

PROGRAMME BOOKLET

Hynes Convention Center
Boston, USA
October 21 - 23, 2019

www.ga4gh.org
@GA4GH
#GA4GHPlenary2019



Table of Contents

Letter from 7th Plenary Committee Chair	4
Letter from GA4GH Chair	5
Agenda	6
Session Overviews	8
Keynote Speakers	14
Plenary Speakers	16
GA4GH Standards	28
Get Involved with GA4GH	32
Venue Map	34

ABOUT 7TH PLENARY

The GA4GH 7th Plenary Meeting will bring together a diverse group of stakeholders for two days of keynotes, talks, updates, and workshops that will focus on advancing development work for the immediate data sharing needs of the community. We are grateful for the support of meeting sponsors, Genome Canada, Microsoft, Illumina, AbbVie, Google Cloud, and Vertex Pharmaceuticals. This meeting was supported by the National Institute of Child Health and Human Development of the National Institutes of Health (NICHD) under award number RFA-HD-17-001.

NEED ASSISTANCE?

If you need anything during your time in Boston, please do not hesitate to reach out. The registration booth will be staffed throughout the meeting and you can always reach us on Twitter (@GA4GH) or via email (info@ga4gh.org).

PROGRAMME COMMITTEE

Robert Freimuth (Chair), Tommi Nyrönen, Heidi Sofia, Stacey Donnelly, Hidewaki Nakagawa, David Siedzik, Nicola Mulder, Madeleine Murtagh, Sarion Bowers, Jeff Gentry, and Peter Goodhand



Welcome to GA4GH 7th Plenary

Dear Colleagues,

Welcome to Boston and the 7th Plenary Meeting of the Global Alliance for Genomics and Health. It is my pleasure to have chaired this year's programme committee—a diverse and engaged group of individuals from across the GA4GH community—and to have led the delivery of what I believe will be a dynamic and engaging agenda over the next two days.



Members of the GA4GH Work Streams will present updates on their activities, including five new approved standards from the GA4GH Connect Strategic Roadmap. We will hear keynote presentations from Eric Lander (Broad Institute of MIT and Harvard), Heidi Rehm (Massachusetts General Hospital), and Julia Stoyanovich (New York University) and participate in breakout sessions and panel discussions on diversity in genomic datasets, future technologies, patient perspectives, collaborative genomic standards development, and more.

I am truly looking forward to engaging with everyone as we work together to enable an exciting new era of genomic research and medicine.

Sincerely,

A handwritten signature in black ink that reads "Robert R. Freimuth". The signature is written in a cursive style.

Robert Freimuth, PhD
GA4GH 7th Plenary Programme Committee Chair
Division of Digital Health Sciences
Enterprise Information and Knowledge Management
Center for Individualized Medicine
Mayo Clinic



Message from the GA4GH Chair

To the GA4GH Community:

I am delighted to welcome you to the GA4GH 7th Plenary Meeting—the fourth under my watch as GA4GH Chair. The first time I wrote one of these letters, we were in Orlando and about to unveil the new GA4GH Connect Strategic Plan. A roadmap of more than two dozen standards for data discovery, access, transfer, storage, and analysis followed soon thereafter. I'm happy to report that progress on that roadmap has been moving at a steady clip. In Basel, last year, we released the first set of approved deliverables and this year we will announce several more.



I invite you to learn more about these deliverables and how you can get involved in their future development—or implement them at your own institution—at the GA4GH Updates session on Tuesday afternoon. You can also talk to the developers directly at the attended poster sessions on Monday and Tuesday evenings.

I am proud of the work our contributors have accomplished and excited to see what they will accomplish next.

Thank you for joining us here in Boston this year. I anticipate the next few days will be filled with excitement, innovation, and collaboration.

Warm Regards,

A handwritten signature in blue ink that reads "Ewan Birney". The signature is written in a cursive, slightly slanted style.

Ewan Birney, PhD
GA4GH Chair
Director, EMBL-EBI



Agenda

TUESDAY, OCTOBER 22, 2019

8:45	Welcome & Opening Remarks (Room 302/304/306)				
9:00	Intro to GA4GH (Room 300)	Data Access & Usage (Room 303)	Collaborative Genomics Standards Development (Room 301)	National Initiatives Workshop (Room 313)	GenoPri (Room 310)
10:30	BREAK				
11:00	Patient Perspectives (Room 303)	The Future of Technology in Genomics (Room 300)	ELIXIR:: GA4GH Strategic Partnership (Room 301)	National Initiatives Workshop (Room 313)	GenoPri (Room 310)
12:30	LUNCH				
13:30	KEYNOTE: JULIA STOYANOVICH “TransFAT: Translating Fairness, Accountability, and Transparency into Data Science Practice” (Room 302/304/306)				
14:15	GA4GH Updates (Room 302/304/306)				
15:15	BREAK				
15:45	GA4GH Updates (Cont’d) (Room 302/304/306)				
17:30	Cocktail & Poster Session				



