental Investigation. 9<sup>th</sup> Annual Neuroscience Symposium, College Station, TX, 2017. (Poster)
- Kashiparekh, A. The Use of Mixl1 in Ex Vivo and Chimeric Organ Regeneration. Undergraduate Research Week, Austin, TX, 2017. (Poster)

## Honours and Awards

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<b>SSIEM Travel Award</b> , Society for the Study of Inherited Errors of Metabolism, Porto	Fall 2024
<b>Guided poster talk</b> , Society for the Study of Inherited Errors of Metabolism, Porto	Fall 2024
<b>Second Highest Ranked Poster</b> , International Network for Fatty Acid Oxidation Research and Management, Porto	Fall 2024
<b>Second place- Three Minute Thesis</b> , University of Pittsburgh Graduate School of Public Health	Spring 2024
<b>Platform talk</b> , American Society of Human Genetics, DC	Fall 2023
<b>Honourable mention</b> , Children's Hospital Research Symposium, Pittsburgh	Fall 2022
<b>GCURS Symposium Travel Award</b> , Rice University	Fall 2018
<b>Charles Ely Lankford Memorial Scholarship</b> , UT Austin	Fall 2018
<b>Undergraduate Research Fellowship</b> , UT Austin	Spring 2018
<b>Liberal Arts Undergraduate Chapter for Research- Research Scholarship</b> , UT Austin	Spring 2018
<b>College of Liberal Arts Undergraduate Research Scholarship</b> , UT Austin	Fall 2018
<b>Scholastic Out-of-State Tuition Waiver from the College of Natural Sciences</b> , UT Austin	
<b>University Honours</b> , UT Austin	
<b>Dean's Honour List</b> , UT Austin	

**Dean's Research Reception List**, UT Austin

**Third Place, Debate competition** Income Tax Department of India

**Third highest scorer for state**, National Science Olympiad, Gujarat

**Honoured and selected to sing in front of the Prime Minister of India**, Gujarat

## Skills

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### **Computational biology:**

- Skilled in analyzing, integrating and interpreting transcriptomics, metabolomics (GCMS), lipidomics (LCMS) and proteomics (MS) data by developing statistical analysis pipeline using Rstudio..
- Proficient in MATLAB, Python (Biopython and PyMOL) and working in a high-performance computing Linux/ Unix environment (Texas Advanced Computing Center, O2 Harvard Medical School Research Computing and HTC at the University of Pittsburgh)
- Experience in working with DNA sequence alignment and analysis tools, RNA secondary structure prediction and covariation analysis softwares, and BLAST, NCBI GenBank, PDB, Ensembl and UCSC Genome browser

### **Wet bench biology:**

- Proficient in gel electrophoresis (agarose and polyacrylamide), PCR, qPCR, Western Blotting, bacterial transformation, Drosophila culture, DNA cloning using plasmid vectors (design, digestion, ligation, elution and miniprep), microscopy (two-photon, confocal and dissection), Immunoprecipitation and Immunohistochemistry.
- Skilled in performing functional assays like ROS measurements, Oxygen Consumption Rate (Seahorse assay), apoptosis assay and wound healing (migration) assays.
- Experience in working with mammalian cell culture (Skin Fibroblasts, HeLa, MCF7, MDCK II, OVCAR-8, 4T1, Beta cells, T cells), and bacterial cell culture (*E. coli*, *ECC15*), *D. melanogaster*, Arabidopsis, *A. domesticus*, *M. musculus* and Humans

### **Chemistry:**

- Proficient in performing protein purification, chromatography, spectrophotometry and titrations

### **Languages:**

Proficient- Gujarati, Hindi, English; Beginner- Sanskrit and German

## Leadership and Volunteering

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**Vice President**, *Student Government Association*, University of Pittsburgh Graduate School of Public Health

**Certified Sexual Assault Counselor**, *Pittsburgh Action Against Rape*, Pittsburgh

**Co-editor in chief**, *The Public Health Journal*, University of Pittsburgh Graduate School of Public Health

**Committee member**, *Inclusivity & Diversity Climate Committee*, University of Pittsburgh Graduate School of Public Health

**Lead Guitarist**, *Wellman Center for Photomedicine at Massachusetts General Hospital- Departmental Band*, Boston

**Mentor and Blogger**, *Students With Ambition Go To College*, Austin

**Honours member**, *Beta Beta Beta Biology Honours Society*, UT Austin

**Research Matching Committee**, *Research Student Advisory Committee*, UT Austin

**Authorized Representative**, *Delete Blood Cancer- Longhorn Chapter*, UT Austin

**Honours member**, *Burnt Orange Society - Office of the Dean of Students*, UT Austin

**Volunteer**, *Breast Cancer Resource Center*, Austin

**Co-organizer**, *TEDxSpeedway Plaza*, UT Austin