GA4GH 14th Plenary Meeting Report



The GA4GH 14th Plenary Meeting, co-hosted by Precision Health Research, Singapore (PRECISE), brought together nearly 600 attendees from across the global genomics and health community for a week of collaboration, technical advancement, and strategic reflection. The GA4GH Plenary meetings are crucial forums to convene people from a wide variety of sectors — including research, healthcare, government, and academia — to discuss the path forward for responsible global genomic and health interoperability.

This meeting marked GA4GH's first Plenary in Asia. During the opening session, GA4GH and PRECISE signed a Memorandum of Understanding for PRECISE to become GA4GH's first Regional Hub Institution, supporting GA4GH's coordination and engagement activities within the Asia-Pacific region. Learn more about the partnership here.

The 14th Plenary meeting centred on six core themes: evolving models of genomic data sharing, AI in genomic medicine, demonstrating value in genomic medicine, embedding genomics into healthcare systems, genomic newborn screening, and genomics at population scale.

Learn more about the meeting in our press release here. The meeting was also highlighted in *AsiaOne*, *BioSpectrum Asia*, and on the PRECISE website. Additionally, welcoming remarks from MDM Rahayu Mahzam, Minister of State, Ministry of Digital Development and Information and Ministry of Health, were published by the Ministry of Health, Singapore.

This year, more than 60 presenters participated in the poster session. To view all posters presented during the meeting, visit the Poster Guide.

We extend our gratitude to this year's meeting sponsors, funders, and Plenary Programme Committee Members for dedicating their time, energy, and resources to shaping this year's event and making the 14th Plenary meeting possible.

View photos from the meeting here and a brief recap video here.

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Plenary Day One (29 September 2026)

Opening remarks

Recording | Slides

The opening session of the Global Alliance for Genomics and Health (GA4GH) 14th Plenary meeting began with remarks from Patrick Tan, Executive Director of Precision Health Research, Singapore (PRECISE) and Heidi Rehm, Chair of GA4GH, who welcomed attendees to GA4GH's first Plenary meeting in Asia.

Afterwards, a signing ceremony of two Memoranda of Understanding commenced: one naming PRECISE as GA4GH's first Regional Hub Institution, based in the Asia-Pacific region, and the other establishing bilateral cooperation on genomic implementation with Genomics Australia.

Next, Shih Wee Seow, Chief of Staff and Senior Director of PRECISE, explained how Singapore has evolved to become a hub for science and health data connectivity. She also discussed PRECISE's

designation as GA4GH's first Regional Hub Institution, which serves to connect people, institutions, and programmes across the Asia-Pacific region.

Angela Page, Chief Strategy and Engagement Officer of GA4GH, provided an overview of GA4GH, diving into its mission to enable an interoperable learning health system through a community-oriented approach to building standards, frameworks, and tools. She ended with a call to action for more programmes and institutions to engage in harmonising and sharing genomic and health data.

Andrew and Tiffany's story

Recording

Andrew and Tiffany shared their family story of how rapid genetic testing diagnosed their daughter's Coffin-Siris syndrome, changing their family's path. Conducted at KK Women's and Children's Hospital, the diagnosis brought clarity, connected them with a supportive online community, and provided the confidence to have a second daughter. Highlighting how global research, databases, and rapid sequencing transformed their lives, the parents expressed gratitude to the medical team, researchers, and registry contributors for giving their family hope, answers, and support.

Panel: Rethinking openness: evolving models of genomic data sharing in a changing global landscape

Session chair: Zinaida Perova (Biomedical Data Hub)

KEYNOTE PRESENTATION

Geoffrey Ginsburg (*All of Us* Research Program / National Institutes of Health): "Advancing global collaboration through trusted genomic data sharing"

Recording | Slides

Geoffrey Ginsburg, Chief Medical and Scientific Officer and Acting Chief Data Officer of the *All of Us* Research Program, highlighted five innovations from *All of Us* that show how data sharing accelerates discovery and health impact. These include: building participant trust as infrastructure, enrolling diverse participants, scaling multimodal data, shifting from a data repository to a cloud platform with shared workspaces, and securing data through tiered access and egress controls. He called GA4GH the "connective tissue" that makes federated, cross-cohort research possible and promoted the S-index as an incentive to reward data reuse. He closed by urging researchers, funders, cohort leaders, and technologists to make cross-cohort analysis routine and to design for AI readiness.

Bron Kisler (ISO Technical Committee 215): “Global -omics standards: overview, data sharing, advancing clinical trials”

Recording | Slides

Bron Kisler, Chair of ISO Technical Committee 215 Genomics Informatics Sub-Committee, highlighted fast-growing global participation — especially from Asia — in developing standards for -omics data sharing and secondary use. Over the years, the subcommittee has coordinated closely with GA4GH. Phenopackets was their first joint standard, and Singapore is now leading a joint project on Whole Genome Sequencing Quality Control standards. Addressing interoperability challenges in precision medicine and clinical trials, Kisler introduced a new initiative hosted by the Pistoia Alliance: leveraging the IEEE BioCompute Object standard to handle complex -omics data missing from traditional CDISC structures.

Emma Tudini (QIMR Berghofer): “Shariant — retrofitting genomic knowledge sharing into a health system”

Recording | Slides

Emma Tudini, Shariant Project Manager, detailed the implementation and challenges of Shariant, a platform designed to facilitate genomic variant data sharing across Australia's federated healthcare system. Operating with a small central team and no national mandate, Shariant achieved widespread adoption by establishing controlled-access governance and building a technical transformation layer to automatically parse unstructured data from diverse lab systems without increasing workload for clinicians. This approach has engaged 18 clinical labs, integrated 48,000 records, resolved half of all variant classification discordances, and expanded Australian contributions to ClinVar from 300 to over 13,000 variants. Despite this success, the platform faces sustainability challenges as Commonwealth funding expires, underscoring the systemic difficulty of securing long-term operational support for vital health infrastructure projects.

Michael Baudis (University of Zurich / Swiss Institute of Bioinformatics): “The GA4GH Beacon protocol: federating data discovery, analysis, and sharing in biomedical genomics”

Recording | Slides

Michael Baudis, Professor of Bioinformatics at the University of Zurich, introduced the evolution of the GA4GH Beacon protocol, emphasising its crucial role in federated biomedical data discovery and sharing across distributed global systems. Initially designed for simple queries regarding the presence of specific genetic variants, the protocol now supports complex queries incorporating rich metadata, data use conditions, and structured data models. Beyond point-variant searches, modern Beacon

implementations enable population allele frequency browsers, privacy-preserving data aggregation, and seamless software integration. Illustrated by projects such as Progenetix and Genomics Thailand, Beacon effectively bridges isolated repositories to advance global research collaboration.

Panel discussion

Recording

The panel addressed key challenges in global genomic data sharing, open-access publishing, and infrastructure sustainability. To overcome barriers for researchers in low- and middle-income countries facing high open access fees, the panelists advocated for international collaboration and urged better-resourced co-authors to cover publishing costs. Regarding standard alignment and funding, experts highlighted the financial scarcity facing cross-boundary organisations such as GA4GH and called on national funding bodies and pharmaceutical entities to better support infrastructure. Additionally, the panel proposed convening journal editors to stop the penalisation of studies that analyse existing datasets rather than generating new data. Finally, the speakers stressed the importance of closing the loop with patients by returning lay summary reports and providing genetic counseling alongside actionable findings.

Panel: AI in health systems: reliability, bias, and clinical trust

Session chair: Susheel Varma (Sage Bionetworks)

KEYNOTE PRESENTATION

Bilal Mateen (PATH): “The future of AI in clinical decision support”

Recording | Slides

In his talk, Bilal Mateen, Chief AI Officer at PATH, warned against overhyping AI in healthcare without rigorous evidence. Highlighting that it took 30 years to prove the clinical value of FDA-approved breast cancer computer-aided detection (CAD), he cautioned that chasing unevaluated large language models (LLMs) risks wasting scarce health resources. Mateen demonstrated that while in-silico benchmarks — such as USMLE multiple-choice tests — fail to reflect real-world medicine, off-the-shelf LLMs outperformed local primary care clinicians in Rwanda on open-ended diagnostic tasks. However, in a real-world cluster randomised trial in Kenya involving nearly 10,000 patients, an AI diagnostic assistant improved clinical documentation and decision-making quality but yielded no meaningful improvement in patient health outcomes, largely because clinicians often ignored the AI's advice. Nevertheless, Mateen noted a promising "learning effect": over time, clinicians repeatedly exposed to AI feedback generated significantly fewer diagnostic errors independently. He concluded that rather

than viewing AI as a permanent diagnostic crutch, healthcare should leverage it as a tool to upskill clinicians while rigorously investigating long-term effects such as cognitive offloading and de-skilling.

Tudor Groza (BII A*STAR / KK Women's and Children's Hospital / Precision Health Research, Singapore [PRECISE]): "Accountable scale: from genomic AI to a national reliability engine"

Recording | Slides

Tudor Groza, Deputy CTO (Health System Integration) at PRECISE and Principal Scientist at A*STAR, addressed the shift of Singapore's precision health research program into Phase 3, targeting 450,000 citizens (~10% of the population) to integrate routine clinical genomics. He emphasised that the primary barrier to mainstream adoption is building "trust infrastructure" within clinical workflows, rather than improving data models or benchmarks. Groza proposed using LLMs as structured reasoning mechanisms, rather than autonomous decision-makers, guided by deterministic ACMG logic and curated knowledge bases to interrogate evidence, preserve uncertainty, and expose reasoning. Achieving 98% concordance across 1,700 variants in local trials, he argued that the goal must be creating governable, contestable, and accountable clinical pathways that continuously learn from expert human disagreement.

Rui-Yao Zhang (Tencent): "From molecules to populations: what makes data 'good' for AI in drug discovery"

In her talk, Rui-Yao Zhang, Head of AI for Life Sciences Business Strategy at Tencent, emphasised that advancing AI in biology depends far more on data quality than just on model architectures. Highlighting Tencent's multi-layered ecosystem — spanning cloud infrastructure, foundation models (e.g. TFold, SC BERT, UniPert G2CP), and scientific workflows such as BioBuddy — she noted that observational data alone fails to capture causal biology. To bridge this, Zhang outlined six essential properties for "model-ready" data to be: interventional, inclusive of negative or failed results, provenance-rich, representative of populations the model will serve, linked across date types, and explicitly consented and governable. Zhang emphasised that better models will come from better data, and better data will come from trust.

Melissa Haendel (University of North Carolina): "Trust relies on evidence and and provenance"

Recording | Slides

Melissa Haendel, Director of Precision Health and Translational Informatics at the University of North Carolina at Chapel Hill, addressed the critical challenges of reliability, bias, and clinical trust in

integrating genomics and AI into point-of-care decision-making. Haendel highlighted that clinical AI models inherit severe systemic biases due to lossy medical coding mappings, variable disease prevalence estimates, inconsistent disease terminologies across databases, and incomplete electronic health record data. To overcome these bottlenecks, she emphasised the need for transparent data provenance, portable cohort definitions, and robust evaluation frameworks. Illustrating how agentic AI and semantic guardrails (such as LinkML) can construct mechanism-based disease classifications, Haendel urged the GA4GH community to modernise governance and standardise evaluation metrics to build trustworthy, evidence-backed clinical tools.

Panel discussion

Recording

The panel explored how GA4GH can maximise its impact on AI implementation across healthcare and life sciences. Bilal Mateen advised GA4GH to focus on its unique niche amidst market noise, while Riu-Yao Zhang emphasised that GA4GH's greatest strength lies in standardising high-quality, international datasets, which are increasingly vital as public training data becomes exhausted. Tudor Groza and Melissa Haendel highlighted the need to structure domain knowledge, noting that ontologies and interoperable schemas serve as essential guardrails for AI models. Haendel reaffirmed that biocurators and robust medical ontologies remain indispensable for safe clinical reasoning, explaining that AI tools rely heavily on human-curated frameworks. Finally, the panel addressed broader ethical and environmental dimensions, discussing the nuances of managing lifetime consent for genomic data and noting that higher-quality data reduces compute requirements, ultimately minimising AI's environmental impact.

