

Joseph K. Aicher

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Education

University of Pennsylvania, Philadelphia, PA

M.D. expected May 2024*

Ph.D. Genomics and Computational Biology, expected June 2022

Thesis topic: “Enhancing molecular diagnosis in children using splicing prediction algorithms”

University of Alabama, Tuscaloosa, AL

B.S. Mathematics and Physics, *summa cum laude*, May 2015

M.A. Mathematics, May 2015

Master’s thesis: “Metabolic network inference with the graphical Lasso”

Publications

Vaquero-Garcia J[†], **Aicher JK**[†], Jewell P[†], Gazzara MR[†], Radens CM, Jha A, Green CJ, Norton SS, Lahens NF, Grant GR, Barash Y. RNA splicing analysis using heterogeneous and large RNA-seq datasets [preprint]. bioRxiv; 2021 [under review at Nat Commun].

Lubin E, Bryant L, **Aicher J**, Li D, Bhoj E. Analysis of histone variant constraint and tissue expression suggests five potential novel human disease genes: H2AFY2, H2AFZ, H2AFY, H2AFV, H1F0. Hum Genet. 2022 Jan 24.

Aicher JK, Jewell P, Vaquero-Garcia J, Barash Y, Bhoj EJ. Mapping RNA splicing variations in clinically accessible and nonaccessible tissues to facilitate Mendelian disease diagnosis using RNA-seq. Genet Med. 2020;22(7):1181-1190.

Jha A, **K Aicher J**, R Gazzara M, Singh D, Barash Y. Enhanced Integrated Gradients: improving interpretability of deep learning models using splicing codes as a case study. Genome Biol. 2020;21(1):149. Published 2020 Jun 19.

Oza VH, **Aicher JK**, Reed LK. Random forest analysis of untargeted metabolomics data suggests increased use of omega fatty acid oxidation pathway in Drosophila melanogaster larvae fed a medium chain fatty acid rich high-fat diet. Metabolites. 2019;9(1):5.

Lawrimore J, **Aicher JK**, Hahn P, Fulp A, Kompa B, Vicci L, Falvo M, Taylor RM, Bloom K. ChromoShake: a chromosome dynamics simulator reveals that chromatin loops stiffen centromeric chromatin. Mol Biol Cell. 2016 Jan 1;27(1):153–166.

Aicher JK. Metabolic network inference with the graphical lasso [master’s thesis]. [Tuscaloosa (AL)]: University of Alabama; 2015. 31 p.

Baker PR, Friederich MW, Swanson MA, Shaikh T, Bhattacharya K, Scharer GH, **Aicher J**, Creadon-Swindell G, Geiger E, MacLean KN, Lee W-T, Deshpande C, Freckmann M-L, Shih L-Y, Wasserstein M, Rasmussen MB, Lund AM, Procopis P, Cameron JM, Robinson BH, Brown GK, Brown RM, Compton AG, Dieckmann CL, Collard R, Coughlin CR, Spector E, Wempe MF, Van Hove JLK. Variant non ketotic hyperglycinemia is caused by mutations in LIAS, BOLA3 and the novel gene GLRX5. Brain. 2014 Feb;137(2):366–379.

* On parental leave during Fall 2020, Summer 2022 semesters

† Co-first author

Oral Presentations

Aicher JK, Bhoj EJ, Barash Y. Fast and scalable RNA-seq splicing analysis for the clinical setting. Wellcome Genomics of Rare Disease. 2020 Mar.

Aicher JK, Bhoj EJ, Barash Y. Analysis of RNA splicing in clinically-accessible and non-accessible tissues: relevance to Mendelian disease diagnosis. Philadelphia Genetics Meeting. 2019 May 15.

Aicher JK, Bhoj EJ, Barash Y. Tools and evaluation for RNA-seq splicing quantification for suspected Mendelian disorders across tissues. Wellcome Genomics of Rare Disease. 2019 Mar 28.

Awards and Honors

NIH Ruth L. Kirschstein NRSA Individual Fellowship F30HD098803 (2019 – Present)

Blavatnik Family Fellowship in Biomedical Research Finalist (2019)

NIH Medical Scientist Training Program Grant T32GM007170 (2015 – 2018)

UA Distinguished Undergraduate Scholar (2015)

UA B. B. Comer Mathematics Medal (2015)

UA Department of Physics and Astronomy Outstanding Senior Award (2015)

UA Department of Physics and Astronomy First Year Student Award (2013)

UA Outstanding Organic Chemistry Student Award (2012)

National Merit Scholarship (2011-2015)

Research Experience

Research Assistant (April 2018 – Present)

Professor Yoseph Barash, Department of Genetics, University of Pennsylvania

Professor Elizabeth Bhoj, Department of Pediatrics, Children's Hospital of Philadelphia

