# **Nathan Samuel Abell**

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ABOUT ME

I am a Ph.D. Candidate in the Department of Genetics at Stanford University School of Medicine, and a member of the Montgomery Lab in the Departments of Pathology and Genetics. I also spend lots of time in the Statistics and Biomedical Data Science departments. I have experience and formal training in experimental molecular biology, bioinformatics/computational biology, and computational/applied statistics. In a past life, I also studied political theory/philosophy and researched comparative constitutional design. My current work involves generating and analyzing massively parallel reporter assays to interpret and validate genetic association studies, particularly eQTL and rare variant regulatory effects. I am also leading the mmPCR-seq component of the eGTEx Consortium which contains high-resolution allele-specific expression measurements for thousands of GTEx samples as well as an ovarian cancer panel.

RESEARCH INTERESTS

Computational and statistical genomics, functional and experimental human genomics, gene regulation, high-throughput genetic measurement, sequencing bioinformatics, applied machine learning in molecular biology, epigenomics and epitranscriptomics, small RNA biology, regularization and dimensionality reduction

**EDUCATION** 

# Stanford University School of Medicine, Stanford, California USA

Ph.D., Genetics, 2021 (in progress)

- Advisor: Stephen Montgomery, Departments of Pathology, Genetics, and Computer Science
- Committee: Michael Bassik, Arend Sidow, Zihuai He

Stanford University, Stanford, California USA

M.S., Statistics, 2018

### University of Texas at Austin, Austin, Texas USA

B.S., Cell and Molecular Biology, 2013

• Advisor: Vishwanath Iyer, Departments of Molecular Biosciences and Oncology

B.A., Political Science, Philosophy, and Plan II Honors, 2013

RESEARCH EXPERIENCE

### Stanford University, Stanford, California USA

Graduate Researcher, Montgomery Lab

January 2016 - present

- Experimental and statistical characterization of human genetic regulation of gene expression with massively parallel reporter assays; focus areas include variation associated with molecular phenotypes like gene expression and organismal phenotypes like cancer
- Data production, bioinformatics, and statistical analysis of the allele-specific expression (ASE) by mmPCR-Seq as a component of the enhancing GTEx (eGTEx) consortium; applications to ASE quantification, inference and interpretation in ovarian cancer
- Other active collaboratory projects include gene expression and genetic association studies in African
  population cohorts and high-throughput comparative quantification of the mutational spectra of various
  CRISPR base editors
- Completed work in the lab includes regulatory analyses of the human retinal pigment epithelium, comparative study of gene expression and splicing changes due to age in a human cohort, and identification of jointly exported small RNAs in human exosomes along with significant contributions to the GTEx v6 and v8 releases

#### Rotation Student, Teruel Lab

#### September 2015 - December 2015

- · Generated/analyzed RNA-Seq and ChIP-Seq data from a differentiating murine adipocyte time-course
- Aggregated and compared large amounts of published sequencing data across a number of -omic dimensions relevant to adipose differentiation to guide experimental design

# University of Texas at Austin, Austin, Texas USA

Bioinformatician, Xhemalce Lab

### September 2014 - Present

- Bioinformatic and statistical analysis of high-dimensional genomic data types, including many flavors
  of non-standard sequencing, mass spectrometry, metabolomics, and some image analysis
- Integration of high-dimensional data to link the genomic effects of perturbations in epitranscriptomic reader and writer enzymes such as BCDIN3D with proliferative and invasive cellular phenotypes
- Maintenance of computational infrastructure on Linux servers at the Texas Advanced Computing Center (TACC), a national research computing cluster
- Use of chemical biological and image analysis tools to probe the interactions of enoxacin, a miRNA
  pathway modulator and potential anti-cancer therapeutic, in human cells; identified its mechanistic
  target as PIWIL3

#### Research Technician, Iyer Lab

# January 2011 - September 2014

- Analysis of data from RNA-seq, RIP-seq, (PAR)CLIP-seq, and ChIP-seq on the TACC cluster supercomputing infrastructure
- Led multiple computational projects involving the extensive use/modification of existing bioinformatics tools and the creation of several novel tools
- Contributed analysis to projects including primary glioblastoma chromatin conformation, primary tumor exome-seq, ENCODE miRNA-seq meta-analysis, and ChIP-seq bias correction algorithm development
- Extensive use of applied machine learning, including statistical and mathematical foundations and practical implementation on clusters

## Research Assistant, CCP

# **January 2009 - January 2011**

 Quantitatively coded dozens of constitutional documents to identify geographic and temporal trends for the Comparative Constitutions Project

#### **PUBLICATIONS**

IN REVIEW: McFadden MJ, McIntyre ABR\*, Gokhale NS\*, **Abell NS**\*, Ipas H, Xhemalce B, Mason CE, Horner SM. Post-transcriptional regulation of antiviral gene expression by N6-methyladenosine. 2020.

