Barry Moore

3417 S. Crestwood Dr. Millcreek, UT 84109 barry.utah@gmail.com | (801) 243-8819 | LinkedIn: https://www.linkedin.com/in/barrymoore66 https://barrymoore.bio/

PROFESSIONAL SUMMARY

Senior Scientist, Precision Genomics at Fabric Genomics, with over two decades of experience in genome informatics, clinical genomics, and rare disease diagnostics. Longtime co-developer of foundational tools including VAAST, VVP, Phevor, and the GEM AI platform, with deep expertise in building scalable NGS analysis workflows and AI-enabled interpretation systems. Fluent across genomics, software engineering, and clinical application, with a track record of translating complex genomic data into actionable insights for patients and healthcare providers. Known for cross-disciplinary collaboration, technical leadership, and delivering innovative solutions that bridge research and clinical practice.

CORE COMPETENCIES

- **Clinical Genomics & Rare Disease Diagnostics** – End-to-end expertise in genome interpretation, variant prioritization, and integration of WGS/RNASeq into clinical workflows.
- **Bioinformatics Software & Pipeline Development** – Design, implementation, and optimization of scalable NGS analysis pipelines with Nextflow, Snakemake, and custom frameworks; extensive experience with GATK, Sentieon, Dragen, and GEM.
- **AI-Enabled Genome Interpretation** – Co-developer of the GEM AI platform; deep experience building and curating training datasets, algorithm refinement, and integration with clinical decision support tools.
- **Cross-Disciplinary Collaboration & Leadership** – Proven ability to bridge genomics, software engineering, and clinical application; mentor to clinicians, analysts, postdocs, and engineers; recognized for delivering complex projects across research and industry teams.
- **Data Science & Systems Integration** – Deep domain expertise in genomic reference datasets, with advanced skills in EHR-based NLP and phenotype-driven analysis. Extensive experience integrating this data into genomic and clinical diagnostic workflows, supported by a strong background in Linux, HPC, and cloud computing.

- **Scientific Communication & Training** – Experienced instructor at Cold Spring Harbor Laboratory and other international courses; frequent collaborator on high-impact publications and grants.

EXPERIENCE

Senior Scientist, Precision Genomics *Fabric Genomics | 2025 – Present*

- Lead development of clinical-grade genomics workflows and AI-enabled variant interpretation systems, with a focus on rare disease diagnostics and scalable, cost-efficient analysis pipelines.
- Build on over 15 years of prior collaboration with Fabric to refine and expand the GEM AI platform, VAAST/VVP variant prioritization stack, and phenotype-driven analysis tools.
- Partner with cross-functional teams, including GeneDx collaborators, to integrate advanced analysis capabilities into production pipelines for clinical reporting.
- Mentor analysts, bioinformaticians, and software engineers in best practices for genomic data analysis, pipeline optimization, and reproducibility.
- Drive innovation at the intersection of software engineering, bioinformatics, and clinical genomics to deliver actionable insights that directly support patient care.

Senior Consultant *Fabric Genomics (formerly Omicia) | 2009 – 2025*

- Long-term contributor to the GEM AI platform and full gene/variant prioritization stack.
- Built and curated all major training datasets for VAAST/VVP/Phevor/GEM.
- Supported collaborations with Genomics England, Rady Children's, GeneDx, ONT and other Fabric partners.
- Built NGS secondary (variant calling) frameworks for Fabric and key customers based on GATK, Sentieon and Dragen.

Director of Research and Science *Utah Center for Genetic Discovery, University of Utah | 2014 – 2025*

- Leadership role in the analytical arm of NeoSeq and GeneKids projects focused on rapid whole genome interpretation for critically ill children.
- Developed clinical interpretation pipelines and SOPs for research-clinical integration of WGS and RNASeq data.
- Created tools like PhenVen for HPO-driven phenotype matching supporting of WGS/RNASeq diagnostics.
- Led RNASeq-based reanalysis efforts to improve diagnostic yield.
- Mentored clinicians, postdocs, graduate students, and analysts across genomics teams.

Research Scientist *Department of Human Genetics, University of Utah | 2006 – 2014*

- Designed and implemented scalable & repeatable WGS pipelines for disease gene discovery.
- On the development team multiple tools (VAAST, VVP, Phevor) adopted in genomic analysis and deployed them in analysis of dozens of large genomic datasets - large and small.
- Led bioinformatics support across collaborations with ARUP, Regeneron, Clinithink, Childhood Liver Disease Research Network and more.

Senior Research Specialist *Department of Human Genetics, University of Utah | 1994 – 2006*

- Self-directed transition from molecular biology to genome informatics during the early genomic era.
- Managed lab operations, trained junior researchers, and built early genome analysis tools.
- Key member of multiple large-scale genomic research initiatives.
- Instructor in numerous CSHL bioinformatics and NGS sequencing courses.

EDUCATION & TRAINING

- M.S., Biology (Molecular Biology & Systematics), Loma Linda University, 1993
- B.S., Chemistry, Southwestern Adventist University, 1990
- Genome Informatics, Cold Spring Harbor Laboratory, 2003
- Summer Institute in Statistical Genetics, University of Washington, 2010

SELECTED PUBLICATIONS

- Reese et al., *Genome Biology*, 2010: Standard Variation File Format
- Yandell et al., *PLoS Comp Biol*, 2008: Disease Alleles in Paralogous Proteins
- Eilbeck et al., *BMC Bioinformatics*, 2009: Quantitative Measures for Genome Annotation
- Moore et al., *Genet Med*, 2011: Variation in 10 Healthy Genomes and Diagnostic Implications
- Hu et al., *Genet Epidemiol*, 2013: VAAST 2.0 Variant Classification
- Flygare et al., *BMC Bioinformatics*, 2018: VAAST Variant Prioritizer (VVP)
- De La Vega et al., *Genome Med*, 2021: AI-Enabled Genome Interpretation
- Moore et al., *Genet Med Open*, 2023: RNASeq Resolves NEB Variant in Nemaline Myopathy
- Jenkins et al., *NPJ Genom Med*, 2025: Utah NeoSeq Program in NICU Genomic Diagnostics

Full list of 76 publications available on request or Google Scholar - https://bit.ly/3IKbfLV Citation metrics (Google Scholar, July 2025): h-index = 31, i10-index = 41, total citations = 6,522

TOOLS & TECHNOLOGIES

- **Languages:** Perl, Python, JavaScript, SQL
- **Linux Admin & Power Tools:** Bash, screen/tmux, git, emacs, cron/at, find, parallel/xargs, sed/awk/CLI Perl, VisiData, jq, sqlite3, rclone/rsync
- **Workflow Systems:** Nextflow, Snakemake, custom scripting
- **Cloud/Infra:** AWS, Docker, Linux HPC clusters
- **Bioinformatics Tools:** GATK, Sentieon, Dragen, samtools, bcftools, bedtools, htslib, vcfanno, GEM, VAAST, VVP, Phevor
- **Data Analysis/Presentation:** iPython/Jupyter, numpy, Pandas, scipy, matplotlib/Seaborn/Plotly, custom dashboards & worksheets using JavaScript, DataTables, HTML/CSS, XlsxWriter, Markdown

PATENTS

US Patent Application No. US20230326547A1 **Systems and Methods for Prioritizing Genome Variants.** Filed October 2023. ***Co-inventor***. https://patents.google.com/patent/US20230326547A1

References available upon request.