Agenda

WEDNESDAY, OCTOBER 23, 2019

9:00	Opening Remarks (Room 302/304/306)
9:15	KEYNOTE: HEIDI REHM “Advancing Medicine Through Data Sharing and Collaboration on a Global Scale” (Room 302/304/306)
10:00	Panel: Collaborating Across Traditional Boundaries (Room 302/304/206)
11:00	BREAK
11:30	Panel: Diversity in Genomics (Room 302/304/306)
12:30	LUNCH
13:30	Panel: The Future of Genomic and Health Data (Room 302/304/306)
14:30	BREAK
15:00	KEYNOTE: ERIC LANDER “The Power of Data Sharing in Genomics and Beyond” (Room 302/304/306)
15:45	Closing Remarks (Room 302/304/306)



Session Overviews

Breakout Session: Data Access & Usage

Tuesday, October 22 • 9:00AM - 10:30AM

Challenges and future solutions to streamlining genomic data access

Chairs: Sarion Bowers (Wellcome Sanger Institute), Tommi Nyrönen (CSC-IT Center for Science)

Speakers: Melanie Courtot (European Bioinformatics Institute), Jonathan Dursi (The Hospital for Sick Children), Maarten Kremers (SURFnet)

Breakout Session: Collaborative Genomics Standards Development

Tuesday, October 22 • 9:00AM - 10:30AM

Discussion on collaboration and alignment across standards development organizations and beyond

Chair: Robert Freimuth (Mayo Clinic)

Speakers: Adam Berger (National Institutes of Health), Bron Kisler (National Institutes of Health), Kim Pruitt (National Institutes of Health), Soichi Ogishima (Tohoku University)

Breakout Session: Intro to GA4GH

Tuesday, October 22 • 9:00AM - 10:30AM

Overview of GA4GH's role in building and maintaining standards to enable global genomic data exchange and interoperability

Speakers: Peter Goodhand (GA4GH), Rishi Nag (GA4GH), Melissa Konopko (GA4GH), Lindsay Smith (GA4GH), Adrian Thorogood (GA4GH)

Breakout Session: Patient Perspectives

Tuesday, October 22 • 11:00AM - 12:30PM

Patient perspectives towards genomics and data sharing

Chairs: Ada Hamosh (Johns Hopkins University), Tiffany Boughtwood (Australian Genomics)

Speakers: Anna Middleton (Wellcome Sanger Institute), Illona Schelle (Inspire2Live), Richard Milne (Wellcome Sanger Institute), Eric Minikel (Broad Institute), Sonia Vallabh (Broad Institute)



Session Overviews

Breakout Session: The Future of Technology in Genomics

Tuesday, October 22 • 11:00AM - 12:30PM

How new technology is shaping genomics

Chairs: Andy Yates (European Bioinformatics Institute), Moran Cabili (Foundation Medicine)

Speakers: Sumit Januar (Global Gene Corp), Patrick Kemmeren (Princess Maxima Center), Marco Roos (Leiden University Medical Center), Barend Mons (Leiden University Medical Center)

GA4GH Updates

Tuesday, October 22 • 2:15PM - 5:30PM

Updates on the GA4GH roadmap and product development

Speakers: GA4GH Work Stream Product Leads

Panel: Collaborating Across Traditional Boundaries

Wednesday, October 23 • 10:00AM - 11:00AM

Building partnerships across geographic, institutional, and industry boundaries

Chair: Kathryn North (Australian Genomics)

Speakers: Julia Wilson (Wellcome Sanger Institute), Ian Dunham (European Bioinformatics Institute), Bronwyn MacInnis (Broad Institute)

Panel: Diversity in Genomics

Wednesday, October 23 • 11:30AM - 12:30PM

The importance of increasing diversity and representation in research populations

Chair: Madeleine Murtagh (University of Newcastle)

Speakers: Calvin Ho (National University of Singapore), Alicia Martin (Broad Institute), Mavis Machirori (University of Newcastle)

Panel: The Future of Genomic and Health Data

Wednesday, October 23 • 1:30PM - 2:30PM

Discussing the future trends impacting genomics and health data research

Chair: Laszlo Radvanyi (OICR)

Speakers: Meng Yang (BGI Big Data Center), Jean-Pierre Hubaux (EPFL), Teresa Cabán Zayas (US Department of Health and Human Services)



6th International Workshop on Genome Privacy and Security (GenoPri'19)