IN REVIEW: Spector LP, Tiffany M, Ferraro NM, **Abell NS**, Montgomery SB, Kay MA. Evaluating the genomic parameters for optimizing rAAV-mediated homologous recombination. 2020.

IN REVIEW: Reinsborough CW\*, Ipas H\*, **Abell NS**\*, Gouws EB, Williams JP, Mercado M, Van Den Berg C, Xhemalce B. BCDIN3D RNA methyltransferase stimulates Aldolase C expression and glycolysis through let-7 microRNA in breast cancer cells. 2020.

Gay NR, Gloudemans M, Antonio ML, Abell NS, Balliu B, Park Y, Martin AR, Musharoff S, Rao A, Aguet F, Barbeira B, Bonazzola R, Hormozdiari F, GTEx Consortium, Ardlie K, Brown CD, Im HK, Lappalainen T, Wen X, Montgomery SB. Impact of admixture and ancestry on eQTL analysis and GWAS colocalization in GTEx. Genome Biology. 2020.

Ferraro NM\*, Strober BJ\*, Einson J, **Abell NS**, Aguet F, Berveira AN, Bucan M, Castel S, Davis JR, Greenwald E, Hess GT, Hilliard AT, Kember RL, Kotis B, Park Y, Peloso G, Ramdas S, Scott AJ, Smail C, Tsang EK, Zekacat S, Ziosi M, Aradhana, TOPMed Lipids Working Group, Ardlie KG, Assimes TL, Bassik MC, Brown CD, Correa A, Hall I, Im HK, Li X, Natarajan P, GTEx Consortium, Lappalainen T, Mohammadi P,

Montgomery SB, Battle A. Transcriptomic signatures across human tissues identify functional rare genetic variation. Science. 2020;369:eaaz5900.

Balliu B, Durrant M, de Goede O, **Abell NS**, Li X, Liu B, Gloudemans MJ, Cook NL, Smith KS, Pala M, Cucca F, Schlessinger D, Jaiswal S, Sabatti C, Lind L, Ingelsson E, Montgomery SB. Genetic dysregulation of gene expression and splicing during a ten-year period of human aging. Genome Biology. 2019;20(1):230.

Reinsborough CW\*, Ipas H\*, **Abell NS**\*, Nottingham RM\*, Yao J, Devanathan SK, Shelton SB, Lambowitz AM, Xhemalce B. BCDIN3D regulates tRNA-His 3 fragment processing. PLOS Genetics. 2019;15(7):e1008273.

Liu B\*, Calton MA\*, **Abell NS**, Benchorin G, Gloudemans MJ, Chen M, Hu J, Li X, Balliu B, Montgomery SB, Vollrath D. Ocular disease mechanisms elucidated by genetics of human fetal retinal pigment epithelium gene expression. Communications Biology. 2019;2:186.

Shelton SB, Shah NM\*, **Abell NS**\*, Devanathan SK, Mercado M, Xhemale B. Crosstalk between the RNA Methylation and Histone-Binding Activities of MePCE Regulates P-TEFb Activation on Chromatin. Cell Rep. 2018;22(6):1374-1383.

**GTEx Consortium**. Genetic effects on gene expression across human tissues. Nature. 2017;550(7675):204-213. [As a member of the Consortium]

Zacharioudakis E, Agarwal P, Bartoli A, **Abell NS**, Kunalingam L, Bergoglio V, Xhemalce B, Miller KM, Rodriguez R. Chromatin Regulates Genome Targeting with Cisplatin. Angew Chem Int Ed Engl. 2017;56(23):6483-6487.

**Abell NS**\*, Mercado M\*, Caeque T, Rodriguez R, Xhemalce B. Click Quantitative Mass Spectrometry Identifies PIWIL3 as a Mechanistic Target of RNA Interference Activator Enoxacin in Cancer Cells. J Am Chem Soc. 2017;139(4):1400-1403.

Tsang EK, **Abell NS**, Li X, Anaya V, Karczewski KJ, Knowles DA, Sierra RG, Smith KS, Montgomery SB. Small RNA Sequencing in Cells and Exosomes Identifies eQTLs and 14q32 as a Region of Active Export. G3 (Bethesda). 2017;7(1):31-39.

Polioudakis D, **Abell NS**, Iyer VR. MiR-191 Regulates Primary Human Fibroblast Proliferation and Directly Targets Multiple Oncogenes. PLoS ONE. 2015;10(5):e0126535.

Polioudakis D, **Abell NS**, Iyer VR. miR-503 represses human cell proliferation and directly targets the oncogene DDHD2 by non-canonical target pairing. BMC Genomics. 2015;16:40.