Panel: From sequencing to outcomes: defining and demonstrating value in genomic medicine

Session chair: Mélanie Courtot (Ontario Institute for Cancer Research / University of Toronto)

KEYNOTE PRESENTATION

Rob Annan (Genome Canada): "The future of genomic medicine: translating innovation into health outcomes"

Recording | Slides

Rob Annan, President and CEO of Genome Canada, detailed Genome Canada's strategy to unify the country's ten provincial healthcare jurisdictions into a coordinated national framework. Addressing cultural and institutional barriers to data sharing, the CA$200 million Canadian Precision Health Initiative (CPHI) will sequence 120,000 whole genomes using GA4GH standards, a mandatory

requirement, while honouring Indigenous data sovereignty. Annan stressed that technical hurdles are manageable, but long-term success hinges on building public trust and maintaining social license amidst post-pandemic skepticism and rising AI safety concerns. Ultimately, Canada's multi-year financial commitment to GA4GH reflects a deep dedication to international collaboration and robust, sovereign data governance.

Ilias Goranitis (University of Melbourne): “Determining and realising the economic value of genomic medicine”

Recording | Slides

Ilias Goranitis, Professor of Economics of Genomics and Precision Medicine at the University of Melbourne, discussed the evolution of economic evaluation in precision health, noting that as genomic applications broaden — from rare disease diagnosis to rapid newborn screening and polygenic scores — the economic mandate shifts from a single-test "evaluation problem" to a system-wide "optimisation problem." Highlighting tools such as the Genomic Utility Valuation Scale, Goranitis demonstrated how functional genomics, long-read sequencing, and automated reanalysis maximised diagnostic yield while optimising healthcare budgets. He stressed that while determining value is a measurement problem solvable through linked real-world data and standardised GA4GH outcome metrics, fully realising that value requires redesigning health systems to ensure equitable access and sustainable financing.

Indri Rooslamiati Supriadi (Center for Biomedical and Genomic Health and Clinical Research Center): “Advancing genomic medicine in a LMIC: Indonesia’s journey from genomes to health outcomes”

Recording | Slides

Indri Rooslamiati Supriadi, Director of the Center for Biomedical and Health Genomics of the Indonesian Ministry of Health, highlighted Indonesia’s initiative to embed genomic medicine into its national health strategy across a diverse population of 287 million people spanning 17,000 islands. Facing resource constraints typical of low- and middle-income countries, the Ministry of Health established the Biomedical and Genome Science Initiative (BGSI) to address major national disease priorities by integrating genomic data collection into existing public health programs, including primary care and national tuberculosis screening. Utilising GA4GH tools and federated frameworks to standardise data governance and access, BGSI demonstrated that implementing scalable, representative clinical genomics is feasible and vital for improving health outcomes in large, multi-ethnic populations.

Kenjiro Kosaki (Keio University): “Mobilising distributed rare disease knowledge through LLM-assisted extraction of interoperable variant-phenotype observation”

Recording | Slides

Kenjiro Kosaki, Professor at Keio University, presented Japan’s Initiative on Rare and Undiagnosed Diseases, which has analysed over 10,000 trios with a nearly 50% diagnostic yield, leading to national health insurance reimbursement for exome sequencing. To mobilise valuable non-English case reports across Asia, Kosaki proposed utilising local, privacy-preserving Large Language Models (LLMs) to perform optical character recognition and translation while extracting phenotype and variant data. To overcome the risk of text rendering errors in human-readable PDFs, Kosaki proposed using QR codes to securely preserve precise machine-readable variant representations, urging GA4GH to help integrate regional non-English patient-level data into the global genomic ecosystem.

Bimal Chaudhari (Nationwide Children’s Hospital): “Programmatic development to deliver rGS and genomic medicine”

Recording | Slides

Bimal Chaudhari, Principal Investigator at Nationwide Children’s Hospital, addressed the system-level failures in rare disease diagnosis, arguing that laboratory turnaround times are often dwarfed by clinical delays before testing is even ordered. In response, Chaudhari outlined Nationwide Children's Hospital's implementation of an Acute Care Genomics Consult Service, which proactively screens all neonatal intensive care unit (NICU) admissions. This structured, early-intervention workflow guarantees rapid genome sequencing within seven days and results within fourteen days. Chaudhari demonstrated that integrating dynamic phenotyping tools and standardised clinical pathways dramatically improved diagnostic speed and yielded cost savings.

Panel discussion

Recording

The panel addressed key challenges in quantifying and realising the value of genomic medicine within health systems globally. Panelists highlighted that the value of rapid genomic testing extends far beyond direct patient diagnosis, creating systemic ripple effects such as reduced ICU bed stays and shortened outpatient wait times. Maintaining political and community support also requires demonstrating real-world impact to taxpayers and decision-makers. The panelists underscored that establishing robust governance and data standards before scaling collection is crucial for building public trust and ensuring long-term program viability, particularly in low-resource contexts.

Plenary Day Two (30 September 2026)

Opening remarks

Recording

Peter Goodhand, CEO of GA4GH, welcomed attendees to day two of the Plenary meeting. He distinguished Plenary — focused on global impact and broader themes — from the upcoming Connect meeting, which involves hands-on collaboration on technical, regulatory, and ethical standards for interoperability. Outlining the day's themes, including embedding genomics in healthcare and population screening, he reflected on the powerful impact of day one's patient story and introduced the next family speaker to keep the community grounded in its core purpose.

Serene's story

Recording

Serene shared her family's decade-long diagnostic odyssey with her son, who was experiencing severe developmental delays. Initially treated broadly as a cerebral palsy patient without a clear diagnosis, her son was enrolled in therapy, early intervention, and non-invasive research programs at KK Women's and Children's Hospital before genomic sequencing identified his condition as PURA syndrome when he was nine. This diagnosis provided clarity, allowing them to transition to active condition management, and connected the family with international support networks such as the PURA Syndrome Foundation.

Panel: Embedding genomics into the healthcare system

Session chair: Alex Wagner (Nationwide Children's Hospital / Ohio State University)

KEYNOTE PRESENTATION

Eric Green (Illumina): "Delivering precision healthcare: embedding genomics into clinical practice"

Recording | Slides

In his presentation, Eric Green, Chief Medical Officer at Illumina, reflected on the field's 23-year evolution from the completion of the Human Genome Project to the routine integration of genomic medicine into clinical practice. Tracing his journey across academia, 15 years as Director of NHGRI, and his role at Illumina, Green highlighted the million-fold drop in sequencing costs — enabling human genomes to be sequenced in seconds rather than years. He pointed to five established pillars of genomic medicine — cancer, rare disease, prenatal testing, pharmacogenomics, and prevention —

while identifying newborn sequencing as the next major frontier. Acknowledging ongoing scientific, diagnostic, and complex societal hurdles such as health equity and data privacy, Green emphasised that frameworks such as GA4GH are vital to ensuring data is safely shared and used to advance global healthcare.

Emma Weisz (Melbourne Genomics Health Alliance): “Embedding genomics into routine general paediatric care”

Recording | Slides

In her presentation, Emma Weisz, a Paediatrician at Monash Health, discussed embedding genomics into routine general paediatric practice in Victoria, Australia. To address long wait times at clinical genetic services, the Australian government enabled paediatricians to order funded genomic tests. Weisz's team employed a "champion model of change" and co-designed step-by-step clinical resources and a dedicated liaison service. Recognising that education alone was insufficient, they created an experiential "paediatric genomics clinic" — pairing paediatricians with genetic counselors and geneticists to learn by doing. This approach achieved a 50% diagnostic yield, boosted paediatrician confidence to practice independently, and demonstrated the value of combining co-design, clinical champions, and hands-on learning to mainstream genomics.

Breana Cham (KK Women’s and Children’s Hospital): “Training and career pathways for genetic counsellors and the genomics workforce”

Recording | Slides

Breana Cham, Lead Genetic Counsellor at KK Women’s and Children’s Hospital, addressed the shortage of genetic counselors in Southeast Asia and focused on workforce development initiatives in Singapore. To meet increasing clinical demands, Cham and her colleagues established training roadmaps, competency frameworks, and entrustable professional activities (EPAs) to support genetic counselors across various career stages. Additionally, they integrated Genetic Counseling Associates to handle routine, administrative clinical tasks, allowing fully trained counselors to manage complex cases and operate counselor-led clinics. Through standardised competency domains and a planned Master of Genetic Counseling program, these strategies aim to optimise workforce efficiency, expand capacity, and ensure high standards of care across the healthcare system.

Catherine Procknow (Epic): “Embedding genomics into EHR software”

Recording | Slides

Catherine Procknow, Lead Software Developer at Epic, discussed how Epic integrates genomic data into electronic health records (EHRs) to drive precision medicine. Procknow emphasised that treating

genomics as discrete, actionable clinical data — rather than static PDF reports — enables automated point-of-care alerts for pharmacogenomics, cancer screening, and targeted therapies. By utilising HL7 standards and collaborating with GA4GH, Epic facilitates interoperable data exchange across systems. Furthermore, Procknow highlighted Epic's "Cosmos" database and "Look-Alikes" tool, which aggregates real-world patient data to connect clinicians treating similar patients, demonstrating how scalable infrastructure and shared standards can continually enhance patient outcomes.

Panel discussion

Recording

The panel addressed key strategies and challenges surrounding the global integration of genomics into routine clinical care. Panelists emphasised that due to wide variations across healthcare systems, making genomic sequencing routine requires generating clear evidence of improved clinical outcomes and cost-effectiveness. Speakers also noted that while asking targeted diagnostic questions is crucial in rare disease care, clinicians must also view patients holistically rather than defining them solely by their genetic data. Finally, the panelists agreed that overcoming clinical adoption barriers requires flexible training, continuous education, and seamless data communication between tertiary centres and primary care providers.

Panel: Genomic newborn screening: from research to routine care

Session chair: Ainsley Newson (University of Sydney)

KEYNOTE PRESENTATION

Robert Green (Mass General Brigham / Harvard Medical School): "Making genomic newborn screening a clinical reality"

Recording | Slides

In his talk, Robert Green, Professor of Medicine at Harvard Medical School, discussed advancing genomic newborn sequencing into routine clinical care through research studies such as BabySeq and BRIDGES. Green highlighted empirical findings from BabySeq, revealing that 11% of healthy newborns carry positive monogenic risk variants, with 35% displaying subtle, subclinical features of the underlying condition. Addressing common clinical concerns, Green noted that learning genomic risk does not increase parental anxiety, disrupt bonding, or trigger excessive downstream medical spending. He introduced the BRIDGES initiative, which integrates genomic screening directly into US public health workflows and frames newborn sequencing as an at-risk assessment model to build lifelong precision healthcare.

Ritu Jain (DEBRA Singapore / Nanyang Technological University)

Recording

Ritu Jain, Vice President of DEBRA Singapore and Assistant Chair and Senior Lecturer at Nanyang Technical University, shared a deeply personal perspective as a rare disease advocate and parent of a daughter with Epidermolysis Bullosa Simplex. She highlighted how genomic screening can drastically shorten the diagnostic odyssey for rare diseases, enabling timely clinical interventions, early monitoring, and informed family planning. However, Jain cautioned against expanding screening without robust safeguards, warning that genetic diagnoses can carry severe cultural stigma, financial burden, and insurance discrimination — factors that led her family to forgo a formal genetic test. Ultimately, she called for placing families and patient perspectives at the centre of national genomic programme planning.

Richard Scott (Genomics England): “Genomic newborn sequencing in England”

Recording | Slides

Richard Scott, Chief Executive Officer of Genomics England, presented the Generation Study, a major initiative evaluating whole genome sequencing for newborns embedded within the UK's National Health Service (NHS). Seeking to recruit 100,000 babies across England, the study screens for over 200 early-onset, actionable rare conditions while contributing data to the National Genomic Research Library to drive ongoing discovery. Scott emphasised that establishing a continuous, evidence-generating system is essential to expanding actionable gene lists over time. Ultimately, the study aims to assess clinical utility, cost-effectiveness, and public acceptability, informing the UK's strategic goal of offering genomic sequencing to all newborns by 2035.

