

Add-On/Ticketed Event Info

Please see more information in ASHG's Online Planner (coming in July).

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Interactive Workshops

Getting Started with Biomedical and Genomic Data in the All of Us Researcher Workbench

Tuesday, November 5: 10:00 am - 12:00 pm

Pricing: \$40 ASHG member, \$55 nonmember. Advance ticket purchase required to attend.

Includes: Entrance to the workshop & access to the OnDemand recording

Speakers: Julie Coleman, Shamika Ketkar, Jinyoung Byun

The NIH's All of Us data is a significant source of biomedical and genomic data with 245,400 short-read whole genome sequences (WGS), 1,040 long-read WGS, 11,400 short-read WGS with structural variant calls, and 312,940 genotyping arrays. The genomic data are from participants who have also shared phenotypic data, including from electronic health records, drug exposures, procedures, physical measurements, Fitbits, and surveys. New data are released every year with ongoing engagement of one million participants planned. Researchers from institutions with Data Use and Registration Agreements (DURAs) access and analyze the data on the online platform, the All of Us Researcher Workbench.

This workshop will take attendees through interactive exercises to become familiar with the All of Us data and begin research with it.

First, attendees will perform a data quest on the All of Us Research Hub to browse and query the available data and to examine current projects.

Then attendees will complete an exercise to learn the unique tools of the Researcher Workbench and how they are used to access and analyze the data. We will demonstrate a GWAS in interactive Jupyter Notebooks using Hail, a cloud-compatible genomic data analytic tool with efficiency and scalability to handle the large dataset. Example notebooks will be provided to the attendees. We will also address importing external (local or cloud-based) data in the Researcher Workbench.

The close of the workshop will be a Q&A session.

A Guide to Card Sort Methods for Engaging Participants and Patients

Tuesday, November 5: 10:00 am - 12:00 pm

Pricing: \$40 ASHG member, \$55 nonmember. Advance ticket purchase required to attend.

Includes: Entrance to the workshop & access to the OnDemand recording Speakers: Karen Meagher, Sara Watson, Anya Prince, Jean Cadigan

This workshop consists of a hands-on demonstration of card sort methods for eliciting stakeholder values and attitudes toward genetics.

Card sort methods provide sorters with a "stack" (virtual or physical) of items and asks them to group the items. In "closed" sorts, sorters are given pre-established categories; in the "open" alternative, sorters develop and articulate their own group concepts. Card sorts have been used in social science and applied ethics research, such as to elicit sorters' views on end-of-life care and scarce resource allocation.

As a case study, attendees will hear about the design and outcomes of an ethical, legal, and social implications (ELSI) project that employed card sort methods as a community engagement activity to discern people's values regarding the development of polygenic scores for social traits.

The workshop will provide a tutorial to apply card sorting for engaging research participants, patients, and communities about genetics. The tutorial will include a take-home user-guide that describes such steps as how to identify card sort goals; select close, open, or hybrid designs; develop a facilitation guide; and analyze results.

The workshop will also include a Q&A session and facilitated breakout groups for attendees to perform card sorting and consider its various strengths and limitations. Attendees will have ample time to develop and practice incorporating card sort methods into their own research, clinical care, or outreach.

Using the New Ensembl Genome Browser and Variant Effect Predictor (VEP) to Analyse and Interpret Genomic Variation Data

Tuesday, November 5: 10:00 am - 12:00 pm

Pricing: \$40 ASHG member, \$55 nonmember. Advance ticket purchase required to attend.

Includes: Entrance to the workshop & access to the OnDemand recording Speakers: Jane Loveland, Ola Austine, Aleena Mushtaq, Louisse Mirabueno

Identifying variants of interest is crucial to understanding the genetic basis of human diseases and phenotypes. However, the prioritisation of variants within large datasets requires the consideration of disparate evidence including functional prediction, allele frequencies, co-occurrence with phenotypes and conservation across species. The Ensembl genome browser and Variant Effect Predictor (Ensembl VEP) enable prediction of the functional consequences of genetic variants in the context of the Ensembl/GENCODE geneset, MANE (Matched Annotation from NCBI and EMBL-EBI) transcripts, regulatory elements and evolutionarily conserved regions. Ensembl also integrates allele frequency, protein function and phenotype association data from many resources, providing an efficient and flexible platform for variant filtering and prioritisation.