OCTOBER 21 - 22, 2019
HYNES CONVENTION CENTER
3RD FLOOR, ROOM 310

KEYNOTE SPEAKERS

Bonnier Berger (MIT)
Mwenz Blell (Newcastle University)
Edward Dove (Edinburgh University)
Amalio Telenti (Vir Biotechnology)

SESSION TOPICS

“People” • Monday, Oct 21 • 2:20PM - 3:05PM
“Regulation” • Monday, Oct 21 • 3:35PM - 4:20 PM
“Models” • Tuesday, Oct 22 • 9:30AM - 10:15AM
“Tools” • Tuesday, Oct 22 • 10:15AM - 10:45AM

View the full program at: genopri.org/program

#GenoPri19





National Initiatives Workshop

OCTOBER 22, 2019
9:00AM - 12:30PM

HYNES CONVENTION CENTER
ROOM 313

All plenary attendees associated with (or interested in) a national genomic data initiative are welcome to join us for a special workshop to discuss opportunities for sharing resources and knowledge

AGENDA OVERVIEW

National Initiatives Toolbox
Federated Data Analysis Across Cohorts
Curation
Genomic Data Access
Future Directions of National Initiatives group

CHAIR

Kathryn North
(Australian Genomics)



ELIXIR::GA4GH

Strategic Partnership

HOW WE'RE WORKING TOGETHER

BREAKOUT SESSION • ROOM 301
OCTOBER 22 • 11:00AM - 12:30PM

ELIXIR, the European infrastructure for life sciences, has been a key contributor to GA4GH deliverables since 2014. A Strategic Partnership between the two organizations was established in 2019 to help coordinate standards development and GA4GH engagement across Europe.

Open to all, this session will focus on:

- ELIXIR and National implementations of approved GA4GH standards
- Collaborating across Driver Projects to drive standards development
- Interoperability of standards from across Work Streams



Global Alliance
for Genomics & Health
Collaborate. Innovate. Accelerate.



Poster Session

Learn about the work of GA4GH and related activities that will enable the responsible, voluntary, and secure sharing of genomic and health-related data to the broader genomics and health community.



SPEAK TO THE POSTER PRESENTERS!

Monday, October 21 • Cocktail Reception (5:45pm - 6:45pm)

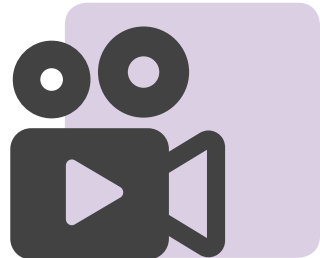
Tuesday, October 22 • Cocktail Reception (5:30pm - 6:30pm)

Posters will remain mounted for the duration of the meeting.

Video Booth

Tell us your story about genomics and health data sharing!

- Why do genomics and data sharing matter?
- Why does GA4GH matter?
- How is your work impacting the world?



Record your thoughts at our video booth!

Monday, October 21 • Cocktail Reception (5:45pm - 7:45pm)

Tuesday, October 22 • Cocktail Reception (5:30pm - 7:30pm)



Keynote Speaker: Eric Lander

The Power of Data Sharing in Genomics and Beyond

Wednesday, October 23
3:00PM - 3:45PM

Eric Lander is president and founding director of the Broad Institute of MIT and Harvard. A geneticist, molecular biologist, and mathematician, he has played a pioneering role in the reading, understanding, and biomedical application of the human genome. He was a principal leader of the Human Genome Project.



Keynote Speaker: Julia Stoyanovich

TransFAT: Translating Fairness, Accountability, and Transparency into Data Science Practice

Tuesday, October 22
1:30PM - 2:15PM

Julia Stoyanovich is an assistant professor of Computer Science and Engineering and of Data Science at New York University. Julia's research focuses on responsible data management and analysis practices: on operationalizing fairness, diversity, transparency, and data protection in all stages of the data acquisition and processing lifecycle.



Keynote Speaker: Heidi Rehm

Advancing Medicine Through Data Sharing and Collaboration on a Global Scale

Wednesday, October 23
9:15AM - 10:00AM

Heidi Rehm is the Chief Genomics Officer in the Department of Medicine and at the Center for Genomic Medicine at the Massachusetts General Hospital. With leadership roles at the Broad Institute, ClinGen, and the Matchmaker Exchange, Rehm is deeply involved in defining standards for the use of next generation sequencing.