Sebastian Lunke (Murdoch Children’s Research Institute): “Genomic newborn screening: lessons from BabyScreen+”

Recording | Slides

Sebastian Lunke, Head of the Division of Genetics and Genomics at the Victorian Clinical Genetics Services, presented findings from Australia's Babyscreen+ pilot study, which performed clinically accredited whole genome sequencing on 1,000 newborns in Victoria for over 600 severe, early-onset conditions. Integrated with standard dry blood spot testing and utilising prenatal online consent, the study achieved an average turnaround time of under 14 days and a 1.6% screen-positive rate. Notably, early detection enabled a life-saving bone marrow transplant for an infant with familial hemophagocytic lymphohistiocytosis (HLH). Participants overwhelmingly supported public, equitable access to genomic screening. Lunke concluded that clear public roadmaps and larger implementation studies are essential to prevent commercialisation from causing healthcare inequities.

Panel discussion

Recording

The panelists addressed key implementation and ethical questions regarding genomic newborn screening (NBS), noting that despite lingering concerns, current legal frameworks — such as US policy coverage and statutory protections in the UK and Australia — largely mitigate genetic discrimination. Broader public support for genomic NBS remains high, though slightly more cautious than among study volunteers, with strong emphasis placed on data privacy and long-term storage. Panelists also discussed that reproductive and newborn screening serve distinct, complementary purposes. Preconception testing aids family planning, whereas genomic newborn screening optimises population health, accelerates treatment development, and enables continuous, lifelong re-analysis of patient data.

Panel: Implementing population genomics: policy, regulation, and sustainable delivery

Session chair: Sue Hill (NHS England)

KEYNOTE PRESENTATION

Ee Shien Tan (Precision Health Research, Singapore [PRECISE]): “The future of population genomics: from evidence to implementation”

Recording | Slides

Ee Shien Tan, Chief Innovation Officer of PRECISE, presented Singapore's National Precision Medicine roadmap, focusing on embedding genomics into population health. Facing a rapidly aging society and rising chronic disease, Singapore progressed from a 10,000-person foundational cohort (Phase 1) to a deeply phenotyped 100,000-person cohort (Phase 2, SG100K). Tan detailed the clinical implementation of Familial Hypercholesterolemia (FH) screening within primary care, which leveraged local data, cost-effectiveness analyses, and simplified referral thresholds to improve early diagnosis and cascade testing. Moving into Phase 3, which aims to sequence ~10% of Singapore's population via hospital-based hubs, Tan introduced the "sequence once, analyse repeatedly" model. She emphasised that expanding into proactive screening, pharmacogenomics, and polygenic risk scores requires robust governance, dynamic consent, equitable access, and public trust.

Surakameth Mahasirimongkol (Department of Medical Sciences, Ministry of Public Health): “Genomics Thailand Phase 2: implementing genomic medicine in Thailand health system”

Recording | Slides

Surakameth Mahasirimongkol, Director of the Information and Communication Technology Center at the Ministry of Public Health of Thailand, discussed Genomics Thailand, a national initiative advancing precision medicine to help extend Thailand's life expectancy to 85. Building on Thailand's Universal Health Coverage, the program began by integrating pharmacogenomic screening to prevent severe adverse drug reactions. The initial $55 million, five-year phase successfully established a reference database of 50,000 whole genomes, achieving a 37% diagnostic yield in rare diseases and expanding free *BRCA1/2* testing for high-risk breast cancer patients. Supported by an expanding workforce, biobank, and data infrastructure, Thailand is now planning Phase 2 to sequence 200,000 additional genomes and broaden clinical implementation nationwide.

Wadha A. Al-Muftah (Qatar Precision Health Institute): “Beyond sequencing: what Qatar is learning from turning a national genome program into national health policy”

Recording | Slides

In her talk, Wadha A. Al-Muftah, Chief Medical Officer of Qatar Precision Health institute, discussed transitioning Qatar’s population genomics program from whole-genome sequencing (35,000 adult genomes) to clinical implementation, public health policy, and sustainable funding. With 5.6% of participants harboring actionable ACMG variants, bridging the gap between research discovery and clinical care requires explicit responsibilities, accredited confirmatory testing, clear referral pathways, and population-wide equity. To scale responsibly — demonstrated through high-risk breast cancer variants and pharmacogenomic models like *CYP2C19* guided clopidogrel prescribing — health systems must fund entire care pathways rather than just initial testing. Ultimately, success relies on population-specific evidence, continuous re-interpretation, participant re-consent, and global knowledge-sharing to maintain public trust.

Michael Cook (Our Future Health): “Our Future Health: a world-leading platform for discovery, aetiological and translational research”

Recording | Slides

Michael Cook, Chief Science Officer of Our Future Health, discussed how Our Future Health, a major non-profit UK research program, is recruiting 5 million adults to accelerate translational clinical research and disease prevention. The multi-ethnic cohort combines baseline surveys, biobanked blood, multi-ethnic array genotyping, and broad electronic health record linkages. With nearly 3 million consented participants and over 1.6 million genotyped datasets generated to date, this research

platform offers unprecedented scale for studying polygenic risk, pharmacogenomics, and complex disease interactions. Additionally, Our Future Health provides a rapid Clinical Research Recruitment Service that researchers can use to selectively invite participants into their own observational studies and clinical trials, a service which is also being used in collaboration with the NHS as part of an evidence-based strategic shift toward proactive, preventive healthcare.

Panel discussion

Recording

This panel discussion focused on translating population genomics into clinical practice, expanding multi-omic research, and adopting global standards. Panelists highlighted plans to return clinical data to participants by integrating genomic insights into unified primary care records, with Singapore and Thailand selecting clinical pilots based on population prevalence, cost-effectiveness, and urgent unmet needs such as rare diseases and cancer. Moving forward, the programmes aim to expand beyond DNA into proteomics, single-cell, and long-read sequencing as sustainable funding allows. Finally, all participating projects reaffirmed their commitment to adopting or aligning with GA4GH and ISO standards, uniting around shared goals such as sequencing every citizen and extending healthy life expectancies globally.

Evening talk show: What will genomic medicine look like in 2035

Recording

In the closing panel discussion on "Genomic Medicine in 2035," four experts shared their visions and challenges for the decade ahead:

- Valis Tanapima, a Bunun Indigenous physician and Chief of the Department of Emergency Medicine at Cheng Hsin General Hospital, Taiwan, emphasised the vital need for Indigenous data sovereignty and meaningful governance, warning that minority populations risk being erased or misclassified under broad categories in global datasets.
- Jantina de Vries, Director of the Ethics Lab and Professor of Medicine at the University of Cape Town, stressed that the key obstacles to equitable genomic medicine are ethical and political rather than technical, pointing out global disparities such as the lack of basic newborn screening and high costs of gene therapies in resource-limited settings.
- Yuki Okada, Professor at Osaka University, outlined computational advances in risk prediction while highlighting the critical responsibility researchers hold in accurately communicating probabilistic uncertainty and limitations to patients and society.
- Lea Starita, Associate Professor at the University of Washington, discussed bridging the interpretation gap by generating comprehensive, ancestry-agnostic functional data to resolve

variants of uncertain significance (VUS) at scale, maximising the clinical utility of population sequencing.

Connect Day One (1 October 2026)

Connect welcome

Recording | Agenda | Slides

The GA4GH Connect Welcome session opened with a technical update from Sasha Siegel, Chief Product Officer, on GA4GH's shift from standards development toward adoption and implementation. Siegel presented updates from the GA4GH Technical Team. New services include a Reference Cloud (with conformance verification to follow), a unified GA4GH Registry, a DOI service for attribution, and an Analytics Dashboard to track uptake, which currently shows 40 registered API implementations and over 1,284 publications mentioning GA4GH. The revised Product Development and Approval Process will require starter kits, conformance tests, and evidence of use. It will also introduce reviews by the Regulatory & Ethics, Data Security, and Artificial Intelligence (AI) Work Streams, and by the Technical Alignment Subcommittee (TASC), which will assess interoperability using its new working definition.

Following Siegel's opening presentation, leaders from each Work Stream and Community of Interest provided updates about the work of their group.

- **Discovery** reported progress on the Experiments Metadata Checklist and Beacon (with v3.0 under development) and highlighted the Variant Level Matching GIF project.
- **Genomic Knowledge Standards** (GKS) welcomed three new product teams (Whole Genome Sequencing Quality Control Standards, File Formats, and Future of VCF), consolidated Variation Representation Specification, Variant Annotation Specification, and Categorial Variation Representation Specification into Genomic Knowledge Models (GKM) with a new starter kit, and launched a new Asia-Pacific Think Tank series.
- **Clinical & Phenotypic Data Capture** (Clin/Pheno) reported on Phenopackets, the Pedigree standard and KIN ontology, and the Human Exposome group's work on standardising place-based environmental exposures.
- **Data Use & Researcher Identity** (DURI) is expanding the Data Use Ontology (DUO) into a broader ecosystem for data access.
- **Data Security** welcomed Crypt4GH into its product portfolio, shared updates on Passport/AAI v2.0, and discussed plans to explore verifiable credentials and agent passports.

- **Regulatory & Ethics** (REWS) highlighted two new products, Position Statement on Genetic Discrimination and an ethical, legal, and social implications (ELSI) decision tool for newborn screening, and outlined current priorities including AI governance, Asian genomic data sharing, policy uptake metrics, and pathogen data sharing.
- **Federated Analysis** (formerly Cloud) now oversees eight APIs, including htsget, RNAget, and refget, and is moving toward an initiative-based model in collaboration with the GA4GH Implementation Forum (GIF).
- **Artificial Intelligence** (AI) introduced a new structure built on three foundational product categories (governance, provenance, and benchmarking), which will underpin applied tools such as an Agentic Harness for natural-language interaction with GA4GH standards and an AI Registry.

Among the Communities of Interest, **Rare Disease** is working toward a standardised head-to-toe phenotype that may soon enter product development, while **Cancer** has engaged with structural variant aggregation efforts and the OncoBeacon project. Both communities are likely to move action-oriented projects into the Work Streams and will hold stakeholder meetings this fall. **Infectious Disease**, a collaboration between GA4GH and PHA4GE, reported on a pathogen genomics domain analysis model to guide future FHIR implementation guides, the identification of 44 data-sharing repositories for detailed assessment, and a pilot applying Phenopackets and PHA4GE standards to typhoid surveillance data from Nepal. **Neuroscience**, now around 40 members, is compiling a state-of-the-field synthesis, collaborating on cohort discovery tools, and seeking members with genomics expertise. Finally, the **Clinical Genomics Laboratory Community** (CGLC), formed in partnership with ClinGen and now over 600 members strong, is developing a data model with GKS to support forthcoming sequence variant classification standards.

Mondo across GA4GH: mapping interoperability with Beacon, VRS/VA, DUO, and FHIR

Recording | Agenda | Slides

Mondo is GA4GH's first formally endorsed externally-developed standard, coordinated by the Monarch Initiative Driver Project. This working session mapped how Mondo interoperates across the GA4GH ecosystem and grows the community that sustains it. Mondo bridges OMIM's Mendelian knowledge toward clinical classifications (ICD-11) on one side, and serves as a disease-concept resource. A Mondo interoperability map was presented to link care and research, incorporating the Variation Representation Specification (VRS), Variant Annotation Specification (VA-Spec), the Data Use Ontology (DUO), Beacon v2.0, and Phenopackets, in partnership with HL7 and OHDSI to support FHIR and OMOP terminology services. Using a coverage-cascade method, a preliminary interoperability map

was presented across these standards, covering where Mondo is already adopted, where Mondo has formalised the knowledge but the standard has not yet bound to it, and where the concept exists only in other upstream sources and Driver Projects, such as OMIM or EGA.

