Drew Hughes

Professional Profile

- Clinical Pathology resident applying machine learning to complex clinical laboratory data to standardize interpretation and increase throughput
- MD/PhD scientist with extensive experience developing and implementing next-generation sequencing assays and bioinformatic analyses
- Motivated by opportunities to work with multidisciplinary teams using machine learning to build next-generation diagnostics and improve patient care

Education

2009–2019 **MD/PhD, Molecular Genetics and Genomics**, *Washington University School of Medicine*, St. Louis, MO.

2004–2008 **BS (*summa cum laude*), Mathematics**, *College of William & Mary*, Williamsburg, VA.

Work Experience

2019–Present **Resident, Clinical Pathology**, *Barnes-Jewish Hospital*, St. Louis, MO.

- Developed machine learning models to automate the interpretation of serum protein electrophoresis to promote result standardization and reduce hands-on time
- Performed analysis for the clinical validation of a new molecular test (ChromoSeq) and prepared the associated regulatory documentation
- Worked with a multidisciplinary team to build an interactive dashboard summarizing respiratory pathogen test results to allow clinicians to visualize trends over time

Research Experience

2014–2017 **Washington University School of Medicine**, *Graduate Research Thesis*, St. Louis, MO.

- Mapped cell type-specific *cis*-regulatory elements in mouse photoreceptors by performing ATAC-seq and RNA-seq on FACS-purified rods and cones
- Developed machine learning models to identify sequence features associated with photoreceptor-specific regions of open chromatin
- Designed and implemented a massively parallel reporter assay to test quantitative models of *cis*-regulatory element activity *in vivo* at single nucleotide resolution

2011 **Washington University School of Medicine**, *Graduate Research Rotation*, St. Louis, MO.

- Used targeted single-cell sequencing to reconstruct the clonal architecture of tumor specimens from patients with secondary acute myeloid leukemia

2006–2008 **College of William & Mary**, *Undergraduate Research Thesis*, Williamsburg, VA.

- Characterized patterns of calcium-mediated signaling in developing excitatory and inhibitory neurons *in vivo* in the African clawed frog

Teaching Experience

2015, 2016 **Washington University School of Medicine**, *Teaching Assistant: Fundamentals of Biostatistics*, St. Louis, MO.

2011 **Washington University School of Medicine**, *Teaching Assistant: Human Anatomy and Development*, St. Louis, MO.

Skills

Data Science: machine learning | statistical modeling | data visualization | R {tidyverse} {tidymodels} {shiny} {Bioconductor} | Bash | Python | SQL | Git | high-performance computing.

Genomics: next-generation sequencing | nanopore sequencing | end-to-end bioinformatic analysis | variant calling | RNA-seq | ATAC-seq | ChIP-seq | massively parallel reporter assays.

Clinical Pathology: molecular diagnostics | clinical assay validation | clinical informatics | Epic | Cerner.

Honors and Awards

2021 Richard Marshall Education Award, American Association for Clinical Chemistry.

2021 Clinical Pathology Trainee of the Month, Department of Pathology & Immunology, Washington University School of Medicine.

2021 Best Abstract (Clinical and Diagnostic Immunology), American Association for Clinical Chemistry.

2021 Most Innovative Abstract, American Society for Histocompatibility and Immunogenetics.

2020 Abstract Achievement Award, American Society of Hematology.

2020 Best Laboratory and Genomic Medicine Grand Rounds Presentation, Department of Pathology & Immunology, Washington University School of Medicine.

2019 Clinical Pathology Trainee of the Month, Department of Pathology & Immunology, Washington University School of Medicine.

2019 Dr. Philip Rosenblatt Award, Department of Pathology & Immunology, Washington University School of Medicine.

2015 Vision Sciences Training Grant, Department of Ophthalmology & Visual Sciences, Washington University School of Medicine.

2014 Best Poster, Molecular Genetics and Genomics/Computational and Systems Biology Annual Retreat, Washington University School of Medicine.

2008 Phi Beta Kappa, College of William & Mary.

2008 Cissy Patterson Prize, Department of Mathematics, College of William & Mary.

2008 Honors, Department of Biology, College of William & Mary.

Peer-Reviewed Publications

2021 **Hughes AEO**, and Jackups R Jr. Clinical Decision Support for Laboratory Testing. *Clinical Chemistry* (in press).

2021 Duncavage EJ, Schroeder MC, O'Laughlin M, Wilson R, MacMillan S, Bohannon A, Kruchowski S, Garza J, Du F, **Hughes AEO**, Robinson J, Hughes E, Heath SE, Baty JD, Neidich J, Christopher MJ, Jacoby MA, Uy GL, Fulton RS, Miller CA, Payton JE, Link DC, Walter MJ, Westervelt P, DiPersio JF, Ley TJ, and Spencer DH. Genome Sequencing as an Alternative to Cytogenetic Analysis in Myeloid Cancers. *The New England Journal of Medicine*, 384(10), 924–935. PMID: 33704937. https://doi.org/10.1056/NEJMoa2024534.

2020 Perez-Cervantes C, Smith LA, Nadadur RD, **Hughes AEO**, Wang S, Corbo JC, Cepko C, Lonfat N, and Moskowitz IP. Enhancer Transcription Identifies *cis*-Regulatory Elements for Photoreceptor Cell Types. *Development*, 147(3), dev184432. PMID: 31915147. https://doi.org/10.1242/dev.184432.