This workshop will guide you through the Ensembl/GENCODE and MANE genesets and how to use the <u>new Ensembl genome browser</u> and Ensembl VEP to annotate and prioritise genetic variants in the context of the human reference genome assemblies and human pangenomes.

There will be a combination of live demonstrations, polling and group exercises, where example clinical datasets will be annotated, with individual variants interpreted and prioritised.

A laptop is required to participate in the hands-on aspects of this workshop.

Building Understanding and Practical Skills in Community Engagement for Genetic Research

Tuesday, November 5: 2:00 pm - 4:00 pm

Pricing: \$40 ASHG member, \$55 nonmember. Advance ticket purchase required to attend.

Includes: Entrance to the workshop & access to the OnDemand recording

Speakers: Mildred Cho, Alham Saadat, Sandra Lee, Maya Sabatello, Kellan Baker

This interactive workshop will use actual cases of community-engaged research (CEnR) to illustrate why and when CEnR is needed to enhance both scientific and ethical quality, and will be led by instructors with diverse expertise and lived experiences.

Part 1 will address fundamental questions that should be asked early in the CEnR planning process. Participants will then learn about different models of CEnR, highlighting when they can and should be used, and use an interactive platform to share their experiences with CEnR. Participants will use a case example of a genetic research project to learn why and how CEnR might be integrated as a key element of research design. The interactive platform will be used to explore issues such as what questions a genetic researcher would want to ask before embarking on CEnR. Participants will discuss key issues such as defining communities, power dynamics, the resources necessary to conduct CEnR, how to build trust, and impacts on genetic research projects.

In Part 2, participants will break into small groups to develop a plan that includes articulation of the rationale for a genetic research project, the goals and appropriate approaches for CEnR, defining relevant communities and identifying community representatives, and the nature of engagement.

In Part 3, small groups will reconvene to present their plans, highlighting their objectives, methods, intended benefits, and anticipated challenges.

Elevating Education: Engaging with Real Data and Tools in the Cloud

Tuesday, November 5: 2:00 pm - 4:00 pm

Pricing: \$40 ASHG member, \$55 nonmember. Advance ticket purchase required to attend.

Includes: Entrance to the workshop & access to the OnDemand recording Speakers: Zelia Worman, Emily Hughes, Cera Fisher, Nathalie Volkheimer

Teaching and learning genomics is hard. Apart from the struggle to master concepts and content, the skills and methods are difficult to comprehend without hands-on experiences. NHLBI BioData Catalyst® (BDC) makes teaching bioinformatics accessible and engaging, maximizing educational impact in a short period. BDC offers pre-installed tools in RStudio, SAS, and Jupyter Notebooks in a unified environment, reducing setup time. In addition, BDC also provides access to over 800 tools in a no-code interface. It supports flexible teaching formats, efficient student progress tracking, and collaborative workspaces for clear, individual contribution in group projects.

This workshop will provide a train-the-trainer intro to BDC for instructors, including brainstorming sessions, course design discussions, and teaching plans for short and long courses.

Participants will leave with a clear path to incorporating cloud-based bioinformatics in their teaching, with:

- · Course materials for students, including test data, methods, and example analyses
- Compute resource with sufficient computational power & software packages for carrying out hands-on exercises
- Real-time monitoring of student progress and review of final work
- Dedicated BDC technical support staff to allow you to concentrate on content and teaching
- You and your students can continue working and learning in the BDC ecosystem after the course

Hidden Features of the UCSC Genome Browser

Tuesday, November 5: 2:00 pm - 4:00 pm

Pricing: \$40 ASHG member, \$55 nonmember. Advance ticket purchase required to attend.

Includes: Entrance to the workshop & access to the OnDemand recording

Speakers: Robert Kuhn

The UCSC Genome Browser is a widely used tool for both clinical genetics and in the research lab. A visualization tool that combines data from a host of projects in an easy-to-use graphical interface, the Browser allows scientists to explore a wide variety of data types, using their own intuition to guide the configuration of the data.

