William P. Bone
wpbone06@gmail.com | 304-904-0147 | github.com/wpbone06

SUMMARY

- Statistical geneticist and bioinformatician with 10 years of research in the genetics of human diseases
- Formal training in genetics, statistics, biology, and biochemistry
- 19 peer-reviewed manuscripts and 2 manuscripts in preparation/review

EDUCATION

University of Pennsylvania, Philadelphia, PA 2017 – Present
PhD candidate in Genomics and Computational Biology
Pursing a Dual Master's Degree in Statistics

St. Mary's College of Maryland, St. Mary's City, MD 2012
Double Major: Biology and Biochemistry

RECENT RESEARCH

University of Pennsylvania, Philadelphia, PA 2017 – Present
Mentored by Dr. Marylyn Ritchie and Dr. Benjamin Voight

Investigating pleiotropy between cardiometabolic traits and complex diseases

- **Goal:** Detect pleiotropic loci associated between cardiometabolic traits and complex diseases to identify potential novel therapeutic targets.
- Using multi-trait GWAS and colocalization analyses, we detected potential therapeutic targets including *DOCK4* for Alzheimer's disease and *PCSK6* for atherosclerosis as well as several other pleiotropic loci.

Developed a pipeline to identify candidate causal genes for GWAS data using eQTL and sQTL data

- **Goal:** Develop a pipeline, ColocQuiaL, to rapidly perform and visualize the results from thousands of colocalization analyses between GWAS data and eQTL (RNA expression quantitative trait loci) and sQTL (RNA splicing quantitative trait loci) data.
- ColocQuiaL can perform colocalization analyses between loci from any GWAS summary statistics file and GTEx v8 single-tissue sQTL and eQTL data. I used different iterations of this pipeline to identify candidate causal genes for GWAS signals. Collaborations across disease areas led to four published manuscripts.

Multi-trait rare variant gene burden tests for cardiometabolic traits

- **Goal:** Perform multi-trait rare variant gene burden tests for cardiometabolic traits to identify novel associated genes with potential to be therapeutic targets.
- Made a pipeline to perform multi-trait rare variant gene burden tests across the genome for biobank electronic health record data and applied it to Penn Medicine Biobank blood lipid levels and liver enzyme levels where we identified novel associated gene *ANKRD36C*.

WORK EXPERIENCE

Recursion Pharmaceuticals, Salt Lake City, UT 2022
Data Science Team

- Worked on computational methods to evaluate CRISPR gene knockout phenotypes.

Agilent Technologies, Lexington, MA 2015-2017
Cartagenia Bench Products

- Worked on the research team for the Cartagenia suite of genetic analysis tools for patient NGS data.
- Developed proof of principle bioinformatics tools and pipelines to test new feature functionality.
- Bioinformatics liaison to customers for feature requests and resolution of informatics hurdles.

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National Institutes of Health, Bethesda, MD 2012 – 2015
Undiagnosed Diseases Program (UDP)

- Collaborated with the Monarch Initiative in the development of Exomiser, a NGS variant prioritizing software suite that compares patient phenotype to known genetic disorders and model organism data.
- Developed a computational pipeline to identify and prioritize potentially causal genetic variants, which led to the discovery of 95 candidate disease variants, and 41 diagnoses.
- Investigated the effects of mutations in the enzyme spermine synthase in human bone marrow stromal cells to study the mechanism of disease for Snyder-Robinson Syndrome.

SELECT PUBLICATIONS (4 OF 21)

- BY Chen*****, **WP Bone*** et al. *ColocQuiaL: A QTL-GWAS colocalization pipeline.* **Bioinformatics**. 2022 Jul. 27. doi:10.1093/bioinformatics/btac512
- TR Bellomo*****, **WP Bone*** et al. *Multi-trait GWAS of atherosclerosis detects novel pleiotropic loci.* **Frontiers in Genetics**. 2022 Feb 02. doi:10.3389/fgene.2021.787545
- **WP Bone** et al. *Multi-trait association studies discover pleiotropic loci between Alzheimer's disease and cardiometabolic traits.* **Alzheimer's Research & Therapy**. 2021 Feb 04. doi:10.1186/s13195-021-00773-z
- **WP Bone** et al. *Computational evaluation of exome sequence data using human and model organism phenotypes improves diagnostic efficiency*. **Genetics in Medicine**. 2015 Nov 12. doi:10.1038/gim.2015.137

AWARDS AND RECOGNITION

CHARGE Meritorious Abstract Award	2021
Blavatnik Family Fellowship Graduate Group Semi-finalist	2021
American Society of Human Genetics Abstract Reviewer's Choice Award	2020
American Heart Association Predoctoral Fellowship Award	2019
NHGRI G.R.E.A.T. Award	2014

CONFERENCES (4 OF 10)

CHARGE Meeting, Virtual Meeting Oct. 7-8, 2021
Presentation: *Multi-trait GWAS of atherosclerosis detects novel loci and potential therapeutic targets*

Vascular Discovery Conference, Virtual Meeting Sep. 22-24, 2021
Moderated Poster Presentation: *Multi-trait GWAS of Atherosclerosis Detects Novel Loci and Potential Therapeutic Targets*

American Society of Human Genetics, Virtual Meeting Oct. 27-30, 2020
Poster Presentation: *Multi-trait GWAS of atherosclerosis and correlated traits detects novel pleiotropic loci*

AMIA Translational Bioinformatics Summit, San Francisco, CA Mar. 23-25, 2015
Peer Learning Workshop: Semantic Phenotyping for Translational Medicine

PRACTICAL SKILLS

- **Practical knowledge of high-performance cluster and cloud-based computing**
- **Coding Languages:** Proficiency in Python, R, and Bash
- **Genomic Software:** Experience with DESeq2, PLINK, GCTA, GATK, bedtools, samtools, and bcftools
- **Genomic Data:** Worked with DNA-seq (exomes, genomes, cfDNA), RNA-seq, and SNP array data

PERSONAL

Co-Founder of Fantasy Soccer Statistics Blog (overthinkingfootball.com), History Podcasts, Board Member of Philadelphia Open Soccer