Polioudakis D, Bhinge AA, Killion PJ, Lee BK, **Abell NS**, Iyer VR. A Myc-microRNA network promotes exit from quiescence by suppressing the interferon response and cell-cycle arrest genes. Nucleic Acids Res. 2013;41(4):2239-54.

TEACHING EXPERIENCE Teaching Assistant, BIOS 201: Next Generation Sequencing

March 2018, 2019, 2020

- Acted as teaching assistant for Stanford's short course on sequencing analysis, covering common bioinformatic procedures like variant calling, gene expression analysis, peak calling, and more
- Assisted with supplementary tutorials introducing Linux, R/RStudio, and the command line

Course Instructor, Big Data in Biology Summer School

July 2014, 2015, 2016

- Co-instructor for Core NGS Analysis Tools at TACC, run by the Center for Computational Biology and Bioinformatics at UT Austin; course included graduate students and post-docs
- Guest lectured for the Introduction to RNA-seq course, titled RIP-Seq Analysis and surveyed an array
  of RNA interaction by sequencing analysis methods like CLIP-seq and PAR-CLIP

Course Instructor, Summer Statistics Institute

July 2013

- Co-instructed the Introduction to NGS Bioinformatics run by the Statistics department; course included graduate students and post-docs
- Methods covered included, quality assessment, alignment, feature quantitation, Linux pipeline execution, variant calling, differential expression, simple statistical testing, and substantially more

# Undergraduate Teaching Assistant

# August 2010 - December 2010

 Acted as teaching assistant for an introductory freshman course on Western feminism with Dr. Lisa Moore, responsible for discussion sections, individualized student tutoring, and assignment grading

# HONORS AND AWARDS

Joint Institute for Metrology in Biology Training Grant, September 2017-present

Center for Systems Biology Seed Grant (\$25000, with Teruel Lab), July 2016

Hertz Foundation Fellowship Semi-Finalist, Nov. 2014

Best Poster Award, Big Data in Biology Symposium, UT Austin, May 2014

Deans Honored Graduate (1% of the graduating class based on research accomplishments), May 2013

Graduation with High Honors (General), May 2013 Graduation with Special Honors (Biology), May 2013

Phi Beta Kappa Membership, May 2013

Deans List (Every semester within the indicated time frame), Sep. 2009 - May 2013

Plan II Honors Research Grant, Sep. 2012

Best Paper Award, 18th Annual Political Science Conference, Illinois State University, Oct. 2010

National Merit Corporate Scholarship, May 2007

#### **PRESENTATIONS**

Poster Presentation: Fine-mapping causal regulatory variants using massively parallel reporter assays JIMB/NIST Annual Meeting, Gaithersburg MD March 2019

Platform Talk: Fine-mapping causal regulatory variants using massively parallel reporter assays Annual Meeting of the American Society for Human Genetics, San Diego CA October 2018

Poster Presentation: Click-qMS Identifies PIWIL3 as a Target of RNA Interference Activator Enoxacin in Cancer Cells

Cell Symposia: Technology. Biology. Data Science, Berkeley CA October 2016

Technical Talk: The MirUtils Suite and its Use in miRNA Quantification

Byte Club, Center for Computational Biology and Bioinformatics, UT Austin November 2014

Poster Presentation: miRNA Profiling of ENCODE Small RNA-seq Data

1st Annual Big Data in Biology Conference, UT Austin June 2014

Thesis Presentation: Transcriptome-Wide RNA Binding Profiles of Argonaute Proteins in Cultured **Human Cell Lines** 

Honors Thesis Symposium, UT Austin May 2013

**OTHER PROFESSIONAL** EXPERIENCE

F&B LLC, Paralegal Assistant, Austin TX USA August 2010 - May 2011 **PBFMC LLC**, Administrative Assistant, Austin TX USA June 2010 - August 2011 Equality Texas, Public Policy/Legislative Intern, Austin TX USA January 2010 - August 2011 Multiple High Schools, Debate Coach, Austin TX USA June 2007 - May 2010 Hammerle Finley LLC, Administrative Assistant, Denton TX USA June 2008 - August 2008

COMPUTING SKILLS • Analysis Packages: Bioconductor, tidyverse, ggplot2, kernlab/e1071/caret, parallel, markdown/knitr/shiny, lme4, glmnet, gptk, htmlwidgets, devtools (many more)

- Languages: R, Python, Bash, Perl, Matlab, Javascript
- Applications: LATEX, Microsoft Office, common bioinformatics tools, high-performance/cluster computing, assorted database, spreadsheet, and presentation software
- Operating Systems: Unix/Linux, OS X, Windows (from most to least experience)

# REFERENCES

Dr. Stephen Montgomery - sbmontgom@stanford.edu Dr. Blerta Xhemalce - b.xhemalce@utexas.edu Dr. Vishwanath Iyer - vishy@utexas.edu

Additional references available upon request