Key takeaways

- Mondo can anchor disease concepts across the patient journey. Following one synthetic rare disease case from clinic note to VRS, Categorical Variation Representation Specification (Cat-VRS), VA-Spec, Phenopackets, DUO, Beacon, FHIR and ICD-11 showed where a Mondo term can already sit (the Cat-VRS condition, the VA-Spec object condition, Phenopackets diagnoses, Beacon filtering terms, DUO disease-specific permissions). It also showed how seldom the research and clinical sides actually exchange data today.
- Cat-VRS and Mondo complement each other: “put Cat-VRS in the disease, not the disease in Cat-VRS”. Versioned Cat-VRS variant sets could help define gene- or mechanism-specific Mondo entities, such as SCN2A gain- versus loss-of-function epilepsies, in the same way a code set defines a condition. Monarch’s new disease-mechanisms resource (DisMech) could connect the two.
- Mapping and translation is the practical gap. Beacon networks need an explicit translation layer between terminologies. Mondo’s SSSOM mappings, which come with full provenance in every release, and a Node Normalizer-style service could provide it. DUO needs Mondo ‘slims’ and a workable, computable meaning of “related diseases” in consent forms.

Priority action items

- Use the new #mondo channel on GA4GH Slack for questions, ideas and follow-up from this session.
- Prototype a Mondo/SSSOM-based term-translation layer for Beacon that runs on the query side or at each implementation, possibly with a dashboard of terms used across Beacons (Mondo team with Michael Baudis and Jordi Rambla).
- Explore Cat-VRS–Mondo alignment (Monarch with GKS/Cat-VRS leads, e.g. Daniel Puthawala). Use versioned Cat-VRS sets as definitional input for Mondo entities, and work out how Mondo IDs should be versioned or obsoleted when those sets change.
- Work with DUO leads (Jonathan Lawson, Gregory Rushton) on Mondo slims for matching data-use permissions to research purposes. Consider a renewed push on machine-readable consent so forms avoid terms like “and related diseases” or “other”.
- Improve cancer coverage in Mondo (NCIt and ICD-O alignment, susceptibility records and context). Volunteers include Hindrik Kerstens and Michael Baudis.

- Consolidate session feedback into the interoperability map, circulate a short recommendations document and start the Monarch-led manuscript, with contribution-based authorship.

GIF Project kick-off: approval into action

Recording | Agenda | Slides

The GA4GH Implementation Forum (GIF) session opened with an overview of its renewed focus on supporting projects that implement GA4GH standards in real-world settings. GIF leadership will hold quarterly project check-ins to identify challenges, facilitate connections, and ensure implementation feedback reaches the relevant Work Streams. Natalie Twine, Mohamed Zahir Alimohamed, and Mónica Muñoz Torres were announced as new GIF Co-Leads, joining Brian O'Connor and Wafaa Rashed, expanding capacity to support the growing project portfolio.

Five new projects illustrated several approaches to testing interoperability. The Federated Data Sharing Between NIH Systems project will use the Data Repository Service (DRS) and Passports through NIH's Researcher Auth Service to support multi-ancestry GWAS across four platforms, producing a reusable example for future cross-platform analyses. The Research Agents Demonstration Using GA4GH APIs project will explore how AI agents can discover data and workflows, access services, and execute analyses using GA4GH APIs, in collaboration with the technical team and AI Work Stream. Two federated learning experiments will test distributed variant pathogenicity prediction and multi-ancestry fine-mapping, using synthetic data to assess both interoperability and whether federated analyses recover the expected scientific results.

Projects in Tanzania and Pakistan highlighted opportunities to incorporate standards while developing local genomic infrastructure. Tanzania's rare disease project will focus on implementing Phenopackets, validating case records, and establishing technical and governance readiness for Beacon v2.0 and potential participation in variant-level matching. Participants recommended engaging with the variant-level matching group early, allowing these activities to develop in parallel. Pakistan's project will begin with a two-centre pilot for hematological disorders, using Beacon v2.0, Phenopackets, DUO, and Passports to enable discovery while retaining local data stewardship, with a longer-term ambition to expand nationally.

Updates from existing projects demonstrated practical progress and areas requiring further support. The Federated Variant-Level Matching network now connects four sites and has supported two gene discoveries, with additional sites approved to join and Phenopackets support developed but not yet deployed across the network. The Federated Imputation Platform is running in production and connects services in Cape Town, Michigan, and TOPMed, allowing users to submit analyses across multiple services and reference panels through one interface. TRE Open Suite reported security vulnerabilities identified through its confidential computing work and outlined efforts with other standards organisations to advance safer approaches.

Across the presentations, recurring needs included implementation guidance, participating sites, governance support, and mechanisms for communicating gaps in standards and implementations. Several projects share technologies and objectives, creating opportunities to reuse tools and learn from one another. GIF co-leads agreed to follow up by mapping project needs and making connections across projects, Work Streams, and the wider implementation community.

Key takeaways

- GIF's core role is to facilitate collaboration and feedback between implementers and standards developers.
- Shared use of Beacon, Phenopackets, DRS, Passports, and execution APIs creates opportunities to reuse approaches across projects.
- Success requires both technical interoperability and workable governance, access, and consent arrangements.
- Projects should validate scientific correctness and reproducibility alongside successful workflow execution.

Priority action items

- Establish quarterly project check-ins and map shared needs to relevant projects, Work Streams, and implementers.
- Publish the new project profiles and contact details on the website.
- Project teams: Recruit implementation partners and test sites, and document API gaps and proposed extensions for Work Stream feedback.

DURI data governance products

Recording | Agenda | Slides

Controlled data access does not scale: manual review of Data Access Requests, repetitive negotiation of data access agreements, and error-prone consent translation create chokepoints for repositories. The GA4GH Data Use & Researcher Identities (DURI) Work Stream is advancing four products toward computable governance, and this working session invited parties invested in computable governance to shape the specifics of each of the following:

Passports — new visa types encoding data-residency policy, building on the data passports framework described by Jonathan Lawson and Tom Conner (https://dx.doi.org/10.2139/ssrn.5372874).

DUO — updates to a streamlined Data Use Ontology, extending its proven machine-readable consent vocabulary (>3,000 dbGaP datasets mapped with 93% parity; adopted by EGA).

DAAO — the new Data Access Agreement Ontology, built on GA4GH Model DAA clauses.

DARO — the new Data Access Request Ontology, derived from a systematic review of DAR forms.

The session aimed to refine proposed solutions for computable data governance through hands-on collaboration on these four DURI products.

Key takeaways

- DUO was recognised as an established foundation for representing computable data use permissions, while DARO and DAAO were discussed as complementary proposals aimed at standardising key components of the data access request and agreement process to improve interoperability and reduce friction in data sharing.
- Researcher Passports and Data Passports were discussed as complementary approaches. Researcher Passports focus on describing who a researcher is and what that researcher is authorised to do, while Data Passports describe governance conditions associated with a dataset.
- Participants emphasised the importance of making governance information consistent, interoperable, and computable, rather than attempting to encode every possible governance scenario or automate all governance decisions.
- There was strong interest in aligning DURI products with existing standards and initiatives, including ODRL, machine-readable consent guidance, verifiable credentials, and related governance frameworks where appropriate.
- Participants noted the challenges of achieving global standardisation of data access processes given differing institutional, legal, and jurisdictional requirements. Data access requests and data access agreements were identified as areas where greater structure and machine-readable representations could help reduce barriers to data sharing.

Priority action items

- Further develop and socialise the tri-ontology proposal around the DARO, DAAO, and DUO to guide upcoming iterations.
- Explore opportunities to align these efforts with existing standards such as ODRL, machine-readable consent guidance, verifiable credentials, and other governance-related frameworks.
- Gather additional community feedback on governance requirements that should be represented, particularly in relation to data access decision-making, policy enforcement, trusted research environments, and federated governance models.

OncoBeacon Network: a practical application of standard Beacon implementation software in (paediatric) cancer genomics

Recording | Agenda | Slides

The GA4GH Beacon standard provides a lightweight data discovery protocol that enables biomedical institutes and genomic resource providers to make their data globally discoverable. A collection of nodes implementing the Beacon API can be utilised to create a Beacon network which can empower researchers to compose research cohorts from multiple centres, e.g. to perform genomic research at scale.

While the default Beacon data model includes several properties relevant for representation of cancer related data, a practical implementation in the Princess Máxima Center found the need to implement some modifications and to specify the use for some general properties, according to the needs of the oncology driven use case, towards a dedicated OncoBeacon specification.

In the interactive, hands-on session, speakers demonstrated how the application of the OncoBeacon specification implements and modifies the standard Beacon model and how the use of standard API and network software has led to a practical Beacon implementation in paediatric oncology.

This session brought together data holders in other (paediatric) oncology centres with whom a collaboration can be established and included presentations of other Beacon resources with relevance to oncogenomic use cases. It focused on the current state of the Beacon data model, its capabilities for querying and representing cancer related data and showcased how modifications developed for the OncoBeacon specification improve utility of the Beacon protocol in a concrete oncogenomic use case.

Key takeaways

- Beacon already works in real-world paediatric oncology, without changing the schema. OncoBeacon at the Princess Máxima Center took a stricter approach. The team listed the attributes they needed, placed them in the Beacon model (using generic “measurements” for things like tumour mutational burden, tumour purity and molecular subtype), and bound both attributes and values to NCIt terms. A proof-of-concept Dutch network now returns tiered responses: Boolean for industry, counts for researchers, record-level for trusted healthcare users. Harmonising data and choosing ontology terms was the hardest part; the technical deployment was smooth.
- Longitudinal events are the most important model gap. Cancer patients can have several primary tumours, relapses, and treatment episodes, and Beacon cannot yet link samples to events over time. Phenopackets “medical actions” offer a route, and alignment with Phenopackets is being assessed for a Beacon v2.0 extension or v3.0. Participants also cautioned against overloading a simple model.
- Networks need shared query conventions and community connections. Participants called for a catalogue of recommended query terms (sex, age groups, disease, gene-level events like TP53

deletion) that every implementation resolves consistently. There was strong interest in linking paediatric cancer initiatives (OncoBeacon, Zero Childhood Cancer, PCGL/Marathon of Hope, ITCC) and in dataset lineage to detect duplicated cohorts.

Priority action items

- Follow up on suggestions to improve the presented changes.
- Develop the proposed changes and create implementation proposals.
- Set up a group (possibly a new Beacon Scout) on recommended query terms and prototype queries, and test them across different implementations.
- Assess adding events or episodes to Beacon (aligned with Phenopackets medical actions) as a non-breaking v2.0 extension before year-end, or otherwise for Beacon v3.0.
- Explore connecting the Zero Childhood Cancer Beacon (Children's Cancer Institute Australia) and PCGL/iCHANGE with the OncoBeacon network, and approach international paediatric cancer initiatives (e.g. ITCC, INSERM–NCI) about a pilot.
- Start collecting implementation metrics (datasets discovered through Beacon, effort, cost, and lessons learned) to demonstrate value.
- Develop the OncoBeacon proposals into implementation proposals for the Beacon specification, and follow up on the suggestions raised.
- Revisit dataset-level provenance and lineage in Beacon to help identify the same data re-shared across resources.

Clinical implementation of genomic discoveries in LMIC

Recording | Agenda | Slides

This session examines the persistent translational gap between genomic discovery and clinical deployment in low- and middle-income countries (LMICs), where infrastructure, regulatory pathways, and population-specific reference data often lag behind high-income settings. Drawing on real-world experience delivering pharmacogenomic and predictive genomic testing across diverse Southeast Asian populations, the session will discuss how GA4GH standards — including the Variation Representation Specification (VRS) for computable variant and genotype representation and emerging PGx data standardisation efforts — can be adapted to support interoperable, equitable clinical decision support where EHR infrastructure and reference databases are underdeveloped or Western-population-biased.

Key takeaways

- There are persistent challenges and cultural sensitivities when translating GA4GH standards into LMIC contexts, some of these are unique to the jurisdictions and some of these are wider challenges across the globe (e.g. genomic familiarity and understanding).
- Evidently, there are different value systems across jurisdictions and thus, communities may wish for different forms of benefits for participating in genomic programmes that are appropriate to their socio-cultural context (e.g. familial ancestry).
- Building sustainable, long-term programmes are crucial not only for the broadening of genomic research but also to maintain sustainable demonstrations of trustworthy behaviour with participants.

Priority action items

- Explore the potential of a GA4GH Implementation Forum (GIF) specifically on LMIC translation of GA4GH products.
- Explore ways of building wider conversation between LMIC jurisdictions to share challenges and solutions that have been found.

