# **Ruth Johnson**

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### **Education**

University of California, Los Angeles

' PhD Computer Science

Los Angeles, CA Sept 2017-present

University of California, Los Angeles

Los Angeles, CA

B.S. Mathematics, Minor in Bioinformatics

Sept 2013-June 2017

Academic Honors.....

- o RECOMB Travel Fellowship (2020)
- o NRT-Modeling and Understanding Human Behavior Fellowship (2018)
- o Ford Fellowship Predoctoral Competition Honorable Mention (2018)
- o NSF Graduate Research Fellowships Program Honorable Mention (2017)
- o Eugene V. Cota-Robles Fellowship, UCLA (2017)

# Research Experiences

#### Machine Learning in Biomedical Lab at UCLA

Los Angeles, CA

Graduate Student Researcher

July 2017 - current

- Leveraging electronic health records to predict common variable immune deficiency and other rare disorders through phenotype risk scores
- Developed Bayesian statistical framework to model genetic architecture of complex traits
- Emphasis on probabilistic modeling, Markov Chain Monte Carlo, variational inference, graphical models
- Under supervision of Prof. Sriram Sankararaman and Prof. Bogdan Pasaniuc

Illumina Foster City, CA

Deep Learning Engineering Intern

June 2018-Sept 2018

- Developed end-to-end parallelizable training data generation pipeline for deep learning base calling models
- Utilized Keras, convolutional neural networks, Python
- Under supervision of Amirali Kia

## **Bioinformatics Lab at UCLA**

Los Angeles, CA

Undergraduate Student Researcher

March 2016 - June 2017

- Constructed software pipeline for fine-mapping analyses with the integration of functional annotation data
- Created Python based data visualization tool that produces publication-ready figures of integrated fine-mapping experiments
- Under supervision of Prof. Bogdan Pasaniuc

# **Sandia National Laboratory**

Albuquerque, NM

R&D Engineering Intern

June 2016 - August 2016

- Researched satellite anomaly detection methods through supervised classification algorithms such as DBSCAN
- Designed components of an object-oriented, dynamic web UI for satellite control systems using ReactJS and ExtJS

### **Journal Publications**

- \* denotes joint authors
- 1. Localizing components of shared transethnic genetic architecture of complex traits from GWAS summary data.
  - Huwenbo Shi\*, Kathryn S Burch\*, <u>Ruth Johnson</u>, Malika K Freund, Gleb Kichaev, Nicholas Mancuso, Astrid M Manuel, Natalie Dong, Bogdan Pasaniuc; *American Journal of Human Genetics* 2020.
- 2. An automated machine learning-based model predicts postoperative mortality using readilyextractable preoperative electronic health record data
  - Brian Hill, Robert Brown, Eilon Gabel, Christine Lee, Maxime Cannesson, Loes Olde Loohuis, <u>Ruth Johnson</u>, Brandon Jew, Uri Maoz, Aman Mahajan, Sriram Sankararaman, Ira Hofer, Eran Halperin; *British Journal of Anaesthesia 2019*.
- 3. **Probabilistic fine-mapping of transcriptome-wide association studies**Nicholas Mancuso, Malika K. Freund, <u>Ruth Johnson</u>, Huwenbo Shi, Gleb Kichaev, Alexander Gusev, and Bogdan Pasaniuc; *Nature Genetics 2019*.
- 4. A unifying framework for joint trait analysis under a non-infinitesimal model Ruth Johnson, Huwenbo Shi, Bogdan Pasaniuc\*, Sriram Sankararaman\*; *Bioinformatics 2019*.
- 5. **Improved methods for multi-trait fine mapping of pleiotropic risk loci** Gleb Kichaev\*, Megan Roytman\*, <u>Ruth Johnson</u>, Eleazar Eskin, Sara Lindström, Peter Kraft, Bogdan Pasaniuc; *Bioinformatics 2017*.

#### **Conference Publications**

- A scalable method for estimating the regional polygenicity of complex traits
   Ruth Johnson, Kathryn S. Burch, Kangcheng Hou, Mario Paciuc, Bogdan Pasaniuc, Sriram Sankararaman; RECOMB 2020.
- 2. A unifying framework for joint trait analysis under a non-infinitesimal model Ruth Johnson, Huwenbo Shi, Bogdan Pasaniuc\*, Sriram Sankararaman\*; ISMB 2018.

### **Preprints**

- 1. Estimation of regional polygenicity from GWAS provides insights into the genetic architecture of complex traits
  - Ruth Johnson, Kathryn S. Burch, Kangcheng Hou, Mario Paciuc, Bogdan Pasaniuc, Sriram Sankararaman; under review at *American Journal of Human Genetics*.
- Prior diagnoses and medications as risk factors for COVID-19 in a Los Angeles Health System
  Timothy S Chang, Yi Ding, Malika K Freund, <u>Ruth Johnson</u>, Tommer Schwarz, Julie M Yabu, Chad
  Hazlett, Jeffrey N Chiang, Ami Wulf, Daniel H Geschwind, Manish J Butte, Bogdan Pasaniuc; <u>medRxiv</u>
  2020.