Welcoming Remarks: David Altshuler



EVP Global Research and Chief Scientific Officer
(**Vertex Pharmaceuticals**),
Strategic Advisory Board Member and Founding Chair (**GA4GH**)



Plenary Speakers

Sergi Beltran

Head of Bioinformatics, Spanish National Genome Analysis Center
Breakout Session: National Initiatives Workshop

Adam Berger

Director of Clinical & Healthcare Research Policy,
National Institutes of Health
Breakout Session: Collaborative Genomics Standards Development

David Bernick

Senior Director, CISO, Broad Institute
GA4GH Updates: Authentication & Authorization Infrastructure

Ewan Birney

Director, European Bioinformatics Institute
Opening & Welcoming Remarks

Tiffany Boughtwood

Program Manager, Australian Genomics
Breakout Session: Patient Perspectives

Sarion Bowers

Head of Policy, Wellcome Sanger Institute
Breakout Session: Data Access and Usage

Moran Cabili

Associate Director, Foundation Medicine
Breakout Session: The Future of Technology in Genomics

Jorge Contreras

Professor, University of Utah
GA4GH Updates: Copyright / IP Policy

Melanie Courtot

Metadata Standards Coordinator, European Bioinformatics Institute
Breakout Session: Data Access and Usage
Breakout Session: ELIXIR::GA4GH Strategic Partnership
GA4GH Updates: Data Use Ontology



Plenary Speakers

Rob Davies

Senior Scientific Manager, Wellcome Sanger Institute
GA4GH Updates: CRAM

Ian Dunham

Open Targets Director, European Bioinformatics Institute
Panel: Collaborating Across Traditional Boundaries

Jonathan Dursi

Architect & Technical Lead, CanDIG, The Hospital for Sick Children
Breakout Session: Data Access and Usage

Stephanie Dyke

Ethics & Policy Researcher, McGill University
GA4GH Updates: GA4GH Passports

Yossi Farjoun

Associate Director of Scientific Computational Methods, Broad Institute
GA4GH Updates: The Future of VCF

Robert Freimuth

Assistant Professor, Mayo Clinic
Breakout Session: Collaborative Genomics Standards Development

David Glazer

Engineering Director, Verily
GA4GH Updates: Data Repository Service API

Peter Goodhand

CEO, Global Alliance for Genomics and Health
Breakout Session: Intro to GA4GH

Melissa Haendel

Co-Director, Oregon Health and Science University
GA4GH Updates: Phenopackets



Plenary Speakers

Ada Hamosh

Professor, Johns Hopkins University
Breakout Session: Patient Perspectives

Reece Hart

Independent Contractor
GA4GH Updates: Variation Representation

Calvin Ho

Assistant Professor, National University of Singapore
Panel: Diversity in Genomics

Oliver Hofmann

Associate Professor, University of Melbourne
Breakout Session: National Initiatives Workshop

Jean-Pierre Hubaux

Professor, Swiss Federal Institute of Technology - EPFL
Panel: The Future of Genomic and Health Data
GA4GH Updates: Data Security Infrastructure Policy

Sumit Jamuar

Chairman & CEO, Global Gene Corp
Breakout Session: The Future of Technology in Genomics

Patrick Kemmeren

Principal Investigator, Princess Máxima Center
Breakout Session: The Future of Technology in Genomics

Bron Kisler

Bioinformatics Specialist, National Institutes of Health
Breakout Session: Collaborative Genomics Standards Development

Bartha Knoppers

Professor and Director, Centre of Genomics and Policy, McGill University
GA4GH Updates: Regulatory & Ethics



Plenary Speakers

Ray Krasinski

Director, Interoperability and Standardization, Philips
GA4GH Updates: Copyright / IP Policy

Melissa Konopko

Technical Work Stream Manager, Global Alliance for Genomics and Health
Breakout Session: Intro to GA4GH

Maarten Kremers

Technical Product Manager, SURFnet
Breakout Session: Data Access and Usage

Jonathan Lawson

Software Product Manager, Broad Institute
GA4GH Updates: Data Use Ontology

Mike Lin

Independent Contractor
GA4GH Updates: htsget API

Mikael Linden

Senior Application Specialist, CSC-IT Center for Science
Breakout Session: ELIXIR::GA4GH Strategic Partnership

Mavis Machirori

Postdoctoral Researcher, Newcastle University
Panel: Diversity in Genomics
Breakout Session: National Initiatives Workshop

Bronwyn MacInnis

Associate Director, Malaria and Viral Genomics, Broad Institute
Panel: Collaborating Across Traditional Boundaries