Building -omics bridges for precision health

Recording | Agenda | Slides

This session examined how GA4GH standards and infrastructure could support the integration of proteomics, microbiome, and spatial -omics data with genomics. Three introductory talks outlined the scientific opportunities and technical challenges, followed by group discussions on clinical metadata, discovery and federated querying, and potential precision health applications. The overall aim was to identify gaps and practical opportunities for collaboration with communities already developing standards for these modalities.

The proteomics presentation highlighted the contrast between established open-data infrastructure and the growing need to manage sensitive human proteomics data under controlled access. Different proteomics data types present different potential identification risks, requiring proportionate governance and better understanding among policy specialists and data access committees. Early work extending the Federated EGA metadata model to support proteomics was presented as a starting point. Potential connections with GA4GH included protein-oriented discovery through Beacon, linking biomarkers to clinical phenotypes through Phenopackets, and closer collaboration with the Proteomics Standards Initiative.

The microbiome and spatial -omics talks illustrated the complexity of extending genomic approaches to other data types. Microbiome analysis requires consideration of species, functional genes, assay

methods, reference databases, and changes over time; residual human DNA in samples also raises privacy concerns. Spatial -omics combines molecular measurements with tissue location, creating opportunities to understand tumour biology, cell interactions, and potential therapeutic targets. Its large datasets and complex annotations introduce substantial storage, access, and computational requirements.

The breakout discussions produced several concrete areas for follow-up. Participants outlined possible Beacon queries involving microbial species, antimicrobial resistance, cell populations, and gene-expression patterns, while noting challenges with changing cell labels and comparable measurements. The clinical metadata group emphasised the need to explain why a test was ordered, the sample source, and how it was performed so clinicians can judge its relevance. Another group identified cancer biomarkers and dementia prevention as potential multi-omics use cases, but highlighted the lack of common reference materials and calibrators as a major barrier to comparing results across platforms. Organisers committed to consolidating the discussions in an after Connect report and passing the query suggestions to the Beacon team.

Key takeaways

- Multi-omics integration requires modality-specific metadata, discovery approaches, and appropriate access controls.
- Existing specialist standards communities provide an important foundation for collaboration.
- Common reference materials and calibration are needed to improve cross-platform comparability.
- Clinical usefulness depends on clearly communicating a test's purpose, provenance, and limitations.

Priority action items

- Share the proposed microbiome and spatial -omics queries with the Beacon team.
- Explore an initiative on common -omics reference materials and calibrators.
- Further develop clinical metadata requirements and potential pilots in cancer biomarkers or dementia prevention.

Agentic science powered by GA4GH standards: an update on AI tools and use cases

Recording | Agenda | Slides

The AI Work Stream recently crafted a strategy and priority areas for the next 12 months of development across three Foundational Pillars (Governance, Provenance, and Benchmarking),

following GA4GH community consultations. Originating from the 35 use cases and projects identified at April Connect 2026 in Montreal. These use cases and projects fit naturally into 7 strategic themes that will set the course for the AI Work Stream's future development activities. We aimed to facilitate discussions on 8 newly proposed AI standards in this session. We also described ownership and accountability for these themes across the wider GA4GH community.

Key takeaways

- AI Accountability Framework: GA4GH is developing standards to hold AI agents accountable like we would humans, with a focus on authorisation, actions, and performance tracking.
- Agentic Harness Prototype: Working prototype enables researchers to interact with GA4GH standards via natural language; reduces complex workflows from days to minutes.
- AI Registry: continue to grow our catalogue of biomedical AI artefacts (models, skills, MCP servers) with provenance and benchmarking metadata to increase transparency and trust.
- Provenance Tracking: Agents "travel" across systems — comprehensive logging of delegation chains, prompts, and decisions are needed to determine accountability.
- Benchmarking Gap: AI benchmarking is fundamentally different from traditional tools due to non-deterministic outputs and lack of ground truth in novel research questions.

Priority action items

- AI product category development; coordinate governance, provenance, benchmarking, and harness work.
- Refine agentic harness and AI registry prototypes; support real-world testing through GA4GH Implementation Forum (GIF) projects.
- Coordinate governance and liability discussion and contribute to provenance and benchmarking work
- Encourage participation in the AI Work Stream's three subgroups; collect more AI artefacts for the AI Registry; solicit feedback on specifications; and define domain-specific benchmarking standards.
- Delivery target: draft specifications and products within 6 to 12 months, targeting the April Connect 2027 meeting.

AI governance in genomics: adapting global frameworks to regional and population-level practice

Recording | Agenda | Slides

This session explored the opportunities and challenges of AI risk governance in genomics and health, bringing together perspectives on regulation, representation, privacy, consent, and benefit sharing. Speakers examined the growing adoption of risk-based AI governance frameworks across jurisdictions, highlighting both their flexibility and their limitations, particularly in relation to power asymmetries, stakeholder participation, and the unique collective risks associated with genomic data. Discussion also focused on the role of federated multi-omic AI models and the need to ensure that underrepresented populations can contribute in ways that are both privacy-preserving and genuinely meaningful, with attention to attribution, consent, withdrawal, representation, and reciprocal benefit. A complementary perspective emphasised the importance of balancing governance with innovation and avoiding unnecessary barriers to the clinical and research benefits of genomic AI. Following these presentations, participants engaged in a hands-on workshop using a genomic AI scenario to identify and assess risks across eight categories, highlighting how risks often overlap and how context, uncertainty, power dynamics, and stakeholder perspectives can shape perceptions of risk probability and severity.

Key takeaways

- Risk-based governance is an increasingly influential approach to AI oversight, but its effectiveness depends on context. How risks are identified, assessed, and managed varies across legal frameworks, use cases, and affected populations.
- Genomic AI risks extend beyond individual-level harms. Risks may affect families, communities, and populations, raising questions about collective interests, data sovereignty, trust, and representation.
- Who defines and assesses risk matters. Existing governance processes often place responsibility on developers and institutions, while affected individuals and communities may have limited influence over which risks are recognised and how they are evaluated.
- Representation is both an equity and performance issue. Underrepresentation in genomic and multi-omic datasets can limit the performance and generalisability of AI models, highlighting the need for both inclusive data practices and appropriate governance mechanisms.
- Several governance tensions remain unresolved. Discussions explored trade-offs involving privacy and attribution, consent and inclusion, withdrawal and model persistence, and benefit sharing and re-identification risk. These issues cannot be addressed through technical solutions alone.
- The workshop exercise demonstrated that risks are interconnected and shaped by perspective. Risks frequently cut across traditional categories, while assessments of likelihood and severity often vary depending on available evidence, expertise, context, and lived experience.

Priority action items

- The Generative AI REWS group will review and synthesise the findings regarding the risks, assessments, observations that were captured during the interactive exercise, and share these outputs with interested participants. Outcomes from the session will help inform the next phase of GA4GH discussions on AI risk governance in genomics and health, including considerations related to representation, participation, and context-sensitive risk assessment.
- Consolidate workshop outputs into a stakeholder map, preliminary risk checklist, and regional-adaptation criteria to guide the next phase of tool development.
- Propose a working group to continue refining the draft community principles and the tool's design criteria.
- Community members are encouraged to share additional feedback on the questions and priorities for further exploration, using the link here.
- Participants interested in continuing these discussions are invited to engage in future activities and potential group efforts emerging from this initiative. For more information, please contact Prof. Ma'n Zawati or rews-coordinator@ga4gh.org.

Measuring the impact of the Regulatory & Ethics Work Stream through nuanced indicators

Recording | Agenda | Slides

Building on the participatory discussion at GA4GH April Connect 2026 in Montreal, this session moved from identifying impact categories to selecting metrics derived from the most promising indicators.

This session invited participants to prioritise elements that will be featured in a GA4GH Regulatory & Ethics Work Stream (REWS) performance metrics policy resource. Participants helped flesh out selected metrics, clarified potential data sources, and identified which indicators could be incorporated into a GA4GH metrics dashboard versus those requiring separate qualitative evaluation.

Key takeaways

- Participants voted on the most useful categories, which wereknowledge production, Cross-Work-Stream integration, and real-world implementation.
- The main missing categories that were raised were equity metrics and public health implementation.

Priority action items

- The feedback from the session will be integrated into the suggested indicators and will be circulated for open comment.
- Further updates from the group and any potential meetings will be shared.

Connect Day Two (2 October 2026)

Bridging regulation and technology: genomic data sharing across Asia

Recording | Agenda | Slides

This session explored the regulatory, ethical, and practical dimensions of genomic data sharing across several Asian jurisdictions, featuring presentations from experts across Taiwan, Hong Kong, Singapore, South Korea, Japan, and mainland China. Discussions highlighted comparative overviews of legal and governance frameworks, common themes and differences in privacy and cross-border sharing, and practical challenges regarding genomic data identifiability, inclusion, and responsible international collaboration.

Key takeaways

- Regulatory approaches to genomic data sharing differ across jurisdictions, reflecting varying priorities related to privacy, ethics, data sovereignty, biosecurity, and research collaboration.
- Trusted research environments (TREs) and other secure-access models are emerging as important mechanisms for enabling collaboration while maintaining appropriate governance and oversight.
- Re-identification risk remains a complex and evolving challenge, with participants noting that risk is highly context-dependent and influenced by population characteristics, data type, and intended use.
- Participants emphasised the importance of inclusion, both through broader representation of additional jurisdictions and through meaningful engagement with underrepresented populations, including Indigenous communities.
- Despite legal, technical, and governance challenges, participants agreed that responsible genomic data sharing is essential to advancing research, improving patient outcomes, and supporting future innovations in genomic medicine.

Priority action items

- Finalise and publish a comparative manuscript examining genomic data sharing regulations across the participating jurisdictions.
- Analyse feedback and survey responses gathered during the session to identify practical challenges and opportunities for genomic data sharing.
- Develop a future white paper focused on implementation considerations, real-world data-sharing practices, and governance needs.
- Continue community engagement through the project's Slack channel and broader collaboration activities.
- Expand participation and explore opportunities for future partnerships, funding, and collaborative research initiatives.

Bring your discovery problem: solved, adaptable, or missing?

Recording | Agenda | Slides

In this session, participants were encouraged to bring a data discovery problem from their own project, with the goal of mapping it against existing GA4GH standards, including Beacon and the Experiments Metadata Checklist. The group went over use cases from the Genomic Data Infrastructure project and Genomics Thailand.

Key takeaways

- Beacon v2.0 is being wrapped up and the focus is moving to v3.0. Before the end of the year, the team plans to move the repository into the GA4GH GitHub organisation, consolidate to v2.2, align with Phenopackets, and update Service Info. The proposed v3.0 scope covers aggregated responses, an OR operator with explicit interpretation in the response, Variation Representation Specification (VRS) v2.0 alignment, RNAget v2.0 integration, and schema and documentation improvements. Tentative items include pathogens, more metadata types, and generating OpenAPI from configuration.
- The Experiments Metadata Checklist aims to connect to existing standards rather than duplicate them. A LinkML formalisation is underway, with a route into the European Nucleotide Archive (ENA) and International Nucleotide Sequence Database Collaboration (INSDC) submission requirements. Participants asked for wider coverage beyond sequencing (arrays and other omics), higher-level instrument categories to avoid an endless catch-up game, and explicit mappings to RO-Crate and MINSEQE.
- Flexibility versus prescription remains the central tension. Communities need extensions (e.g. pathogens, allele frequencies, transcriptomics). One option is standardised, versioned mappings

and schemas outside of the core specification that implementations can opt into. Implementers also want to describe dataset metadata once and expose it both as Health DCAT-AP and through Beacon, and there was a renewed call for shared query terms.