- Developing and validating tissue- and variant-aware predictive models of alternative splicing using deep learning and diverse publicly available datasets
- Integrating leading splicing models into clinical human genetics pipelines to prioritize potential disease-causing intronic and exonic variants from patient exomes

Rotation Student (August 2017 – March 2018)

Professor Jennifer Phillips-Cremins, Department of Bioengineering, University of Pennsylvania

- Investigated interplay between disease-associated genetic variation, 3D chromatin structure, and gene expression in the context of sporadic and familial neurodegenerative disease

Rotation Student (June 2016 – August 2016)

Professor John Maris, Department of Pediatrics, Children's Hospital of Philadelphia

- Developed, simulated, and optimized pipelines for the analysis of RNA-Seq data from patient-derived tumor xenografts

Research Assistant (October 2013 – May 2015)

Professor Laura Reed, Department of Biology, University of Alabama

Professor Song Song, Department of Mathematics, University of Alabama

- Investigated the application of high-dimensional statistics techniques for use in understanding biological pathways and biomarker discovery
- Analyzed effects of diet and genetic variation on metabolic disease in *Drosophila* using metabolomics and gene expression studies, leading to publication

REU Student Researcher (May 2013 – August 2013, May 2014 – August 2014)

Professor Russell M. Taylor II, Department of Computer Science, University of North Carolina Chapel Hill

- Created integrated workflow for evaluating simulations and experiments studying the yeast mitotic spindle including BrightPixel, an ImageJ plugin for tracking multiple objects in 3D fluorescence microscopy videos
- Developed ChromoShake, a parallel simulation framework for studying Langevin dynamics of large biological polymers on multicore processors and GPUs, leading to publication

Research Assistant (August 2012 – May 2013, August 2013 – November 2013)

Professor William Butler, Department of Physics, University of Alabama

- Created and numerically implemented tight-binding models for understanding the properties of magnetic alloys
- Performed first-principles calculations using density functional theory to elucidate information about the properties of magnetic materials relevant to spintronics
- Investigated potential applications of topological insulators in semiconductors

Research Assistant (December 2011 – May 2012)

Professor Jerome Busenitz, Department of Physics, University of Alabama

- Measured and analyzed the activity of radioactive sources as part of the Double Chooz collaboration
- Assisted in the construction and calibration of a NaI scintillation detector
- Modeled the dynamics of the NaI detector for different radioactive sources with Monte Carlo methods using Geant4

Pediatrics Student Research Program Intern (June 2011 – July 2011)

Professor Johan Van Hove, Department of Clinical Genetics and Metabolism, University of Colorado Denver

- Applied molecular biology and bioinformatics techniques to identify novel disease-causing genes and mutations of variant non-ketotic hyperglycinemia, leading to publication

Research Assistant (January 2011 – June 2011, May 2012 – July 2012)

Professor Manuel Lladser, Department of Applied Mathematics, University of Colorado Boulder

- Researched techniques in probability and statistics relevant to problems in computational biology and theoretical computer science

Work Experience

Data Scientist Intern (May 2021 – August 2021)

Amazon

- Developed methods using NLP and causal inference to identify/prioritize new business opportunities for Amazon private labels

Learning Assistant (August 2012 – May 2014)

Professors William Butler and Tim Mewes, Department of Physics, University of Alabama

- Junior level electrodynamics sequence and honors introduction to electricity and magnetism
- Created new course material to encourage and evaluate student learning
- Designed and implemented lab experiments to encourage inquiry-based learning
- Delivered weekly tutorial and review sections and assisted in-class lectures

Activities

Penn GCB Student Programming Tutorial Lecturer (August 2019, August 2020)

Penn GCB Executive Committee (August 2018 – August 2020)

Penn GCB Programming Bootcamp TA (July 2018 – August 2018)

Penn MSTP Speaker Selection Committee (September 2017, September 2018)

Penn MSTP Steering Committee Student Representative (August 2017 – August 2019)

Huey/Comegys Health Sciences Exploration Program (January 2016 – May 2019)

Pi Mu Epsilon Math Honor Society (March 2012 – May 2015)

- President (Fall 2013 – May 2015)

Society of Physics Students (Fall 2011 – May 2015)

- President (Spring 2014 – May 2015)
- Vice President (Spring 2013 – Spring 2014)
- Secretary (Spring 2012 – Spring 2013)

Every Move Counts Chess Education Initiative (January 2015 – May 2015)

Discovery Buddies Matthews Elementary School Outreach (February 2014 – December 2014)

Volunteer AP Tutor at Hillcrest High School (February 2014 – May 2014)

Computer Skills

General programming languages: Python, C++, SQL, Fortran, Java, CUDA, OpenCL

Mathematical programming languages: R, NumPy, MATLAB

Machine learning: PyTorch, TensorFlow, scikit-learn

Bioinformatics: NGS analysis (e.g. RNA-Seq, WES, Hi-C), Bioconductor, ImageJ