Alicia Martin

Instructor, Broad Institute
Panel: Diversity in Genomics



Plenary Speakers

Ellen McDonagh

Head of Curation, Genomics England

Breakout Session: National Initiatives Workshop

Anna Middleton

Head of Society & Ethics Research, Wellcome Sanger Institute

Breakout Session: Patient Perspectives

Richard Milne

Senior Social Scientist, Wellcome Sanger Institute

Breakout Session: Patient Perspectives

Eric Minikel

Associated Scientist, Broad Institute

Breakout Session: Patient Perspectives

Barend Mons

Professor, Leiden University Medical Center

Breakout Session: The Future of Technology in Genomics

Madeleine Murtagh

Professor, Newcastle University

Panel: Diversity in Genomics

Rishi Nag

Technical Work Stream Manager, Global Alliance for Genomics and Health

Breakout Session: Intro to GA4GH

Hidewaki Nakagawa

Team Leader, RIKEN Center for Integrative Medical Sciences

Breakout Session: National Initiatives

Tommi Nyrönen

Head of ELIXIR Finland Node, CSC-IT Center for Science

Breakout Session: Data Access and Usage

GA4GH Updates: GA4GH Passports



Plenary Speakers

Kathryn North

Lead Investigator, Australian Genomics

Breakout Session: National Initiatives Workshop

Panel: Collaborating Across Traditional Boundaries

Brian O'Connor

Director, University of California Santa Cruz

GA4GH Updates: Workflow Execution Service API

Soichi Ogishima

Professor, Tohoku University

Breakout Session: Development of Genomics Standards

Sabine Österle

Scientific Project Manager, SIB-Swiss Institute of Bioinformatics

Breakout Session: National Initiatives Workshop

Kim Pruitt

Director, Data Services, National Center for Biotechnology Information

Breakout Session: Collaborative Genomics Standards Development

Laszlo Radvanyi

President & Scientific Director, Ontario Institute for Cancer Research

Breakout Session: The Future of Genomic and Health Data

Jordi Rambla

Team Leader, Centre for Genomic Regulation

Breakout Session: ELIXIR::GA4GH Strategic Partnership

GA4GH Updates: Beacon API

Marco Roos

Assistant Professor, Leiden University Medical Center

Breakout Session: The Future of Technology in Genomics

Augusto Rendon

Director of Bioinformatics, Genomics England

Breakout Session: National Initiatives Workshop



Plenary Speakers

Gary Saunders

Human Data Coordinator, ELIXIR

Breakout Session: ELIXIR::GA4GH Strategic Partnership

Ilona Schelle

Patient Advocate, Inspire2Live

Breakout Session: Patient Perspectives

Alexander Senf

Project Leader, European Bioinformatics Institute

GA4GH Updates: Crypt4GH

Per Sikora

Co-Chair, Genomic Medicine Sweden

Breakout Session: National Initiatives

Lindsay Smith

Technical Work Stream Manager, Global Alliance for Genomics and Health

Breakout Session: Intro to GA4GH

Dylan Spalding

Coordinator, European Bioinformatics Institute

Breakout Session: ELIXIR::GA4GH Strategic Partnership

Zornitza Stark

Clinical Geneticist, Victorian Clinical Genetics Service

Breakout Session: National Initiatives Workshop

Adrian Thorogood

Work Stream Manager, Global Alliance for Genomics and Health

Breakout Session: Intro to GA4GH

Juha Törnroos

Software Architect, ELIXIR Finland

Breakout Session: ELIXIR::GA4GH Strategic Partnership



Plenary Speakers

Sean Upchurch

Software Developer, California Institute of Technology
GA4GH Updates: RNAget API

Sonia Vallabh

Group Lead, Broad Institute
Breakout Session: Patient Perspectives

Susheel Varma

ELIXIR Competence Centre Project Manager,
European Bioinformatics Institute
Breakout Session: ELIXIR::GA4GH Strategic Partnership

Craig Voisin

Software Engineer, Google
GA4GH Update: GA4GH Passports

Julia Wilson

Associate Director, Wellcome Sanger Institute
Panel: Collaborating Across Traditional Boundaries

Meng Yang

Vice Director, BGI Big Data Center
Panel: The Future of Genomic and Health Data

Andy Yates

Team Lead, European Bioinformatics Institute
Breakout Session: The Future of Technology in Genomics
GA4GH Updates: refget API

Denis Yuen

Senior Software Developer, Ontario Institute for Cancer Research
GA4GH Updates: Tool Registry Service API

Teresa Cabán Zayas

Chief Scientist, US Department of Health and Human Services
Panel: The Future of Genomics and Health Data



Subscribe to the Monthly GA4GH GDPR Briefs!

Launched in September 2018 by the GA4GH Regulatory & Ethics Work Stream, the monthly GA4GH GDPR Brief features articles relating to the impact of the European General Data Protection Regulation (GDPR) on international health research and data sharing.

Past Topics include:

- When can I rely on broad consent for research?
- Are pseudonymised data within the GDPR's scope?
- How can researchers approach international data transfers under the GDPR?
- What is the difference between research ethics consent and data protection consent?