Priority action items

- The Experiments Metadata Checklist team aims to map the dictionary to RO-Crate and MINSEQE, consider arrays and other omics, add higher-level instrument categories, and continue the LinkML formalisation and INSDC/ENA alignment.
- The Beacon team aims to:
    - Wrap up the current version by the end of the year, including: moving the repository to the GA4GH GitHub organisation, releasing v2.5.
    - Define and agree on the scope of Beacon v3.0, in line with the new GA4GH product development and approval process, separating committed features from tentative community requests.
    - Capture allele-frequency extension needs, missingness and genotype counts, and allele-frequency use cases (e.g. Genomics Thailand, Armenia, Genome of Europe).
    - Explore exposing dataset metadata once in several formats (Health DCAT-AP and Beacon), plus a possible files endpoint to support RO-Crate-style packaging.

Achieving clinical application of GA4GH products — a triad

Recording | Agenda | Slides

Building on the outcomes of previous Connect sessions that identified barriers to realising the potential of GA4GH standards in clinical genomics, this workshop shifted toward actionable execution.

Fifteen previously identified action items were categorised across three pillars, including:

1. Leadership and governance (e.g. demonstrating return on investment, regulatory compliance)
2. Technical alignment (e.g. internal harmonisation, building APIs/translators for HL7/FHIR compatibility, bridging data mapping gaps)
3. Showcase of external clinical implementations (e.g. developing tangible clinical use cases, refining feedback loops with the GA4GH Implementation Forum [GIF] and the National Initiatives Forum [NIF]).

The discussion focused on the “how” aspect of achieving application of GA4GH products in clinical infrastructure, and captured feedback from the Asian genomics community.

Key takeaways

- Recognition of the importance to intentionally work towards clinical integration should be a key objective of the GA4GH strategic plan.
- Develop concrete ideas and action items that can help guide GA4GH towards initiatives that will lower barriers to and encourage integration within clinical systems.
- Build the critical mass needed within the GA4GH community to grow and sustain efforts to bridge GA4GH products into clinical systems.
- Design for longitudinal reuse: “sequence once, analyse repeatedly.” National programmes presented at NIF and Plenary (Precision Health Research, Singapore [PRECISE], NHS England, Hong Kong Genome Project, Genome Canada, Mass General Hospital) and described genomic data used across a lifetime and across generations: newborn screening, pharmacogenomics, carrier status, cancer risk and cascade testing. Results must stay findable, linkable, and re-evaluable for decades. Narrative PDF reports and VCFs in the electronic health record (EHR) cannot support this today.
- Getting the Variation Representation Specification (VRS) into clinical systems is a top priority, together with reanalysis. The tables repeatedly named three gaps: variant reclassification and reanalysis as knowledge changes, clinical labs reporting VRS identifiers so clinicians can look up current knowledge in real time, and longitudinal phenotype representation. Patient identity linkage, family history, and diagnosis-coding support were also raised.
- Meet clinical systems where they are. Clinical systems are governed by regulated standards (e.g. IEC/ISO, FDA clearance) and departmental procurement, and there is no “genomic medicine department” that would buy a genomics stack. Mapping clinical data into GA4GH formats was seen as a barrier to adoption. The favoured approach is shims and query layers between FHIR and GA4GH APIs, with GA4GH filling gaps that clinical standards do not cover.

Priority action items

- Synthesise the report-outs into the next round of discussion and a community-driven mandate for GA4GH leadership.
- Scope out a FHIR to Beacon/Phenopackets shim for variant-level matching as a near-term option, with native FHIR import/export in Beacon or a SMART on FHIR app as longer-term goals. Steps towards Beacon FHIR support are already underway.
- Promote VRS adoption by clinical laboratories and in FHIR genomics reporting, so that variants in the electronic health record can be linked to current knowledge. (ClinVar, gnomAD, and other platforms are already VRS-enabled.)

- Invite contributors to the Phenopackets work on temporal elements to support longitudinal representation; compare with OMOP's longitudinal model.
- Explore family history standardisation (e.g. the Pedigree/KIN work) as a clear clinical gap, building on partnerships mentioned with Dana Farber, Epic and others.
- Explore regulatory and vendor pathways (e.g. certification, working with other standards development organisations such as ISO, engagement with EHR and device vendors). Hold virtual follow-up sessions on the agenda items not reached, with a focus on cross-Work Stream collaboration and developer-clinician feedback loops.

Pathogen genomic data sharing: aligning technical traceability with governance standards

Recording | Agenda | Slides

The Pathogen Genomic Data Sharing Group has developed five institutional strategies for operationalising equitable data sharing: governance structures, contractual benefit-sharing, traceability, monitoring and verification, and partnerships and attribution. Building on this work, the session drew on examples from pathogen genomic data repositories and platforms to explore how traceability (tracking where data comes from and how they are later used) can be supported in practice. The featured presentations by data contributors and repository and platform operators explored how pathogen genomic data and associated information are managed, including how information about data origin and their contributors is recorded and kept connected to genomic records as they are shared. The session provided practical context to inform the group's ongoing examination of how repository policies and other documentation addresses traceability.

Key takeaways

- Governance questions from the group's early work on traceability and the relevant considerations raised include:
    1. What information needs to remain traceable, and for what purposes?
        - Provenance and contributor information can be associated with genomic records in different ways and may not all persist through sharing and reuse.
    2. Who is responsible for maintaining the information needed to link data to their origin, contributors, and, where relevant, later use?
        - Responsibility may be distributed across contribution, submission, repository/platform processes, and conditions governing later use.
    3. What do repository and platform policies and agreements need to specify about tracking subsequent access to and reuse of data?

    - Information about later access or reuse may not always remain visible or linked back to the original record or contributor.
- Data generators, especially in low- and middle-income countries (LMICs), often receive little recognition, limited access to downstream analyses, and limited access to resulting vaccines and products.
- When pathogen genomic data are submitted to repositories through large consortia, laboratories that collected the samples or generated the data may not be clearly credited in the resulting records, making it difficult to cite their contributions in grant applications.
- Although an accession ID can identify a genomic data record in a repository, it does not necessarily ensure that the original data contributors are credited. One suggestion raised was for researchers conducting secondary analyses to contact and acknowledge those contributors. The role of funders and journals in supporting and encouraging attribution was also highlighted.
- Beyond attribution, participants also discussed traceability in the context of federated data sharing. While traceability has been central to the World Health Organization's Pathogen Access and Benefit-Sharing (PABS) negotiations, it is unclear how federated models from human genomics can support the rapid sharing of pathogen genomic data during emergencies.
- Other issues included trade-offs between open and restricted data sharing and the distinction between researcher attribution and benefit-sharing for communities.

Priority action items

- Use the practical examples and issues raised during the session to refine the governance questions guiding the group's examination of repository policies and other documentation.
- Convene a focused follow-up discussion with the GA4GH–PHA4GE Infectious Disease Community and relevant experts to assess whether the questions raised during the session capture the key governance issues arising in practice, identify any gaps, and determine which questions warrant further examination.

Expanding pipelines for rare familial cancer genomics to Asian and other ethnic populations

Recording | Agenda | Slides

The session explored how variant interpretation pipelines can better account for populations that are underrepresented in genomic reference datasets. The St. Jude team presented GRACE — the Germline Recognition, Annotation, and Classification Engine — and its founder-aware extension, which brings together ancestry-aware quality control, population-specific allele frequencies, family relationships, and variant classification. The aim is to make these methods available through a

portable, containerised pipeline that institutions can run locally, enabling broader evaluation without requiring them to share individual-level data.

Presentations from Botswana illustrated why broad ancestry categories and existing reference databases can provide insufficient context for interpretation. Research initially focused on HIV progression in children revealed substantial genetic diversity and population structure that were poorly represented in available datasets. Some variants that appeared rare in external references proved common locally, changing their interpretation. These findings helped build the case for the Botswana 50,000 Genomes Study. Speakers also described interest in using Beacon and trusted research environments to support sharing of aggregate allele frequencies and collaboration across African initiatives.

The Uganda Genome Resource presentation emphasised the ethical and social implications of studying populations at finer resolution. Participants discussed risks of re-identification, community stigmatisation, and findings affecting families or groups beyond the individuals enrolled. Meaningful, ongoing community involvement, clear data sharing arrangements, and equitable benefits were identified as essential. Discussion also highlighted how controlled access and time-limited embargoes can protect opportunities for local researchers to analyse their own data in settings with unequal analytical capacity.

The session also examined the reproducibility of AI-assisted analysis. Testing of the GRACE founder-aware workflow produced differences between users despite using the same inputs and instructions, demonstrating that packaging an analysis as an AI skill does not guarantee consistent results. The presenter invited collaborators to benchmark the pipeline on their own cohorts and help refine its calibration. A final discussion considered whether limited phenotypic information could accompany allele frequencies to improve interpretation, while recognising that appropriate access would depend on consent and the risk of identifying small communities.

Key takeaways

- Population-specific context is essential for accurate variant interpretation: broad ancestry categories and underrepresentative reference datasets can make locally common variants appear rare, underscoring the need for more diverse genomic resources and ancestry-aware pipelines.
- Collaboration must balance data utility with community protections and local equity: sharing aggregate allele frequencies could improve interpretation, supported by meaningful community involvement, consent-based access, and opportunities for local researchers to analyse their own data.
- Portable pipelines need rigorous validation: GRACE's locally deployable approach enables evaluation without sharing individual-level data, but variability in AI-assisted results highlights the need for cross-cohort benchmarking, calibration, and reproducibility testing.

Priority action items

- Discuss forming a GA4GH cross-Work-Stream working group with the Genomic Knowledge Standards, Discovery, Regulatory & Ethics, Clinical & Phenotypic Data Capture, and Data Security Work Streams to map existing non-disease allele-frequency resources for underrepresented populations, define gaps, and establish a federated data discovery road map using Beacon v2.0 and the Variation Representation Specification (VRS) makes sense.
- Gain feedback on various benchmarking and governance protocols for AI-assisted variant interpretation, in collaboration with the Data Security and Regulatory & Ethics Work Streams, covering data security, provenance, ancestry-aware performance evaluation, and human oversight. Draft initial guidelines for community review before the next GA4GH Plenary meeting.

GKS products, road map, and community feedback: implementation support across Work Streams

Recording | Agenda | Slides

Over the past six months since April Connect 2026, the Genomic Knowledge Standards (GKS) Work Stream has welcomed three new product groups from the Large Scale Genomics Work Stream: Whole Genome Sequencing Quality Control Standards (WGS-QC), File Formats (consisting of SAM/BAM/CRAM, VCF and BED) and the Future of VCF. Meanwhile, the longer-standing GKS product teams of VRS (Variant Representation Specification), Cat-VRS (Categorical Variant Representation Specification), and VA-Spec (Variant Annotation Specification) are coordinating efforts under the shared banner of the Genomic Knowledge Model (GKM), reflecting the high degree of integration between these products. In this GKS Work Stream session, the group reviewed recent progress across the various product teams, presented the road map ahead, and opened the floor for discussion on community priorities. The session also covered challenges related to implementation of GA4GH products, including engaging implementers and developing implementation resources.

Key takeaways

- GKS has grown, and community feedback keeps shaping it. WGS-QC, File Formats and Future of VCF have joined the Work Stream, and VRS, Cat-VRS, and VA-Spec now work together as the Genomic Knowledge Model (GKM). The teams are working towards new releases for VRS v2.1/2.1.1, Cat-VRS v1.1.1, and VA-Spec v1.1.
- WGS-QC v1.0 is an approved GA4GH product, and adoption is the next step. v1.0 covers germline short-read WGS through metric definitions, a Nextflow reference implementation, and functional-equivalence benchmarking, and PRECISE already uses it in vendor deliverables. Next steps are including somatic and long-read metrics, increasing uptake (e.g. by vendors, data producers and repositories), obtaining ISO approval through the

cross-standards-development-organisation (SDO) effort, and integration with Data Connect, the Data Repository Service (DRS), and Experiments Metadata Checklist (ExpMeta). Slido polls gathered views on core metrics.

- Implementers want tooling for adoption more than new features. Priorities raised include: integrating VRS in widely used tools (e.g. Ensemble Variant Effect Predictor, the Sequence Variant Classification v4.0 data model, Variant-Level Matching, Phenopackets); adding structural variant aggregation; including pharmacogenomics diplotypes, phasing, and versioned star alleles; human-readable VRS-aligned names; polygenic risk scores; VRS-to-VCF round-tripping (AnyVar); and a case-level evidence bridge between VA-Spec and Phenopackets. GKM plans to meet by project rather than product and to publish a public road map and status board.