### **Oral Presentations**

- 1. A scalable method for estimating the regional polygenicity of complex traits RECOMB, July 2020. Virtual meeting.
- 2. Leveraging electronic health record signatures identify undiagnosed patients with Common Variable Immunodeficiency Disease
  - Undiagnosed Diseases Network Steering Committee Meeting, March 2020. Los Angeles, CA, USA. (cancelled due to COVID-19)

3. Leveraging electronic health record signatures identify undiagnosed patients with Common Variable Immunodeficiency Disease

Institute for Quantitative and Computational Biosciences - Research Seminar, February 2020. Los Angeles, CA, USA.

4. Electronic health record signatures identify undiagnosed patients with of CVID

Medical and Population Genetics seminar - Computational Genomics and Health, November 2019. Los Angeles, CA, USA.

5. Dissecting the genetic architecture of complex traits through local polygenicity using summary statistics from genome-wide association studies

Biology of Genomes 2019, May 2019. Long Island, NY, USA.

6. Dissecting the genetic architecture of complex traits through local polygenicity using summary statistics from genome-wide association studies

Medical and Population Genetics, May 2019. Los Angeles, CA, USA.

7. A scalable Bayesian model for estimating the genetic architecture of complex traits using summary statistics from GWAS

Probabalistic Modeling in Genomics Meeting 2018, November 2018. Long Island, NY, USA.

- 8. A unifying framework for joint trait analysis under a non-infinitesimal model ISMB 2018, July 2018. Chicago, IL, USA.
- 9. Combining genetic correlation and colocalization into a unifying model Medical and Population Genetics, May 2019. Los Angeles, CA, USA.
- CANVIS: Correlation Annotation VISualization RECOMB Genetics Satellite Meeting, July 2017. Los Angeles, CA, USA.

### **Poster Presentations**

1. Fine-scale estimates of regional polygenicity provide insights into the genetic architecture of complex traits

Ruth Johnson, Kathryn S. Burch, Kangcheng Hou, Bogdan Pasaniuc, Sriram Sankararaman; Annual meeting of the American Society of Human Genetics, Oct 2019. Houston, TX, USA.

- 2. An analysis of the genetic overlap of 20 complex traits under a non-infinitesimal model Ruth Johnson, Huwenbo Shi, Kathryn Burch, Bogdan Pasaniuc, Sriram Sankararaman; Annual meeting of the American Society of Human Genetics, Oct 2018. San Diego, CA, USA.
- 3. Integrative fine-mapping of 34 complex phenotypes
  Ruth Johnson, Gleb Kichaev, Kathryn Burch, Bogdan Pasaniuc; Annual meeting of the American Society
  of Human Genetics, Oct 2017. Orlando, FL, USA.
- 4. Leveraging functional annotations in fine-mapping of causal variants for complex traits
  Ruth Johnson, Gleb Kichaev, Kathryn Burch, Bogdan Pasaniuc; UCLA Undergraduate Research Poster
  Day, May 2017. Los Angeles, CA, USA.

\*Dean's Prize Science Award Honoring Outstanding Undergraduate Researcher

5. Visualizing correlated causal variants

Ruth Johnson, Gleb Kichaev, Bogdan Pasaniuc; Annual meeting of the American Society of Human Genetics, October 2016. Vancouver, CN.

# **Mentoring Experience**

- o Jessie Chen. Undergraduate student, Bruins in Genomics Summer Program.
  - Project title: "ATLAS-hub: an R Shiny App for PheWAS results on the ATLAS BioBank"
  - Research Excellence award and Top Presentation award
- o Mario Paciuc. Undergraduate student, Rice University.

Project title: "Genetic correlation of complex traits under a non-infinitesimal model"

- co-author on Johnson et al. RECOMB 2020
- award for Distinction in Research and Creative Works from the Department of Statistics
- o Gary Hu. Undergraduate student, Bruins in Genomics Summer Program.

Project title: "Trans-ethnic genetic overlap in complex traits."

- o Hugo Mainguy. Undergraduate student, Bruins in Genomics Summer Program.
  - Project title: "Assessing the overlap of complex traits through the shared proportion of causal SNPs and genetic correlation"
- o Engineering Undergraduate Research Program Graduate Student Mentor.
  - Taught weekly workshops about scientific presentations and guided 10-15 students through creating abstracts, posters, and presentations about their research projects.

# **Software**

o **BEAVR** Estimating regional polygenicity

Software that estimates the proportion of causal variants (*i.e.* polygenicity) within a given region from GWAS summary statistics and in-sample LD.

https://github.com/bogdanlab/BEAVR

o UNITY Quantifying genetic overlap of complex traits

Software that uses a fully Bayesian framework to calculate the proportion of shared causal variants between two complex traits through GWAS summary statistics. The method also explicitly models the genetic correlation present between both traits.

https://github.com/bogdanlab/UNITY

o CANVIS Fine-mapping visualization

A fine-mapping tool that visually summarizes an integrative fine-mapping experiment. The tool provides visual representation of the local correlation structure (LD), the functional annotations used, as well as association statistics and posterior probabilities for each SNP.

https://github.com/bogdanlab/PAINTOR/tree/master/CANVIS