To view past GDPR briefs, visit: bit.ly/GDPRBriefs

GA4GH.ORG/SUBSCRIBE



GA4GH Timeline 2018-2019

OCTOBER 2018

refget, Beacon & WES APIs approved as GA4GH standards

JANUARY 2019

GA4GH publishes review of national genomic data initiatives in AJHG

FEBRUARY 2019

Data Use Ontology approved as new GA4GH technical standard

APRIL 2019

David Bernick & Jean-Pierre Hubaux named Data Security Work Stream co-leads

SEPTEMBER 2019

Updated GA4GH Regulatory & Ethics policies released

DECEMBER 2018

Heidi Rehm joins GA4GH Executive Team, Tommy Nyrönen & Michael Baudis named Work Stream Leads

FEBRUARY 2019

Eight new GA4GH Driver Projects announced

MARCH 2019

Extensions to the GA4GH Beacon API released

MAY 2019

ELIXIR & GA4GH establish strategic partnership

OCTOBER 2019

Six new deliverables approved as GA4GH standards



GA4GH 2019 DR

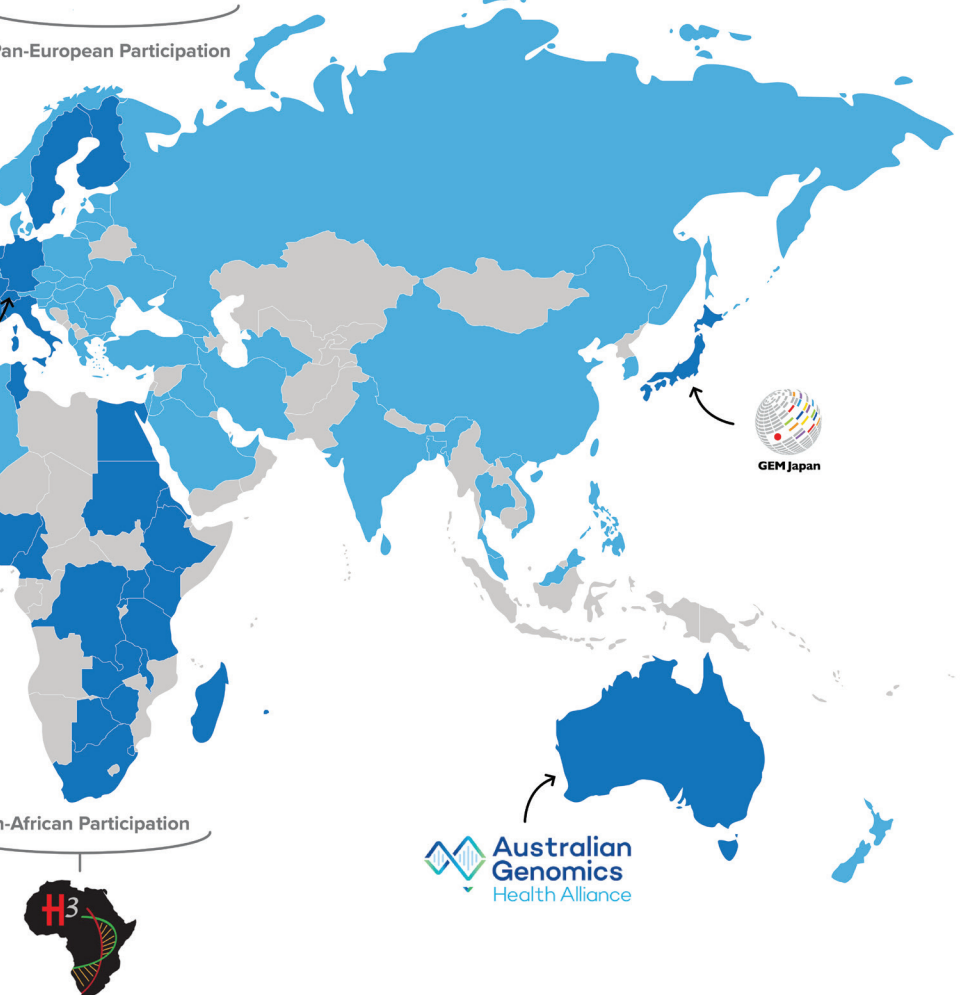


RIVER PROJECTS



- Direct Engagement
- Indirect Engagement

pan-European Participation



pan-African Participation



GA4GH Approved Standards

GA4GH approved standards enable responsible storing, discovering, accessing, and sharing of genomic and health-related data. Learn more: ga4gh.org/toolkit

BEACON API

A web-accessible service, the Beacon API offers global discovery of genomic variant data. bit.ly/GA4GHBeacon