Priority action items

- Move GKM to project-based meetings (bringing VRS, Cat-VRS and VA-Spec leads together around implementer projects) and publish a public road map and status board.
- Develop a star-allele model with ClinPGx, inviting input from the Hong Kong Genome Institute and MaveDB.
- Provide a mapping from VRS v2.0 to v2.1 for ReferenceLengthExpression repeat-subunit changes, for existing implementers.
- Support AnyVar adopters (e.g. Oslo University Hospital), making sure everyone uses consistent reference sequence sets and tests Cat-VRS in AnyVar.
- Promote the Structural Variant Aggregation Interest Group and the new APAC GKS Think Tank series.
- Circulate a follow-up poll on WGS QC, develop long-read and somatic metrics, pursue the ISO cross-SDO route, and hold an Open House webinar.
- Recruit volunteers for the File Formats and Future of VCF groups through the interest form for a hackathon or Think Tank (on topics such as CRAM next-generation compression, VCF scalability, validator).

Genomic metadata across the lifecycle

Recording | Agenda | Slides

Genomic data is only as findable and reusable as the metadata describing how it was produced, and aligning that metadata with the demands of AI is central to GA4GH's mission. This session walked through the early data lifecycle by area of metadata capture, mapping GA4GH standards to AI-readiness layers. The session also explored frameworks such as FAIRSCAPE (using RO-Crate, PROV-O/EVI) to capture the provenance, ethics, and computability needed to make data genuinely

AI-ready. The session brought people together to form consensus around practical metadata requirements and prioritise engagement with local and broader Asian genomics communities, so regional needs and use cases can shape the recommendations.

Key takeaways

- GA4GH covers much of the data lifecycle, but dataset-level provenance has no GA4GH standard. Pedigree/KIN, Phenopackets, Experiments Metadata Checklist (ExpMeta), Whole Genome Sequencing Quality Control (WGS QC) standards, and the Cloud APIs (WES, TRS, DRS, TES) cover families, participants, biosamples, experiments, quality control, and analysis. Describing a dataset's origin, funding, population, contributors, and context remains a gap that participants repeatedly raised. The April Connect 2026 discussion in Montreal has not yet had an owner to carry it forward.
- 'AI-ready' is mostly good data practice made explicit. The Bridge2AI criteria define seven dimensions (FAIRness, provenance, characterisation, pre-model explainability, ethics, sustainability, and computability) with a 28-criterion rubric, plus genomic-specific recommendations developed with Alex Wagner's team. RO-Crate packaging with PROV-O and EVI, and LinkML 'Datasheets for Datasets' aligned with RO-Crate and Croissant, show how to capture deep provenance. Participants noted that the same metadata makes data reusable by people too.
- Reuse, don't reinvent, and take ownership. Participants pointed to RO-Crate (and a possible GA4GH profile), RAiD, ROR, and ORCID for crediting contributors, NLM's DATM dataset metadata schema and MINSEQE. They also warned against putting AI over unharmonised data due to access and risk. There was support for a GA4GH study group to produce a landscape review and recommendations, not a new standard, and several volunteers came forward.

Priority action items

- Establish a GA4GH study group on metadata across the data lifecycle with a defined scope and output (e.g. landscape review and white paper), meeting every two to three months at rotating times that suit APAC time zones.
- Run a landscape assessment covering RO-Crate (including a possible GA4GH profile), DATM, MINSEQE, Health DCAT-AP, RAiD/ROR/ORCID, and existing consortium schemas, identifying what falls between existing GA4GH standards.
- Map WGS QC outputs to the Bridge2AI QC recommendations, building on the JSON-LD output of the reference implementation.
- Coordinate with the AI Work Stream, the Technical Alignment Subcommittee (TASC), the Ethical Provenance Toolkit team from the Regulatory & Ethics Work Stream, the ExpMeta product team, and the Beacon product team so that recommendations line up across GA4GH.

- Collect implementation success stories to share with the community.

Building a more representative human reference: a working session for joining the Human Pangenome Project

Recording | Agenda | Slides

The session focused on two complementary goals of the Human Pangenome Project (HPP): constructing a globally representative reference and enabling researchers and clinicians to use it. Speakers outlined the need to coordinate international initiatives that differ in sequencing methods, data access arrangements, consent, and infrastructure. HPP provides a framework for collaboration on protocols, quality assessment, tools, and education, with participation open to both data contributors and those developing analyses or applications.

The HPP telomere-to-telomere (T2T) task team described its work toward a stable reference release, with an ambition to include approximately 1,000 diploid genomes, or 2,000 phased haplotypes. Shared, openly consented 1000 Genomes cell lines provide a practical foundation for international collaboration, while additional samples undergo consent compatibility and representation review. Speakers emphasized that contributions can include sequencing, assembly, software development, and methodological expertise. Scaling complete genome assembly remains challenging, making shared workflows, benchmarking, and capacity building important parts of the effort.

Updates from Japan and Singapore showed how local initiatives can complement the global reference and support clinical research. Japan's work is motivated partly by unresolved rare disease cases and plans to reanalyse existing short-read data using pangenome resources. Singapore described a combination of deeply characterised reference genomes and larger sequencing cohorts, spanning both open and controlled-access data. Discussion cautioned against treating national or broad population labels as complete descriptions of genetic diversity and emphasised the importance of capturing variation within those categories.

Consent, privacy, and governance were major themes throughout the session. Speakers discussed enabling voluntary participation in open-reference research while clearly explaining long-term data availability, re-identification risks, and implications for relatives. Participants raised unresolved questions about withdrawal, GDPR, data sovereignty, and the governance of a stable international reference. HPP described ongoing consent review and legal consultation, alongside a developing privacy working group to explore the use of restricted data in pangenome research.

The final part of the session covered practical adoption. Tools can already use pangenome information to improve mapping and variant calling while returning results in familiar reference coordinates, helping users incorporate these approaches into existing workflows. However, this does not fully capture sequences and structural variation absent from conventional references, and reporting that information remains an active development area. Discussion concluded with interest in a task team or

pilot bringing together groups with local pangenomes and long-read datasets to explore interoperability, quality requirements, and ways to incorporate local data into the global reference within secure environments.

Key takeaways

- A representative pangenome requires coordinated international contributions and attention to diversity within populations.
- Stable releases, quality standards, and usable tools are essential for broader adoption.
- Compatibility with existing reference coordinates supports uptake but does not capture all the benefits of pangenomes.
- Integrating open and controlled-access resources remains a significant technical and governance challenge.

Priority action items

- HPP and partners aim to continue coordinated sequencing, assembly, benchmarking, and consent review toward a stable release.
- Interested participants are welcome to join the HPP community and developing privacy working group
- Governance teams aim to continue addressing consent compatibility, withdrawal, and jurisdiction-specific requirements for the reference resource.

Federated Analysis hackathon: ship real code, together

Recording Part 1 | Recording Part 2 | Agenda | Slides

The GA4GH Federated Analysis Work Stream held a hackathon to focus on federated learning (FL). FL enables collaborative model training across platforms without sharing raw data, improving privacy and leveraging GA4GH standards such as the Workflow Execution Service (WES), Task Execution Service (TES), Data Repository Service (DRS), Passports, and the Authorisation and Authentication Infrastructure (AAI). The session provided a collaborative space to prioritise a backlog of follow-up work, and to conduct a hackathon to turn one to three high-priority items into prototypes or draft pull requests.

Key takeaways

- The work of the Federated Analysis Work Stream (FAWS) is shifting from API-by-API focused work to project-based collaboration with GA4GH Implementation Forum (GIF) projects.

- Emerging standards for FAWS to look at include Portable Format for Bioinformatics (PFBs) and Mesh Cards.
- The Reference Cloud was successfully deployed locally using Docker Compose.
- A "Poke Chop" framework was introduced to scan for privacy risks in workflow outputs.
- An experimental integration of language models with WES API was explored for future workflow execution.
- WES API conformance testing is ongoing, with partial v1.1 compliance.
- The GA4GH Service Registry ecosystem needs clarity on service deployment vs. codebase definitions.
- A "Federation of federations" model was proposed for local-to-global registry interoperability.
- Authorisation passing methods are being refined for a Passports v2.0 and AAI release

Priority action items

- Reference Cloud and deployment
    - Continue WES installation and tooling improvements.
    - Implement local directory support for DRS and containerised Postgres.
    - Develop orchestrator components and htsget integration.
    - Develop and improve Reference Cloud Starter Kit (Docker container/Helm chart) for federated FAWS API deployment.
- Security and conformance
    - Perform security checks on the WES API backend and refine the Poke Chop framework.
    - Develop security guidelines and vulnerability checking procedures for WES implementations.
    - Complete WES conformance testing of the version 1.1 specification.
- Registry and interoperability
    - Investigate GA4GH Service Registry API status and clarify service vs. codebase distinctions.
    - Explore language model integration with the WES API and address deployment risks.
- Documentation and onboarding
    - Review the Federated Analysis Work Stream website and documentation; identify gaps for newcomers.

- Document Mesh Cards ecosystem descriptions and clarify how components relate to each other.
- Refine authorisation passing structures for Passports and AAI v2.0 release.

GKS Starter Kit and real-world implementations

Recording | Agenda | Slides

Adopting GA4GH standards can feel daunting to newcomers. Implementers repeatedly mention that the hardest part is not the specification, but the on-ramp: knowing where to start, having runnable examples, and being able to show colleagues that others have already succeeded.

This session introduced the Genomic Knowledge Standards Starter Kit, driven by real-world datasets represented using the Genomic Knowledge Model (GKM) specifications: the Variation Representation Specification (VRS), Categorical Variation Representation Specification (Cat-VRS), and Variant Annotation Specification (VA-Spec). A collection of real-world implementation vignettes using these data, along with a lightweight Python toolkit that enables these analyses, were also shared. Together, these components provide a comprehensive introduction to the GKM, designed to let implementers spin up and demonstrate GA4GH Genomic Knowledge Standards on a local machine in minutes. The session included a live demonstration of how to get started with real-world data from resources informing variant interpretation such as ClinVar, CIViC, and MaveDB.

Key takeaways

- The Starter Kit gives the GKM a practical on-ramp. It has three pillars: data (bundles), tools (the ga4gh.gkm Python toolkit, alongside the VRS, Cat-VRS and VA-Spec reference implementations), and user stories. Bundles use local JSON pointers so each object is defined once and referenced elsewhere, which cut MaveDB's representative dataset by about 90%. The toolkit loads, validates, resolves, and normalises bundles against the latest releases.
- Real production resources already ship GKM data. ClinVar-GKM publishes all of ClinVar (SCVs, VCVs, RCVs) as a single cross-linked JSON bundle plus typed Parquet, with monthly full releases and weekly deltas; Epic and the ClinGen Linked Data Hub plan to use it; and MaveDB uses VRS, Cat-VRS and VA-Spec to share functional evidence with ClinVar, ClinGen, VEP, and others. CIViC shows how its native concepts map onto the GKM.
- In terms of feedback, participants asked for query recipes (Parquet/DuckDB, JSON pointers as foreign keys), shared keying conventions so bundles from different resources stay interoperable, and a broader catalogue of ecosystem tools (e.g. AnyVar for annotation use cases). They also wanted a clear value proposition compared with existing ClinVar XML pipelines. Some questioned whether the name 'Starter Kit' undersells bundles, which is the core way to share GKM data.

Priority action items

- Add query recipes to the Starter Kit (e.g. converting bundle sections to tables, JSON pointers to foreign keys, DuckDB/Parquet examples).
- Draft conventions for shared bundle sections and keys (e.g. genes keyed by HGNC ID, shared variant and location sections), and consider a tool that compacts inline documents into bundles.
- Catalogue the wider GKM tool ecosystem (including AnyVar, vrs-python, cat-vrs-python, va-spec-python, and the toolkit) with real-world examples, and revisit the 'Starter Kit' name and framing.
- Encourage early adopters to test ClinVar-GKM and report issues or upvote road map items on GitHub.
- Invite the community to share user stories through pull requests, suggestions for more datasets, and producer bundles for the public bundle repository. Continue exploring Parquet distribution for very large datasets.