To keep the Browser interface from becoming overwhelming, it has been necessary to make only a subset of features obvious to the user. Based on years of experience in workshops, we know that many of the most interesting features have not been discovered by even experienced users. This workshop will begin with a demonstration of these features, including and especially, recently released user-controlled options.

Workshop participants will use these features in service of solving problems. Interactive elements will include exercises designed to put the new features into practice. Participants will create, save and share their sessions with each other. The workshop is designed for the new generation of geneticists who have not grown up with the Browser during its 24 years of development, highlighting data sets that are used most frequently in interpreting variants, both CNVs and SNVs.

Participants will learn how to use the highlight tool enhance their Browser view, how to save and share sessions and how to export high-resolution images for publication. Participants should bring a fully charged laptop and expect to participate.

Luncheons

CDC Professional Development Workshop Luncheon

Wednesday, November 6: 11:45am - 1:15pm

Pricing: \$45 ASHG member, \$60 nonmember. Advance ticket purchase required to attend.

Includes a boxed lunch.

Speakers: TBD

Description coming soon.

Diversity, Equity, and Inclusion Luncheon

Wednesday, November 6: 11:45 am - 1:15 pm

Pricing: \$45 ASHG member, \$60 nonmember. Advance ticket purchase required to attend.

Includes a boxed lunch.

Speakers: TBD

The Diversity, Equity, and Inclusion Luncheon is a ticketed event that provides an exceptional venue for networking and showcasing ASHG programs and partnerships focused on advancing diversity, equity, and inclusion in human genetics and genomics.

Addressing the Challenges of Polygenic Scores in Human Genetic Research

Thursday, November 7: 11:45am - 1:15pm

Pricing: \$45 ASHG member, \$60 nonmember. Advance ticket purchase required to attend.

Includes a boxed lunch.

Speakers: TBD

ASHG Professional Practice and Social Implications (PPSI) committee-led discussion with a speaker panel and audience engagement on a topic related to the committee's ongoing work in the ethical, legal, and social implications of human genetics research.

Behind the Scenes: Publications Workshop Luncheon

Friday, November 8: 11:45am - 1:15pm

Pricing: \$45 ASHG member, \$60 nonmember. Advance ticket purchase required to attend.

Includes a boxed lunch.

Speakers: TBD

This workshop will provide you with information to help navigate the scientific publication process. Editors from several journals, including *AJHG* and *HGG Advances*, will give you a behind-the-scenes view of scientific publishing, covering topics such as data sharing, open access publication, and the ins and outs of the peer review process. There will be plenty of time for questions and networking. This session is geared towards trainees and junior faculty who are relatively new to publishing.

Receptions

Trainee Reception (open to trainee and early career attendees only)

Tuesday, November 5: 7:00 pm - 8:30 pm

Pricing: \$45 ASHG member, \$60 nonmember. Advance ticket purchase required to attend.

Includes heavy appetizers and one drink ticket (beer, wine, and soft drinks only).

Speakers: TBD

This fun event provides an opportunity to network with other trainees and get advice from potential mentors at different career stages. Enjoy getting to see and meet one another at ASHG while enjoying beverages and food!

Diversity, Equity, and Inclusion Reception

Wednesday, November 6: 7:00 pm - 8:30 pm

Pricing: \$45 ASHG member, \$60 nonmember. Advance ticket purchase required to attend.

Includes heavy appetizers and one drink ticket (beer, wine, and soft drinks only).

Speakers: TBD

The Reception will recognize and celebrate the importance of diversity, equity, and inclusion, as well as provide networking for the research and training community.

Continuing Education Credits

	EARLY	ADVANCE	LATE / ONSITE
	(Through Aug	(Aug 21 – Oct	(Starting Oct 1
	21 at 5pm ET)	1 at 5pm ET)	at 5pm)
CME Credits (for those with a valid ACCME license)	\$85	\$105	\$125

CEU Credits for Genetic Counselors (for	\$85	\$105	\$125
those with a valid NSGC license)			

Attendees can earn continuing education contact hours for California Licensed Clinical and Molecular Laboratory Directors (P.A.C.E.®) at no additional cost, however, an <u>additional form must be completed</u> and valid P.A.C.E.® license information must be provided.