

Human Pangenome Workshop: An Introduction to the Human Pangenome Tools and Workflows

Workshop Description

The release of the first human reference assembly was an important achievement for genome sciences and medicine. However, the use of a single reference genome in bioinformatics can lead to reference bias and miss important information about genome variability and relationships. New assembly methods have made it easier to generate high-quality complete genome assemblies, and using a pangenome graph that expresses many genomes and their mutual alignment can help address these issues ([Liao et al. Nature 2023](#)). This workshop offers an introduction of this new reference resource to genomics researchers and biocurators who want to get a head start on learning about the Human Pangenome, using and interpreting graph-based reference datasets, and gaining familiarity with pangenome analysis tools and workflows.

Presentations are supported by members of the Human Pangenome Project, a GA4GH Driver Project, including the Human Pangenome Reference Consortium (HPRC) (<https://humanpangenome.org/>), which is an NHGRI-funded effort to represent a global genomic diversity. Further, we will provide an overview of the African Pangenome Effort and efforts from the H3ABionet/eLwazi training opportunities and positive outcomes. This workshop is designed to provide an introduction to efforts to construct a pangenome and the emerging tooling ecosystem to map and call variants.

Intended Audience:

The intended audience for this workshop is genomics researchers and professionals who would like to: 1) Learn more about the Human Pangenome Reference; and 2) Find tools and tutorials to support genomic analyses in the future. Participants will benefit from prior knowledge of basic file formats (e.g. FASTA and VCF) and concepts.

Workshop learning objectives

By the end of this workshop, participants will be able to

- Understand why the Human and Africa Pangenome Projects were established and how they address the limitations of single reference genomes.
- Apply computational approaches to analyze pangenomes by building and interpreting graph-based representations of diverse human genome assemblies.

Workshop Schedule of Events

10:35 Introduction to the Human Pangenome Project & Future HPRC Data Release:
Karen Miga

11:00 Africa Pangenome Project & H3ABionet/eLwazi: *Mohammed Farahat*

11:30 Building and Analyzing Pangenome Graphs: *Erik Garrison & Andrea Guarracino*

12:00: End

Practical Course Instructors

Erik Garrison

[<egarris5@uthsc.edu>](mailto:egarris5@uthsc.edu)

*Associate Professor, Department of Genetics, Genomics and Informatics
University of Tennessee Health Science Center*

[GitHub](#)

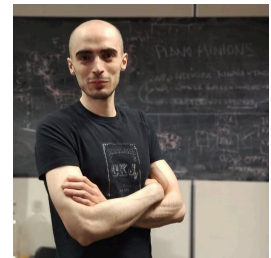


Andrea Guarracino

[<aguarra1@uthsc.edu>](mailto:aguarra1@uthsc.edu)

*Postdoctoral Scholar, Department of Genetics, Genomics and Informatics
University of Tennessee Health Science Center*

[GitHub](#)



Mohammed Farahat

[<mohammed.farahat@uct.ac.za>](mailto:mohammed.farahat@uct.ac.za)

*Postdoctoral Fellow, Computational Biology Division, Department of
Integrative Biomedical Sciences, University of Cape Town.*

[GitHub](#)



Karen Miga

[<khmiga@ucsc.edu>](mailto:khmiga@ucsc.edu)

*Associate Professor, Biomolecular Engineering Department
University of California, Santa Cruz*

Associate Director, UC Santa Cruz Genomics Institute

HPRC Pangenome Resources [GitHub](#)