CRYPT4GH 2019

An encrypted container file format that keeps genomic data secure throughout its lifetime. github.com/samtools

DATA SECURITY INFRASTRUCTURE POLICY 2019

Best practices for standards development and implementation to facilitate responsible sharing and processing of genomic data. bit.ly/GA4GHdsip

DATA USE ONTOLOGY 2019

Semantically tag datasets with restrictions about their usage. bit.ly/GA4GHDUO

FRAMEWORK FOR RESPONSIBLE SHARING OF GENOMIC AND HEALTH-RELATED DATA SHARING

Foundational principles and core elements for responsible data sharing, guided by human rights.

bit.ly/GA4GHframework

HTSGET API

A protocol for secure, efficient and reliable access to sequencing read and variation data. bit.ly/GA4GHhtsget



GA4GH Approved Standards

PHENOPACKETS 2019

A file format for exchanging high-level and deep clinical phenotype information. github.com/phenopackets

REFGET API

A framework to retrieve reference sequences without ambiguity from different database identifiers or aliases. bit.ly/GA4GHrefget

TOOL REGISTRY SERVICE (TRS) API 2019

An API that enables the portable exchange of tools and workflows. bit.ly/GA4GHtrs

UPDATED CONSENT & PRIVACY AND SECURITY POLICIES 2019

Guidance on autonomous decision-making; confidentiality and integrity of data services; and privacy for data sharing. bit.ly/GA4GHconsent2019 and bit.ly/GA4GHdataps

VARIATION AND READ FILE FORMATS

File formats for storing raw genomic read data (SAM, BAM, and CRAM) and for representing genomic variation (VCF and BCF). bit.ly/htspecs and bit.ly/GA4GHvcf

VARIATION REPRESENTATION (VR) 2019

A standard, extensible data model and message schema supporting the precise representation of genetic variation. bit.ly/GA4GHvr

WORKFLOW EXECUTION SERVICE (WES) API

An API allowing researchers to execute the same scientific tools and workflows across different environments. bit.ly/GA4GHWES



Standards Coming Soon...

GA4GH PASSPORTS

This system will allow users to establish identity and credentials claims to acquire access across datasets.

bit.ly/GA4GHpassports

AUTHENTICATION AND AUTHORIZATION INFRASTRUCTURE (AAI)

AAI is the GA4GH standard technical profile for authenticating the identity of individuals seeking to access data and services offered by the Driver Projects, and for authorizing access in accordance with applicable Driver Project policies. bit.ly/GA4GHaaai

DATA REPOSITORY SERVICE (DRS) API


An API that provides a common way to refer to and access data regardless of platform. bit.ly/GA4GHdrs



GA4GH Roadmap

The GA4GH Connect Strategic Roadmap

presents planned standards and frameworks to meet the data sharing needs of the genomics community. Originally rolled out in 2018, the Roadmap is now being updated for 2020, using both a bottom-up approach driven by the GA4GH Work Streams, as well as a top-down approach driven by a gap analysis led by Heidi Rehm (MGH, Broad Institute, GA4GH) and Andrew Morris (HDR UK).