2020 **Hughes AEO**, Webber DM, Wallace MA, Johnson C, Burnham CA, and Anderson NW. Comparable Detections of Viral Pathogens in Lower Respiratory Tract Specimens with the BioFire Respiratory Panel 2 and the BioFire Pneumonia Panel. *Journal of Clinical Microbiology*, 58(6), e00254-20. PMID: 32269103. https://doi.org/10.1128/JCM.00254-20.

2020 Volkov LI, Kim-Han JS, Saunders LM, Poria D, **Hughes AEO**, Kefalov VJ, Parichy DM, and Corbo JC. Thyroid Hormone Receptors Mediate Two Distinct Mechanisms of Long-Wavelength Vision. *Proceedings of the National Academy of Sciences of the United States of America*, 117(26), 15262–15269. PMID: 32541022. https://doi.org/10.1073/pnas.1920086117.

2019 Murphy DP, **Hughes AEO**, Lawrence KA, Myers CA, and Corbo JC. *Cis*-Regulatory Basis of Sister Cell Type Divergence in the Vertebrate Retina. *Elife*, 8, e48216. PMID: 31633482. https://doi.org/10.7554/eLife.48216.

2018 **Hughes AEO**, Myers CA, and Corbo JC. A Massively Parallel Reporter Assay Reveals Context-Dependent Activity of Homeodomain Binding Sites In Vivo. *Genome Research*, 28(10), 1520–1531. PMID: 30158147. https://doi.org/10.1101/gr.231886.117.

2017 **Hughes AEO**, Enright JM, Myers CA, Shen SQ, and Corbo JC. Cell Type-Specific Epigenomic Analysis Reveals a Uniquely Closed Chromatin Architecture in Mouse Rod Photoreceptors. *Scientific Reports*, 7, 43184. PMID: 28256534. https://doi.org/10.1038/srep43184.

2016 Shen SQ, Myers CA, **Hughes AEO**, Byrne LC, Flannery JG, and Corbo JC. Massively Parallel *cis*-Regulatory Analysis in the Mammalian Central Nervous System. *Genome Research*, 26(2), 238–55. PMID: 26576614. https://doi.org/10.1101/gr.193789.115.

2015 Young AL, Wong TN, **Hughes AEO**, Heath SE, Ley TJ, Link DC, and Druley TE. Quantifying Ultra-Rare Pre-Leukemic Clones Via Targeted Error-Corrected Sequencing. *Leukemia*, 29(7), 1608–11. PMID: 25644247. https://doi.org/10.1038/leu.2015.17.

2014 **Hughes AEO**, Magrini V, Demeter R, Miller CA, Fulton R, Fulton LL, Eades WC, Elliott K, Heath S, Westervelt P, Ding L, Conrad DF, White BS, Shao J, Link DC, DiPersio JF, Mardis ER, Wilson RK, Ley TJ, Walter MJ, and Graubert TA. Clonal Architecture of Secondary Acute Myeloid Leukemia Defined by Single-Cell Sequencing. *PLOS Genetics*, 10(7), e1004462. PMID: 25010716. https://doi.org/10.1371/journal.pgen.1004462.

2012 Ramos E, Levinson BT, Chasnoff S, **Hughes AEO**, Young AL, Thornton K, Li A, Vallania FL, Province M, and Druley TE. Population-Based Rare Variant Detection Via Pooled Exome or Custom Hybridization Capture with or without Individual Indexing. *BMC Genomics* 13, 683. PMID: 23216810. https://doi.org/10.1186/1471-2164-13-683.

Presentations

2021 **Hughes, AEO**, Farnsworth, CW, and Gronowski, AM. Automated Interpretation of Serum Protein Electrophoresis. Poster presentation, *American Association for Clinical Chemistry*, September 26–30, Atlanta, GA.

2021 **Hughes, AEO**, Montgommery, MC, Liu, C, and Weimer, ET. Allele-Specific Quantification of HLA Transcript Isoforms by Nanopore Long-Read Sequencing. Poster presentation, *American Society for Histocompatibility and Immunogenetics*, September 27–October 1, Orlando, FL.

2021 **Hughes, AEO**, Farnsworth, CW, and Gronowski, A. Automated Interpretation of Serum Protein Electrophoresis. Poster presentation (virtual), *Association for Pathology Informatics Summit*, May 18–21, Pittsburgh, PA.

2020 **Hughes, AEO**, Montgommery, MC, Liu, C, and Weimer, ET. Allele-Specific HLA Expression with Nanopore Long-Read Sequencing. Poster presentation (virtual), *American Society of Hematology Annual Meeting*, December 5–8, San Diego, CA.

2020 **Hughes, AEO**, Webber, DM, Wallace, MA, Johnson C, Burnham, CAD, and Anderson NW. Combining the BioFire Pneumonia Panel with Culture Optimizes Lower Respiratory Pathogen Testing. Poster presentation (virtual), *American Society for Microbiology Microbe*, July 17–18, Chicago, IL.

2020 **Hughes, AEO**, Webber, DM, Wallace, MA, Johnson C, Burnham, CAD, and Anderson NW. Combining the BioFire Pneumonia Panel with Culture Optimizes Lower Respiratory Pathogen Testing. Poster presentation (virtual), *The Academy of Clinical Laboratory Physicians and Scientists Annual Meeting*, June 25, Iowa City, IA.

2016 **Hughes, AEO**, Enright, JM, Myers, CA, and Corbo, JC. Nrl mediates widespread changes in the epigenomic landscape of mouse photoreceptors. Poster presentation, *The Biology of Genomes*, May 10–14, Cold Spring Harbor, NY.

Peer Review Activities

Referee: *Clinical Chemistry*, *The Journal of Applied Laboratory Medicine*, and *Journal of Clinical Microbiology*.