Passport/AAI v2.0 and beyond

Recording | Agenda | Slides

This session provided an update on the ongoing development of Passport and AAI v2.0, focusing on the motivations for the proposed changes as well as updates aimed at improving scalability, security, and implementation flexibility across federated research ecosystems. Participants discussed challenges associated with the current Passports model, particularly the growing size and complexity of authorisation payloads, and explored a proposed task-specific token approach for managing authorisation information more efficiently. The discussion also examined Passport Lite, which would decouple authentication from authorisation, as well as parallel efforts to explore Verifiable Credentials as a complementary approach for representing visas and authorisation information.

Key takeaways

- Discussions explored the motivations for Passport and AAI v2.0, including opportunities to simplify implementation, improve scalability, strengthen security, and address lessons learned from existing deployments.
- The proposed Task-Specific Token approach was presented as a mechanism for reducing Passport size by enabling authorisation information to be scoped to a specific task or request.
- Discussions examined Passport Lite, a model that would separate authentication from authorisation and provide greater flexibility for ecosystems that manage authorisation outside of the Passport infrastructure.

- Ongoing work on Verifiable Credentials was presented as a complementary effort that could provide an alternative approach for representing visas and authorisation information.
- Community feedback was identified as an important part of the process as the Work Stream continues to refine proposals, develop proof-of-concept implementations, and evaluate different deployment models.

Priority action items

- Continue refining the Passport and AAI v2.0 proposals, including the Task-Specific Token architecture and related authorisation workflows.
- Develop a proof-of-concept ecosystem and associated benchmarks to evaluate performance, scalability, and security considerations ahead of April Connect 2027.
- Aim for a potential v2.0 release by the forthcoming Plenary.
- Explore edge cases involving hybrid deployments, including scenarios that combine task-specific tokens and full Passports, as well as downstream token exchange and progressive downscoping.
- Expand current resources, including documentation, FAQs, implementation guidance, and reference materials to support adopters.
- Review the PR issues #137 and #122. A rendered web view is also available online: https://ga4gh.github.io/aai/2.0-draft-agearh-tst/.

National Initiatives Forum (NIF)

Recording | Slides

NIF Day 2026 brought together 112 in-person participants from 25 nations to share progress, challenges, and practical approaches to implementing genomics at population scale. Across sessions on infrastructure, genomic diversity, and clinical implementation, a consistent message emerged: sequencing capacity must be matched by sustainable funding, effective governance, usable data systems, workforce development, and evidence of patient benefit. Presentations illustrated different national approaches while discussions identified opportunities to reuse resources, strengthen international collaboration, and guide the development and implementation of GA4GH standards. Equity and public trust were recurring themes, encompassing population representation, meaningful community involvement, and ensuring that the benefits of genomic developments reach those contributing their data.

State of the National Initiatives Forum

The opening session reaffirmed NIF's mission to facilitate the delivery of genomics into healthcare and promote safe and responsible sharing of genomic and clinical data globally. The chairs described NIF as a forum for exchanging implementation experience, identifying common challenges, developing collaborations, and feeding real-world requirements into GA4GH Work Streams. The refreshed governance structure, including regular NIF Council meetings and a plan to launch a renewed Steering Committee, was introduced as a means of maintaining engagement and shaping future activities.

An opening poll highlighted the wide range of maturity across initiatives, from programmes preparing to launch to established clinical services and large research resources. Common challenges included funding and sustainability, data sharing and interoperability, governance, and clinical implementation. Participants also identified experience they could share, including governance frameworks, community engagement approaches, education resources, and evidence of clinical and non-clinical value. These contributions informed lunchtime topic tables and provided input for future NIF priorities.

Genomic infrastructure: architecture, access, and equity

Four presentations explored infrastructure choices and the trade-offs between security, usability, scalability, and sustainability.

- **Genomics England**: Richard Scott described the transition to a fully cloud-based research environment, separating the data layer from research tools and using GA4GH standards to support flexibility and reduce dependence on individual providers. This approach allows internal teams to focus increasingly on data quality and curation.
- **German Human Genome Archive (GHGA)**: Juliane Winkelmann outlined a federated infrastructure with regional data hubs, centrally coordinated access, and continued control by submitting institutions. GHGA's connection to Germany's clinical sequencing programme illustrated how reimbursement requirements can incentivise the submission of genomic and clinical data.
- **H3Africa**: Nicola Mulder described efforts to connect datasets across countries with differing infrastructure, legal requirements, and resources. Priorities include harmonisation, locally relevant reference resources, workforce capacity, and lightweight tools for federated analysis, including in settings where cloud use is impractical.
- **Federated EGA**: Mallory Freeberg outlined the development of a network of repositories that retain data within local jurisdictions while sharing metadata for discovery. Reusable software, transparent policies, shared values, and collaboration between nodes were central to the network's development.

During the discussion, participants emphasised that infrastructure decisions depend on national circumstances and sustainable operating models. Cloud services can reduce the burden of maintaining infrastructure, but recurring costs, procurement arrangements, and funding uncertainty can make on-premises approaches more practical elsewhere. Staffing, cybersecurity, data stewardship, and ongoing operations need to be included in cost assessments.

Metadata collection remains a substantial challenge: rich data models are only useful if contributors can populate them. Examples of useful approaches included dedicated data stewards, collaborative curation events, and adaptable models that can accommodate new data types.

The discussion also distinguished technical connectivity from practical research interoperability. Connecting repositories does not automatically make their data straightforward to analyse together; progress requires specific use cases and feedback from researchers. Participants called for more implementation kits, clear documentation, and peer support, alongside simple, scalable standards. Participants discussed data downloading and data visiting as models whose suitability depends on consent, governance, and the responsibility of data controllers.

Genomic diversity: building more diverse and representative genomic reference resources

This session examined how national initiatives can improve the representation and usefulness of genomic reference resources:

- **Human Pangenome Project**: Karen Miga described progress towards a more complete and diverse reference resource, incorporating high-quality telomere-to-telomere assemblies. She highlighted opportunities to improve variant detection and interpretation and encouraged initiatives to begin pilot work with the available resources and tools.
- **Federated gnomAD and variant-level matching**: Heidi Rehm outlined how locally processed, aggregate allele frequencies can expand gnomAD's population representation without transferring individual-level data. She also presented the Federated Variant-Level Matching network, which uses GA4GH Beacon and Phenopackets to connect variant and phenotype information for rare disease interpretation.
- **Pan-Canadian Genome Library**: Guillaume Bourque described efforts to bring together genomic and health data while embedding diversity and inclusion. The presentation highlighted Indigenous-led governance arrangements, the need for better sociodemographic data collection, and exploratory work on pangenome approaches.
- **Precision Health Research, Singapore (PRECISE)**: Nicolas Bertin presented genomic diversity work in Singapore, including contributions from Chinese, Malay, and Indian populations, long-read sequencing, structural variant and repeat analysis, pangenome resources, and participation in federated gnomAD. He also outlined the programme's transition towards

hospital-based recruitment and the repeated clinical use of genomic information over a patient's lifetime.

Several themes emerged from the discussion:

- **Technical readiness and adoption**
    - Participants explored the implications of combining short- and long-read data, different sequencing platforms, and different analytical pipelines. Quality metrics and provenance will be essential to distinguish genuine biological differences from technical artefacts. Pangenome adoption also raises questions about variant representation, annotation, and compatibility with existing clinical workflows.
    - Accessible tools were viewed as essential for wider adoption, particularly in laboratories with limited bioinformatics support. Participants stressed that advanced resources must be available through practical interfaces and established workflows, alongside training and adequate investment.
- **Participation, trust, and benefit sharing**
    - Staffing constraints, competing priorities, local computing requirements, and consent compatibility were identified as barriers to participation. Initiatives were encouraged to engage early with international projects so that local databases, consent processes, and pipelines can be designed with future interoperability in mind. The discussion also stressed the importance of accommodating multiple sequencing technologies, particularly those used in lower-resource settings.
    - Participants highlighted the need to recognise diversity within countries and broad ancestry categories, and to involve communities in determining how their data are governed and used. Historical experiences of extractive research, concerns about commercial use, and Indigenous data sovereignty require sustained engagement. Demonstrating benefits to contributing communities was considered central to trust, while participants also noted the responsibility to enable sufficient data use to fulfil the purpose of participants' contributions.

Integrating genomics into healthcare: what is actually working?

This session focused on the full pathway from genomic data to clinical action, including service design, evaluation, workforce readiness, and securing funding. Presentations illustrated different national implementation approaches and learnings.

- **England**: Dame Sue Hill outlined plans for an NHS genomics population health service extending into primary and community care. Priority applications included inherited disease risk,

pharmacogenomics, and polygenic risk scores, supported by phased implementation, workforce education, digital infrastructure, and evaluation of outcomes and equity. Rich Scott described the complementary evidence-generation role of Genomics England's Generation Study and developing Adult Population Genomics Programme.

- **Hong Kong**: Brian Chung described an evaluation programme covering technical performance, clinical utility, and patient and societal outcomes. Findings on diagnosis, clinical management, genomic literacy, and psychosocial effects are informing discussions about integration into routine care.
- **Sweden**: Anna Lindstrand presented a national long-read sequencing implementation project conducted alongside standard care. The work highlighted benefits for phasing, structural variant interpretation, and reducing follow-up testing, while demonstrating the importance of local reference datasets, clinical validation, interpretation tools, and staff training.
- **Indonesia**: Ines Atmosukarto described how the Biomedical and Genome Science Initiative (BGSi) developed sequencing, biobanking, and data infrastructure alongside hospital and primary care partnerships. Pharmacogenomics was presented as an initial route to demonstrating locally relevant clinical and economic value, with results prioritised around actionable medicines available within the national formulary.
- **Aotearoa New Zealand**: Christopher Print described using the Beyond 1 Million Genomes maturity model to assess national readiness and build collaboration. Work with Māori leaders added an Indigenous genomics and equity domain. A contribution from Huti Watson posed the question of whether a separate domain was sufficient, or whether shared authority, Indigenous knowledge, data sovereignty, and equity should shape the entire framework.

Several themes emerged from the discussion:

- **Population-specific evidence**: Participants questioned how reliably pharmacogenomic findings transfer across populations. Better ancestry-aware interpretation, clinical outcome data, and documentation of adverse drug reactions are needed.
- **Long-term clinical use:** Newborn sequencing was discussed as a possible foundation for lifelong genomic care, while acknowledging that adoption requires evidence and formal screening decisions. Systems also need to accommodate future sequencing technologies and multi-omics.
- **Integration with clinical systems**: Participants identified a continuing gap between genomic resources and information clinicians can act on within electronic health records. Collaboration with record-system and decision-support vendors was seen as important for making standards adoption more consistent.

- **Evidence and practical collaboration**: Shared economic evaluations, implementation resources, and direct support between initiatives could help programmes build investment cases. Small collaborations between interested countries were suggested as a practical starting point.
- **Education, engagement, and workforce**: The discussion supported complementary approaches involving schools, professional training, public engagement, and clear communication with policymakers. Genetic counsellors were recognised as essential, with scarce specialist capacity focused on complex needs and other healthcare staff trained for defined tasks. Participants cautioned against overselling genomics and stressed the importance of listening to public concerns.

Work plan and next steps

The closing session positioned NIF as both a source of implementation feedback for GA4GH and a community through which initiatives can learn from one another. The value of sharing evidence, adapting approaches from comparable health systems, using implementation kits, and designing infrastructure and services for future needs was highlighted. Collaboration on federated gnomAD and New Zealand's use of the maturity model were cited as examples of relationships developed through NIF leading to practical work.

Participants ranked topics to inform the 2026-2027 work plan. Data infrastructure, interoperability, and collaboration, together with clinical implementation, emerged as leading priorities. Other priorities included governance, consent and public trust; national strategies; funding, evidence and sustainability; equity and genomic diversity; and workforce development. The closing discussion emphasised that all of the priority topics remained important to the work of national initiatives, and how opportunities for sharing evidence and strengthening peer relationships across the global landscape is key to sustained progress.

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