**Learn more about
the GA4GH Roadmap:
ga4gh.org/roadmap**



Get Involved with GA4GH

JOIN A WORK STREAM

Cloud
Discovery
Large Scale Genomics

Rishi Nag
rishi.nag@ga4gh.org

Data Use & Researcher
Identities
Genomic Knowledge
Standards
Data Security

Melissa Konopko
melissa.konopko@ga4gh.org

Regulatory & Ethics

Adrian Thorogood
adrian.thorogood@ga4gh.org

Clinical & Phenotypic
Data Capture

Lindsay Smith
lindsay.smith@ga4gh.org



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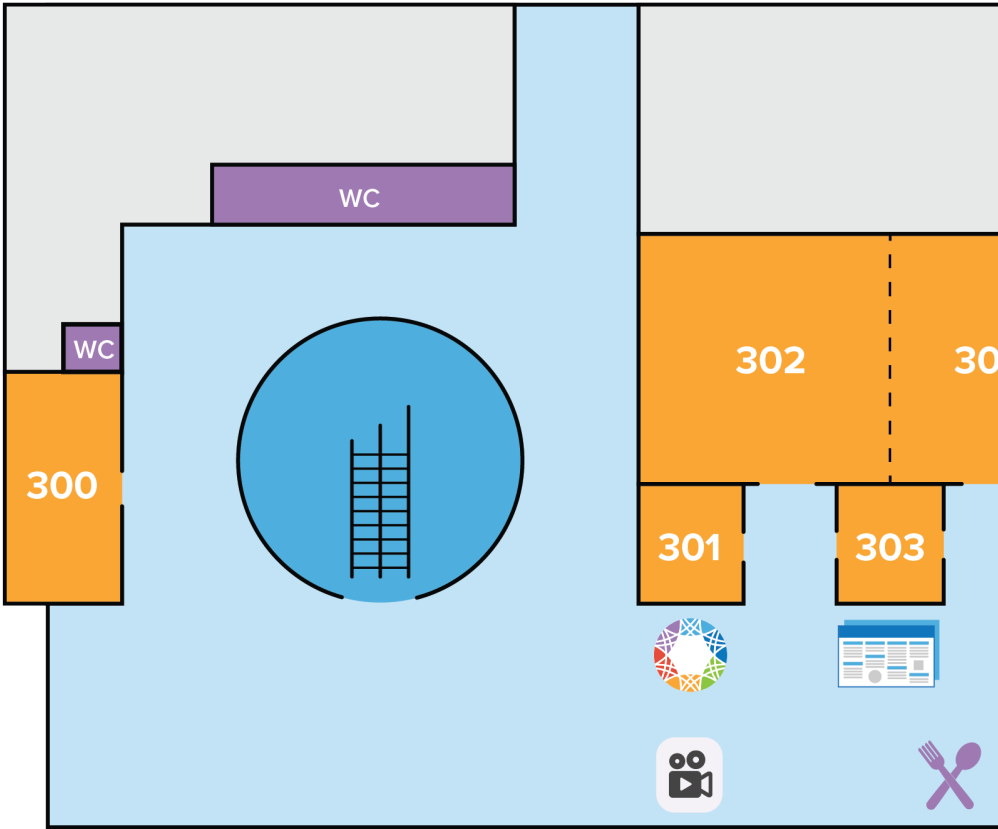
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Venue Map: Hynes Conve

3RD FLOOR • 900 BOYLSTON ST



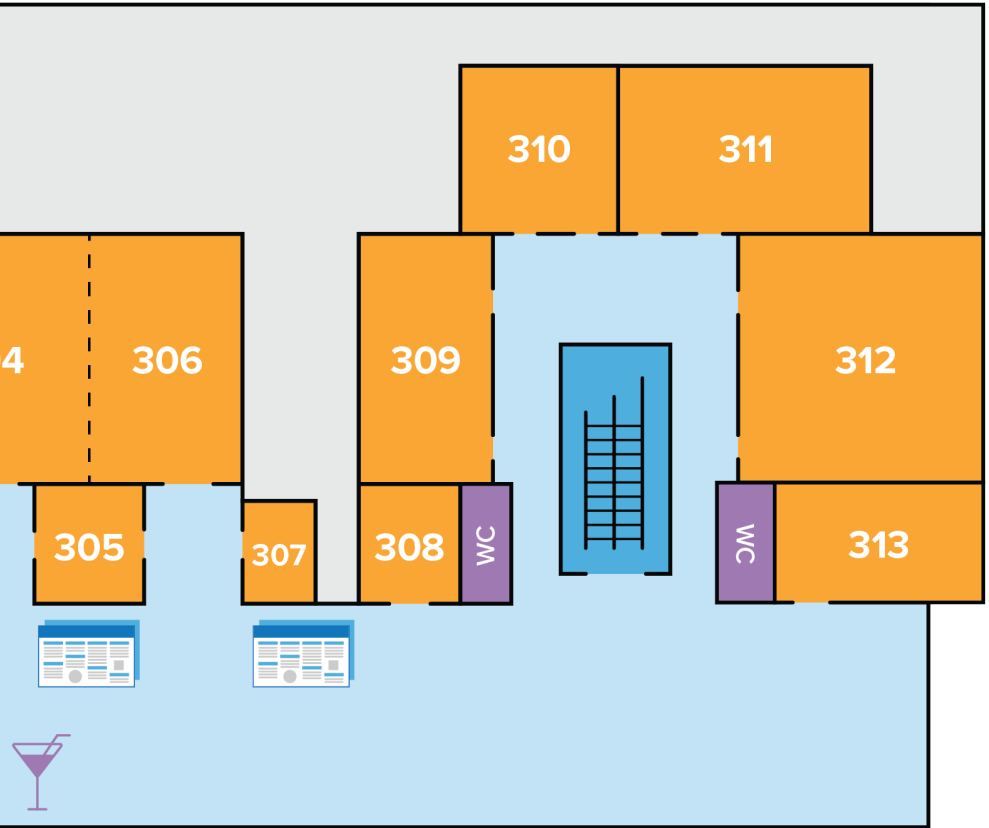
Registration



Poster Session



Convention Center



Meals



Cocktail Reception



Photo Booth





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